# 13th Manchester Dysmorphology Conference

## Manchester Conference Centre

## 28th to 31st October 2008

MANCHESTER 1824

Central Manchester and Manchester Children's University Hospitals NHS Trust

nowgen
A Centre for Genetics in Healthcare

# 13th Manchester

# Dysmorphology Conference

## 28th – 31st October 2008


Conference Organisers
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# PROGRAMME

## TUESDAY 28th October

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|---|---|
| 4.00 pm | Registration |
| 6.45 pm | **SUPPER** |
| **Chairman -** | **Han Brunner & Elizabeth Thompson** |
| 8.00 pm | UNKNOWNS SESSION |

**WEDNESDAY 29th October**

**Chairman - Peter Farndon**

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|---|---|
| 9.00 am | Mutational hot-spot and functional studies of the *TCF4* gene in Pitt-Hopkins syndrome<br>*J AMIEL* |
| 9.15 am | Cornelia de Lange syndrome: What causes the mild phenotypes?<br>*G GILLESSEN-KAESBACH* |
| 9.30 am | Bohring-Opitz syndrome<br>*R HASTINGS* |
| 9.45 am | Donnai-Barrow syndrome and *LRP2* mutations<br>*B POBER* |
| 10.00 am | Microdeletions in the *COH1* gene as a cause of Cohen syndrome<br>*T DE RAVEL* |
| 10.15 am | **COFFEE** |

**Chairman – Raoul Hennekam**

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| 10.45 am | CLOVE syndrome – a distinct, recognizable syndrome<br>*L BIESECKER* |
| 11.15 am | Congenital cutis laxa: a follow-up study of eighteen patients<br>*L VAN MALDERGEM* |
| 11.30 am | Domain-specific mutations in *FBN1* cause a congenital form of scleroderma: Stiff Skin Syndrome<br>*B LOEYS* |
| 11.45 am | The 'neuro-cardio-facial-cutaneous syndromes': clinical classification and genotype phenotype correlations<br>*M ZENKER* |
| 12.00 pm | Cardion-facio-cutaneous syndrome: Does genotype predict phenotype?<br>*J ALLANSON* |
| 12.15 pm | Developmental ocular disease and the RPE: Complicated at best.<br>*G BLACK* |
| 12.30 pm | **LUNCH** |

**Chairman - Michael Wright**

2.00 pm *ADAMTSL2* mutations in geleophysic dysplasia lead to a dysregulation of TGF-β signalling pathway
*V CORMIER-DAIRE*

2.20 pm Germline mutations in a tumour suppressor cause X-linked osteopathia striata congenita with cranial sclerosis
*S ROBERTSON*

2.45 pm Spondylo-megaepiphyseal-metaphyseal dysplasia: a rare but distinct skeletal disorder
*G MORTIER*

3.00 pm Evc is a tissue specific positive mediator of Hedgehog signalling
*J GOODSHIP*

3.15 pm Some aspects of temporal changes in the skeletal dysplasias
*C HALL*

3.30 pm **TEA**

**Chairman - Eamonn Maher**

4.00 pm Unifying germline mutation and neoplasia to genetic events occurring in the same cell: the example of thantophoric dysplasia type II
*A WILKIE*

4.15 pm Maternally inherited mental retardation and dysmorphism caused by a missense mutation in the genomically imprinted potassium channel *KCNK9*
*S SHALEV*

4.30 pm Mutations in *ZFP57* are associated with hypomethylation of multiple imprinted loci in patients with Transient Neonatal Diabetes (TND1)
*K TEMPLE*

4.45 pm Clinical and genetic characterization of 13 BWS twins.
*S MAAS*

5.00 pm Constitutional 11p15 abnormalities, including heritable imprinting centre mutations, cause non-syndromic Wilms tumour
*N RAHMAN*

5.15 pm Biallelic loss of function of the promyelocytic leukaemia zinc finger (*PLZF*) gene causes severe skeletal defects and genital hypoplasia
*D WIECZOREK*

7.00 pm **SUPPER**

8.30 pm **POSTER SESSION**

9.30 pm **Informal Surprise Items**

**THURSDAY 30th October**

**Chairman - Gunnar Houge**

8.45 am Loss-of-function mutation in the dioxygenase-encoding, obesity-associated FTO gene causes severe ante and post natal growth retardation and multiple malformations
*A MUNNICH*

9.00 am Molecular connections between the Usher, Joubert, and Bardet Biedl ciliary networks. Implications for the clinical spectrum of ciliopathies
*H BRUNNER*

9.15 am Finding the gene causing renal-heptic-pancreatic dysplasia in a consanguineous Norwegian family
*T FISKERSTRAND*

9.30 am Mutations of CASK cause a novel X-linked syndrome with microcephaly and disproportionate brainstem and cerebellar hypoplasia
*W DOBYNS*

9.45 am Tubulin Alpha 1A (*TUBA1A)* mutations: delineation of the phenotype
*D PILZ*

10.00 am The CDKL5 disorder: a description of patients identified in Cardiff
*H ARCHER*

10.15 am **COFFEE**

**Chairman - Ben Hamel**

10.45 am Rocks, dots and bands:Intracranial calcification as a diagnostic handle
*Y CROW*

11.00 am Inherited disorders of the Neuromuscular Junction and Multiple Pterygium Syndrome / Fetal Akinesia Sequence
*J VOGT*

11.15 am Severe arthrogryposis caused by GLE1 gene mutations – genotype-phenotype correlation
*J IGNATIUS*

11.30 am Phenotypic spectrum of Microcephallic osteodysplastic primordial dwarfism type Majewski II caused by mutations in the Pericentrin (PCNT) Gene
*A RAUCH*

11.45 am Defective ATR-dependent DNA damage signalling is associated with Microcephalic Dwarfism
*A JACKSON*

12.00 pm Genotype, neurological and clinical phenotype of patients with microcephalia vera or microcephaly with simplified gyral pattern due to ASPM gene mutations
*A VERLOES*

12.30 pm **LUNCH**

**Chairman - Stanislas Lyonnet**

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| 2.00 pm | The fruitfly *Drosophila melanogaster* as a model organism for genetic disorders<br>*A SCHENCK* |
| 2.30 pm | Of Mice, men and Williams syndrome<br>*M TASSABEHJI* |
| 2.45 pm | Prevention of the Treacher Collins syndrome in mice through inhibition of p53 function<br>*M DIXON* |
| 3.00 pm | An atypical familial 4p16.3 deletion associated with mild Wolf-Hirschhorn syndrome 3D facial shape analysis<br>*K DEVRIENDT* |
| 3.15 pm | The future of molecular dysmorphology<br>*J BURN* |
| 3.30 pm | **TEA** |
| 4.00 pm – 5.00 pm | **POSTER VIEWING** |
| 7.45 pm | **RECEPTION AND CONFERENCE DINNER**<br>**WHITWORTH ART GALLERY** |

**FRIDAY 31st October**

**Chairman - Connie Schrander-Stumpel**

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| 8.45 am | Mutations in the aristaless-related homeobox gene *ALX3* cause craniorhiny<br>*M LEES* |
| 9.00 am | FAF1 a new gene for Cleft Palate and Pierre Robin Sequence<br>*M GHASSIBE* |
| 9.15 am | Disruption of very distant highly conserved non-coding elements on either side of the *SOX9* gene is associated with Pierre Robin sequence<br>*S LYONNET* |
| 9.30 am | A new Pierre Robin syndrome caused by 5q23 deletions<br>*D FITZPATRICK* |
| 9.45 am | Screening of X-linked mental retardation patients using exon resolution arrayCGH<br>*L BRUETON* |
| 10.00 am | The 7q11.23 microduplication: is there a syndrome?<br>*N VAN DER AA* |
| 10.15 am | **COFFEE** |

**Chairman - David Fitzpatrick**

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|---|---|
| 10.45 am | 22q11 duplication syndrome: ascertaining the truth<br>*K PRESCOTT* |
| 11.00 am | Haploinsufficiency of the gene Quaking (QKI) is associated with the 6q terminal deletion syndrome<br>*H VAN ESCH* |
| 11.15 am | Further delineation of the 15q13 micro-deletion and duplication syndrome<br>*T KLEEFSTRA* |
| 11.30 am | Homozygous deletion for the 15q13.3 microdeletion syndrome causes profound mental retardation with Angelman/Rett-like features<br>*M SPLITT* |
| 11.45 am | Clinical and molecular delineation of the 17q21.31 microdeletion syndrome<br>*B DE VRIES* |
| 12.00 pm | **LUNCH** |

**END OF MEETING**