9th Manchester Birth Defects Conference

Chancellors Conference Centre
7th - 10th November 2000

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 7TH November

4.00 pm onwards – Registration

6.45 pm **BUFFET SUPPER**

Chairman - Michael Baraitser

8.00 pm UNKNOWNS SESSION

WEDNESDAY 8TH November

Chairman - Peter Farndon

9.00 am The BOR syndrome : Spectrum and variants
H BRUNNER

9.15 am Mutation of a novel WD-repeat protein gene in Allgrove (Triple A) syndrome
S LYONNET

9.30 am Nine cases of lymphedema–lymphangiectasia-mental retardation (Hennekam) syndrome
R HENNEKAM

9.45 am Hutchinson Gilford Progeria – clinical and molecular genetic findings
G WOODS

10.00 am Gerodermia Osteodysplastica
H STEWART

10.10 am Setleis syndrome: 3 new cases and a review of the literature
J MCGAUGHRAN

10.20 am Coffin-Siris syndrome
K METCALFE

10.30 am **COFFEE**

Chairman - Connie Schrander-Stumpel

11.00 am Genotypic and phenotypic spectrum in the tricho-rhino-phalangeal syndromes type I and III
G GILLESSEN-KAESBACH

11.15 am Wolf-Hirschhorn syndrome: developmental outcome and number of congenital anomalies correlate with deletion size
N SHANNON

11.30 am Ring chromosome 22: four children with similar subtle dysmorphic features and delayed speech
H KÄÄRIÄINEN

11.45 am	Familial Turner syndrome – a new family and review of the literature *S KANT*
12.00 pm	46, XY Sex Reversal in a family with an unbalanced 9;21 chromosomal translocation and variable phenotype *W LAM*
12.15 pm	Rubinstein-Taybi syndrome: clinical, cytogenetic and molecular studies *D LACOMBE*
12.30 pm	Noonan Syndrome: the changing face *J ALLANSON*
12.45 pm	**LUNCH**
Chairman -	**David Fitzpatrick**
2.00 pm	Embryology of the heart *K SULIK*
2.30 pm	Establishing the left-right body plan *J GOODSHIP*
3.00 pm	Left Hypoplastic Heart: a genetic aetiology *D WILSON*
3.15 pm	Clinical and molecular genetic studies in patients with suspected Barth syndrome *R NEWBURY-ECOB*
3.30 pm	Loss-of-function mutations in the cathepsin c gene result in periodontitis and palmoplantar keratosis (Papillon-Lefevere syndrome) *N THAKKER*
3.45 pm	**TEA**
Chairman -	**Robin Winter**
4.15 pm	The McKusick-Kaufman Bardet-Biedl syndrome spectrum: allelic disorders caused by mutations in the MKKS gene *L BIESECKER*
4.30 pm	Chromosomal translocations and ocular anterior segment development *R JAMIESON*
4.45 pm	Two sibs with different MR/MCA syndromes based on different unbalanced karyotypes as a result of a familial cryptic translocation *E BIJLSMA*
5.00 pm	Retinal phenotype in ring chromosome 17 *G BLACK*
5.10 pm	X-linked mental retardation with Dandy-Walker complex *N PHILIP*

5.25 pm X-linked mental retardation with Marfanoid Hapitus maps to Xq22-Xq25 in a 4 generation Scottish family
M PORTEOUS

5.40 pm Sphrintzen-Goldberg syndrome followed up during 24 years
C STOLL

5.50 pm **END OF SESSION**

6.00 pm **Computer faces demonstration – Gus Ferguson and David Fitzpatrick**

6.45 pm **SUPPER**

Chairman - Peter Turnpenny

8.00 pm A prospective, population based assessment of developmental delay and dysmorphism in children born to mothers with epilepsy
F JAMES

8.15 pm Fetal anticonvulsant syndrome – a retrospective study
U KINI

8.30 pm The Aberdeen anticonvulsants in pregnancy study: adverse fetal effects of anticonvulsant drug exposure in a population cohort
J DEAN

8.45 pm Cemental Dysplasias – Non-Genetic and Genetic
R GORLIN

THURSDAY 9TH November

Chairman - Han Brunner

9.00 am From Uniparental Disomy to an Imprinted Gene – the TNDM Story
K TEMPLE

9.15 am A novel mechanism for Angleman syndrome
A BARNICOAT

9.30 am Rett and Angelman syndrome: overlapping disorders
J CLAYTON-SMITH

9.45 am X-linked severe mental retardation and progressive neurological disorder in males caused by a mutation in MECP2, the Rett syndrome gene
K DEVRIENDT

10.00 am A novel syndrome of osteopetrosis, lymphoedema, ectodermal dysplasia and immune deficiency ascribed to mutations of NEMO, the gene for Incontinentia Pigmenti
A MUNNICH

10.15 am An unclassified X-linked immunodeficiency syndrome with signs of hypohidrotic ectodermal dysplasia in a Norwegian family does not map to ED1
H-K ØRSTAVIK

10.30 am **COFFEE**

Chairman - Willie Reardon

11.00 am Assessment of CNS anomalies in relation to normal development
J GOLDEN

11.30 am Preliminary phenotype analysis and syndrome delineation in 250 patients with polymicrogyria
W DOBYNS

12.00 pm Severe cortical malformation and microcephaly: due to maternal hyperthermia during pregnancy
D PILZ

12.15 pm X-linked lissencephaly with absent corpus callosum and ambiguous genitalia (XLAG). Clinical, MRI and neuropathological findings
A TOUTAIN

12.30 pm New MCA/MR syndrome with distinct facial appearance, broad and webbed neck, hypoplastic inverted nipples, skeletal anomalies, epilepsy, and pachygyria of the frontal lobes
J-P FRYNS

12.45 pm **LUNCH**

Chairman - Mike Patton

2.00 pm Somitogenesis
O POURQUIÉ

2.30 pm Three families with renal anomalies, bifid gynaecological tract and skeletal abnormalities and a segregation pattern suggestive of autosomal dominant inheritance
T COLE

2.45 pm A novel brachydactyly/polydactyly syndrome caused by a missense mutation in HOXD13
F GOODMAN

3.00 pm P63 Mutations: genotype/phenotype correlation
B HAMEL

3.15 pm A strategy for identifying new candidate genes for human cleft lip
D FITZPATRICK

3.30 pm A genetic study of parietal foramina: insights into development of the skull
A WILKIE

3.45 pm **TEA**

3.45-5.30 pm Poster Session (authors present until 4.30 pm)

6.45 pm **SUPPER**

Chairman - Jill Clayton-Smith

8.30 pm Surprise Items and late bar

FRIDAY 10TH November

Chairman - Raoul Hennekam

8.45 am Ischio-Pubic-Patella syndrome is surving campomelic dysplasia!
S MANSOUR/C HALL

9.00 am Linkage of the gene for otopalatodigital syndrome type 2 (OPD2) to a 3cM interval in distal Xq28; support for allelism with OPD1
S ROBERTSON

9.15 am Human genetic insights regarding the processing and activity of two secreted regulators of skeletal growth and homeostasis, cartilage-derived morphogenetic protein-1 and noggin
M WARMAN

9.30 am The phenotype of Stickler syndrome due to mutations in the COL2A1 gene
G MORTIER

9.45 am Novel COL 2A1 mutation in a family with retinal detachment, brachydactyly and a non-Stickler phenotype
H HUGHES

10.00 am Molecular analysis of Ellis-van Creveld syndrome: redefining the short-rib polydactyly spectrum?
M WRIGHT

10.15 am Diastrophic dysplasia and lethal spur type hypophosphatasia in three closely related families?
R SALONEN

10.30 am Thrombocytopenia-Absent Radius Syndrome (TAR): A clinical genetic study
L GREENHALGH

10.45 am **COFFEE**

Chairman - Margo Whiteford

11.15 am Genito-Patellar syndrome: a new condition comprising absent patellae, scrotal hypoplasia, renal anomalies, facial dysmorphism and mental retardation
V CORMIER-DAIRE

11.30 am Mapping the gene for Raine syndrome, a rare lethal autosomal recessive condition
V MURDAY

11.45 am Prenatal femoral abnormalities – natural history, prognosis and management
L CHITTY

12.00 pm Robin-Moebius Sequence : nosology
A VERLOES

12.15 pm Game, set and CATCH to Sedláčková
P TURNPENNY

END OF MEETING

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