MANCHESTER 1824
The University of Manchester

Central Manchester and Manchester Children's University Hospitals
NHS Trust

11th Manchester Birth Defects Conference

Chancellors Conference Centre
9th - 12th November 2004

nowgen
North West Genetics Knowledge Park

PROGRAMME

TUESDAY 9TH November

4.00 pm onwards – Registration

6.45 pm **BUFFET SUPPER**

Chairman -

8.00 pm UNKNOWNS SESSION

WEDNESDAY 10TH November

Chairman - Peter Farndon

9.00 am WT1 mutation is a cause of Congenital Diaphragmatic hernia associated with Meacham syndrome
W REARDON

9.15 am Nance-Horan syndrome: molecular spectrum and clinical data in 16 independent families
A TOUTAIN

9.30 am Loss of function mutations in *MAN1* result in osteopoikilosis, Buschke-Ollendorff syndrome and melorheostosis
G MORTIER

9.45 am RAPADILINO syndrome: clinical and molecular analysis
H KÄÄRIÄINEN

10.00 am The real Norman Roberts syndrome
G WOODS

10.15 am **COFFEE**

Chairman - Stan Lyonnet

10.45 am PA26 is a candidate gene for human heterotaxia: human and zebrafish studies
K DEVRIENDT

11.00 am Clinical and molecular study of 53 patients with Oculo-Auriculo-Vertebral Spectrum. Development of a clinical scoring system and a new classification
G GILLESSEN-KAESBACH

11.15 am *SALL4* deletions are a common cause of Okihiro and acro-renal-ocular syndromes and confirm haploinsufficiency as the pathogenetic mechanism
J KOHLHASE

11.30 am Genotype-Phenotype correlations for GL13: An update
L BIESECKER

11.45 am	Multicentric study of Baraitser-Winter syndrome: critical review, further delineation and re-definition in 24 patients *A VERLOES*
12.00 pm	Hutchinson-Gilford progeria: Early presentation with sclerodermoid skin changes in four children *L WILSON*
12.15 pm	Break
12.30 pm	**LUNCH**
Chairman -	**Alan Fryer**
2.00 pm	Exploring the genetics of developmental eye anomalies *V VAN HEYNINGEN*
2.35 pm	Is it developmental? Unravelling disorders of anterior ocular embryogenesis *G BLACK*
3.00 pm	Malformations Associated with *SOX2* Haploinsufficiency: Symmetry in Ectodermal and Endodermal Patterning? *D FITZPATRICK*
3.15 pm	Molecular Genetics of MICRO syndrome and genotype-phenotype correlations *I ALIGIANIS*
3.30 pm	**TEA**
Chairman -	**Keon Devriendt**
4.00 pm	ADAMTS 10 mutations in the autosomal recessive Weill-Marchesani syndrome *V CORMIER-DAIRE*
4.15 pm	The CHARGE syndrome after the identification of the *CHD7* gene *B DE VRIES*
4.30 pm	The syndromic forms of Leber congenital amaurosis *A MUNNICH*
4.45 pm	Apert syndrome and the selfish testis *A WILKIE*
5.00 pm	From syndrome families to functional genomics *H BRUNNER*
5.15 pm	**POSTER SESSION**
6.30 pm	**SUPPER**
Chairman -	**Karen Temple**
8.00 pm	Two Research Databases for Birth Defects *M PATTON*
8.15 pm	Quiz *C HALL*

THURSDAY 11th November

Chairman - Geoff Woods

9.00 am — Bilateral Frontoparietal Polymicrogyria (BFPP) and *GPR56* mutations
D PILZ

9.15 am — Microcephalin – linking human brain size and chromosome condensation
A JACKSON

9.30 am — Identification of a new gene involved in a severe variant of Rett syndrome
H VAN ESCH

9.45 am — Angelman Syndrome due to UBE3A mutations, phenotype, genotype and differential diagnosis
J CLAYTON-SMITH

10.00 am — A new syndrome of neonatal diabetes, developmental delay, and epilepsy due to mutations in *KCNJ11*, a recently identified gene for neonatal diabetes
K TEMPLE

10.15 am — Two female monozygotic triplets discordant for a methylation dependent syndrome
S KANT

10.30 am — **COFFEE**

Chairman - Han Brunner

11.00 am — Genetic causes of vascular malformations
M VIKKULA

11.40 am — TGF-beta signalling in hereditary haemorrhagic telangiectasia, pulmonary arterial hypertension and cardiac development
R HARRISON

11.55 am — Milroy Disease – Phenotypic features in 71 individuals with *VEGFR3* mutations
S MANSOUR

12.10 pm — Macrocephaly-Cutis Marmorata Telangiectatica Congenita Syndrome: a report of ten cases, including cranial MRI findings, and a review of the literature.
K LEASK

12.30 pm — **Lunch**

Chairman -	**Trevor Cole**
2.00 pm	Genotype-phenotype correlation at the PHOX2B locus allows prediction of tumour risk in patients with congenital central hypoventilation syndrome *J AMIEL*
2.15 pm	Mosaic variegated aneuploidy and cancer predisposition caused by biallelic BUB1B mutations *N RAHMAN*
2.30 pm	Epigenotyping as a tool for the prediction of tumour risk and tumour type in Beckwith-Wiedemann syndrome (BWS) patients *S MAAS*
2.45 pm	High Incidence of phenotype abnormalities in patients who had cancer as a child *R HENNEKAM*
3.00 pm	**TEA, POSTERS**
	Coach trip to: **Lowry Centre/** **Imperial War Museum**
7.00 pm	**SUPPER**
Chairman -	**Jill Clayton - Smith**
8.30 pm	**Surprise Items and late bar**

FRIDAY 12th November

Chairman - Helena Kaariainen

9.00 am	Genotype-phenotype studies in three families with mutations in the polyglutamine-binding protein 1 gene (PQBP1) *B HAMEL*
9.15 am	PQBP1 mutations and dysmorphology in X-linked mental retardation *R STEVENSON*
9.30 am	Sharpening the focus – An X-chromosome tiling array elucidates the genetic basis of disease in a family with visual impairment and short stature *H FIRTH*
9.45 am	Further delineation of the clinical phenotype associated with mutations in the *OPHN1* gene: a clinically recognizable syndrome *N PHILIP*
10.00 am	Molecular Basis for a new disorder: Proximal Myopathy and Paget Disease of the Bone with or without Frontotemporal Dementia *V KIMONIS*
10.15 am	**COFFEE**

Chairman - Andrew Wilkie

10.45 am	Targeted treatment or population supplements: can the promise of birth defect prevention with folic acid supplements be realised? *J BURN*
11.00 am	Molecular determinants of atrial septal defect *R NEWBURY-ECOB*
11.30 am	Systematic assessment of *atypical* deletions reveals genotype-phenotype correlation in 22q11.2 *A RAUCH*
11.45 am	Comparison between array CGH and MAPH/MLPA in MR/MCA patients *E BIJLSMA*
12.00 pm	Primary lymphoedema and laterality disturbance *J GOODSHIP*
12.15 pm	**END OF MEETING**
12.30 pm	**LUNCH**

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