Central Manchester and Manchester Children's University Hospitals NHS Trust

NHS

12th Manchester Birth Defects Conference

Manchester Conference Centre

21st - 24th November 2006

MANCHESTER 1824

nowgen
North West Genetics Knowledge Park

12th Manchester Birth Defects Conference

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Conference Organisers: Dian Donnai
& Jill Clayton-Smith

Conference Co-Ordinator: Gill Reed

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PROGRAMME

TUESDAY 21st November

4.00 pm	Registration
6.45 pm	**SUPPER**
Chairman -	**Han Brunner and Karen Temple**
8.00 pm	UNKNOWNS SESSION

WEDNESDAY 22nd November

Chairman -	**Peter Farndon**
9.00 am	Evaluation of the phenotypic spectrum associated with mutations in the TGFβ receptor genes *B LOEYS*
9.15 am	Facial morphology and clinical phenotypes associated with mutations in TGFBR1 and TGFBR2 *L ADES*
9.30 am	Homozygosity for a *FBN1* missense mutation: clinical and molecular evidence for a recessive Marfan syndrome *B HAMEL*
9.45 am	The clinical and radiological phenotype of Shprintzen-Goldberg syndrome: five new cases *S MANSOUR/C HALL*
10.00 am	Contiguous gene deletion within chromosome 10q is associated with the Bannayan-Riley-Ruvalcaba and juvenile polyposis of infancy phenotypes *S LYONNET*
10.15 am	**COFFEE**
Chairman –	**Judith Allanson**
10.45 am	Genetic disorders caused by mutations in RAS-MAPK pathway genes *E LEGIUS*
11.15 am	Geneotype, phenotype correlation in Costello Syndrome *B KERR*
11.30 am	Mutation screening and phenotype analysis in 48 patients with a clinical diagnosis of CFC or Costello syndromes and no mutation in PTPN11 and HRAS *A VERLOES*
11.45 am	Myopathy with excess of muscle spindles, hypertrophic cardiomyopathy, and Noonan-like phenotype is caused by mutations in HRAS *I VAN DER BURGT*
12.00 pm	The natural history of Noonan syndrome: a long-term follow-up study *A SHAW*
12.30 pm	**LUNCH**

Chairman - Judith Goodship

2.00 pm Phenotypes and mechanisms in defective A-type lamins associated syndromes
N LEVY

2.30 pm Extreme phenotypic diversity and non-penetrance in families with the R644C mutation of the *LMNA* gene
J RANKIN

2.45 pm CHARGE syndrome, searching for the mild end of the phenotype
M JONGMANS

3.00 pm CHARGE syndrome and *CHD7* gene – mutational and phenotypic analysis in Finnish patients
H KÄÄRIÄINEN

3.15 pm Craniosynostosis and phenotypic variability – contribution of FGFR and ephrin-B1 mutations
A WILKIE

3.30 pm **TEA**

Chairman - Sarah Smithson

4.00 pm Haploinsufficiency for LEMD3: what are the phenotypes?
G MORTIER

4.15 pm WNT7A and limb development: intra-family, inter-family and inter-species variability
G WOODS

4.30 pm Sequencing *EVC* and *EVC2* identifies mutations in two-thirds of Ellis-van Creveld syndrome patients
J GOODSHIP

4.45 pm Nail patella syndrome: first evidence for the involvement of the *LMX1B* mutation location in the risk of developing nephropathy
E BONGERS

5.00 pm Broadening the clinical spectrum of CDMP-1 mutations
M HOLDER-EPINASSE

5.15 pm The radiological profile of Schinzel-Gideon syndrome
W REARDON

7.00 pm **SUPPER**

THURSDAY 23rd November

Chairman - Helen Stewart

9.00 am — Peters' Plus syndrome is caused by mutations in *B3GALTL*, a putative glycosyltransferase
S OBERSTEIN

9.15 am — The role of the BCL6 co-repressor (BCOR) in ocular and lens development
G BLACK

9.30 am — *RAB3GAP1*: Micro syndrome and *RAB3GAP2*: Martsolf syndrome
I ALIGANIS

9.45 am — A novel autosomal recessive gene locus causes anophthalmia, congenital heart defects, diaphragmatic hernia, alveolar capillary dysplasia, and mental retardation
A RAUCH

10.00 am — Pierson syndrome: clinical features and mutational spectrum in four families
M SURI

10.15 am — Clinical and laboratory characteristics of Ohdo syndrome (Say-Barber/Biesecker/Young-Simpson type)
R DAY

10.30 am — **COFFEE**

Chairman - Andrew Wilkie

11.00 am — The filaminopathies: the cytoskeleton regulates morphogenesis
S ROBERTSON

11.30 am — Genotype-phenotype correlations in filaminopathies A
D LACOMBE

11.40 am — Allelic disorders associated with mutations in filamin B (*FLNB*)
J GRAHAM

11.50 am — The male face of MECP2: Double gene dosage leads to a specific MR phenotype; towards a detailed genotype-phenotype correlation
H VAN ESCH

12.05 pm — Mutations in the gene encoding the sigma 2 subunit of the Adapter Protein 1 Complex, *AP1S2*, cause X-linked mental retardation
L RAYMOND

12.20 pm — Lessons from Opitz FG syndrome and Lujan syndrome
R STEVENSON

12.35 pm — **LUNCH**

Chairman - David FitzPatrick

2.00 pm 'From genotype to phenotype – how array-CGH is changing the process of syndrome identification'
H FIRTH

2.15 pm Array-CGH: A novel tool in genetic diagnosis of individuals with congenital heart defects
K DEVRIENDT

2.30 pm A new chromosome 17q21.31 microdeletion syndrome associated with a common inversion polymorphism
B DE VRIES

2.40 pm The evolution of the phenotype in the 17q21.31 microdeletion syndrome
J HURST

2.50 pm Loss-of-Function mutations in euchromatin histone methyl transferase 1 (EHMT1) cause the 9q34 subtelomeric deletion syndrome
H BRUNNER

3.05 pm **TEA**

Chairman - Koen Devriendt

3.35 pm Atypical 22q11.2 deletions distal to the 3Mb region commonly deleted in VCFS/DGS syndrome are associated with a distinct phenotype
N PHILIP

3.50 pm Maternal uniparental disomy 14 [upd(14)mat] presents with an age dependent phenotype. Detection of 12 new patients by DNA methylation assay.
G GILLESSEN-KAESBACH

4.05 pm Paternal UPD 14: Review of 6 cases and further refinement of the critical region for the clinical and radiological phenotype
A OFFIAH

4.20 pm The hypomethylation syndrome presenting as transient neonatal diabetes mellitus (TNDM)
K TEMPLE

4.35 pm Beckwith-Wiedemann syndrome after assisted reproduction – the influence on phenotype
S BOWDIN

4.50 pm Genetic analysis of idiopathic hemihyperplasia patients with and without tumours
S MAAS

8.00 pm **CONFERENCE DINNER**

FRIDAY 24th November

Chairman - Julie McGaughran

8.45 am	Don't take lab results for gospel truth: lessons from our mistakes in oxidative phosphorylation deficiency *A MUNNICH*
9.00 am	The multiple pterygium syndromes: Phenotype, genotype and the role of the embryonal acetylcholine receptor γ subunit (CHRNG) *L BRUETON*
9.15 am	The clinical and molecular phenotype of Aicardi-Goutières syndrome *Y CROW*
9.30 am	Mutations in each of three Ribonuclease H2 subunits cause Aicardi-Goutières syndrome and mimic congenital viral brain infection *A JACKSON*
9.45 am	*PHOX2B* mutations trigger a wide spectrum of ANS disorders ranging from dysgenetic malformations to tumour predisposition *J AMIEL*
10.00 am	Mutations of the ARX homeobox gene, a paradigm for one gene with many phenotypes *W DOBYNS*
10.15 am	**COFFEE**

Chairman - John Burn

10.45 am	Molecular basis of Cornelia de Lange syndrome *V CORMIER-DAIRE*
11.00 am	Large phenotypic variability associated with germline *RASA1* mutations *N REVENCU*
11.15 am	Disruption of long-range control of SOX9 as a cause of Pierre Robin sequence *D FITZPATRICK*
11.30 am	Mutation of the HYLS1 gene causes the multiple malformations of hydrolethalus syndrome *R SALONEN*
11.45 am	Congenital malformations in pregnancies of diabetic women, are they the result of teratogenesis or mutagenesis? *T COLE*
12.00 pm	**LUNCH**

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