3rd MANCHESTER BIRTH DEFECTS CONFERENCE

October 25th – 28th 1988

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

3RD MANCHESTER BIRTH DEFECTS CONFERENCE

October 25th - 28th 1988

Holly Royde
Conference Centre
Palatine Road
Manchester 20

Conference Organizer : Dr Dian Donnai

Conference Secretary : Mrs Val Warren

Regional Genetic Service
St Mary's Hospital
Hathersage Road
Manchester M13 OJH
UK

PROGRAMME

TUESDAY

3.00	Onwards	Registration
6.45	pm	Supper
7.30	pm	Dysmorphology Club Meeting

WEDNESDAY

CHAIRMAN - P Farndon

9.00	am	Introduction and announcements. D Donnai
9.05	am	Fetal dysmorphology : The new challenge. J Burn
9.30	am	The California Birth Defects Monitoring Program. C Curry
9.55	am	Congenital anomalies : An increasingly important cause of mortality and workload in a neonatal intensive care unit. J G Hall, E W Ling
10.15	am	COFFEE
11.00	am	Prenatal diagnosis of the caudal deficiency-asplenia syndrome. R F Mueller, P S Vinall, G Batcup, M D Shields
11.15	am	Two siblings with Walker-Warburg syndrome - Problems in prenatal diagnosis? A Mustonen, R Herva, P Kirkinen, P Jouppila, J Leisti
11.30	am	The Baller-Gerold syndrome : report of two cases and further delineation of the syndrome. S M Huson, J Tolmie, P Galea, C M Hall, R M Winter
11.45	am	Turner's syndrome : Errroneous diagnosis on buccal smear. M A Patton, E M T Pyta, R Patel
12.00	noon	The recognition of germline mosaicism. J H Edwards
12.30	pm	LUNCH

WEDNESDAY continued

CHAIRMAN - H Kingston

2.00 pm — The natural history of Lowe (oculocerebrorenal) syndrome.
C M E McKeown

2.30 pm — Concordant and discordant MZ twins in Beckwith syndrome - related but different aetiologies?
P W Lunt, J Clayton-Smith, A P Read, D Donnai

2.45 pm — Neurofibromatosis - Noonan syndrome - independent segregation of the two conditions within a family?
J Clayton-Smith, D Donnai

3.00 pm — TEA

3.30 pm — Investigations into the causes of neural tube defects in mice in men.
M Seller

4.00 pm — MASA syndrome : further clinical delineation and chromosomal location.
R M Winter, M N Patterson, M V Bell, S M Huson, K E Davies

4.15 pm — A case of kyphomelic dysplasia.
I K Temple, E Thompson, G Bridgeman, M E Pembrey

6.30 pm — SUPPER

7.45 pm — The tongue, its use and abuse.
R J Gorlin

8.45 pm — BREAK

9.00 pm — Unknown cases

1) A possible new X linked dysmorphic retardation syndrome in 3 generations of one family.
J Burn, E Elliot, E V Davison

2) A new lethal short limbed dwarfism - case report.
W C Tsang, K Tse, A S Chau

3) Inheritance of a bizarre eye muscle disorder.
L S Chitty, M Baraitser

4) A previosly unrecognised spondyloepiphyseal dysplasia with associated dysmorphic features.
S Huson, C M Hall, R M Winter

5) Schwartz-Jampel Syndrome. H E Hughes

THURSDAY

CHAIRMAN - I Young

9.00 am	A foetal diagnosis of the BBB syndrome. A Hockey, A Murphy
9.15 am	An extension of the phenotype description of Binders syndrome. O W J Quarrell, M Kock, H E Hughes
9.30 am	Mid-line facial defects with ocular colobomata : a new syndrome? I K Temple, J Burn, M Baraitser
9.50 am	A new autosomal dominant tricho-dental dysplasia. D J Eteson, R D Clark
10.10 am	Gestational hyperthermia as a possible cause for Moebius syndrome. J M Graham, M J Edwards, A K Iofolla, A H Lipson, W S Webster
10.30 am	COFFEE
11.00 am	45 Dutch patients with Rubinstein-Taybi syndrome. R C M Hennekam, J C A Baselier, E M M I Beijaert, M J Van Den Boogaard, J Jansma, J M Van Doorne, V V Thorbecke-Nilson, H Veerman, B J Sibbles, A Bos, H G Van Spijker
11.30 pm	The changing face : Rubinstein-Taybi syndrome. J E Allanson
12.00 noon	Changing patterns in metacarpophalangeal profiles. P F Dijkstra, R Hennekam
12.30 pm	LUNCH
1.45 pm	Some craniofacial syndromes. R J Gorlin
2.45 - 5.00 pm	Posters Trade display Book display Computer demonstrations Slide projectors and viewing boxes available for informal discussion of cases
	TEA served at 3.30 pm

THURSDAY continued

6.30	pm	SUPPER
		CHAIRMAN - M Pembrey
7.45	pm	Discussion of posters
8.45	pm	BREAK
9.00	pm	J Burn - surprise item.
		R Winter - vote of thanks

FRIDAY

CHAIRMAN - H Hughes

9.00 am — Radial aplasias and associated anomalies - a review of 25 undiagnosed patients.
J Hurst, M Baraitser, R M Winter

9.20 am — The phenotype of 45,X/46,XY mosaicism: an analysis of prenataly diagnosed cases.
H J Chang, R D Clark, H Bachman

9.40 am — Vitelline placentation in man.
R C Rogers, R E Stevenson, R A Saul, M C Phelan

9.55 am — Congenital shortness of the costocoracoid ligament.
J G Hall, S Bamforth

10.15 am — COFFEE

10.45 am — Gorlin's syndrome : Macrocephaly and developmental delay in childhood.
P A Farndon

11.00 am — Duchenne muscular dystrophy in South Africa : Molecular management.
R Ballo

11.15 am — Possible human homologues for the mouse mutant disorganisation.
R M Winter, D Donnai

11.30 am — Disorganisation : a model for "early amnion rupture"?
D Donnai, R M Winter

12.00 noon — LUNCH

END OF MEETING