**General Information General Information** 

This international conference (now in its 47th year) is a forum for all workers in human and medical genetics to review advances and develop research collaborations. The conference has become one of the premier events in the field of human genetics with over 3.000 delegates, more than 100 oral presentations, 15 workshops, and 9 educational sessions. The ESHG conference is where the latest developments in human genetics are discussed, and where professionals from all parts of human genetics meet.

#### **Programme**

- Invited Plenary lectures and Symposia
- ESHG Award and Mendel lectures
- "Educational Track" throughout the meeting
- Workshops
- Concurrent Sessions of submitted abstracts
- Poster presentations of submitted abstracts
- Young Scientist and Poster Awards
- Conference Fellowships for young researchers from central and eastern Europe, as well as Fellowships for **National Societies**
- Corporate Satellites
- · Over 150 exhibitors from all over the world

Further details The website www.eshg.org/eshg2014 will open in

November 2013

**Abstract** 

Online abstract submission via www.eshg.org/eshg2014

submission Closing date: February 14, 2014

Scientific & Administrative

ESHG 2014 c/o Vienna Medical Academy Alser Strasse 4, 1090, Vienna, Austria

Tel: +43 1 405 13 83 16 Conference Email: conference@eshg.org **Secretariat** 

Exhibition, Sponsoring, Rose International

P.O.Box 93260, 2509 AG The Hague Corporate The Netherlands

**Satellites** Tel: +31 70 383 8901

Email: eshg@rose-international.com

Further information on programme, registration and abstract submission:

www.eshg.org/eshg2014

#### ESHG Scientific Programme Committee 2013 - 2014 (as per date of printing)

Brunhilde Wirth. Chair Germany Antonio Amoroso, Local Host United Kingdom Jeffrey Barrett Alexis Brice France Martina Cornel The Netherlands Paul de Bakker The Netherlands Hélène Dollfus France David Fitzpatrick **United Kingdom** Maurizio Genuardi **Daniel Grinberg** Spain Gunnar Houge Norway Erik Iwarsson Sweden Xavier Jeunemaitre France Lidia Larizza Italy Jose Carlos Machado Portugal Giovanni Neri Italy Finland Minna Nyström Francesc Palau Spain Aarno Palotie United Kingdom Anita Rauch Switzerland Peter N. Robinson Germany Marco Seri Italy Joris Veltman The Netherlands Joris Vermeesch Belgium

#### **Co-Chairs EMPAG Scientific Programme Committee**

Tara Clancy United Kingdom Elisabetta Razzaboni

#### ESHG Annual Meetings Committee 2013 - 2014

President: Han Brunner, NL Helena Kääriäinen. FI President-elect: Vice-President: Stan Lvonnet, FR Secretary General: Gunnar Houge, NO Deputy Secretary General: Karin Writzl, SI Andrew Read, UK Chair of the AMC: Chair of the SPC: Brunhilde Wirth, DE Local Host: Antonio Amoroso, IT **Executive Officer:** Jerome del Picchia, AT Advisor: Jantie de Roos, NL Mirjam Uebelhör, AT Advisor:

The European Society of Human Genetics promotes research in basic and applied human and medical genetics and facilitates contact between all persons who share these aims.

# ESHG 2014 EMPAG



Milan, Italy, May 31 - June 3 EUROPEAN SOCIETY OF HUMAN GENETICS

## **European Human Genetics Conference**

in conjunction with the

European Meeting on Psychosocial Aspects of Genetics



www.eshg.org

### Invitation to the 47<sup>th</sup> European Human Genetics Conference

Preliminary Schedule ESHG 2014

Dear Colleagues and Friends,

It was 1997 in Genoa the last time when the European Society of Human Genetics met in Italy, so it was time to be back. On behalf of the Italian Society of Human Genetics (SIGU). I am honored to invite you to attend the European Human Genetic Conference 2014 that will be held in Milan May 31 - June 3, 2014.

Milan, known as the industrial capital of Italy, also treasures a notable artistic as well as scientific tradition. Just think of Leonardo da Vinci, the visionary genius, author of the Codex Atlanticus and of the Last Supper, that you will be able to admire in the Biblioteca Ambrosiana and in the Refectory of S. Maria delle Grazie, respectively. In that tradition, Milan is today the place of many Universities as well as a host of research institutes, providing the right climate for an international scientific meeting, such as the ESHG Conference. Likewise, the Italian Society of Human Genetics will contribute to the success of the Conference with a large participation of its members and with all possible measures of support.

Human genetics continues to progress at an unprecedented pace and every annual meeting promises to bring new data to the attention of the scientific community, fostering productive debates and further progress. In this respect, the Milan Conference will not differ from previous ones. Therefore, I expect a numerous and enthusiastic participation. I can promise that your visit will be a memorable one.

Arrivederci a Milano in 2014!

Antonio Amoroso President, Società Italiana di Genetica Umana

Cover picture by courtesy of Liz Steel

| Thon  | reliminary programme is subject to alterati   | ion D                | Place          | consult the website as of Neve   | mbor 2013               |   |                         |       |  |
|-------|---|----------------------|----------------|--|-------------------------|---|-------------------------|-------|--|
| Time  | Saturday, May 31  |                      | Time           | Sunday, June   |                         | Monday, June  | 2                       | Time  | Tuesday, June 3  |
| 10.15 | Registration opens  Educational Sessions  ES1: The platelets planet: from diagnosis to  | 0                    | 08.30          | Concurrent Symposia S01: Towards Genomic Personalised Medicine S02: Functional genomics S03: Neuronal Migration disorders S04: Computational Analysis of Gene Networks S05: Early development & preimplantation genetics  Educational Session ES5: Mosaicism in human disease                  |                         | Concurrent Symposia S11: Rare copy number variants in common traits S12: Epigenetic basis of disease S13: Non-invasive prenatal testing, joint with EMPAG S14: Rapid genome diagnostics  Educational Session ES7: From mutation identification to therapy |                         | 09.00 | Plenary Session 3  Debate: What IF (incidental findings), joint with EMPAG |
|       | therapy of inherited thrombocytopenias  | lites                | 10.00          | College Broak  |                         | Coffee Break  Poster Viewing with Authors   |                         |       |  |
|       | ES2: Genetic predictions scores in common diseases: are they of any value? ES3: What's new in Next Generation Sequencing? ES4: DNA Repair and genomic instability | Corporate Satellites | 10.30          |  |                         |   |                         | 11.00 | Coffee Break  Concurrent Sessions C19 - C24  from submitted abstracts      |
|       | Workshop W01: Disease of the year: Rasopathies  | S 1                  | 11.45          | Free Poster viewing - Lunch  | Corporate<br>Satellites | Free Poster viewing - Lunch   | Corporate<br>Satellites |       |  |
| 12.00 | Lunch Break   | 1                    | 13.30          | Concurrent Sessions (  | C07 - C12               | Concurrent Sessions   | C13- C18                | 12.30 | Lunch Break  |
| 14.00 | Welcoming Addresses<br>Opening  | 1                    | 15.00          | from submitted abstracts   |                         | from submitted abstracts  Vitamin Break   |                         | 13.30 | Plenary Session 4 Mendel Lecture   |
| 14.30 | Opening Plenary Session 1   |                      | 15.30          | <b>Workshops</b> W02: Dysmorphology 1 W03: ENSEMBL   |                         | Workshops W08: Dysmorphology 2 W09: Genome Browser UCSC   |                         | 14.15 | Plenary Session 5 EJHG Nature Awards                                       |
| 16.00 | Coffee Break  |                      |                | W04: Practical Bioinformatics: Whole exome   |                         | W10: Array CGH  |                         |       | Young Investigator Awards  |
| 16.30 | Plenary Session 2 What's new? from submitted abstracts  |                      |                | sequence analysis W05: Quality assurance W06: Community genetics W07: Preimplantation EMPAG W1: The impact of risk reducing surgery  |                         | W11: Clinical Cancer Genetics Club W12 Preconception and prenatal screening W13: Next Generation Sequencing EMPAG W2: New research proposals/research networking  |                         |       | Poster Awards<br>ESHG Award Lecture<br>Closing                             |
|       |   |                      |                | Corporate Satellites   |                         | Corporate Satellites  |                         | 15.45 | End of Meeting   |
| 18.00 | Coffee Break  |                      | 17.00          | Coffee Break   |                         | Coffee Break  |                         |       |  |
| 18.30 | Concurrent Sessions C01 - C06 from submitted abstracts  |                      | 17.30          | Concurrent Symposia S06: Risk perception & risk communication, joint with EMPAG S07: Therapy for human genetic diseases S08: Population genetics: from Neanderthals to a globalized world S09: Advances & new challenges in genetics of cardiovascular diseases S10: New Mutational Mechanisms |                         | Concurrent Symposia S15: Networks and pathways in genetic diseases S16: The new RNA world S17: Tumour heterogeneity S18: A novel class of disease of lipid metabolism S19: Three sequencing approaches in complex disease                                 |                         |       | Abstract submission deadline   |
| 20.00 | Networking Mixer  |                      |                | Educational Sess<br>ES6: How to be successful in ra<br>Identification  |                         | Educational Ses<br>ES8: Current developments in lega<br>Untangling the law & what it  | l aspects of genetics:  |       | act gubruary   |
|       | Networking Mixer  | 4                    | 10 00          |  |                         |   |                         |       |  |
|       | Networking Mixer  | <u> </u>             | 19.00<br>21.00 | Corporate Satelli  | ites                    | Corporate Satel  Networking Par   |                         | -     | Abstr Pe   |