

European Society of Human Genetics - ESHG

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European Human Genetics Conference 2014 - May 31 to June 3, Milan, Italy

Abstract Submission Countdown: 31 days left to submit your abstract!

ESHG Abstract Submission closes Friday, February 14, 2014, 24.00 hrs CET

The Scientific Programme Committee (SPC) invites abstracts to be submitted for inclusion in the programme as posters or spoken presentations in 24 Concurrent Sessions.

Abstracts can be submitted only online via the website:

https://www.eshg.org/abstracts2014.0.html

Extended deadline for submission: Friday, February 14, 2014, 24.00 hrs CET

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Invitation to Milan

Dear colleagues,

The last time the European Society of Human Genetics met in Italy, it was 1997 in Genoa, so it is time to come back. On behalf of the Italian Society of Human Genetics (SIGU), I am honoured 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 ESHG 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 the 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, Societa Italiana di Genetica Umana and Local Host of the ESHG 2014

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ESHG Scientific Programme

Dear Colleagues,

The Scientific Programme Committee has again endeavoured to set up an exciting programme at the cutting-edge of current human genetics research.

The programme includes Plenary Sessions, Concurrent Symposia, Concurrent Sessions from submitted abstracts, Workshops, Educational Sessions (more education-focussed, broader-view-type of sessions, scheduled throughout the meeting in parallel to the Symposia) and Poster Sessions with ample viewing time.

In particular, we are proud to announce, that the ESHG and the American Society of Human Genetics (ASHG) will be cooperating on a scientific level in the form of joint sessions at their annual meetings. The first in the hopefully very interesting series of joint sessions will be a debate on Tuesday, June 3, 2014: WHat IF...? (Incidental Findings).

Plenary Lectures include

- **P1 Opening Plenary Session** (M. Tartaglia, E. Cattaneo, L. Naldini)
- **P2 What's New?** (from submitted abstracts)
- P3 What IF...? (Incidental Findings), an interactive debate joint with ASHG and EMPAG (A. Clarke, M. Cornel, R. Green, S. Kingsmore, M. Kriek (tbc), A. Munnich, Moderated by H. Brunner)
- **P4 Mendel Lecture** (M. Capecchi)
- **P5 ESHG Award Lecture** (M. Stratton)

Concurrent Symposia include

- **S01. Towards Genomic Personalised Medicine** (M. Summar, A. Antonidou, C. Klaver)
- **S02. Functional genomics** (B. Deplancke, M. Georges, J. Marioni
- **S03. Neuronal Migration disorders** (K. Kutsche, A. Wynshaw-Boris, J. Chelly)
- S04. Computational Analysis of Gene Networks (C. Webber, H. Yu, R. Sharan)
- **S05.** Early development and preimplantation genetics (R. Pera, T. Voet)
- **S06. Risk perception and risk communication, joint with EMPAG** (B. Peshkin, C. Julian-Reynier, D. Timmermans)
- **S07.** Therapy for human genetic diseases (A. Ballabio, E.H. Epstein, R. Semple)
- **S08. Population genetics: from Neanderthals to a globalized world** (I. Pe'er, S. Myers, P. Gasparini)
- S09. Advances and new challenges in genetics of cardiovascular diseases joint with the European Society of Cardiology (ESC) (H. Watkins, M.V. Holmes, P. Charron)

- **S10.** New Mutational Mechanisms (E. Cuppen, S. Nik-Zainal, J.O. Korbel)
- S11. Rare copy number variants in common traits (X. Zhang, B.Keavney, C. Thiel)
- S12. Epigenetic basis of disease (A.P. Feinberg, A. Ferguson-Smith, P.A. Jones)
- **S13. Non-invasive prenatal testing, joint with EMPAG** (L. S Chitty, D.W. Bianchi, K. Ormond)
- **S14. Rapid genome diagnostics** (J. O'Halloran, S. Kingsmore, C.U. Köser)
- S15. Networks and pathways in genetic diseases (C. Petit, J. Surrallés, P.J. Scambler)
- **S16.** The new RNA world (S. Gustincich, D. Gabellini, L. Pellizoni)
- **S17. Tumour heterogeneity** (A. Bardelli, A. Marusyk, K. Pantel)
- S18. A novel class of disease of lipid metabolism (H.H. Freeze, F. Mochel, F.M. Vaz)
- **S19.** Three sequencing approaches in complex disease (N. Soranzo, T. Lappalainen, F. Cucca)

All details on the programme can be found at https://www.eshg.org/programme2014.0.html.

With best regards and my sincere wishes for a successful year ahead,

Brunhilde Wirth Chair of the ESHG Scientific Programme Committe

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"Educational Track" throughout the Meeting

The successful series of Educational Sessions will be continued with

- ES1. The platelets planet: from diagnosis to therapy of inherited thrombocytopenias (A. Savoia, C.L. Balduini)
- ES2. Genetic prediction scores in common diseases: are they of any value? (Samuli Ripatti, A. Hingorani)
- ES3. What's new in Next Generation Sequencing? (A. Hoischen, J. Lundeberg)
- ES4. DNA repair and genomic instability (J. Jiricny, J. Hoeijmakers
- **ES5. Mosaicism in human disease** (N. Spinner, M.F. Jonkmann)
- **ES6.** How to be successful in rare disease gene identification (F. Alkuraya, K. Boycott)
- **ES7. From mutation identification to therapy** (M.R. Hayden, C. Bear)
- ES8. Current developments in legal aspects of genetics: Untangling the law and what it means for you (D. Barton, D. Townend)

Further details on educational sessions can be found at https://www.eshg.org/education2014.0.html.

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Workshops

2014 Workshops include:

- WS01. Disease of the year: Rasopathies (Organiser: G. Neri)
- WS02. Dysmorphology 1 (Organisers: D. Donnai & J. Clayton-Smith)
- WS03. ENSEMBL

- WS04. Practical Bioinformatics: Whole exome sequence analysis (Organiser: N. Robinson)
- WS05. Quality assurance (Organiser: E. Dequeker)
- WS06. Community genetics (Organiser: M. Cornel)
- WS07. Preimplantation (Organisers: J. Vermeesch, E. Iwarsson)
- WS08: Dysmorphology 2 (Organisers: D. Donnai & J. Clayton-Smith)
- WS09: Genome Browser UCSC (Organiser: R. Kuhn)
- WS10: Array CGH (Organisers: N. de Leeuw & C. van Ravenswaaij-Arts)
- WS11: Clinical Cancer Genetics Club (Organisers: M. Genuardi & D. Stoppa-Lyonnet)
- WS12 Preconception and prenatal screening (Organiser: M. Macek Jr.)
- WS13: Next Generation Sequencing (Organiser: J. Veltman)

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ESHG Young Investigator Awards

The ESHG awards prizes of EUR 500.- for outstanding research by young scientists presented as a spoken contribution at the conference, namely:

- Young Investigator Awards for Outstanding Science
- Isabelle Oberlé Award for Research on Genetics of Mental Retardation
- Lodewijk Sandkuijl Award for the best talk in Statistical Genetics
- Vienna Medical Academy Award for the best talk in translational genetics/genetic therapy
- ESHG Poster Awards

All young scientists submitting spoken presentations are encouraged to apply. The nominee must be first author (i.e. presenting author) on an abstract submitted for spoken presentation and should **not be more than four years post-doctoral and not a Principal Investigator (P.I.)**.

The best scored abstracts in each topic, which could not be selected for oral presentation will automatically compete for the **ESHG Poster Award**.

Authors wishing to be considered for these awards should complete the appropriate boxes during <u>abstract submission</u>.

Closing date: February 14, 2014

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ESHG Conference Fellowships

A number of **conference fellowships** will be awarded by the Scientific Programme Committee for applicants from Central and Eastern Europe. Authors wishing to be considered for a fellowship should complete the appropriate box during <u>abstract submission</u>. The nominee must be first author (i.e. presenting author) of an abstract submitted for either oral or poster presentation, and should be not more than six years postdoctoral.

Closing date: February 14, 2014

The ESHG is glad to make available to **each European National Human Genetics Society, one fellowship of EUR 800.-** (& free registration) for a young researcher (<35 years, member of the

society) having shown excellence in his or her field in the past year. **The allocations of these fellowships are at the discretion of each National Society**. Please contact your local society for further details and on how to be a candidate.

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Registration

Online Registration is open. Considerable discounts apply to **ESHG members**. Dedicated fees for post-docs, students and counselors are available.

Special rates apply for ESHG members (Join/Renew your membership).

Please note that higher fees apply for *on-site registrations*.

Registration forms can be found at https://www.eshg.org/registration2014.0.html

Deadline for registration at early fee: March 31, 2014

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Technical Exhibition and Satellite Symposia

The European Human Genetics Conference 2014 will host a large industry exhibition, where the latest technologies will be shown. In addition, there will be a number of company satellites during the conference, always of great interest to the participants.

For further information see <u>the list of exhibitors</u> as to date, as well as the <u>company satellites</u> <u>information</u>.

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European Meeting on Psychosocial Aspects of Genetics 2014



Dear Colleagues,

The biennial European Meeting on Psychosocial Aspects of Genetics (EMPAG) will be held on May 31-June 3, 2014 in Milan, Italy. It will be held concurrently with the European Society of Human Genetics Conference, and there will be a number of joint sessions with the ESHG, these include two symposia and one plenary session □.

The EMPAG meetings have gained a reputation as a key event for those involved in psychosocial research as well as those providing clinical services, in particular genetic counselling. As usual, colleagues from a diverse range of disciplines will be welcome.

The deadline for abstract submission will be February 14, 2014, and the EMPAG website will be accessible via the ESHG website http://www.eshg.org/empag2014

We look forward to seeing you in Milan.

Tara Clancy (Manchester, UK) and Elisabetta Razzaboni (Modena, Italy)

Co-chairs of the EMPAG Scientific Programme Committee

EMPAG SPC Members

Cathrine Bjorvatn (Bergen, Norway) Ignacio Blanco (Barcelona, Spain) Nina Hallowell (Cambridge, UK) Maria Regele (Bolzano, Italy) Sam Riedijk (Rotterdam, Netherlands)

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More Information

All available information can be found on the regularly updated website.

European Human Genetics Conference 2014: www.eshg.org/eshg2014

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