15th Manchester Dysmorphology Conference

22nd – 25th October 2012

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Conference Organisers
Dian Donnai
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PROGRAMME

Monday 22nd October

From
3.00 pm Registration

6.30 pm SUPPER

Chairman - ***Karen Temple***

8.00 pm (1) Syndromology through the Retrospectoscope – another family with the distal ("atypical") 16p11.2 microdeletion
S McKee

8:15 pm (2) Dysmorphology at a distance: results of a web-based diagnostic service
Sofia Douzgou

8:30 pm (3) Early results from DDD study – UK-wide collaborative venture to decipher the genomic architecture of developmental disorders
Helen Firth

8:45 pm (4) Skeletome project: a community-driven knowledge curation platform for skeletal dysplasias
Andreas Zankl

9:00 pm 10 selected unknowns

Tuesday 23rd October

Chairman - ***Dian Donnai***

8:30 am (5) Schinzel-Giedion syndrome: defining the clinical spectrum
Bregje Van Bon

8:45 am (6) Cantú syndrome: clinical and skeletal features in 20 patients
Ingrid Scurr

9.00 am (7) Dominant missense mutations in ABCC9 cause Cantú syndrome
Mieke Van Haelst

9.15 am (8) De Novo mutations in MLL Cause Wiedemann-Steiner syndrome
Wendy Jones

9:30 am (9) Phenotypic spectrum and GPC3 mutations in a series of 41 patients with Simpson-Golabi-Behmel syndrome
Annick Toutain

9:45 am (10) Clinical and mutational spectrum of disease in Cornelia de Lange syndrome and associated phenotypes
Gemma Poke

10:00 am (11) Mutations in RAD21 and HDAC8 cause a Cornelia de Lange syndrome-like phenotype
Gabriele Gillessen-Kaesbach

10:15am **COFFEE**

Chairman – ***David Fitzpatrick***

10.45 am (12) Coffin-Siris and Nicolaides-Baraitser syndromes – clinical phenotypes of 43 previously unreported patients and mutational spectrum of the SWI/SNF complex
Dagmar Wieczorek

11.00 am (13) Coffin-Siris syndrome: a genotype and phenotype study
Gijs Santen

11:15 am (14) Exome sequencing reveals novel loss of function SMARCE1 mutations in patients with multiple spinal meningiomas
Bill Newman

11.30 am (15) Clinical and molecular review of 28 Myhre syndrome cases
Valerie Cormier-Daire

11:45 pm (16) Leri's pleonosteosis results from defective SMAD signalling
Sid Banka

12.00 pm (17) Recurrent de novo mutations in PACS1 cause defective cranial neural crest migration and define a new intellectual disability syndrome
Han Brunner

12:15 pm (18) Involvement of kinesin family members KIF44 and KIF5C in intellectual disability and synaptic function
Tjitske Kleefstra

12:30 pm **LUNCH**

Chairman - **Gunnar Houge**

1.45 pm (19) Smith-Magenis & Potocki-Lupski Syndromes. What have we learned?
Jim Lupski

2.30 pm (20) Increased sensitivity to DNA damage in a recessive form of Weaver syndrome caused by functional loss of an E3 ubiquitin ligase
Bernd Wollnik

2.45 pm (21) Loss of function mutations in TGFB2 cause Loeys-Dietz syndrome
Bart Loeys

3.00 pm	(22)	Wang syndrome (paternal UPD14-like phenotype): clinical spectrum, molecular pathology and prognosis ***Abhijit Dixit***
3.15 pm	(23)	6q24 transient neonatal diabetes mellitus (TNDM) – 16 years of data collection ***Karen Temple***
3:30 pm	(24)	Identification of a novel cause of urofacial (Ochoa) syndrome confirming genetic heterogeneity ***Helen Stuart***
3.45 pm	(25)	Genetics of primary lymphoedema – the story so far ***Sahar Mansour***
4:10 pm	**TEA & POSTER VIEWING (EVEN NUMBERS)**	
7.00 pm	**SUPPER AT THE PLACE (Transport details TBA)**	

Wednesday 24th October

Chairman - ***Koen Devriendt***

8:30 am (26) Mandibulofacial dysostosis with microcephaly caused by EFTUD2 mutations has a distinct and recognisable phenotype
Susan White

8:45 am (27) EFTUD2 haploinsufficiency leads to syndromic EA
Muriel Holder-Espinasse

9.00 am (28) Four new craniosynostosis syndromes identified by exome or whole genome sequencing
Andrew Wilkie

9.15 am (29) Heterogeneity of mutational mechanisms and modes of inheritance in auriculo-condylar syndrome
Jeanne Amiel

9:30 am (30) Specific phenotypes from ubiquitous/essential genes: lessons from the branchial arch and limb bud
David Fitzpatrick

9:45 am (31) Why is T2* so valuable for diagnosing progressive encephalopathy?
Nathalie Boddaert

10.00 am (32) Let us not be blind to optic atrophy
Arnold Munnich

10:15 am (33) LIG4 mutations are a common cause of microcephalic primordial dwarfism
Andrew Jackson

10:30 am **COFFEE**

Chairman - ***Willie Reardon***

11.00 am (34) The spectrum of conditions cause by mosaic and germline mutations in the AKT/PIK3CA/MTOR pathway: connecting malformations to cancer
Leslie Biesecker

11.15 am (35) A case of bilateral hemi-megalencephaly and comparison with the brain malformation in 5 other similar cases, a new sub-phentopye
Anand Saggar

11.30 am (36) Genotype-phenotype analysis of PI3K pathway genes in megalencephaly and related overgrowth syndromes
William Dobyns

11:45 pm (37) Aicardi-Goutieres syndrome: treating an autoimmune disease with antiretrovirals. Surely some mistake?
Yanick Crow

12.00 pm (38) 5H syndrome and the emergence of the phenotype of nucleolus disorders of ribosome biogenesis
Geoff Woods

12.15 pm (39) KAT6B related phenotypes
Jill Clayton-Smith

12:30 pm **LUNCH & POSTER VIEWING (odd numbers)**

3:00 pm **TEA**

Chairman - **Michael Wright**

3.15 pm (40) Clinicial spectrum and molecular pathogenesis of osteopathia striata with cranial sclerosis
Stephen Robertson

3.30 pm (41) Severe brachydactyly and absent ossification of several tubular bones in hands and feet as distinctive features of a rare skeletal dysplasia
Geert Mortier

3:45 pm	(42)	Spondyloepimetaphyseal dysplasia with abnormal dentition (SEMDAD) ***Mohnish Suri***
4.00 pm	(43)	A distinctive autosomal recessive osteocutaneous syndrome with unusual face caused by a mutation in POC1A ***Stavit Shalev***
4.15 pm	(44)	New short rib-polydactyly syndrome in humans and yeti mouse caused by mutations in WDR35 / Wdr35 and abnormal ciliogenesis ***Ravi Savarirayan***
4.30 pm	(45)	Investigation of the cellular pathways linking tartrate resistant acid phosphates (TRAP), type 1 interferon and human lupus ***Tracy Briggs***
4:45 pm	(46)	Recessive mutations in KIAA1632 cause Vici syndrome, a multisystem disorder with defective autophagy ***Shehla Mohammed***
7.00 pm		Coaches leave the Conference Centre and The Place for The Monastery
7.30 pm		**Drinks Reception**
8.00 pm		**CONFERENCE DINNER AT THE MONASTERY (also known as Gorton Monastery)**

Thursday 25th October

Chairman - ***Daniela Pilz***

8.30 am (47) Weaver syndrome: the clinical and mutational spectrum associated with EZH2 alterations
Kate Tatton-Brown

8.45 am (48) E3 ubiquitin ligase UBE3B is mutated in an autosomal-recessive blepharophimosis-intellectual disability syndrome
Lina Basel-Vanagaite

9.00 am (49) Exome sequencing in severe unspecific intellectual disability broadens phenotype of well-known disease entities
Anita Rauch

9.15 am (50) Making headway with the molecular and clinical definition of rare genetic disorders with intellectual disability
Marjolein Willemsen

9.30 am (51) Mutations in PIGO, a member of the GPI anchor synthesis pathway cause hyperphosphatasia with mental retardation
Denise Horn

9:45 am (52) A founder mutation in a transposable element impacts a long non-coding RNA in the autosomal recessive Ravine progressive encephalopathy
Stanislas Lyonnet

10.00 am (53) A de novo missense variant in PIEZO2, a mechanically activated cation channel, detected in a mother and son with arthrogryposis and agenesis of the cruciate ligaments
Gunnar Houge

10:30 am **COFFEE**

Chairman - ***Han Brunner***

11:00 am (54) Germline mutations in DIS3L2 cause the Perlman syndrome of overgrowth and cancer susceptibility
Eamonn Maher

11.15 am (55) The genetic base of TAR syndrome: compound inheritance of a low-frequency regulatory SNP and a rate null mutation / deletion
Ruth Newbury-Ecob

11.30 am (56) Split hand / split foot malformation (SHFM; ectrodactyly)
Peter Turnpenny

11.45 am (57) Clinical and molecular findings in patients with KBG syndrome
Natalie Canham

12:00 am (58) Mutations in KANSL1 cause the 17q21.31 microdeletion syndrome
Bert de Vries

12:15 am (59) Noonan-like syndrome with loose anagen hair: further delineation of the phenotype
Martin Zenker

12:30 pm **LUNCH & END OF MEETING**