# 14th Manchester Dysmorphology Meeting

11th - 14th October 2010

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# Dysmorphology Conference

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Conference Organisers
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**PROGRAMME**

**Monday 11th October**

4.00 pm Registration

6.45 pm **SUPPER**

**Chairman – Han Brunner, Elizabeth Thompson**

8.00 pm UNKNOWNS SESSION

**Tuesday 12th October**

**Chairman – Julie McGaughran**

9.00 am (1) Identification of 3MC syndrome genes reveals a novel role for complement pathway components in development and disease
*M LEES / P BEALES*

9.15 am (2) Tufting enteropathy and a skeletal disorder due to homozygous EpCAM
*T COLE*

9.30 am (3) Six cases with MEF2C haploinsufficiency
*E BIJLSMA*

9.45 am (4) Manitoba-Oculo-Tricho-Anal (MOTA) syndrome is caused by mutations in FREM1
*A SLAVOTINEK*

10.00 am (5) Allelic NFIX mutations trigger either a Sotos-like or a Marshall-Smith syndrome depending on impact on non-sense mediated MRNA decay
*V CORMIER-DAIRE*

10.15 am (6) Seckel syndrome with deficiency of the ATR signalling pathway
*M SURI*

10.30 am **COFFEE**

**Chairman – Willie Reardon**

11.00 am (7) Mutations in HPSE2 cause urofacial syndrome
*W NEWMAN*

11.15 am (8) Bilateral renal agenesis / hypoplasia / dysplasia (BRAHD)
*D FITZPATRICK*

11.30 am (9) Mutations in a regulator of Cdc42 and Rac1 GTPases, cause scalp and transverse truncating limb birth defects
*R TREMBATH*

11.45 am (10) TUBB2B mutations and cortico-cerebral dysgeneses
*D PILZ*

12.00 pm (11) Germline gain-of-function mutations of ALK lead to abnormal central nervous system development
*J AMIEL*

12.15 pm (12) Clinical features of the Frank-Ter Haar syndrome
*J HURST*

12.30 pm **LUNCH**

**Chairman – Ruth Newbury-Ecob**

2.00 pm (13) Comments on the Nosology of Genetic Skeletal Disorders 2010
*A SUPERTI-FURGA*

2.30 pm (14) Mutations in PITX1 cause a human patellar malformation syndrome
*E BONGERS*

2.45 pm (15) Mutations in the DDR2 gene cause SMED with short limbs and abnormal calcifications
*A RAAS-ROTHSCHILD*

3.00 pm (16) Mesomelia-synostoses syndrome results from deletion of SULF1

and SLC05A1 genes at 8q13
*A VERLOES*

3.15 pm (17) Clinical and molecular findings on 20 patients with fibrodysplasia ossificans progressiva
*I STEFANOVA*

3.30 pm **TEA**

**Chairman – Sarah Smithson**

4.00 pm (18) Mutation in the 3'UTR of the HDAC6 gene abolishing the post-transcriptional regulation mediated by hsa-miR-433 is linked to a new form of dominant X-linked chondrodysplasia
*D LACOMBE*

4.15 pm (19) The 12q14 microdeletion syndrome: an update on genotype and phenotype and relevance to bone dysplasias and growth disorders
*G MORTIER*

4.30 pm (20) dREAMS: extracting the knowledge behind radiological language
(21) *C HALL / I BAMSEY*

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| 5.00 pm - 6.30 pm | **Poster Session with Authors (even numbers)** |
| 7.30 pm | **SUPPER (The Place)** |

**Wednesday 13th October**

**Chairman – David Fitzpatrick**

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| 9.00 am | (22) | Lenz-like syndrome in a large pedigree suggestive of X-linked inheritance is caused by a 2.3 Mb deletion at 14q22.2q23.1 involving OTX2<br>*A TOUTAIN* |
| 9.15 am | (23) | Hypomorphic mutations in NSDHL associated with CK syndrome<br>*L RAYMOND* |
| 9.30 am | (24) | Male phenotype associated with loss of function alleles at the FLNA locus<br>*S ROBERTSON* |
| 9.45 am | (25) | Female phenotype of RSK2 mutations and its implications for genetic counselling<br>*A RAUCH* |
| 10.00 am | (26) | Whole ARX gene duplications are associated with a mild or normal phenotype<br>*N PHILLIP* |
| 10.15 am deficiency | (27) | Clinical presentation and therapeutic approach of coenzyme Q10<br>*A MUNNICH* |
| 10.30 am | **COFFEE** | |

**Chairman – Gunnar Houge**

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| 11.00 am | (28) | Will Next Generation Sequencing change the practice of genetics<br>*H BRUNNER* |
| 11.15 am | (29) | Massively parallel sequencing of exons on the X chromosome: successes and promising leads<br>*L BIESECKER* |
| 11.30 am | (30) | Application of next generation sequencing in the diagnosis of bone dysplasias<br>*A ZANKL* |
| 11.45 am | (31) | Use of exome sequencing to identify GJC2: gene responsible for causing four-limb primary lymphoedema<br>*F CONNELL* |
| 12.00 pm | (32) | Hyperphosphatasia-mental retardation syndrome is caused by mutations of PIGV<br>*D HORN* |
| 12.15 pm | (33) | When one single X-ray gives the diagnostic clue<br>*A BOTTANI* |
| 12.30 pm | **LUNCH AND POSTER VIEWING** | |

**Chairman – Leslie Biesecker**

2.00 pm (34) Long range dysregulation of gene expression at the SOX9 locus
*S LYONNET*

2.15 pm (35) Dysregulation but not deletion of LRFN5 is associated with autism
*G HOUGE*

2.30 pm (36) A paradoxical genotype-phenotype correlation for EFNB1 mutations: worse outcome in mosaic than constitutionally-deficient males
*A WILKIE*

2.45 pm (37) Imprinting disorders finding out why? The clinical impact of methylation testing at multiple imprinted loci
*K TEMPLE*

3.00 pm **TEA**

**Chairman – Bronwyn Kerr**

3.30 pm (38) Novel and extended phenotypes in patients with germline mutation in SHOC2
*E BURKITT-WRIGHT*

3.45 pm (39) Costello syndrome and germline HRAS mutations: phenotypic and genotypic variations
*K GRIPP*

4.00 pm (40) What is the significance of genetic heterogeneity in Fraser syndrome?
*M ZENKER*

4.15 pm (41) New FRAS1 and FREM2 gene mutations in Fraser syndrome using Roche 454 GS-FLX system
*M van HAELST*

4.30 pm **Poster Session with Authors (odd numbers)**

8.00 pm **CONFERENCE DINNER – IMPERIAL WAR MUSEUM**

**Thursday 14th October**

**Chairman – Emma Hobson**

8.45 am (42) Tbox genes in development and disease
*R NEWBURY-ECOB*

9.00 am (43) Nablus mask-like facial syndrome and blepharo-naso-facial syndrome are the same entity
*J ALLANSON*

9.15 am (44) 10q24 duplication: 13 new cases identified on either Q-PCR or Array – CGH allow prenatal diagnosis and broadening the clinical spectrum
*M HOLDER-ESPINASSE*

9.30 am (45) Autosomal recessive mental retardation caused by COMMD4 mutation in an Israeli Arab family
*S SHALEV*

9.45 am (46) Delineation and expansion of the phenotypic effects of bi-allelic mutations in G6PC3
*S BANKA*

10.00 am (47) Pericentrin mutations in microcephalic osteodysplastic primordial dwarfism type II
*A JACKSON*

10.15 am **COFFEE**

**Chairman – Judith Allanson**

10.45 am (48) Mutations in ABHD12 cause the neurodegenerative disease PHARC: first inborn error of endocannabinoid metabolism
*T FISKERSTRAND*

11.00 am (49) Mutations in GRIN2A and GRIN2B encoding regulatory subunits of NMDA receptors cause variable neurodevelopmental phenotypes
*D WIECZOREK*

11.15 am (50) SOBP gene mutation causes autosomal recessive syndromic and nonsyndromic mental retardation and is highly expressed in the limbic system
*L BASEL-VANAGAITE*

11.30 am (51) Recessive mutations in the OCLN gene cause band-like calcification and polymicrogyria with simplified gyration
*M O'DRISCOLL*

11.45 pm (52) PQBP1-linked XMR: moving border between non-specific X-linked MR and syndromic XMR
*L VAN MALDERGEM*

12.00 pm **LUNCH AND END OF MEETING**