17th Manchester Dysmorphology Conference

7th - 10th November 2016

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Dysmorphology Conference

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Conference Organisers
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PROGRAMME

Monday 7th November

From 3.30 pm		**Registration**
6.30 pm		**SUPPER**
Chairman –		**JILL CLAYTON-SMITH**
8.00 pm	(1)	Evidence of KPNA7 variants in Gomez-Lopez-Fernandez syndrome ***Sofia Douzgou***
8:10 pm	(2)	The phenotype resulting from mutations in *PUF60*-a regulator of the Spliceosome ***Karen Low***
8:20 pm	(3)	Chromatin modulator *TLK2*: a novel intellectual disability gene with a distinct phenotype ***Melissa Lees***
8:30 pm	(4)	Further delineation of the phenotypic spectrum seen in patients with mutations in the LARP7 gene, (Alazami syndrome) ***Kate Chandler***
8:40 pm	(5)	The clinical phenotype due to mutation in Histone 4 ***Jane Hurst***
8:50 pm	(6)	A new mitochondrial disorder with effects ranging from neonatal demise to sudden death after ingesting small amounts of alcohol ***Angus Dobbie***
9:00 pm	(7)	Disruption of POGZ is associated with intellectual disability and autism spectrum disorders ***Marjolein Willemsen***
9:10 pm	(8)	Mitochondrial dysfunction results in Perrault syndrome (sensorineural hearing loss and premature ovarian Insufficiency) ***William Newman***

Tuesday 8th November

Chairman – GUNNAR HOUGE

9.00 am	(9)	RNA processing defects and polyadenylation site mutations in *NAA10* Cause X-linked anophthalmia ***Leslie Biesecker***
9.15 am	(10)	De novo gain-of-function mutations in the epigenetic regulator *SMCHD1* cause Bosma arhinia microphthalmia syndrome ***Jeanne Amiel***
9:30 am	(11)	Mutations in *MYT1*, encoding the myelin transcription factor 1, are a rare cause of Goldenhar syndrome within the RA signaling pathway ***Didier Lacombe***
9:45 am	(12)	Matrix metallopeptidase 21 *(MMP21)* is mutated in human heterotaxy and is an essential determinant of vertebrate left-right asymmetry ***Stanislas Lyonnet***
10:00 am	(13)	Gillespie Syndrome Unravelled ***Frances Elmslie***
10:15 am	(14)	The challenge of translating NGS from bench to medical genetic service ***Arnold Munnich***
10:30 am	**COFFEE**	

Chairman – DIAN DONNAI

11.00 am	(15)	Heterozygous mutations in MAP3K7 coding for transforming growth factor β activated kinase 1 cause cardiospondylocarpofacial syndrome ***Valérie Cormiere-Daire***
11:15 am	(16)	Molecular Mechanisms in Cerebro-Costo-Mandibular syndrome and other craniofacial disorders caused by spliceosomal defects ***Madeleine Tooley***
11.30 am	(17)	Homozygous nonsense variant in *LTBP1* is associated with a syndrome of craniosynostosis, cleft palate, brachydactyly, dysmorphism and learning disability ***Kay Metcalfe***
11:45 pm	(18)	Biallelic mutations in CDC45 cause a phenotypic spectrum ranging from Meier-Gorlin syndrome through to syndromic coronal craniosynostosis ***Louise Wilson***
12:00 pm	(19)	Multiple rare actionable mutations identified by whole exome or genome sequencing of patients with craniosynostosis ***Andrew Wilkie***
12.15 pm	(20)	RSPONDIN2 Drives Lung and Limb Development independently of LGR4/5/6 ***Bruno Reversade***
12:30 pm	**LUNCH**	

Chairman – SIDDHARTH BANKA

2.00 pm	(21)	Intellectual disability and altered brain size in patients with activating mutations in mTOR pathway genes ***Han Brunner***
2.15 pm	(22)	Two novel EIF2S3 mutations associated with X-linked syndromic intellectual disability with severe microcephaly, growth retardation and epilepsy ***Stéphanie Moortgat***
2:30 pm	(23)	Life with a 5th nucleotide: *ITPA* deficiency causes Martsolf syndrome with dilated cardiomyopathy ***David Fitzpatrick***
2:45 pm	(24)	*UNC80* mutations lead to a recognizable syndrome with persistent hypotonia, encephalopathy, severe intellectual disability and postnatal growth retardation ***Trine Prescott***
3:00 pm	(25)	Mutations in FZD3 cause a novel recessive syndrome with severe hydrocephalus and other brain malformations, limb contractures and variable cystic dysplastic kidneys cause a novel ***Cynthia Curry***
3.15 pm	(26)	Is PEHO distinct enough to be a “syndrome” or is it just a “phenotype”? ***Geoff Woods***
3:30 pm		**TEA & POSTER VIEWING (EVEN NUMBERS)**
6-8pm		**Manchester Art Gallery Visit** **Drinks/Canapes/Viewings**

Wednesday 9th November

Chairman – KATE TATTON-BROWN

9.00 am	(27)	Leucine as a targeted mTOR pathway therapy for *TBCK*-related intellectual disability, a novel progressive syndrome of hypotonia, developmental delay, and dysmorphic facial features ***Elizabeth Bhoj***
9.15 am	(28)	Biallelic mutations in the Integrator complex subunits genes *INTS1* and *INTS8* associated with intellectual disability and brain developmental disorder ***Grazia Mancini***
9:30 am	(29)	Heterozygous mutations in the protein kinase domain of *CDK13* cause a syndromic form of intellectual disability ***Mohnish Suri***
9:45 am	(30)	A genotype-first approach identifies gain-of-function mutations of *TFE3* in a novel syndrome with intellectual disability, seizures, facial dysmorphism,short stature and obesity ***Daphné Lehalle***
10.00 am	(31)	Synaptotagmin-1 mutation is a recurrent disorder of neurotransmitter release ***Kate Baker***
10:15 am	(32)	Reverse phenotyping of novel genes for Intellectual Disability: from TRIO to PPM1D ***Bert de Vries***
10:30 am	**COFFEE**	

Chairman – TJITSKE KLEEFSTRA

11.00 am	(33)	Phenotype and natural history in 101 individuals with Pitt-Hopkins syndrome through an internet questionnaire system ***Emilia Bijlsma***
11.15 am	(34)	Longitudinal clinical information on 119 individuals with Koolen-de Vries syndrome entered and updated by the family of the affected individual in the GenIDA patient registry ***David Koolen***
11.30 am	(35)	Characterizing The Morbid Genome of Ciliopathies ***Fowzan Alkuraya***
11:45 pm	(36)	Novel phenotypes associated with mutations in the KAT6A gene ***Ruth Newbury-Ecob***
12.00 pm	(37)	Lessons from gene panel analysis in 1,000 patients with early-onset seizure and severe developmental delay disorders ***Richard Scott***
12.15 pm	(38)	The Genomic Architecture of Developmental Disorders: Insights for clinical practice from analysis of ~8000 trios in the DDD study ***Helen Firth***
12:30 pm	**LUNCH**	

Chairman – WILLIE REARDON

2:00 pm	(39)	Evidence that bi-allelic mutations in NPR3 result in a peculiar phenotype with tall stature, arachnodactyly, long halluces and multiple extra epiphyses in hands and feet ***Geert Mortier***
2:15 pm	(40)	From fibrous overgrowth to connective tissue destruction: The pterygiae-fibroma to lipodystrophy-acroosteolysis spectrum caused by gain-of function PDGFRB mutations ***Gunnar Houge***
2:30 pm	(41)	Vosoritide for children with achondroplasia: updates from an ongoing phase 2 clinical trial ***Ravi Savarirayan***
2:45 pm	(42)	Mutations in genes encoding the condensin complexes cause microcephaly through decatenation failure at mitosis ***Jennie Murray***
3:00 pm	(43)	ARCN1 mutations in a dominant form of microcephalic dwarfism ***Sandrine Passemard***
3:15 pm	(44)	Genome stability genes for primordial dwarfism ***Andrew Jackson***
3:30 pm		**POSTER VIEWING (odd numbers)**

Chairman – MICHAEL WRIGHT

4:00 pm	(45)	*ZMYND11* case series: delineating the phenotype and genotype-phenotype correlations ***Michael Parker***
4.15 pm	(46)	A Genetic and Phenotypic Study of 78 Individuals with Wiedmann-Steiner syndrome and a wider study of hypertrichosis ***Wendy Jones***
4.30 pm	(47)	Rare non-coding mutations extend the mutational spectrum in the *PGAP3* subtype of Hyperphosphatasia with Mental Retardation syndrome ***Denise Horn***
4:45 pm	(48)	Impaired Kennedy pathway phospholipid biosynthesis due to *EPT1* mutation underlies a complex form of hereditary spastic paraplegia ***Emma Baple***
5:15 pm	(49)	Pathogenic variants in *HTRA2*, *RTN4IP1* and *CHCHD10* genes cause distinct clinical phenotypes associated with mitochondrial dysfunction ***Charu Deshpande***

7.30 pm **CONFERENCE DINNER AT THE PLACE**

Thursday 10th November

Chairman – RUTH NEWBURY-ECOB

9.00 am	(50)	Lymphatic –Related Hydrops Fetalis: two new genetic disorders ***Sahar Mansour***
9.15 am	(51)	Loss of function mutations in Carboxypeptidase D cause a new syndrome with recognizable dysmorphisms, lymphedema and sensorineural hearing loss ***Umut Altunoglu***
9.30 am	(52)	Mutations in the box C/D snoRNA U8 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts ***Yanick Crow***
9:45 am	(53)	The deubiquitinase OTULIN is an essential negative regulator of inflammation and autoimmunity ***Eamon Maher***
10.00 am	(54)	Human USP18 deficiency underlies Type 1 interferonopathy leading to severe pseudo-TORCH syndrome ***Marije Meuwissen***
10:15 am	(55)	Loss-of-function mutations in the X-linked gene BGN cause a severe syndromic form of thoracic aortic aneurysms and dissections ***Bart Loeys***
10:30 am	**COFFEE**	

Chairman – HAN BRUNNER

11:00 am	(56)	Mutations in KDM3B cause intellectual disability and are associated with genetic cancer predisposition ***Illja Diets***
11.15 am	(57)	Phenotype, Cancer Risks and Surveillance in Beckwith-Wiedemann Syndrome Depending on Molecular Genetic Subgroups ***Saskia Maas***
11.30 am	(58)	Multilocus imprinting disorders and new imprinting phenotypes ***Karen Temple***
11.45 am	(59)	Truncating mutations on the paternal allele of *MAGEL2*, a gene within the Prade-Willi locus, cause Schaaf-Yang syndrome ***Emmelien Aten***
12:00 am	(60)	Isolated PREPL deficiency, a differential diagnosis for patients with a Prader-Willi-like phenotype ***Isabelle Maystadt***
12:15 am	(61)	Duplicated enhancer region upstream of the *CTSB* gene segregates with Keratolytic Winter Erythema in South African and Norwegian families ***Torunn Fiskerstrand***
12:30 pm	**LUNCH & END OF MEETING**	