16th Manchester Dysmorphology Conference

10th–13th November 2014

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Dysmorphology Conference

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Conference Organisers
Dian Donnai
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PROGRAMME

Monday 10th November

From 3.30 pm		Registration
6.30 pm		**SUPPER**
Chairman –		**David Fitzpatrick**
7.45 pm	(1)	Deciphering Developmental Disorders Study: Detailed analysis of 1133 trios and Preliminary Analysis of 4000 trios ***Helen Firth***
8.00 pm	(2)	New severe learning difficulty syndrome with skeletal features due to *de novo* missense mutation in the polycomb group ring finger protein 2 (PCGF2) gene ***Michael Wright***
8.10 pm	(3)	Clinical phenotype associated with heterozygous *de novo* mutations in DNM1 ***Helen Stewart***
8.20 pm	(4)	Mutations in *PURA* are a cause of developmental delay – a new syndrome? ***Diana Baralle***
8.30 pm	(5)	A Recurrent mutation in COL4A3BP causes a new syndrome with developmental delay ***Diana Johnson***
8.40 pm		10 selected unknowns

Tuesday 11th November

Chairman – Dian Donnai

8:45 am	(6)	Mutations in *CKAP2L*, the Human Ortholog of the Mouse Radmis Gene, Cause Filippi Syndrome ***Bill Newman***
9.00 am	(7)	Further delineation of the KBG syndrome phenotype caused by *ANKRD11* aberrations ***Charlotte Ockeloen***
9.15 am	(8)	Tricho-Rhino-Phalangeal syndrome: Genotypes and Phenotypes in a large series ***Saskia Maas***
9.30 am	(9)	Mutations in the DNA methyltransferase gene *DNMT3A* cause an overgrowth syndrome with intellectual disability ***Katrina Tatton-Brown***
9:45 am	(10)	'Malan syndrome' – Sotos-like overgrowth with *de novo NFIX* sequence variants and deletions in six new patients and a review of the literature ***Richard Scott***
10.00 am	(11)	Diagnosis of Angelman-like disorders using a next generation sequencing panel approach ***Jill Clayton-Smith***
10:15am	**COFFEE**	

Chairman – Jill Clayton-Smith

10.45 am	(12)	Next generation clinical genetics in a consanguineous population: limitless opportunities ***Fowzan Alkuraya***
11.15 am	(13)	Compound heterozygosity of low-frequency promoter deletions and rare loss-of-function mutations in *TXNL4A* alters spliceosome assembly and causes Burn-McKeown syndrome (BMKS) ***Dagmar Wieczorek***
11:30 am	(14)	Abnormal endothelin1 signalling and mandibulofacial dysostoses in Human ***Jeanne Amiel***
11.45 am	(15)	Mutations in *C1orf85* associated with a novel craniofacial fibrous dysplasia syndrome with unusual palate and hand abnormalities ***Christopher Barnett***
12.00 pm	(16)	*BRF1* mutations alters RNA polymerase II-dependent transcription and cause neurodevelopmental anomalies ***Maria Lisa Dentici***
12.15 pm	(17)	Tessier type 3 facial clefts and choanal stenosis: a distinct genetic entity? ***Melissa Lees***
12:30 pm	**LUNCH**	

Chairman – Gunner Houge

1.45 pm	(18)	*De novo* mutations in CCND2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome ***Eamonn Sheridan***
2.00 pm	(19)	Four new cases of *PIK3R2*-related megalencephaly and polymicrogyria ***Andrew Fry***
2.15 pm	(20)	Rationalizing PIK3CA overgrowth spectrum phenotyping and therapeutics ***Leslie Biesecker***
2.35 pm	(21)	Activating mutations of *PIK3CA*, *PIK3R2*, *AKT3*, *CCND2* and other PI3K-AKT pathway genes result in a spectrum of megalencephaly and segmental overgrowth syndromes ***Bill Dobyns***
2.55 pm	(22)	A postzygotic dominant-negative *RHOA* mutation in a mosaic odonto-cerebro-cutaneo-ocular dysplasia ***Bénédicte Demeer***
3.10 pm	(23)	A novel gene causing an autosomal recessive form of infection-triggered encephalopathy ***Kay Metcalfe***
3.25 pm	(24)	Genetic heterogeneity in Hyperphosphatasia with Mental Retardation Syndrome due to mutations in PGAP3, a member of the GPI anchor synthesis pathway ***Denise Horn***
3.40 pm		**TEA & POSTER VIEWING (EVEN NUMBERS)**
7.30 pm		**SUPPER AT THE PLACE**

Wednesday 12th November

Chairman – Stanislas Lyonnet

8.45 am	(25)	The value of Whole Genome Sequencing for Intellectual Disability diagnosis ***Han Brunner***
9.00 am	(26)	Multifactorial familial intellectual disability: A clinical study in schools for special education ***Koen Devriendt***
9.15 am	(27)	The UK10K Coloboma Study ***David Fitzpatrick***
9.30 am	(28)	Identification of novel syndromal obesity genes by custom targeted exon-centric Next-Generation-Sequencing of 582 (candidate) genes ***Mieke Van Haelst***
9.45 am	(29)	Exome Analysis for Gene Discovery and Diagnosis in Multiple Pterygium Syndrome and Related Disorders ***Eamonn Maher***
10.00 am	(30)	Excess of *de novo* variants in genes involved in chromatin remodeling and regulation of transcription in patients with marfanoid habitus and intellectual disability ***Laurence Faivre***
10.15 am	(31)	Yet another gene for Loeys-Dietz syndrome… ***Bart Loeys***
10:30 am	**COFFEE**	

Chairman – Kate Chandler

11.00 am	(32)	Cerebro-Costo-Mandibular syndrome: clinical and radiological findings in a series of UK patients ***Madeleine Tooley***
11.15 am	(33)	*XYLT1* mutations in Desbuquois dysplasia further support proteoglycan synthesis impairment in disorders with multiple dislocations ***Valérie Cormier-Daire***
11.30 am	(34)	Evidence that brachyolmia with amelogenesis imperfecta can be caused by a defect in the TGFbeta signaling pathway ***Geert Mortier***
11.45 am	(35)	Homozygous Mutations in *TMCO1* cause cerebro-facio-thoracic dysplasia: Craniofacial Dysmorphism, Skeletal Anomalies, And Mental Retardation Syndrome; CFSMR ***Yasmin Alanay***
12.00 pm	(36)	Axial SMD and Zach Beighton enchondromatosis: a clinical and radiological spectrum? ***Sarah Smithson***
12.15 pm	(37)	Thalidomide Embryopathy: a re-evaluation of the clinical phenotype and updated consideration of the differential diagnosis ***Emma Baple, Christine Hall***
12.35 pm	(38)	Molecular Mechanisms in Brittle Cornea Syndrome **Graeme Black**
12.50 pm		**LUNCH & POSTER VIEWING (odd numbers)**
3.00 pm		**TEA**

Chairman – Daniela Pilz

3.15 pm	(39)	Pain and the Dysmorphologist ***Geoff Woods***
3.30 pm	(40)	*De novo* and recurrent missense mutations in the regulatory B56δ and scaffolding Aα subunits of protein phosphatase 2A cause a syndromic form of intellectual disability ***Sarju Mehta***
3.45 pm	(41)	New scenarios and therapeutic approaches in mitochondrial cardiomyopathies ***Arnold Munnich***
4.00 pm	(42)	Dysmorphology in a dish: an iPSC model for the *MECP2* duplication syndrome ***Hilde Van Esch***
4.15 pm	(43)	Human type I interferonopathies ***Yanick Crow***
4.30 pm	(44)	The origins of mutation and the new dysmorphology of the testis ***Andrew Wilkie***
4.45 pm	(45)	Neu-Laxova Syndrome is a heterogeneous metabolic disorder caused by defects in enzymes of the L-serine biosynthesis pathway ***Martin Zenker***
7.00 pm		**Tour of Town Hall (optional)**
7.30 pm		**Civic Reception**
8.00 pm		**CONFERENCE DINNER AT MANCHESTER TOWN HALL**

Thursday 13th November

Chairman – Sofia Douzgou

8.45 am	(46)	Haploinsufficiency of MECP2 modifier *SIN3A* causes mild intellectual disability by affecting cortical integrity ***Tjitske Kleefstra***
9.00 am	(47)	Mutations in PLK4, a master regulator of centriole biogenesis, cause microcephaly, growth failure and retinal dystrophy ***Andrew Jackson***
9.15 am	(48)	Loss of *CNKSR2* causes intellectual disability, attention deficit-hyperactivity and language regression ***Sarah Bowdin***
9.30 am	(49)	DYRK1A haploinsufficiency, a novel frequent cause of microcephaly and developmental delay? ***Muriel Holder***
9.45 am	(50)	ARID2 is a novel gene causing syndromic intellectual disability resembling Coffin-Siris syndrome ***Katharina Steindl***
10.00 am	(51)	Getting to new genes for Intellectual Disability: *DEAF1* and *YY1* ***Bert de Vries***
10.15 am	(52)	Mutations in the non-homologous end joining gene, XRCC4, in a patient with microcephalic primordial dwarfism ***Charu Deshpande***
10.30 am	**COFFEE**	

Chairman – Siddharth Banka

11.00 am	(53)	Looking for Persistent Truncus Arteriosus (PTA) genes ***Stanislas Lyonnet***
11.15 am	(54)	Patients with Bainbridge-Ropers syndrome caused by mutations in *ASXL3* show clinically recognizable phenotypic features ***Alma Kuechler***
11.30 am	(55)	Temple Baraitser syndrome – an overview of the phenotype and genotype ***Michael Gabbett***
11.45 am	(56)	Delineation of two novel disease entities with borderline IQ caused by 5p deletions limited to *CTNND2* or the MRIII region, respectively ***Anita Rauch***
12.00 pm	(57)	Heterozygous mutations in *MED13L* and *SYT1* result in a distinctive phenotype falling within chromosome 1p36 deletion syndrome ***Marcella Zollino***
12.15 pm	(58)	Heterozygous mutation in *ALDH18A1* is the cause of cataracts, motor system disorder, short stature, learning difficulties, and skeletal abnormalities: (SPG9) ***Jane Hurst***
12.30 pm	(59)	Identification of a novel gene causing a recognizable and distinct autosomal recessive intellectual disability and ataxia syndrome with cerebellar atrophy, relative macrocephaly, and coarse facial features ***Sérgio Sousa***
12.45 pm		**LUNCH & END OF MEETING**