ESHG

THE EUROPEAN SOCIETY OF HUMAN GENETICS

EMPAG
EUROPEAN MEETING ON PSYCHOSOCIAL ASPECTS OF GENETICS

EUROPEAN HUMAN GENETICS CONFERENCE 2014

in conjunction with the

European Meeting on Psychosocial Aspects of Genetics 2014

May 31 - June 3 , Milan, Italy

PROGRAMME

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1
cont.	**C18 Large scale genomics** Chair: O. Zuffardi, H. Scheffer	**C19 Internal organs** Chair: M. Zollino, B. Melegh	**C20 Basic mechanisms in genetics** Chair: B. Franco, S. Lyonnet	**C21 Rasopathies and CDG** Chair: F. Sangiuolo, K. Writzl	**C22 Returning results: Ethical and legal issues,** joint with EMPAG Chair: F. Faravelli, M. Cornel
11.45	**C18.4** **Planar cell polarity gene mutations contribute to the etiology of human Neural Tube Defects** ***Patrizia De Marco**, E. Merello, G. Piatelli, A. Cama, Z. Kibar, V. Capra;* *Genova, Italy*	**C19.4** **Identification and functional characterization of ESR2, a new disease gene for 46,XY disorders of sex development (DSD)** ***Dorien Baetens***, T. Guran, L. De Cauwer, L. Looijenga, K. De Bosscher, M. Cools, E. De Baere;* *Ghent, Belgium*	**C20.4** **Pseudoautosomal region 1 length polymorphism in the human population** ***Martin A. Mensah***, M.S. Hestand, M.H.D. Larmuseau, M. Isrie, N. Vanderheyden, M. Declercq, E.L. Souche, J. Van Houdt, R. Stoeva, H. Van Esch, K. Devriendt, T. Voet, R. Decorte, P.N. Robinson, J.R. Vermeesch;* *Leuven, Belgium*	**C21.4** **A mutation in PAK3 with a dual molecular effect deregulates the RAS/MAPK pathway and drives an X-linked syndromic phenotype** ***Pamela Magini***, T. Pippucci, I. Tsai, S. Coppola, E. Stellacci, A. Bartoletti-Stella, D. Turchetti, C. Graziano, G. Cenacchi, I. Neri, D.M. Cordelli, V. Marchiani, R. Bergamaschi, G. Gasparre, G. Neri, L. Mazzanti, A. Patrizi, E. Franzoni, G. Romeo, D. Bordo, M. Tartaglia, N. Katsanis, M. Seri;* *Bologna, Italy*	**C22.4** **International views on sharing incidental findings from whole genome research** ***Anna Middleton**, M. Parker, C. Wright, H. Firth, E. Bragin, M. Hurles, O. DDD Project;* *Cambridge, United Kingdom*
12.00	**C18.5** **Clinical exome sequencing for cerebellar ataxia and spastic paraplegia reveals novel gene-disease associations and uncovers unanticipated rare disorders** ***Erik-jan Kamsteeg**, B.P. van de Warrenburg, S.T. de Bot, M.A.A.P. Willemsen, S. Vermeer, M.I. Schouten, R. Meijer, M. Pennings, C. Gilissen, H. Scheffer;* *Nijmegen, Netherlands*	**C19.5** ***LRP5* variants associated with development of polycystic kidney and liver disease** ***Wybrich R. Cnossen***, R.H.M. te Morsche, A. Hoischen, C. Gilissen, H. Venselaar, S. Mehdi, C. Bergmann, M. Losekoot, M.H. Breuning, D.J.M. Peters, J.A. Veltman, J.P.H. Drenth;* *Nijmegen, Netherlands*	**C20.5** **Comparative proteomic analysis of different fragile X syndrome cell lines** *S. Lanni, F. Palumbo, M. Goracci, G. Mancano, A. Vitali, V. Marzano, F. Iavarone, F. Vincenzoni, M. Castagnola, P. Chiurazzi,* ***Elisabetta Tabolacci**, G. Neri;* *Rome, Italy*	**C21.5** **Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis** ***Francesca Pantaleoni***, M. Jaiswal, E. Flex, S. Martinelli, M. Strullu, E.K. Fansa, A. Caye, A. De Luca, F. Lepri, L. Pannone, S. Paolacci, G. Bocchinfuso, C. Rossi, A. Farrotti, O. Fenneteau, B. Brethon, P. Cianci, E. Di Schiavi, A. Selicorni, B. Dallapiccola, I.C. Cirstea, L. Stella, M. Zenker, B.D. Gelb, H. Cavé, M.R. Ahmadian, M. Tartaglia;* *Roma, Italy*	**C22.5** **Newborn screenings and whole genome sequencing: the real need of a genuine public involvement** ***Marta Tomasi**, A. Santosuosso;* *Trento, Italy*
12.15	**C18.6** **WES detects disease causing SNVs and CNVs in Primary immunodeficiencies** ***Hanne S. Sorte**, A. Stray-Pedersen, P.S. Samarakoon, L. Forbes, T. Gambin, O.K. Rødningen, I.C. Hanson, L.M. Noroski, C. Davis, F. Seeborg, S.K. Nicholas, J.W. Caldwell, N.Y. Chokshi, D. Bayer, C.R. Beck, T.J. Vece, W. Wiszniewski, S.J. Penney, S.N. Jhangiani, D. Muzny, L.O. Mæhle, A. Patel, H.C. Erichsen, T.G. Abrahamsen, J. Buchner, G.E. Tjonnfjord, P. Aukrust, L.T. Osnes, M.A. Kulseth, D.E. Undlien, W.T. Shearer, B. Fevang, R.A. Gibbs, R. Lyle, J.S. Orange, J.R. Lupski;* *Oslo, Norway*	**C19.6** **Digenic model in Alport syndrome** ***Maria Antonietta Mencarelli***, M. van Geel, H. Storey, C. Fallerini, L. Dosa, M. Antonucci, F. Cetta, A. van den Wijngaard, S. Yau, F. Mari, M. Bruttini, F. Ariani, K. Dahan, B. Smeets, F. Flinter, A. Renieri;* *Siena, Italy*	**C20.6** **RNA-DNA Differences in Endoplasmic Reticulum Stress Response** ***Allison L. Richards***, S. Liu, Z. Zhu, V.G. Cheung;* *Ann Arbor, United States*	**C21.6** **A New Mouse Model for Costello Syndrome** ***Tania Sorg**, B. Arveiler, M. Birling, G. Bou About, M. Champy, F. Dupuy, I. Goncalves, M. Jagla, H. Jacobs, H. Meziane, G. Pavlovic, N. Philip, F. Radvanyi, R. Rossignol, M. Roux, S. Sigaudy, Y. Herault, D. Lacombe;* *Illkirch, France*	**C22.6** **Current Developments in the Regulation of Direct-to-Consumer Genetic Testing in Europe** ***Louiza M. Kalokairinou***, H.C. Howard, P. Borry;* *Leuven, Belgium*
12.30 - 13.30	*Lunch Break on Level 1 & 2*				

Presentations highlighted by an asterisk (*) and a grey background are from Young Investigator Award Finalists.

17:30 - 19:00 **EES2 - EMPAG Educational Session: Qualitative and quantitative methods in psychosocial research** **Room Amber 3+4**

Chair: B. Ignacio, C. Bjorvatn

EES2.1 Qualitative and quantitative methods in psychosocial research
K. O'Doherty,
Guelph, Canada

Bettina Meiser;
Randwick, Australia

20:30 *Networking party*

Tuesday, June 3, 2014

09:00 - 10:30 **ESHG-ASHG Building Bridges Session PL3: „Towards finding global agreement on..." What IF... (Incidental Findings), an interactive Debate - joint with ESHG** **Gold Room**

Moderator: **Han Brunner, The Netherlands**

Discussants:

- ***Angus Clarke**, United Kingdom*
- ***Martina Cornel**, The Netherlands*
- ***Robert Green**, United States*
- ***Stephen Kingsmore**, United States*
- ***Marjolijn Kriek**, The Netherlands*
- ***Arnold Munnich**, France*

10:30 - 11:00 *Coffee break*

11:00 - 12:30 **C22 - Returning results: Ethical and legal issues, joint with ESHG** **Space 1**

Chair: F. Faravelli, M. Cornel

11:00 **C22.1 The impact of reporting exome and whole genome sequencing: Predicted frequencies of primary, secondary and incidental findings based on modelling**
***Leslie Burnett**, L.C. Ding, R.M. Lew, D. Chesher, A.L. Proos;*
Sydney, Australia

11:15 **C22.2 Defending the child's right to an open future concerning genetic information.**
***Annelien L. Bredenoord**, M.C. de Vries, J.J. van Delden;*
Utrecht, Netherlands

11:30 **C22.3 Implementation of a duty-to-recontact system in molecular and clinical genetics: perspectives from professionals and patients**
***Mirjam Plantinga**, W. Lamers, A.V. Ranchor, M.A. Verkerk, E. Birnie, I.M. van Langen;*
Groningen, Netherlands

11:45 **C22.4 International views on sharing incidental findings from whole genome research**
***Anna Middleton**, M. Parker, C. Wright, H. Firth, E. Bragin, M. Hurles, O. DDD Project;*
Cambridge, United Kingdom

12:00 **C22.5 Newborn screenings and whole genome sequencing: the real need of a genuine public involvement**
***Marta Tomasi**, A. Santosuosso;*
Trento, Italy

12:15 **C22.6 Current Developments in the Regulation of Direct-to-Consumer Genetic Testing in Europe**
***Louiza M. Kalokairinou**, H.C. Howard, P. Borry;*
Leuven, Belgium

End of Meeting

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Final Programme