Sunday, June 13, 2010, 15.00 - 16.30 hrs

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WS1. Dysmorphology Workshop 1 (D . Donnai, J . Clayton-Smith) — Room F1+F2+F3

The Organisers of the Dysmorphology Workshop invite clinicians to submit rare known and unknown cases with dysmorphic syndromes before the workshop. Please bring a short case presentation on a USB stick from 12.30 - 13.00 hrs to room F1+F2+F3. Maximum time for presentation: 5 minutes.

WS2. Workshop on recent progress and developments in QAu in Europe (E . Dequeker, M . Morris) — Room F6

15.00 Introduction : E Dequeker
15.05 The European pilot EQA for molecular karyotyping demonstrates large variation in outcome and quality across laboratories. Joris Vermeesch, Leuven, Belgium
15.20 The availability of reference materials for genetic testing, David Barton, Dublin, Ireland
15.35 Providing Quality Information To Patients: Experience Of The EuroGentest Project, Alastair Kent, Genetic Interest Group, UK
15.50 The European laboratory Quality Assurance database, Mariana Jovanovic, Paris, France
16.05 A standardized framework for the validation and verification of clinical molecular genetic tests, Chris Mattocks, Salisbury, UK
16.20 Conclusion M Morris

WS3. Genetic Education (P . Farndon, T. Ozcelik) — Room G3

This workshop covers topics from genetic education relating to high schools, pre-registration health professionals, impact on clinical service and the availability of resources. To encourage discussion, please look at the relevant posters before coming to the session; additional material will be presented.

- DNA Day:a good starting point to disseminate genetic education in high school (Poster P01.19)
  B. Zanini, Genova, Italy
- Influencing how genetics is taught in UK secondary schools: The Nowgen Schools Genomics Programme (Poster P01.18)
  L. E. Holmes, Nowgen Centre for Genetics in Healthcare & Manchester Academic Health Science Centre, Manchester United Kingdom
- Achieving change in clinical management: results from a genetics education intervention (Poster P01.33)
  M. Bishop, NHS National Genetics Education and Development Centre, Birmingham, United Kingdom
- A practice framework for promoting appropriate reporting and use of molecular genetic test results: combining education, test result reporting, and information resources (Poster P01.56)
  I.M. Lubin, Centers for Disease Control and Prevention, Atlanta, GA, United States
- Eurogene: a pan-european e-learning Service in human genetics (Poster P01.22)
  M. Dutto, European Genetics Foundation, Bologna, Italy

Discussion:
Can using MedEdWorld help build a community of practice in genetics education? (Peter Farndon)
What proposals should be made for pre-conference workshops and sessions on genetics education at the International Congress of Human Genetics 2011? (Sylvia Metcalfe)

WS4. Rapid aneuploidy detection: trouble shooting. (T .H . Bui) — Room F4+F5

MLPA. *Brigitte Faas (Nijmegen, TheNetherlands)*
QF-PCR. *Vincenzo Cirigliano (Barcelona, Spain)*
QF-PCR stand alone. *The-Hung Bui (Stockholm, Sweden)*

WS5. DNA diagnostics - how to make sense from soooo many genes, variants and databases (J . den Dunnen) — Room G1+G2

Current developments in sequencing technology generate an ever-increasing flood of DNA sequence variants. When in the past we could only afford to analyse the protein coding part of a clear candidate disease gene, we can now perform a genome-wide exome analysis. As a consequence we detect more and more variants all demanding careful analysis; might it have functional (pathogenic) consequences or not? In the first half of the session some current standards, possibilities and developments will be presented. In the second half of the session we will discuss the subject with the audience, focusing on current obstacles, limitations and tools that are lacking; „what do we need to keep track of the deluge of data we are faced with".

- The HGVS recommendations for the description of sequence variants and gene sequence variant databases (LSDBs), *Johan den Dunnen*
- A new standard - the Locus Reference Genomic (LRG) DNA sequence format, *Raymond Dalgleish*
- Using gene variant databases (LSDBs) to store and analyse sequence variants; pathogenic or not?, *Speaker t.b.a.*

WS6. Implications of next generation sequencing for clinical practice - A debate (J . Veltman, H. Scheffer) — Congress Hall

Next generation sequencing is rapidly becoming the method of choice in genetic research and is likely to be quickly adopted in DNA diagnostics. During this workshop we will discuss the impact of this development for the medical field. We will debate possible scenarios for the future with experts from different field of genetics as well as with the audience.
Possible scenarios:
Genome sequencing will be the first and only tool in genetic diagnostics
Diagnostic genome sequencing will be done by commercial companies
Genome sequencing will not be used diagnostically in the next 5 years
Genome sequencing will open genetic diagnostics to many more common diseases
Clinical geneticist will not counsel patients before they have genome sequence available
The role of clinical geneticist is over, genome sequencing data will be interpreted by referring clinicians with aid from bioinformaticians and statisticians
Legal/ethical/political restrictions will limit the implementation of genome sequencing in diagnostics

Invited debaters:
Clinical genetics: *Prof. Koen Devriendt*
Research: *Prof. Xavier Estivill*
Company: *Representative from Complete Genomics*
DNA diagnostics: *Prof. Gert Matthijs*
Ethics: *Prof. Anne Cambon*
Prof. James Lupski

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WS7. Dysmorphology Workshop 2 (D . Donnai, J . Clayton-Smith) — Room F1+F2+F3

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WS8. Complex Genetics beyond GWAS (C . van Duijn, C . Janssens) — Room F4+F5

15.00 - 15:30 Finding the Missing Heritability - *Prof Dr Cornelia M van Duijn, Erasmus MC, Rotterdam*
15.30 - 16.00 Recent developments in GENETIC risk prediction studies - *Dr A. Cecile J.W. Janssens, Erasmus MC, Rotterdam*
16.00 - 16:30 Discussion

WS9. Community Genetics - Cardiogenetics (M . Cornel, U . Kristoffersson) — Room F6

15:00-15:20 Cardiac genetics: the first 250 clinic patients (P01.07), *Julie McGaughran, Brisbane, Australia*
15:20-15:40 Genetic counselling and testing in Cardiomyopathies (P01.44), *N.Marzilliano, Milano, Italy*
15:40-16:00 Genetic counselling and testing in inherited monogenic cardiac disorders: interviews with patients and their families (P01.43), *S. Fokstuen, Geneva, Switzerland.*
16:00-16:30 Forum discussion: Future of cardiogenetic testing in the light of the fast developments of whole genome technologies:
- rapid decline of price of whole genome information
- difficulty to interpret this information
- lack of clinical utility for most of the information, especially if the meaning is not clear
- apparent clinical utility in some fields (cardiogenetics in Lancet Paper Ashley et al.)
- where will we find the personnel to analyse and discuss whole genome information?

Additional participants to the forum: *A. Clarke*, Clinical Genetics, Cardiff University, Cardiff, UK, *H. Meijers-Heijboer*, Clinical Genetics, VU University Medical Centre, Amsterdam, Netherlands, *G. de Wert*, Ethics of Reproductive Medicine, University of Maastricht, Netherlands

WS10. Legal regulation for genetic testing (M . Macek, Jr . L. Lwoff) — Room G1+G2

Introduction:
- Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes, 15'
Laurence Lwoff (Bioethics Division, Health and Bioethics Department, Council of Europe, Strasbourg, France)

Examples of national legal provisions related to genetic testing (in alphabetical order):
- Dietmar Vybiral (Abteilung für Gentechnik, Bundesministerium für Gesundheit, Vienna, Austria), 10'
- Holger Tönnies (Robert Koch-Institut, Geschäftsstelle Gendiagnostik-Kommission, Berlin, Germany), 10'
- Jorge Sequeiros (UnIGENe, IBMC, Universidade de Porto, Portugal), 10'
- Michael Morris (Labo de Diagnostic Moléculaire, Service de Médecine Génétique, Hôpitaux Univ de Genève, Switzerland), 10'

Case presentation:
- Genetic variants as extenuating circumstance: a controversial legal proceeding in Italy, 10'
Francesca Forzano, Pascal Borry, Anne Cambon-Thomsen, Shirley V Hodgson, Aad Tibben, Petrus de Vries, Carla van El and Martina Cornel (S.S.D.Genetica Medica, E.O. Ospedali Galliera, Genova, Italy)

WS11. Chemoprevention and chemotherapy of hereditary cancers (J . Lubinski, J . Burn) — Room G3

Chemoprevention in cancer family syndromes.
John Burn, Institute of Human Genetics, Newcastle University, Newcastle upon Tyne, United Kingdom

Selenium and Cancer risk in BRCA1 carriers.
Jan Lubinski, Internation Hereditary Cancer Center, Pomeranian Medical University, Szczecin, Poland

Chemotherapy depending on genetic constitutional and somatic changes.
Evgeny Imyanitov, N.N. Petrov Institute of Oncology, St Petersburg, Russia

Chemotherapy with cisplatinum for BRCA1 carriers.
Tomasz Byrski, Internation Hereditary Cancer Center, Pomeranian Medical University, Szczecin, Poland

WS12. Molecular cytogenetics (J . Vermeesch, N. de Leeuw) — Congress Hall

15.00-15.15 Universal reference samples for diagnostic copy number variation analysis (Poster P03.017)
D. E. Barton, J. de Ligt, J. Y. Hehir-Kwa, C. Brady, J. A. Veltman
15.15-15.30 Parental insertional balanced translocations are an important cause of apparently de novo CNVs in patients with developmental anomalies (Poster P03.064)
B. A. Nowakowska, N. de Leeuw, R. Thoelen, R. Pfundt, H. Mieloo, B. de Vries, J. P. Fryns, J. R. Vermeesch
15.30-15.45 Array CGH; an excellent tool in the search for low-level mosaics (Poster P03.084)
B. Sikkema-Raddatz, L. Leegte, K. Kok, C. van Ravenswaaij-Arts, R. Sinke, T. Dijkhuizen
15.45-16.00 Genome wide 250k SNP array analysis for diagnosing pregnancies with ultrasound anomalies (Poster P05.61)
B. H. W. Faas, R. Pfundt, I. van der Burgt, A. J. A. Kooper, J. Hehir-Kwa, A. P. T. Smits, N. de Leeuw
16.00-16.15 Molecular karyotyping using SNP-array analysis in 100 cases of prenatal diagnosis (Poster P05.62)
M. S. Rocca, L. Esposito, P. Gasparini, V. Pecile
16.15-16.30 Single cell array CGH applied in pre-implantation genetic diagnosis
E. Vanneste, Thierry Voet, J. R. Vermeesch