EUROPEAN HUMAN GENETICS CONFERENCE 2014

in conjunction with the European Meeting on Psychosocial Aspects of Genetics 2014

May 31 - June 3, Milan, Italy

PROGRAMME
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<th>Time</th>
<th>Gold Room</th>
<th>Space 3+4</th>
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| 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. Sangiuliano, K. Witzel | 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. Platei, A. Cama, Z. Kibar, V. Capra; Genova, Italy | C19.4 Identification and functional characterization of ESRR2, a new disease gene for 46,XY disorders of sex development (DSD)  
Dorien Baetens*, T. Gurau, L. De Cauwer, L. Loojenga, K. De Bosscher, M. Coois, E. De Baere; Ghent, Belgium | C20.4 Pseudautosomal region 1 length polymorphism in the human population  
| 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.J.A.P. Willemsen, S. Vermeersch, M.J. Schouten, R. Meijer, M. Penning, C. Gillissen, H. Schellen, Nijmegen, Netherlands | C19.5 LRP5 variants associated with development of polycystic kidney and liver disease  
S. Lanini, F. Palumbo, M. Goracci, G. Mancano, A. Vitalli, V. Marzano, F. Iavarone, F. Vincenzoni, M. Castagnola, P. Chiarotti, Elisabetta Tabocchi; G. Neri; Rome, Italy | C21.5 Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaeomagenesis  
Marta Tomasi, A. Santosussi; Trento, Italy |
| 12.15 | C18.6 WES detects disease causing SNVs and CNVs in Primary immunodeficiencies  
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. Dahlan, B. Smets, F. Flinter, A. Renieri; Siena, Italy | C20.6 RNA-DNA Differences in Expression of Core Curricular Stress Response  
Alison L. Richards*, S. Liu, Z. Zhu, V.G. Cheung; Ann Arbor, United States | C21.6 A New Mouse Model for Costello Syndrome  
Louiza M. Kalokairinou*, H.C. Howard, P. Bony; Leuven, Belgium |

Lunch Break on Level 1 & 2

Presentations highlighted by an asterisk (*) and a grey background are from Young Investigator Award Finalists.
**Programme EMPAG 2014 - Tuesday**

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

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**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.G. 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. Birmie, 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 Tomasí,* 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

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*End of Meeting*
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Italian Society of Human Genetics (SIGU)

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Final Programme