# 4th MANCHESTER BIRTH DEFECTS CONFERENCE

October 2nd – 5th 1990


Holly Royde
Conference Centre
Palatine Road
Manchester 20 U.K.

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Conference Organizer : Dr Dian Donnai

Conference Secretary : Mrs Val Warren

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

PROGRAMME

Tuesday

4.00 pm Onwards - Registration

6.45 pm BUFFET SUPPER

7.30 pm Dysmorphology Club Meeting

Wednesday

Chairman - P Farndon

9.00 am Introduction and announcements
D Donnai

9.05 am Limb reduction defects of the upper limbs.
U G Froster-Iskenius

9.25 am Jeune's asphyxiating thoracic dystrophy. I Dijkstra

9.45 am Cartilage-Hair hypoplasia. I van der Burgt

10.05 am A new patient with bone dysplasia, midface hypoplasia and deafness. H Kaarianinen

10.20 am Chondrodysplasia punctata and Larsen syndrome in second cousins. J C S Dean

10.35 am COFFEE

11.00 am The Noonan phenotype in NFI individuals.
A F Colley

11.20 am Neurofibromatosis - mechanism/risk of mutation and segmentation. S Huson

11.40 am Some Other Overgrowth Syndromes. T R P Cole

12 noon Megalencephaly - presentation of five cases. J Zaremba

12.15 pm Protean problems in Proteus syndrome. J Burn

12.30 pm Hypertrophic cardiomyopathy in Noonan syndrome.
M Sharland

12.45 pm LUNCH

Chairman - M Pembrey

2.00 pm Guest Lecture
G Turner and M Partington

2.45 pm Waardenburg syndrome and piebaldism. P Beighton

3.05 pm X-Linked ocular albinism with sensorineural deafness.
I Winship

3.20 pm A new syndrome with Potter sequence, CHD and characteristic facies. P Meinecke

3.35 pm TEA

4.00 pm An embryological look at polydactylies and syndactylies. R Winter

4.30 pm Genetic aspects of non-Apert acrocephalosyndactylies.
L A Brueton

4.55 pm Anthropometric craniofacial pattern profiles.
J E Allanson

5.15 pm Macrocephaly : A genetic factor in NTD. R E Stevenson

5.30 pm Hydrolethalus syndrome; Prenatal and neurological findings. R Salonen

6.45 pm SUPPER

8.00 pm 'Nobody knows the trauma I've seen'. R Gorlin

8.45 pm BREAK

9.00 pm The dysmorphology detective. J Aase

Thursday

Chairman - M Baraitser

9.00 am A possible role for surface ectodermal placodes.
R D Clark

9.20 am Sporadic patterns of anomalies due to maternal hyperthermia. J Graham

9.40 am Genomic imprinting, MZ twinning and X-inactivation.
J G Hall

10.00 am Genetic heterogeneity. J H Edwards.

10.20 am COFFEE

11.50 am A clinical and genetic study of Angelman syndrome. J Clayton-Smith

11.10 am Miller-Dieker syndrome. C A Oley

11.25 am Peters' anomaly - short stature and Peters' plus. E Thompson

11.40 am Do all males with OFD II have OFD II? J Goodship

11.55 pm Dysmorphic features in variegate porphyria. I Winship

12.10 pm Incontinentia pigmenti. D Donnai

12.25 pm LUNCH

Chairman - C Berry

1.45 pm Guest lecture. R J Gorlin

2.15 pm A family with severe X-linked arthrogryposis. R C M Hennekam

2.30 pm PWS with joint contractures. H Hughes

2.45 pm A new syndrome of deafness, Perthe's disease and developmental delay. L S Chitty

3.00 pm Cleft lip and the "unknowns". S E Holder

3.15-5 pm Posters
Trade display
Computer demonstrations
Slide projectors and viewing boxes available for informal case discussion.
TEA served at 3.30 pm

6.00 pm Wine reception

6.30 pm SUPPER

7.30 pm Poster discussion. M Pembrey

8.00 pm 'Mistakes men have made' M Sellar

8.45 pm BREAK

9.00 pm SURPRISE ITEM

Friday

Chairman - M Partington

9.00 am The limitations of prenatal diagnosis. I D Young

9.15 am Paternal origin of chromosome 4 in Wolf-Hirschhorn syndrome. O W J Quarrell

9.30 am Dysmorphic features and diagnosis of DiGeorge syndrome. D I Wilson

9.45 am What happens to dysmorphic newborns? F Child

10.00 am EDV-assisted dysmorphology database. S Stengel-Rutkowski

10.15 am COFFEE

10.45 am Guest lecture. R Happle

11.30 am Congenital papular skin lesions in a family. B D Hall

11.45 am Association between trichothiodystrophy and DNA repair defect. J L Tolmie, A R Lehmann

12.05 pm Cornelia de Lange syndrome - a clinical review. M Ireland

12.20 pm Does Moebius syndrome exist? K MacDermott

12.35 pm Retinal manifestations in carriers of Norries disease. P W Lunt

12.50 pm LUNCH

End of Meeting