DIAN DONNAI

8th Manchester Birth Defects Conference

Chancellors Conference Centre
10th to 13th November 1998

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

8th MANCHESTER BIRTH DEFECTS CONFERENCE

10th - 13th NOVEMBER 1998

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 10th November

4 pm Onwards - Registration

6.45 pm **BUFFET SUPPER**

Chairman - Robin Winter/Michael Baraitser

8.00 pm UNKNOWNS SESSION

WEDNESDAY 11th November

Chairman - Peter Farndon

9.00 am A review of 59 consecutive cases of Wilms tumour
T. COLE

9.15 am Hypomelanosis of Ito and malignancy
J. McGAUGHRAN

9.30 am PTEN mutations in two macrocephaly/hamartoma syndromes, Cowden disease and Bannayan-Zonana syndrome
D. LACOMBE

9.45 am Nijmegen breakage syndrome: A single mutation of Slavic origin in NBSI gene among Polish NBS families
K. CHRZANOWSKA

10.00 am Atypical chromosome breakage syndrome?
I. VAN DER BURGT

10.15 am Severe Rothmund-Thomson-like phenotype with enteropathy
R. WINTER

10.30 am COFS syndrome may be allelic with Cockayne syndrome
J. GRAHAM

10.45 am **COFFEE**

Chairman - Han Brunner

11.15 am Demonstration of an electronic database to aid diagnosis of skeletal dysplasias
C. HALL

11.40 am Facial dysmorphy in Stickler and Marshall syndromes
H. KÄÄRIÄINEN

11.55 pm Clinical and radiographic features of hypochondroplasia
G. MORTIER

12.10 pm Stuve-Wiedemann syndrome - overlap with Schwartz-Jampel syndrome Type 2
A. MUNNICH

12.25 pm Heterogeneity in autosomal recessive spondylocostal dysostosis
P. TURNPENNY

12.35 pm Spondylo-epi-metaphyseal dysplasia
P. BEIGHTON

12.45 pm **LUNCH**

Chairman - Raoul Hennekam

2.00 pm Molecular and clinical aspects of the brachydactylies
M. WARMAN

2.30 pm Novel HOXA13 mutations and Hand-Foot-Genital syndrome
F. GOODMAN

2.45 pm Upper limb malformations associated with mutations in human TBX5 and TBX3 genes
R. NEWBURY-ECOB

3.00 pm Radial defects and increased levels of follicle stimulating hormone due to spermatogenic failure - A new syndrome?
H. BRUNNER

3.15 pm Variability of the Acrocallosal syndrome
R. KÖNIG

3.30 pm X chromosome inactivation in carriers of Barth syndrome
K-H. ØRSTAVIK

3.45 pm **TEA**

Chairman - Helen Kingston

4.15 pm Molecular defects associated with anterior segment dysgenesis
G. BLACK

4.30 pm A new classification of congenital eye defects
D. MORRISON

4.45 pm — Severe manifestation of Cranio-frontonasal dysplasia
C. SCHRANDER-STUMPEL

5.00 pm — Related males with congenital dyserythropoietic anemia, limb defects, growth hormone deficiency and hypogonadism
H. TORIELLO

5.15 pm — Arterial tortuosity syndrome
M. SURI

5.25 pm — Adams-Oliver and Cutis Marmorata Telangiectasia Congenita are manifestations of a single mutant gene
H. SANTOS

5.35 pm — Two cases of Epidermolysis Bullosa Simplex
J. MORTON

5.45 pm — **END OF SESSION**

6.30 pm — **SUPPER**

Chairman - John Burn

7.45 pm — The Proteus syndrome
L. BIESECKER

8.00 pm — Autoimmunity in Noonan syndrome
M. PATTON

8.15 pm — Familial Noonan-like syndrome with cardiomyopathy and abnormal cardiac excitation
S. DAVIES

8.30 pm — Facio-audio-symphalangism syndrome
T. MONTGOMERY

8.45 pm — Perrault syndrome - Report of three families
C. GARDINER

THURSDAY 12th November

Chairman - Andrew Wilkie

8.45 am — Deletions of distal chromosome 8p
K. DEVRIENDT

9.15 am — Evidence for a specific locus for isolated cleft palate on 2q32-33
C. BREWER

9.30 am Clinical features in four children with a subtelomeric 1p36.3 microdeletion
I. YOUNG

9.45 am Terminal 1q deletion syndrome
A. TOUTAIN

10.00 am An epidemiological study of Wolf-Hirschhorn syndrome
N. SHANNON

10.15 am Failure to thrive and transient chromosome abnormalities
E. BIJLSMA

10.30 am **COFFEE**

Chairman - Connie Schrander-Stumpel

11.00 am Angelman syndrome - genotype and phenotype
J. CLAYTON-SMITH

11.15 am LISI and XLIS mutations cause most classical lissencephaly, but different patterns of malformation
D. PILZ

11.30 am Cutis verticis gyrata with congenital brain anomalies
F. BEEMER

11.45 am Studies of autosomal recessive primary microcephaly
G. WOODS

12.00 pm The childhood type of myotonic dystrophy: clinical and genetic aspects
C. DE DIE-SMULDERS

12.15 pm Syringomyelia, hemihypertrophy and the hemi 3 syndrome
J. COOK

12.30 pm Can we prevent Spina Bifida?
J. BURN

12.45 pm **LUNCH**

Chairman - Dian Donnai

2.00 pm Abnormalities associated with GLI3 mutations
L. BIESECKER

2.30 pm PAX2 mutation spectrum and expression in early human development
S. LYONNET

2.45 pm Gene expression in human embyros
D. WILSON

3.00 pm Association of the INS VNTR with size at birth
M. PEMBREY

3.15 pm Apert Syndrome: Insights into mechanisms of malformation
A. WILKIE

3.30 pm Craniosynostosis - "The Manchester Experience"
K. METCALFE

3.45 pm **TEA**

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

6.45 pm **SUPPER**

Chairman - Jill Clayton-Smith

8.30 pm Surprise Items and late bar

FRIDAY 13th November

Chairman - Bronwyn Kerr

8.45 am Fetal anticonvulsant syndrome - A clinical study of 57 cases
J. DEAN

9.00 am Lethal Arthrogryposis Multiplex Congenita - A review of 61 cases
L. BRUETON

9.10 am Hypercoagulable factors in neonatal stroke
C. CURRY

9.20 am Transient Neonatal Diabetes Mellitus - A clinical and molecular study
K. TEMPLE

9.35 am Left isomerism sequence is associated with maternal IDDM
M. SPLITT

9.45 am Pregnancy outcome after sonographic identification of short femora
L. CHITTY

10.00 am Prenatal diagnosis of birth defects in an unselected population over six years
P. BOYD

10.15 am Milroy's Lymphoedema presenting as non-immune hydrops in pregnancy
S. LYNCH

10.30 am **COFFEE**

Chairman - Marcus Pembrey

11.00 am A mélange of rare syndromes
R. GORLIN

11.15 am Onychodysplasia, localised hirsutism and learning difficulties with normal stature and relative macrocephaly
L. WILSON

11.30 am Popliteal pterygium syndrome: A clinical and molecular study
M. LEES

11.45 am The small patella syndrome
B. HAMEL

12.00 pm Phenotype in 45,X/47,XXX: comparison with other types of Turner Syndrome
J. TOLMIE

12.15 pm Clinical governance for dysmorphology services
P. LUNT

END OF MEETING