10th Manchester Birth Defects Conference

Chancellors Conference Centre
19th - 22nd November 2002

University Department Of Medical Genetics
and Regional Genetics Service
St Mary's Hospital
Manchester, UK

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CHANCELLORS CONFERENCE CENTRE
The University of Manchester
Chancellors Way, Moseley Road,
Fallowfield, Manchester M14 6NN

CONFERENCE ORGANISERS: DIAN DONNAI & JILL CLAYTON-SMITH

CONFERENCE CO-ORDINATOR: GILL REED

UNIVERSITY DEPARTMENT OF MEDICAL GENETICS
AND REGIONAL GENETICS SERVICE
St Mary's Hospital
Hathersage Road
Manchester M13 0JH

PROGRAMME

TUESDAY 19TH November

4.00 pm onwards – Registration

6.45 pm **BUFFET SUPPER**

Chairman - Jill Clayton-Smith

8.00 pm Should dysmorphologists break the rules?
T COLE

UNKNOWNS SESSION

WEDNESDAY 20TH November

Chairman - Dian Donnai

9.00 am Molecular epidemiology of craniosynostosis: an 8 year prospective study
A WILKIE

9.15 am Treacher Collins syndrome: Molecular spectrum and genotype-phenotype correlation in 42 patients.
G GILLESSEN-KAESBACH

9.30 am Costello syndrome: the face
J ALLANSON

9.45 am A clinical study of Kabuki syndrome
S WHITE

10.00 am Mapping without metaphases: Haploinsufficiency for SOX2 associated with anophthalmia
D FITZPATRICK

10.15 am Mutation of ALMS1, a large gene with a tandem repeat of 47 amino acids, causes Alström syndrome
D WILSON

10.30 am **COFFEE**

Chairman - Karen Temple

11.00 am Inherited disorders of hemidesmosomes and desmosomes
J McGRATH

11.35 am Familial partial lipodystrophy: another family
B HAMEL

11.50 am Keratosis follicularis spinulosa decalvans cum ophiasi: what's in a name?
J OOSTERWIJK

12.05 pm Aplasia cutis congenita, limb anomalies and hepatoportal sclerosis: a novel entity or a variant of Adams-Oliver syndrome?
S LYONNET

12.20 pm Hypertrichosis, growth failure and mental retardation: a new syndrome?
R NEWBURY-ECOB

12.35 pm Hypertrichosis, mental retardation, dysmorphic features and abnormally shaped corpus callosum in three unrelated females
H KÄÄRIÄINEN

12.45 pm **LUNCH**

Chairman - David Fitzpatrick

2.00 pm Disorders of chromatin structure and modification in human genetics
W BICKMORE

2.40 pm Clinical and molecular pathology associated with the *MECP2* gene
D RAVINE

3.00 pm Abnormal chromosome condensation on chromosome 4p16.1 linked with familial inherited microtia
K DEVRIENDT

3.15 pm Assisted reproductive technologies (ART) and Beckwith-Wiedemann syndrome: a case for systematic review
L BRUETON

3.30 pm Another case of imprinting defect in a girl with Angelman syndrome conceived by intracytoplasmic sperm injection from a father with normal sperm
H-K ØRSTAVIK

3.40 pm **TEA**

Chairman - Daniela Pilz

4.00 pm Mutations of *ARX* cause a wide range of phenotypes including X-linked mental retardation, West syndrome or cryptogenic infantile spasms, other epileptic encephalopathies, agenesis of the corpus callosum, X-linked lissencephaly with abnormal genitalia, and hydranencephaly
B DOBYNS

4.25 pm ARX, a novel forebrain transcription factor involved in X-linked mental retardation
H VAN ESCH

4.40 pm Spastic paraplegia, mental retardation and glaucoma. A rare X-linked or autosomal recessive condition
A TOUTAIN

4.55 pm Defective glycosylation as a key to understanding Walker-Warburg syndrome
H BRUNNER

5.10 pm The molar tooth sign and malformation occurs in four well-defined disorders including Dekaban-Arima, Joubert, Senior-Löken and Váradi (OFD6) syndromes, as well as in four other possible syndromes
B DOBYNS

5.25 pm Down-regulation of cholesterol synthesis pathway as a possible aetiological cause of the phenotype seen in Trisomy 18
W LAM

5.40 pm **POSTER SESSION**

6.30 pm **SUPPER**

Chairman - Julie McGaughran

8.00 pm Linking phenotype and gene expression databases
R WINTER

8.15 pm Thirteen year clinical follow up and molecular characterization of a family with Noonan/Leopard syndrome and a PTPN11 mutation
C CURRY

8.30 pm PTPN11 mutations in patients with features of Noonan syndrome and/or NF1 and/or LEOPARD syndrome
I VAN DER BURGT

8.45 pm Redefining the boundaries of Noonan syndrome
M PATTON

THURSDAY 21ST November

Chairman - Michael Wright

8.45 am Disparate phenotypes caused mutations in the Wnt co-receptor LRP5
M WARMAN

9.15 am Homozygosity mapping of a Dyggve-Melchior-Clausen syndrome gene to chromosome 18q21.1
V CORMIER-DAIRE

9.30 am Acrocapitofemoral dysplasia: a new autosomal recessive skeletal dysplasia
G MORTIER

9.45 am Leri-Weill dyschondrosteosis – a clinical and molecular study
D SHEARS

10.00 am Greenberg dysplasia (HEM) and lethal X-linked dominant chondrodysplasia punctata are both due to defects in the sterol pathway and have similar radiological findings
S MANSOUR

10.15 am MSX2 mutations in Parietal Foramina-Cleidocranial dsyplasia
S GARCIA-MINAUR

10.30 am **COFFEE**

Chairman - John Burn

11.00 am Multiple epiphyseal dysplasia and related conditions: phenotypes and genotypes
M BRIGGS

11.30 am Spondyloepimetaphyseal dysplasia with dislocations (Type Hall)
C HALL

11.45 am Homozygosity for a truncating mutation in *MESP2*, a transcription factor whose murine homologue alters rostro-caudal polarity in somitogenesis, is a cause of spondylocostal dysostosis 'type 2' in man
P TURNPENNY

12.00 pm Longterm follow up in Chondrodysplasia Punctata, Tibia-metacarpal type (CD-TM), demonstrating natural history
R SAVARIRAYAN

12.15 pm Amino acid substitutions in HOXA13 and HOXD13: novel phenotypes and mechanisms
F GOODMAN

12.30 pm Mutations in *SALL4* on chromosome 20 cause Okihiro syndrome
W REARDON

12.45 pm Abnormal vitreoretinal architecture associated with *PTCH/ptc* mutation implicates Hh signalling in mammalian retinal differentiation
G BLACK

1.00 pm **Lunch**

Chairman - Mohnish Suri

2.15 pm Identification of microcephalin, a protein implicated in determining the size of the human brain
A JACKSON

2.30 pm Coupling energy metabolism and brain development: Amish microcephaly
L BIESECKER

2.45 pm A silent coding change altering splicing of ATR, a DNA repair gene, is a cause of Seckel syndrome
J GOODSHIP

3.00 pm Premature centromere division in children with microcephaly, growth failure and Wilms' tumour
S SMITHSON

3.15 pm IQ's and microcephaly
G WOODS

3.30 pm Large scale deletions and *SMADIP1* truncating mutations in syndromic Hirschsprung disease with midline structure involvement
J AMIEL

3.45 pm Antenatal manifestations of mitochondrial respiratory chain deficiency
A MUNNICH

4.00 pm	**TEA**
4.00-5.30 pm	Poster Sessions (authors present until 4.45 pm)
7.00 pm	**SUPPER**
Chairman -	**Shane McKee/Alex Magee**
8.30 pm	Fibrillin and the Pharaohs – the archaeodysmorphology of Akhenaten *S MCKEE*
	Surprise Items and late bar

FRIDAY 22ND November

Chairman -	**Helen Hughes**
8.45 am	The phenotypic and molecular spectra of *NSD1* aberrations in overgrowth syndromes *N RAHMAN*
9.00 am	Benign familial macrocephaly – the Wessex PTEN experience *K TEMPLE*
9.15 am	Overgrowth in a child with 15q-trisomy, and growth retardation in a child with 15q-monosomy: a crucial role for IGF1R gene dosage in the regulation of growth *H FIRTH*
9.30 am	Overgrowth syndrome with disproportionately long great toes: report of three patients including one with a balanced chromosomal rearrangement *N PHILIP*
9.40 am	Teebi hypertelorism syndrome: further reports *A VERLOES*
9.50 am	Mental retardation and blepharophimosis in two patients: overlapping features between Young-Simpson and Ohdo syndrome *M HOLDER-EPINASSE*
10.00 am	Examination of the fetal spine – an aid to antenatal diagnosis *L CHITTY*
10.15 am	**COFFEE**

Chairman - Bronwyn Kerr

10.45 am The gene for Van der Woude syndrome
M DIXON

11.00 am Responsiveness of isolated and non-isolated neural tube defects to folic acid intervention
R STEVENSON

11.15 am A novel 5q35.3 microdeletion syndrome
A RAUCH

11.30 am The 'Trisomy 9p' syndrome: review of the critical region
T de RAVEL

11.45 pm 10q- syndrome
M IRVING

12.00 pm Behavioural features of FG syndrome (FGS), compared with Down syndrome (DS), Prader-Willi syndrome (PWS), non-specific mental retardation (NSMR), and Williams syndrome (WS)
J GRAHAM

12.15 pm Feedback from dysmorphologists' survey
T COLE

12.30 PM END OF MEETING

LUNCH

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