7th Manchester Birth Defects Conference

Holly Royde Conference Centre
8th to 11th October 1996

Regional Genetics Service
St Mary's Hospital
Manchester, UK

7th MANCHESTER BIRTH DEFECTS CONFERENCE

OCTOBER 8TH - 11TH 1996

HOLLY ROYDE
CONFERENCE CENTRE
PALATINE ROAD
MANCHESTER 20

CONFERENCE ORGANISERS: DIAN DONNAI & JILL CLAYTON-SMITH

CONFERENCE SECRETARY: GILL REED

REGIONAL GENETICS SERVICE
ST MARY'S HOSPITAL
HATHERSAGE ROAD
MANCHESTER M13 0JH
UNITED KINGDOM

PROGRAMME

TUESDAY 8th OCTOBER

4 pm Onwards - Registration

6.45 pm **BUFFET SUPPER**

Chairman - Michael Baraitser

8 pm A mélange of rare syndromes
R. GORLIN

8.20 pm Syndrome diagnosis: An instructive case
S.M. HUSON

8.25 pm Diagnosis of Williams syndrome by FISH - An Australasian Consortium Study
A. HOCKEY

8.35-10 pm Dysmorphology cases (5 minutes for each).

WEDNESDAY 9th OCTOBER

Chairman - Peter Farndon

9.00 am A clinical and molecular study of Proteus syndrome
E. HATCHWELL

9.15 am Symptoms and course of three patients with Proteus syndrome
R. KONIG

9.30 am Deafness in Epidermal Naevus syndrome: An under-reported feature which may show a physical association between the naevus site and deafness
T. COLE

9.45 am Six further patients with macrocephaly and cutis marmorata - a distinctive overgrowth syndrome
J. CLAYTON-SMITH

10.00 am Spectrum of malformations and developmental outcome in 500 patients with 22q11 deletions: a European collaborative study
A.K. RYAN

10.15 am Molecular analyses in Williams syndrome and identification of the LIM-kinase gene within the deleted region
K. METCALFE

10.30 am **COFFEE**

WEDNESDAY 9th OCTOBER cont

Chairman - Elizabeth Thompson

11.00 am Facial dysmorphism in disorders of oxidative phosphorylation
A. MUNNICH

11.15 am Stuve-Wiedeman syndrome and defect of the mitochondrial respiratory chain
N. PHILIP

11.30 am Two rare biochemical dysmorphic syndromes with lessons for the dysmorphologist
I. YOUNG

11.45 pm Pendred syndrome: Evidence for genetic homogeneity and refinement of linkage
R. TREMBATH

12.00 pm Phenotypes and pitfalls in Pendred syndrome
W. REARDON

12.15 pm The Toriello-Carey syndrome: three further cases and review
R.F. MUELLER

12.30 pm **LUNCH**

Chairman - Marcus Pembrey

2.00 pm Imprinting and overgrowth in Beckwith-Wiedemann syndrome
W. REIK

2.30 pm Simpson-Golabi-Behmel syndrome, overgrowth and the glypican gene
R. HUGHES-BENZIE

3.00 pm Paternal duplication of 11p15.5 and Beckwith-Wiedemann syndrome
A. SLAVOTINEK

3.10 pm Deletion screening of the GPC3 gene in Simpson-Golabi-Behmel syndrome
M. IRELAND

3.25 pm Evidence for an imprinted region of the human genome at 6q22-23 and mapping of the gene for Transient Neonatal Diabetes
I.K. TEMPLE

3.40 pm **TEA**

WEDNESDAY 9th OCTOBER cont

Chairman - Robin Winter

4.10 pm A clinical study of Silver-Russell syndrome
S.M. PRICE

4.25 pm A clinical review of the Floating Harbor syndrome in seven individuals aged 4-26 years
J.A. HURST

4.40 pm Progeroid syndromes: the clinical spectrum
A. BARNICOAT

4.55 pm Short stature, dysmorphic facies and food aversion: a distinct phenotype?
M. PORTEOUS

5.10 pm Rothmund Thomsen syndrome; eight cases and review
C.G. WOODS

5.25 pm Ivemark Asplenia syndrome and anophthalmia in siblings. A possible contiguous gene syndrome on 14Q
H.G. BRUNNER

5.40 pm **END OF SESSION**

6.30 pm **SUPPER**

Chairman - Judith Allanson

7.45 pm Another approach to dysmorphology
R.C.M. HENNEKAM

8.05 pm Non-random involvement of chromosome regions in reported cytogenetic deletion and duplication syndromes - a search for an explanation
R.M. WINTER

8.25 pm Follow-up in a dysmorphology clinic: what is the diagnostic return?
H.E. HUGHES

8.40 pm Chromosome abnormalities and neural tube defects
M.J. SELLER

8.55 pm The caudal dysgenesis syndromes
S.A. LYNCH

9.05 pm Neural tube defects and Klippel-Feil syndrome
J. McGAUGHRAN

THURSDAY 10th OCTOBER

Chairman - Dian Donnai

8.45 am — Insight into PAX 6 and aniridia using mouse models
P. RASHBASS

9.15 am — 5q13: a locus for genes involved in visual impairment
G. BLACK

9.30 am — The phenotype of deletion 2q37 syndrome
P.D. TURNPENNY

9.45 am — Phenotype analysis of 32 patients with Albright's hereditary osteodystrophy and GS alpha gene mutations
L. WILSON

10.00 am — Obesity and mental retardation. Established conditions and undefined syndromes
G. GILLESSEN-KAESBACH

10.15 am — Bardet-Biedl syndrome: A survey of clinical features amongst patients residing in the United Kingdom
P.L. BEALES

10.30 am — **COFFEE**

Chairman - Karen Temple

11.00 am — Genetics of Hirschsprung's disease
S. LYONNET

11.30 am — Dysmorphic facies, mental retardation and Hirschsprung disease: one or two syndromes?
H. KAARIAINEN

11.45 am — Synpolydactyly (SPD) phenotypes correlate with size of expansion in HOXD13 polyalanine tract
F.R. GOODMAN

12.00 pm — Apert syndrome: a model system for the study of human mutation and malformation
A. WILKIE

12.15 pm — Further evidence for phenotypic and cytogenetic overlap between Baller-Gerold and Roberts syndromes
E. THOMPSON

12.25 pm — New syndromes involving craniosynostosis
D. LACOMBE

THURSDAY 10th OCTOBER cont

12.35 pm **LUNCH**

Chairman - Andrew Wilkie

2.00 pm Lessons from the Lissencephaly Research Project: five new malformation syndromes
W.B. DOBYNS

2.15 pm Winter-Tsukahara syndrome: a new type of lissencephaly?
S.F. SLANEY

2.30 pm Mapping of two neuronal migration syndrome genes: the LIS1 gene in chromosome 17p13.3 and the XLIS gene in Xq22.3-23
W.B. DOBYNS

2.45 pm Lissencephaly syndromes: Does the face reflect the brain?
J.E. ALLANSON

3.00 pm Integrin expression and function in early neural development
C. FFRENCH-CONSTANT

3.15 pm Developmental delay and mild learning disability associated with Arachnoid cysts exemplify difficulties with diagnosis of macroscopic cerebral dysplasia
J.L. TOLMIE

3.30 pm Bowen syndrome: Report of a case and review of the literature
H.V. TORIELLO

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

8.00 pm Amber
R. GORLIN

8.30 pm Surprise Items and late bar

FRIDAY 11th OCTOBER

Chairman - Alan Fryer

8.45 am Treacher-Collins syndrome
M. DIXON

9.00 am A distinctive blepharophimosis-retardation syndrome: The Say-Barber-Biesecker ("smooth face") type
P. MEINECKE

9.10 am Holoprosencephaly in a family with a balanced paracentric inversion of chromosome 13q
L.A. BRUETON

9.20 am Parietal foramina and multiple exostoses - familial inheritance
S.J. DAVIES

9.35 am A dominant syndrome of joint restriction, tight skin and bulky muscles: A mild variety of Moore-Federman syndrome with normal height?
N. DENNIS

9.45 am Heterozygous manifestations in the heritable disorders of the skeleton
P. BEIGHTON

10.00 am Heterogeneity in Meckel syndrome
R. SALONEN

10.15 am **COFFEE**

Chairman - John Burn

10.45 am Familial Angelman syndrome in eight affected relatives with "unrecognizable" phenotypes
I. VAN DER BURGT

11.00 am True hermaphrodites and 46,XX males in the same family: The result of an autosomal mutation
L.S. CHITTY

11.15 am Multiple congenital anomaly (MCA) syndromes associated with abnormal Mullerian development: A clinical and genetic review
D. KUMAR

11.30 am Otopalatodigital syndrome Type II, Yunis-Varon syndrome and severe Melnick-Needle syndrome: Are they really distinct entities?
A. VERLOES

11.45 am Psychological aspects of children with Noonan syndrome
M.A. PATTON

FRIDAY 11th OCTOBER cont

12.00 pm Clinical manifestations in 100 South African patients with Fetal Alcohol syndrome
D. VILJOEN

12.15 pm Hyperemesis gravidarum and congenital vertical nystagmus: in utero Wernickes encephalopathy
S.M. HUSON

12.30 pm **LUNCH**

END OF MEETING