6th Manchester Birth Defects Conference

Holly Royde Conference Centre
1st to 4th November 1994

Regional Genetics Service
St. Mary's Hospital
Manchester, UK

6th MANCHESTER BIRTH DEFECTS CONFERENCE

NOVEMBER 1ST - 4TH 1994

HOLLY ROYDE
CONFERENCE CENTRE
PALATINE ROAD
MANCHESTER 20

CONFERENCE ORGANISERS: DIAN DONNAI & JILL CLAYTON-SMITH

CONFERENCE SECRETARY: LUCY REDSHAW

REGIONAL GENETICS SERVICE
ST MARY'S HOSPITAL
HATHERSAGE ROAD
MANCHESTER, M13 OJH
UNITED KINGDOM

PROGRAMME

TUESDAY 1st NOVEMBER

4.00pm Onwards - Registration

6.45pm **BUFFET SUPPER**

7.30pm Dysmorphology Club Meeting

Chairman - H. HUGHES, C. OLEY

WEDNESDAY 2nd NOVEMBER

Chairman - P. FARNDON

8.50am Introduction and Announcements
D. DONNAI

9.00am Aarskog-Scott syndrome; mutations in rho/rac gene
R. E. STEVENSON

9.15am A clinical and genetic study of Aarskog syndrome
L. LOGIE

9.30am Lumping Juberg-Marsidi syndrome and ATR-X
A. MUNNICH

9.45am An ATR-X phenocopy
R. J. GIBBONS

9.55am A patient with disturbed cell death?
R. HENNEKAM

10.05am Genetic aspects of campomelic dysplasia
S. MANSOUR

10.20am Yet another ectodermal dysplasia?
A. CLARKE

10.30am **COFFEE**

Chairman - A. FRYER

11.00am The Curry Jones syndrome
I. K. TEMPLE

11.15am	Long term survival in Pallister-Hall syndrome *A. VERLOES*
11.30am	Lumping and splitting in Pallister-Hall syndrome *J. M. GRAHAM*
11.45am	LEOPARD syndrome with craniosynostosis *R. D. CLARK*
12.00 noon	FSH muscular dystrophy; mutation and hypothesis *P. W. LUNT*
12.15pm	GI anomalies in the branchial arch syndrome *J. M. McGAUGHRAN*
12.25pm	Single central incisor and choanal atresia *A. BANKIER*
12.35pm	**LUNCH**
Chairman -	**A. WILKIE**
2.00pm	Review of holoprosencephaly in West of Scotland *M. L. WHITEFORD*
2.15pm	Holoprosencephaly with micro-/anophthalmia *G. GILLESSEN-KAESBACH*
2.30pm	Neural development in the tenascin-minus transgenic mouse *C. ffRENCH CONSTANT*
2.45pm	Clinical and molecular studies of X-linked spastic paraplegia, hydrocephalus and MASA syndrome *C. SCHRANDER-STUMPEL*
3.00pm	A clinical study of lissencephaly in England and Wales *D. T. PILZ*
3.15pm	**TEA**
Chairman -	**M. PEMBREY**
3.45pm	Homeobox genes in the fore/mid brain and face *M. W. J. FERGUSON*
4.15pm	Holoprosencephaly as a genetic model for normal craniofacial development *M. MUENKE*

4.45pm The molecular genetics of lissencephaly
W. B. DOBYNS

5.30pm **END OF SESSION**

6.30pm **SUPPER**

Chairman - S. HUSON

8.00pm Evidence in favour of intermittent neural tube closure in humans
M. J. SELLER

8.15pm Prevention of neural tube defects: towards a molecular explanation
A. J. COPP

8.30pm Human dermatosparaxis - EDS Type VIIC
W. REARDON

8.45pm A melange of rare syndromes
R. J. GORLIN

THURSDAY 3rd NOVEMBER

Chairman - M. BARAITSER

9.00am The clinical phenotypes of classical and mild De Lange syndrome
M. IRELAND

Craniofacial measurements in De Lange syndrome
J. E. ALLANSON

9.25am Aplasia cutis congenita with epibulbar dermoids
D. VILJOEN

9.35am Ocular, facial, dental, cardiac anomalies with normal intelligence
J. C. OOSTERWIJK

9.45am Oral clefting in the Leicester Asian population
M. BARROW

9.55am Craniofacial complications of NFI
S. HUSON

10.05am Kabuki syndrome: clinical features
S. J. DAVIES

10.20am Intestinal neuronal dysplasia, short stature etc: mutation in RET gene
M. PATTON

10.30am **COFFEE**

Chairman - J. TOLMIE

11.00am AD coloboma with cleft lip and palate: clinical and linkage analysis
A. O. M. WILKIE

11.15am Insitu expression of TUPLE-I and T10
D. I. WILSON

11.30am Molecularly defined Yq deletions in ten patients
H. KAARIAINEN

11.45am Skewed X-inactivation in Beckwith Wiedemann twins
K. H. ORSTAVIK

11.55am Clinical variability in branchial arch syndromes
T. R. P. COLE

12.05pm Autozygosity study of laterality sequence in Pakistani Muslim population
M. PENMAN-SPLITT

12.15pm Characterisation of deletions and translocations involving 6p
F. FLINTER

12.25pm Deletions of the elastin gene in Williams syndrome
F. GREENBERG

12.40pm **LUNCH**

Chairman - I. YOUNG

2.00pm Phenotype to genotype in Crouzon syndrome
W. REARDON

2.20pm Pfeiffer syndrome: clinical and molecular findings
M. MUENKE

2.35pm Localisation of the gene for Saethre-Chotzen syndrome
R. M. WINTER

2.50pm A clinical study of Apert syndrome
S. F. SLANEY

3.05pm Phenotype genotype correlation in Waardenburg syndromes types I and II
A. P. READ

3.30pm Kodak Digital Imaging Solution *C. GUNNER*

3.45-5.30pm **POSTER SESSION** (authors present until 4.30pm)
Book display

6.45pm **SUPPER**

Chairman - J. CLAYTON-SMITH

8.00pm The Evil Eye
R. J. GORLIN

8.30pm Surprise Items and late bar

FRIDAY 4th NOVEMBER

Chairman - A. COLLEY

9.00am The natural history of Fragile X syndrome
S. STENGEL-RUTKOWSKI

9.10am Prenatal diagnosis of fetal anomalies
R. SALONEN

9.25am Termination on medical grounds in N. England
J. BURN

9.40am AR fetal ace inhibitor-like syndrome
D. KUMAR

9.50am Arthrogryposis with antibodies that inhibit fetal AChR function
A. VINCENT

10.05am Limb, facial and renal anomalies: association with maternal hypertension treatment
J. A. HURST

10.15am Gigantism
P. BEIGHTON

10.30am **COFFEE**

Chairman - R. HENNEKAM

11.00am Heterogeneity in Holt Oram syndrome
R. NEWBURY-ECOB

11.10am Gingival fibromatosis "plus"
I. STOLTE-DIJKSTRA

11.20am	Schinzel (Ulnar-Mammary) syndrome *R. KONIG*
11.30am	A fetus with X;1 translocation and Norrie disease *A. C. BERRY*
11.40am	A further case of Mulvihill-Smith syndrome *J. C. S. DEAN*
11.50am	MCP pattern profile *P. MEINECKE*
12.00noon	Nijmegen breakage syndrome *I. Van Der BURGT*
12.10pm	Radial ray defects, renal ectopia, duodenal atresia etc: extended spectrum for Fanconi anaemia *D. G. R. EVANS*
12.30pm	**LUNCH**

END OF MEETING