
MDC'18
18TH MANCHESTER DYSMORPHOLOGY CONFERENCE · 5-8 NOVEMBER 2018
DIAN DONNAI
RINCIPA

18th Manchester Dysmorphology Conference

5 – 8th November 2018

Conference Organisers
Jill Clayton-Smith
Sofia Douzgou
Siddharth Banka

PROGRAMME

MONDAY 5TH NOVEMBER

From 3.00 pm **Registration – Ballroom entrance**

Ballroom

Chairs JULIE MCGAUGHRAN and HELEN STUART

5:30pm	(A1)	Further delineation of the MECP2 duplication syndrome phenotype in 59 French male patients, with a particular focus on morphological and neurological features ***Salima El Chehadeh***
5:45pm	(A2)	TCF20 loss-of-function mutations associated with intellectual disability - a study of 19 patients identified by whole exome sequencing through the DDD Study ***Shane McKee***
6:00pm	(A3)	Type 1 interferonopathy due to germline STAT2 gain-of-function mutation ***Tracy Briggs***
6:15pm	(A4)	Delineation of phenotypes associated with TBL1XR1 mutations ***Diana Johnson***
6:30pm	(A5)	De novo missense variants in TUBA1A – whole exome sequencing in a mixed neurodevelopmental cohort expands the genotypic and phenotypic spectrum ***Abhijit Dixit***
6:45pm	(A6)	PCYT2 mutations cause an early-onset complex hereditary spastic paraplegia syndrome highlighting the importance of the Kennedy pathway ***John McDermott***

7:00pm	(A7)	Further delineation of the Aarskog-Scott syndrome phenotype in a series of 79 male patients with a FGD1 mutation: recommendations for diagnosis and management ***Annick Toutain***
7:15pm		**SUPPER - The Whitworth Suite**
8:15pm		Unknowns' session

Chairs JILL CLAYTON-SMITH and SOFIA DOUZGOU

TUESDAY 6TH NOVEMBER

Ballroom

Chairs MOHNISH SURI and ALAIN VERLOES

9:00am	(B1)	Delineating the clinical spectrum due to heterozygous TRAF7 missense mutations: A series of 24 cases ***Jeanne Amiel***
9:15am	(B2)	From Amelia to Polymelia ***Bruno Reversade***
9:30am	(B3)	Non-syndromic craniosynostosis: mutations in PRRX1, SIX1 and SMAD6 ***Andrew Wilkie***
9:45am	(B4)	Germline loss-of-function mutations in EPHB4 cause a second form of capillary malformation-arteriovenous malformation (CM-AVM2) deregulating RAS-MAPK signaling ***Nicole Revencu***
10:00am	(B5)	Unraveling the genetic architecture of bladder exstrophy epispadias complex ***William Newman***
10:15am	(B6)	The genetics of Hirschsprung disease: transethnic analyses of isolated cases and new candidate gene for syndromic cases. ***Stanislas Lyonnet***

10:30am **COFFEE - The Whitworth Suite**

Ballroom

Chairs LAURENCE FAIVRE and LUCY RAYMOND

11:00am	(C1)	Prioritising genes of interest from whole genome sequences to maximise diagnostic yield in developmental disorders in the 100,000 Genomes Project ***Richard Scott***
11:15am	(C2)	NIPT as first screening of trisomies 13, 18 and 21 in a genetic center in Belgium: retrospective study of 12 060 tests ***Colombine Meunier***
11:30am	(C3)	Accurate molecular detection and functional validation of PI3K-AKT-MTOR pathway in focal malformations of cortical development: novel genetic insights ***Ghayda Mirzaa***
11:45am	(C4)	The landscape of pathogenic copy number variations in healthy, reproducing females. Results from 1 year genome-wide noninvasive prenatal testing ***Koenraad Devriendt***
12:00pm	(C5)	The DDD study – Delivering Discovery & Diagnosis in rare disease ***Helen Firth***
12:15pm	(C6)	What Have We Learned from The First 2,500 Diagnostic Exomes in Saudi Arabia? ***Fowzan Alkuraya***
12:30pm		**LUNCH - The Whitworth Suite**
1:30pm		**POSTER VIEWING (ODD NUMBERS)**

Ballroom

Chairs TJITSKE KLEEFSTRA and SIDDHARTH BANKA

3:00pm	(D1)	Examination of the landscape of histone lysine methylases and demethylases in human developmental disorders leads to identification of novel syndromes ***Victor Faundes***
3:15pm	(D2)	Phenotypic spectrum of novel intellectual disability syndrome due to de novo variants in KMT2E ***Anne O'Donnell-Luria***
3:30pm	(D3)	Further delineation of the phenotypic spectrum of the KAT6B gene ***Ruta Marcinkute***
3:45pm	(D4)	Missense mutations disrupting the ATPase domain of CHD3 cause a novel neurodevelopmental syndrome with intellectual disability, macrocephaly and impaired speech and language ***Sandra Whalen***
4:00pm	(D5)	A recurrent de novo missense variant in SMARCB1 causes severe intellectual disability and hydrocephalus due to choroid plexus hyperplasia ***Trine Prescott***
4:15pm	(D6)	Constitutive HIST1H1E variants and an emerging phenotype characterised by a distinctive facial gestalt and an intellectual disability ***Kate Tatton-Brown***
4:30pm	(D7)	De novo germline variants in Histone 3 Family 3A (H3F3A) and Histone 3 Family 3B (H3F3B) cause a severe neurodegenerative disorder and functional effects unique from their somatic mutations ***Elizabeth Bhoj***

4:45pm	(D8)	Gain of function DNMT3A mutations cause microcephalic dwarfism and hypermethylation of Polycomb-regulated regions ***Andrew Jackson***
5:00pm	**End of session**	
7:00pm	**DINNER – COPPER FACE JACKS PUB (BASEMENT)**	

WEDNESDAY 7TH NOVEMBER

Ballroom

Chairs GEERT MORTIER and BRUNO REVERSADE

9:00am	(E1)	Frontometaphyseal dysplasia: Expanding Phenotypic, Locus and Allelic Heterogeneity ***Stephen Robertson***
9:15am	(E2)	SLC10A7 mutations in human and mouse cause a skeletal dysplasia with amelogenesis imperfecta mediated by GAG biosynthesis defects ***Valerie Cormier-Daire***
9:30am	(E3)	A recurrent de novo heterozygous COG4 variant leads to Saul-Wilson syndrome, disruption of vesicular trafficking, perturbed Golgi structure and altered glycosylation of a secreted proteoglycan ***Carlos Ferreira***
9:45am	(E4)	SPONASTRIME Dysplasia is caused by biallellic mutations in TONSL ***Melissa Lees***
10:00am	(E5)	The Homozygous Variant c.797G>A/p.(Cys266Tyr) in PISD is Associated with a Spondyloepimetaphyseal Dysplasia with Large Epiphyses and Disturbed Mitochondrial Function ***Kerstin Kutsche***
10:15am	(E6)	Drug repurposing for rare diseases- MCDS therapy, a phase I/II trial of carbamazepine in patients with Metaphyseal Dysplasia Type Schmid as an example. ***Michael Wright***

10:30am **COFFEE- The Whitworth Suite**

Ballroom

Chairs DANIELA PILZ and ANITA RAUCH

11:00am	(F1)	Eyes in a Dish: Modelling Eye Malformations Using Optic Vesicle Organoid Culture ***David FitzPatrick***
11:20am	(F2)	RAB18 Micro syndrome mechanism ***Eamonn Sheridan***
11:35am	(F3)	Deep exploration of a CDKN1C mutation causing a mixture of Beckwith-Wiedemann and IMAGe syndromes revealed a novel transcript associated with developmental delay ***Siren Berland***
11:45am	(F4)	Non-penetrance of a frameshifting SHANK3 deletion is associated with compensatory mechanisms in both alleles ***Gunnar Houge***
11:55am	(F5)	A targeted therapeutic for Proteus syndrome - preliminary results ***Leslie Biesecker***
12:10Pm	(F6)	Targeted therapy in patients with PIK3CA-related overgrowth syndrome ***Guillaume Canaud***
12:30pm		**LUNCH - The Whitworth Suite**
1:30pm		**POSTER VIEWING (EVEN NUMBERS)**

Ballroom

Chairs DIAN DONNAI and GIJS SANTEN

3:00pm	(G1)	Next Generation Phenotyping using DeepGestalt in Clinic and Research ***Karen Gripp***
3:25pm	(G2)	Computer-assisted Image Analysis in Exome Diagnostics ***Peter Krawitz***
3:50pm	(G3)	Clinical Phenotyping of Bohring-Opitz Syndrome Using a Registry and Facial Photographs ***Wen Hann-Tan***
4:00pm	(G4)	Characterization of Glycosylphosphatidylinositol Biosynthesis Defects by Clinical Features, Flow Cytometry, and Automated Image Analysis ***Denise Horn***
4:15pm	(G5)	Next generation phenotyping of 7,697 individuals with intellectual disability ***Bert de Vries***
4:30pm	(G6)	Nuclear factor one (NFI) deficiency – a new family of overlapping developmental disorders ***Martin Zenker***
4:45pm	(G7)	Back to the future-a clinical diagnosis is the key to a rapid answer in urgent genomic analysis ***Jane Hurst***
5:00pm	**End of session**	
7:00pm	**CONFERENCE DINNER – Ballroom**	

THURSDAY 8TH NOVEMBER

Ballroom

Chairs JULIA RANKIN and ANDREW FRY

9:00am	(H1)	Genetic studies of cases with Bohring Opitz , Bohring Opitz like and Crisponi syndrome associated with ASXl1 and KLHL7 mutations ***Ruth Newbury-Ecob***
9:15am	(H2)	Gating-affecting mutations in KCNK4cause a recognizable neurodevelopmental syndrome ***Francesca Radio***
9:30am	(H3)	Genetic variants in components of the NALCN-UNC80-UNC79 ion channel complex cause a broad clinical phenotype (NALCN channelopathies) ***Dagmar Wieczorek***
9:45am	(H4)	TMX2 gene variants cause primary microcephaly, cortical malformation and epileptic encephalopathy with abnormal Ca2+-dependent regulation of protein folding at the ER-mitochondria interface ***Grazia Mancini***
10:00am	(H5)	Loss of function mutations in the nuclear pore complex protein NUP188 causes a novel syndrome of neurodegeneration, cataracts and facial dysmorphisms ***Sarah Sheppard***
10:15am	(H6)	Are congenital Freys syndrome, MiTES and HSAN-8 different presentation of the same thing, or just different? ***Geoff Woods***
10:30am		**COFFEE - The Whitworth Suite**

Ballroom

Chairs KERSTIN KUTSCHE and SAHAR MANSOUR

11:00am	(I1)	De novo missense variants in RAC3 cause a novel neurodevelopmental syndrome with brain malformations ***Tiong Yang Tan***
11:15am	(I2)	Mutations in the Rac-GEF and Spectrin Repeat Domains of TRIO produce Clinically Distinct Neuro-Behavioural Syndromes ***Diana Baralle***
11:30am	(I3)	A clinical and molecular study of LZTR1-associated Noonan syndrome ***Helen Stewart***
11:45am	(I4)	Recurrent de novo mutations in SLC25A24 cause Fontaine syndrome ***Karin Writzl***
12:00pm	(I5)	Growth disrupting mutations in epigenetic regulatory molecules are associated with abnormalities of epigenetic aging ***Emma Baple***
12:15pm	(I6)	DNA Polymerase epsilon deficiency causes IMAGe Syndrome with variable immunodeficiency ***Jennie Murray***
12:30pm		**LUNCH - The Whitworth Suite**