CURRICULUM VITAE

MEHUL TULSIDAS DATTANI

MBBS DCH FRCP FRCPCH MD FMEDSCI

Personal Details

Nationality	British
Present Appointment	**Professor of Paediatric Endocrinology** UCL Institute of Child Health, London/ Honorary Consultant in Paediatric and Adolescent Endocrinology, Great Ormond Street Hospital for Children and University College London Hospital
Date of Appointment	October 1st 2006
Department	Molecular Basis of Rare Diseases Section Genetics and Genomic Medicine Programme UCL GOS Institute of Child Health University College London 30 Guilford Street London WC1N 1EH
Telephone no.	0207 905 2657
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GMC Registration no.	2942717
Good Clinical Practice (GCP) Refresher Date	May 22 2022

Education/Qualifications

Education	Whitmore High School, Harrow Pinner Sixth Form College The Middlesex Hospital Medical School
Qualifications	1984 Bachelor of Medicine, Bachelor of Surgery (MB BS; London) 1987 Diploma in Child Health (DCH; London) 1988 Membership of Royal College of Physicians (MRCP; London) 1994 Doctor of Medicine (MD; London): Measurement of growth hormone bioactivity using a lactogenic eluted stain bioassay (ESTA) 1999 Fellowship of Royal College of Paediatrics and Child Health (FRCPCH; London) 2000 Fellowship of Royal College of Physicians (FRCP; London) 2023 Fellowship of the Academy of Medical Sciences 2023
Distinctions/Merits	1981 Merit in Pharmacology 1984 Merit in Clinical Pharmacology and Therapeutics

Professional History

Previous Appointments

01/08/84-31/01/85	House Physician, Whittington Hospital, London
01/02/85-31/07/85	House Surgeon, St Alban's City Hospital, Hertfordshire
01/02/86-30/04/86	Casualty Officer, Whittington Hospital, London
01/05/86-31/10/86	Senior House Officer (SHO) Paediatrics, Edgware General Hospital, London
01/11/86-30/04/87	SHO Paediatric Endocrinology, The Middlesex Hospital, London
01/05/87-31/10/87	SHO Neonatology, Bristol Maternity Hospital, Bristol
01/02/88-31/07/88	SHO Paediatric Cardiology/Respiratory Medicine, The Brompton Hospital, London
01/08/88-31/01/89	SHO Paediatric Haematology & Nephrology, Hospital for Sick Children, Great Ormond Street, London
01/02/89-28/02/90	Registrar in General Paediatrics, Edgware General Hospital, London
01/03/90-28/02/91	Registrar in Paediatric Endocrinology/Oncology, The Middlesex Hospital, London
01/03/91-31/08/94	Research Fellow in Paediatric Endocrinology, The Middlesex Hospital, London
01/09/94-30/09/98	Clinical Lecturer in Paediatric Endocrinology, Institute of Child Health, London, and Great Ormond Street Children's Hospital, London
01/10/98-30/09/04	Senior Lecturer in Paediatric Endocrinology, Institute of Child Health, London; Honorary Consultant in Paediatric Endocrinology, Great Ormond Street Children's Hospital and University College London Hospital, London
01/10/04-30/09/06	Reader in Paediatric Endocrinology, Great Ormond Street Children's Hospital and University College London Hospital, London

Other Appointments and Affiliations

Membership of learned societies

American Endocrine Society
British Society for Paediatric Endocrinology and Diabetes
European Society for Paediatric Endocrinology
Society for Endocrinology
Royal College of Physicians
Royal College of Paediatrics and Child Health
Academic Paediatrics Association
Growth Hormone Research Society
European Society for Endocrinology

Membership of Advisory Boards

Member of Medical Research Council College of Experts (2005 - 2009)
Member of GAIN (Genentech and Ipsen Network) International Advisory Board (2004 - 2007)
Member of Growth Hormone Advisory Group for submission to the National Institute of Clinical Excellence
Member of Advisory Boards to Ferring, Ipsen, Novo, Pfizer and Serono

Editorial Boards and others

Member of Editorial Board for *Clinical Endocrinology* (2004-2007). This is the premier British Clinical Endocrinology Journal.
Member of Editorial Board for *Hormone Research* (2007 onwards). This is the journal of the European Society for Paediatric Endocrinology.
Member of the Editorial Board for *Journal of Paediatric Endocrinology and Metabolism* (2007 onwards).
Member of the Editorial Board for *International Journal of Paediatric Endocrinology* 2009-2015.
Associate Editor "Growth and Growth factors" and "Pituitary Disease", *Yearbook of Paediatric Endocrinology* 2007-12.
Appointed as a Non-Executive Director of The Company of Biologists Limited 2007 - 2013.
Appointed to the Advisory Board of *Disease Models and Mechanisms* 2007 - 2013.
Associate Editor *Hormone Research* – lead Editor for Mini-Reviews (2008 onwards).
Member of the editorial board for *Journal of Clinical Endocrinology and Metabolism* (2008 - 2012). This is the premier international journal of clinical endocrinology.
Member of Editorial Board of *Frontiers in Paediatric Endocrinology* (2010-3).
Specialty Chief Editor of *Frontiers in Genomic Endocrinology* (2010-3).
Specialty Chief Editor of *Frontiers in Paediatric Endocrinology* (2013-2015).
Advisory Board to *Nature Reviews in Endocrinology* (2012 onwards)
Senior Editor *Endocrinology, Diabetes and Metabolism Case Reports* (2013 - 2017)
Associate Editor *Journal of Pediatric Endocrinology and Metabolism* (2015 - 2019)
Senior Editor, *Endocrine Connections* (2021-current)

Examining Boards

Examiner to the Royal College of Paediatrics and Child Health (MRCPCH, DCH)
Member of Specialist Question-writing Group, RCPCH, 2007 – 2010
Appointed Senior Examiner RCPCH, 2018

Peer Review Activities

1. Grant Peer Reviews

I have reviewed grants for the following organizations:

Action Research, Birmingham Children's Hospital Children Nationwide, Child Health Research Appeal Trust (CHRAT), BBSRC, European Society for Pediatric Endocrinology, Medical Research Council UK, Inserm, Michigan Diabetes and Endocrinology Centre, GENOPAT, Qatar Foundation, Wellcome Trust

2. Review of Articles

I have refereed articles for the following journals:

Acta Paediatrica, *American Journal of Human Genetics, Archives of Diseases in Childhood, Canadian Journal of Medicine, Clinical Dysmorphology, Clinical Endocrinology, Developmental Biology*, *Endocrinology*, *European Journal of Endocrinology, European Journal of Medical Genetics*, *Genome Medicine*, *Growth Hormone and IGF1 Research*, *Hormone Research, Human Molecular Genetics, Journal of Clinical Endocrinology and Metabolism, Journal of Clinical Investigation, Journal of Medical Genetics, Journal of Pediatrics*, *Journal of Pediatric Endocrinology and Metabolism, Molecular and Cellular Endocrinology*, *Lancet*, *Lancet Diabetes and Endocrinology*, *Lancet Child and Adolescent Health*, *Nature Reviews in Endocrinology, Neuroendocrinology, NEJM, Pediatric Research, Pituitary, PLOS Genetics*

3. Abstract marking

I have marked abstracts submitted for annual meetings of the following societies:

British Society for Paediatric Endocrinology and Diabetes, European Society for Paediatric Endocrinology, International Society for Endocrinology, Society for Endocrinology, European Federation of Endocrine Societies, Endocrine Society USA, European Society for Endocrinology.

4. PhD examinations:

Appointed as examiner to 11 PhDs:
Dr. Nicola Solomon
Dr. Michelle Pelling
Dr. Dan Hanson
Dr. Harvinder Chahal
Dr. Claire Hughes
Dr. Yu Sun
Dr. Seley Gharanei
Dr. Julia Kowalczyck
Dr. Nitash Zwaveling-Soonawala
Dr. Reena Perchard
Dr. Cecile Brachet

5. Appointments to Peer Review Committees

1. Appointed Chair of the Scientific Evaluation Committee for the EU-funded E-Rare Call for "European research projects on rare diseases" 2007.
2. Appointed to Local ACCEA committee based at GOSH 2007-present.
3. Appointed to CEA committee of the British Society for Paediatric Endocrinology and Diabetes

4. Invited to give evidence at "Growth Hormone Therapy and Biosimilars in Clinical Practice Stakeholder Survey and Parliamentary Summit" meeting, Houses of Parliament, February 24 2009
5. Appointed to MCRN/BSPED CSG 2010-current
6. Appointed to Clinical Reference Group for Specialist Commissioning Paediatric Medicine, 2013-2016
7. Member of Scientific Advisory Board, Alliance4Rare, The Eva Luise and Horst Köhler Foundation for Rare Diseases

Prizes, Awards and other Honours

Prizes

Royal College of Paediatrics and Child Health 1999: Donald-Paterson Prize for best scientific paper published over the preceding two years [Mutations in the homeobox gene *HESX1/Hesx1* associated with septo-optic dysplasia in human and mouse. *Nature Genetics* **19(2):** 125-133].

European Society for Paediatric Endocrinology 1998: Henning-Andersen Prize for best abstract (*HESX1:* a novel homeobox gene implicated in septo-optic dysplasia).

European Society for Paediatric Endocrinology 2010: Henning-Andersen Prize for best abstract (Enhancement of the canonical Wnt pathway in Rathke's pouch results in pituitary tumours reminiscent of human adamantinomatous craniopharyngioma; awarded to Dr. Carles Gaston-Massuet)

British Society for Paediatric Endocrinology and Diabetes 2010: Award for best abstract to Dr. Carles Gaston-Massuet (Enhancement of the canonical Wnt pathway in Rathke's pouch results in pituitary tumours reminiscent of human adamantinomatous craniopharyngioma)

Endocrine Society USA 2011: Young Investigator Award to Dr. Carles Gasto-Massuet (**The Wnt/β-Catenin effector Tcf3 is required for normal hypothalamic-pituitary development)**

Royal College of Paediatrics and Child Health 2013: Donald-Paterson Prize for best scientific paper published over the preceding two years to Dr. Emma Webb [Webb EA, O'Reilly MA, Clayden JD, Seunarine KK, Chong WK, Dale N, Salt A, Clark CA, **Dattani MT** (2012) Effect of growth hormone deficiency on brain structure, motor function and cognition. *Brain 135 (Part 1): 216-227*]

Other Prize-winning abstracts

Kelberman D, Mundlos S, Grueters A, **Dattani MT** (2008) Polyalanine tract mutations within the transcription factor SOX3 are associated with variable hypopituitarism. 90th Annual Meeting of the Endocrine Society USA, San Francisco, California, USA. **Awarded Best Poster Prize in section.**

Schoenmakers N, Bochukova E, Agostini M, Schoenmakers E, Rajanayagam O, Keogh J, Henning E, Reinemund J, Gevers E, Sarri M, Offiah A, Albanese A, Halsall D, Schwabe J, Bain M, Lindley K, Muntoni F, Vargha-Khadem F, **Dattani MT**, Farooqi IS, Gurnell M, Chatterjee K (2012) Growth retardation and severe constipation due to the first human, dominant negative thyroid hormone receptor α mutation. Annual Meeting of the Society for Endocrinology, Harrogate, UK. **Winner Young Endocrinologist Clinical Prize.**

Andoniadou C, Gaston-Massuet C, Reddy R, Jacques T, **Dattani M**, Martinez-Barbera JP (2012) New insights into the molecular and cellular pathogenesis of human craniopharyngioma: do pituitary stem cells underlie the origin of these tumours. 15th International and 14th European Congress of Endocrinology (ICE/ECE 2012). **Winner of best basic science poster prize.**

Sun Y, Bak B, Schoenmakers N, van Trotsenburg ASP, Oostdijk W, Voshol P, Cambridge E, White JK, le Tissier P, Mousavy Gharavy SN, Martinez-Barbera JP, Stokvis-Brantsma WH, Vulsma T, Kempers MJ, Persani L, Campi I, Bonomi M, Beck-Peccoz P, Zhu H, Davis TME, Hokken-Koelega ACS, Del Blanco DG, Rangasami JJ , Ruivenkamp CAL, Laros JFL, Kriek M, Kant SG, Bosch CAJ, Biermasz NR, Appelman-Dijkstra NM, Corssmit EP, Hovens GCJ, Pereira AM, den Dunnen JT, Breuning MH, Hennekam RC, Chatterjee KK*, **Dattani MT***, Wit JM*, Bernard DJ* (*Co-Senior Authors) (2013) Loss-of-function mutations in IGSF1 cause a novel, X-linked syndrome of central hypothyroidism and testicular enlargement. Annual Meeting of the Society for Endocrinology. **Winner of Clinical Endocrinology Trust Prize for best clinical abstract.**

Andoniadou CL, Le Tissier PR, Pevny LH, **Dattani MT**, Martinez-Barbera JP (2013) The SOX2+ population of the adult murine pituitary includes stem cells with paracrine tumor-inducing potential. 95th Annual Meeting of the Endocrine Society USA, San Francisco, USA. **Winner of Mara E. Lieberman Memorial Travel Grant Award for outstanding abstract submitted by a woman.**

Gaston-Massuet C, Le Tissier PR, **Dattani MT**, Martinez-Barbera JP (2013) Genetic and in vitro inhibition of the Wnt/β catenin pathway results in amelioration of adamantinomatous craniopharyngioma. 95th Annual Meeting of the Endocrine Society USA, San Francisco, USA. **Winner of Outstanding Abstract Award.**

Gregory L, Rhodes S, Humayun K, Levy MJ, Greening J, **Dattani MT** (2014) Novel Lethal Form of Hypopituitarism Associated with the First Recessive LHX4 Mutation. 96th Annual Meeting of the Endocrine Society USA, Chicago, USA. **Winner of Outstanding Abstract Award.**

Gregory LC, Shah P, Sanner J, Arancibia M, Hurst J, Jones W, Spoudeas H, Le Quesne Stabej P, Ocaka L, Loureiro C, Martinez-Aguayo A, Williams H, **Dattani MT (2018)** Mutations In MAGEL2. And L1CAM Are Associated With Congenital Hypopituitarism And Arthrogryposis Annual Meeting of the Endocrine Society USA, Chicago, USA. **Winner of Outstanding Abstract Award.**

Gregory LC, A novel missense variant in the gene encoding Fatty Acid Synthase (FASN) associated with a unique multi-system disorder including hypopituitarism and hypoparathyroidism. (2022) Annual Meeting of the Endocrine Society USA, Atlanta, USA. **Winner of Outstanding Abstract Award.**

Awards/Achievements

Endocrine Society USA 1998: Quest Diagnostics Young Investigator Award (*HESX1:* a novel homeobox gene implicated in septo-optic dysplasia).

Appointed as NIHR Investigator, 2006

BSPED Merck-Serono award for best research submission
£25,000
Dr. Vaitsa Tziaferi, **Professor Mehul Dattani (PI)**

2012: Listed in *The Times* Top 100 UK Children's Specialists List

2014: Syndrome named after me: Webb-Dattani syndrome

2014: One of Best Reviewer of the Year awards for my services to *Journal of Clinical Endocrinology and Metabolism*

2018-2023: Gold Clinical Excellence Award – awarded by ACCEA

2021: Award of Scientific and Clinical Excellence in Paediatric Endocrinology by the Greek Paediatric and Adolescent Endocrine Society – Kalaitzoglou Prize

2022: Visionary Award 2022 Human Growth Foundation USA

2022: ESPE Research Award for substantial achievement in the field of paediatric endocrinology

2023: Awarded Fellowship of the Medical Academy of Sciences (FMedSci)

2023: Awarded Honorary Membership of the Italian Society for Paediatric Endocrinology and Diabetes (SIEDP)

Grants

2024-2026	ESPE Research Fellowship awarded to Dr. Dogus Vurrali: "Congenital hypopituitarism: Genetic and Clinical Characterization of a Large Nationwide Cohort" €140000; **Sponsor Mehul Dattani**
2024-2025	Donation of £100000 by the St. Piers family for the project "Optimisation of biochemical control in children and young people with congenital adrenal hyperplasia using novel biochemical assays" **Mehul Dattani (PI)**
2023-2026	GOSH Children's Charity/SPARKS Understanding the role of cellular senescence in Rasopathies £249,917.99 **Mehul Dattani (CoI)**
2023-2024:	GOSH BRC Minimally invasive cortisol and 17OHP monitoring in Congenital Adrenal Hyperplasia £14,500 **Mehul Dattani (PI)**
2022-2024:	Medical Research Foundation Research into mutations causing pituitary endocrine disorders directly or indirectly affecting growth and development £55,000 **Mehul Dattani (PI)**
2022-2029:	NIHR Discontinuation of GH treatment in pubertal children £1,852,244.98 **Mehul Dattani (PI)**
2021-2023:	ROHHAD Association ROHHAD syndrome: Towards a Better Understanding £131,844.55 **Mehul Dattani (PI)**
2020-2025:	Great Ormond Street Hospital Children's Charity: Molecular Basis of Rare Endocrine Developmental Disorders £500,000 – later used towards funding Dr. Naresh Hanchate **Mehul Dattani (PI)**
2021-2024	MRC Clinical Academic Research Partnership Scheme Award to Dr. Harshini Katugampola: 3 year funding £220,000 Elucidating and modelling complex

postnatal steroid hormone changes in preterm infants. **Supervisor/ICH Partner Mehul Dattani**

2021-2026 International Fund-raising Congenital Adrenal Hyperplasia (IFCAH)
Establishing novel improved biomarker profiles in congenital adrenal hyperplasia. Euros 110,000 Awarded to Nils Krone, **Mehul Dattani (CoI)**

2018-2021 Children with Cancer
Post-doctoral Fellowship Markers of Hypothalamic Dysfunction in children with hypothalamo-pituitary axis tumours or pituitary maldevelopment £250,000 Manuela Cerbone, **Mehul Dattani (Sponsor)**

2018-2021 European Commission
European Registeries for Rare Endocrine Conditions (EuRREcA)
Co-applicant – GOSH portion of Euros 12,689.83 (Total Euros 159,507)

2017-2020 GOSH Children's Charity
Research Leader Award: Investigation of Disorders of forebrain, eye and pituitary development £94,275 **Mehul Dattani**

2017-2019 Medical Research Foundation
Research into mutations causing pituitary endocrine disorders directly or indirectly affecting growth and development £55,000 **Mehul Dattani (PI)**

2016-2017 GOSH Children's Charity
Great Ormond Street Hospital Research Capacity Building £84,827 **Mehul Dattani (PI)**

2015-2017 Medical Research Foundation
Research into mutations causing pituitary endocrine disorders directly or indirectly affecting growth and development £100,000 **Mehul Dattani (PI)**

2015-2018 Children with Cancer UK
Characterization and assessment of potential biomarkers and novel drug targets for childhood craniopharyngioma £458,728
Juan Pedro Martinez-Barbera (PI), **Mehul Dattani (Co-I)**

2013-2015 GOSH CC Midline Brain Tumour Interest Group £29,730 **Mehul Dattani (PI)**

2014-2017 Action Medical Research
Investigating the protective effect of Non-steroidal Anti-inflammatory drugs (NSAIDs) in craniopharyngioma tumours. £190,000
Carles Gaston-Massuet (PI), Tim Warner (Co-I), **Mehul Dattani (Co-I)**

2013-2014 GOSH Children's Charity Clinical Research Starter Grant
Quantification of oxytocin and arginine-vasopressin in patients with complex hypothalamo-pituitary disorders including septo-optic dysplasia £49,734
Professor Mehul Dattani (PI), Dr. JP Martinez-Barbera, Dr. Helen Aikenhead

2013 -2015 British Society for Paediatric Endocrinology & Diabetes Research Award to Dr. Hoong Wei Gan
Quantification of oxytocin and arginine-vasopressin concentration in patients with complex hypothalamo-pituitary disorders including septo-optic dysplasia and central nervous system tumours

£15,000
Professor Mehul Dattani (Co-investigator)

2013-2015 Great Ormond Street Hospital for Children Biomedical Research Centre Grant
Quantification of oxytocin and arginine-vasopressin concentration in patients with complex hypothalamo-pituitary disorders including septo-optic dysplasia and central nervous system tumours
£10,000
Professor Mehul Dattani (PI)

2013-2016 BUPA Cromwell Hospital Grant
Quantification of oxytocin and arginine-vasopressin concentration in patients with complex hypothalamo-pituitary disorders including septo-optic dysplasia and central nervous system tumours
£177 597
Professor Mehul Dattani (Sponsor)

2012-2015 Children with Cancer UK Charity/GOSH Children's Charity
£79,152
Dr. JP Martinez-Barbera (PI), **Professor Mehul Dattani (Co-investigator)**

2012-2015 GOSH Children's Charity
Research Leader Award: Investigation of Disorders of forebrain, eye and pituitary development
£433,771
Professor Mehul Dattani

2011-2012: GOSH Children's Charity
Research Leader Award: Investigation of Disorders of forebrain, eye and pituitary development
£130,702
Professor Mehul Dattani

2011-2013 Novo Nordisk
Nordinet International Outcomes Study
£73,092
Professor Mehul Dattani

2011-2013 Pfizer
KIGS International Outcomes study
£73,092
Professor Mehul Dattani

2011-2014 MRC Australia
Functional analysis of a novel genetic mouse model for congenital growth hormone deficiency
Australian $501299.00 - £259,112
Assoc Professor Paul Thomas (PI), **Professor Mehul Dattani** (Co-I)

2010-2011: GOSH Children's Charity
Investigation of Disorders of forebrain, eye and pituitary development
£129,314
Professor Mehul Dattani

2010-2011 Biomedical Research Centre GOSH/ICH
Genotype-Phenotype correlations in Craniopharyngioma
£3000

Dr. Esieza Ikazoboh, **Professor MT Dattani**

2009-2012 BSPED Merck-Serono award
Investigation of the role of Kallmann genes in the aetiology of hypopituitarism
£25,000
Dr. Vaitsa Tziaferi, **Professor Mehul Dattani (PI)**

2009-2011 Birth Defects Foundation
Investigation of the genetic basis of congenital hypopituitarism in a cohort of patients with complex developmental disorders
£120,000
Professor MT Dattani (PI), Dr. Maria Bittner-Glindzicz

2009-2012 Wellcome Trust
The role of Sox2/SOX2 in hypothalamo-pituitary development
£359,039
Professor MT Dattani (PI), Dr. Juan-Pedro Martinez-Barbera

2008-2009 Biomedical Research Centre GOSH/ICH
Establishing a molecular diagnosis of hypogonadotrophic hypogonadism
£3000
Dr. Vaitsa Tziaferi, **Professor MT Dattani**

2008-2009 ESPE Research Unit Small Grant 2008-2009
Endocrine and Genetic Characteristics of Uganda Pygmies
€7,500 (£6,140)
Professor Zeev Hochberg, Dr. Edison Mworozi, Dr. Cecilia Camacho-Hubner, **Professor MT Dattani**

2008-2010 Child Growth Foundation
An investigation into the role of growth hormone on higher functioning in children
£53,148
Professor MT Dattani (PI), Professor PC Hindmarsh, Dr. A Salt, Dr. N Dale

2007-2010 Child Growth Foundation
An investigation into the role of growth hormone on higher functioning in children
£305,000
Professor MT Dattani (PI), Professor PC Hindmarsh, Dr. A Salt, Dr. N Dale

2006-2008 European Society for Paediatric Endocrinology
Investigation of the role of the transcription factors *SOX2* and *SOX3* in disorders of eye, forebrain and pituitary development in humans
600,000 DK
Dr. S Alatzoglou, **Professor MT Dattani (Sponsor of study)**

2006-2009 Wellcome Trust
Characterisation and functional analysis of Hesx1-interacting proteins in mouse and human
£235,911
Dr. JP Martinez-Barbera (Principal Investigator, [PI]), **Professor MT Dattani (Co-investigator)**

2005-2009 Child Health Research Appeal Trust award for a PhD studentship
Microarray Comparative Genomic Hybridization in the detection of

submicroscopic chromosomal aberrations in patients with complex disorders of pituitary development
£40,000
Dr. Maria Bitner-Glindzicz (PI), **Professor MT Dattani (Co-investigator)**

2004-2007 Medical Research Council
The role of SOX3 in disorders of forebrain and pituitary development in humans
£260,724
Professor MT Dattani (PI), Dr. M Bitner-Glindzicz

2004-2007 Child Health Research Appeal Trust award for a PhD studentship
Role of *Hesx1* in forebrain and pituitary development in mouse and man
£40,000
Dr. JP Martinez-Barbera (PI), **Professor MT Dattani (Co-investigator)**

1999-2004 Medical Research Council
Genetic Evaluation of Homeobox Proteins in Anterior Pituitary Development (Career Establishment Grant)
£598,796
Professor MT Dattani (PI)

2002-2006 Eli Lilly
Optimisation of Growth Hormone treatment in short children born small for gestational age based on a growth prediction model
£12,000
Professor MT Dattani (PI)

2002-2003 Medical Research Council
Genotype-Phenotype Correlation in children with congenital hypopituitarism
£42,556
Professor MT Dattani (PI)

2003-2005 Child Growth Foundation
Genotype-Phenotype Correlation in children with congenital hypopituitarism
£50,000
Professor MT Dattani (PI)

2002-2003 Child Health Research Appeal Trust (Pump-priming award)
The genetic basis of disorders of sexual differentiation
£8,788
Dr. JC Achermann (PI), Dr. CE Brain, Dr. PC Hindmarsh, **Professor MT Dattani (Co-investigator)**

1998-2001 Child Health Research Appeal Trust award for a PhD studentship:
The role of homeobox genes *Hesx1*, *Pit1*, *Prop1* and *Lhx3* in human pituitary development.
£38,250
Professor MT Dattani (PI), Professor MA Preece

1999-2000 Child Health Research Appeal Trust (Pump-priming award)
The role of the homeobox genes *Pit1*, *Prop1* and *Lhx3* in human pituitary gland development
£16,000
Professor MT Dattani (PI)

1999 European Society for Paediatric Endocrinology

Analysis of the role of the *PIT1*, *PROP1* and *LHX3* genes in Combined Pituitary Hormone Deficiency
$10,000
Professor MT Dattani (PI)

1992 European Society for Paediatric Endocrinology
Establishment of an Eluted Stain Assay for the measurement of GH in human serum
$10,000
Professor MT Dattani (PI)

1995-2005 Travel grants from Endocrine Society, BSPED, ESPE, Society for Endocrinology, UCL to Group
£6000

Invited lectures

1) Speaker on: "Congenital Disorders of Hypothalamo-Pituitary Development" at the Paediatric Research Symposium, University of Leipzig, Germany, March 2024.
2) Speaker on: "The genetic complexity of Growth Hormone Deficiency and related disorders – the plot thickens" at the International Conference of Bone, Growth and Growth Plate, Manchester, February 2024.
3) Speaker on: SOD: Lessons learnt from a Single Centre Cohort" and "SOD: Future Directions" at a Webinar on Septo-Optic Dysplasia for ENDO-ERN, January 2024.
4) Plenary speaker on "*Congenital Hypopituitarism: Etiology, Diagnosis and Treatment*" at the 15th Annual Meeting of the Brazilian Congress of Paediatric Endocrinology and Metabolism, Belo Horizonte, Brazil, December 2023
5) Speaker on: "*Long-Acting Growth Hormone*" at Pfizer India Webinar, October 2023
6) Speaker on: *"Molecular defects in isolated GHD and multiple pituitary hormone deficiencies*" at GroAcademy meeting, Dubai, UAE, October 2023
7) Speaker on: *"GHD Treatment: Landscape of future therapies"*at GroAcademy meeting, Dubai, UAE, October 2023
8) Plenary Speaker on: "*Congenital Hypopituitarism*" at the Swedish Society for Paediatric Endocrinology and Diabetes, Stockholm, Sweden, November 2023
9) Plenary Speaker on: "*Congenital Hypopituitarism: Discoveries and Challenges*" at SIEDP, Italian Society for Paediatric Endocrinology and Diabetes, Bologna, Italy, November 2023
10) Speaker on: "*Long-acting Growth Hormone*" at SIEDP, Italian Society for Paediatric Endocrinology and Diabetes, Bologna, Italy, November 2023
11) Speaker on "*Management of SGA*" in GH Deficieny webinar, October 2023
12) Speaker on "*GHD: Challenges and Unmet Needs in Children*" and Chair at "Growth Hormone Deficiency Roundtable" webinar June 2023
13) Speaker on "*Research Advances in the Growth Mechanism and Treatment of Noonan Syndrome*"- Chinese Society of Paediatric Endocrinology and Metabolism Webinar, May 2023.
14) Plenary Speaker on "*Molecular Basis of Congenital Hypopituitarism*"at the 50th Annual Meeting of the Hellenic Endocrine Society, Thessaloniki, Greece, May 2023
15) Speaker on "*Clinical Approach to Congenital Hypopituitarism*" Nordic Webinar Endocrinology, April 2023
16) Speaker on "*Rare forms of Congenital Adrenal Hyperplasia*" at 2022 International Forum on Standardized Diagnosis and Treatment of Children's Growth and Development, December 2022, China (Virtual Meeting).

17) Speaker on "*Congenital Hypothalamic disorders*" at ESPE Science Symposium, Netherlands, 2022

18) Speaker on "*Recent advances in understanding Congenital Hypopituitarism*" 14th Pituitary and Hypothalamus Meeting, Montpelier, France, September 2022

19) Speaker on "*Molecular Basis of Congenital Hypopituitarism*"at the Annual Meeting of the European Society for Paediatric Endocrinology, Rome, Italy September 2022

20) Speaker on "*Molecular Basis of Congenital Hypopituitarism*" at the Annual Meeting of the Endocrine Society USA, Atlanta, USA, June 2022

21) Speaker on "*GHD Treatment Challenges and Future solutions at Pfizer UAE Virtual Meeting*", May 2022

22) Speaker (virtual) on "*Genetics of Hypothalamo pituitary disorders - Bedside implications*" at the Pearls in Paediatric Endocrinology CME meeting at the Bai Jerbai Wadia Hospital for Children, Mumbai, May 2022

23) Speaker on "*The management of growth in Noonan syndrome*" at the 25th NovoNordisk Endocrine Nurse Workshop, Stratford, UK April 2022

24) ENDO TODAY Live Symposium, Hot Topics in Growth and Pituitary Disease: Speaker on "*Update on Congenital Hypopituitarism*" December 02 2021

25) Speaker on "*The management of growth in Noonan syndrome*" Noonan Syndrome Association, December 06 2021

26) Annual Scientific Meeting of the Irish Endocrine Society 2021: Speaker on: "*Congenital Hypopituitarism: Novel Insights into management*"

27) Asia-Pacific Paediatric Endocrine Society (APPES) Fellows School 2021: Speaker on "*Management of Hypopituitarism*" November 06 2021

28) Growth Hormone Research Society, India, 5th Annual Meeting: Speaker on "*Value of MRI in the management of GHD and associated conditions*". October 10 2021

29) Greek Paediatric and Adolescent Endocrine Society Annual Meeting: Speaker on "*Genetics of Hypopituitarism*" September 11 2021 Award of Scientific and Clinical Excellence in Paediatric Endocrinology – Kalaitzoglou Prize

30) Paediatric Endocrine Education Dinner Speech, Queensland Ghildren's Hospital, Australia: Speaker on "*Septo-Optic Dysplasia*" July 14 2021

31) ESE Spotlight on Science: Speaker on "*Masterclass in Congenital Hypopituitarism*" July 12 2021

32) 2021 Human Growth Foundation (HGF) Endocrine Education Growth Lecture Series Speaker on "*Congenital Hypopituitarism: Recent Advances in Management*" June 14 2021

33) University of Michigan Department of Human Genetics Seminar Series: Speaker on "*The Molecular Basis of Congenital Hypopituitarism: Novel Genes, Novel Phenotypes*" May 11 2021

34) RSM/Society for Endocrinology *Clinical Update in Genetics and Endocrinology meeting*: Speaker on "*Genetics of Pituitary Disorders*" May 06 2021

35) American Pediatric Endocrine Society (PES) International Plenary Speaker on "*Genetics of Pituitary Development"*, May 01 2021

36) Meet the Professor speaker on "*Management of Pituitary Hormone Replacement Through Transition From Adolescence to Young Adulthood*" at the Annual Endocrine Society meeting USA, 2021 – March 23 2021

37) Speaker on "*Diagnosis and management of GH deficiency*" at the 15th SEHA International Paediatric conference (GOSH Lecturer), March 19 2021

38) Speaker on "*Genotype-Phenotype Correlations in Congenital Hypopituitarism*" at the Postgraduate school of Endocrinology and Metabolic Disease, University of Milan, February 08 2021

39) Speaker on "*Genetics of Hypopituitarism*" at the Inaugural Virtual Academic meeting of the Indian Society of Paediatric and Adolescent Endocrinology, January 30 2021

40) Speaker on "*Developmental disorders of the Pituitary*" at University of Glasgow Paediatric Endocrinology Research Group Online meeting, November 19th 2020

41) Speaker in the "Advances in Growth Disorders" symposium at the Spanish Paediatric Endocrinology Society Annual meeting on "*New Aspects of Congenital Hypopituitarism*", Online Meeting, October 15 2020

42) Speaker on "*Diagnosis and management of GHD – an update for 2020 at "Growth and Thyroid*" webinar for the Egyptian Paediatric Endocrinology Society, September 24 2020

43) Speaker on "*Genetics of Growth*" in the **"**Linear growth: past, present and future" sympsium at the 7th international meeting on Nutrition and Growth, London, UK

44) Webinar on "*Congenital Hypopituitarism – an update on Approaches to Diagnosis and Treatment*?" Turkish Paediatric Endocrinology meeting June 26 2020

45) Speaker on "*Molecular Genetics of Congenital Hypopituitarism*" at the Online ViSPET Teaching course at Sheffield University, June 25th 2020

46) Speaker at the Finnish Society for Pediatric Endocrinology Spring Meeting 2020, March 12 2020 (meeting in Lapland but my contribution was online due to COVID-19). 2 lectures:
a) Development of the pituitary and hypothalamus
b) Management of Hypopituitarism

47) Speaker on: *Noonan Syndrome – an Endocrine perspective into a genetic disorder* in a symposium entitled "Noonan Syndrome: A multi-disciplinary approach" at the 25th Novo Nordisk® Endocrine Workshop, Thursday 27 - Friday 28 February 2020, Stratford upon Avon

48) Speaker at P-E Connect meeting on "*Molecular Basis of Congenital Hypopituitarism*", Rome, January 2020

49) Meet the Expert session on "*Management of Hypopituitarism*" at the Annual EDGE Meeting, Rome, December 2019

50) Inquisitor Session, Annual meeting of the British Society for Paediatric Endocrinology, Cardiff, November 2019. I was "blind" questioned on a range of endocrine scenarios by Dr. Tim Cheetham, Consultant Paediatric Endocrinologist at Newcastle.

51) Speaker on "*Molecular Basis of Hypopituitarism – unravelling of a complex condition*' at the Annual Latin America Society pf Paediatric Endocrinology meeting, Florianopolis, Brazil, November 2019

52) Speaker on "*Molecular Basis of Congenital Hypopituitarism*" at the FASEB The Growth Hormone (GH)/ Prolactin (PRL) Family in Biology and Disease Conference, Fort Lauderdale, Florida, USA July 2019

53) Speaker at "Growth Disorders: Theory to Practice" meeting, New Delhi, India, April 2019 – 2 lectures:
a) Optimization of GH therapy in GHD
b) Genetics of GHD

54) Speaker on "*Congenital Hypopituitarism – New Perspectives*" at 6th ENDOPED Conference, Timisoara, Romania, April 2019

55) Speaker at NovoNordisk Endocrinology Update Meeting on "Use of MRI in the management of GHD" February 2019

56) Speaker on "*Genetics of Pituitary Hormone Deficiencies*" at the European Neuroendocrine Association, Wroclaw, Poland, October 2018

57) Speaker on "*What is Normal Growth – understanding normal growth*" and "*Perspectives on managing growth disorders*"at the IPSEN International Children's Growth Awareness day, Paris, France, September 2018

58) Speaker on "*Genetics of Hypothalamo-pituitary disease*" at the Paediatric Research Symposium, Leipzig, Germany, July 2018

59) Speaker on "*Challenges in the diagnosis of growth hormone deficiency*" at the International EDGE meeting, Rio de Janeiro, Brazil, June 2018

60) Speaker on "*Challenges in the diagnosis of growth hormone deficiency*" at the Hong Kong Society for Paediatric Endocrinology and Metabolism, Hong Kong, June 2018

61) Speaker on "*Molecular Basis of Congenital Hypopituitarism*" at the Forum on Growth Hormone Research 2018, Kobe, Japan, June 2018
62) Speaker on "*Hypogonadotrophic Hypogonadism*" at the International Workshop on Sex Development and Maturation, Lubeck, Germany, February 2018
63) Speaker and Facilitator on the Henning Andersen course, Amsterdam, Netherlands, December 2017
64) Facilitator of workshops at the Growth Hormone Research Society Consensus Meeting on GH Biomarkers, Aarhus, Denmark, November 2017
65) Speaker on "*Congenital Hypopituitarism*" at the Ferring GOSH Endocrinology Update, November 2017
66) Speaker on "*Congenital Hypopituitarism*" at the 11th Pituitary and Hypothalamus workshop meeting, Jerez, Spain, September 2017
67) Lecturer and Faculty at the Speaker on "*Disorders of Puberty*" at the British Paediatric Endocrinology Training School, Stratford, UK, July 2017
68) Plenary speaker on "*Management of GHD*" at the Romanian Society for Endocrinology, Cluj-Napoca, Romania, June 2017
69) Speaker on "Management of Hypopituitarism" at the West Midlands Regional Training Afternoon, June 2017
70) Speaker on "*Congenital Hypopituitarism*" in INKEP meeting, Essen, Germany, June 2017
71) Coordinator of Severe Insulin Resistance Expert Group, London, January 2017
72) Speaker on "*Molecular Basis of Pituitary Developme*nt" at the Memorial Symposium for Primus Mullis, Berne, Switzerland, January 2017
73) Speaker on "*Congenital Hypopituitarism*" at the Seminaire D'Endocrinologie Pediatrique et Developpement, Paris, France, January 2017
74) Visiting Professor in Hong Kong, December 2016
75) Speaker on "*Diagnosis and Management of Hypopituitarism*" and "*Disorders of Sex Development*" at the Chinese Society for Paediatric Endocrinology and Metabolism (CSPEM) CME Forum, Wuhan, China, December 2016
76) Speaker on "*Genetics of Hypopituitarism*" at INKEP meeting, Paris, France, December 2016
77) Keynote lecture speaker at the 2016 Henning Andersen course, Zurich December 2016
78) Speaker at the 2nd Middle Eastern Endocrine Forum, Dubai, UAE, November 2016
79) Plenary Speaker at 2016 Congress of the Latin-American Society for Pediatric Endocrinology: *Congenital Hypopituitarism* and *Management of Craniopharyngioma*, Buenos Aires, November 2016
80) Speaker on "*Effect of GH deficiency on brain structure and cognition*" at the Medical Dialogue on Growth and Metabolism meeting, Amsterdam, Netherlands, November 2016
81) Plenary speaker on "*Diagnosis of GH deficiency and related disorders*"and MTE on "*Optimization of GH therapy*" at the Endocrine Society of India ESICON 2016, NewDelhi, October 2016
82) Speaker on "*Congenital Hypopituitarism*" at the 10th Kent, Surrey and Sussex Paediatric and Neonatal Research Day, Brighton, October 2016
83) Speaker on "*Genetics of Hypothalamic Hamartoma*" at Symposium on Hypothalamic Hamartoma, London, September 2016
84) Speaker on "*Congenital Hypopituitarism*" at the INKEP meeting, London June 2016
85) Speaker on "*Congenital Hypopituitarism*" in"Hot Topics in Paediatric Endocrinology", Stockholm, Sweden, May 2016
86) Speaker on "*Genetics of Pituitary Disorders*" at the Italian Society for Paediatric Endocrinology and Diabetology Summer School, May 2016
87) Speaker on *Congenital Hypopituitarism* at Scottish Paediatric Endocrine Group meeting, Stirling, Scotland, May 2016
88) Facilitator and Speaker at GH workshop, Pfizer Best Practices and Controversies

in GH meeting, Berlin, May 2016

89) Speaker on "*Congenital Hypopituitarism*" at the annual meeting of the Endocrine Society, Boston, USA, April 2016

90) Speaker on "*Genetics of hypothalamo-pituitary-GH axis*" at **ENDOCONNECT** meeting, Bled, Slovenia, 2016

91) Speaker on "*Why should paediatric and adult endocrinologists talk to each other?*" at the **Society for Endocrinology Clinical Update,** Birmingham 2016

92) Speaker on "*Genetics of Pituitary Disorders*" at **COST meeting for GnRH Network**, Budapest, Hungary, April 2016

93) Speaker on "*Congenital Hypopituitarism*" at the Rare Diseases Day 2016, Brandenburg Academy of Sciences (BBAW), Berlin, 2016

94) Speaker on "*Management of Congenital Hypopituitarism*" at the National Clinicopathological Pituitary Conference, February 2016

95) Speaker on "*Congenital Hypopituitarism – clinical management*" at the **21st Annual Novo Nordisk** meeting in Stratford, 2015

96) Speaker on "*Novel Insights into Congenital Hypopituitarism*" at the **43rd Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes**, Sheffield, UK, 2015

97) Speaker on "*Genetic Regulation of growth*" at the **NordiUp meeting**, Tehran, Iran, 2015

98) Speaker on "*Genetics of Growth Hormone Deficiency"* at the **GH in Paediatric Practice meeting**, Cairo, Egypt, 2015

99) Speaker on "*GHD: Are we treating the right patients?*" at the **Second International GH Academy meeting**, Munich, Germany, 2015

100) Speaker on
   a. "*Congenital Hypopituitarism: New genes, new phenotypes*"
   b. "*An Update on Disorders of Sex Development*"
   at the **2nd Sandoz EDGE meeting**, Berlin, Germany, 2015

101) Speaker on "*Role of genetic factors in the aetiology of Hypopituitarism*" at the **GH and Growth Factors – Metabolic Disorders Course for Post-graduates**, Gothenberg, Sweden, 2015

102) Speaker on "*Hypopituitarism: diagnosis to optimal management*" at **Summit for Growth meeting**, Istanbul, Turkey, April 2015

103) Speaker on "*Genetics of Pituitary Disorders*" at **COST meeting for GnRH Network**, Prato, Italy, April 2015

104) Speaker at Endocrinology of the Newborn and the Infant, **Ipokrates Foundation Clinical Seminar**, Athens, Greece, 2015:
   a) "*Congenital Hypopituitarism*"
   b) "*Managing rare congenital growth disorders*"

105) Speaker on "*Novel Insights into Congenital Hypopituitarism*" at the **Paediatric Endocrinology 2015: An Update for Paediatricians and Endocrinologists**, Prague, Czech Republic, 2015

106) Speaker on "*Management of Congenital Hypopituitarism*" at the **1st Sandoz EDGE meeting**, Vienna, Austria, 2014

107) Speaker on: "*Molecular basis of hypothalamo-pituitary developmental disorders*" at the **1st Ferring Update in Paediatric Endocrinology** meeting, London, 2014

108) Speaker on "*Genetics and Phenotypes in Congenital Hypothalamo-Pituitary Disorders*" at the **Neuroendocrine Association Annual Meeting**, Sofia, Bulgaria, 2014

109) Speaker on "*Pituitary Development*" at the **Manchester Endocrine Group**, Manchester, UK, 2014

110) Speaker on "*Pituitary Development*" at the **Brecon Paediatric Endocrine and Diabetes Group Annual Meeting**, Llandrindod, Wales, 2014

111) Speaker on "*Congenital Hypothalamo-pituitary disorders*" at **Edinburgh Paediatric Seminars** meeting, Edinburgh, UK, 2014

112) Speaker at **Novo Paediatric Endocrinology Growth Hormone Meeting**, Istanbul 2014:

a. "*Disorders of Hypothalamo-Pituitary development*"
b. "*Growth Hormone Deficiency: Clinical and Laboratory diagn*osis"
c. "*Congenital Hypopituitarism: Clinical Management*"

113) Speaker on "*GHD: Better Outcomes through Treatment Optimization: What is best practice?*" at the **First International GH Academy meeting**, Zurich, Switzerland, 2014

114) Speaker on "*Clinical phenotypes in Septo-optic Dysplasia*" at the **Annual Pituitary Workshop**, Edinburgh, UK, 2014

115) Speaker on "*Investigating growth failure*" at the **3rd Practical Aspects in Paediatric Endocrinology**, London, UK, 2014.

116) Speaker on "*Pituitary Development*" at the **Annual INKEP meeting**, Genova, Italy 2014

117) Speaker on "*Role of transcription factors in the aetiology of Hypopituitarism*" at the **GH and Growth Factors – Metabolic Disorders Course for Post-graduates**, Gothenberg, Sweden, 2014

118) Speaker at the **Israeli Paediatric Endocrine Society Annual Meeting**, Israel, 2014
a. *"Update on Diagnosis of GHD"*
b. *"Recent Advances in our understanding of the molecular basis of Hypothalamo-pituitary disorders"*

119) Speaker on "*Hypogonadism and its managemen*t" at the **First conference on Endocrinology in Paediatric Practice**, Santiago, Chile, 2013

120) Speaker on "*Management of growth failure*" at the **First conference on Endocrinology in Paediatric Practice**, Santiago, Chile, 2013

121) Speaker on *"Growth Disorders: a research update*" - **Fiona Hurel lecture at the Child Growth Foundation, UK** 2013.

122) Speaker on "*Novel genes involved in hypopituitarism*" at the **Joint Meeting of the European Society of Paediatric Endocrinology and the Paediatric Endocrine Society**, Milan, Italy, 2013.

123) Speaker on: "*Pituitary*" at the Yearbook symposium **51st Annual Meeting of the European Society for Paediatric Endocrinology,** Leipzig, Germany, 2013.

124) Speaker on "*Interpreting Dynamic Function Tests (Growth and Puberty)*" at the **3rd Practical Aspects in Paediatric Endocrinology**, London, UK, 2013.

125) Speaker on **"***Congenital Hypopituitarism – an Update***"** at the 95th **Annual Meeting of the Endocrine Society**, San Francisco, USA, 2013.

126) Speaker on **"***Congenital Hypopituitarism: novel insights into the aetiology***"**, at the **NordiScience symposium**, San Francisco, USA, 2013.

127) Speaker on **"***Wnt/Beta-Catenin signaling in Craniopharyngioma***"** at the **13th International Pituitary Congress",** San Francisco, USA, 2013.

128) Speaker on "*Pituitary Development*" at the **Annual INKEP meeting**, Rome, Italy 2013

129) Speaker on "*Role of transcription factors in the aetiology of Hypopituitarism*" at the **GH and Growth Factors – Metabolic Disorders Course for Post-graduates**, Gothenberg, Sweden, 2013.

130) Speaker on **"***Congenital Hypopituitarism – an Update***"** at the **IGSF1 MiniSymposium**, Leiden, Netherlands, 2013.

131) Speaker on "*Growth Hormone and its use in Growth Disorders*" at the **Health Agenda Symposium on Growth Hormone**, London 2012

132) Speaker at **1st International Saudi Society of Endocrinology and Metabolism Conference**, Riyadh, Saudi Arabia, 2012:
a. *Congenital Hypopituitarism* – Plenary Lecture
b. *Novel Insights into Management of Growth Hormone Deficiency* – Workshop
c. *Case Studies in Multiple Pituitary Hormone Deficiencies* – Workshop

133) Speaker on: "*Genetic causes of pituitary hormone deficiency*" at the **Masterclass in Paediatric Endocrinology – new genetic insights in growth disorders**, Utrecht, Netherlands, 2012.

134) Speaker on: "*Pituitary Development and Function*" at the CME day of the **40th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes**, 2012.

135) Speaker on: "*Congenital Disorders of Hypothalamo-Pituitary Develo*pment – *an Update*" at the **6th Annual Workshop on Hypothalamo-pituitary Disorders**, Lauret, France, 2012.

136) Speaker at the University of Parma on *Congenital Hypopituitarism*, Parma, Italy, 2012.

137) Speaker on: "*Pituitary*" at the Yearbook symposium **51st Annual Meeting of the European Society for Paediatric Endocrinology,** Leipzig, Germany, 2012.

138) Speaker on **"***Genetics of Hypopituitarism***"** at the **"GH- IGFs system interplay in the human being: Novel insights from clinical practice**" symposium to celebrate 50 years of the University of Navarre, Pamplona, Spain, 2012

139) Speaker on "*Recent Advances in Growth*" at the joint RSM-RCPCH "**Recent Advances in Paediatrics**" course, Royal Society of Medicine, London, 2012

140) Speaker on "*Genetic Aspects of Pituitary Disease*" at the "**Endocrinology in Clinical Genetics Summer School**" for SpRs, London, 2012

141) Speaker on "*Growth Disorders*" at the **PUNCH** Charity meeting, London, July 2012

142) Speaker on "*Genetic causes of Hypopituitarism*" at **Annual Retreat for Paediatric Endocrine Trainees,** Warwickshire, 2012

143) **Speaker on "***Strategies to detect a new gene and prove its importance*" at the **ESPE Science School**, Heraklion, Crete, 2012

144) Speaker on "*Pituitary gland development: update*" at the **6th Advanced ESPE Seminar in Developmental Endocrinology**, Bern, Switzerland, 2012.

145) Speaker on "*Role of transcription factors in the aetiology of Hypopituitarism*" at the **GH and Growth Factors – Metabolic Disorders Course for Post-graduates**, Gothenberg, Sweden, 2012.

146) **Niall O'Meara Lecture**, Study Day of the Irish Endocrine Society 2012: "*Congenital Hypopituitarism – the Plot Thickens*", Royal College of Physicians, Dublin, Ireland.

147) Lecture on "*Congenital Hypopituitarism – the Plot Thickens*" **OCDEM Lecture Series**, Oxford University, October 2011.

148) Lecture on "*Endocrinology Research at GOSH*" – **GOSH Open Day** 2011.

149) Lecture on "Growth Disorders" at the **Reflectorium**, Holten, Netherlands, 2011.

150) Speaker on: "*Pituitary*" at the Yearbook symposium **50th Annual Meeting of the European Society for Paediatric Endocrinology,** Glasgow, Scotland, 2011.

151) Speaker on "*Growth and the Hypothalamo-pituitary axis*" at the **Overseas Meeting of the Royal Society for Medicine Paediatrics and Child Health Section, Athens, Greece, 2011**

152) Speaker on "*Pituitary Development*" at the **Annual INKEP meeting**, Paris, France, 2011

153) Lecture on "*Growth Disorders in Child*hood" at the **Paediatric Endocrine Study Day**, Lister Hospital, Hertfordshire, September 2011.

154) Speaker on "*Growth Disord*ers" at the **PUNCH** Charity meeting, London, July 2011

155) Speaker on "*Molecular Basis of Craniopharyngioma*" at the **Annual SIOP meeting**, Liverpool, 2011

156) Speaker on "*Role of transcription factors in the aetiology of Hypopituitarism*" at the **GH and Growth Factors – Metabolic Disorders Course for Post-graduates**, Gothenberg, Sweden, 2011

157) Speaker on "*Growth and the Hypothalamo-pituitary axis*" at the **Overseas Meeting of the Royal Society for Medicine Paediatrics and Child Health Section, Athens, Greece, 2011**

158) Speaker on "*Pituitary Development*" at the Annual INKEP meeting, Paris, France, 2011

159) Speaker on "*Role of transcription factors in the aetiology of Hypopituitarism*" at

the **GH and Growth Factors – Metabolic Disorders Course for Post-graduates**, Gothenberg, Sweden, 2010

160) Speaker on: "*Genetics of Hypopituitarism*" at Malmoinides Hospital, New York, USA, March 2011

161) Speaker on: "*Overview and Clinical Use of Diagnostic Tests in the Assessment of Short Stature*", 3rd Merck Serono Growth Hormone Symposium, Lisbon, Portugal, 2011

162) Speaker on "*Role of transcription factors in the aetiology of Hypopituitarism*" at the **GH and Growth Factors – Metabolic Disorders Course for Post-graduates**, Gothenberg, Sweden, 2010

163) Speaker on: "*Pituitary Development*" at the **European Society for Paediatric Endocrinology Summer School**, Prague, 2010

164) Speaker on: "*Growth and Growth Factors*" at the Yearbook symposium **49th Annual Meeting of the European Society for Paediatric Endocrinology**, Prague, Czechoslovakia, 2010

165) Speaker on "*Role of transcription factors in the aetiology of Hypopituitarism*" at the **GH and Growth Factors – Metabolic Disorders Course for Post-graduates**, Gothenberg, Sweden, 2010

166) Speaker on: "*The Genetic Basis of Hypopituitarism*" at the **UPDATE IN ENDOCRINOLOGIA PEDIATRICA Il ruolo della Biologia Molecolare, Bari, Italy, 2010**

167) Speaker on: "*Role of transcription factors in early pituitary development"* at the **International Congress for Endocrinology**, Kyoto, Japan, 2010

168) Speaker on: "*Role of transcription factors in early pituitary developmen*t" at the **International Symposium on Pediatric Endocrinology Official ICE 2010 Satellite Symposium**, Tokyo, Japan, 2010

169) Speaker on: "*Genetic defects leading to GH deficiency and related phenotypes*" at the **18th Novo Nordisk Symposium on GH and metabolism**, Berlin, Germany, 2010

170) Speaker on "*Developmental disorders of the pituitary and hypothalamus***" at the Neonatal Update 2009 – The Science of Newborn Care meeting, Imperial College, 2009**

171) Speaker on: "*Investigation of growth disorders in the peri-pubertal period*" at the 37th **Annual Meeting of the British Society for Paediatric Endocrinolog**y, Reading, UK, 2009

172) Speaker on: "*Physiology and Developmental Biology of the Pituitary gland*" at the **CME training day for the British Society for Paediatric Endocrinology**, Reading, UK, 2009

173) Speaker on "*Septo-optic dysplasia: from laboratory to clinic*" at the **Annual meeting of the National Cooperative Growth Study**, Phoenix, Arizona, 2009

174) Speaker on "*Genetics of Hypopituitarism*" in the **Department of Human Genetics, University of Michigan**, Ann Arbor, Michigan, USA, 2009

175) Speaker on "*Clinical management of Hypopituitarism*" in the **Department of Pediatric Endocrinology, University of Michigan**, Ann Arbor, Michigan, USA, 2009

176) Speaker on: "*Short and Tall stature*" at the **Paediatric SpR Endocrinology Training Day, Royal Society for Medicine**, 2009

177) Speaker on *"Septo-optic dysplasia"* at the **SERAP meeting**, ICH, 2009

178) Speaker in: "*Development of the anterior pituitary gland*" at the **Ipsen UK Research Forum**, Edinburgh, UK, 2009

179) Speaker on: "*Growth and Growth Factors*" at the **8th Joint Meeting of the European Society for Paediatric Endocrinology and the Lawson-Wilkins Society for Endocrinology**, New York, USA, 2009

180) Speaker on the "*Genetic Ontogeny of Pituitary Development*" at the **91st Annual Meeting of the Endocrine Society USA,** Washington, USA, 2009

181) Speaker on: "*Approaching short stature in Malta*" at the **Malta meeting of the Royal Society for Medicine (Paediatric section)**, Malta, 2009

182) Speaker on: "*Congenital hypopituitarism – a genetic disease*" at the **Human Genetics Division meeting**, University of Southampton, April 2009

183) Speaker on: "*Management of Growth Disorders -2009 Update*" at the **Arab Health Congress**, Dubai, UAE, 2009

184) Symposium International Congress of Endocrinology, Rio de Janeiro, Brazil, 2008

185) Speaker on: "*Growth and Growth Factors*" at the **47th Annual Meeting of the European Society for Paediatric Endocrinology**, Istanbul, Turkey, 2008

186) Speaker on: "*Congenital Hypopituitarism: the tip of the iceberg*" in the ***Stem cells, differentiation and cell networks in the pituitary*** workshop, Montreal, Canada, 2008

187) Speaker on: "*Management of Craniopharyngioma*" in a Meet the Professor session at the **90th Annual Meeting of the Endocrine Society USA,** San Francisco, USA, 2008

188) Visiting Professor at Massachusettes General Hospital and Harvard University, Boston, USA, 2008

189) Speaker on: "*Pituitary Development*" at the **10th European Congress of Endocrinology**, Berlin, Germany, 2008

190) Speaker on: "*Role of transcription factors in midline CNS and pituitary defects*" at the **Endocrine Involvement in Developmental Syndromes workshop**, Rome, Italy, 2008

191) Speaker on: "*Endocrine Genetics for the Neuroradiologist*" at the **European Course in Pediatric Neuroradiology**, London, UK, 2008

192) Speaker on: "*Candidate Gene Approach*" at the **10th KIGS/KIMS Expert meeting**, Florence, Italy, 2008

193) Speaker on: "*Developmental Endocrinology, Embryogenesis, Tumorigenesis*" at **the Developing Brain in Neuro-oncology meeting,** Cambridge, UK, 2008

194) Speaker on: "*Signalling processes of the embryonic and adult pituitary gland*" at the **Winter and Summer School SIEDP –First Edition**, Riccione, Italy, 2008

195) Speaker on: "*Genetics of Hypopituitarism*" at Malmoinides Hospital, New York, USA, February 2008

196) Speaker on "*Genetics of Hypopituitarism*" at the **Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes**, Cambridge, UK, 2007

197) Speaker on: "*Growth and Growth Factors*" at the **46th Annual Meeting of the European Society for Paediatric Endocrinology**, Helsinki, Finland, 2007

198) Speaker on: "*Hypopituitarism: Oddities – Congenital causes*" at the **39th International Symposium: GH and Growth Factors in Endocrinology and Metabolism**, Berlin, Germany, 2007

199) Speaker on: "*Novel Insights into Hypothalamic and Pituitary Development*" at the **Evolving Biology of Growth and Metabolism** meeting, Lisbon, Portugal, 2007

200) Speaker on: "*Hypopituitarism: from the laboratory to the clinic*" at the **3rd European Society for Endocrinology Postgraduate Course in Endocrinology**, Ohrid, Macedonia, 2007

201) Speaker on: "*Genetics of Septo-optic Dysplasia*" at the FIUMPPE meeting, Catania, Sicily, 2007

202) Speaker on: *"Pituitary Development and Hypopituitarism: From laboratory to clinic"* at the **European Neuroendocrinology Association**, Athens, Greece, 2006

203) Speaker on: *"Pituitary Development: From Mouse to Human"* at the **Australasian Paediatric Endocrine Group Annual Meeting (Plenary lecture),** Hobart, Tasmania, Australia, 2006

204) Speaker on: *"Management of Hypopituitarism: How I do it"* at the **APEG Clinical Fellows Meeting**, Hobart, Tasmania, Australia, 2006

205) Speaker on: *"Hypopituitarism: From genes to clinical findings"* at the **XIII meeting of the Societe Internationale Neuroendocrinology (SINE) (Plenary Lecture)** Porto Alegre, Brazil, 2006

206) Speaker on*: "Craniopharyngioma and Rathke's pouch cyst: molecular and aetiological findings"* at the **XIII meeting of the Societe Internationale Neuroendocrinology (SINE)** Porto Alegre, Brazil, 2006

207) Speaker on: *"The role of genetic testing in pituitary disease"* at the **EFES Regional Postgraduate course in Clinical Endocrinology**, Belgrade, Serbia, 2006

208) Speaker on: "*Genetics of Hypopituitarism*" at the **Endodays 2005 Meeting of the Finnish Endocrine Society**, Helsinki, Finland, 2005

209) Speaker on: **"***Genetic determinants of short stature***"** at the **Annual Meeting of the Italian Society for Paediatric Endocrinology and Diabetes (SIEDP) (Plenary session),** Sardinia, 2005

210) Speaker on: "*Developmental and Genetic Causes of Hypopituitarism*" at the **Annual Meeting of the Endocrine Society USA,** San Diego, USA, 2005

211) Speaker on: "*Indications and Expectations of Growth Hormone*" at the **Joint Overseas Meeting of the Royal Society for Medicine Paediatrics and Child Health Section and the Portuguese Association of Paediatrics,** Lisbon, May 2005

212) Speaker on: *"Defining Idiopathic Short Stature"* at **"Advances in the Understanding of Growth, Growth Hormone Therapy and Paediatric Endocrine Disorders",** Royal Society of Medicine, March 2005

213) Speaker on: a."*Novel insights into the management of disorders of sexual differentiation" b."Phenotype variation in congenital hypopituitarism"* at the **Fifth International Update in Paediatric Endocrinology**, Bangalore, India, August 2004

214) Speaker on *"Genetic Forms of Hypopituitarism"* in **"Abnormalities of Pituitary Development and Function: Mechanism and Management"**, Hazlewood Castle, Yorkshire, June 2004

215) Speaker on *"Phenotypic Variation in Genetic GHD"* in **"Disorders of Growth and Abnormalities of Growth Hormone Secretion: From Deficiency to Excess",** The Landmark Hotel, London, May 2004

216) Speaker on *"Regulation of Hormone Action: Novel insights from human disease models*" **in Satellite Workshop "Growth Hormone and Insulin-like Growth Factors: Growth and Beyond"** Geneva, Switzerland, May 2004

217) Speaker on *"The Pituitary"* in: **Molecular Biology for the Endocrinologist: New insights into etiology and prognosis (Plenary Session), 15th International Symposium on Growth Hormone and Endocrinology,** Malta, April 2004

218) Speaker on *"Recent Advances in Pituitary Development Using Murine and Human Models"* in: Symposium on **Neural Migration in Neuroendocrine systems**, **23rd Joint Meeting of the British Endocrine Societies with the European Federation of Endocrine Societies,** Brighton, March 2004

219) Speaker on "*Genetic and Endocrine advances in children with hypopituitarism*" in: **International Symposium on "Controversial Issues in Paediatric Endocrinology"***,* St. Bartholomew's Hospital, London, February 2004

220) Speaker on "*Evaluation of short stature in a 3rd world country*" in **"17th International Biennial Paediatric Conference"**, Lahore, Pakistan, February 2004

221) *"What hormones are needed for post-natal growth?"* In **"How Humans Grow"**: Joint Meeting of the Endocrinology and Diabetes and Paediatrics and Child Health Sections, Royal Society of Medicine, December 2003

222) *"Genetics of Pituitary Disease"* – **Society for Endocrinology Endocrine Nurses Training Course**, September 2003

223) *"Genetics of Growth hormone deficiency"* at the **1st Novo Nordisk meeting on *"Adult consequences of childhood endocrine disorders",*** Manchester, February 2003

224) *"Genetic causes of hypopituitarism"* in **5th Pituitary Clinicopathological Conference 2003: Plenary lecture**

225) *"Developmental Causes of Hypopituitarism"* at the 34th International Symposium

"GH and growth factors in Endocrinology and Metabolism", Budapest, Hungary, October 2002

226) "*Genetics of Hypopituitarism*" at the 5th International Congress of Neuroendocrinology: Symposium **"Genetics of Neuroendocrine Diseases",** Bristol, UK September 2002

227) *"Role of HESX1 in forebrain and pituitary development"* in the **First International Workshop Genetics of Growth Disorders,** Sitges, Spain, April 2002

228) Invited speaker on *"Genetics of Growth Disorders"* and *"Genetics of Disorders of Sexual Differentiation"* at **7th UAE Paediatric Conference,** Dubai, April 2002

229) Invited speaker on: *a) Genetics of Hypopituitarism b) Diagnosis of GH deficiency* at the **Genesis Investigators Meeting,** Dubai, May 2001

230) Invited speaker on *"Role of HESX1 in disorders of forebrain and pituitary development"* **at the NovoNordisk New Millenium Symposium on Growth**, Tokyo, Japan, September 2000

231) Invited speaker on *"Role of HESX1 in disorders of pituitary development"* at the **Lilly International Symposium on Growth and Sexual Development**, Fukuokwa, Japan, October 2000

232) Invited speaker on *"HESX1 and Septo-Optic Dysplasia"* at the GENESIS Investigators meeting: **XXVIIIth Eli Lilly International Symposium on Endocrinology and Development,** Sorrento, Italy, May 2000

233) Invited speaker on "*Genetic Disorders of Hypothalamo-pituitary development*" at the **Annual Meeting of the European Society for Paediatric Endocrinology,** Warsaw 1999

234) Invited speaker on *"Role of HESX1 in disorders of eye development"* at the **International EU meeting on "Developmental Genetic Disorders in Childhood involving the Eye",** Vilnius, Lithuania, June 1999

235) Invited speaker on *"Role of HESX1 in Septo-Optic Dysplasia"* at the **27th International Symposium GH and Growth Factors in Endocrinology and Metabolism**, Nice, April 1999; Plenary symposium

236) Invited speaker on "*Role of HESX1 in disorders of forebrain and pituitary development"* at the **XVIIth Eli Lilly International Symposium on Endocrinology and Development,** Amsterdam, March 1999

237) Invited speaker on "*Bioassay of Growth Hormone"* at the Workshop on *"New frontiers in growth hormone assays: can consensus be reached"?* at: **Growth hormone and growth factors in Endocrinology and Metabolism, 20th International Symposium**, Berlin, September 1995

238) Invited speaker on "*Physiology of Normal Growth*" at the Symposium on **"Growth hormone therapy of slow growth" at the XXIst International Congress of Paediatrics**, Cairo, Egypt, 1995

Academic Supervision

PhD students

1. **1998-2002**
   Dr. David McNay: Primary Supervisor
   PhD awarded June 2003: *Role of the homeobox gene HESX1 in hypothalamo-pituitary development in humans*
   Current post: Post-doctoral Fellow in Developmental Biology (Dr. SL Ang laboratory), National Institute of Medical Research, Mill Hill

2. **2000-2003**
   Dr. Luciani Carvalho (University of Sao Paolo, Brazil): Affiliate Supervisor 2001-2002

PhD awarded December 2003: *Functional investigation of the 126T mutation in HESX1*
Current post: Post-doctoral Fellow in the laboratory of Professor Sally Camper, University of Michigan, USA

3. **2004-2007**
Ms. Ezat Sajedi: Secondary Supervisor (Dr. JP Martinez-Barbera is the primary supervisor)
PhD awarded April 2008: *Role of Hesx1 in forebrain and pituitary development in mouse and man*

4. **2000-2004**
Dr. Maria Cundall: Affiliate Supervisor 2003-2004 (Dr. Karen Woodward was the primary supervisor)
PhD awarded 2006
Duplications of X chromosome associated with human disease

5. **2007-2010**
Dr. Ritika Kapoor: Secondary Supervisor (Dr. Khalid Hussain is the primary supervisor)
PhD awarded 2010
Genotype-phenotype correlations in children with congenital hyperinsulinism

6. **2005-2008**
Mr. James Turton: Secondary Supervisor (Dr. P Letissier at the MRC National Institute for Medical Research, Mill Hill, UK is the primary supervisor)
PhD awarded 2010
Investigation into the functional consequences of mutations of the transcription factor POU1F1 associated with Combined Pituitary Hormone Disease

7. **2008-2011**
Dr. Bruno Ferraz de Souza: Secondary supervisor (Dr. John Achermann is the primary supervisor)
PhD awarded 2011
The role of SF1 in adrenal development

8. **2005-2009**
Ms. Emanuela Spadoni: Secondary Supervisor (Dr. M Bitner-Glindzicz is the primary supervisor)
Microarray Comparative Genomic Hybridization in the detection of submicroscopic chromosomal aberrations in patients with complex disorders of pituitary development

9. **2007-2010**
Dr. Emma Webb: Primary Supervisor
PhD awarded 2013
Role of growth hormone on higher functioning in children
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10. **2006-2008**
Dr. Sandy Alatzoglou: Primary Supervisor
PhD awarded 2015
Investigation of the role of the transcription factors SOX2 and SOX3 in disorders of eye, forebrain and pituitary development in humans

12. **2007-2008**
Dr. Sara Tomaselli (affiliate student): Secondary supervisor (Dr. John Achermann is the primary supervisor)

13. **2008-2011**
Ms. Sujata Jayakody: Secondary supervisor (Dr. Juan-Pedro Martinez Barbera is the primary supervisor)
PhD awarded June 2012
The role of Hesx1 in hypothalamic development

14. **2011-2014**
Ms. Maha Sherif: Secondary supervisor (Dr. Khalid Hussain is the primary supervisor)
PhD awarded 2015
Genetic Aetiology of Diabetes Mellitus and Sensorineural Deafness

15. 2012-2015
Ms. Louise Gregory: Primary Supervisor "*Identification of novel molecular mechanisms in the aetiology of Congenital Hypopituitarism*"
PhD awarded 2016

16. **2013-2016**
Dr. Hoong Wei Gan: Primary Supervisor "*Investigation of Hypothalamic Obesity*"
PhD awarded 2020

17. **2018-2024**
Dr. Manuela Cerbone: Primary Supervisor "*Unravelling the Hypothalamic Syndrome*"

18. **2018-2022**
Dr. Sinead McGlacken Byrne: Secondary Supervisor "*Human Ovary Development and Dysfunction*"

MD Students

1. **2002-2005**
Dr. Ameeta Mehta: Primary Supervisor
MD awarded 2007: *Clinical and Genetic Characterization of Patients with Hypopituitarism*

2. **2017-2022**
Dr. Elim Man: Secondary Supervisor
An Investigation of Multidisciplinary Pathways in Paediatric DSD: Evaluation in Tertiary Centres in the United Kingdom and Hong Kong

Post-doctoral staff

1. **2000-2005**
Dr. Kathryn Woods

Dr. Woods was appointed as a post-doctoral fellow in our laboratory having completed her PhD under the supervision of Dr. Judith Goodship. Dr. Woods studied a number of genes implicated in hypopituitarism associated with midline defects. She performed a number of functional studies that have resulted in publications in high

impact journals such as *Journal of Clinical Investigation* and *American Journal of Human Genetics*. She is currently a scientist at the University of Southampton.

2. **2004-2008**
Dr. Dan Kelberman

Dr. Kelberman joined our laboratory at the end of 2004 and has successfully identified mutations in the transcription factors SOX2 and SOX3. He has performed a number of functional studies into the role of these factors in normal and abnormal CNS and pituitary development. His data were published in a number of high impact journals (*Journal of Clinical Investigation*, *Human Molecular Genetics*, *Journal of Clinical Endocrinology and Metabolism*). He has recently been appointed a senior post-doctoral scientist in the Ulverscroft Visual Sciences group at the Institute of Child Health London.

3. **2002-2006**
Professor John Achermann

I was Sponsor for a Clinician Scientist Fellowship [The NR5A subfamily of orphan nuclear receptors in human development and disease 2002-2006 (£329,000)] awarded to Dr. Achermann by the Wellcome Trust. Dr. Achermann is a Reader within the department, and is currently studying the role of genes encoding a number of transcription factors and other proteins in the development of the human reproductive system. I have also recruited a number of patients with disorders of sexual differentiation, many of whom have been found to harbour interesting mutations within genes implicated in normal sexual differentiation. A number of manuscripts have been published in high impact journals. Dr. Achermann has been awarded a Senior Clinical Fellowship with the Wellcome Trust, and more recently Professorship at ICH.

4. **2002-2005**
Professor Khalid Hussain

I supervised Dr. Hussain as a Clinical Lecturer at ICH between 2002 and 2005. During this period he built up his clinical and research portfolio which enabled him to establish himself as a full-time NHS consultant with considerable expertise in the area of congenital hyperinsulinism. More recently, he has been successful in obtaining a DH Senior Lectureship and more recently Professorship at ICH.

5. **2008-2012**
Dr. Mark McCabe

Dr. McCabe joined our laboratory at the end of 2008 and has successfully identified mutations in the FGF signaling pathway associated with complex midline defects of the brain and pituitary gland. These data will be submitted to high impact journals in due course. He is also working on the identification of partners and targets of the transcription factor SOX3.

6. **2007-2010**
Dr. Michelle O'Reilly

Dr. O'Reilly is a post-doctoral fellow with considerable expertise in the area of neurocognition. She is working on the effects of growth hormone on neurocognition.

7. **2007-2010**
Dr. Stephen O'Riordan

Dr. Stephen O'Riordan originally came to ICH as an ESPE Clinical Fellow studying the long-term complications of diabetes mellitus in children, focusing particularly on early detection of complications using novel Vitamin E markers. He has now been appointed as a Consultant Paediatric Endocrinologist in Cork.

8. **2017-current**
   Dr. Louise Gregory

   Dr. Louise Gregory started off as a Research Associate and the obtained her PhD in 2017. Following this, she has been appointed as a post-doctoral fellow in our laboratory at ICH.

9. **2021 – current**
   Dr. Naresh Hanchate

   Dr. Hanchate was appointed as a UCL Excellence Fellow and works on stress and the endocrine pathways impacting on this.

Research Associates

1. James Turton 2000 – 2005
2. Louise Gregory 2009-2012
3. Claire Leeson 2014-2015

Research Fellows

2003-2005
Dr. Rajeeb Rashid

Dr. Rashid was a Specialist Registrar who wished to gain experience in Molecular Medicine. He gained experience in the discipline by screening for mutations in the *GHRHR* gene in children with isolated growth hormone deficiency (IGHD).

2006
Katie Jordan

Ms. Jordan worked as a summer student in our laboratory and worked on the role of GH1 in the aetiology of IGHD.

MSc Students

2002-2003
Dr. Aileen Alston

Dr. Alston was a student on the clinical MSc course at ICH who under my supervision performed a retrospective analysis of children with congenital hypothyroidism as her dissertation thesis. She was awarded a Distinction for the thesis.

2007-2009
Dr. Vaitsa Tziaferi

Dr. Tziaferi is an Academic Clinical Fellow supervised by myself and was a student on the clinical MSc course at ICH who under my supervision performed a genetic analysis of patients with SOD/CPHD. She has identified mutations in a number of novel genes that were previously associated with Kallmann syndrome in patients with SOD/hypopituitarism. She was awarded a Distinction for the thesis, and we are currently preparing a manuscript based on her data.

2009 – 2011
Dr. Esi Iskeboah

Dr. Esi Iskeboah is an Academic Clinical Fellow supervised by myself and was a student on the clinical MSc course at ICH. I co-supervised her with Dr. Helen Spoudeas on a project evaluating children with craniopharyngioma.

Medical Students

2000
Ms. Neils Hauf (Medical Student, Berlin, Germany)
Ms. Hauf studied the role of *BMP4* in hypothalamo-pituitary disease during her 3-month attachment.

2004
Mr. Tom Winter (Medical Student, Cardiff Medical School)
Mr. Winter studied the role of the transcription factor *GLI2* in hypothalamo-pituitary disease during an eight-week attachment, and his findings will be submitted for publication in the near future.

2005
Mr. Cick Pouw (Medical student from Leiden University, Netherlands)
Mr. Pouw studied the role of *LHX4* in children with hypopituitarism (manuscript in preparation).

2006 Ms. Sabeen Ghause (CHRAT Summer Studentship Medical Student)
Ms. Ghause studied the promoter region of *SOX2* in children with eye and pituitary abnormalities, and her data will lead to a publication.

2007 Ms. Shireen Beebeejaun (CHRAT Summer Studentship Medical Student)
Ms. Beebeejaune studied the role of novel candidate genes in the aetiology of hypopituitarism.

2008 Dr. Leonidas Panagiotakopoulos (Affiliate Research student)
Dr. Panagiotakopoulos is studying the role of the Sonic Hedgehog pathway in pituitary disease.

ESPE Clinical Fellows:

Dr. John Torpiano, Malta, 2002-2003
Dr. Rocio Riatto Della Coletta, Brazil, 2009
Dr. Maria Guemes, Hospital Infantil Universitario Gregorio Marañon, Madrid, Spain, 2012
Dr. Manuela Cerbone, Naples, Italy 2013
Dr. Aleksandra Janchewska, Macedonia, 2014
Dr. Sukran Poyrazoglu, Istanbul, Turkey, 2014
Dr. Nicola Improda, Naples, Italy, 2014-5
Dr. Dogus Vurralli, Turkey, 2021-2

Academic Clinical Fellows

Dr. Vaitsa Tziaferi
Dr. Esi Iskeboah

Teaching/Educational Activity

1. Trainer in Paediatric Endocrinology at Great Ormond Street Hospital

As a Consultant in Paediatric Endocrinology, I am responsible for the Clinical Training of Senior House Officers and Specialist Registrars in Paediatric Endocrinology at GOSH. I am an Educational Supervisor for the ward Specialist Registrar. I have supervised the following SpRs over the last four years:

1) Dr. Ameeta Mehta
2) Dr. Rakesh Amin
3) Dr. Saji Alexander
4) Dr. Evangelia Charmandari
5) Dr. James Greening
6) Dr. Catherine Peters
7) Dr. Myra Poon
8) Dr. Raja Padidela
9) Dr. Li Chan
10) Dr. Sakthivel Gnanasambandam
11) Dr. Sophie Khadr
12) Dr. Sandy Alatzoglou
13) Dr. Renuka Dias
14) Dr. Vaitsa Tziaferi
15) Dr. Evelien Gevers (Trust Fellow)
16) Dr. Benila Ravindranathan
17) Dr. Phillip Murray
18) Dr. Claire Hughes
19) Dr. Rachel Besser
20) Dr. Harshini Katumgopala
21) Dr. Pratik Shah
22) Dr. Sandra Walton-Betancourth
23) Dr. Sasha Howard
24) Dr. George Paltoglou
25) Dr, Meera Shaunak

I have also been involved in training a number of Visiting Fellows:

1) Dr. Evelien Gevers – Senior Research Fellow, MRC National Institute for Medical Research; 2004-present
2) Dr. Ashiya Ali, SpR at Chelsea and Westminster Hospital
3) Dr. Radhika Putha, SpR at Chelsea and Westminster Hospital
4) Dr. Joanne Baker, Consultant Paediatrician, Kent and Canterbury Hospital; 2006 - present
5) Dr. Cristina Iglesias, Madrid, Spain; 08/01/2008 – 29/02/2008
6) Dr. Dina Cortez, Denmark; 25/02/2008 – 07/03/2008
7) Dr. Maite Echeverría, Madrid, Spain; 01/03/2008 – 30/04/2008
8) Dr. Konstantina Kosta, Registrar, Thessaloniki, Greece; 2008 – present
9) Dr. Sharmila Nambiar, SpR, QE II Hospital, Hemel Hempstead; 2009- present
10) Dr. Laura Manjon, Salamanca, February – April 2010, SpainDr. Santhosh Olety, SpR, Luton and Dunstable Hospital

11) Dr. Rodrigo Bancalari, Clinical Fellow, Santiago, Chile; 2010
12) Dr. Ved Arya, SpR, Southend Hospital, 2011
13) Dr. Pietro Lazzeroni, University of Padua, 2011
14) Dr. Shirley Wong, Queen Elizabeth Hospital, Hong Kong, 2011
15) Dr. Nicoletta Cresta, Naples, 2011
16) Dr. Hessa Al-Otaibi, Saudi Arabia, 2012
17) Dr. Sachin Mittal, India, RCPCH Bill Marshall Fellow 2014
18) Dr. Abhishek Kulkarni, India, RCPCH Visiting Fellow 2014
19) Dr. Jasmine Chow, Hong Kong, 2014-5
20) Dr. Rajni Sharma, India, RCPCH Visiting Fellow 2015
21) Dr. Gloria Pang, Hong Kong, 2015
22) Dr. Anton Holmgren, Stockholm, 2016
23) Dr. Chiara Guzzetti, Cagliari, 2016
24) Dr. Tansit Saengkaeuw, Bangkok, Thailand, 2016
25) Dr. Anna Malmer, Sweden, 2017
26) Dr. Julia Hoppmann, Leipzig, Germany, 2017
27) Dr. Ana Martins, Lisbon, Portugal, 2017
28) Dr. Silvana Calulo, Milan, Italy, 2017
29) Dr. Elisa Lambach, Brazil, 2017
30) Dr. Khadija Humayun, Pakistan, 2017
31) Dr. Grazia Morandi, Verona, Italy, 2017
32) Dr. Belen Roldan Martin, Madrid, Spain, 2017
33) Dr. Eleni Rapti, Thessaloniki, Greece, 2017
34) Dr. Beatriz Corredor, Madrid, 2017
35) Dr. Cecilia Cionna, Ancona, Italy, 2018
36) Dr. Jimena Lopez Cadal, Buenos Aires, Argentina, 2018
37) Dr. Giusy Patti, Genoa, Italy, 2017-2018
38) Dr. Hannah Tan, RCPCH Bill Marshall Fellow, Malaysia, October – December 2018
38) Dr. Federica Verdecchia, Rome, Italy 01/02/2019-31/07/2019
39) Dr. Elena Galazzi, Milan, Italy 03/12/2018-31/05/2019
40) Dr. Nese Akcan, Nicosia, Cyprus, 01/04/2019-30/06/2019
41) Dr. Arliena Amin, Putrajaya Hospital, Kuala Lumpur, Malaysia, 02/01/2020 – 13/03/2020; 01/02/22 - 17/05/22
42) Dr. Queenie See, Hong Kong, 03/02/2020 – 31/07/2020
43) Dr. Giulia Zichichi, University of Milan, 08/01/2020-09/03/2020
44) Dr. Tiziana Camia, University of Genoa, Italy 15/04/2023-31/08/23
45) Dr. Francesca Mainieri, University of Chieti, Italy 01/09/23 – 19/03/23

The training takes the form of ward-based teaching, out-patient supervision with a post-clinic meeting whereby all the patients are discussed, and ward round teaching.
I initiated and supervised the teaching programme at GOSH and UCLH for several years. This takes the form of monthly trainee Academic afternoons, held in conjunction with the Paediatric Endocrinology team at St. Bartholomew's Hospital. The meeting is well-attended by Paediatric Endocrine trainees from all over London. I participate actively in the teaching programme and give lectures as well as supervise presentations by junior staff.

2. Organizer of Pan-London Paediatric Endocrinology Group Meetings

I organized 4-monthly joint meetings between GOSH, UCLH, St. Bartholomew's, Kings College and St. George's Hospitals until the COVID-19 Pandemic. The meetings act as a forum to share ideas, to discuss difficult and interesting patients and to present research. The meetings are well-attended by the Paediatric Endocrine community in London, and form an important training component of the curriculum in Paediatric Endocrinology.

3. Teaching on post-graduate courses

I have taught regularly on the MRCPCH courses at ICH as well as on the PASTEST course. Additionally I used teach regularly on the MSc in Clinical Paediatrics at ICH as well as the MRes course. I regularly lectured on growth for a BDA course attended by Paediatric Dietitians.

4. Creator of Endocrine Protocols in clinical use at Great Ormond Street Hospital

I have compiled detailed protocols and guidelines that are currently in use in the Paediatric Endocrinology Department at GOSH and UCLH. These protocols and guidelines cover a wide range of endocrine conditions, and have been distributed widely to trainees as well as collaborating centers and outreach clinics both nationally and internationally (Malta, UAE). They have been updated recently and we have incorporated a major teaching element to the protocols.

5. Examiner to the Royal College of Paediatrics and Child Health (MRCPCH, DCH)

I am an examiner for the Royal College of Paediatrics and Child Health and regularly examine for the MRCPCH and DCH examinations. This is critical for the normal training and education of Paediatricians, and to ensure that future consultants will have met the high standard of practice that has been established by the RCPCH. Previously (1996), I organized the clinical MRCP Paediatrics Part II examination at GOSH for Professor Charles Brook, who was the host examiner. Appointed Senior Examiner 2019.

6. Appointed to Specialty Question Writing Group for MRCPCH

7. Teaching of Nursing Staff

I teach nursing staff at GOSH on an informal basis. Additionally, I teach members of the nursing staff on the Society for Endocrinology Nurses Meeting on a regular basis.

8. Teaching undergraduates at UCL

Approximately 3-4 undergraduate medical students from UCL are attached to the Endocrinology Unit at GOSH over a year. I coordinate the attachments and ensure that the medical students will have satisfactory exposure to all areas of Paediatric Endocrinology. Additionally, we have had a number of medical students who have wished to undertake laboratory work in our group (see above under “Academic Supervision”).

9. Appointed as Faculty to Endocrine Educational Initiative 2008

This initiative by Pfizer promotes the education and support of both trainees as well as new and established consultants. The focus is on equipping trainees/consultants with skills in management and to help trainees prepare for their consultant posts.

10. Organization of Scott Rivkees symposium at ICH – March 30-31 2009

I organised a 2-day symposium in paediatric endocrinology in honour of Professor Scott Rivkees from Yale University.

11. Founding member and Co-Chair of Ipsen Knowledge Exchange Programme (INKEP), member of steering committee.

This is an educational programme that includes Scandinavian paediatric endocrinologists as well as the Paediatric team at GOSH/UCLH. The annual meeting aims to foster an exchange of ideas and to establish collaborations as well as to present difficult clinical cases which then benefit from input by a number of experts in the field. The programme has expanded to include a number of European countries recently.

12. Lecturer on Short Stature at PUNCH meeting, London, 2011, 2012, 2013.

13. Initiated International Endocrine Update meetings twice a year at GOSH – sponsored by Ferring

Enabling Activity

Local

1. Academic and Clinical Lead in Endocrinology, UCL Institute of Child Health, London, and Great Ormond Street Hospital for Children, London
Management of a large clinical and research active Endocrinology Department. 8 Consultant staff, 8 trainees, 10 clinical nurse specialists and 8 scientific staff. I have expanded the department with the addition of 2 new consultants and 2 new clinical nurse specialists, as well as a Data Manager.

2. 2014-2018 I was appointed Section Head of the Genetics and Epigenetics section within the Programme of Genetics and Genomic Medicine in ICH.

2. 2003 – present Member of the Academic Board, University College London
I am a member of the Academic Board at UCL and therefore participate actively in the administrative activities at UCL.

3.Administration of Postgraduate Training programme in Paediatric Endocrinology
I was previously responsible for the initiation and organization of the Paediatric Endocrinology teaching programme at GOSH/UCLH. Additionally the various teaching activities at the London Centre for Paediatric Endocrinology are open to other Endocrine Centres within London. I arranged external funding (£3000/year) for these meetings on a recurrent basis.

4. Coordination of Joint Laboratory Meetings between ICH and UCLH
I organized the programme of fortnightly academic meetings in Endocrinology within ICH and UCL. This allowed interaction between the various academic groups in Endocrinology, namely forebrain and pituitary development, sexual differentiation, pancreatic disease and fetal growth and its impact on disease.

5**.** Organization of Pan-London Paediatric Endocrinology group meetings
These 4 monthly meetings allow interaction between Paediatric Endocrinologists in London at both Consultant and trainee level. They are well-attended and give rise to collaborative research studies as well as acting as an important forum for our junior staff to present data and patients of interest.

6**.** Organization of GOSH/UCLH Paediatric/Adolescent/Adult Endocrinology interface meetings
I have been instrumental in promoting interaction between the Adult and Paediatric/Adolescent Endocrinology teams at GOSH/UCLH by organizing 4 monthly meetings between the two Units. This allowed a seamless transition of Paediatric Endocrine patients through to the adult service, in addition to promoting clinical and academic collaboration between the disciplines.

7. Supervision of Audit projects
I have supervised numerous audit projects.

8. Appointed to Local ACCEA committee based at GOSH 2007-2010.

9. Appointed to the Rare Diseases Research Centre steering group: 2011 – 2013.

10. I am an Appraiser for GOSH, UCL and UCLH, and previously undertook 6-10 appraisals per year of colleagues.

National/International

1. Membership of the MRC College of Experts

   I was previously a member of the newly established MRC College of Experts. This is an organization that fulfills the important role of peer review for the Medical Research Council.

2. Peer Review Activities

   I am actively involved in peer review activities both at the national and international levels. This includes reviewing grants, manuscripts for journals such as the American Journal of Human Genetics, Journal of Clinical Endocrinology and Metabolism, Lancet etc, and marking abstracts for national and international meetings.

3. International Advisory Board membership

   I was previously the UK member of the Genentech and Ipsen Network Advisory Board (GAIN). The Board membership comprised leading Endocrinologists from the US and Europe (UK, Germany, France, Spain and Italy).

4. Editorial Board membership

   I have previously been a member of the Editorial Board for Clinical Endocrinology, which is the premier journal for clinical endocrinology in the UK. As a member of the editorial team, I reviewed manuscripts on a regular basis and attended Board meetings. I have also been appointed as Associate Editior for *Hormone Research*, and *Journal of Paediatric Endocrinology and Metabolism.* I was previously a member of the Editorial Board of the *Journal of Clinical Endocrinology and Metabolism* for four years. I was a member of the Advisory Boards of the recently launched *Disease Models and Mechanisms* and am a member of the Advisory Board for *Nature Reviews in Endocrinology.* I have been the Chief Section Editor for *Frontiers in Genomic Endocrinology.*

   Currently, I am Senior Editor for Endocrine Connections, and an Associate Editor on Hormone Research . Additionally, I serve on the Editorial Board of *JCEM Case Reports.*

5. Examining activities

   I have been appointed as an Examiner to the Royal College of Paediatrics and Child Health. This involves regular examining activities. I examine for the Diploma in Child Health and the Membership of the Royal College of Paediatrics and Child Health at least annually. I have also examined a number of PhD theses.

6. Participation in National and International Meetings

   I have chaired scientific sessions at major national and international meetings such as the International Society for Endocrinology, British Society for Paediatric Endocrinology, the European Society for Paediatric Endocrinology, British Endocrine Societies and the Endocrine Society, USA.

7. Outreach Service for training and clinical management

As part of my clinical role at GOSH/UCLH, I offer an outreach service to a number of hospitals both within the UK and abroad. These include visits to the UAE and Malta. The outreach service is crucial not only in terms of offering a comprehensive service to the patients, but also for training of local Paediatric Endocrinologists. They include out-patient teaching as well as formal lectures on various aspects of Paediatric Endocrinology. As a result of the outreach service, we have been able to accommodate trainees in our Department from the local hospitals. One such trainee, Dr. John Torpiano, was awarded an ESPE Clinical Training Fellowship in our Department from 2001-2002, and has now established himself as a Paediatric Endocrinologist locally in Malta, with support from our Department. Additionally, the outreach clinics have led to the recruitment of a number of patients for our research studies and have resulted in publications in high profile journals such as the *American Journal of Human Genetics* and the *Journal of Clinical Investigation*.

8. Appointments Advisory Committees

I have sat on a number of AACs for the appointment of Consultant Paediatricians in the UK.

9. CSAC Training site visits

I have performed site visits to assess training of Paediatric Endocrinologists at tertiary centres such as the Royal London/St. Bartholomew's Hospital.

10. Interaction with Patient Support Groups

I am a medical advisor to the national support group for patients with Septo-optic Dysplasia (FOCUS). I have also given talks to the Child Growth Foundation and the Bardet Biedl Support Group. This entails attendance at the meetings of the support group as well as speaking about our research into various endocrine conditions at these meetings. I also interact with the Living with CAH support group and am a founding member oof the ROHHAD International Consoortium. I am a medical advisor to the Noonan Syndrome Association.

11. Interaction with other national endocrine centres

I have been invited to give lectures on my research at a number of national Endocrinology centres. These include Manchester, Cardiff, Southampton, Birmingham, Cambridge, Oxford, Bristol and Newcastle. These visits have fostered collaborations between our laboratory group and geneticists and endocrinologists at these centres.

12. Appointed Chair of the Scientific Evaluation Committee for the EU-funded E-Rare Call for "European research projects on rare diseases" 2007.

13. Invited to give evidence at "Growth Hormone Therapy and Biosimilars in Clinical Practice Stakeholder Survey and Parliamentary Summit" meeting, Houses of Parliament, February 24 2009.

14. Founding member and Co-Chair of Ipsen Knowledge Exchange Programme (INKEP), member of steering committee. I organized the inaugural meeting in October 09-10 2008 at ICH. This educational initiative promotes clinical and research interactions between Scandinavia and the UK. I have organized several meetings in London subsequently.

15. Chairman and organizer of the 16th NovoNordisk Paediatric Endocrine workshop, Stratford-upon-Avon, December 2009. This was a 2-day meeting and I organized the programme and invited the speakers etc. The meeting was highly successful with excellent feedback.

16. Chairman-elect of the British Society for Paediatric Endocrinology and Diabetes 2009-2010.

17. Chairman of the British Society for Paediatric Endocrinology and Diabetes, 2010 – 2013. As the Chair, I represented the Executive Committee on the CSAC Advisory Group for the training of juniors, the Programme Organizing Committee, the Clinical Committee, the MCRN-CSG for Diabetes and Endocrinology, and the Clinical Excellence Awards committee.

18. Member of the European Society for Paediatric Endocrinology (ESPE) Research Unit Committee, 2011- 2013.

19. Host to the 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, 2011. This was widely regarded as one of the most successful BSPED meetings, with the highest number of delegates and submitted abstracts.

21. Chair of a meeting on: **"Improving patient outcomes in human growth hormone."** December 2012.

22. Appointed member of **Clinical Reference Group in Paediatric Medicine for National Commissioning** 2013 - 2019.

23. Member of the Programme Organizing Committee, European Society for Paediatric Endocrinology, 2011 - 2019.

24. **Chair of the Programme Organizing Committee, European Society for Paediatric Endocrinology**, 2013 - 2019.

25. Member of Paediatric steering group of EU – supported COST action on **Hypogonadotrophic Hypogonadism.**

26. Founder member and Steering group member of the newly established **EMA supported EnPrEMA initiative in Paediatric Endocrinology and Diabetes (CADET).**

26. Principle Coordinator of Chapter on Hypothalamo-pituitary disease for the International Classification of Paediatric Endocrine Disease
I led a team of 4 internationally renowned consultants in this initiative.

27.. I also established the annual **"International GHD Academy"** for newly appointed endocrinologists from all over the world, together with an adult endocrinologist.

28. I also initiated twice yearly international educational meetings at GOSH sponsored by Ferring.

29. **Paediatric member AAGBI Steroid Working Party**

30. **100000 Genomes**
Deputy Lead Growth and Endocrine Disorders subdomain, member Paediatric Cross-cutting GECIP domain Oversight committee for 100000 genome initiative; member of Endocrine subdomain in Endocrine and Metabolism GECIP. In 2015 I wrote the Data Model form for Short stature syndromes for Genomics England.

31. Steering Committee member for **EDGE (Endocrinology Debate and Exchange)** meetings sponsored by Sandoz (2013- current).

32. Steering Committee member of **IPSEN iNCGS** (International Cooperative Growth Study) initiative (2015-current).

33. Steering Committee member of **Best Practice and Controversies in Growth Hormone Treatment in Children Meeting,** May 2016

34. Steering Committee member of the **Severe Insulin Resistance** meeting, January 2017

35. Chair of Scientific Organizing Committee, **Endocrine First Global Summit,** October 2017

35. Member of **Global Norditropin Advisory Board**, December 2017

35. Steering Committee member of the **Italian Debate for Endocrinology Experts Meeting,** April 2018 -current

36. Member of Steering Committee of **ENDO-ERN (European Reference Network). Co-Chair of Pituitary Main Thematic Group**

37. Lead of GOSH-UCLH HCP of Endo-ERN

38. Norrie Disease Foundation Medical Advisory Board member

39. Invited member Italian SOD/Pituitary Family Association

40. Appointed Senior Examiner at RCPCH, 2019

41. External Examiner for 1st Hong Kong Paediatric Endocrine Exit Exam, Hong Kong 2022

Contributions to the European Society for Paediatric Endocrinology (ESPE):

I have been a member of ESPE since 1990. I have attended the Annual Meeting continuously since then, with only one exception. I have submitted numerous abstracts to the meeting over the years, and I have been awarded the Henning Anderson prize for best abstract on two occasions (1998, 2010). I have contributed to the meetings as Session Chair as well as speaker on several occasions. I have been a member of a number of committees as well as scientific activities, including Faculty member of several ESPE schools and scientific meetings. These are summarised below.

1. Member, ESPE Research Unit (2010-2013)
2. Member, ESPE Program Organizing Committee (POC; 2011-2019)
3. Chair of the ESPE Program Organizing Committee (2014-2019)
4. Associate-Editor, ESPE-Yearbook of Pediatric Endocrinology (2006-2013)
5. Associate Editor, Hormone Research in Paediatrics, (2008-current)
6. Supervision of ESPE Research Fellow – Dr. Alatzoglou
7. Supervision of eight ESPE Clinical Fellows
8. Faculty, ESPE Summer School (2010, 2019)
9. Faculty, ESPE Advanced Seminar in Developmental Endocrinology (2012)
10. Faculty, ESPE Science School (2012)
11. Member ESPE Council (2013-2021)
12. Programme organisation of ESPE Connect Online 2020
13. Host and programme organisation ESPE 2021 Online
14. Organisation and Chair of ESPE Connect Webinar on Noonan Syndrome, October 12 2021
15. Annual Meeting Host ESPE 2024, Liverpool, UK

As POC Chair, I have organized seven highly successful meetings including Dublin in 2014, Barcelona in 2015, Paris in 2016, Athens in 2018 and Vienna in 2019. In 2020 and

2021, together with POC Chair Professor Nils Krone, we adapted the Annual ESPE meeting to an online format. The meetings were attended by a large number of delegates and the feedback was generally very good.

I have also contributed as a POC member to the joint meeting with the Pediatric Endocrine Society held in Washington in 2017. I served as one of the two ESPE representatives for the joint IPOC for the IMPE meeting held in Buenos Aires in 2023.

Medicolegal Work

I have advised as an expert witness in a number of medico-legal cases that have involved the endocrine system. I am familiar with the process of preparing a medicolegal report. My main area of research expertise lies in the field of disorders of hypothalamo-pituitary structure and function, although my clinical expertise spans the whole of paediatric endocrinology.

Clinical Practice

I practice as a Consultant in Paediatric and Adolescent Endocrinology at Great Ormond Street Children's Hospital and at University College London Hospital and have under my care a large number of patients with common and uncommon endocrine disorders. I served as the Head of Clinical Service in Endocrinology at GOSH from 2006-2023. My clinical workload is equivalent to a full-time NHS consultant.

I also practice as a Consultant in Paediatric Endocrinology at the following out-reach hospitals:
Kent and Canterbury Hospital, Canterbury, Kent
Lister Hospital, Stevenage (until 2010)
West Middlesex Hospital, London (until 2018)
The Whittington Hospital, London
St. Luke's Hospital, Malta
Dubai Hospital, UAE (until 2017)
Luton and Dunstable Hospital
Northwick Park Hospital

I run and manage an internationally recognized tertiary paediatric endocrine service based at 2 main sites (GOSH and UCLH) with an emphasis on growth, pituitary, adrenal, thyroid and pancreatic disorders. This activity is undertaken predominantly as an outpatient specialty with inpatient activity centred mainly on investigative endocrinology and patients with persistent hyperinsulinaemic hypoglycaemia. On the whole, I look after children with a range of complex endocrine disorders. My paediatric patients are followed up at Great Ormond Street Hospital, whereas the adolescent patients are followed up at UCL Hospitals. Recently, the inpatient workload has increased considerably as a result of referrals of complex endocrine problems from a variety of specialities throughout GOSH and externally, and as a result of the retirement of a colleague who was been replaced. My clinical workload (contract with GOSH and UCLH of 6PAs) is equal to that of a full-time NHS consultant. Between 2010-11, I saw 1140/3664 outpatient consults and looked after 481/1078 inpatient admissions in Endocrinology.

As a result of my clinical role, I have embarked on a number of clinical studies. These include the following:

1. Principal UK clinical investigator in the following studies sponsored by Eli Lilly and Company:
    a. The Genetics and Neuroendocrinology of Short Stature International Study

b. Optimisation of Growth Hormone Treatment in Short Children Born Small for Gestational Age Based on a Growth prediction Model

2. Chief Investigator (UK) on the International Cooperative Growth Study (iNCGS) Post-Marketing Surveillance Programme for Nutropin

3. Investigator Pfizer KIGS study - Post-Marketing Surveillance Programme for Genotropin

4. Co-Investigator on 18BB32 Extension of Dasiglucagon for Treatment of Children with Congenital Hyperinsulinism (IRAS: 249715) and 18BB39 A Two-Period, Open-label Trial Evaluating the Efficacy and Safety of Dasiglucagon (IRAS: 248535)

5. A phase 3, randomized, multicenter, open-label, crossover study assessing subject perception of treatment burden with use of weekly growth hormone (somatrogon) versus daily growth hormone (genotropin®) injections in children with growth hormone deficiency Pfizer Somatrogon Study C0311002. PI

6. Defining the evidence base for health care delivery in congenital adrenal hyperplasia – a multi-centre CAH-UK (Children & Adolescents with CAH in the UK) initiative CAH-UK – Investigator. Co I

7. Chief Investigator NIHR UK Bioresource Rare Diseases – Septo-optic Dysplasia PI

8. Chief Investigator NIHR funded GHD Reversal study - £1,852,244.98

9. UK Chief Investigator Pfizer PROGRES study

10. UK Chief Investigator Crinecerfont CAH 2006 Study sponsored by Neurocrine. A Randomized, Double-Blind, Placebo-Controlled Study to Evaluate the Safety and Efficacy of Crinecerfont (NBI-74788) in Pediatric Subjects with Classic Congenital Adrenal Hyperplasia, Followed by Open-Label Treatment

11. RM-493-040 Study sponsored by Rhythm Pharmaceuticals. A Phase 3, Double Blind, Randomized, Placebo-Controlled Trial to Evaluate Setmelanotide in Obesity. PI

International Outreach Service

I provided an annual outreach service to Dubai Hospital between 2004 and 2017. I attended 4 all day clinics during a 5 day stay and provided 2 lectures to the Paediatricians. I advised on patients with all types of endocrine conditions including growth, thyroid and pubertal disorders, obesity, disorders of calcium homeostasis and Disorders of Sex Development. The service was established by Dr. Hassan Mundi in collaboration with me and is now very busy.

Additionally, I provide an annual outreach service to Mater Dei Hospital in Malta, where I attend 3 all day clinics during a 3 day stay. I see patients with a wide variety of endocrine conditions during each visit. This service was established with Dr. John Torpiano and now runs very successfully. We are currently collaborating on research studies which will bring a research aspect to the service.

Research Activity

General

The Developmental Endocrinology Research Group at ICH and the London Centre for Paediatric Endocrinology is one of the largest grouping of clinical and basic scientists studying Developmental Endocrinology in the UK. We have established ourselves as the leading centre for the clinical and genetic evaluation of patients with congenital pituitary disorders in the UK and internationally, and receive DNA samples from leading national and international Endocrine centres. Close links existed between my group and the Divisions of Molecular Endocrinology and Developmental Genetics at the MRC National Institute for Medical Research at Mill Hill, and subsequently the group of Professor Robin Lovell-Badge at the Crick Institute in London. The collaboration epitomizes the concept of translational research.

Experience

My training as a Research Fellow in Endocrinology (1991-1994) under the supervision of Professor Charles Brook, Dr. Peter Hindmarsh and Dr. Nick Marshall at the Middlesex Hospital enabled me to become fully conversant with cell culture techniques and assay methodology and validation, and led to the award of an MD degree. Subsequently, my post-doctoral secondment to the National Institute of Medical Research (1996-1998) enabled me to become fully conversant with a range of techniques in molecular biology and allowed me to develop my research interest in pituitary development.

Research Programme

The aim of my main research programme is to understand the mechanisms underlying the development of the forebrain, the hypothalamus and the pituitary gland, as well as other rare endocrine disorders (eg thyroid and adrenal). This includes factors involved in hypothalamic development, early anterior pituitary development and specification of the cell types contained within the anterior pituitary.

The Research Programme focuses on the following:

1. The role of transcription factors and other developmental genes in forebrain, pituitary and eye development

The basis for this work is the unrivalled DNA sample – patient phenotype database that we have created which now consists of over 2000 DNA samples. Using this detailed resource we have identified a number of novel dominant and recessive mutations within a number of genes and investigated the functional consequences of the mutations in detail. In collaboration with Dr. Rosa Beddington and Professor Iain Robinson at the MRC National Institute for Medical Research, we were the first to describe a genetic basis for septo-optic dysplasia (SOD). In particular, we demonstrated that Hesx1/HESX1 acts as a repressor of transcription and mutations were associated with SOD. Subsequently, we identified mutations in *HESX1* in association with combined pituitary hormone deficiency (CPHD) and isolated GH deficiency (IGHD). These findings have been published in *Nature Genetics*, *Human Molecular Genetics, Development* and the *Journal of Clinical Investigation*. The penetrance of the mutations within *HESX1* is highly variable, suggesting the role of other genes and/or environmental factors. In collaboration with Prof. Juan-Pedro Martinez in the Division of Developmental Biology at ICH, we inserted the human mutations into murine models and established the effects of these on forebrain and pituitary development and on neuroendocrine function.

We have also identified mutations within a novel gene *SOX2* in a cohort of patients with anophthalmia and hypopituitarism, in association with other features such as oesophageal atresia, agenesis of the corpus callosum and developmental delay and learning difficulties. These studies have been performed in collaboration with **Professor Iain Robinson and Dr. Robin Lovell-Badge at the National Institute for Medical Research**, who showed that mice that are heterozygous for a null mutation within the gene manifest dysgenesis of Rathke's pouch in addition to poor growth and impaired fertility. We have shown a role for SOX2 in progenitor cell proliferation in the developing and post-natal pituitary, and more recently, we have shown that SOX2 can

impact on GnRH neurons and also on pituitary development. We plan to embark on studies that will further elucidate the role of Sox2/SOX2 in murine and human development, as well as identification of partners and putative targets of the protein. Since SOX2 is implicated in stem cell differentiation as well as possible neuronal migration, these studies will cast light on the processes implicated in neuronal stem cell differentiation and neuronal migration in humans.

In collaboration with **Professor Iain Robinson and Dr Robin Lovell-Badge at the National Institute for Medical Research and Dr. Paul Thomas at the Murdoch Institute in Melbourne, Australia**, we investigated the role of a novel gene *SOX3* in forebrain and pituitary development. The gene is located on the X chromosome. We identified a polyalanine expansion in a pedigree in which 3 boys are affected with pituitary hypoplasia. Additionally, collaborative studies with **Dr. Karen Woodward and Dr. Maria Bitner-Glindzicz in the Department of Molecular Genetics, ICH,** suggested that duplications of the gene may be associated with a phenotype that encompasses hypothalamo-pituitary disease. We showed that infundibular, and consequently pituitary, development is critically dependent upon the dosage of SOX3, and these data were published in the *American Journal of Human Genetics*. We are currently attempting to further elucidate the phenotypes associated with *SOX3* mutations in humans, and to identify putative partners and targets of this gene.

2. Hypothalamic Factors and the Phenotype

As a result of the above observations we have screened a number of genes that are implicated in forebrain and pituitary development for mutations. Given the expression patterns of a number of developmental genes within the prospective forebrain and developing pituitary, we have selected these as candidate genes for septo-optic dysplasia and related disorders. We have now identified a number of variations in many of these genes (*GLI2, OTX2*). Functional studies of these changes are currently being undertaken. We have recently identified mutations in a novel pathway implicated in hypothalamic development. Mutations in *ARNT2* are associated with a novel syndrome characterized by multiple pituitary hormone deficiency, renal and urinary tract abnormalities and seizures with post-natal microcephaly.

3. Specification of Anterior Pituitary Cells

Mutations within a number of genes are known to be associated with combined pituitary hormone deficiency (CPHD) or isolated growth hormone deficiency (IGHD). These include *Prophet of Pit1* (*PROP1*), *POU1F1*, *LHX3*, *LHX4*, *GHRHR* and *GH*-1. We have screened a large cohort of patients within each phenotypic category. We have identified a number of known and novel mutations within *LHX3, PROP1*, *POU1F1*, *GH1* and *GHRHR*. These studies represent the largest cohort of patients screened for mutations within these genes.

4. Identification of novel candidate genes in hypopituitarism and related disorders

As a result of our genetic studies, we have now accumulated over 2000 DNA samples from patients with hypopituitarism that is isolated or associated with other phenotypic features. A number of these samples are derived from familial cases. Additionally, we have identified several de novo chromosomal translocations in association with hypopituitarism. Using techniques such as homozygosity mapping, exome and genome sequencing, microarray comparative genomic hybridization and a candidate gene approach, we have identified several novel genes that are implicated in these disorders. These studies have led to the identification of several novel genetic pathways implicated in hypothalamo-pituitary development. I have also described a number of novel deisease entities associated with novel molecular mechanisms.

5. Refining the Definition of the Human Phenotypes: Clinical studies of children with congenital growth hormone deficiency.

We are currently performing studies on children with congenital GHD, either in isolation or associated with other pituitary hormone deficiencies and/or midline defects. We have established a clinical base for children with these disorders and are currently defining the clinical phenotypes in detail. Such studies include a re-evaluation of the assessment of neuro-endocrine function and the methods of assessing the gonadal axis in infants and children. Our studies are giving us novel insights into hypothalamo-pituitary function so that we are now re-defining protocols for the investigation and treatment of these children. Given the unique nature of Paediatric practice at Great Ormond Street Hospital, we are in a position to collaborate extensively with other groups such as **Opthalmology, Genetics, Neurology, Neuroradiology and Developmental Paediatrics**. We have identified a high incidence of Autistic Spectrum Disorder in these patients and are currently evaluating this further in collaboration with **Dr. Alison Salt and Dr. Naomi Dale at the Wolfson Institute.** Recently, in collaboration with **Dr. Chris Clark and Dr. Naomi Dale at ICH,** we have used newer modalities of imaging such as Diffusion Tensor Imaging and Volumetric analysis to identify specific abnormalities in various brain structures such as the hippocampus and basal ganglia in children with IGHD. We have correlated these to specific neurodevelopmental deficits in the children with IGHD.

6. Investigation of Hypothalamic Dysfunction
We are currently investigating patients with hypothalamic dysfunction to identify novel treatment modalities for this rare but debilitating and life-threatening condition. We are focusing on the investigation of hypothalamic obesity as well as delineating the anatomy of the hypothalamus on MRI scanning.

7. Congenital Adrenal Hyperplasia
This is a condition characterized by adrenal insufficiency, often associated with significant co-morbidities including hyperandrogenism with severe virilisation, obesity, precocious puberty and short stature. The condition can be highly challenging and careful monitoring and management is required. We aim to identify novel biomarkers and novel methods of measuring known biomarkers in order to optimise disease control. These studies will be performed in collaboration with scientists at UCL GOS ICH and clinical scientists based at Sheffield University.

Management activity

I was Specialty Lead/Head of Clinical Service in Endocrinology at GOSH from 2006 to 2023. The endocrine team consists of 10 consultants, 7 SpRs/Clinical Fellows, 7 CNS and 2 SHOs. I led the Department on my own between 2006 and 2019, and was then co-Lead until April 2023. I was previously Head of the Section of Genetics and Epigenetics in Health and Disease (GEHD) within the Genetics and Genomic Medicine Programme at the UCL Institute of Child Health. As Section Lead, I had overall responsibility for the GEHD Section, which includes 8 PIs including 7 Professors. Under my leadership, the Endocrinology department has expanded by 4 further consultants as well as 5 new members of the CNS team and a data manager. I was responsible for the appraisal and mentoring of senior members of staff, and also appraised other academic members of staff. I was responsible for both NHS and academic job planning for the Department. I attended monthly senior staff meetings as well as team meetings for the rest of the department. I was adopted onto the Variability and Flow management process in the hospital and was on the Steering Group for the project. I participated in the E3 Senior Leaders Course in 2009. I was appointed to the Steering Group for the Rare Diseases Centre which will be established at Great Ormond Street Hospital for Children. I was Chairman of the British Society for Paediatric Endocrinology and Diabetes from 2010-2013. I have been a member of the Research Unit and Programme Organizing Committees of the European Society for Paediatric Endocrinology (ESPE), and was Chair of the Programme Organising Committee of ESPE from 2013-2019. I have been a member of ESPE Council since 2013. I am currently Host-Elect of the Annual Meeting of ESPE to be held in Liverpool, UK, in September 2024.

Media Exposure

I have been involved with the media on several occasions, providing advice on a range of paediatric endocrine conditions such as septo-optic dysplasia, skeletal dysplasias and precocious puberty. I have appeared on the Today programme for Radio 4, newspapers such as the *Daily Mirror*, the *Daily Express, Guardian* and the *Sunday Times*, and specialist television programmes such as *Extraordinary 10 year olds: remarkable kids in Britain, Harvey and I* and *Embarrassing Bodies*.

Publications

A. Book Chapters

1. Savage MO, **Dattani MT**, Perry LA, Donaldson MC, Grant DB, Hughes IA, Brook CGD, Chaussain JL (1995) Clinical spectrum, endocrine characteristics and aspects of therapy in patients with 5-alpha reductase deficiency. *In: Les ambiguites sexuelles.* Ed. JL Chaussain, IU Roger, Publi-fusion, Cahors pp. 19-28.

2. Dattani MT, Preece MA Physical Growth and Development (1998) *In: Forfar and Arneil's Textbook of Paediatrics, 5th edition.* pp. 349-380. Churchill Livingstone, London.

3. Dattani MT, Brook CGD Precocious puberty (1998). *In: Estrogens and Progestogens in Clinical Practice, 1st edition* pp. 495-507. Churchill Livingstone International.

4. Dattani MT The measurement of growth hormone (1999) *In: Growth hormone therapy in KIGS: 10 years experience* pp. 43-53. Johann Ambrosius Barth Verlag, Leipzig.

5. Dattani MT, Brook CGD Adolescent Health Problems (2000). *In: Harrison's Principles of Internal Medicine 15th edition* pp.31-36. Ed. Braunwald E, Hauser SL, Fauci AS, Kasper DL, Longo DL, Jameson JL McGraw-Hill.

6. Dattani MT Tests in Paediatric Endocrinology (2001) *In: Clinical Paediatric Endocrinology 4th edition* pp. 467-495 Ed. CGD Brook, PC Hindmarsh Blackwell Science Limited.

7. Dattani MT Septo-optic dysplasia and associated brain malformations (2001) *In: Endocrine Development: Hypothalamo-pituitary development,* pp. 77-93, Ed: R. Rappaport, S. Amselem, Karger.

8. Dattani MT, Hindmarsh PC Premature sexual maturation (2003) *In: Oxford Textbook of Endocrinology and Diabetes* pp 1046-1063, Ed: S. Shalet, JAH Wass Oxford University Press.

9. Woods K, **Dattani MT** Transcription factors involved in disorders of forebrain and pituitary development (2003) *In: Molecular Basis of Inborn Errors of Development* pp. 540-551, Ed. C Epstein, R Erickson, A Wynshaw-Boris, Oxford University Press.

10. Dattani MT, Hindmarsh PC Growth hormone deficiency in children (2005) In*: DeGroot's Endocrinology 5th edition* pp. 733-754, Ed. S Melmed, Elsevier, New York.

11. Dattani MT, Hindmarsh PC Normal and abnormal puberty (2005) *In: Clinical Paediatric Endocrinology 5th edition* pp. 183-210, Eds Brook CGD, Brown R and Clayton PE, Blackwell Science, Oxford.

12. Mehta A, **Dattani MT** Congenital Disorders of the hypothalamo-pituitary axis (2005) *In: Clinical Paediatric Endocrinology 5th edition* pp. 67-89, Eds Brook CGD, Brown R and Clayton PE, Blackwell Science, Oxford.

13. Dattani MT, Grant D, Baumer H, Mallam K, Brook CGD Endocrinology (2007*) In: The Great Ormond Street Colour Handbook of Paediatrics and Child*

Health, Ed. Strobel S, Marks S, Smith PK, El Habbal MH, Spitz L, Manson Publishing Ltd.

14. Dattani MT, Gevers E, Hindmarsh PC Growth and growth factors (2007) *In: Yearbook of Paediatric Endocrinology 2007***,** Ed. JC Carel, Z Hochberg, Karger.

15. Kelberman D, **Dattani MT** Transcription factors involved in disorders of forebrain and pituitary development (2008) *In: Molecular Basis of Inborn Errors of Development*, Ed. C Epstein, R Erickson, A Wynshaw-Boris, Oxford University Press.

16. Kelberman D, **Dattani MT** SOX3 and infundibular hypoplasia (2008) *In: Molecular Basis of Inborn Errors of Development*, Ed. C Epstein, R Erickson, A Wynshaw-Boris, Oxford University Press.

17. Gevers E, **Dattani MT** Growth and growth factors (2008) *In: Yearbook of Paediatric Endocrinology 2008***,** Ed. JC Carel, Z Hochberg, Karger.

18. Mehta A, Gevers E, **Dattani MT** Congenital Disorders of the hypothalamo-pituitary axis (2009) *In: Clinical Paediatric Endocrinology 6th edition* Eds Brook CGD, Brown R and Clayton PE, Blackwell Science, Oxford.

19. Alatzoglou KS, **Dattani MT** Acquired Disorders of the hypothalamo-pituitary axis (2009) *In: Clinical Paediatric Endocrinology 6th edition* Eds Brook CGD, Brown R and Clayton PE, Blackwell Science, Oxford.

20. Dattani MT, Tziaferi V, Hindmarsh PC Normal and abnormal puberty (2009) *In: Clinical Paediatric Endocrinology 6th edition* Eds Brook CGD, Brown R and Clayton PE, Blackwell Science, Oxford.

21. Gevers E, **Dattani MT** Growth and growth factors (2009*) In: Yearbook of Paediatric Endocrinology 2009***,** Ed. JC Carel, Z Hochberg, Karger.

22. Dattani MT, Hindmarsh PC Growth hormone deficiency in children (2011) In*: DeGroot's Endocrinology 6th edition* Ed. S Melmed, Elsevier, New York.

23. Webb E, **Dattani MT** In: Diagnosis of growth hormone deficiency (2010) In: Current Indications for Growth Hormone Therapy 2nd Edition, Endocrine Development 18: 55-66 Ed. PC Hindmarsh

24. Martinez-Aguayo, A, **Dattani MT**, Achermann JC (2010) Gonadotropin hormones: Disorders *Encyclopedia of Life Sciences*

25. Gevers E, **Dattani MT** Growth and growth factors (2010) *In: Yearbook of Paediatric Endocrinology 2010***,** Ed. JC Carel, Z Hochberg, Karger.

26. Dattani MT, Fisher D, Hindmarsh PC Neonatal Endocrinology (2011) In: Williams Textbook of Endocrinology, Ed. S Melmed,

27. Alatzoglou KS, **Dattani MT** (2011) *In: Oxford Textbook of Endocrinology and Diabetes 2nd edition* Ed: JAH Wass Oxford University Press.

28. Gevers E, **Dattani MT** Growth and growth factors (2011) *In: Yearbook of Paediatric Endocrinology 2011***,** Ed. JC Carel, Z Hochberg, Karger.

29. Tziaferi V, **Dattani MT** (2011) Embryology, Anatomy and Physiology of the Pituitary gland *In: Endocrine Surgery in Children,* Ed. Daniel Ledbetter and Paul Johnson.Springer Press.

30. Gevers E, Gaston-Massuet C, **Dattani MT** Growth and growth factors (2012) *In: Yearbook of Paediatric Endocrinology 2012***,** Ed. K Ong, Z Hochberg, Karger.

31. Bancaleri R, Gregory L, McCabe M, Dattani MT (2012) Pituitary Development: An Update. In: *Endocrine Development* **23:**1-15 Ed. PE Mullis

32. Gevers E, Gaston-Massuet C, **Dattani MT** Growth and growth factors (2013) In: *Yearbook of Paediatric Endocrinology 2013***,** Ed. K Ong, Z Hochberg, Karger.

33. McCabe MJ, **Dattani MT** Genetic aspects of hypothalamic and pituitary gland development (2013) In: *Clinical Neuroendocrinology*, Ed. M. Korbonitz, E. Fliers, JA Romijn Elsevier Science Publishers

34. Backeljauw PF, **Dattani MT**, Rosenfeld R, Cohen P Disorders of Growth Hormone/Insulin-Like Growth Factor Secretion and Action (2013) In: *Pediatric Endocrinology*, Ed. MJ Sperling

35. Peters CJ, **Dattani MT** Endocrinology (2013) In: *GOSH Handbook of Paediatrics and Child Health*

36. Miller B, Frohnert B, **Dattani MT** Pituitary and Hypothalamus (2013) In: *Pediatric Endocrinology and Inborn Errors of Metabolism Ed.* K Sarafoglou, Hoffman G, Roth K

37. Alatzoglou KS, Kelberman D, **Dattani MT** SOX3 and Infundibular Hypoplasia (2014) In: Epstein's Inborn Errors of Development, Eds. RP Erickson, A. Wynshaw-Boris

38. Kelberman D, **Dattani MT** *EMX2* **and** *HESX1* and Type I Schizencephaly and Septo-Optic Dysplasia (2014) In: Epstein's Inborn Errors of Development, Eds. RP Erickson, A. Wynshaw-Boris

39. Alatzoglou KS, **Dattani MT** Growth Hormone Deficiency in Childhood (2014) In: Jameson/De Groot Endocrinology Adult and Pediatric 7th Edition.

40. Gevers E, Fisher D, **Dattani MT** Fetal and Neonatal Endocrinology (2014) In: Jameson/De Groot Endocrinology Adult and Pediatric 7th Edition.

41. Gevers E, **Dattani MT** Endocrinology of Fetal Development (2015) In: Williams Textbook of Endocrinology 13th Edition.

42. Gan HW, Spoudeas H, **Dattani MT** Endocrine Deficits in Human Craniopharyngioma (2017) In: Basic Research and Clinical Aspects of Adamantinomatous Craniopharyngioma, Eds. C Andoniadou, JP Martinez-Barbera.

43. Frohnert BI, Miller BS, **Dattani MT** *Developmental Disorders of the Anterior Pituitary* (2017) In: Essential Paediatric Endocrinology and Metabolism, Eds. K. Sarafoglou.

44. Gregory L, Dattani MT *Development of the Pituitary and Genetic forms of Hypopituitarism* (2018) In: Oxford Textbook of Endocrinology and Diabetes, Ed. John Wass

45. Gan HW, Cerbone M, Dattani MT *Hypothalamic dysfunction (Hypothalamic syndromes)* (2018) In: Oxford Textbook of Endocrinology and Diabetes, Ed. John Wass

46. Paltoglou G, Katugampola H, Dattani MT *Genetic and Hormonal Control of Growth* In: Encyclopedia of Endocrine Disease 2nd edition, Ed. Ilpo Huhtaniemi, Elsevier publishers

47. Gregory L, Dattani MT *Genomics of the Pituitary Gland* In: Pituitary Disorders of Childhood: Diagnosis and Clinical Management, Ed. Brenda Kohn, Springer Publishers

48. Katugampola H, Cerbone M, Dattani MT *Hypothalamus and Pituitary Physiology*, In: Maternal-Fetal and Neonatal Endocrinology, Eds C Kovacs, C Deal

49. Cerbone M, Katugampola H, Dattani MT *Hypothalamus and Pituitary Disorders*, In: Maternal-Fetal and Neonatal Endocrinology, Eds C Kovacs, C Deal

50. Katugampola H, Gevers E, Dattani MT (2018) *Endocrine Disorders in the Neonate* In: Williams Textbook of Endocrinology, Ed Shlomo Melmed

51. Baron J, Jorge A, Dattani MT, Grimberg A *Disorders of Childhood Growth* (2020) Sperling: *Pediatric Endocrinology*, 5th edition

52. Bancaleri R, Dattani MT *Pituitary Development and Physiology* Libro Endocrinología Pediátrica Práctica (in press)

53. Dastamani A, Dattani MT *Growth Disorders* De Groot's Endocrinology 8th edition (In Press)

54. Katugampola H, El-Khairi R, Dattani MT *Neonatal endocrinology* De Groot's Endocrinology 8th edition (In Press)

55. Katugampola H, El-Khairi, Dattani MT (2022) *Endocrine Disorders in the Neonate* In: Williams Textbook of Endocrinology, Ed Shlomo Melmed

B. Refereed articles

Total Citations: 18,984

h-index: 75
i10 index: 209

1. **Dattani MT**, Pringle PJ, Hindmarsh PC, Brook CGD (1992) What is a normal stimulated growth hormone concentration? *Journal of Endocrinology* **133:**447-450.

2. **Dattani MT**, Hindmarsh PC, Weir T, Robinson ICAF, Brook CGD, Marshall NJ (1993) Enhancement of growth hormone bioactivity by zinc in the eluted stain assay system (ESTA). *Endocrinology* **133:**2803-2808.

3. **Dattani MT,** Hindmarsh PC, Brook CGD, Robinson ICAF, Marshall NJ (1994) Inhibition of growth hormone bioactivity by recombinant human growth hormone-binding protein in the eluted stain assay (ESTA) system. *Journal of Endocrinology* **140:**445-453.

4. Ealey PA, Yateman M, Sandhu R, **Dattani M,** Hassan M, Holt S, Marshall NJ (1995) Development of an eluted stain bioassay (ESTA) for hGH. *Growth Regulation* **5:**36-44.

5. **Dattani MT,** Hindmarsh PC, Brook CGD, Robinson ICAF, Kopchick JJ, Marshall NJ (1995) G120R, a human growth hormone antagonist, shows zinc-dependent agonist and antagonist activity on Nb2 cells. *Journal of Biological Chemistry* **270:**9222-9226.

6. **Dattani MT,** Hindmarsh PC, Pringle PJ, Brook CGD, Marshall NJ (1995) The measurement of GH bioactivity in patient serum using an eluted stain assay (ESTA). *Journal of Clinical Endocrinology and Metabolism* **80:**2675-2683.

7. **Dattani MT,** Ealey PA, Pringle PJ, Hindmarsh PC, Brook CGD, Marshall NJ (1996) An investigation into the lability of the bioactivity of human growth hormone using the ESTA bioassay. *Hormone Research* **46 (2):**64-73.

8. **Dattani MT,** Winrow AP, Tuil'Pakov A, Pringle PJ, Hindmarsh PC, Brook CGD, Marshall NJ (1996) Evaluation of GH responses to pulsed GHRH administration using the MTT-ESTA bioassay. *European Journal of Endocrinology* **135:**87-95.

9. Lindley KJ, Dunne MJ, Kane C, Shepherd RM, Squires PE, James RFL, Johnson PRV, Eckhardt S, Wakeling E, **Dattani M,** Milla PJ, Aynsley-Green A (1996) Ionic control of □ cell function in nesidioblastosis. A possible therapeutic role for calcium channel blockade. *Archives of Disease in Childhood* **74:**373-378.

10. Wong NACS, Ahlquist JAO, Camacho-Hubner C, Goodwin CJ, **Dattani M,** Marshall NJ, Wass JAH (1997) Acromegaly or chronic renal failure: a diagnostic dilemma. *Clinical Endocrinology* **46:** 221-226.

11. **Dattani MT,** Martinez-Barbera JP, Thomas PQ, Brickman JB, Gupta R, Martensson I, Toresson H, Fox M, Wales JKH, Hindmarsh PC, Krauss S, Beddington RSP, Robinson ICAF (1998) Mutations in the homeobox gene *HESX1/Hesx1* associated with septo-optic dysplasia in human and mouse. *Nature Genetics* **19(2):** 125-133.

12. Mitchell H, **Dattani MT,** Nanduri V, Hindmarsh PC, Preece MA, Brook CGD (1999) Failure of IGF-1 and IGFBP-3 to diagnose growth hormone insufficiency. *Archives of Disease in Childhood* **80 (5):** 443-448.

13. Nuoffer JM, Fluck C, Deladoey J, Eble A, **Dattani MT**, Mullis PE (2000) Regulation of human GH receptor gene transcription by 20 and 22 kDa human hepatoma cell line. *Journal of Endocrinology* **165 (2):** 313-320.

14. Thomas PQ, **Dattani MT**, Brickman JM, McNay D, Warne G, Zacharin M, Cameron F, Hurst J, Woods K, Dunger D, Stanhope R, Forrest S, Robinson ICAF, Beddington RSP (2001) Heterozygous *HESX1* mutations associated with isolated congenital pituitary hypoplasia and septo-optic dysplasia. *Human Molecular Genetics* **10:**39-45.

15. Charmandari E, **Dattani MT**, Perry LA, Hindmarsh PC, Brook CGD (2001) Kinetics and effects of percutaneous administration of dihydrotestosterone. *Hormone Research* **56(5-6):** 177-181.

16. Brickman JM, Clements M, Tyrell R, McNay D, Woods K, Warner J, Stewart A, Beddington RSP, **Dattani MT** (2001) Molecular effects of novel mutations in

Hesx1/HESX1 associated with human pituitary disorders. *Development* **128:**5189-5199.

17. Mongan NP, Jaaskelainen J, Green K, Schwabe JW, Shimura N, **Dattani MT**, Hughes IA (2002) Two de novo mutations in the androgen receptor gene cause the complete androgen insensitivity syndrome in a pair of twins. *Journal of Clinical Endocrinology and Metabolism* **87 (3):** 1057-1061.

18. Karavanaki K, Divoli A, Dattani M, Briassoulis G, Theodorou V, Hatzara V, Avlonitis S (2002) Multiple pituitary hormone abnormalities, fever, behavioural problems, seizures and apnoeic spells in a 6 year old girl. *Hormones (Athens)* Apr-Jun **1(2):** 121-125.

19. Creighton S, Ransley P, Duffy P, Wilcox D, Mushtaq I, Cuckow P, Woodhouse C, Minto C, Stanhope R, Hughes I, **Dattani M**, Hindmarsh P, Brain C, Achermann J, Conway G, Liao LM, Barnicoat A, Perry L (2003) Regarding the consensus statement on 21-hydroxylase deficiency from the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology. *Journal of Clinical Endocrinology and Metabolism* **88(7):**3455.

20. Wakeling E, **Dattani MT**, Bloch-Zupan A, Winter RM, Holder SE (2003) Septo-optic dysplasia, subglottic stenosis and skeletal abnormalities: a case report. *Clinical Dysmorphology* **12 (2):** 105-109.

21. Roche EF, Charmandari E, **Dattani MT**, Hindmarsh PC (2003) Blood pressure in children and adolescents with congenital adrenal hyperplasia (21-hydroxylase deficiency): a preliminary report. *Clinical Endocrinology* **58 (5):**589-596.

22. Carvalho LR, Woods KS, Mendonca BB, Marcal N, Zamparini AL, Stifani S, Brickman JM, Arnhold IJP, **Dattani MT** (2003) A homozygous mutation (I26T) in the engrailed homology domain of HESX1 associated with evolving hypopituitarism due to impaired repressor-corepressor interaction. *Journal of Clinical Investigation* **112 (8):**1192-1201.

23. Jackson RS, Creemers JWM, Farooqi IS, Raffin-Sanson M, Varro A, Dockray GJ, Holst JJ, Brubaker PL, Corvol P, Polonsky KS, Ostrega D, Becker KL, Bertagna S, Hutton JC, White A, **Dattani MT**, Hussain K, Middleton S, Nicholl TM, Lindley KJ, O'Rahilly S (2003) Severe small intestinal dysfunction accompanies the complex endocrinopathy of human proprotein convertase 1 deficiency. *Journal of Clinical Investigation* **112 (10):**1550-1560.

24. Mehta A, Hindmarsh PC, Stanhope RG, Brain CE, Preece MA, **Dattani MT** (2003) Is the thyrotrophin-releasing hormone test necessary in the diagnosis of central hypothyroidism in children? *Journal of Clinical Endocrinology and Metabolism* **88 (12):** 5696-5703.

25. **Dattani MT,** Preece MA (2004) Growth Hormone Deficiency and related disorders: insights into aetiology, diagnosis and treatment. *Lancet* **363:** 1977-1987.

26. Mehta A**,** Viner R, Christie D, Newson T, **Dattani MT** (2004) An unusual case of an atypical eating disorder masquerading as a serious multi-systemic illness. *Acta Paediatrica* **93(5):**714-716**.**

27. Metherell LA, Savage MO, **Dattani MT**, Walker J, Clayton PE, Clark AJL (2004) Absence of *TPIT* (*Tbx19*) gene mutations in most patients with isolated ACTH deficiency. *European Journal of Endocrinology* **151(4):**463-465**.**

28. Mehta A, Hindmarsh PC, **Dattani MT** (2005) An update on the biochemical diagnosis of congenital ACTH insufficiency. *Clinical Endocrinology* **62(3):**307-314**.**

29. Turton J, Mehta AM, Raza J, Woods KS, Tiulpakov A, Cassar J, Chong K, Thomas PQ, Enice M, Ammini AC, Bouloux PM, Starzyk J, Hindmarsh PC, **Dattani MT** (2005) Mutations within the transcription factor PROP1 are rare in a cohort of patients with sporadic Combined Pituitary Hormone Deficiency (CPHD). *Clinical Endocrinology (Oxf).* **63(1):**10-8.

30. Hussain K, Cosgrove KE, Shepherd RM, Luharia A, Smith VV, Kassem J, Gregory JW, Sivaprasadarao A, Christensen HT, Jacobsen BB, Brusgard K, Glaser B, Maher EA, Lindley KJ, Hindmarsh PC, **Dattani MT**, Dunn M (2005) Hyperinsulinaemic hypoglycaemia in Beckwith-Wiedemann syndrome (BWS) due to defects in the function of pancreatic {beta}-cell ATP-sensitive K+ channels. *J Clin Endocrinology Metabolism* **90(7):**4376-82.

31. Woods KS, Cundall M, Turton J, Rizotti K, Mehta A, Palmer R, Wong J, Chong WK, Al-Zyoud M, El-Ali M, Otonkoski T, Martinez-Barbera JP, Thomas PQ, Robinson ICAF, Lovell-Badge R, Woodward KJ, **Dattani MT** (2005) Over- and under-dosage of SOX3 is associated with infundibular hypoplasia and hypopituitarism. *American Journal of Human Genetics* **76(5):**833-49.

32. Turton JPG, Reynaud R, Mehta A, Torpiano J, Saveanu A, Woods KS, Tiulpakov A, Hamilton J, Attard-Montalto S, Parascandalo R, Vella C, Clayton PE, Shalet S, Barton J, Brue T, **Dattani MT** (2005) Novel mutations within the *POU1F1* gene associated with variable Combined Pituitary Hormone Deficiency (CPHD) *J Clin Endocrinology Metabolism* **90(8):**4762-70.

33. Sami D, Taylor D, Saunders D, Thompson D, Russell-Eggitt I, Nischal K, Jeffery G, **Dattani M**, Clement R, Liasis A (2005) The spectrum of Achiasmia: congenitally reduced chiasmal decussation. *British Journal of Opthalmology* **89(10):**1311-17.

34. **Dattani MT** Growth Hormone Deficiency and Combined Pituitary Hormone Deficiency: Does the Genotype matter? (2005) *Clinical Endocrinology (Oxf).* **63(2):**121-30.

35. Mehta A, Hindmarsh PC, Stanhope RG, Turton JPG, Cole TJ, Preece MA, **Dattani MT** (2005) The role of Growth Hormone in determining Birth Size and early Postnatal Growth, using Congenital Growth Hormone Deficiency (GHD) as a model. *Clinical Endocrinology (Oxf).* **63(2):**223-31.

36. Hindmarsh PC, **Dattani MT** (2006) Use of growth hormone in children. *Nat Clin Pract Endocrinol Metab* **2(5):** 260-268.

37. Kelberman D, Rizzoti K, Avilion A, Bitner-Glindzicz M, Cianfarani S, Collins J, Chong WK, Kirk JM, Achermann JC, Ross R, Carmignac D, Lovell-Badge R, Robinson IC, **Dattani MT** (2006) Mutations within Sox2/SOX2 are associated with abnormalities in the hypothalamo-pituitary-gonadal axis in mice and humans. *J Clin Invest* **116 (9):**2442-2455.

38. Lin L, Conway GS, Hill NR, **Dattani MT**, Hindmarsh PC, Achermann JC (2006) A homozygous R262Q mutation in the gonadotropin-releasing hormone receptor (GNRHR) presenting as constitutional delay of growth and puberty with subsequent borderline oligospermia. *J Clin Endocrinology and Metabolism* **91(12):** 5117-5121.

39. Turton JPG, Buchanan CR, Robinson ICAF, Aylwin SJB, **Dattani MT** (2006) Evolution of gonadotropin deficiency in a patient with Type II autosomal dominant growth hormone deficiency. *European Journal of Endocrinology* **155(6)**: 793-799

40. Kelberman D, **Dattani MT** (2006) The role of transcription factors implicated in anterior pituitary development in the aetiology of congenital hypopituitarism. *Annals of Medicine* 38 (8): 560-577.

41. Lin L, Hindmarsh PC, Metherell LA, Alzyoud M, Al-Ali M, Brain CE, Clark AJL, **Dattani MT**, Achermann JC (2007) Severe loss-of-function mutations in the adrenocorticotropin receptor (ACTHR, MC2R) can be found inpatients with salt-losing adrenal hypoplasia. *Clinical Endocrinology* **66(2):** 205-210.

42. McNay DEG, Turton JP, Kelberman D, Woods KS, Brauner R, Papadimitriou A, Keller E, Keller A, Hauf N, Krude H, Shalet SM, **Dattani MT** (2007) *HESX1* mutations are an uncommon cause of septo-optic dysplasia and hypopituitarism. *J Clin Endocrinology and Metabolism* **92 (2):** 691-697.

43. Lin L, Ercan O, Raza J, Burren CP, Creighton SM, Auchus RJ, **Dattani MT**, Achermann JC (2007) Variable phenotypes associated with aromatase (CYP19) insufficiency in humans. *J Clin Endocrinology and Metabolism* **92 (3):** 982-990.

44. Lin L, Philibert P, Ferraz-de-Souza B, Kelberman D, Homfray T, Albanese A, Molini V, Sebire NJ, Einaudi S, Conway GS, Hughes IA, Jameson JA, Sultan C, **Dattani MT**, Achermann JA (2007) Heterozygous missense mutations in steroidogenic Factor 1 (SF1/Ad4BP, NR5A1) are associated with 46 XY disorders of sex development with normal adrenal function. *J Clin Endocrinology and Metabolism* **92 (3):** 991-999.

45. Farooqi IS, Wangensteen T, Collins S, Kimber W, Matarese G, Keogh JM, Lank E, Bottomley B, Lopez-Fernandez J, Ferraz-Amaro I, **Dattani MT**, Ercan O, Myhre AG, Retterstol L, Stanhope R, Edge JA, McKenzie S, Lessan N, Ghodsi M, De Rosa V, Perna F, Fontana S, Barroso I, Undlien DE, O'Rahilly S (2007) Clinical and molecular genetic spectrum of congenital deficiency of the leptin receptor. *N Engl J Med* **356 (3):** 237-247.

46. Andoniadou CL, Signore M, Sajedi E, Gaston-Massuet C, Kelberman D, Burns AJ, Itasaki N, **Dattani M**, Martinez-Barbera JP (2007) Lack of the murine homeobox gene Hesx1 leads to posterior transformation of the anterior forebrain. *Development* 134 (8): 1499-1508.

47. Vallette-Kasic S, Couture S, Balsalobre A, Gauthier Y, Metherell L, **Dattani M**, Drouin J (2007) The TPIT gene mutation M86R associated with isolated ACTH deficiency interferes with protein: protein interactions. *J Clin Endocrinol Metab* **92(10):** 3991-3999.

48. Petkovic V, Lochmatter D, Turton J, Clayton PE, Trainer PJ, **Dattani MT**, Eble A, Robinson IC, Fluck C, Mullis PE (2007) Exon splice enhancer mutation (GH-E32A) causes autosomal dominant GH deficiency. *J Clin Endocrinol Metab* **92(11):** 4427-4435.

49. Sajedi E, Gaston-Massuet C, Andoniadou CL, Signore M, Hurd PJ, **Dattani M**, Martinez-Barbera JP (2008) DNMT1 interacts with the developmental transcriptional repressor HESX1. *Biochim Biophys Acta* **1783 (1):** 131-143.

50. Raz B, Janner M, Petkovic V, Lochmatter D, Eble A, **Dattani MT**, Hindmarsh PC, Fluck CE, Mullis PE (2008) Influence of growth hormone receptor d3 and full length isoforms on growth hormone response and final height in patients with severe growth hormone deficiency. *J Clin Endocrinol Metab* **93(3):** 974-980.

51. Fauquier T, Rizzoti K, **Dattani M**, Lovell-Badge R, Robinson ICAF (2008) A *Sox2*-expressing progenitor cell generates all the major cell types in the adult mouse pituitary gland. *Proceedings of the National Academy of Sciences USA* **105(8):** 2907-2912.

52. Al-Gazali L, Hertecant J, Algawi K, El Teraifi H, **Dattani MT** (2008) A new autosomal recessive syndrome of ocular colobomas, icthyosis, brain malformations and endocrine abnormalities in an inbred Emirati family. *American J of Medical Genetics* **146 (7):**813-819.

53. Kelberman D, de Castro SCP, Huang S, Crolla JA, Palmer R, Gregory JW, Taylor D, Cavallo L, Faienza MF, Fischetto R, Achermann JC, Martinez-Barbera JP, Rizzoti K, Lovell-Badge R, Robinson ICAF, Gerrelli D, **Dattani MT** (2008) *SOX*2 plays a critical role in the development of the pituitary, forebrain and eye during human embryonic development. *J Clin Endocrinol Metab* **93(5):** 1865-1873.

54. Rajab A, Kelberman D, de Castro SC, Biebermann H, Shaikh H, Pearce K, Hall CM, Shaikh G, Gerrelli D, Grueters A, Krude H, **Dattani MT** (2008) Novel mutations in *LHX3* are associated with hypopituitarism and sensorineural hearing loss. *Human Molecular Genetics* **17(14):** 2150-9.

55. Elder CJ, Barton JS, Brook CG, Preece MA, **Dattani MT**, Hindmarsh PC (2008) A randomized study of the effect of two doses of biosynthetic human growth hormone on final height in children with familial short stature. *Hormone Research* **70 (2)**: 89-92.

56. Coelho R, Brook CG, Preece MA, Stanhope RG, **Dattani MT**, Hindmarsh PC (2008) A randomized study of two doses of biosynthetic human growth hormone on final height of pubertal children with growth hormone deficiency. *Hormone Research* **70(2):** 85-88.

57. Chen R, Hussain K, Al-Ali M, **Dattani MT**, Hindmarsh P, Jones PM, Marsh P (2008) Neonatal and late-onset diabetes mellitus caused by failure of pancreatic development: report of 4 more cases and a review of the literature. *Pediatrics* **121(6):** e1541-7.

58. Deeb A, Jaaskelainen J, **Dattani M**, Whitaker HC, Costigan C, Hughes IA (2008) A novel mutation in the human androgen receptor suggests a regulatory role for the hinge region in amino-terminal and carboxy-terminal interactions. *Journal of Clinical Endocrinology and Metabolism* **93(10**): 3691-3696.

59. Sajedi E, Gaston-Massuet C, Signore M, Andoniadou CL, Kelberman D, Castro S, Etchevers HC, Gerrelli D, **Dattani MT**, Martinez-Barbera JP (2008) Analysis of mouse models carrying the I26T and R160C substitutions in the transcriptional repressor HESX1 as models for septo-optic dysplasia and hypopituitarism. *Disease Models and Mechanisms* **1(4-5):** 241-254.

60. Kelberman D, Turton J, Woods K, Mehta A, Al-Khawari M, Greening J, Swift P, Otonkoski T, Rhodes S, **Dattani M** (2009) Molecular analysis of novel *PROP1* mutations associated with Combined Pituitary Hormone Deficiency. *Clinical Endocrinology* **70(1):** 96-103.

61. Segal TY, Mehta A, Anazodu A, Hindmarsh PC, **Dattani MT** (2009) Role of GnRH and HCG stimulation tests in differentiating patients with hypogonadotropic hypogonadism [HH] from those with Constitutional Delay of Growth and Puberty [CDGP] *Journal of Clinical Endocrinology and Metabolism* **94(3):** 780-785.

62. Alatzoglou KS, Kelberman D, **Dattani MT** (2009) Role of Sox/SOX proteins in normal pituitary development. *Journal of Endocrinology* 200(3): 245-258.

63. de Smith AJ, Purmann C, Walters RG, Ellis RJ, Holder SE, Van Haelst MM, Brady AF, Fairbrother UL, **Dattani M**, Keogh JM, Henning E, Yeo GS, O'Rahilly S, Froguel P, Farooqi IS, Blakemore AI (2009) A deletion of the HBII-85 class of small nucleolar RNAs (snoRNAs) is associated with hyperphagia, obesity and hypogonadism. *Human Molecular Genetics* **18(17):** 3257-3265.

64. Padidela R, Kelberman D, Press M, Al-Khawari M, Hindmarsh PC, **Dattani MT** (2009) Mutation in the TBCE gene is associated with Hypoparathyroidism-Retardation-Dysmorphism syndrome featuring pituitary hormone deficiencies and hypoplasia of the anterior pituitary and the corpus callosum. *J Clin Endocrinol Metab* **94(8):** 2686-2691.

65. Mehta A, Hindmarsh PC, Mehta H, Turton JP, Russell-Eggitt I, Taylor D, Chong WK, **Dattani MT** (2009) Congenital hypopituitarism: clinical, molecular and neuroradiological correlates. *Clinical Endocrinology* **70(1):** 96-103.

66. Alatzoglou KS, Turton J, Kelberman D, Clayton PE, Mehta A, Buchanan C, Aylwin S, Crowne EC, Christesen HT, Hertel T, Trainer P, Savage MO, Raza J, Banerjee K, Sinha S, Ten S, Mushtaq T, Brauner R, Cheetham T, Hindmarsh PC, Mullis PE, **Dattani MT (**2009) Expanding the spectrum of mutations in *GH1* and *GHRHR*: Genetic screening in a large cohort of patients with Congenital Isolated Growth Hormone Deficiency (IGHD). *J Clin Endocrinol Metab* **94(9):** 3191-3199.

67. Alatzoglou KS, Hindmarsh PC, Brain C, Torpiano J, **Dattani MT** (2009) Acanthosis Nigricans and Insulin Sensitivity in patients with Achondroplasia and Hypochondroplasia due to FGFR3 mutations. *J Clin Endocrinol Metab* **94(10):** 3959-3963.

68. Webb E, **Dattani MT** (2009) Septo-optic dysplasia. *European Journal of Human Genetics* **18(4)**:393-397.

69. Kelberman D, Rizzoti K, Lovell-Badge R, Robinson ICAF, **Dattani MT** (2009) Genetic Regulation of pituitary gland development in human and mouse. *Endocrine Reviews* **30(7):** 790-829.

70. Gaston-Massuet C, Kelberman D, **Dattani M**, Martinez-Barbera JP (2009) Absence of SIX3 mutations in patients with congenital hypopituitarism. *Am J Med Genet A* **149A (12):** 2874-6.

71. Howard S, Raine J, **Dattani MT** (2009) Corneal rupture in a child with Down syndrome and hyperthyroidism. *BMJ Case Reports* pii: bcr08.2008.0842. doi: 10.1136/bcr.08.2008.0842. Epub 2009 Mar 5.

72. Petkovic V, Godi M, Pandey AV, Lochmatter D, Buchanan CR, **Dattani MT**, Eble A, Fluck CE, Mullis PE (2010) Growth hormone (GH) deficiency Type II: a novel GH-1 gene mutation (GH-R178H) affecting secretion and action. *J Clin Endocrinol Metab* 95(2): 731-9.

73. Peters C, Langham S, Mullis PE, **Dattani MT** (2010) Use of combined liothyronine and thyroxine therapy for consumptive hypothyroidism associated with hepatic haemangiomas in infancy. *Hormone Research Paediatrics* 74(2):149-152.

74. Igreja S, Chahal HS, King P, Bolger GB, Srirangalingam U, Guasti L, Chapple JP, Trivellin G, Gueorguiev M, Guegan K, Stals K, Khoo B, Kumar AV, Ellard S, Grossman AB, Korbonits M; International FIPA Consortium (2010) Characterization of aryl hydrocarbon receptor interacting protein (AIP) mutations in familial isolated pituitary adenoma families. *Hum Mutation* 31(8):950-960.

75. O'Riordan SM, **Dattani MT**, Hindmarsh PC (2010) Cystic-fibrosis related diabetes in childhood. *Hormone Research Paediatrics* 73(1):15-24.

76. Sharma G, Muller D, O'Riordan S, Bryan S, Hindmarsh P, **Dattani M**, Mills K (2010) A novel method for the direct measurement of urinary conjugated metabolites of alpha-tocopherol and its use in diabetes. *Mol Nutr Food Research* 54(5):599-600.

77. Gozzi T, Fluck C, L'allemand D, **Dattani MT**, Hindmarsh PC, Mullis PE (2010) Do centimetres matter? Self-reported versus estimated height measurements in parents? *Acta Paedi*atr 99(4): 569-574.

78. Webb EA, O'Reilly MA, Orgill J, Dale N, Salt A, Gringras P, **Dattani MT** (2010) Rest-activity disturbances in children with septo-optic dysplasia characterized by actigraphy and 24 hour plasma melatonin profiles. *J Clin Endocrinol Metab* 95(10): E198-203.

79. Bockenhauer D, Van't Hoff W, **Dattani M**, Lehnhardt A, Subtirelu M, Hildebrandt F, Bichet DG (2010) Secondary nephrogenic diabetes insipidus as a complication of inherited renal diseases. *Nephron Physiology* 116 (4): 23-29.

80. Morkane C, Raptis D, Brain C, Spoudeas H, Dattani M, Hindmarsch P, Piero A, DeCoppi P, Kurzawinski TR (2010) Prophylactic thyroidectomy in children with Multiple Endocrine Neoplasia type 2. *Regul Pept* 164(1):9.

81. Schoenmakers E, Agostini M, Mitchell C, Schoenmakers N, Papp L, Rajanayagam O, Padidela R, Robertson V, Ceron-Gutierrez L, Doffinger R, Prevosto C, Luan J, Montano S, Lu J, Castanet M, Clemons N, Groeneveld M, Castets P, Karbaschi M, Aitken S, Dixon A, Williams J, Campi I, Blount M, Burton H, Muntoni F, O'Donovan D, Dean A, Warren A, Brierley C, Baguley D, Guicheney P, Fitzgerald R, Coles A, Gaston H, Todd P, Holmgren A, Khanna K, Cooke M, Semple R, Halsall D, Wareham N, Schwabe J, Grasso L, Beck-Peccoz P, Ogunko A, **Dattani M**, Gurnell M, Chatterjee K (2010) Mutations in the selenocysteine insertion sequence binding protein 2 gene lead to a multisystem selenoprotein deficiency disorder in humans. *J Clin Invest* 120 (12): 4220-35.

82. Alatzoglou S, **Dattani MT** (2010) Genetic causes and treatment of isolated growth hormone deficiency—an update. *Nature Reviews in Endocrinology* 6(10): 562-576.

83. Ferraz-de-Souza B, Lin L, Shah S, Jina M, Hubank M, **Dattani MT**, Achermann JC (2011) CHIP-on chip analysis reveals angiopoietin 2 (Ang2, ANGPT2) as a novel target of steroidogenic factor-1 (SF-1, NR5A1) in the human adrenal gland. *FASEB J* 25(4): 1166-75.

84. Idkowiak J, O'Riordan S, Reisch N, Malunowicz EM, Collins F, Kerstens MN, Köhler B, Graul-Neumann LM, Szarras-Czapnik M, **Dattani M**, Silink M, Shackleton

CH, Maiter D, Krone N, Arlt W (2011) Pubertal presentation in seven patients with congenital adrenal hyperplasia due to P450 Oxidoreductase deficiency. *J Clin Endocrinol Metab* 96(3): E453-62.

85. Ferraz-de-Souza B, Hudson-Davies RE, Lin L, Parnaik R, Hubank M, **Dattani MT**, Achermann JC (2011) Sterol O-acyltransferase 1 (SOAT1, ACAT) is a novel target of steroidogenic factor-1 (SF-1, NR5A1, Ad4BP) in the human adrenal. *J Clin Endocrinol Metab* 96(4): E663-8.

86. Alatzoglou KS, Kelberman D, Cowell CT, Palmer R. Arnhold IJ, Melo ME, Schnabel D, Grueters A, **Dattani MT** (2011) Increased transactivation associated with SOX3 polyalanine tract deletion in a patient with hypopituitarism. *Journal of Clinical Endocrinology and Metabolism* 96(4): E685-690.

87. Raffan E, Soos MA, Rocha N, Tuthill A, Thomsen AR, Hyden CS, Gregory JW, Hindmarsh P, **Dattani M**, Cochran E, Al Kaabi J, Gorden P, Barroso I, Morling N, O'Rahilly S, Semple RK (2011) Founder effect in the Horn of Africa for an insulin receptor mutation that may impair receptor recycling. *Diabetologia* 54(5): 1057-65.

88. Gault EJ, Perry RJ, Cole TJ, Casey S, Paterson WF, Hindmarsh PC, Betts P, Dunger DB, Donaldson MD; British Society for Paediatric Endocrinology and Diabetes. (2011) Effect of oxandrolone and timing of pubertal induction on final height in Turner syndrome: randomized, double blind, placebo-controlled trial. *British Medical Journal* 342 d.1980 doi: 10.1136/bmj.d1980 .

89. Gaston-Massuet C, Andoniadou CL, Signore M, Jayakody SA, Charolidi N, Kyeyune R, Vernay B, Jacques TS, Taketo MM, Le Tissier P, **Dattani MT**, Martinez-Barbera JP (2011) Increased Wingless (Wnt) signaling in pituitary progenitor/stem cells gives rise to pituitary tumors in mice and humans. *Proc Natl Acad Sci USA* 108(28): 11482-7.

90. Franca MM, Jorge AA, Alatzoglou KS, Carvalho LR, Mendonca BB, Audi L, Carrascosa A, **Dattani MT**, Arnhold IJ (2011) Absence of GH-releasing hormone (GHRH) mutations in selected patients with isolated GH deficiency. *Journal of Clinical Endocrinology and Metabolism* 96 (9): E1457-60.

91. McCabe MJ, Gaston-Massuet C, Tziaferi V, Gregory LC, Alatzoglou KS, Signore M, Puelles E, Gerrelli D, Farooqi IS, Raza J, Walker J, Kavanaugh SI, Psai PS, Pitteloud N, Martinez-Barbera JP, **Dattani MT** (2011) Novel *FGF8* mutations associated with recessive holoprosencephaly, craniofacial defects, and hypothalamo-pituitary defects. *Journal of Clinical Endocrinology and Metabolism* 96(10): E1709-18.

92. Alatzoglou KS, Andoniadou CL, Kelberman D, Buchanan C, Crolla J, Arriazu MC, Roubicek M, Martinez-Barbera JP, **Dattani MT** (2011) SOX2 haploinsufficiency is associated with slow-progressing Hypothalamus-pituitary tumours. *Human Mutation* 32(12): 1376-80.

93. Bochukova E, Schoenmakers N, Agostini N, Schoenmakers E, Rajanayagam O, Keogh JM, Henning E, Reinemund J, Gevers E, Sarri M, Downes K, Offiah A, Albanese A, Halsall D, Schwabe J, Bain M, Lindley K, Muntoni F, Vargha Khadem F, **Dattani M**, Farooqi S, Gurnell M, and Chatterjee K (2012) A mutation in the thyroid hormone receptor alpha gene. *New England Journal of Medicine* 366(3): 243-9.

94. Webb EA, O'Reilly MA, Clayden JD, Seunarine KK, Chong WK, Dale N, Salt A, Clark CA, **Dattani MT** (2012) Effect of growth hormone deficiency on brain structure, motor function and cognition. *Brain* **135 (Part 1):** 216-227.

95. Turton JP, Strom M, Langham S, **Dattani MT**, Le Tissier P (2011) Two novel mutations in the *POU1F1* gene generate null alleles through different mechanisms leading to Combined Pituitary Hormone Deficiency. *Clinical Endocrinology* **76(3):** 387-393.

96. Miletta MC, Scheidegger UA, Giordano M, Bozzola M, Pagani S, Bona G, **Dattani M**, Hindmarsh PC, Petkovic V, Oser-Meier M, Flück CE, Mullis PE (2011) Association of the (CA)(n) repeat polymorphism of Insulin-like-Growth Factor-I (IGF-I) and -202 A/C IGF Binding Protein-3 (IGFBP3) Promoter Polymorphism with Adult height in patients with severe growth hormone deficiency. *Clinical Endocrinology* doi: 10.1111/j.1365-2265.2011.04267.

97. Krone N, Reisch N, Idkowiak J, Dhir V, Ivison HE, Hughes BA, Rose IT, O'Neil DM, Vijzelaar R, Smith MJ, Macdonald F, Cole TR, Adolphs N, Barton JS, Blair EM, Braddock SR, Collins F, Cragun DL, **Dattani M**T, Day R, Dougan S, Feist M, Gottschalk ME, Gregory JW, Haim M, Harrison R, Olney AH, Hauffa BP, Hindmarsh PC, Hopkin RJ, Jira PE, Kempers M, Kerstens MN, Khalifa MM, Köhler B, Maiter D, Nielsen S, O'Riordan SM, Roth CL, Shane KP, Silink M, Stikkelbroeck NM, Sweeney E, Szarras-Czapnik M, Waterson JR, Williamson L, Hartmann MF, Taylor NF, Wudy SA, Malunowicz EM, Shackleton CH, Arlt W (2012) Genotype-Phenotype Analysis in Congenital Adrenal Hyperplasia due to P450 Oxidoreductase Deficiency *Journal of Clinical Endocrinology and Metabolism* 97(2):E257-267.

98. El-Khairi R, Parnaik R, Duncan AJ, Lin L, Gerrelli D, **Dattani MT**, Conway GS, Achermann JC (2012) Analysis of LIN28A in early human ovary development and as a candidate gene for primary ovarian insufficiency. *Molecular and Cellular Endocrinology* 351(2): 264-8.

99. Raivio T, Avbelj M, McCabe MJ, Romero CJ, Dwyer AA, Tommiska J, Sykiotis GP, Gregory LC, Diaczok D, Tziaferi V, Elting MW, Padidela R, Plummer L, Martin C, Feng B, Zhang C, Zhou QY, Chen H, Mohammadi M, Quinton R, Sidis Y, Radovick S, **Dattani MT**, Pitteloud N (2012) Genetic overlap in Kallmann syndrome, Combined Pituitary Hormone Deficiency, and Septo-optic Dysplasia. *Journal of Clinical Endocrinology and Metabolism* 97(4): E694-9.

100. Andoniadou CL, Gaston-Massuet C, Reddy R, Schneider RP, Blasco MA, Le Tissier P, Jacques TS, Pevny LH, **Dattani MT**, Martinez-Barbera JP (2012) Identification of novel pathways involved in the pathogenesis of human adamantinomatous craniopharyngioma. *Acta Neuropathologica* 124 (2): 259-271.

101. Bawazir WM, Gevers EF, Flatt JF, Ang AL, Jacobs B, Oren C, Grunewald S, **Dattani M**, Bruce LJ, Stewart GW (2012) An infant with pseudohyperkalemia, hemolysis and seizures:Cation-leaky GLUT1-deficiency syndrome due to a SLC2A1 mutation. *Journal of Clinical Endocrinology and Metabolism* 97(6): E987-993.

102. Prasov L, Masud T, Khaliq S, Mehdi SQ, Abid A, Oliver ER, Silva ED, Lewanda A, Brodsky MC, Borchert M, Kelberman D, Sowden JC, **Dattani MT**, Glaser T (2012) ATOH7 mutations cause autosomal recessive persistent hyperplasia of the primary vitreous. *Human Molecular Genetics* **21(16):** 3681-3694.

103. Peters C, Hill N, **Dattani MT**, Matthews D, Hindmarsh PC (2012) Deconvolution analysis of 24h serum cortisol profiles informs the amount and distribution of hydrocortisone replacement therapy. *Clinical Endocrinology* **78(3):** 347-351.

104. Jayakody S, Andoniadou CL, Gaston-Massuet C, Signore M, Cariboni A, Bouloux PM, Le Tissier P, Pevny L, **Dattani MT**, Martinez-Barbera JP (2012) Severe pituitary hypoplasia and decreased GnRH neurogenesis in a mouse model of *SOX2*-associated hypopituitarism in humans. *Journal of Clinical Investigation* **122(10):** 3635-46.

105. Sun Y, Bak B, Schoenmakers N, van Trotsenburg ASP, Oostdijk W, Voshol P, Cambridge E, White JK, le Tissier P, Mousavy Gharavy SN, Martinez-Barbera JP, Stokvis-Brantsma WH, Vulsma T, Kempers MJ, Persani L, Campi I, Bonomi M, Beck-Peccoz P, Zhu H, Davis TME, Hokken-Koelega ACS, Del Blanco DG, Rangasami JJ , Ruivenkamp CAL, Laros JFL, Kriek M, Kant SG, Bosch CAJ, Biermasz NR, Appelman-Dijkstra NM, Corssmit EP, Hovens GCJ, Pereira AM, den Dunnen JT, Breuning MH, Hennekam RC, Chatterjee KK*, **Dattani MT***, Wit JM*, Bernard DJ* (*Co-Senior Authors) (2012) Loss-of-function mutations in IGSF1 cause a novel X-linked syndrome of central hypothyroidism and testicular enlargement *Nature Genetics* **44(12):** 1375-1381.

106. Sharma G, Muller DP, O'Riordan SM, Bryan S, **Dattani MT**, Hindmarsh PC, Mills K (2013) Urinary conjugated α-tocopheronolactone-a biomarker of oxidative stress in children with type 1 diabetes. *Free Radic Biol Med* **55:**54-62.

107. McCabe MJ, Gaston-Massuet C, Gregory LC, Alatzoglou KS, Tziaferi V, Sbai O, Rondard P, Masumoto KH, Nagano M, Shigeyoshi Y, Pfeifer M, Hulse T, Buchanan CR, Pitteloud N, Martinez-Barbera JP, **Dattani M** (2013) Variations in PROKR2, but not PROK2, are associated with hypopituitarism and septo-optic dysplasia. *J Clin Endocrinol Metab* 98(3): E547-557.

108. Gregory LC, Gevers EF, Baker J, Kasia T, Chong K, Josifova DJ, Caimari M, Bilan F, McCabe MJ, **Dattani MT** (2013) Structural pituitary abnormalities associated with CHARGE Syndrome. *J Clin Endocrinol Metab* 98(4): E737-743.

109. Webb EA, O'Reilly MA, Clayden JD, Seunarine KK, Dale N, Salt A, Clark CA, **Dattani MT** (2013) Reduced ventral cingulum integrity and increased behavioral problems in children with isolated optic nerve hypoplasia and mild to moderate or no visual impairment. *PLOS One* **8(3):** e59048.

110. Ramachandrappa S, Raimondo A, Cali AM, Keogh JM, Henning E, Saeed S, Thompson A, Garg S, Bochukova EG, Brage S, Trowse V, Wheeler E, Sullivan AE, **Dattani M**, Clayton PE, Datta V, Bruning JB, Wareham NJ, O'Rahilly S, Peet DJ, Barroso I, Whitelaw ML, Farooqi IS (2013) Rare variants in Single-minded 1 (SIM1) are associated with severe obesity *Journal of Clinical Investigation* **123 (7):** 3042-3050.

111. Subbarayan A, **Dattani MT**, Peters CJ, Hindmarsh PC (2013) Cardiovascular risk factors in children and adolescents with congenital adrenal hyperplasia due to 21-hydroxylase deficiency. *Clinical Endocrinology* **80(4):**471-7.

112. Moran C, Schoenmakers N, Agostini M, Schoenmakers E, Offiah A, Kydd A, Kahaly G, Mohr-Kahaly S, Rajanayagam O, Lyons G, Wareham N, Halsall D, **Dattani M**, Hughes S, Gurnell M, Park SM, Chatterjee K (2013) An adult female with resistance to thyroid hormone mediated by defective thyroid hormone receptor α. Journal of Clinical Endocrinology and Metabolism **98 (11):** 4254-61.

113. Kido Y, Gordon CT, Sakazume S, Ben Bdira E, **Dattani M**, Wilson LC, Lyonnet S, Murakami N, Cunningham ML, Amiel J, Nagai T (2013) Further characterization of atypical features in auriculocondylar syndrome caused by recessive PLCB4 mutations. American Journal of Medical Genetics A **161 (9):** 2339-2346.

114. Webb EA, AlMutair A, Kelberman D, Bacchelli C, Chanudet E, Lescai F, Andoniadou CL, Banyan A, Swaid A, Alrifai MT, Alahmesh MA, Balwi M, Mousavy-Gharavy SN, Lukovic B, Burke D, McCabe MJ, Kasia T, Kleta R, Stupka E, Beales PL, Thompson DA, Chong WK, Alkuraya FS, Martinez-Barbera JP, Sowden JC, **Dattani MT** (2013) *ARNT2* mutation causes hypopituitarism, postnatal microcephaly, visual and renal anomalies. *Brain* **136 (Pt10):** 3096-3105.

115. Andoniadou C, Matsushima D, Mousavy-Gharavy SN, Signore M, Mackintosh AI, Schaeffer M, Gaston-Massuet C, Mollard P, Jacques TS, Le Tissier P, **Dattani MT**, Pevny L, Martinez-Barbera JP (2013) The Sox2+ population of the adult murine pituitary includes progenitor/stem cells with tumour-inducing potential. *Cell Stem Cell* **13(4):** 433-445.

116. Joustra SD, Schoenmakers N, Persani L, Campi I, Bonomi M, Radetti G, Beck-Peccoz P, Zhu H, Davis TM, Sun Y, Corssmit EP, Appelman-Dijkstra NM, Heinen CA, Pereira AM, Varewijck AJ, Janssen JA, Endert E, Hennekam RC, Lombardi MP, Mannens MM, Bak B, Bernard DJ, Breuning MH, Chatterjee K, Dattani MT, Oostdijk W, Biermasz NR, Wit JM, van Trotsenburg AS (2013) The IGSF1 deficiency syndrome: Characteristics of male and female patients. *Journal of Clinical Endocrinology and Metabolism* **98(12):** 4942-52.

117. Alatzoglou KS, Webb E, LeTissier P, **Dattani MT** (2014) Isolated Growth Hormone Deficiency in childhood and adolescence: an update. *Endocrine Reviews* **35 (3):** 376-432.

118. Alatzoglou K, Azriyanti A, Rogers N, Ryan F, Curry N, Noakes C, Bignell P, Hall G, Littooij A, Saunders D, Thomas P, Stewart H, **Dattani M** (2014) Sox3 deletion in mouse and human is associated with persistence of the craniopharyngeal canal. *Journal of Clinical Endocrinology and Metabolism* **99(12):** E2702-8.

119. Gregory LC, Gaston-Massuet C, Andoniadou CL, Carreno G, Webb EA, Kelberman D, McCabe MJ, Panagiotakopoulos L, Saldanha JW, Spoudeas HA, Torpiano J, Rossi M, Raine J, Canham N, Martinez-Barbera JP, **Dattani MT** (2014) The role of the Sonic Hedgehog signalling pathway in patients with midline defects and congenital hypopituitarism. *Clinical Endocrinology* **82(5):**728-38.

120. Formosa N, Torpiano J, Allgrove J, **Dattani MT** (2014) Anticonvulsant Treatment Associated with Intractable Hypocalcaemia in a Female Child with Hypoparathyroidism *Hormone Research in Paediatrics* **83(1):** 62-66.

121. Sinha CK, Decoppi P, Pierro A, Brain C, Hindmarsh P, Butler G, **Dattani M**, Spoudeas H, Kurzawinski TR (2014) Thyroid Surgery in Children: Clinical Outcomes. *European Journal of Paediatric Surgery* **25(5):**425-9.

122. Senniappan S, Wood D, Hakeem V, Stoneham S, Freeman A, **Dattani M** (2014) Gonadotrophin-independent precocious puberty associated with later diagnosis of testicular embryonal carcinoma. *Hormone Research in Paediatrics* **82(4):**272-7.

123. Simpkin A, Cochran E, Cameron F, **Dattani M**, de Bock M, Dunger DB, Forsander G, Guran T, Harris J, Isaac I, Hussain K, Kleta R, Peters C, Tasic V, Williams R, Yap Kok Peng F, O'Rahilly S, Gorden P, Semple RK, Bockenhauer D (2014) Insulin receptor and the kidney: nephrocalcinosis in patients with recessive *INSR* mutations. *Nephron Physiolo*gy (*in press*).

124. Alagaratnam S, Brain C, Spoudeas H, **Dattani MT**, Hindmarsh P, Allgrove J, Van't Hoff W, Kurzawinski TR (2014) Surgical treatment of children with

hyperparathyroidism: Single centre experience. *Journal of Paediatric Surgery* **49(11):** 1539-43.

125. Hufnagel RB, Arno G, Hein ND, Hersheson J, Prasad M, Anderson Y, Krueger LA, Gregory LC, Stoetzel C, Jaworek TJ, Hull S, Li A, Plagnol V, Willen CM, Morgan TM, Prows CA, Hegde RS, Riazuddin S, Grabowski GA, Richardson RJ, Dieterich K, Huang T, Revesz T, Martinez-Barbera JP, Sisk RA, Jefferies C, Houlden H, **Dattani MT**, Fink JK, Dollfus H, Moore AT, Ahmed ZM (2014) Neuropathy target esterase impairments cause Oliver-McFarlane and Laurence-Moon syndromes. *J Med Genet* **52(2):**85-94.

126. Amores-Hernandez I, Besser RE, **Dattani MT**, Inwald DP (2015) A 12-year-old child with fever, headache, shock, and coma. *Lancet* **385:** 9967.

127. Gregory LC, Humayun KN, Turton JPG, McCabe MJ, Rhodes SJ, **Dattani MT** (2015) Novel lethal form of congenital hypopituitarism associated with the first recessive *LHX4* mutation. *Journal of Clinical Endocrinology and Metabolism* **100(6):** 2158-2164.

128. Wiskin AE, Katugampola H, **Dattani MT**, Mutalib M (2015) Water Toxicity During Antroduodenal Manometry. *Journal of Clinical Gastroenterology* **49(8):** 715-6.

129. Boehm U, Bouloux PM, **Dattani M**, de Roux N, Dodé C, Dunkel L, Dwyer AA, Giacobini P, Hardelin JP, Juul A, Maghnie M, Pitteloud N, Prevot V, Raivio T, Tena-Sempere M, Quinton R, Young J (2015) Congenital hypogonadotrophic hypogonadism: pathogenesis, diagnosis, and treatment. *Nature Reviews Endocrinology Consensus Statement* **11(9):**547-64.

130. Waelchi R, Williams J, Cole T, **Dattani M**, Hindmarsh P, Kennedy H, Martinez A, Khan S, Semple R, White A, Sebire N, Healy E, Moore G, Kinsler V (2015) Growth and hormonal profiling in children with congenital melanocytic naevi. *British Journal of Dermatology* **173(6):**1471-8.

131. McCabe M, Youli H, Gregory LC, Gaston-Massuet C, Alatzoglou KS, Saldanha JW, Gualtieri A, Thankamony A, Hughes I, Townshend S, Martinez-Barbera JP, Bouloux PM, **Dattani MT** (2015) Novel *in vitro* functional assay used for the assessment of *KAL1* variants in three females with septo-optic dysplasia (SOD). *Molecular and Cellular Endocrinology* **417:**63-72.

132. Schoenmakers N, Alatzoglou KS, Chatterjee VK, **Dattani MT** (2015) Recent advances in central congenital hypothyroidism. J Endocrinol **227 (3):** R51-71.

133. Gaston-Massuet C, McCabe MJ, Scagliotti V, Young RM, Carreno G, Gregory LC, Jayakody SA, Pozzi S, Gualtieri A, Basu B, Koniordou M, Wu CI, Bancalari RE, Rahikkala E, Veijola R, Lopponen T, Graziola F, Turton J, Signore M, Mousavy Gharavy SN, Charolidi N, Sokol SY, Andoniadou CL, Wilson SW, Merrill BJ, ***Dattani MT, *Martinez-Barbera JP** (2016) **(***Co-Senior Authors) Transcription factor 7-like 1 is involved in hypothalamo-pituitary axis development in mice and humans. *Proc Natl Acad Sci USA* **13(5):**E548-57.

134. Joustra SD, Heinen CA, Schoenmakers N, Bonomi M, Ballieux BE, Turgeon MO, Bernard DJ, Fliers E, van Trotsenburg AS, Losekoot M, Persani L, Wit JM, Biermasz NR, Pereira AM, Oostdijk W, Aisenberg J, van den Akker EL, Bergadá I, Bocca G, Braslavsky D, Callewaert B, Cummings EA, Cuppen MP, **Dattani M**, Domené HM, van der Heyden JC, van Hulle S, Jacobs MA, Links TP, Lunshof L, Mul D, Neijens FS, Pedro HF, Salerno M, de Schepper J, Voorhoeve PG, Zidell AS,

van der Zwaag PA, Zwaveling-Soonawala N; IGSF1 clinical care group. (2016) IGSF1 deficiency: lessons from an extensive case series and recommendations for clinical management. *J Clin Endocrinol Metab. (in press)*.

135. Chesover A, **Dattani MT** (2016) Evaluation of Growth Hormone Stimulation Testing in Children. *Clin Endo* **84(5):**708-14.

136. Fang Q, Benedetti AF, Ma Q, Gregory L, Li JZ, **Dattani M**, Sadeghi-Nejad A, Arnhold IJ, de Mendonça BB, Camper SA, Carvalho LR (2016) *HESX1* Mutations in Patients with Congenital Hypopituitarism: Variable Phenotypes with the Same Genotype. *Clinical Endocrinology* **85(3):**408-14.

137. Marx-Berger D, Milford DV, Bandhakavi M, Van't Hoff W, Kleta R, **Dattani M**, Bockenhauer D (2016) Tolvaptan is successful in treating inappropriate antidiuretic hormone secretion in infants. Acta Paediatrica **105(7):**e334-7.

138. Güemes M, Murray PG, Brain CE, Spoudeas HA, Peters CJ, Hindmarsh PC, **Dattani MT** (2016) Management of Cushing syndrome in children and adolescents: experience of a single tertiary centre. European Journal of Paediatrics **175(7)**:967-76.

139. Nicholas AK, Jaleel S, Lyons G, Schoenmakers E, **Dattani MT**, Crowne E, Bernhard B, Kirk J, Roche EF, Chatterjee VK, Schoenmakers N (2016) Molecular spectrum of TSHβ subunit gene defects in central hypothyroidism in the UK and Ireland. *Clinical Endocrinology* **86(3):**410-418.

140. Gregory LC, Alatzoglou KS, McCabe MJ, Hindmarsh PC, Saldanha JW, Romano N, Le Tissier P, **Dattani MT** (2016) Partial loss of function of the GHRH Receptor leads to mild Growth Hormone Deficiency. *J Clin Endocrinol Metab* **101(10):**3608-3615.

141. Nicholas AK, Serra EG, Cangul H, Alyaarubi S, Ullah I, Schoenmakers E, Deeb A, Habeb AM, AlMaghamsi M, Peters C, Nathwani N, Aycan Z, Saglam H, Bober E, **Dattani M**, Shenoy S, Murray PG, Babiker A, Willemsen R, Thankamony A, Lyons G, Irwin R, Padidela R, Tharian K, Davies JH, Puthi V, Park SM, Massoud AF, Gregory JW, Albanese A, Pease-Gevers E, Martin H, Brugger K, Maher ER, Chatterjee K, Anderson CA, Schoenmakers N (2016) Comprehensive screening of eight known causative genes in congenital hypothyroidism with gland-in-situ. *J Clin Endocrinol Metab* **101(12):**4521-4531.

142. Huang-Doran I, Tomlinson P, Payne F, Gast A, Sleigh A, Bottomley W, Harris J, Daly A, Rocha N, Rudge S, Clark J, Kwok A, Romeo S, McCann E, Müksch B, **Dattani M**, Zucchini S, Wakelam M, Foukas LC, Savage DB, Murphy R, O'Rahilly S, Barroso I, Semple RK (2016) Insulin resistance uncoupled from dyslipidemia due to C-terminal PIK3R1 mutations. *JCI Insight.* **1(17):**e88766.

143. Chow J, Rahman J, Achermann JC, **Dattani MT**, Rahman S (2017) Mitochondrial disease and endocrine dysfunction. *Nat Rev Endocrinol.* **13(2):**92-104.

144. Gevers EF, Meredith S, Shah P, Torpiano J, Peters C, Sebire NJ, Slater O, White A, **Dattani MT** (2017) Cushing syndrome in a child due to pro-opiomelanocortin (POMC) secretion from a yolk sac tumor. *Eur J Endocrinol*. **176(2):**K1-K7

145. Hawton K, Walton-Betancourth S, Rumsby G, Raine J, **Dattani M** (2017) Growth Hormone With Aromatase Inhibitor May Improve Height in CYP11B1 Congenital Adrenal Hyperplasia. *Pediatrics* **139(2)**. pii: e20160730. doi: 10.1542/peds.2016-0730.

146. Improda N, Ponmani C, Schoenmakers N, Senniappan S, Atterbury A, Barnicoat A, Chatterjee K, **Dattani MT** (2017) Intrafamilial Phenotypic Variability and Consequences of Non-Compliance with Treatment in Congenital Adrenal Hyperplasia and Congenital Hypothyroidism within a Single Family. *Hormone Research in Paediatrics* **88(2):**172-178.

147. Liao L, Gan HW, Hwa V, **Dattani M**, Dauber A (2017) Two Siblings with a Mutation in CCDC8 Presenting with Mild Short Stature: A Case of 3-M Syndrome. *Hormone Research in Paediatrics* **88(5):** 364-370**.**

148. Morandi G, Cerbone M, Lamback EB, Rapti E, **Dattani MT** (2018) 45,X/46,XY Mosaicism Presenting With Isolated Unilateral Cryptorchidism and a Normal Blood Karyotype. *J Clin Endocrinol Metab* **103(6):**2079-2082.

149. Klammt J, Neumann D, Gevers EF, Andrew SF, Schwartz ID, Rockstroh D, Colombo R, Sanchez MA, Vokurkova D, Kowalczyk J, Metherell LA, Rosenfeld RG, Pfäffle R, **Dattani MT,** Dauber A, Hwa V (2018) Dominant-negative STAT5B mutations cause growth hormone insensitivity with short stature and mild immune dysregulation. *Nat Commun.* 2018 May **29;9(1):**2105. doi: 10.1038/s41467-018-04521-0.

150. Martin CA, Sarlós K, Logan CV, Thakur RS, Parry DA, Bizard AH, Leitch A, Cleal L, Ali NS, Al-Owain MA, Allen W, Altmüller J, Aza-Carmona M, Barakat BAY, Barraza-García J, Begtrup A, Bogliolo M, Cho MT, Cruz-Rojo J, Dhahrabi HAM, Elcioglu NH; GOSgene, Gorman GS, Jobling R, Kesterton I, Kishita Y, Kohda M, Le Quesne Stabej P, Malallah AJ, Nürnberg P, Ohtake A, Okazaki Y, Pujol R, Ramirez MJ, Revah-Politi A, Shimura M, Stevens P, Taylor RW, Turner L, Williams H, Wilson C, Yigit G, Zahavich L, Alkuraya FS, Surralles J, Iglesais A, Murayama K, Wollnik B, **Dattani M**, Heath KE, Hickson ID, Jackson AP (2018) Mutations in TOP3A Cause a Bloom Syndrome-like Disorder *Am J Hum Genet.* 2018 Aug 2;103(2):221-231. doi: 10.1016/j.ajhg.2018.07.001. Epub 2018 Jul 26.

151. Roche EF, McGowan A, Koulouri O, Turgeon MO, Nicholas AK, Heffernan E, El-Khairi R, Abid N, Lyons G, Halsall D, Bonomi M, Persani L, **Dattani MT**, Gurnell M, Bernard DJ, Schoenmakers N (2018) A novel IGSF1 mutation in a large Irish kindred highlights the need for familial screening in the IGSF1 deficiency syndrome. *Clin Endocrinol (Oxf).* 2018 Aug 7. doi: 10.1111/cen.13827. [Epub ahead of print].

153. Dastamani A, Malhorta N, Güemes M, Morgan K, Rees CM, **Dattani M**, Shah P (2018) Post-Prandial Hyperinsulinaemic Hypoglycaemia after Oesophageal Surgery in Children *Hormone Research in Pediatrics* 2018 Aug 9:1-5. doi: 10.1159/000491647.

154. Dastamani A, Güemes M, Pitfield C, Morgan K, Rajab M, Rottenburger C, Bomanji J, De Coppi P, **Dattani M**, Shah P (2018) The Use of a Long-Acting Somatostatin Analogue (Lanreotide) in Three Children with Focal Forms of Congenital Hyperinsulinaemic Hypoglycaemia *Hormone Research in Pediatrics* 2018 Aug 16:1-6. doi: 10.1159/000491101.

155. Biebermann H, Kleinau G, Schnabel D, Bockenhauer D, Wilson LC, Tully I, Kiff S, Scheerer P, Reyes M, Paisdzior S, Gregory JW, Allgrove J, Krude H, Mannstadt M, Gardella TJ, **Dattani M***, Jüppner H*, Grüters A* (2018) A new multi-system disorder caused by the Gαs mutation p.F376V. *J Clin Endocrinol Metab* 2018 Oct 11. doi: 10.1210/jc.2018-01250.

156. Persani L, Brabant G, **Dattani M**, Bonomi M, Feldt-Rasmussen U, Fliers E, Gruters A, Maiter D, Schoenmakers N, van Trotsenburg ASP (2018) 2018 European Thyroid Association (ETA) Guidelines on the Diagnosis and Management of Central Hypothyroidism. *European Thyroid Journ*al 2018 Oct;7(5):225-237. doi:10.1159/000491388.

157. Ali SR, Bryce J, Cools M, Korbonits M, Beun JG, Taruscio D, Danne T, **Dattani MT**, Dekkers O, Linglart A, Netchine I, Nordenstrom A, Patocs A, Persani L, Reisch N, Smyth A, Sumnik Z, Visser WE, Hiort O, Pereira AM, Ahmed SF (2018) The current landscape of European registries for rare endocrine conditions. 2018 *European Journal of Endocrinology* Nov 1. pii: EJE-18-0861. doi: 10.1530/EJE-18-0861.

158. **Dattani MT** Letrozole: a new treatment for delayed puberty in boys? *Lancet Child Adolesc Health.* 2019 Feb;3(2):60-62. doi: 10.1016/S2352-4642(18)30405-X. Epub 2019 Jan 4.

159. Kiff S, Babb C, Guemes M, Dastamani A, Gilbert C, Flanagan SE, Ellard S, Barton J, **Dattani M**, Shah P Partial diazoxide responsiveness in a neonate with hyperinsulinism due to homozygous ABCC8 mutation. *Endocrinol Diabetes Metab Case Rep*. 2019 Feb 11;2019. pii: EDM180120. doi: 10.1530/EDM-18-0120.

160. Gregory LC, Ferreira CB, Young-Baird SK, Williams HJ, Harakalova M, van Haaften G, Rahman SA, Gaston-Massuet C, Kelberman D, GOSgene, Qasim W, Camper SA, Dever TE, Shah P, Robinson ICAF, **Dattani MT** Impaired EIF2S3 function associated with a novel phenotype of X-linked hypopituitarism with glucose dysregulation. *EBioMedicine.* 2019 Apr;42:470-480. doi: 10.1016/j.ebiom.2019.03.013. Epub 2019 Mar 14.

161. Cerbone M, Clement E, McClatchey M, Dobbin J, Gilbert C, Keane M, Boukhibar L, Williams H, Gagunashvili A, **Dattani MT**, Hurst J, Shah P. Sotos Syndrome Presenting with Neonatal Hyperinsulinaemic Hypoglycaemia, Extensive Thrombosis, and Multisystem Involvement. Horm Res Paediatr. 2019 Mar 15:1-7. doi: 10.1159/000496545. [Epub ahead of print] PMID:30879005

162. Wei W, Tuna S, Keogh MJ, Smith KR, Aitman TJ, Beales PL, Bennett DL, Gale DP, Bitner-Glindzicz MAK, Black GC, Brennan P, Elliott P, Flinter FA, Floto RA, Houlden H, Irving M, Koziell A, Maher ER, Markus HS, Morrell NW, Newman WG, Roberts I, Sayer JA, Smith KGC, Taylor JC, Watkins H, Webster AR, Wilkie AOM, Williamson C; NIHR BioResource–Rare Diseases; 100,000 Genomes Project–Rare Diseases Pilot, Ashford S, Penkett CJ, Stirrups KE, Rendon A, Ouwehand WH, Bradley JR, Raymond FL, Caulfield M, Turro E, Chinnery PF. Germline selection shapes human mitochondrial DNA diversity. *Science*. 2019 May 24;364(6442). pii: eaau6520. doi: 10.1126/science.aau6520. Epub 2019 May 23. PMID: 31123110

163. Hiort O, Cools M, Springer A, McElreavey K, Greenfield A, Wudy SA, Kulle A, Ahmed SF, Dessens A, Balsamo A, Maghnie M, Bonomi M, **Dattani M**, Persani L, Audi L; COST Actions DSDnet and GnRH Network as well as the European Reference Network for Rare Endocrine Conditions (Endo–ERN). Addressing gaps in care of people with conditions affecting sex development and maturation. *Nat Rev Endocrinol.* 2019 Aug 12. doi: 10.1038/s41574-019-0238-y. [Epub ahead of print] Review.

164. Bacila I, Adaway J, Hawley J, Mahdi S, Krone R, Patel L, Alvi S, Randell T, Gevers E, **Dattani M**, Cheetham T, Kyriakou A, Schiffer L, Ryan F, Crowne E, Davies JH, Ahmed SF, Keevil B, Krone N. Measurement of salivary adrenal-specific androgens as biomarkers of therapy control in 21-hydroxylase deficiency. *J Clin*

Endocrinol Metab. 2019 Jul 30. pii: jc.2019-00031. doi: 10.1210/jc.2019-00031. [Epub ahead of print]

165. Gregory LC, Shah P, Sanner JRF, Arancibia M, Hurst J, Jones WD, Spoudeas H, Le Quesne Stabej P, Williams HJ, Ocaka LA, Loureiro C, Martinez-Aguayo A, **Dattani MT**. Mutations in MAGEL2 and L1CAM Are Associated With Congenital Hypopituitarism and Arthrogryposis *J Clin Endocrinol Metab.* 2019 Dec 1;104(12):5737-5750. doi: 10.1210/jc.2019-00631.

166. Krieger TG, Moran CM, Frangini A, Visser WE, Schoenmakers E, Muntoni F, Clark CA, Gadian D, Chong WK, Kuczynski A, **Dattani M**, Lyons G, Efthymiadou A, Varga-Khadem F, Simons BD, Chatterjee K, Livesey FJ. Mutations in thyroid hormone receptor α1 cause premature neurogenesis and progenitor cell depletion in human cortical development. *Proc Natl Acad Sci U S A.* 2019 Nov 5;116(45):22754-22763. doi: 10.1073/pnas.1908762116. Epub 2019 Oct 18.

167. Gregory LC, **Dattani MT**. The molecular basis of congenital hypopituitarism and related disorders. *J Clin Endocrinol Metab.* 2019 Nov 8. pii: dgz184. doi: 10.1210/clinem/dgz184. [Epub ahead of print]

168. Tahoun M, Chandler JC, Ashton E, Haston S, Hannan A, Kim JS, D'Arco F, Bockenhauer D, Anderson G, Lin MH, Marzouk S, Saied MH, Miner JH, **Dattani M**, Waters AM. Mutations in *LAMB2* associate with albuminuria and Optic Nerve Hypoplasia with Hypopituitarism. *J Clin Endocrinol Metab.* 2019 Nov 26. pii: dgz216. doi: 10.1210/clinem/dgz216. [Epub ahead of print]

169. Marques P, Caimari F, Hernández-Ramírez LC, Collier D, Iacovazzo D, Ronaldson A, Magid K, Lim CT, Stals K, Ellard S, Grossman AB, Korbonits M; FIPA Consortium. Significant Benefits of AIP Testing and Clinical Screening in Familial Isolated and Young-onset Pituitary Tumors *J Clin Endocrinol Metab.* 2020 Jun 1;105(6):e2247-60. doi: 10.1210/clinem/dgaa040.PMID: 31996917

170. Woodcock T, Barker P, Daniel S, Fletcher S, Wass JAH, Tomlinson JW, Misra U, **Dattani M**, Arlt W, Vercueil A. Guidelines for the management of glucocorticoids during the peri-operative period for patients with adrenal insufficiency: Guidelines from the Association of Anaesthetists, the Royal College of Physicians and the Society for Endocrinology UKAnaesthesia. 2020 May;75(5):654-663. doi: 10.1111/anae.14963. Epub 2020 Feb 3.PMID: 32017012 Review.

171. Hietamäki J, Gregory LC, Ayoub S, Iivonen AP, Vaaralahti K, Liu X, Brandstack N, Buckton AJ, Laine T, Känsäkoski J, Hero M, Miettinen PJ, Varjosalo M, Wakeling E, ***Dattani MT**, *Raivio T (*Joint last authors) Loss-of-Function Variants in TBC1D32 Underlie Syndromic Hypopituitarism. J Clin Endocrinol Metab. 2020 Jun 1;105(6):1748-58. doi: 10.1210/clinem/dgaa078.PMID: 32060556

172. Cerbone M, Güemes M, Wade A, Improda N, **Dattani M** Endocrine morbidity in midline brain defects: Differences between septo-optic dysplasia and related disorders *Lancet EClinicalMedicine*. 2020. PMID: 32140665.

173. Hyperinsulinemic hypoglycemia in children and adolescents: Recent advances in understanding of pathophysiology and management**.** Güemes M, Rahman SA, Kapoor RR, Flanagan S, Houghton JAL, Misra S, Oliver N, **Dattani MT**, Shah P.Rev Endocr Metab Disord. 2020 Dec;21(4):577-597. doi: 10.1007/s11154-020-09548-7.PMID: 32185602.

174. Eggermann T, Elbracht M, Kurth I, Juul A, Johannsen TH, Netchine I, Mastorakos G, Johannsson G, Musholt TJ, Zenker M, Prawitt D, Pereira AM, Hiort

O; European Reference Network on Rare Endocrine Conditions (ENDO-ERN. Genetic testing in inherited endocrine disorders: joint position paper of the European reference network on rare endocrine conditions (Endo-ERN). *Orphanet J Rare Dis.* 2020 Jun 8;15(1):144. doi: 10.1186/s13023-020-01420-w.PMID: 32513286

175. Turro E, Astle WJ, Megy K, Gräf S, Greene D, Shamardina O, Allen HL, Sanchis-Juan A, Frontini M, Thys C, Stephens J, Mapeta R, Burren OS, Downes K, Haimel M, Tuna S, Deevi SVV, Aitman TJ, Bennett DL, Calleja P, Carss K, Caulfield MJ, Chinnery PF, Dixon PH, Gale DP, James R, Koziell A, Laffan MA, Levine AP, Maher ER, Markus HS, Morales J, Morrell NW, Mumford AD, Ormondroyd E, Rankin S, Rendon A, Richardson S, Roberts I, Roy NBA, Saleem MA, Smith KGC, Stark H, Tan RYY, Themistocleous AC, Thrasher AJ, Watkins H, Webster AR, Wilkins MR, Williamson C, Whitworth J, Humphray S, Bentley DR; NIHR BioResource for the 100,000 Genomes Project, Kingston N, Walker N, Bradley JR, Ashford S, Penkett CJ, Freson K, Stirrups KE, Raymond FL, Ouwehand WH Whole-genome sequencing of patients with rare diseases in a national health system. *Nature*. 2020 Jul;583(7814):96-102. doi: 10.1038/s41586-020-2434-2. Epub 2020 Jun 24.PMID: 32581362

176. de Jong MC, Gaze MN, Szychot E, Rozalén García V, Brain C, **Dattani M**, Spoudeas H, Hindmarsh P, Abdel-Aziz TE, Bomanji J, Shankar A, Stoneham S, Morley S, Beale T, Jawad S, Otero S, Proctor I, Amin S, Butler G, Hewitt RJ, Kurzawinski TR.Treating papillary and follicular thyroid cancer in children and young people: Single UK-center experience between 2003 and 2018 *J Pediatr Surg.* 2021 Mar;56(3):534-539. doi: 10.1016/j.jpedsurg.2020.07.034. Epub 2020 Aug 4.PMID: 32838975

177. Galazzi E, Improda N, Cerbone M, Soranna D, Moro M, Fatti LM, Zambon A, Bonomi M, Salerno M, **Dattani M**, Persani L Clinical benefits of sex steroids given as a priming prior to GH provocative test or as a growth-promoting therapy in peripubertal growth delays: Results of a retrospective study among ENDO-ERN centres**.** Clin Endocrinol (Oxf). 2021 Feb;94(2):219-228. doi: 10.1111/cen.14337. Epub 2020 Oct 15.PMID: 32969044

178. Lamback EB, Chiarini S, Roposch A, **Dattani MT** (2020) Congenital growth hormone deficiency associated with hip dysplasia and Legg-Calve-Perthes disease. Clin Endocrinol (Oxf). 2021 Apr;94(4):590-597. doi: 10.1111/cen.14365. Epub 2020 Nov 20.PMID: 33147364

179. Kostopoulou E, Dastamani A, Güemes M, Clement E, Caiulo S, Shanmugananda P, **Dattani M**, Gilbert C, Hurst JA, Shah P Syndromic Forms of Hyperinsulinaemic Hypoglycaemia-A 15-year follow-up Study. Clin Endocrinol (Oxf). 2021 Mar;94(3):399-412. doi: 10.1111/cen.14393. Epub 2021 Jan 31.PMID: 33345357

180. Di Cicco E, Moran C, Visser WE, Nappi A, Schoenmakers E, Todd P, Lyons G, **Dattani M**, Ambrosio R, Parisi S, Salvatore D, Chatterjee K, Dentice M Germ Line Mutations in the Thyroid Hormone Receptor Alpha Gene Predispose to Cutaneous Tags and Melanocytic Nevi**.** Thyroid. 2021 Mar 13. doi: 10.1089/thy.2020.0391. Online ahead of print.PMID: 33509032

181. Persani L, Bonomi M, Cools M, **Dattani M**, Dunkel L, Gravholt CH, Juul A ENDO-ERN expert opinion on the differential diagnosis of pubertal delay. Endocrine. 2021 Mar;71(3):681-688. doi: 10.1007/s12020-021-02626-z. Epub 2021 Jan 29.PMID: 33512657

182. Bosch I Ara L, Katugampola H, Dattani MT. Congenital Hypopituitarism During the Neonatal Period: Epidemiology, Pathogenesis, Therapeutic Options, and Outcome

Front Pediatr. 2021 Feb 2;8:600962. doi: 10.3389/fped.2020.600962. eCollection 2020.PMID: 33634051

183. Gualtieri A, Kyprianou N, Gregory LC, Vignola ML, Nicholson JG, Tan R, Inoue SI, Scagliotti V, Casado P, Blackburn J, Abollo-Jimenez F, Marinelli E, Besser REJ, Högler W, Karen Temple I, Davies JH, Gagunashvili A, Robinson ICAF, Camper SA, Davis SW, Cutillas PR, Gevers EF, Aoki Y, Dattani MT, Gaston-Massuet C Activating mutations in BRAF disrupt the hypothalamo-pituitary axis leading to hypopituitarism in mice and humans**.** Nat Commun. 2021 Apr 1;12(1):2028. doi: 10.1038/s41467-021-21712-4.

184. Gregory LC, Gergics P, Nakaguma M, Bando H, Patti G, McCabe MJ, Fang Q, Ma Q, Ozel AB, Li JZ, Poina MM, Jorge AAL, Benedetti AFF, Lerario AM, Arnhold IJP, Mendonca BB, Maghnie M, Camper SA, Carvalho LRS, **Dattani MT.** The phenotypic spectrum associated with OTX2 mutations in humans. Eur J Endocrinol. 2021 May 25;185(1):121-135. doi: 10.1530/EJE-20-1453.PMID: 33950863.

185. Backeljauw P, Cappa M, Kiess W, Law L, Cookson C, Sert C, Whalen J, **Dattani MT.** Impact of short stature on quality of life: A systematic literature review. Growth Horm IGF Res. 2021 Apr-Jun;57-58:101392. doi: 10.1016/j.ghir.2021.101392. Epub 2021 Apr 30.PMID: 33975197.

186. Coutant R, Bosch Muñoz J, Dumitrescu CP, Schnabel D, Sert C, Perrot V, **Dattani M.** Effectiveness and Overall Safety of NutropinAq® for Growth Hormone Deficiency and Other Paediatric Growth Hormone Disorders: Completion of the International Cooperative Growth Study, NutropinAq® European Registry (iNCGS). Front Endocrinol (Lausanne). 2021 May 25;12:676083. doi: 10.3389/fendo.2021.676083. eCollection 2021.PMID: 34113318.

187. Shariq OA, Lines KE, English KA, Jafar-Mohammadi B, Prentice P, Casey R, Challis BG, Selberherr A, Boon H, Cranston T, Ryan FJ, Mihai R, Healy U, Kurzawinski T, **Dattani MT**, Bancos I, Dy BM, Lyden ML, Young WF Jr, McKenzie TJ, Richards D, Thakker RV. Surgery. Multiple endocrine neoplasia type 1 in children and adolescents: Clinical features and treatment outcomes. 2021 Jun 25:S0039-6060(21)00453-0. doi: 10.1016/j.surg.2021.04.041. Online ahead of print.PMID: 34183184.

188. Buonocore F, Maharaj A, Qamar Y, Koehler K, Suntharalingham JP, Chan LF, Ferraz-de-Souza B, Hughes CR, Lin L, Prasad R, Allgrove J, Andrews ET, Buchanan CR, Cheetham TD, Crowne EC, Davies JH, Gregory JW, Hindmarsh PC, Hulse T, Krone NP, Shah P, Shaikh MG, Roberts C, Clayton PE, **Dattani MT**, Thomas NS, Huebner A, Clark AJ, Metherell LA, Achermann JC. Genetic Analysis of Pediatric Primary Adrenal Insufficiency of Unknown Etiology: 25 Years' Experience in the UK. J Endocr Soc. 2021 May 11;5(8):bvab086. doi: 10.1210/jendso/bvab086. eCollection 2021 Aug 1.PMID: 34258490.

189. Di Cicco E, Moran C, Visser WE, Nappi A, Schoenmakers E, Todd P, Lyons G, Dattani M, Ambrosio R, Parisi S, Salvatore D, Chatterjee K, Dentice M. Germ Line Mutations in the Thyroid Hormone Receptor Alpha Gene Predispose to Cutaneous Tags and Melanocytic Nevi. *Thyroid*. 2021 Jul;31(7):1114-1126. doi: 10.1089/thy.2020.0391. Epub 2021 Mar 13.PMID: 33509032.

190. Hage C, Gan HW, Ibba A, Patti G, Dattani M, Loche S, Maghnie M, Salvatori R. Advances in differential diagnosis and management of growth hormone deficiency in children**.** *Nat Rev Endocrinol.* 2021 Oct;17(10):608-624. doi: 10.1038/s41574-021-00539-5. Epub 2021 Aug 20.PMID: 34417587.

191. Saengkaew T, Patel HR, Banerjee K, Butler G, Dattani MT, McGuigan M, Storr HL, Willemsen RH, Dunkel L, Howard SR. Genetic evaluation supports differential diagnosis in adolescent patients with delayed puberty. *Eur J Endocrinol*. 2021 Oct 8;185(5):617-627. doi: 10.1530/EJE-21-0387.PMID: 34403359.

192. 100,000 Genomes Project Pilot Investigators, Smedley D, Smith KR, Martin A, Thomas EA, McDonagh EM, Cipriani V, Ellingford JM, Arno G, Tucci A, Vandrovcova J, Chan G, Williams HJ, Ratnaike T, Wei W, Stirrups K, Ibanez K, Moutsianas L, Wielscher M, Need A, Barnes MR, Vestito L, Buchanan J, Wordsworth S, Ashford S, Rehmström K, Li E, Fuller G, Twiss P, Spasic-Boskovic O, Halsall S, Floto RA, Poole K, Wagner A, Mehta SG, Gurnell M, Burrows N, James R, Penkett C, Dewhurst E, Gräf S, Mapeta R, Kasanicki M, Haworth A, Savage H, Babcock M, Reese MG, Bale M, Baple E, Boustred C, Brittain H, de Burca A, Bleda M, Devereau A, Halai D, Haraldsdottir E, Hyder Z, Kasperaviciute D, Patch C, Polychronopoulos D, Matchan A, Sultana R, Ryten M, Tavares ALT, Tregidgo C, Turnbull C, Welland M, Wood S, Snow C, Williams E, Leigh S, Foulger RE, Daugherty LC, Niblock O, Leong IUS, Wright CF, Davies J, Crichton C, Welch J, Woods K, Abulhoul L, Aurora P, Bockenhauer D, Broomfield A, Cleary MA, Lam T, **Dattani M**, Footitt E, Ganesan V, Grunewald S, Compeyrot-Lacassagne S, Muntoni F, Pilkington C, Quinlivan R, Thapar N, Wallis C, Wedderburn LR, Worth A, Bueser T, Compton C, Deshpande C, Fassihi H, Haque E, Izatt L, Josifova D, Mohammed S, Robert L, Rose S, Ruddy D, Sarkany R, Say G, Shaw AC, Wolejko A, Habib B, Burns G, Hunter S, Grocock RJ, Humphray SJ, Robinson PN, Haendel M, Simpson MA, Banka S, Clayton-Smith J, Douzgou S, Hall G, Thomas HB, O'Keefe RT, Michaelides M, Moore AT, Malka S, Pontikos N, Browning AC, Straub V, Gorman GS, Horvath R, Quinton R, Schaefer AM, Yu-Wai-Man P, Turnbull DM, McFarland R, Taylor RW, O'Connor E, Yip J, Newland K, Morris HR, Polke J, Wood NW, Campbell C, Camps C, Gibson K, Koelling N, Lester T, Németh AH, Palles C, Patel S, Roy NBA, Sen A, Taylor J, Cacheiro P, Jacobsen JO, Seaby EG, Davison V, Chitty L, Douglas A, Naresh K, McMullan D, Ellard S, Temple IK, Mumford AD, Wilson G, Beales P, Bitner-Glindzicz M, Black G, Bradley JR, Brennan P, Burn J, Chinnery PF, Elliott P, Flinter F, Houlden H, Irving M, Newman W, Rahman S, Sayer JA, Taylor JC, Webster AR, Wilkie AOM, Ouwehand WH, Raymond FL, Chisholm J, Hill S, Bentley D, Scott RH, Fowler T, Rendon A, Caulfield M. 100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care - Preliminary Report. *N Engl J Med.* 2021 Nov 11;385(20):1868-1880. doi: 10.1056/NEJMoa2035790.PMID: 34758253.

193. Whittaker DE, Oleari R, Gregory LC, Le Quesne-Stabej P, Williams HJ; GOSgene, Torpiano JG, Formosa N, Cachia MJ, Field D, Lettieri A, Ocaka LA, Paganoni AJ, Rajabali SH, Riegman KL, De Martini LB, Chaya T, Robinson IC, Furukawa T, Cariboni A, Basson MA, Dattani MT. A recessive PRDM13 mutation results in congenital hypogonadotropic hypogonadism and cerebellar hypoplasia**.** J Clin Invest. 2021 Dec 15;131(24):e141587.doi:10.1172/JCI141587.PMID: 34730112.

194. Teoh LJ, Solebo AL, Rahi JS; British Childhood Visual Impairment and Blindness Study Interest Group. Temporal trends in the epidemiology of childhood severe visual impairment and blindness in the UK Br J Ophthalmol. 2021 Dec 23:bjophthalmol-2021-320119. doi: 10.1136/bjophthalmol-2021-320119. Online ahead of print.PMID: 34949577.

195. McGlacken-Byrne SM, Le Quesne Stabej P, Del Valle I, Ocaka L, Gagunashvili A, Crespo B, Moreno N, James C, Bacchelli C, **Dattani MT**, Williams HJ, Kelberman D, Achermann JC, Conway GS. ZSWIM7 Is Associated With Human Female Meiosis and Familial Primary Ovarian Insufficiency J Clin Endocrinol Metab. 2022 Jan 1;107(1):e254-e263 doi:10.1210/clinem/dgab597.PMID: 34402903.

196. Akin L, Rizzoti K, Gregory LC, Corredor B, Le Quesne Stabej P, Williams H, Buonocore F, Mouilleron S, Capra V, McGlacken-Byrne SM, Martos-Moreno GÁ, Azmanov DN, Kendirci M, Kurtoglu S, Suntharalingham JP, Galichet C, Gustincich S, Tasic V, Achermann JC, Accogli A, Filipovska A, Tuilpakov A, Maghnie M, Gucev Z, Gonen ZB, Pérez-Jurado LA, Robinson I, Lovell-Badge R, Argente J, **Dattani MT.** Pathogenic variants in RNPC3 are associated with hypopituitarism and primary ovarian insufficiency. Genet Med. 2022 Feb;24(2):384-397. doi: 10.1016/j.gim.2021.09.019. Epub 2021 Nov 30. PMID: 34906446.

197. McGlacken-Byrne SM, Del Valle I, Le Quesne Stabej P, Bellutti L, Garcia-Alonso L, Ocaka L, Ishida M, Suntharalingham JP, Gagunashvili A, Ogunbiyi OK, Mistry T, Buonocore F, GOSgene, Crespo B, Nadjeda Moreno N, Niola P, Brooks T, Brain CE, Dattani MT, Kelberman D, Vento-Tormo R, Lagos CF, Livera G, Conway GS, Achermann JC. Pathogenic variants in the human m^6A reader *YTHDC2* are associated with primary ovarian insufficiency**.** *JCI Insights* (in press).

198. Cerbone M, Katugampola H, Simpson HL, **Dattani MT.** APPROACH TO THE PATIENT: Management of pituitary hormone replacement through transition. J Clin Endocrinol Metab. 2022 Mar 9:dgac129. doi: 10.1210/clinem/dgac129. Online ahead of print. PMID: 35262704.

199. Teare H, Argente J, **Dattani M**, Leger J, Maghnie M, Sherlock M, Ali GC, Francombe J, Marjanovic S. Challenges and improvement needs in the care of patients with central diabetes insipidus. *Orphanet J Rare Dis.* 2022 Feb 16;17(1):58. doi: 10.1186/s13023-022-02191-2.PMID: 35172866.

200. Shariq OA, Lines KE, English KA, Jafar-Mohammadi B, Prentice P, Casey R, Challis BG, Selberherr A, Boon H, Cranston T, Ryan FJ, Mihai R, Healy U, Kurzawinski T, Dattani MT, Bancos I, Dy BM, Lyden ML, Young WF Jr, McKenzie TJ, Richards D, Thakker RV Corrigendum to 'Multiple endocrine neoplasia type 1 in children and adolescents: Clinical features and treatment outcomes' [*Surgery* 171 (2021) 77-87] *Surgery*. 2022 Jun;171(6):1708-1711. doi: 10.1016/j.surg.2022.03.013. Epub 2022 Apr 12.PMID: 35428475

201. van Haalen FM, Kaya M, Pelsma ICM, Dekkers OM, Biermasz NR, Cannegieter SC, Huisman MV, van Vlijmen BJM, Feelders RA, Klok FA, Pereira AM; Endo-ERN Cushing and Thrombosis study group. Current clinical practice for thromboprophylaxis management in patients with Cushing's syndrome across reference centers of the European Reference Network on Rare Endocrine Conditions (Endo-ERN). Orphanet J Rare Dis. 2022 May 3;17(1):178. doi: 10.1186/s13023-022-02320-x.PMID: 35505430

202. Solebo AL, Teoh L, Rahi JS; British Childhood Visual Impairment and Blindness Study Interest Group. The role of screening and surveillance in the detection of childhood vision impairment and blindness in the UK. Arch Dis Child. 2022 Sep;107(9):812-817. doi: 10.1136/archdischild-2021-323470. Epub 2022 May 5.PMID: 35512795

203. Bezen D, Kutlu O, Mouilleron S, Rizzoti K, **Dattani M**, Guran T, Yeşil G. A homozygous Y443C variant in the RNPC3 is associated with severe syndromic congenital hypopituitarism and diffuse brain atrophy. Am J Med Genet A. 2022 Sep;188(9):2701-2706. doi: 10.1002/ajmg.a.62888. Epub 2022 Jul 6.PMID: 35792517

204. Bacila I, Lawrence NR, Mahdi S, Alvi S, Cheetham TD, Crowne E, Das U, **Dattani MT**, Davies JH, Gevers E, Krone RE, Kyriakou A, Patel L, Randell T, Ryan F, Keevil B, Ahmed SF, Krone NP. Health Status of Children and Young Persons with Congenital Adrenal Hyperplasia in the UK (CAH-UK): a cross-sectional

multi-centre study. Eur J Endocrinol. 2022 Aug 1:EJE-21-1109. doi: 10.1530/EJE-21-1109. Online ahead of print.PMID: 36001026.

205. Maniatis AK, Carakushansky M, Galcheva S, Prakasam G, Fox LA, Dankovcikova A, Loftus J, Palladino AA, de Los Angeles Resa M, Turich Taylor C, **Dattani MT**, Lebl J.J. Treatment Burden of Weekly Somatrogon vs Daily Somatropin in Children With Growth Hormone Deficiency: A Randomized Study Endocr Soc. 2022 Sep 10;6(10):bvac117. doi: 10.1210/jendso/bvac117. eCollection 2022 Oct 1.PMID: 36101713.

206. McGlacken-Byrne SM, Abdelmaksoud A, Haini M, Palm L, Ashworth M, Li J, Wang W, Wang X, Wang J, Callaghan B, Kinsler VA, Faravelli F, **Dattani MT.** Mosaic PRKACA duplication causing a novel and distinct phenotype of early-onset Cushing's syndrome and acral cutaneous mucinosis. Eur J Endocrinol. 2022 Dec 1;187(6):K55-K61. doi: 10.1530/EJE-22-0287.PMID: 36691942.

207. Zamanipoor Najafabadi AH, van der Meulen M, Priego Zurita AL, Faisal Ahmed S, van Furth WR, Charmandari E, Hiort O, Pereira AM, Dattani M, Vitali D, de Graaf JP, Biermasz NR; MTG6 Pituitary of Endo-ERN. Starting point for benchmarking outcomes and reporting of pituitary adenoma surgery within the European Reference Network on Rare Endocrine Conditions (Endo-ERN): results from a meta-analysis and survey study. Endocr Connect. 2022 Dec 13;12(1):e220349. doi: 10.1530/EC-22-0349. Print 2023 Jan 1.PMID: 36327151 Free PMC article. Review.

208. Man E, Mushtaq I, Barnicoat A, Carmichael P, Hughes CR, Davies K, Aitkenhead H, Amin R, Buchanan CR, Cherian A, Costa NJ, Creighton SM, Duffy PG, Hewson E, Hindmarsh PC, Monzani LC, Peters CJ, Ransley PG, Smeulders N, Spoudeas HA, Wood D, Hughes IA, Katugampola H, Brain CE, *Dattani MT, *Achermann JC. A Single-Center, Observational Study of 607 Children and Young People Presenting With Differences of Sex Development (DSD) J Endocr Soc. 2022 Oct 28;7(1):bvac165. doi: 10.1210/jendso/bvac165. eCollection 2022 Nov 17.PMID: 36419940 Free PMC article.*Joint Corresponding authors.

209. Teoh LJ, Solebo AL, Rahi JS; British Childhood Visual Impairment and Blindness Study Interest Group 2023 Temporal trends in the epidemiology of childhood severe visual impairment and blindness in the UK. Br J Ophthalmol. May;107(5):717-724. doi: 10.1136/bjophthalmol-2021-320119. Epub 2021 Dec 23.PMID: 34949577.

210. Gan HW, Leeson C, Aitkenhead H, Dattani M. Inaccuracies in plasma oxytocin extraction and enzyme immunoassay techniques 2023 Compr Psychoneuroendocrinol. 2023 Jun 8;15:100188. doi: 10.1016/j.cpnec.2023.100188. eCollection 2023 Aug.PMID: 37360277.

211. Brettell E, Högler W, Woolley R, Cummins C, Mathers J, Oppong R, Roy L, Khan A, Hunt C, Dattani M; G. H. D. study group. The Growth Hormone Deficiency (GHD) Reversal Trial: effect on final height of discontinuation versus continuation of growth hormone treatment in pubertal children with isolated GHD-a non-inferiority Randomised Controlled Trial (RCT) Trials. 2023 Aug 21;24(1):548. doi: 10.1186/s13063-023-07562-z.PMID: 37605233.

212. Aung Y, Kokotsis V, Yin KN, Banerjee K, Butler G, Dattani MT, Dimitri P, Dunkel L, Hughes C, McGuigan M, Korbonits M, Paltoglou G, Sakka S, Shah P, Storr HL, Willemsen RH, Howard SR. Key features of puberty onset and progression can help distinguish self-limited delayed puberty from congenital hypogonadotrophic hypogonadism Front Endocrinol (Lausanne). 2023 Aug 28;14:1226839. doi: 10.3389/fendo.2023.1226839. eCollection 2023.PMID: 37701896 Free PMC article.

213. Gregory LC, Cionna C, Cerbone M, **Dattani MT.** Genet Med. 2023 Identification of genetic variants and phenotypic characterization of a large cohort of patients with congenital hypopituitarism and related disorders. Sep;25(9):100881. doi: 10.1016/j.gim.2023.100881. Epub 2023 May 8.PMID: 37165954.

214. Backeljauw PF, Andrews M, Bang P, Dalle Molle L, Deal CL, Harvey J, Langham S, Petriczko E, Polak M, Storr HL, Dattani MT. Challenges in the care of individuals with severe primary insulin-like growth factor-I deficiency (SPIGFD): an international, multi-stakeholder perspective. Orphanet J Rare Dis. 2023 Oct 7;18(1):312. doi: 10.1186/s13023-023-02928-7.PMID: 37805563.

215. Gan HW, Cerbone M, Dattani MT. Appetite- and weight-regulating neuroendocrine circuitry in hypothalamic obesity. Endocr Rev. 2023 Nov 29:bnad033. doi: 10.1210/endrev/bnad033. Online ahead of print.PMID: 38019584.

216. Lawrence NR, Bacila I, Dawson J, Mahdi S, Alvi S, Cheetham TD, Crowne E, Das U, Dattani MT, Davies JH, Gevers E, Krone RE, Patel L, Randell T, Ryan FJ, Keevil B, Ahmed SF, Krone NP. Quality of Life in Children and Young People With Congenital Adrenal Hyperplasia-UK Nationwide Multicenter Assessment J Clin Endocrinol Metab. 2023 Dec 21;109(1):e336-e346. doi: 10.1210/clinem/dgad405.PMID: 37439248.

217. Veligratli F, Alexandrou D, Shah S, Amin R, **Dattani M**, Gan HW, Famuboni A, Lopez-Garcia C, Trompeter R, Bockenhauer D. Tolvaptan and urea in paediatric hyponatraemia. Pediatr Nephrol. 2024 Jan;39(1):177-183. doi: 10.1007/s00467-023-06091-w. Epub 2023 Jul 19.PMID: 37466863 .

218. Malhotra N, Yau D, Cunjamalay A, Gunasekara B, S A, Gilbert C, Morgan K, Dattani M, Dastamani A. Low-dose diazoxide is safe and effective in infants with transient hyperinsulinism**.** Clin Endocrinol (Oxf). 2024 Feb;100(2):132-137. doi: 10.1111/cen.14987. Epub 2023 Dec 7.PMID: 38059644

C. Invited Commentaries/Reviews

1. **Dattani MT,** Brook CGD (1996) Neonatal Screening for Congenital Hypothyroidism: Outcomes. *Current Opinion in Paediatrics* **8 (4):**389-395.

2. Strasburger CJ, **Dattani MT** (1997) New growth hormone assays: potential benefits. *Acta Paediatrica* (Suppl) **423:** 5-11.

3. **Dattani MT,** Martinez-Barbera JP, Thomas PQ, Brickman JB, Gupta R, Wales JKH, Hindmarsh PC, Beddington RSP, Robinson ICAF (1999) *HESX1*: a novel gene implicated in a familial form of septo-optic dysplasia. *Acta Paediatrica* **88 (supp 433):** 49-55.

4. **Dattani MT**, Robinson ICAF (2000) The molecular basis for developmental disorders of the pituitary gland in man. *Clinical Genetics* **57:** 337-346.

5. **Dattani MT,** Martinez-Barbera JP, Thomas PQ, Brickman JB, Gupta R, Wales JKH, Hindmarsh PC, Beddington RSP, Robinson ICAF (2000) Molecular Genetics of Septo-optic dysplasia. *Hormone Research* **53 (suppl 1):** 26-33.

6. **Dattani MT** Septo-optic dysplasia: From Mouse to Man (2001) *Clinical Paediatric Endocrinology* 10(1): 89-95.

7. **Dattani MT** Disorders of pituitary gland development (2001) *Clinical Paediatric Endocrinology* 10(16): 91-99.

8. **Dattani MT** GH deficiency might be associated with normal height in *PROP1* deficiency (2002) *Clinical Endocrinology* **57 (2):** 157-158.

9. **Dattani MT**, Robinson ICAF *HESX1* and septo-optic dysplasia (2002) *Reviews in Endocrine and Metabolic Disorders* **12: 3(4):** 289-300.

10. **Dattani MT** Structural hypothalamic defects (2002) *Journal of Paediatric Endocrinology and Metabolism* **15 (suppl 5):**1423-1424.

11. **Dattani MT** DNA testing in patients with GH deficiency at the time of transition (2003) *Growth Hormone and IGF Research* **13 (Suppl A):** S122-129.

12. **Dattani MT** A novel pituitary phenotype due to mutation in a novel gene (2003) *Journal of Paediatric Endocrinology and Metabolism* **16 (9):** 1207-1209.

13. **Dattani MT** Novel insights into the aetiology and pathogenesis of hypopituitarism (2004) *Hormone Research* **62 (suppl 3):**1-13.

14. Kelberman D, **Dattani MT** Genetics of Septo-optic Dysplasia (2007) *Pituitary* **10(4):**393-407.

15. Kelberman D, **Dattani MT** Hypothalamic and pituitary development: novel insights into the aetiology (2007) *European Journal of Endocrinology* **157 Suppl 1**: S3-14.

16. Kelberma**n** D**, Dattani MT** Hypopituitarism oddities: congenital causes (2007) *Hormone Research* **68 Suppl (5):** 138-144.

17. Kelberma**n** D**, Dattani MT** Septo-optic dysplasia – novel insights into the aetiology (2008) *Hormone Research* **69 (5):** 257-265.

18. Mehta A, **Dattani MT** Developmental disorders of the hypothalamus and pituitary gland associated with congenital hypopituitarism (2008) *Best Pract Res Clin Endocrinol Metab* **22(1):** 191-206.

19. Tziaferi V, Kelberman D, **Dattani MT** The role of SOX2 in hypogonadotrophic hypogonadism (2008) *Sex Dev* **2(4-5):** 194-199.

20. **Dattani MT** The candidate gene approach to the diagnosis of monogenic disorders (2009) *Hormone Research* **71 Suppl 2:** 14-21.

21. Kelberman D, **Dattani MT** Role of transcription factors in midline central nervous system and pituitary defects (2009) *Endocrine Dev* **14:**67-82.

22. Kumaran A, **Dattani MT** Future of growth hormone therapy (2009) *Indian J of Paediatrics* (in press).

23. Mehta A, **Dattani MT** GH deficiency in children (2009) *BMJ Point of Care* (online article).

24. Alatzoglou KS, **Dattani MT** (2009) Genetic forms of hypopituitarism and their manifestation in the neonatal period. *Early Human Development* **85(11):** 705-712.

25. McCabe M, Alatzoglou KS, **Dattani MT** (2011) Septo-optic dysplasia and other midline defects: the role of transcription factors: HESX1 and beyond. *Best Practice and Research: Clinical Endocrinology and Metabolism* 25(1): 115-24.

26. Alatzoglou KS, **Dattani MT** (2012) Phenotype-Genotype Correlations in Congenital Isolated Growth Hormone Deficiency. Indian Journal of Paediatrics 79(1): 99-106.

27. Peters CJ, **Dattani MT** (2012) Interpretations:IGF1. Arch Dis Child Educ Pract Ed (*in press*).

28. McCabe MJ, Bancalari RE, **Dattani MT** (2014) Diagnosis and evaluation of hypogonadism Pediatric Endocrinology Reviews Feb;11 Suppl 2:214-29.

29. Alatzoglou KS, Kular D, **Dattani MT** (2015) Autosomal Dominant Growth Hormone Deficiency (Type II) Pediatric Endocrinology Reviews **12(4):** 347-355.

30. Murray PG, **Dattani MT**, Clayton PE (2015) Controversies in the diagnosis and management of growth hormone deficiency in childhood and adolescence. Archives of Disease in Childhood **101(1):**96-100.

31. **Dattani MT** (2015) Decade in review-paediatric endocrinology: New genes, new therapies. *Nature Reviews in Endocrinology* **11(11):**638-9.

32. Flück CE, **Dattani M**, Mullis A (2016) Primus-Eugen Mullis, November 1954 - January 2016. Hormone Research in Paediatrics 85(3):219-20.

33. **Dattani M**, Mullis A, Flück CE (2016) Primus-Eugen Mullis, MD (1954-2016). Paediatric Endocrinology Revs 13(4):712-3.

34. Cerbone M, **Dattani MT** (2017) Progression from isolated growth hormone deficiency to combined pituitary hormone deficiency. *Growth Horm IGF Res.* Dec;37:19-25.

35. Corredor B, **Dattani M**, Gertosio C, Bozzola M (2018) Tall Stature: A challenge for clinicians. *Current Pediatric Reviews* 2018 Nov 4. doi: 10.2174/1573396314666181105092917.

36. Aftab S, **Dattani MT**. Pathogenesis of Growth Failure in Rasopathies *Pediatr Endocrinol Rev*. 2019 May;16(Suppl 2):447-458. doi: 10.17458/per.vol16.2019.ad.pathogenesisrasopathies. PMID: 31115196

37. Alatzoglou KS, Gregory LC, **Dattani MT** (2020) Development of the Pituitary Gland. Compr Physiol. 2020 Mar 12;10(2):389-413. doi: 10.1002/cphy.c150043.

Books

1. Handbook of Paediatric Endocrinology 2012 Mehul Dattani, Charles Brook; Publishers Blackwell.

2. Neuroendocrine Disorders in Children 2016 Editors: Mehul Dattani, Peter Hindmarsh, Lucinda Carr, Iain Robinson; Publishers Mac Keith Press.

3. Clinical Paediatric Endocrinology 7th Edition 2020 Editors: Mehul Dattani, Charles Brook; Publishers Blackwell.

4. Management of Endocrine Disorders in Children and Adolescents 1st Edition 2024 Editors: Mehul Dattani, Mohamad Maghnie; Publishers Springer

Abstracts (2000 onwards only)

Oral Communications

1. Dhillo WS, Turton J, **Dattani MT**, Cassar J (2000) A novel *PROP1* mutation causing combined pituitary hormone deficiency. 191st Meeting of the Society for Endocrinology (London, UK).

2. Turton JPG, McNay D, Woods K, Cassar J, Bouloux PM, **Dattani MT** (2000) Screening for mutations in *PROP1* in children with hypopituitarism. Annual meeting of the British Society for Paediatric Endocrinology (Birmingham, UK).

3. McNay D, Woods K, Thomas P, Kirk J, Dunger D, Clayton P, Stanhope R, **Dattani M** (2000) Mutational analysis of *HESX1* within septo-optic dysplasia (SOD). Annual meeting of the British Society for Paediatric Endocrinology (Birmingham, UK).

4. Brickman JM, Clements M, Tyrell R, Beddington RSP, Robinson ICAF, **Dattani MT** (2000) The N-terminus of the homeodomain protein *HESX1* is implicated in promoter-specific repression and dimerization. 11th International Congress of Endocrinology, (Sydney, Australia).

5. Turton JPG, McNay D, Woods K, Cassar J, Bouloux PM, Attard-Montalto S, Tuilpakov A, Hindmarsh PC, **Dattani MT** (2001) Mutations within *PROP1* and *PIT1* are rare causes of Combined Pituitary Hormone Deficiency (CPHD) 20th Joint Meeting of the British Endocrine Societies, Belfast, UK.

6. Carvalho LR, Woods KS, Zamparini AL, Mendonca BB, Brickman JM, Arnhold IJ, **Dattani MT** (2002) A novel homozygous mis-sense mutation (I26T) in the repressor domain of the transcription factor HESX1 is associated with evolving combined pituitary hormone deficiency (CPHD) and ectopic posterior lobe (PL) in the absence of a forebrain defect. 84th Annual Meeting of the Endocrine Societies, San Francisco, USA.

7. Mehta A, Hindmarsh P, Stanhope R, Preece M, Brain C, **Dattani M** (2002) Central Hypothyroidism: Variable responses to the TRH test. 30th Annual Meeting of the British Society for Paediatric Endocrinology, Plymouth, UK.

8. Woods KS, Turton PG, Carvalho LR, Arnhold IJP, Mendonca BB, Marcal N, Stifani S, Brickman J, **Dattani MT** (2003) Mutations within HESX1 in CPHD/IGHD are associated with impaired repression due to compromised interaction with the Groucho corepressor. 85th Annual Meeting of the Endocrine Society, Philadelphia, USA.

9. Alexander S, Rumsby G, Honour J, Hakeem V, **Dattani M**, Achermann J (2003) Partial loss of function mutations (delF54) in CYP17 can present with microphallus. 31st Meeting of the British Society for Paediatric Endocrinology and Diabetes, Edinburgh, UK.

10. Mehta A, Hindmarsh PC, **Dattani MT** (2003) ACTH insufficiency: a diagnostic dilemma. 31st Meeting of the British Society for Paediatric Endocrinology and Diabetes, Edinburgh, UK.

11. Turton JPG, Woods KS, Otonkoski T, **Dattani MT** (2004) Combined Pituitary Hormone Deficiency (CPHD) due to a complete deletion of *PROP1*. 86th Annual Meeting of the Endocrine Society, New Orleans, USA.

12. Mehta A, Turton J, **Dattani MT**, Hindmarsh PC (2004) Rapid waxing and waning in size of the pituitary mass may precede complete involution in patients with *PROP1* mutations. 43rd Annual Meeting of the European Society for Paediatric Endocrinology, Basel, Switzerland.

13. Turton JPG, Woods KS, Cundall M, Palmer R, Al-Zyoud M, Al-Ali M, Otonkoski T, Woodward K, **Dattani MT** (2004) Over- and under-dosage of the transcription factor *SOX3* can be associated with X-linked hypopituitarism in the absence of mental retardation. 43rd Annual Meeting of the European Society for Paediatric Endocrinology, Basel, Switzerland.

14. Achermann JC, Ercan O, Raza J, Lin L, **Dattani MT** (2004) Partial aromatase deficiency due to point mutations in *CYP19*. 43rd Annual Meeting of the European Society for Paediatric Endocrinology, Basel, Switzerland.

15. Lin L, Hindmarsh PC, **Dattani MT**, Achermann JC (2004) A homozygous R262Q mutation in the gonadotrophin-releasing hormone receptor (GnRHR) is associated with absent gonadotrophin pulsatility and delayed puberty/hypogonadotrophic hypogonadism. 43rd Annual Meeting of the European Society for Paediatric Endocrinology, Basel, Switzerland.

16. Lin L, Al-Ali M, Alzyoud M, Hindmarsh PC, Brain CE, Metherall L, Clark AJ, **Dattani MT**, Achermann JC (2004) Mutations in the Adrenocorticotrophin Receptor (*ACTHR, MC2R*) can be associated with disturbed renin-sodium homeostasis. 43rd Annual Meeting of the European Society for Paediatric Endocrinology, Basel, Switzerland.

17. Woods K, Turton J, Marcel N, Stefani S, Rhodes S, Brickman J, **Dattani MT** (2004) The N-terminus of the transcriptional repressor HESX1 facilitates interaction with the homeodomain activator PROP1. 12th International Congress of Endocrinology, Lisbon, Portugal.

18. Woods K, Turton J, Cundall M, Al-Zyoud M, Palmer R, El-Ali M, Otonkoski T, Woodward K, **Dattani MT** (2004) Duplication and polyalanine tract expansion within SOX3 associated with X-linked panhypopituitarism in the absence of mental retardation (MR). 12th International Congress of Endocrinology, Lisbon, Portugal.

19. Lin L, Ercan O, Raza J, **Dattani MT**, Achermann JC (2005) Variable phenotypes associated with 46XX aromatase (CYP19) insufficiency. 87th Annual Meeting of the Endocrine Society USA, San Diego, USA.

20. Kelberman D, Woods KS, McNay DEG, Turton JPG, Shalet SM, Rhodes SJ, Stifani S, Brickman JM, **Dattani MT** (2005) Functional analysis of *HESX1* mutations identifies functional domains of this homeodomain repressor. 87th Annual Meeting of the Endocrine Society USA, San Diego, USA.

21. Turton JPG, Mehta A, Woods KS, Shaltout AA, Al-Khawari M, Swift PG, Amselem S, Rhodes SJ, **Dattani MT** (2005) Molecular analysis of novel *PROP1* mutations associated with Combined Pituitary Hormone Deficiency (CPHD). 87th Annual Meeting of the Endocrine Society USA, San Diego, USA.

22. Vallette-Kasic S, Couture C, Pulichino A, Metherell L, Clark A, **Dattani MT**, Drouin J (2005) The M86R *TPIT* mutation associated with isolated ACTH deficiency interferes in protein-protein interactions. 87th Annual Meeting of the Endocrine Society USA, San Diego, USA.

23. Turton JPG, Reynaud R, Mehta A, Torpiano J, Saveanu A, Woods KS, Tiulpakov A, Zdravkovic V, Hamilton J, Parascandalo R, Vella C, Clayton PE, Shalet S, Barton J, Brue T, **Dattani MT** (2005) Novel POU1F1 mutations and phenotypic variability in a cohort of patients with Combined Pituitary Hormone Deficiency. ESPE/LWPES 7th Joint Meeting Paediatric Endocrinology, Lyon, France.

24. Kelberman D, Rizzotti K, Collins J, Carmignac D, Cianfarani S, Achermann J, Stanhope R, Bitner-Glindzicz M, Lovell-Badge R, Robinson ICAF, **Dattani MT** (2005) Mutations within the transcription factor SOX2 are associated with variable hypopituitarism in mouse and human. ESPE/LWPES 7th Joint Meeting Paediatric Endocrinology, Lyon, France.

25. Regan F, de Kardanet M, **Dattani M**, Sommer A, Attie KM, Dunger DB (2006) rhIGF-1/rhIGFBP3 treatment of patients with severe insulin resistance syndromes: preliminary data. 88th Annual Meeting of the Endocrine Society, Boston, USA.

26. Kelberman D, Rizzoti K, Bitner-Glindzicz M, Cianfarani S, Collins J, Chong KW, Kirk JMW, Ross R, Carmignac D, Lovell-Badge R, Robinson ICAF, **Dattani MT** (2006) Mutations within Sox2/SOX2 are associated with hypopituitarism in mouse and humans. 88th Annual Meeting of the Endocrine Society, Boston, USA.

27. Kelberman D, Castro S, Gerreli D, Palmer R, Crolla JA, Rizzoti K, Lovell-Badge R, Robinson ICAF, Taylor D, Gregory JW, **Dattani MT** (2007) *SOX2* is expressed in the forebrain and pituitary during human embryonic development and may be implicated in the Wnt--catenin pathway. 89th Annual Meeting of the Endocrine Society, Toronto, Canada.

28. Petkovic V, Lochmatter D, Clayton PE, **Dattani MT,** Eble A, Fluck C, Mullis PE, Thevis M 2007 A novel mutation in GH molecule (GH-E32A) affecting the correct GH mRNA splicing presented in a pedigree with IGHD type II. 46th Annual Meeting of the European Society for Paediatric Endocrinology, Helsinki, Finland.

29. Kelberman D, Castro S, Gerreli D, Palmer R, Crolla JA, Rizzoti K, Lovell-Badge R, Robinson ICAF, Taylor D, Gregory JW, **Dattani MT** (2007) *SOX2* is expressed in the forebrain and pituitary during human embryonic development and may be implicated in the Wnt--catenin pathway. 46th Annual Meeting of the European Society for Paediatric Endocrinology, Helsinki, Finland.

30. Alatzoglou KS, Turton JPG, Clayton PE, Mehta A, Buchanan C, Aylwin S, Crowne EC, Christesen HT, Hertel T, Trainer P, Savage MO, Raza J, Banerjee K, Sinha S, Ten S, Cheetham TD, Hindmarsh PC, Mullis PE, **Dattani MT** (2008) Congenital Isolated Growth Hormone Deficiency: Something Old, Something New. 90th Annual Meeting of the Endocrine Society USA, San Francisco, California, USA.

31. Petkovic V, Godi M, Lochmatter D, Turton JPG, Alatzoglou S, **Dattani MT**, Fluck C, Mullis PE (2008) A novel mutation in GH molecule (GH-R178H) affecting the correct Zn2+-induced dimerization and condensation in secretory granules presented in a patient with GH deficiency. 47th Annual Meeting of the European Society for Paediatric Endocrinology, Istanbul, Turkey.

32. Avbelj M, Romero C, Tziaferi V, McCabe M, Zhang C, Sidis Y, Plummer L, Elting M, Martin C, Zou Q, Mohammed M, **Dattani M**, Radovick S, Pitteloud N (2009) New loci for congenital hypopituitarism: overlap with Kallmann syndrome. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

33. Alatzoglou KS, Mehta A, Webb EA, Hindmarsh PC, **Dattani MT** (2009) Severe midline abnormalities result in a distinct spectrum of endocrinopathies: implications for genetic diagnosis and follow up. 37th meeting of the British Society for Paediatric Endocrinology and Diabetes, Reading, Berkshire.

34. Webb EA, O'Reilly M, Orgill J, Dale N, Salt A, Gringras P, **Dattani MT** (2009) Melatonin secretion in children with sleep disturbance and Septo-optic dysplasia. 37th meeting of the British Society for Paediatric Endocrinology and Diabetes, Reading, Berkshire.

35. Webb EA, O'Reilly M, Seunarine K, Clayden J, Dale N, Salt A, Clark C, **Dattani MT** (2009) Parahippocampal aberrations in children with growth hormone deficiency: A diffusion tensor imaging study. 37th meeting of the British Society for Paediatric Endocrinology and Diabetes, Reading, Berkshire.

36. Padidela R, Al-Ali N, Schoenmakers E, Agostini M, Rajanayagam O, **Dattani MT**, Chatterjee VKK (2009) A multisystem disorder associated with defective selenoprotein synthesis and a thyroid signature. 37th meeting of the British Society for Paediatric Endocrinology and Diabetes, Reading, Berkshire.

37. Gaston-Massuet C, Andoniadou CL, Signore M, Jayakody S, Charolidi N, Le Tissier P, **Dattani MT**, Martinez-Barbera JP (2010) Enhancement of the canonical Wnt pathway in Rathke's pouch results in pituitary tumours reminiscent of human adamantinomatous craniopharyngioma. 92nd Annual Meeting of the Endocrine Society USA, San Diego, California, USA.

38. McCabe MJ, Tziaferi T, Gaston-Massuet C, Gregory LC, Walker J**,** Tsai PS[4], Pitteloud N, Martinez-Barbera JP, **Dattani MT** (2010) Mutations in the Fibroblast Growth Factor 8 (*FGF8*) gene*,* are associated with complex midline and hypothalamo-pituitary defects. 49th Annual Meeting of the European Society for Paediatric Endocrinology, Prague, Czechoslovakia.

39. Webb,EA, O'Reilly, MA, Seunarine K, Clayden J, Dale, N, Salt, A, Clark C, **Dattani, MT**. Diffusion tensor imaging reveals specific white matter abnormalities in children with IGHD. 49th Annual Meeting of the European Society for Paediatric Endocrinology, Prague, Czechoslovakia.

40. Gevers E, **Dattani MT** (2010) growth hormone induces hepatic stat5 phosphorylation in early postnatal life in mice. 49th Annual Meeting of the European Society for Paediatric Endocrinology, Prague, Czechoslovakia.

41. Gaston-Massuet C, Andoniadou CL, Signore M, Jayakody S, Charolidi N, Le Tissier P, **Dattani MT**, Martinez-Barbera JP (2010) Enhancement of the canonical Wnt pathway in Rathke's pouch results in pituitary tumours reminiscent of human adamantinomatous craniopharyngioma. 49th Annual Meeting of the European Society for Paediatric Endocrinology, Prague, Czechoslovakia. **Awarded Henning Andersen prize for best Basic Science abstract.**

42. O'Reilly M, Webb E, Dale N, Salt A, **Dattani MT** (2010) Growth hormone deficiency in children is associated with selective cognitive deficits. 38th Annual Meeting of the British Society for Paediatric Endocrinology and Metabolism, Manchester, UK.

43. Gaston-Massuet C, Andoniadou C, Signore M, Jayakody S, Charolidi N, Le Tissier P, **Dattani MT**, Martinez-Barbera JP (2010) Enhancement of the canonical Wnt pathway in Rathke's pouch results in pituitary tumours reminiscent of human adamantinomatous craniopharyngioma. 38th Annual Meeting of the British Society for Paediatric Endocrinology and Metabolism, Manchester, UK. **Winner of best Abstract award.**

44. Alatzoglou KS, Kelberman D, Spadoni E, Gaston-Massuet C, Woods K, Nataranjan A, Maghnie M, Bitner-Glindzicz M, Dattani M (2010) Wide range of eye abnormalities in patients with hypopituitarism; is this showing a novel genetic aetiology? 38th Annual Meeting of the British Society for Paediatric Endocrinology and Metabolism, Manchester, UK.

45. Alatzoglou KS, Kosta K, Gkourogianni A, Hindmarsh PC, **Dattani MT** (2011) Investigation of premature adrenarche reveals a high incidence of congenital adrenal hyperplasia. 38th Annual Meeting of the British Society for Paediatric Endocrinology and Metabolism, Manchester, UK.

46. McCabe M, Gaston-Massuet C, Tziaferi V, Gregory L, Alatzoglou K, Signore M, Farooqi S, Raza J, Walker J, Kavanaugh S, Tsai PS, Pitteloud N, Martinez-Barbera JP, **Dattani MT** (2011) Mutations in the gene encoding the fibroblast growth factor 8 (FGF8) are associated with complex midline defects including recessive holoprosencephaly and hypothalamo-pituitary dysfunction. Annual meeting of the British Endocrine Societies, Birmingham, UK.

47. Gaston-Massuet C, KonIordou M, Andoniadou CL, McCabe M, Jayakody S, Le Tissier P, **Dattani M**, Martinez-Barbera JP (2011) The Wnt/-Catenin effector Tcf3 is required for normal hypothalamo-pituitary development. 93rd Annual Meeting of the Endocrine Society USA, Boston, USA.

48. McCabe M, Gevers E, Gregory L Baker J, Josifov D, **Dattani MT** (2011) A novel missense sequence variant in CHD7 associated with hypopituitarism. 50th Annual meeting of the European Society for Paediatric Endocrinology, Glasgow, UK.

49. Alagaratnam S, Brain C, Spoudeas H, **Dattani MT,** Hindmarsh PC, Allgrove J, Van't Hoff W, Kurzawinski T (2011) Surgical treatment of children with hyperparathyroidism: single centre experience. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

50. Andoniadou CL, Gaston-Masuet C, LeTissier P, **Dattani MT** & Martinez-Barbera1 JP (2011) Isolation and characterisation of tumorigenic progenitors/stem cells with a stabilizing mutation in β-catenin, in a mouse model of human adamantinomatous craniopharyngioma. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

51. Schoenmakers N, Bochukova E, Agostini M, Schoenmakers E, Rajanayagam O, Henning E, Gevers E, Sarri M, Offiah A, Albanese A, Halsall D, Schwabe J, Bain M, Lindley K, Muntoni F, Vargha-Khadem F, **Dattani MT**, Farooqi IS, Gurnell M, Chatterjee K (2011) Growth retardation and severe constipation due to the first human, dominant negative thyroid hormone receptor α mutation. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

52. Hindmarsh PC, Hill N, **Dattani M**, Peters C, Charmandari E, Matthews D (2011) Deconvolution analysis of 24 h serum cortisol profiles informs the amount and distribution of hydrocortisone replacement therapy. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

53. Gregory LC, Alatzoglou KS, **Dattani MT** (2011) Mild GH deficiency due to two novel homozygous mutations in the gene encoding GHRH receptor (GHRHR) in a single family. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

54. McCabe M, Gregory L, Gaston-Massuet C, Sbai O, Rondard P, Pfeifer M, Hulse T, Buchanan C, Pitteloud N, Martinez-Barbera JP, **Dattani MT** (2011) Mutations in *PROKR2* but not *PROK2* are associated with congenital hypopituitarism and septo-optic dysplasia. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

55. Webb E, Clayden J, Edmonds CJ, Seunarine K, Singhal A, Lanigan J, Lucas A, Clark C, Isaacs E, **Dattani MT** (2011) A selective effect of IGFBP3 on brain volumes in healthy children. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

56. Alatzoglou KS, Andoniadou CL, Kelberman D, Kim H, Pedersen-White P, Layman L, Martinez-Barbera JP, **Dattani MT** (2011) Novel *SOX2* mutation: from clinical phenotype to identification of new molecular mechanisms of SOX2 action and interactions. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

57. Schoenmakers N, Bochukova E, Agostini M, Schoenmakers E, Rajanayagam O, Keogh J, Henning E, Reinemund J, Gevers E, Sarri M, Offiah A, Albanese A, Halsall D, Schwabe J, Bain M, Lindley K, Muntoni F, Vargha-Khadem F, **Dattani MT**, Farooqi IS, Gurnell M, Chatterjee K (2012) Growth retardation and severe constipation due to the first human, dominant negative thyroid hormone receptor α mutation. Annual Meeting of the Society for Endocrinology, Harrogate, UK. **Winner Young Endocrinologist Clinical Prize.**

58. Schoenmakers N, Bochukova E, Agostini M, Schoenmakers E, Rajanayagam O, Keogh J, Henning E, Reinemund J, Gevers E, Sarri M, Offiah A, Albanese A, Halsall D, Schwabe J, Bain M, Lindley K, Muntoni F, Vargha-Khadem F, **Dattani MT**, Farooqi IS, Gurnell M, Chatterjee K (2012) Growth retardation and severe constipation due to the first human, dominant negative thyroid hormone receptor α mutation.15th International and 14th European Congress of Endocrinology (ICE/ECE 2012), Florence, Italy.

57. Sun Y, Bak B, Schoenmakers N, van Trotsenburg ASP, Oostdijk W, Voshol P, Cambridge E, White JK, le Tissier P, Mousavy Gharavy SN, Martinez-Barbera JP, Stokvis-Brantsma WH, Vulsma T, Kempers MJ, Persani L, Campi I, Bonomi M, Beck-Peccoz P, Zhu H, Davis TME, Hokken-Koelega ACS, Del Blanco DG, Rangasami JJ , Ruivenkamp CAL, Laros JFL, Kriek M, Kant SG, Bosch CAJ, Biermasz NR, Appelman-Dijkstra NM, Corssmit EP, Hovens GCJ, Pereira AM, den Dunnen JT, Breuning MH, Hennekam RC, Chatterjee KK*, **Dattani MT***, Wit JM*, Bernard DJ* (*Co-Senior Authors) (2012) Loss-of-function mutations in *IGSF1* cause central hypothyroidism and testicular enlargement: a novel X-linked syndrome. Annual Meeting of the Endocrine Society USA, Houston, Texas, USA.

58. Sun Y, Bak B, Schoenmakers N, van Trotsenburg ASP, Oostdijk W, Voshol P, Cambridge E, White JK, le Tissier P, Mousavy Gharavy SN, Martinez-Barbera JP, Stokvis-Brantsma WH, Vulsma T, Kempers MJ, Persani L, Campi I, Bonomi M, Beck-Peccoz P, Zhu H, Davis TME, Hokken-Koelega ACS, Del Blanco DG, Rangasami JJ , Ruivenkamp CAL, Laros JFL, Kriek M, Kant SG, Bosch CAJ, Biermasz NR, Appelman-Dijkstra NM, Corssmit EP, Hovens GCJ, Pereira AM, den Dunnen JT, Breuning MH, Hennekam RC, Chatterjee KK*, **Dattani MT***, Wit JM*, Bernard DJ* (*Co-Senior Authors) (2013) Loss-of-function mutations in IGSF1 cause a novel, X-linked syndrome of central hypothyroidism and testicular enlargement. Annual Meeting of the Society for Endocrinology. **Winner of Clinical Endocrinology Trust Prize for best clinical abstract.**

59. Gaston-Massuet C, Andoniadou C, **Dattani M**, Martinez-Barbera JP (2012) Genetic and in vitro inhibition of the Wnt/beta catenin pathway results I amelioration of

adamantinomatous craniopharyngioma. 51st Annual Meeting of the European Society for Paediatric Endocrinology, Leipzig, Germany.

60. Gevers E, Fan Y, Sperling M, **Dattani M** (2012) Growth plate specific reduction/deletion of growth hormone (GH) receptor (GHR) results in reduced tibia and femur growth. 51st Annual Meeting of the European Society for Paediatric Endocrinology, Leipzig, Germany.

61. Webb E, Kelberman D, Al Mutair A, Andoniadou C, Bacchelli C, Chanudet E, Kleta R, Lescai F, Stupka E, Beales P, Sowden E, Martinez-Barbera JP, **Dattani MT** (2012) A novel syndrome characterized by hypothalamic hormonal insufficiency, neonatal seizures, congenital abnormalities of the kidneys and urinary tract, and obesity due to mutation in a gene regulating hypothalamic development. 40th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Leeds, UK.

62. Schoenmakers N, Bak B, Sun Y, van Trotsenburg ASP, Oostdijk W, Voshol P, Persani L, Davis TME, le Tissier P, Mousavy Gharavy SN, Martinez-Barbera JP, Appelman-Dijkstra NM, Pereira AM, den Dunnen JT, Breuning MH, Hennekam RC, Chatterjee KK*, **Dattani MT***, Wit JM*, Bernard DJ* (2012) 40th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Leeds, UK.

63. Alatzoglou KS, Turton JPG, Kelberman D, McCabe MJ, Gregory LC, Webb EA, McNay DEG, Woods KS, Mehta A, **Dattani MT** (2012) Genetic screening in a cohort of 2030 patients with congenital hypopituitarism; current knowledge and future directions. 40th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Leeds, UK.

64. Kelberman D, Webb E, Al Mutair A, Bacchelli C, Chanudet E, Lescai F, Andoniadou CL, Banyan AHA, Al Swaid A, Alrifai M, AlBalwi M, Lukovic B, Burke D, Heales S, McCabe MJ, Kasia T, Kleta R, Stupka E, Beales PL, Thompson DA, Chong KW, Alkuraya F, Martinez-Barbera JP,Sowden E, **Dattani MT** (2013) ARNT2 deficiency causes a multisystem disorder with hypothalamic insufficiency. 95th Annual Meeting of the Endocrine Society USA, San Francisco, USA.

65. Andoniadou CL, Le Tissier PR, Pevny LH, **Dattani MT**, Martinez-Barbera JP (2013) The SOX2+ population of the adult murine pituitary includes stem cells with paracrine tumor-inducing potential. 95th Annual Meeting of the Endocrine Society USA, San Francisco, USA.

66. Gaston-Massuet C, Le Tissier PR, **Dattani MT**, Martinez-Barbera JP (2013) Genetic and in vitro inhibition of the Wnt/β catenin pathway results in amelioration of adamantinomatous craniopharyngioma. 95th Annual Meeting of the Endocrine Society USA, San Francisco, USA.

67. Gaston-Massuet C, McCabe M, Wu Chun I, Mousavy N, Sokol SY, **Dattani MT**, Martinez-Barbera JP (2013) The repressor activity of the Wnt/β-catenin effector Tcf3/TCF7L1 is required for normal hypothalamo-pituitary development. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy.

68. Webb E, Almutair A, Kelberman D, Bacchelli C, Chanudet E, Lescai F, Andoniadou C, Burke D, McCabe M, Kasia T, Kleta R, Alkuraya F, Martinez-Barbera JP, Sowden JC, **Dattani MT** (2013) Novel syndrome of microcephaly, endocrine, visual and renal abnormalities cuased by ARNT2 mutation. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy.

69. Joustra S, Schoenmakers N, Oostdijk W, Biermasz NR, Bonomi M, Radetti G, Persani L, Campi I, Pereira AM, Varewijck A, Janssen JAMLL, Chatterjee K, **Dattani MT**, van

Trotsenburg ASP, Wit JM (2013) The IGSF1 deficiency syndrome: clinical and biochemical characteristics of male and female patients. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy.

70. Guemes M, Cerbone M, **Dattani MT** (2013) Clinical, Biochemical and Radiological characterization of a cohort of patients with Septo-optic Dysplasia and multiple pituitary hormonal deficiencies. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Brighton, UK.

71. Schoenmakers N, H Cangul, A Nicholas, E Schoenmakers, G Lyons, **M Dattani**, C Peters, S Langham, A Habeb, A Deeb, V Puthi, S-M Park, E Maher, VK Chatterjee (2013) A comprehensive next generation sequencing-based strategy for genetic diagnosis in congenital hypothyroidism (CH). Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Brighton, UK.

72. Gregory L, Rhodes SJ, Levy MJ, Greening J, Humayun K, **Dattani MT** (2013) Novel lethal form of hypopituitarism associated with the first recessive *LHX4* mutation. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Brighton, UK.

73. Gaston-Massuet C, McCabe M, **Dattani M**, Martinez-Barbera JP (2013) The repressor activity of the Wnt/β-catenin effector Tcf3/TCF7L1 is required for normal hypothalamo-pituitary development. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Brighton, UK.

74. Gregory L, Humayun K, McCabe M, Turton JPG, Levy MJ, Greening J, Martinez-Barbera JP, Rhodes SJ, **Dattani MT** (2014) Novel lethal form of hypopituitarism associated with the first recessive *LHX4* mutation. 96th Annual Meeting of the Endocrine Society USA, Chicago, USA.

75. Guemes M, Cerbone M, Bagkeris M, Gregory LC, Kasia T, **Dattani M** (2014) Clinical and Neuroradiological characteristics in children and adolescents with Septo-Optic Dysplasia, Multiple Pituitary Hormone Deficiencies and Optic Nerve Hypoplasia: Experience from a Single Tertiary Centre. 53rd Annual Meeting of the European Society for Paediatric Endocrinology, Dublin, Ireland.

76. Gregory L, McCabe M, Bancalari R, Tziaferi V, Spoudeas H, **Dattani M** (2014) Novel Genetic Variants in a Cohort of Paediatric and Adolescent Patients with Hypogonadotrophic Hypogonadism and Kallmann Syndrome. 53rd Annual Meeting of the European Society for Paediatric Endocrinology, Dublin, Ireland.

77. Carreno G, Andoniadou C, Heywood W, Mills K, **Dattani M**, Martinez-Barbera JP (2014) Sonic Hedgehog is Required for Cell Specification of Rathke's Pouch Progenitors During Normal Development and is Over-Expressed in Adamantinomatous Craniopharyngioma. 53rd Annual Meeting of the European Society for Paediatric Endocrinology, Dublin, Ireland.

78. Sherif M, Demirbilek H, Cayir A, Ozbek MN, Baran RT, Cebeci AN, Tahir S, Rahman S, **Dattani M**, Hussain K (2014) Two Novel Homozynous Mutations in WFS1 Gene in Two Turkish Families with Mild Phenotypic Expression of Wolfram Syndrome. 53rd Annual Meeting of the European Society for Paediatric Endocrinology, Dublin, Ireland.

79. Alatzoglou KS, Andoniadou CL, Kelberman D, Kim HG, Botse-Baidoo E, Pedersen-White J, Layman L, Martinez-Barbera JP, **Dattani MT** (2014) Novel SOX2 Mutation: Identification of New Molecular Mechanisms of SOX2 Action and Interactions. 53rd Annual Meeting of the European Society for Paediatric Endocrinology, Dublin, Ireland.

80. Cerbone M, Guemes M, Kasia T, **Dattani MT** (2014) Endocrine Features of a Large Cohort of Children with Septo-Optic Dysplasia and Congenital Multiple Pituitary Hormonal Deficiencies. 53rd Annual Meeting of the European Society for Paediatric Endocrinology, Dublin, Ireland.

81. Gregory LC, Rhodes SJ, Humayun KN, Levy MJ, Greening J, **Dattani MT** (2014) Novel Lethal Form of Hypopituitarism Associated with the First Recessive LHX4 Mutation. 96th Annual Meeting of the Endocrine Society, Chicago, USA.

82. Gaston-Massuet C, McCabe MJ, Gregory LC, Scagliotti V, **Dattani MT** (2014) Mutations in the Wnt/B-Catenin Effector Tcf3/TCF7L1 Are Associated with Septo-Optic –Dysplasia in Mouse and Human. 96th Annual Meeting of the Endocrine Society, Chicago, USA.

84. Gaston-Massuet C, McCabe MJ, Scagliotti V, Gregory LC, Jayakody S, Martinez-Barbera JP, **Dattani MT** (2015) Mutations in the Wnt/β-catenin effector Tcf3/TCF7L1 are associated with septo-optic dysplasia in mouse and humans. 97thAnnual meeting of the Endocrine Society USA, San Diego, USA.

85. Besser R, Gregory L, Davies J, **Dattani MT** (2015) Mutations in *BRAF* are associated with septo-optic dysplasia and cardiofaciocutaneous syndrome. 54TH Annual Meeting of the European Society for Paediatric Endocrinology, Barcelona, Spain.

86. Pease-Gevers E,…..**Dattani MT**, Hwa V (2015) Heterozygous dominant negative negative STAT5B variants associated with short stature and GH insensitivity. 54TH Annual Meeting of the European Society for Paediatric Endocrinology, Barcelona, Spain.

87. Gregory L, Besser R, Davies J, **Dattani MT** (2015) Mutations in *BRAF* are associated with septo-optic dysplasia and cardiofaciocutaneous syndrome. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Sheffield, UK.

139. Tully I, ……..Dattani MT, Gregory J (2017) A novel distinct syndrome of Nephrogenic syndrome of inappropriate anti-diuresis (NSIAD), precocious puberty, parathyroid hormone insensitivity and developmental delay associated with a novel p.F376V mutation in *GNAS.* Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Newcastle, UK.

140. Kyprianou N……Dattani MT (2017) The MAPK effector B-Raf is essential for hypothalamic-pituitary axis development and activating mutations in *BRAF* cause Congenital Hypopituitarism. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Newcastle, UK.

141. Nicole Goff, Harshini Katugampola, Elena Monti, Katherine Taylor, Rakesh Amin, Peter Hindmarsh, Catherine Peters, Shah Pratik, Helen Spoudeas, Mehul Dattani, Jeremy Allgrove, Caroline Brain (2018) Management of severe, protracted hypocalcaemia in patients undergoing thymus transplantation in a tertiary centre: a 10-year experience. 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

142. Antonia Dastamani, Eirini Kostopoulou, Emma Clements, Silvana Caiulo, Prateek Shanmugananda, Kate Morgan, Clare Gilbert, Mehul Dattani, Sarah Flanagan, Sian Ellard, Jane Hurst, Pratik Shah (2018) Genotype and Phenotype Correlation in Syndromic Forms of Hyperinsulinaemic Hypoglycaemia – a 10-year follow-up study in a tertiary centre. 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

143. Salma R. Ali, Jillian Bryce, Martine Cools, Marta Korbonits, Johan G. Beun, Domenica Taruscio, Felix Beuschlein, Thomas Danne, Mehul Dattani, Olaf Dekkers, Agnès Linglart, Irene Netchine, Anna Nordenstrom, Attila (2018) Awareness &

Participation In Rare Disease Registries Within The European Reference Network On Rare Endocrine Conditions (Endo-ERN) 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

144. Louise C Gregory, Shah Pratik, Juliane RF Sanner, Monica Arancibia, Jane Hurst, Wendy D Jones, Helen Spoudeas, Polona Le Quesne Stabej, Louise Ocaka, Carolina Loureiro, Alejandro Martinez-Aguayo, Hywel Williams, Mehul T Dattani (2018) Mutations in MAGEL2 and L1CAM are associated with congenital hypopituitarism and arthrogryposis. 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

145. Manuela Cerbone, Maria Güemes, Angie Wade, Nicola Improda, Mehul T Dattani (2018) CAN NEUROIMAGING PREDICT ENDOCRINE MORBIDITY IN CONGENITAL HYPOTHALAMO-PITUITARY (H-P) DISORDERS? 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

146. Irina-Alexandra Bacila, Jo Adaway, James Hawley, Sundus Mahdi, Carlo L Acerini, Ruth Krone, Leena Patel, Sabah Alvi, Tabitha Randell, Evelien Gevers, Mehul Dattani, Timothy Cheetham, Andreas Kyriakou, Lina Schiffer, Fiona Ryan, Elizabeth Crowne, Justin H Davies, S Faisal Ahmed, Brian Keevil, Nils P Krone (2019) Development of novel non-invasive strategies for monitoring of treatment control in patients with congenital adrenal hyperplasia. 58th Annual Meeting of the European Society for Paediatric Endocrinology, Vienna, Austria.

147. Salma Ali, Jillian Bryce, Tom Muir, Akanimo Okure, Martine Cools, Thomas Danne, Mehul Dattani, Olaf Dekkers, Olaf Hiort, Agnès Linglart, Irène Netchine, Anna Nordenström, Attila Patócs, Alberto Pereira, Luca Persani, Nicole Reisch, Arlene Smyth, Zdeněk Šumnik, Domenica Taruscio, W. Edward Visser, S.Faisal Ahmed (2019) 58th Annual Meeting of the European Society for Paediatric Endocrinology, Vienna, Austria.

148. Leyla Akin, Louise Gregory, Federica Buonocore, GOSgene Group, Selim Kurtoglu, Mustafa Kendirci, Z. Burçin Gonen, Robin Lovell-Badge, Karine Rizzoti, Mehul Dattani (2019) A novel minor spliceosome defect associated with growth hormone deficiency (GHD) and primary ovarian insufficiency (POI) 58th Annual Meeting of the European Society for Paediatric Endocrinology, Vienna, Austria.

149. Cecilia Cionna, Manuela Cerbone, Louise C Gregory, Mehul T Dattani (2019) Phenotypic characterization of a large pediatric cohort of patients with genetic forms of congenital hypopituitarism. 58th Annual Meeting of the European Society for Paediatric Endocrinology, Vienna, Austria.

150. Prentice P, Aftab S, Atterbury A, Langham S, Peters C, Shah P, Tollerfield S, Wade S, Katugampola H, Dattani M (2019) Recombinant human Insulin-like growth factor-1 (rhIGF-1) therapy for IGF-1 deficiency: a 15-year experience in a tertiary care centre. 47th Annual Meeting of the British Society for Paediatric Endocrinology.

151. Gregory LC, Gergics P, Patti G, McCabe MJ, Maghnie M, Spadoni E, Camper SA, Dattani MT (2020) *OTX2* mutations in Congenital Hypopituitarism. Annual Meeting of the Endocrine Society 2020 (ONLINE presentation).

152. Gregory LC, Krywawych S, Eaton S, Bitner-Glindzicz M, Rahman S, GOSGene, Clayton P, Robinson ICAF, Dattani M (2022) A novel missense variant in the gene encoding *Fatty Acid Synthase* (*FASN*) associated with a unique multi-system disorder including hypopituitarism and hypoparathyroidism. 60th Annual Meeting of the European Society for Paediatric Endocrinology, Rome, Italy.

153. McGlacken-Byrne S, Abdelmaksoud A, Haini M, Palm L, Ashworth M, Li J, Wang W, Wang X, Wang J, Callaghan B, Kinsler V, Faravelli S, Dattani M (2022) Mosaic *PRKACA* duplication causing a novel and distinct phenotype of early-onset Cushing syndrome and acral cutaneous mucinosis. 60th Annual Meeting of the European Society for Paediatric Endocrinology, Rome, Italy.

154. Shaunak M, McGlacken-Byrne S, Dattani M (2022) Two cases on the Carney Complex spectrum secondary to *PRKACA/PRKAR1A* variants presenting with Cushing Syndrome in childhood. 48th Annual Meeting of the British Society for Paediatric Endocrinology, Belfast.

155. Kokotsis V, Burchett C, Butler G, Dattani MT, Hughes S, McGuigan M, Shah P, Willemsen R, Howard S (2022) Central Delayed puberty in Adolescence: Differentiating the phenotypes of Congenital Hypogonadotropic Hypogonadism and Self-Limited Delayed Puberty. 48th Annual Meeting of the British Society for Paediatric Endocrinology, Belfast.

156. Gregory LC, Dattani MT (2023) Targeted gene panel screening in 144 congenital hypopituitarism patients, incorporating 135 known and novel genes implicated in hypopituitarism and/or hypothalamo-pituitary development. 61st Annual Meeting of the European Society for Paediatric Endocrinology, The Hague, Netherlands.

157. Cerbone M, D'Arco F, Spoudeas HA, Clark C, Dattani MT (2023) Pituitary size on volumetric MRI predicts the severity of the neuroendocrine phenotype in populations at risk. 61st Annual Meeting of the European Society for Paediatric Endocrinology, The Hague, Netherlands.

158. McGlacken-Byrne SM, Del Torres I, Suntharalingham JP, Buonocore F, HDBR, Crespo B, Moreno N, Simcock I, Arthurs O, Xenakis T, Niola P, Brooks, Dattani MT, John C Achermann JC (2023) Mapping the transcriptomic landscape of early human fetal ovary development through a clinically-focused lens. 61st Annual Meeting of the European Society for Paediatric Endocrinology, The Hague, Netherlands.

159. Napoli F, Cerbone M, Shaikh MG, Buonocore F, Angelelli A, Fava D, Tuli T, Angela Pistorio A, Dattani MT, Maghnie M (2023) Clinical phenotypes of a multicentric cohort of ROHHAD patients. 61st Annual Meeting of the European Society for Paediatric Endocrinology, The Hague, Netherlands.

160. Lawrence N, Bacila I, Ji X, Ahmed SF, Alvi S, Bath L, Blair J, Cheetham T, Crowne E, Davies J, Dattani M, Gevers E, Krone R, Patel L, Thankamony A, Randell T, Ryan *F,* Elford S, Blackett S, Krone NP (2023) National service evaluation project analysing the quality of care for children and young people with congenital adrenal hyperplasia in the United Kingdom: Data from patients and clinicians. 61st Annual Meeting of the European Society for Paediatric Endocrinology, The Hague, Netherlands.

Poster Presentations

1. Brickman JM, Clements M, Tyrell R, Woods K, Beddington RSP, Robinson ICAF, **Dattani MT** (2000) The N-terminus of the homeodomain protein HESX1 is implicated in promoter-specific repression and dimerization. 20th Joint Meeting of the British Endocrine Societies, Belfast, UK.

2. Turton JP, McNay D, Woods K, Cassar J, Bouloux PM, Attard-Montalto S, Tuilpakov A, Hindmarsh PC, **Dattani MT** (2001) Mutations within PROP1 and PIT1 are rare causes of combined pituitary hormone deficiency (CPHD). 83rd Annual meeting of the Endocrine Society, Denver, Colorado, USA.

3. Turton JP, McNay D, Woods K, Cassar J, Bouloux PM, Attard-Montalto S, Tuilpakov A, Hindmarsh PC, **Dattani MT** (2001) Mutations within PROP1 and PIT1 are rare causes of combined pituitary hormone deficiency (CPHD). 6th Joint Meeting of the Lawson-Wilkins and European Societies of Paediatric Endocrinology, Montreal, Canada.

4. McNay D, Woods K, Turton JP, Shalet S, **Dattani MT** (2001) Mutational and functional analysis of HESX1 within SOD. Annual Meeting of the American Society for Human Genetics, San Diego, USA.

5. Woods K, McNay D, Turton J, **Dattani MT** (2001) Septo-optic dysplasia: a multi-genic disorder. Annual Meeting of the American Society for Human Genetics, San Diego, USA.

6. Woods KS, Carvalho L, Turton JPG, McNay DEG, Thomas PQ, Cameron F, Zacharin M, Brickman JM, **Dattani MT** (2002) Loss of repression by HESX1 mutations – a novel mechanism for GHD/CPHD. 84th Annual Meeting of the Endocrine Societies, San Francisco, USA.

7. Turton JPG, McNay DEG, Woods KS, Preece MA, **Dattani MT** (2002) Screening for genetic mutations in patients with CPHD/GHD. 84th Annual Meeting of the Endocrine Societies, San Francisco, USA.

8. Woods KS, Turton JPG, McNay DEG, **Dattani MT** (2002) Mutational analysis of genes implicated in forebrain and pituitary development within septo-optic dysplasia. 84th Annual Meeting of the Endocrine Societies, San Francisco, USA.

9. Deeb A, Jaaskelainen J, Martin H, Dattani M, Hughes IA (2003) A mutation in the hinge region of the human androgen receptor: Functional effects in vitro and in vivo. 85th Annual Meeting of the Endocrine Society, Philadelphia, USA.

10. Mehta A, Hindmarsh PC, Stanhope RG, Brain CE, Preece MA, **Dattani MT** (2003) Central Hypothyroidism: Variable responses to the TRH test. 85th Annual Meeting of the Endocrine Society, Philadelphia, USA.

11. Mehta A, Hindmarsh PC, Stanhope RG, Preece MA, **Dattani MT** (2003) Characteristics of growth hormone deficient patients with early growth failure. 85th Annual Meeting of the Endocrine Society, Philadelphia, USA.

12. Turton JP, Mehta A, Woods KS, Cassar J, Hindmarsh PC, **Dattani MT** (2003) Mutations within the transcription factor PROP1 are associated with a pituitary mass that can rapidly involute. 85th Annual Meeting of the Endocrine Society, Philadelphia, USA.

13. Mehta A, Hindmarsh PC, Stanhope RG, Preece MA, **Dattani MT** (2003) Characteristics of growth hormone deficient patients with early growth failure. Annual Meeting of the European Society for Paediatric Endocrinology, Ljubljana, Slovenia.

14. Hussain K, Roebuck JD, **Dattani MT**, Hindmarsh PC, Lindley KJ (2003) Trans-hepatic pancreatic venous sampling (PVS) to differentiate focal and diffuse forms of hyperinsulinism in infancy (HI): the case for specialist centres. Annual Meeting of the European Society for Paediatric Endocrinology, Ljubljana, Slovenia.

15. Hussain K, Cosgrove KE, Smith V, Gregory JW, Christesen HT, Jacobsen BB, Brusgaard K, **Dattani MT**, Hindmarsh PC, Lindley KJ, Dunne MJ (2003) Hyperinsulinaemic hypoglycaemia in Beckwith-Weidemann syndrome (BWS) due to defects in pancreatic -cell ATP-sensitive K+ channels. Annual Meeting of the European Society for Paediatric Endocrinology, Ljubljana, Slovenia.

16. Metherell LA, Savage MO, **Dattani MT**, Walker J, Clayton PE, Clark AJL (2003) Absence of TPIT (TBX19) gene mutations in most patients with isolated ACTH deficiency. 42nd Annual Meeting of the European Society for Paediatric Endocrinology, Ljubljana, Slovenia.

17. Lindley KJ, **Dattani MT**, Jackson RS, Farooqi IS, Creemers JW, Rahier J, O'Rahilly S (2003) Intestinal failure with intractable neonatal diarrhoea and hypoglycaemia resulting from congenital deficiency of Prohormone Convertase 1.42nd Annual Meeting of the European Society for Paediatric Endocrinology, Ljubljana, Slovenia.

18. Woodward K, Cundall M, Turton JPG, Woods KS, Palmer R, Otonkoski T, **Dattani MT** (2004) Sub-microscopic chromosome Xq27.1 duplications that include the SOX3 gene are associated with hypopituitarism. 86th Annual Meeting of the Endocrine Society, New Orleans, USA.

19. Turton JPG, Woods KS, Al-Zyoud M, Woodward K, **Dattani MT** (2004) A Polyalanine tract expansion within SOX3 is associated with X-linked panhypopituitarism in the absence of mental retardation. 86th Annual Meeting of the Endocrine Society, New Orleans, USA.

20. Mehta A, Hindmarsh PC, **Dattani MT** (2004) ACTH Insufficiency – a diagnostic dilemma. 86th Annual Meeting of the Endocrine Society, New Orleans, USA.

21. Lin L, Rumsby G, Honour JW, Hakeem V, **Dattani MT**, Achermann JC (2004) Micropenis or Hypospadias due to a partial loss of function mutation (del54F) in CYP17. 86th Annual Meeting of the Endocrine Society, New Orleans, USA.

22. Woods KS, Turton JPG, Marcal N, Stefani S, Rhodes SJ, Brickman JM, **Dattani MT** (2004) The N-terminus of the transcriptional repressor HESX1 facilitates interaction with the homeodomain activator PROP1. 86th Annual Meeting of the Endocrine Society, New Orleans, USA.

23. Mehta A, Hindmarsh PC, **Dattani MT** (2004) Congenital ACTH Insufficiency: A diagnostic dilemma. 43rd Annual Meeting of the European Society for Paediatric Endocrinology, Basel, Switzerland.

24. Segal TY, Mitchell H, Anazodo A, Chiang W, Hindmarsh PC, **Dattani MT** (2004) Receiver Operating Characteristic (ROC) curve assessment of the 3 day and 3 week human chorionic gonadotrophin test in pubertal delay. 43rd Annual Meeting of the European Society for Paediatric Endocrinology, Basel, Switzerland.

25. Turton JPG, Mehta A, Woods KS, Swift P, Otonkoski T, Al-Khawari M, **Dattani MT** (2004) Novel mutations within PROP1 associated with combined pituitary hormone deficiency (CPHD). 43rd Annual Meeting of the European Society for Paediatric Endocrinology, Basel, Switzerland.

26. Clayton PE, Turton J, Trueman JA, Price DA, Trainer P, **Dattani MT** (2004) Normal Growth Hormone (GH) levels during a glucagon stimulation test in adulthood associated with autosomal dominant GH deficiency. 43rd Annual Meeting of the European Society for Paediatric Endocrinology, Basel, Switzerland.

27. Turton JPG, McNay DEG, Woods KS, Shalet SM, **Dattani MT** (2005) Mutational screening of coding and regulatory regions of the homeobox gene HESX1 in a cohort of patients with septo-optic dysplasia and hypopituitarism. 87th Annual Meeting of the Endocrine Society USA, San Diego, USA.

28. Turton JPG, Rasheed R, Mehta A, Woods KS, Hertel NT, Clayton PE, Trainer PJ, Buchanan CR, Raza J, Crowne E, Aylwin SJB, Mullis PE, Preece MA, Robinson ICAF, **Dattani MT** (2005) Type II autosomal dominant GHD is the most common genetic finding in a cohort of patients with IGHD. 87th Annual Meeting of the Endocrine Society USA, San Diego, USA.

29. Mehta A, Hindmarsh PC, **Dattani MT** (2005) The gonadotrophin (Gn) releasing hormone (GnRH) test during the infantile gonadotrophin surge in children with hypothalamo-pituitary disorders (HPD) and/or midline forebrain defects (MFD). 87th Annual Meeting of the Endocrine Society USA, San Diego, USA.

30. Turton JPG, Rasheed R, Clayton PE, Trainer PJ, Raza J, Crowne E, Hertel NT, Preece MA, Buchanan CR, Aylwin SJB, Mehta A, **Dattani MT** (2005) Splicing mutations within GH-1 are the commonest genetic cause of IGHD. ESPE/LWPES 7th Joint Meeting Paediatric Endocrinology, Lyon, France.

31. Turton J, Mehta A, Woods KS, Swift P, Al-Kuwari M, Amselem S, Rhodes SG, **Dattani MT** (2005) Novel genetic mechanisms leading to PROP1 deficiency. ESPE/LWPES 7th Joint Meeting Paediatric Endocrinology, Lyon, France.

32. Kelberman D, Woods KS, McNay DEG, Turton JPG, Shalet SM, Rhodes SJ, Stifani S, Brickman JB, **Dattani MT** (2005) Analysis of mutations in the homeodomain repressor HESX1 identifies separate functional domains. ESPE/LWPES 7th Joint Meeting Paediatric Endocrinology, Lyon, France.

33. Lin L, Rumsby G, Honour J, Techatraisak K, Hall C, Mushtaq I, Hakeem V, Taylor N, Butler G, Papari-Zareei M, **Dattani MT**, Auchus R, Achermann J (2005) A spectrum of phenotypes in 46, XY individuals with combined 17α-hydroxylase/17, 20-lyase deficiency. ESPE/LWPES 7th Joint Meeting Paediatric Endocrinology, Lyon, France.

34. Ward ST, Hindmarsh PC, **Dattani MT** (2006) Nursing involvement in the management of two children, presenting in infancy with clinical features of precocious puberty, both identified to have an underlying diagnosis of hypothalamic hamartoma. 88th Annual Meeting of the Endocrine Society, Boston, USA.

35. Kelberman D, Rajab A, Biebermann H, Shaikh H, Castro S, Pearce K, Turton JPG, Gerrelli D, Hall CM, Gruters A, Krude H, **Dattani MT** (2007) Intragenic deletion within

LHX3 associated with an unusual hypopituitary phenotype. 89th Annual Meeting of the Endocrine Society, Toronto, Canada.

36. Kelberman D, Rajab A, Biebermann H, Shaikh H, Castro S, Pearce K, Turton JPG, Gerrelli D, Hall CM, Gruters A, Krude H, **Dattani MT** (2007) Intragenic deletion within LHX3 associated with an unusual hypopituitary phenotype. 46th Annual Meeting of the European Society for Paediatric Endocrinology, Helsinki, Finland.

37. Alatzoglou KS, Spoudeas H, Ell P, Hindmarsh PC, **Dattani MT** (2007) Retesting of GHD patients: more questions than answers? 46th Annual Meeting of the European Society for Paediatric Endocrinology, Helsinki, Finland.

38. **Dattani MT**, Patel A, Hindmarsh PC (2007) Cortisol circadian rhythms differ between children and adults. 46th Annual Meeting of the European Society for Paediatric Endocrinology, Helsinki, Finland.

39. Mehta A, Hindmarsh PC, Chong K, **Dattani MT** (2007) Structure-function relationships within the hypothalamo-pituitary axis in patients with and in those at risk of hypopituitarism. 46th Annual Meeting of the European Society for Paediatric Endocrinology, Helsinki, Finland.

40. Alatzoglou KS, Hindmarsh PC, Brain C, Torpiano J, **Dattani MT** (2008) Acanthosis nigricans and insulin sensitivity in patients with skeletal dysplasia. 90th Annual Meeting of the Endocrine Society USA, San Francisco, California, USA.

41. Kelberman D, Mundlos S, Grueters A, **Dattani MT** (2008) Polyalanine tract mutations within the transcription factor SOX3 are associated with variable hypopituitarism. 90th Annual Meeting of the Endocrine Society USA, San Francisco, California, USA. **Awarded Best Poster Prize in section.**

42. Turton JPG, LeTissier PR, **Dattani MT** (2008) Severe Combined Pituitary Hormone Deficiency (CPHD) associated with compound heterozygosity for two novel loss of function mutations within POU1F1. 90th Annual Meeting of the Endocrine Society USA, San Francisco, California, USA.

43. Turton JPG, **Dattani MT**, Le Tissier PR (2008) Demonstration of dominant negative activity of POU1F1 mutations in the GH3 pituitary-derived cell line. 90th Annual Meeting of the Endocrine Society USA, San Francisco, California, USA.

44. Webb EA, **Dattani MT**, Hindmarsh PC, Gadian D, Fischl B, Singhal A, Lucas A, Isaacs E (2008) Relationship between IGF1, IGFBP3 and cognition in a cohort of preterm individuals. 90th Annual Meeting of the Endocrine Society USA, San Francisco, California, USA.

45. Alatzoglou KS, Turton JPG, Savage MO, Raza J, Banerjee K, Sinha S, Ten S, Cheetham T, **Dattani MT** (2008) Novel mutations in GHRHR associated with Type 1B IGHD. 47th Annual Meeting of the European Society for Paediatric Endocrinology, Istanbul, Turkey.

46. Alatzoglou KS, Hindmarsh PC, Brain C, Torpiano J, **Dattani MT** (2008) Acanthosis nigricans and insulin sensitivity in patients with skeletal dysplasia. 47th Annual Meeting of the European Society for Paediatric Endocrinology, Istanbul, Turkey.

47. Peters CJ, Spada A, Korbonits M, White A, Powell MP, **Dattani MT** (2009) Macroprolactinoma associated with aggressive Cushing's disease in a 12 year old child. 91st Annual Meeting of the Endocrine Society USA, Washington, USA.

48. Padidela R, Al-Ali N, Schoenmakers E, Agostini M, Rajanayagam O, **Dattani MT**, Chatterjee VKK (2009) A multisystem disorder associated with defective selenoprotein synthesis and a thyroid signature. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

49. O'Reilly M, Webb E, Dale N, Salt A, **Dattani M** (2009) Growth hormone deficiency is associated with selective deficits of memory and executive function in children. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

50. O'Riordan SMP, Arlt W, Dattani M (2009) P450 Oxidoreductase deficiency at puberty in a 46 XY individual with Antley-Bixler syndrome. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

51. Alatzoglou KS, Mehta A, Webb EA, **Dattani MT** (2009) Comparison of the spectrum of endocrinopathies in children with severe midline defects. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

52. Alatzoglou AS, Kelberman D, Cowell CT, Arnhold IJP, Melo ME, Mundlos S, Schnabel D, Grueters A, **Dattani MT** (2009) Variability in the length of the polyalanine (PA) tract of SOX3 in patients with congenital hypopituitarism is associated with variable functional and phenotypic effects. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

53. Alatzoglou KS, Kelberman D, Buchanan C, **Dattani MT** (2009) Genetic screening for variability in regulatory regions of SOX2 and implications for hypothalamo-pituitary development. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

54. Alatzoglou KS, Mohan R, Ward S, Bridges N, Hindmarsh PC, **Dattani MT** (2009) Outcome of rhGH treatment in patients with achondroplasia and skeletal dysplasias. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

55. Christesen HBT, Brusgaard K, Thisted E, Turton J, Hertel T, Johansen KB, Hansen D, **Dattani MT** (2009) Ten family members with a growth hormone gene mutation and treatment with growth hormone since 1976. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

56. Kosta K, Gkourogianni A, Alatzoglou KS, Hindmarsh PC, **Dattani MT** (2009) Investigation of premature adrenarche reveals a high incidence of congenital adrenal hyperplasia (CAH). 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

57. Panagiotakopoulos L, Webb EA, Ebstein DJ, **Dattani MT** (2009) How commonly are mutations in the sonic hedgehog signaling pathway found in individuals with septo-optic dysplasia and holoprosencephaly. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

58. Gevers EF, **Dattani MT**, Waters MJ, Robinson ICAF (2009) Growth hormone (GH) signaling in early life in rodents. 8th Joint meeting of the Lawson-Wilkins Pediatric

Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

59. Gozzi T, Tonella P, Scheidegger U, Fluck CE, L'Allemand D, **Dattani MT**, Hindmarsh PC, Mullis PE (2009) Do centimeters matter: inaccuracy of self-reported and estimated height measurements in parents? 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

60. Webb EA, Mehta A, **Dattani MT** (2009) Can the phenotype of septo-optic dysplasia at presentation be used to predict the severity of associated hormonal abnormalities, developmental delay, obesity, sleep and behavioural disorders? 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

61. Alatzoglou KS, Arriazu MC, Crolla JA, Roubicek ME, **Dattani MT** (2009) Incomplete progress through puberty and a large cystic lesion in the hypothalamo-pituitary area in a patient with a heterozygous SOX2 deletion. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

62. Peters CJ, Langham SA, Mullis PE, **Dattani MT** (2009) The use of combined liothyronine and thyroxine therapy for consumptive hypothyroidism associated with hepatic haemangiomas in infancy. 8th Joint meeting of the Lawson-Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, New York, USA.

63. Alatzoglou KS, Arriazu MC, Crolla JA, Roubicek ME, Buchanan C, **Dattani MT** (2010) First report of SOX2 loss of function associated with a large hypothalamo-pituitary tumour; further insights into the role of SOX2 in pituitary development. SfE BES Annual meeting, Manchester, UK.

64. Alatzoglou KS, Kelberman D, Buchanan C, **Dattani MT** (2010) Genetic screening for variability in regulatory regions of SOX2 and implications for hypothalamo-pituitary development. SfE BES Annual meeting, Manchester, UK.

65. Idkowiak J, O'Riordan S, Reisch N, Dhir V, Malunowicz EM, Kerstens M, Maiter D, Collins F, Sillink M, Dattani M, Shackleton CHL, Krone N, Arlt W (2010) Pubertal presentation of P450 oxidoreductase deficiency during puberty. SfE BES Annual meeting, Manchester, UK

66. Gevers E, **Dattani MT** (2010) Growth hormone induces hepatic stat5 phosphorylation in early postnatal life in mice. 92nd Annual Meeting of the Endocrine Society USA, San Diego, California, USA.

67. McCabe MJ, Tziaferi V, Gaston-Massuet C, Gregory LC, Walker J, Tsai P, Pitteloud N, Martinez-Barbera JP, **Dattani MT** (2010) Mutations in the gene encoding Fibroblast Growth Factor 8, FGF8, are associated with complex midline and hypothalamo-pituitary defects. 92nd Annual Meeting of the Endocrine Society USA, San Diego, California, USA.

68. Idkowiak J, O'Riordan S, Malunowicz EM, Collins F, Kerstens M, Reisch N, Szarras-Czapnik M, Maiter D, Sillink M, Dattani M, Shackleton CHL, Krone N, Arlt W (2010) Pubertal presentation in congenital adrenal hyperplasia due to P450 oxidoreductase deficiency. 92nd Annual Meeting of the Endocrine Society USA, San Diego, California, USA.

69. Gregory LC, McCabe MJ, Mutair A, Darzy K, **Dattani MT** (2010) Novel PROP1 mutations in patients with combined pituitary hormone deficiency (CPHD). 49th Annual Meeting of the European Society for Paediatric Endocrinology, Prague, Czechoslovakia.

70. Logan K, Peters C, Hindmarsh P, Dattani M (2011) Optimisation of treatment in children with 21-hydroxylase deficiency using cortisol profiling. 38th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Manchester, UK.

71. Alatzoglou KS, Mohan R, Ward S, Bridges N, Brook CGD, Hindmarsh PC, **Dattani MT** (2011) Outcome of rhGH treatment in patients with achondroplasia and skeletal dysplasia. 38th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Manchester, UK.

72. Ikazoboh EC, Spoudeas HA, **Dattani MT** (2011) Endocrine, hypothalamic and neurodevelopmental outcomes following treatment for craniopharyngiomas. 38th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Manchester, UK.

73. Dias RP, Hakeem V, Peters C, Baker J, Hindmarsh PC, **Dattani MT** (2011) An unusual spectrum of phenotype in autoimmune polyendocrinopathy syndrome type 1: a case series of 5 patients within a single centre. 38th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Manchester, UK.

74. Andoniadou CL, Gaston-Massuet C, Le Tissier P, Dattani M, Martinez-Barbera JP (2011) Cancer stem cells with a stabilising mutation in beta-catenin are implicated in the aetiology of human adamantinomatous craniopharyngioma. Annual meeting of the British Endocrine Societies, Birmingham, UK.

75. Alatzoglou KS, Kelberman D, Spadoni E, Gaston-Massuet C, Woods K, Maghnie M, Bitner-Glindzicz M, **Dattani MT** (2011) Wide range of eye abnormalities in patients with hypopituitarism; implications for diagnosis and treatment. Annual meeting of the British Endocrine Societies, Birmingham, UK.

76. Jayakody SA, Andoniadou CL, Signore M, Gaston-Massuet C, Pevny L, Dattani M, Martinez-Barbera JP (2011) Conditional deletion of Sox2 leads to pituitary hypoplasia and abnormal terminal differentiation of the Pit1 cell lineage. 93RD Annual Meeting of the Endocrine Society USA, Boston, USA.

77. Gregory LC, Webb EA, Panagiotakopoulos L, **Dattani MT** (2011) Mutations in the Sonic Hedgehog signalling pathway in patients with hypopituitary phenotypes. 93RD Annual Meeting of the Endocrine Society USA, Boston, USA.

78. Andoniadou CL, Gaston-Massuet C, Le Tissier P, **Dattani MT**, Martinez-Barbera JP (2011) Understanding the cellular origin of pituitary tumours: isolation and characterization of putative tumorigenic progenitors/stem cells from a mouse model of human adamantinomatous craniopharyngioma. 93RD Annual Meeting of the Endocrine Society USA, Boston, USA.

79. Gevers EF, Tziaferi V, Ravindranathan B, **Dattani MT** (2011) Pituitary masses in children and young people – to operate or not? 93RD Annual Meeting of the Endocrine Society USA, Boston, USA.

80. Sharma G, Muller D, Dattani M, O'Riordan S, Hindmarsh PC, Mills K (2011) Urinary conjugated metabolites of -tocopherol (Vitamin E) in children and young people with Type 1 Diabetes Mellitus. 93RD Annual Meeting of the Endocrine Society USA, Boston, USA.

81. El-Khairi R, Parnaik R, Lin L, **Dattani MT**, Conway GS, Achermann JC (2011) LIN28 in human ovary development and function. 93RD Annual Meeting of the Endocrine Society USA, Boston, USA.

82. Coletta RRD, Redington C, Webb E, **Dattani MT** (2011) Factors associated with obesity in children with septo-optic dysplasia. 93RD Annual Meeting of the Endocrine Society USA, Boston, USA.

83. Franca MM, Jorge AAL, Carvalho LR, Mendonca BB, Audi L, Carrascosa A, **Dattani MT**, Arnhold IJP (2011) Absence of GH-releasing hormone (GHRH) mutations in selected patients with isolated GH deficiency. 93RD Annual Meeting of the Endocrine Society USA, Boston, USA.

84. Alatzoglou KS, Andoniadou CL, Kelberman D, Kim HG, Botse-Baidoo E, Pedersen-White JR, Layman LC, Martinez-Barbera JP, **Dattani MT** (2011) Clinical manifestations of a novel SOX2 mutation may result from failure to repress -catenin-mediated target activation: suggestion for a new mechanism for the interaction between SOX2 and -catenin. 93RD Annual Meeting of the Endocrine Society USA, Boston, USA.

85. Webb E, Redington C, **Dattani MT** (2011) What does prolactin measurement add to the evaluation of pituitary hormone function? 50th Annual meeting of the European Society for Paediatric Endocrinology, Glasgow, UK.

86. Gregory L, Webb E, Panagiotakopoulos L, **Dattani MT** (2011) Mutations in the Sonic Hedgehog signalling pathway in patients with hypopituitary phenotypes. 50th Annual meeting of the European Society for Paediatric Endocrinology, Glasgow, UK.

87. Alatzoglou KS, Kelberman D, Spadoni E, Gaston-Massuet C, Woods K, Maghnie M, Bitner-Glindzicz M, **Dattani MT** (2011) Wide spectrum of eye abnormalities in patients with hypopituitarism; implications for diagnosis and treatment. 50th Annual meeting of the European Society for Paediatric Endocrinology, Glasgow, UK.

88. Alagaratnam S, Brain C, Spoudeas H, **Dattani MT**, Allgrove J, Hindmarsh PC Kurzawinski T, Van't Hoff W (2011) Surgical treatment of children with hyperparathyroidism: a single centre experience. 50th Annual meeting of the European Society for Paediatric Endocrinology, Glasgow, UK.

89. Ikazoboh E, **Dattani MT**, Spoudeas H (2011) Endocrine, hypothalamic and neurodevelopmental outcomes following treatment of craniopharyngioma. 50th Annual meeting of the European Society for Paediatric Endocrinology, Glasgow, UK.

90. Webb E, Clark C, Edmonds C, Isaacs E, Singhal A, Lanigan J, Lucas A, **Dattani MT** (2011) Serum IGFBP3 concentrations are associated with basal ganglia volumes in a cohort of normal children. Annual meeting of the European Society for Paediatric Endocrinology, Glasgow, UK.

91. Mutair M, Albanyan A, Suwaid A, Alkuraya F, Webb E, Kelberman D, **Dattani MT** (2011) Congenital Combined Pituitary Hormone Deficiency (CPHD), dysmorphic features; severe developmental delay, seizure disorder, blindness and neurogenic bladder: a new disorder. Annual meeting of the European Society for Paediatric Endocrinology, Glasgow, UK.

92. Tziaferi V, Gregory L, McCabe M, Spoudeas H, **Dattani MT** (2011) Analysis of Kallmann syndrome genes in a paediatric and adolescent cohort with HH. Annual meeting of the European Society for Paediatric Endocrinology, Glasgow, UK.

85. El-Khairi R, Parnaik R, Lin L, **Dattani MT**, Conway G & Achermann JC (2011) LIN28 in human ovary development and as a candidate gene for primary ovarian insufficiency. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

86. McCabe M, Gregory L, Hu Y, Thankamony A, Hughes I, Townshend S, Bouloux PM, **Dattani MT** (2011) Novel *KAL1* mutations associated with septo-optic dysplasia in three female patients. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

87. Gregory L, Webb EA, Panagiotakopoulos L, **Dattani** MT (2011) Mutations in the Sonic Hedgehog signalling pathway in patients with congenital hypopituitarism. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

88. Balapatabendi M, Fisher G, Browning M, Green M, **Dattani MT**, Greening J, O'Riordan SMO (2011) Diabetes insipidus, immunodeficiency and colitis in infancy. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

89. Webb E, Lazze P, Reddington C, **Dattani MT** (2011) What does prolactin measurement add to the evaluation of pituitary hormone function? 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

90. Kumaran A, Lazzeroni P, Brain C, Hussain K, Kapoor R, **Dattani M** (2011) CHARGE syndrome: experience of a tertiary Endocrine Centre. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

91. Tziaferi V, Spoudeas H, McCabe M, Gregory L, **Dattani MT** (2011) A case of familial isolated hypogonadotrophic hypogonadism due to FGFR1 G687R mutation. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

92. Prete FP, MorkaneC, Watkinson J, Shaw N, Alvi S, Squire R, Harrison B, Wales J, Clayton P, Morrison P, Carson D, Brain C, Hindmarsh P, **Dattani M**, Spoudeas H, Buchanan C, Albanese A, Amin R, Pierro A & Kurzawinski T (2011) Prophylactic thyroidectomy in children with MEN2 in the United Kingdom. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

93. Gevers E, Jacobs B, Flatt J, Bruce L, Grunewald S, **Dattani MT,** Stewart G (2011) (Pseudo)hyperkalaemia caused by stomatin deficient cryohydrocytosis due to GLUT1 deficiency. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

94. Gevers E, Kelberman D, Aylwin S, Buchanan C, Waterham H, **Dattani MT (**2011) Mevalonic aciduria in a pedigree with presumed GH-insensitivity. 39th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, London, UK.

95. McCabe-MJ, Hu Y, Gregory LC, Thankamony A, Hughes IA, Townshend S, Bouloux PM, **Dattani MT** (2012) Novel *KAL1* sequence variants associated with septo-optic dysplasia (SOD) in three female patients. Annual Meeting of the Endocrine Society USA, Houston, Texas, USA.

96. Alatzoglou KS, Turton JPG, Kelberman D, McCabe MJ, Gregory LC, Webb EA, McNay DEG, Woods KS, Mehta A, **Dattani MT** (2012) Genetic screening in a cohort of 2081 patients with congenital hypopituitarism; current knowledge and future directions. Annual Meeting of the Endocrine Society USA, Houston, Texas, USA.

97. McCabe MJ, Gaston-Massuet C, Gregory LC, Tziaferi V, Alatzoglou KS, Sbai O, Rondard P, Masumoto KH, Nagano M, Shigeyoshi Y, Pfeifer M, Hulse T, Buchanan C, Pitteloud N, Martinez-Barbera JP, **Dattani MT** (2012) Variations in *PROKR2*, but not *PROK2*, are associated with hypopituitarism and septo-optic dysplasia. Annual Meeting of the Endocrine Society USA, Houston, Texas, USA.

98. Gregory LC, Alatzoglou KS, McCabe MJ, Letissier P, **Dattani MT** (2012) Mild GH deficiency due to two novel homozygous mutations in the gene encoding Growth-Hormone Releasing Hormone receptor (*GHRHR*) in a single family. Annual Meeting of the Endocrine Society USA, Houston, Texas, USA.

99. Gregory LC, McCabe MJ, Gevers E, Baker J, Caimari M, **Dattani MT** (2012) Missense sequence variants in *CHD7* associated with hypopituitarism. Annual Meeting of the Endocrine Society USA, Houston, Texas, USA.

100. Andoniadou C, Gaston-Massuet C, Reddy R, Jacques T, **Dattani M**, Martinez-Barbera JP (2012) New insights into the molecular and cellular pathogenesis of human craniopharyngioma: do pituitary stem cells underlie the origin of these tumours. 15th International and 14th European Congress of Endocrinology (ICE/ECE 2012). **Winner of best basic science poster prize.**

101. Gregory LC, McCabe MJ, Alatzoglou KS, Letissier P, **Dattani MT** (2012) Mild GH deficiency due to two novel homozygous mutations in the gene encoding Growth-Hormone Releasing Hormone receptor (*GHRHR*) in a single family. 51st Annual Meeting of the European Society for Paediatric Endocrinology, Leipzig, Germany.

102. McCabe-MJ, Hu Y, Gregory LC, Thankamony A, Hughes IA, Townshend S, Bouloux PM, **Dattani MT** (2012) Novel *KAL1* sequence variants associated with septo-optic dysplasia (SOD) in three female patients. 51st Annual Meeting of the European Society for Paediatric Endocrinology, Leipzig, Germany.

103. Alatzoglou KS, Turton JPG, Kelberman D, McCabe MJ, Gregory LC, Webb EA, McNay DEG, Woods KS, Mehta A, **Dattani MT** (2012) Genetic screening in a cohort of 2030 patients with congenital hypopituitarism; current knowledge and future directions. 51st Annual Meeting of the European Society for Paediatric Endocrinology, Leipzig, Germany.

104. Hawton K, Raine J, **Dattani MT** (2012) The use of growth hormone and anastrozole can help optimize linear growth in congenital adrenal hyperplasia due to CYP11B1 mutations. 40th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Leeds, UK.

105. Senniapan S, Hakeem V, Wood D, Stoneham S, **Dattani MT** (2012) Gonadotropin-independent precocious puberty associated with later diagnosis of testicular embryonal carcinoma. 40th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Leeds, UK.

106. Gevers E, Shah P, Meredith S, Torpiano J, Slater O, White A, **Dattani M** (2012) Cushing syndrome due to POMC secretion from an abdominal yolk sac tumour in a two year old child. 40th Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Leeds, UK.

107. Subbarayan A, Dattani MT, Peters CJ, Hindmarsh PC (2013) Hypertension and obesity in children and adolescents with congenital adrenal hyperplasia due to 21-hydroxylase deficiency. 95th Annual Meeting of the Endocrine Society USA, San Francisco, USA – Featured Poster.

108. Gevers E, Shah P, Meredith S, Torpiano J, Peters CJ, Sebire N, Slater O, White A, **Dattani MT** (2013) Cushing syndrome due to POMC secretion from a malignant yolk sac tumor in a two year old child. 95th Annual Meeting of the Endocrine Society USA, San Francisco, USA.

109. Dunger DB, **Dattani MT**, Netchine I, Karres J, Tomasi P (2013) European Research network in diabetes and endocrinology. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy.

110. Gevers EF, Shah P, Meredith S, Torpiano J, Peters CJ, Sebire N, White A, Slater O, **Dattani MT** (2013) Cushing syndrome due to POMC secretion from a malignant yolk sac tumor in a two year old child. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy.

111. Logan KM, Boto L, Lazzeroni P, Hill NR, Peters CJ, Matthews DR, Charmandari E, Riepe FG, **Dattani MT**, Hindmarsh PC (2013) Definition of a practical sampling interval in 24h serum cortisol profiling to optimize treatment in children with 21-hydroxylase deficiency. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy.

112. Alatzoglou KS, McCabe MJ, Gregory LC, Spadoni E, Martinez-Barbera JP, Maghnie M, **Dattani MT** (2013) Contribution of *OTX2* mutations in the aetiology of congenital hypopituitarism in a selected cohort of patients: novel changes and functional consequences. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy.

113. Gregory L, Humayun K , McCabe MJ, Greening J, Rhodes SJ, Levy MJ, **Dattani MT** (2013) A novel homozygous *LHX4* mutation associated with severe panhypopituitarism leading to neonatal death. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy.

114. Gevers EF, Ahmed S, Logan K, Lazzeroni P, Hindmarsh PC, **Dattani MT** (2013) Optimization of hydrocortisone treatment in children with hypopituitarism using 24-hour serum cortisol profiling. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy.

115. Guemes M, Murray P, Brain CE, Peters CJ, Spoudeas HA, Hindmarsh PC, **Dattani MT** (2013) Cushing syndrome in children and adolescents: a retrospective review at presentation, diagnosis, management and outcome. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy.

116. Baranowski ES, Bahillo-Curieses P, Ehtisham S, Murray P, Achermann JC, **Dattani MT**, Hughes C, Taylor A, Parajes S, Krone N (2013) Functional analysis of novel mutations inactivating the HSD3B2 gene. 9th Joint Meeting of Paediatric Endocrinology, Milan, Italy.

117. Sherif MM, Hadeed I, Nessa A, Rahman SA, Arya VB, Senniappan S, **Dattani M**, Hussain K (2013) Two families with diabetes mellitus and sensorineural deafness.

118. Guemes M, Murray P, Brain CE, Peters CJ, Spoudeas HA, Hindmarsh PC, **Dattani MT** (2013) Reliability of diagnostic tests for paediatric Cushing syndrome. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Brighton, UK.

119. Ponmani C, Senniapan S, Schoenmakers N, Nicholas AK, Chatterjee K, **Dattani MT** (2013) Severe 21-hydroxylase deficiency Congenital Adrenal Hyperplasia and Congenital Hypothyroidism due to Thyroglobulin mutations: 2 distinct genetic disorders with phenotypic variability within a single family. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Brighton, UK.

120. Hughes C, **Dattani MT** (2013) Growth hormone neurosecretory dysfunction may be associated with structural abnormalities of the hypothalamic-pituitary axis. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Brighton, UK.

121. Alatzoglou KS, McCabe MJ, Gregory LC, Spadoni E, Martinez-Barbera JP, Maghnie M, **Dattani MT** (2014) Contribution of OTX2 Mutations in the Etiology of Congenital Hypopituitarism: Novel Changes and Functional Consequences. 96th Annual Meeting of the Endocrine Society, Chicago, USA.

122. Ponmani C, Atterbury A, Seniappan S, Schoenmakers N, Nicholas AK, Chatterjee K, **Dattani M** (2014) Severe 21-Hydroxylase Deficiency Congenital Adrenal Hyperplasia and Congenital Hypothyroidism due to Thyroglobulin Mutations in a Single Family: Two Distinct Genetic Disorders with Phenotypic Variability within a Single Family. 53rd Annual Meeting of the European Society for Paediatric Endocrinology, Dublin, Ireland.

123. Maris I, Howard C, Bogue C, Morrissey R, Gregory LC, O'Connell SM, **Dattani MT,** O'Riordan SMP (2014) Clinical Phenotype and Complications, Endocrinopathies and Neuroimaging Findings in a Case Series of SOD. 53rd Annual Meeting of the European Society for Paediatric Endocrinology, Dublin, Ireland.

124. Bhandari J, **Dattani MT**, Nanduri V (2014) Aldosterone synthase deficiency due to a novel mutation in *CYP11B2.* Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Winchester, UK.

125. Losa L, Beisti A, **Dattani M** (2014) Relationship beween IGF-1 concentration and growth velocity in infants and toddlers. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Winchester, UK.

126. Chesover AD, **Dattani MT** (2014) UK Growth Hormone Stimulation Test Survey Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Winchester, UK.

127. Gregory L, McCabe M, Bancalari R, Tziaferi V, Spoudeas H, **Dattani M** (2015) Genetic Screening of children and adolescents with Kallmann syndrome/Hypogonadotrophic Hypogonadism. 97th Annual meeting of the Endocrine Society USA, San Diego, USA.

128. Hufnagel RB, Arno G, Hein ND, Hersheson J, Prasad M, Anderson Y, Krueger LA, **Gregory LC**, Stoetzel C, Jaworek TJ, Hull S, Li A, Plagnol V, Willen C, Morgan TM, Prows CA, Hegde RS, Riazuddin S, Grabowski GA, Richardson RR, Dieterich K, Huang T, Revesz T, Martinez-Barbera JP, Sisk RA, Jefferies C, Houlden H, **Dattani MT**, Fink JK, Dollfus H, Moore AT, Ahmed ZM (2015) Neuropathy target esterase impairments cause Oliver-McFarlane and Laurence-Moon Syndromes. 97thAnnual meeting of the Endocrine Society USA, San Diego, USA.

129. Gevers E, Ahmed S, Logan K, Hindmarsh PC, **Dattani MT** (2015) Optimization of hydrocortisone treatment in children with hypopituitarism using 24 hour serum cortisol profiling. 97thAnnual meeting of the Endocrine Society USA, San Diego, USA.

130. Gan HW……**Dattani MT** (2015) Hypothalamic obesity, hyperphagia and hyperinsulinaemia: time for a paradigm shift in assumptions? 54TH Annual Meeting of the European Society for Paediatric Endocrinology, Barcelona, Spain.

131. Katumgopala H……**Dattani MT** (2015) Atypical features in patients with Leprechaunism suggesting a wide clinical spectrum of disease. 54TH Annual Meeting of the European Society for Paediatric Endocrinology, Barcelona, Spain. (MINI-POSTER)

132. Improda N,…….**Dattani MT** (2015) Recombinant human insulin-like growth factor 1 treatment in patients with insulin receptor mutations resulting in Donoghue syndrome: a 10 year experience in a tertiary centre. 54TH Annual Meeting of the European Society for Paediatric Endocrinology, Barcelona, Spain.

133. Kular D, Baker J, **Dattani MT** (2015) GH deficiency and phenotypic features in four cases of 22q11.2 deletion syndrome. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Sheffield, UK.

134. Gregory L,……Dattani MT (2016) Mutations in *BRAF* Are Associated with Septo-Optic Dysplasia and Cardiofaciocutaneous Syndrome. 98th Annual meeting of the Endocrine Society, Boston, USA.

135. Gregory L,……Dattani MT (2016) A Novel Mutation in Eukaryotic Translation Initiation Factor 2 Subunit 3 (*EIF2S3*) Is Associated with Severe Hypoglycaemia and X-Linked Hypopituitarism. 98th Annual meeting of the Endocrine Society, Boston, USA.

136. Gan HW, …….Dattani MT (2016) Oxytocin Deficiency Is Associated with Hyperphagia and Weight Gain in Hypothalamic and Common Obesity: Preliminary Data from a First-in-Humans Proof-of-Concept Study. 98th Annual meeting of the Endocrine Society, Boston, USA.

137. Monti E…….**Dattani MT** (2016) 5-alpha reductase deficiency: insights into the diagnosis and management of a rare condition. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Nottingham, UK.

138. Gregory L……..**Dattani MT** (2016) A mutation in eukaryotic translation initiation factor 2 subunit 3 (EIF2S3) associated with a novel syndrome of X-linked hypopituitarism and glucose dysregulation. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Nottingham, UK.

139. Morandi G, Rapti E, Alatzoglou S, Chong K, Shah P, Arya V, Dattani MT (2017) Long-term follow up of patients with holoprosencephaly in a single tertiary care centre. International Meeting of Paediatric Endocrinology, Washington, USA.

140. Walton-Betancourth S, Guemes M, Monte E, Buttigieg M, Torpiano J, Shah P, Dattani MT (2017) Is growth hormone insensitivity a cause of impaired growth in congenital tufting enteropathy? International Meeting of Paediatric Endocrinology, Washington, USA.

141. Caiulo S, Gan HW, Hughes C, Amin R, Spoudeas H, Peters C, Hindmarsh PC, Shah P, Dattani MT (2017) Growth hormone neurosecretory dysfunction as part of the spectrum of growth hormone deficiency disorders which benefit from growth hormone treatment. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Newcastle, UK.

142. Dastamani A, Kiff S, Guemes M, Gilbert C, Morgan K, Kapadia S, Iznaola CA, Caiulo S, De Coppi P, Dattani MT, Shah P (2017) Management and Clinical Outcomes of children with focal forms of Congenital Hyperinsulinism. International Meeting of Paediatric Endocrinology, Washington, USA.

143. Pradeep S, Guemes M, Dattani M, Shah P (2017) Congenital hypopituitarism and hyperinsulinaemic hypoglycaemia. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Newcastle, UK.

144. Caiulo S, Gan HW, Hughes C, Amin R, Spoudeas H, Peters C, Hindmarsh PC, Shah P, Dattani MT (2017) Growth hormone neurosecretory dysfunction as part of the spectrum

of growth hormone deficiency disorders which benefit from growth hormone treatment. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Newcastle, UK.

145. Gan HW, Leeson C, Aikenhead H, Farooqi S, Spoudeas HA, Dattani MT (2017) Quantification of appetite-regulating hormones in hypothalamic and simple obesity. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Newcastle, UK.

146. Monti E, ……..Dattani MT (2017) Can the TSH index be used as a predictor of central hypothyropidism in children? Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Newcastle, UK.

147. Macgloin H, Rangasami J, Dattani MT (2017) Albright's hereditary osteodystrophy associated with resistance to insulin and thyroid hormone in 3 male siblings. Annual Meeting of the British Society for Paediatric Endocrinology and Diabetes, Newcastle, UK.

148. Miller BS, Tan. JWL, Parween S, Eblé A, BMA, H, Ternand, C, Gregory LC, Dattani MT, Pandey AV (2018) Autosomal Dominant Growth Hormone Deficiency Due To A Novel c.178G&gtA Mutation In The gh1 Gene Causing Instability Of The Mutant GH Protein (p.Ala34Thr). Annual Meeting Endocrine Society USA, Chicago, USA

149. Gregory LC, Shah P, Sanner J, Arancibia M, Hurst J, Jones W, Spoudeas H, Le Quesne Stabej P, Ocaka L, Loureiro C, Martinez-Aguayo A, Williams H, Dattani MT. (2018) Mutations in *MAGEL2* and *L1CAM* are associated with congenital hypopituitarism and arthrogryposis. Annual Meeting Endocrine Society USA, Chicago, USA

150. Neyman A, Dattani MT, Kaefer M, Martinez A, Eugster E (2018) Use of a GnRH analog to suppress the mini-puberty of infancy in infants with disorders of sex development. Annual Meeting of the Pediatric Endocrine Society, USA.

151. Regis Coutant, Jordi Bosch Muñoz, Cristina Dumitrescu, Dirk Schnabel, Caroline Sert, Valerie Perrot, Mehul Dattani (2018) Year-one Effectiveness and Overall Safety of NutropinAq® for Growth Hormone Deficiency (GHD) and Other Paediatric Growth Disorders: Completion of the International Cooperative Growth Study (iNCGS) European Registry (2018). 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

152. Bradley Miller, Jimmy Tan, Shaheena Parween, Andree Eble, Christine Ternand, Louise Gregory, Mehul Dattani, Amit Pandey (2018) Autosomal Dominant Growth Hormone Deficiency due to a novel c.178G>A mutation in the GH1 gene causing instability of the mutant GH protein (p.Ala34Thr). 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

153. Hoong-Wei Gan, Clare Leeson, Helen Aitkenhead, Sadaf Farooqi, Helen Spoudeas, Mehul Dattani (2018) Towards a greater understanding of the pathophysiology of obesity: hypothalamic obesity as a model of dysregulation of appetite and metabolic homeostasis. 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

154. Antonia Dastamani, Neha Malhotra, Maria Guemes, Kate Morgan, Clare Rees, Mehul Dattani, Pratik Shah (2018) Post-prandial hyperinsulinaemic hypoglycaemia after oesophageal surgery in children. 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

155. Manuela Cerbone, Maria Güemes, Nicola Improda, Mehul T Dattani (2018) GROWTH PATTERN AND FINAL HEIGHT OUTCOME IN CHILDREN WITH

SEPTO-OPTIC DYSPLASIA AND ISOLATED HYPOPITUITARISM TREATED WITH rhGH IN A SINGLE CENTRE. 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

156. Katugampola H, Guemes M, Aftab S, Malhotra N, Morgan K, Bockenhuaer D, Dattani M, Shah P (2018) Nifedipine therapy in hyperinsulinaemic hypoglycaemia due to mutations in the PMM2 gene improves fast tolerance, stabilises blood glucose profile, and enables rationalisation of treatments for glycaemic control and hypertension: the first reported trial in 3 patients in a tertiary centre. 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

157. Alistair Calder, Antonia Dastamani, Helen Spoudeas, Mehul Dattani (2018) Validation of an automated method (BoneXpert) for the determination of bone age in paediatric endocrinology - A single centre experience. 57th Annual Meeting of the European Society for Paediatric Endocrinology, Athens, Greece.

158. Sommayya Aftab, Nicole Goff, Shirley Langham, Rakesh Amin, Peter Hindmarsh, Caroline Brain, Pratik Shah, Helen Spoudeas, Mehul Dattani, Austen Worth, Harshini Katugampola, Catherine Peters (2019) Thyroid dysfunction in patients following thymus transplantation in a tertiary centre: a 10-year experience. 58th Annual Meeting of the European Society for Paediatric Endocrinology, Vienna, Austria.

159. Elena Galazzi, Nicola Improda, Manuela Cerbone, Davide Soranna, Mirella Moro, Letizia Maria Fatti, Antonella Zambon, Mariacarolina Salerno, Mehul Dattani, Luca Persani (2019) ROLE OF PRIMING IN PERI-PUBERTAL GROWTH DELAYS: PRELIMINARY RESULTS OF A LARGE MULTICENTER STUDY. 58th Annual Meeting of the European Society for Paediatric Endocrinology, Vienna, Austria.

160. Prentice P, Bockenhauer D, Dattani MT (2019) Growth and Growth Hormone abnormalities in Bartter Syndrome types 3 and 4. 47th Annual Meeting of the British Society for Paediatric Endocrinology.

161. Mauri Carakushansky, Adriana Dankovčíková, Mehul Dattani, Larry A. Fox, Sonya Galcheva, Jan Lebl, Jane Loftus, Aristides Maniatis, Andrew Palladino, Gnanagurudasan Prakasam, Maria Resa, Carrie Turich Taylor (2021) Perception of Treatment Burden With Once Weekly Somatrogon vs Daily Genotropin in Pediatric Patients With Growth Hormone Deficiency: Results From a Randomized Phase 3 Study. Annual Meeting of the Endocrine Society 2021 (Online).

162. Danielle Whittaker, Roberto Oleari, Louise C. Gregory, Polona Le Quesne-Stabej, Hywel Williams, John Torpiano, Nancy Formosa, Mario J. Cachia, Daniel Field, Antonella Lettieri, Louise Ocaka, Lisa DeMartini, Sakina Rajabali, Kimberley L. Riegman, Alyssa J.J. Paganoni, Iain Robinson, Takahisa Furukawa, Anna Cariboni, M. Albert Basson, Mehul Dattani Recessive *PRDM13* mutations result in hypogonadotropic hypogonadism and cerebellar hypoplasia. Annual Meeting of the Endocrine Society 2021 (Online).

163. McGlacken-Byrne S, Gregory L, Roberts R, ClementsE, Wakeling E, Katugampola H, Dattani MT (2022) Use of the U.K. 100,000 Genomes Project to identify the genetic basis of childhood pituitary disorders within a tertiary paediatric endocrinology centre. Annual Meeting of BSPED 2022, Belfast.

164. Gregory LC, Krywawych S, Eaton S, Bitner-Glindzicz M, Rahman S, GOSGene, Clayton P, Robinson ICAF, Dattani M (2022) A novel missense variant in the gene encoding *Fatty Acid Synthase* (*FASN*) associated with a unique multi-system disorder including hypopituitarism and hypoparathyroidism. 60th Annual Meeting of the European Society for Paediatric Endocrinology, Rome, Italy.

165. Malhotra N, Cerbone M, Dattani M (2022) Spectrum of endocrinopathy in children with an ectopic posterior pituitary. 60th Annual Meeting of the European Society for Paediatric Endocrinology, Rome, Italy.

166. Shaunak M, Zichichi G, Peters CJ, Brain CE, Dattani M (2022) A single centre experience of aromatase inhibitors to limit bone age advancement in pre-pubertal boys with adrenal disorders causing androgen excess. 60th Annual Meeting of the European Society for Paediatric Endocrinology, Rome, Italy.

167. McGlacken-Byrne S, Gregory L, Roberts R, ClementsE, Wakeling E, Katugampola H, Dattani MT (2022) Use of the U.K. 100,000 Genomes Project to identify the genetic basis of childhood pituitary disorders within a tertiary paediatric endocrinology centre. 60th Annual Meeting of the European Society for Paediatric Endocrinology, Rome, Italy.

168. Dattani M, Dyer L, Friedman B, Hsiao J, Khadgawat R, Korth-Bradley J, Monica Nijher M, Roland C, Rosenfeld R, Taylor CT, Cara JF, Wajnrajch MP (2023) Clinical and Immunological Response to Somatrogon in Two Siblings with a Homozygous Whole Gene Deletion of the Growth Hormone 1 Gene. Annual Meeeting of the Endocrine Society USA.

169. Geffner ME, Maniatis A, Ibanez L, La Torre D, Huang C, Darendelieler F, Dattani M, Maghnie M, Phillip M, Horikawa R, Gomez R, Viswanathan S, Carlsson M, Wajnrajch MP (2023) PROGRES, a multi-country, non-interventional, prospective study of patients receiving human growth hormone treatment under routine clinical care: Study update. 61st Annual Meeting of the European Society for Paediatric Endocrinology, The Hague, Netherlands.

170. Cerbone M, D'Arco F, Spoudeas HA, Clark C, Dattani MT (2023) Application of novel clinical and imaging tools to identify and grade hypothalamic disease in populations at risk. 61st Annual Meeting of the European Society for Paediatric Endocrinology, The Hague, Netherlands.