5th Manchester Birth Defects Conference

13th to 16th October 1992

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

5TH MANCHESTER BIRTH DEFECTS CONFERENCE

October 13th - 16th 1992

Holly Royde
Conference Centre
Palatine Road
Manchester 20

Conference Organiser : Dr Dian Donnai

Conference Secretary : Lucy Redshaw

Regional Genetic Service
St. Mary's Hospital
Hathersage Road
Manchester M13 0JH
United Kingdom

PROGRAMME

TUESDAY

4.00pm Onwards - Registration

6.45pm **BUFFET SUPPER**

7.30pm Dysmorphology Club Meeting

WEDNESDAY

Chairman - P. FARNDON

9.00am Introduction and announcements
D. DONNAI

9.05am A clinical and genetic study of Holt-Oram syndrome
R. A. NEWBURY-ECOB

9.20am RAPADILINO syndrome
H. KAARIAINEN

9.35am Incontinentia pigmenti, clinical and genetic study
S. LANDY

9.50am Ocular findings in Incontinentia Pigmenti
J. DICKINSON

10.05am X-linked alpha thalassemia/mental retardation - phenotype
A. O. M. WILKIE

10.20am X-linked alpha thalassemia/mental retardation - localisation to Xq
R. J. GIBBONS

10.35am. **COFFEE**

11.00am Classification of severe OI by fetal radiography
C. M. HALL

11.15am Atelosteogenesis type III
A. HOCKEY

11.30am Imprinting in Albrights hereditary osteodystrophy
S. J. DAVIES

11.45am Brachymelic dwarfism bowed femora syndactylies - a new lethal syndrome
P. MEINECKE

12 noon Unusual lethal skeletal dysplasias
R. A. NEWBURY-ECOB

12.15pm Spondylocostal dysplasia and spina bifida
A. J. VAN ESSEN

12.30pm The OSSUM database of skeletal dysplasias
P. TURNPENNY

12.45pm **LUNCH**

Chairman - M. PEMBREY

2.00pm Man-Mouse homology with special reference to disease
A G SEARLE

2.30pm PAX genes in development
A. P. READ

3.00pm Man-Mouse homology - update on dysmorphology
R. WINTER

3.30pm **TEA**

4.00pm X-inactivation and monozygotic twinning
J. BURN

4.15pm Hypothesis: Monozygous twinning and discordant cell lines
J. G. HALL

4.30pm Neuronal migration defects: aetiology and recurrence risks
J. HURST

4.45pm Tobacco smoke inhalation and embryo development in the mouse
M. J. SELLER

5.00pm Allantoic steal and vitelline steal: developmental anomalies
R. E. STEVENSON

5.15pm Is anticipation seen in dysmorphology?
P. W. LUNT

6.45 **SUPPER**

8.00pm Ivory talk
R. J. GORLIN

THURSDAY

Chairman - R.C.M HENNEKEM

9.00am CHO deficient glycoprotein syndrome
S.E. HOLDER

9.15am Drash syndrome
R. F. MUELLER

9.30am Branchio-Oto renal syndrome
R. KONIG

9.45am Profound MR syndrome with dysmorphic features
J. P. FRYNS

10.00am A distinct blepharophimosis syndrome
J. CLAYTON-SMITH

10.15am "Founder effect" in dysmorphology. Marden-Walker and psuedo-trisomy 18 syndromes
A. VERLOES

10.30am **COFFEE**

11.00am Fetal causes of stillbirth
R. SALONEN

11.15am Ectodermal dysplasia with single incisor - a type III collagen defect?
R. M. WINTER

11.30am Restrictive dermopathy and fetal akinesia deformation sequence.
J. C. S. DEAN

11.45am Simpson-Golabi-Behmel syndrome: clinical aspects
M. IRELAND

12 noon Simpson-Golbi-Behmel syndrome: molecular studies
R. M. HUGHES-BENZIE

12.15pm Dubowitz syndrome and alcohol embryopathy
S. STENGEL-RUTOWSKI

12.30pm **LUNCH**

Chairman - S. HUSON

1.30pm Clinical and genetic studies on Noonan syndrome
I VAN DER BURGT

1.45pm Natural history of Noonan syndrome
M. A. PATTON

2.00pm LEOPARD syndrome
I. K. TEMPLE

2.15pm Costello syndrome with hypertrophic cardiomyopathy
N. PHILIP

2.30pm Sotos syndrome: evolution of facial phenotype
J. E. ALLANSON

2.45pm Hemimegalencephaly - an isolated abnormality?
C. A. OLEY

3.00pm Encephalocraniocutaneous lipomatosis and oculocerebrocutaneous syndrome. A different diagnosis?
J. C. OOSTERWIJK

3.15-5.30pm **POSTER SESSION**

Computer demonstration
Book display

6.00pm **WINE RECEPTION**

6.30pm **SUPPER**

8.00pm Surprise Items and Late Bar

FRIDAY

Chairman J. ALLANSON

9.00am Duplication 17p11.2p2 as a contiguous gene duplication syndrome
F. GREENBERG

9.15am The clinical phenotype of 22q11 deletion
J. GOODSHIP

9.30am Severe phenotype with ring (X) chromosomes
N. R. DENNIS

9.45pm Kabuki make up syndrome in a child with 6q monosomy/12q trisomy
P. E. JARDINE

10.00am Smith-Magenis syndrome
F. GREENBERG

10.15am **COFFEE**

10.45am Syndrome diagnosis - Parent grief or Physician glory?
H. E. HUGHES Discussion *L. KERZIN-STORRAR*

11.15am Seven patients with Peters'-plus syndrome
R. C. M. HENNEKAM

11.30am Unusual but treatable cause of arthrogryposis
S. M. HUSON

11.45pm Arthrogryposis, renal Fanconi syndrome and cholestasis: Nezelof syndrome
O. W. J. QUARRELL

12noon PEHO syndrome
L. CHITTY

12.15pm Schwartz-Jampel syndrome
E. M. THOMPSON

12.30pm **LUNCH**

END OF MEETING