THE EUROPEAN SOCIETY OF HUMAN GENETICS

NEWSLETTER

No. 27 - January 2016

Presidential Address

by Feliciano Ramos, President of the ESHG

Genetics goes partnering

Dear ESHG members and readers:

Welcome to this new issue of the ESHG Newsletter ! and thank you to take some of your time to read it. I am sure if you do, your time has not been wasted.

Nowadays, in most of its fields, Medicine can hardly progress without the help of (Human) Genetics. This poses a big responsibility for human geneticists when collaborating with other medical specialists in the management of patients and families. This responsibility extends to the whole society, since each of us are part of a team of healthcare providers in our workplace. ESHG has been taking over this challenge for many years, helping and guiding human genetics professionals to deal with their tasks of everyday life at work all over Europe, and even beyond its political borders. The influence of the ESHG outside of the EU has been increasing year after year, thanks to the efforts of all the ones that one day served as members of the Board or the Committees. Our Annual Conferences have been steadily gaining in prestige and respect of our colleagues around the globe, due to the top-quality science that has been shared there. I hope this exciting trend continues for many years to come, specially now that we are approaching our 50th Anniversary (Copenhagen, 2017). This President is much conscious of our leading position and, with the help of the Board and the Committees, and the collaboration of all of you as members, I will work as hard as I can to keep ESHG at the frontline.

I have proposed some changes to the ESHG Board and some of them have already being implemented. One is to look for that young ESHG members play a more relevant role in our Annual Conference by including the "young investigator price" awardees of the previous year as co-chairs of oral sessions/symposia/workshops in the following year´s Conference. This will be, hopefully, first implemented in Barcelona 2016, thanks to the good work of our current SPC. We (senior and "not-so-young" members) all know that the new generations of human geneticists will likely be the leading force of our profession. We want to look for them and recognize them.

Relating to that, we are considering to appoint one or two (clinical and/or laboratory) young ESHG members to sit in the Board. I (we) think that this decision will help to bring "fresh air" (i.e. new ideas) to our Society. Hopefully we will be able to make it possible soon.

Another issue I am interested in as President is to broaden the influence of ESHG in human genetics across EU borders. I found a way to start after I had my last conversation with Dr. Aida Falcón, current President of the RELAGH (Latin-American Society of Human Genetics). She was worried about several issues that needed to be addressed in her Society, one was the problems they have had with the RELAGH website which has been virtually inactive for years. I promised her to bring that to the ESHG Board to find the best way we could help, maybe offering our technical and professional experience. Education of young human geneticists in Latin-America was something we discussed as well and, since ESHG offers excellent courses and educational activities, I think it is worthy to talk to our recently renewed Education Committee, chaired by Prof. Han Brunner, who I am sure will be willing to collaborate in that matter. Besides, and since we share the same language with most of the Latin-American countries, the Spanish Society of Human Genetics (AEGH) will be happy to help with translation of educational material. Needless to say that ESHG will be happy to help, if it is requested, any other Human Genetics Societies which may need our help.

Last but not least, and going back to the beginning of my address, I personally believe that our interactions and collaboration with another medical specialists will be exponentially increasing, most of the times because they will look for us and we must be there to share our expertise and professionalism for the good of the patients and families affected by genetic conditions. Such interaction could be extended to our and their Annual Conferences by organizing a sort of "building bridges" session with other specialties strongly related to Genetics (... is there any that is not?). There has been a good example of this in our last Conference in Glasgow where a session was organized in collaboration with the European Society of Cardiology. I hope our SPC will help us to make this possible in future Conferences as well as that Human Genetics will be present in the Annual Conferences/Meetings of other european medical specialties.

As the first ESHG President from Spain I would like to mention that we have not had the specialty of Genetics officially recognized until August 2014 and we are now ready to start the training of our future human geneticists. I know there are still some EU countries where the specialty of (Human) Genetics is not yet officially recognized (i.e. Belgium, Greece, Croatia, ...) to which we must offer all our help and efforts to their respective National Societies to convince their governments to grant official recognition. The European Board of Medical Genetics (EBMG) is working in the different fields of our specialty to establish professional standards of education, training and practice and deliver an EU certification for professionals working in Europe.

Remember to renew your ESHG membership 2016 online:

www.eshgorg/join0.0.html

I do not want to finish without mentioning the last consensus document we have prepared (mostly work of the PPPC) on "Responsible implementation of expanded carrier screening", which is ready to be delivered. I am sure the new Secretary General, Karin Writzl, who has replaced Gunnar Houge, now our Deputy Secretary General (thank you Gunnar for all your fantastic work!), will inform you about other interesting tasks in which ESHG is currently working on. Thanks to all the members of the current Board who have worked hard and timely these first months in office, specially to Jerome del Picchia, our Executive Officer and his team, who simply "make our lives easy".

Hope to see you in Kyoto, next April 2016, at the International Congress of Human Genetics -IFHGS- Meeting (www.ichg2016.org) and in May 2016 at the European Human Genetics Conference in Barcelona, Spain (www.eshg.org/eshg2016)

Feliciano J. Ramos
President of the ESHG

Information on the "IVD Directive"

by David Barton

Everybody knows about drug licensing - a company may not put a drug on the market in the EU unless it has been approved by the European Medicines Agency. You may not be aware that there is a similar regulatory framework for medical devices (everything from breast implants to scalpels) and for in vitro diagnostic medical devices (also known as IVDs). The IVD regulations cover all items used in laboratory diagnostics (from blood collection tubes to large biochemical analyzers) except for general laboratory reagents.

The EU has for many years been in the process of updating the regulatory framework for IVDs, and this process has reached a crucial stage. The Commission published a proposed new Regulation in 2012; the European Parliament proposed over 700 amendments in 2013. Now, the governments (European Council) have published their proposals. Geneticists have been involved in shaping and influencing these proposals for over 7 years now, through EuroGentest and the ESHG's PPPC.

Why are these regulations of interest to clinical and laboratory geneticists? Well, there are several reasons:

1) The Health Institution Exemption

The current regulations or "IVD Directive" give a broad exemption to Health Institution laboratories to manufacture their own tests in house. Unless you are using a CE-marked IVD "kit", you are availing of this exemption, whether you realize it or not.

The new Regulation will tighten up on this exemption in several ways:

a) The exemption will only be available to accredited laboratories
b) Labs manufacturing their own kits or adapting commercial kits will have to justify the need for these kits and will have to comply with a rage of requirements regarding the design and manufacture of the kits. This will involve providing information each year to their national regulatory authorities and may involve inspection of their labs.

2) Requirement for "Genetic Counselling"

The European Parliament has proposed sweeping requirements for those involved in ordering genetic tests, with detailed counseling to be provided by the person who carries out the test and a ban on determination of foetal sex except in cases of serious X-linked disorders. This latter provision would outlaw all genomic prenatal diagnosis, including karyotyping. These alarming provisions are opposed by the Council and the Commission as well as by the ESHG, the PHG Foundation, Wellcome and other prominent organizations. The ESHG has commissioned a legal opinion which asserts that these proposals go beyond the EU's legal powers and would be open to challenge in European courts. However, the Parliament's negotiators are sticking to their guns and it is likely that a compromise will have to be found. It is vital that any such compromise enables appropriate access to genetic testing with a level of genetic counselling proportionate to the risks, and does not stifle innovation.

3) Regulation of Companion Diagnostics

The Parliament has proposed that all genetic tests and all companion diagnostics (i.e. tests used to determine a patient's suitability for a specific drug) should only be available on prescription - that is, they would have to be ordered by a doctor. The ESHG supports this proposal, but it has been argued that this position is too inflexible, and that the proposal should be amended to allow (for example) Genetic Counsellors to order tests, and that individual member states could vary the requirements in the interest of public health.

4) Definition of an IVD Device

The parliament approved a proposal to expand the definition of an IVD device to include devices "with a direct or indirect medical purpose". This has caused some alarm as it is thought that lifestyle apps might then come under the regulation, potentially stifling innovation and over-regulating an important industry which carries little risk to EU consumers. It is important to note here that an app which assesses the characteristics of a mole on your skin and, based on this, advises you to go (or not to go) to your doctor is clearly a medical device. It is not an IVD, by the way, as it is used directly on the "patient" and not on a sample in a lab. On the other hand an app which calculates calories consumed or burnt is less likely to be classified as a medical device. However, this broader definition would also potentially bring "lifestyle genetic testing" such as nutrigenomics under the remit of the Regulation - would this be a good thing?

What happens next?

The next stage of the process is that the Commission, Parliament and Council get together in a "Trilogue" to hammer out a compromise Regulation. The Medical Devices Regulation will be negotiated in parallel, in what promises to be a difficult set of negotiations This process got underway in October and will take several months. After that, the final text would have to be approved by all three bodies and check for legal issues etc before implementation, perhaps by the end of 2016. There would then be a derogation period of 3-5 years for existing tests, but new tests would have to comply sooner than this.

A link to the ESHG website for more information is at www.eshg.org/566.0.html

David Barton
Director of the Division of Molecular Genetics at the Irish National Centre for Medical Genetics in Dublin and ESHG Member

Report from the European Board of Medical Genetics - September 2015

by Heather Skirton, Chair of the EBMG

Latest activities of the EBMG

- There has been consolidation of the registration activities for the Clinical Laboratory Geneticist and Genetic Nurse and Counsellor branches, with satisfactory levels of applications again this year.
- We continue to develop the registration systems for Clinical Laboratory Geneticists and Genetic Nurses and Counsellors. This year, online systems for applications have been developed in conjunction with the ESHG to facilitate the application process for both the applicants and the EBMG assessors.
- We are aware of the possibilities for using the European Professional Qualifications Directive to support professionalization of genetics healthcare in Europe. However, at present we do not have sufficient countries with formal systems in place to support recognition of the Clinical Laboratory Geneticist, Genetic Counsellor or Genetic Nurse as an EU designated specialist profession.
- At the ESHG meeting in Glasgow we ran a workshop to disseminate information about the branches and registration systems.
- We have formed a Complaints Panel, with one member from each professional branch, to deal with any complaints that a registered ErCLG or ERGC/ERGN is not working according to EBMG professional standards. The Complaints process is published on the EBMG website.

a) Clinical Laboratory Geneticist Professional Branch Board

Chair: Thomas Liehr (Germany); Co-Chair: Isabel Carreira (Portugal)

Members: Dilek Aktas (Turkey), Egbert Bakker (The Netherlands), Marta Rodríguez de Alba (Spain), Domenico Coviello (Italy), Lina Florentin (Greece), Martina Rincic (Croatia), Hans Scheffer (The Netherlands).

It is well-known that there is a need for a European recognition of laboratory specialization in our field. The goal of the European Board of Medical Genetics (EBMG), Professional Branch Board for Clinical Laboratory Geneticists (ErCLG) is to provide a system for registration of professionals working in different countries and certifying those being at the same level of education and competences in human genetics laboratory diagnostics. Our efforts are in connection with the implementation of the so-called "green paper", i.e. Professional Qualifications Directive (EU Directive 2013/55/EU) in January 2016 by the European parliament. To the best of our knowledge we will be able to get the ErCLG certificate recognized only after national CLG titles are recognized in 9 member states of the EU by the corresponding national parliaments; at present this is only the case for Hungary.

Nonetheless we are proud to announce that in 2016 we anticipate there will be >300 ErCLGs recognized by the EBMG Professional Branch Board. The first round of application was possible 2014/2015 and 247 applicants were awarded the registration. In the actual 2015/2016 round there are 60 applicants. These applications are presently under evaluation.

Some statistics can be provided as follows. Successful applicants came in 2014/15 from Austria, Belgium, Czech Republic, Finland, Germany, Hungary, Italy, Lithuania, Macedonia, Netherlands, Poland, Portugal, Slovenia, Sweden, and Switzerland. In 2015/16 applications were received additionally from Cyprus, France, Greece, Latvia, Spain, United Kingdom and even Saudi Arabia. Among applicants the female to male ratio is ~2:1, PhD to MSc ratio 4:1, average age of the applicants in 2014/15 was 57 years with a range from 30 to 70 years of age.

It is possible to apply for 5 types of ErCLG:

1. ErCLG - general (172 approved in 2014/15 round),
2. ErCLG - focus on Clinical Genetics (15 approved in 2014/15 round),
3. ErCLG - focus on Molecular Genetics (50 approved in 2014/15 round),
4. ErCLG - focus on Biochemical Genetics (4 approved in 2014/15 round), and
5. ErCLG focus on Tumorgenetics (6 approved in 2014/15 round).

The next round of applications will close 15 September 2016. The prerequisites for application and how to apply can be found on https://www.eshg.org/clg.0.html; please read especially https://www.eshg.org/587.0.html and check under which of three categories falls the country you apply from (https://www.eshg.org/666.0.html). Overall, if you find yourself eligible we cordially invite you to apply for ErCLG registration in September 2016.

b) Genetic Nurse and Counsellor Professional Branch Board

Chairs: Milena Paneque (Portugal) and Ramona Moldovan (Romania)

Members: Inga Bjornevoll (Norway), Christophe Cordier (France), Irene Feroce (Italy), Debby Lambert (Ireland), Clara Serra (Spain), Heather Skirton (UK).

In June 2015 Milena Paneque and Ramona Moldovan became co-chairs of the branch board.

The activities of the past six months have been:

1. An online application system has been designed and will be operational for the full applications to be submitted January 2016.
2. National registration systems offered in Canada, South Africa and Australasia have been assessed and approved. This means applicants working in Europe with registration from US, UK, Canada, South Africa or Australasia can be registered with the EBMG.
3. Requirements and forms for the assessment of new courses that could be used by applicants under the grandfather B clause have been developed.
4. A letter to the editor outlining the development of the European registration system has been accepted by EJHG.
5. We have had a further 21 applications for registration this year and these are being assessed for eligibility.

c) Medical Geneticist Professional Branch Board

Chair: Ulf Kristoffersson (Sweden)

Members: Beata Lipska (Poland) , Béla Melegh (Hungary), Feliciano Ramos (Spain), Alessandra Renieri (Italy), Peter Turnpenny (UK).

A memorandum of understanding detailing the relationship between the UEMS and the EBMG branch has been written and agreed by the EBMG, UEMS section and the ESHG. Bela Melegh has agreed to lead the UEMS section, while Ulf Kristoffersson leads the EBMG Medical section.

Further work has been undertaken to consolidate the curriculum for medical specialists in genetics and obtain approval from relevant parties.

Finally, our thanks to Jerome del Picchia and the staff of the VMA for their support of the administrative work.

Heather Skirton
Chair, European Board of Medical Genetics

Meet the new ESHG Board Members

Kristiina Aittomäki
Helsinki, Finland

Kristiina Aittomäki is the Head of Genetics in the Helsinki University Hospital and Professor of Clinical Genetics at the University of Helsinki. She studied medicine in the University of Helsinki and did first specialist training in gynecology and obstetrics. In her thesis, supervised by professor Albert de la Chapelle, she studied the genetics of premature ovarian failure identifying a recessive form of primary ovarian failure caused by mutations in the FSHR gene. This, with previous interest in genetis, lead to specialist training in Clinical Genetics. She has previously held a senior lecturer position in medical genetics at the University of Helsinki with a senior consultant position in the Helsinki Unviersity Hospital for several years. She has also worked as the Head of Familial Cancer Unit at Victorian Clinical Genetic Services (presently Genetic Health Victoria), Melbourne, Australia. Her research interests include reproductive genetics (a great challenge, as it turns out) and cancer genetics. Kristiina Aittomäki is the secretary of the UEMS Section of Clinical Genetics.

"Finland has 5.2 million people and 0.5 million summer cottages. As so many Finns, we have a summer cottage with a sauna by a lake. This is where we spend most of our free time, preferably reading a good book, a long-lasting interest of mine".

Jill Clayton-Smith
Manchester, United Kingdom

Jill Clayton-Smith has worked in the field of Clinical Genetics since 1987 and has a special interest in genetic causes of intellectual disability and syndrome diagnosis. She is a Consultant and Honorary Professor working within the Manchester Centre For Genomic Medicine in the North West of England. Jill has been an active member of the ESHG for many years as one of the convenors of the annual ESHG Dysmorphology workshops. Her main clinical interests over the years have included clinical and genetic studies of Angelman syndrome and related disorders , paediatric ophthalmic syndromes, and study of effects of exposure to antiepileptic drugs in utero. She has always been keen to work closely with laboratory and other colleagues to translate research findings into clinical practice. From 2007 Jill led the DYSCERNE project , one of the pilot European Reference Networks to enhance diagnosis and management of rare genetic syndromes. From 2013-2015 she was President of the UK Clinical Genetics Society. As part of this role she led a working group looking at the evolving role of the Clinical Geneticist , an area of interest and importance to all of us as we move into the era of exome and genome sequencing. Throughout her career Jill has maintained an interest in genetic education for lay, under-graduate and postgraduate groups. She is one of the organisers of the annual Manchester Dysmorphology Course and most recently has been involved in developing and delivering the new Manchester MsC in Genomic Medicine, one of several such courses being commissioned by Health Education England to increase knowledge of genomics for non-geneticists . Jill hopes that one of the more specific roles she will take on within the ESHG is looking at wider access to genomic education and looks forward to working with the board as our specialty evolves.

Johan den Dunnen
Leiden, The Netherlands

Intrigued by a molecule called DNA I decided to start a study biology at the Nijmegen University. After finishing my study I did my PhD at the same university, specializing in molecular biology. My next stop was Leiden where I applied my expertise as a postdoc to work on Duchenne muscular dystrophy. It were the early days of the Human Genome Project, DNA technology and application became booming and my fascination for DNA became chronic. Over the years I became interested in innovating technology and applied this to study genetic disease, muscular dystrophy in particular. When the internet emerged I started to share our knowledge (technology and DNA variants revealed) on-line and be-came involved in standardisation (HGVS nomenclature) and databases (LOVD).

As a Board member of the ESHG my special interest is ways to further improve DNA-based diagnostics, data sharing and public awareness. DNA diagnostics is possible because we share, worldwide, what we know about genes, variants and phenotypes. Without sharing, DNA diagnostics would not exist. Unfortunately sharing is not (yet) the standard and with simple methods we can achieve significant progress, which would directly benefit the patients and their families. With personal genome sequencing on the horizon we need to share our knowledge in such a way that

lay people can access and understand it. With their DNA read, people will search the internet for reliable information to understand what all these variants mean. In addition we have to explain what you know and what you do not know when you know your DNA. Work enough.

Robert Hofstra
Rotterdam, The Netherlands

I have been involved in human genetics since 1989 starting as PhD student in Groningen and now heading, since 2012, the clinical genetics department in Rotterdam (the Netherlands). From doing bench work myself, managing research projects to running a department with all the tasked coupled to that (research, teaching and patients care).

National tasks: Over the years I also have been involved in many national committees for human genetics, among which was being president of the Dutch Society of Human Genetics (Nederlandss Vereniging voor Humane Genetica, NVHG) 2006-2012 www.nvhg-nav.nl. In this period we hosted the ESHG in Amsterdam (2011)

Why ESHG? Now I have settled in Rotterdam, I would like to take up a new challenge. In particular in the time frame were human genetics is becoming more and more integrated in patient care and clinical management. All this is extremely challenging however many important issues need to be addressed. I do believe that the ESHG should be a forerunner in all this and I would like to contribute to this; promote and support the scientific and educational ESHG values, and discuss and define public and professional ESHG policies.

André Reis
Erlangen-Nürnberg, Germany

André studied medicine in Göttingen and Lübeck and was awarded an M.D. degree in 1986. After his specialty training in Göttingen and Berlin he became associate professor for medical genetics at the Humboldt University in Berlin in 1998. Since 2000 he is full professor and director of the Institute of Human Genetics at the University of Erlangen-Nürnberg (FAU). André is a member of the German National Academy of Sciences and various other Academies. His research centres on elucidation of the molecular basis of both monogenic and complex traits and genotype/phenotype correlation with a focus on intellectual disability, glaucoma and psoriasis. His bibliography lists more than 280 original publications that received in excess of 12,000 citations.

He served as a member of the ESHG Scientific Programme Committee (SPC) from 2002-2006 and for two terms as president of the German Society of Human Genetics (GfH) from 2008-2012. During that time he was also local host of the 2012 ESHG Annual Meeting in Nuremberg. More recently he was elected vice-president of the section „Clinical Genetics" of the European Union of Medical Specialists (UEMS).

In the Board, André wishes to contribute his scientific and professional experience to help shape the Societies future. In the era of next generation sequencing, human genetics has become ever more important for all medical specialities. Therefore it is now essential to strengthen our discipline and to guide human genetics into its growing role in medicine without forgetting our strong foundation in research.

Zeynep Tümer
Copenhagen, Denmark

I am a medical doctor (MD, PhD, DMSc) working with medical and molecular genetics for 25 years. I graduated from the Egean University, Medical Faculty, İzmir, Turkey; and I completed my doctor of philosophy (PhD) studies and doctoral dissertation in medical sciences (DMSc) at the University of Copenhagen, Denmark. I am currently holding a professorship in applied human molecular genetics at the Kennedy Center, Department of Clinical Genetics, Copenhagen University Hospital, Rigshospitalet, Denmark. My main research interest is identifying genetic/epigenetic mechanisms involved in human neurodevelopmental disorders through investigation of cytogenetic abnormalities, copy number variations or sequence changes using chromosome microarray methodologies and next-generation sequencing (NGS) based technologies, with the aim of implementing in clinical diagnostics. Our group works with different neurodevelopmental disorders including Tourette syndrome and co-morbidities, Menkes disease, and intellectual disabilities.

„How my mom looks like"
Bulut, October 2002

We have a large network and collaboration with researchers especially in Europe through COST-networks, Marie Curie programs and several FP7/Horizon 2020 applications mainly on congenital imprinting disorders and Tourette syndrome. During the last 8 years our research received more that 16.000.000 Danish krones as support. Furthermore, I have a vast experience in pre- and post-graduate teaching at the University of Copenhagen and in research supervision (PhD, PostDoc, MSc, bachelor etc.).

My biggest and most successful genetic project has been my son who is 18 years old now.

I thank you all for supporting me in the board elections and I will do my best to contribute to the work of the Society.

February 29, 2016 is Rare Disease Day!

by Lara Chappell, Communications Director, EURORDIS

The rarest day of the year, 29 February is Rare Disease Day. This day, which brings rare diseases to the spotlight, is a EURORDIS initiative recognised all over the world as an opportunity to raise awareness about rare diseases and the cause. Anyone can participate, including researchers and academics by showing their solidarity for people living with a rare disease and their families.

Patient Voice: Join Us in Making the Voice of Rare Diseases Heard

2016 marks the ninth edition of Rare Disease Day, organised and

coordinated by EURORDIS, the European patient organisation for Rare Diseases. In addition to media attention, thousands of events will take place around the world on all continents and in hundreds of cities. This year's theme, 'Patient Voice' recognises the crucial role that patients play in voicing their needs, both with regards to new therapies and initiatives, and in instigating change that improves their own lives but also others and the lives of their families and carers.

This sentiment is strengthened by the Rare Disease Day slogan, 'Join us in making the voice of rare diseases heard'. People living with a rare disease are often isolated and the wider community can help break this isolation and bring rare diseases into the spotlight.

How Can Researchers and Academics Participate in Rare Disease Day?

Researchers and academics are particularly encouraged to participate in or organise Rare Disease Day events to educate and share knowledge on the diseases. All organised events are posted on rarediseaseday.org and range from conferences and symposiums, to presentations at the pub!

Some examples of events that research teams are holding in 2016:

PubHD, held in Leicester, UK, is inviting 3 researchers to give 5 minute presentations in the pub with nothing but a whiteboard. The challenge is to keep everyone interested and inform them about your rare disease research, all over a friendly pint!

Emory University Genetics Lab is running a social media campaign about Rare Diseases during the month of February posting information daily for patients to find online communities like those on RareConnect or patient organisations in their area. They will also publish a blog about the day.

Finally, we expect several lab teams, or individuals and families to send us pictures, raising and joining hands to show their solidarity! Send yours today!

Eurordis Events on the Occasion of Rare Disease Day

Multi-stakeholder Symposium

EURORDIS is holding a Rare Disease Day event on 'Improving Patient Access to Rare Disease Therapies' in Brussels on 24-25 February. This multi-stakeholder symposium is a unique insight for researchers, not only into the process of value determination, appraisal, pricing and reimbursement for rare disease therapies, but also into what patients are looking for in new therapies and the difficulties they currently experience in accessing them. It provides a space for all stakeholders to gain common understanding and agreement on the determinants of value, and of the assessment and appraisal methods for orphan medicinal products. Researchers will be challenged to evaluate and price orphan medicinal products from different stakeholders' perspectives, via simulation exercises. Registration is required to attend this event. The keynote speeches, including the Vytenis Andruikaitis, European Commissioner of Health and Food Safety, will be live-streamed on February 24. Please visit rarediseaseday.org for more information.

EURORDIS Awards

EURORDIS will also be live streaming the 5th edition of the EURORDIS Awards taking place in Brussels on 23 February from 17-18:30 (GMT+1). Some of the previous recipients of the Scientific Award include Professor Kate Bushby, Professor Hans-Hilger Ropers and Ségolène Aymé, MD, PhD. More information about the awards can be found here: www.eurordis.org/eurordis-awards

Opportunities for Researchers at the European Conference on Rare Diseases and Orphan Products

If it's too late for you to schedule a trip to Brussels, the European Conference on Rare Diseases & Orphan Products in Edinburgh next May is an excellent opportunity to present research and network with others in the rare disease field. It provides an exceptional opportunity in which the whole of the rare disease field gathers to monitor and benchmark initiatives in the field of rare diseases. In addition to over 80 speakers and the opportunity to present research, ECRD 2016 is holding a Research Speed Networking session, a form of professional 'matchmaking'. Prior to the event, interested delegates will be asked to fill in a survey, and using their answers, will be introduced to people working in a similar field , whether they are patients, researchers or industry. This event is aimed at the sharing of information and ideas in the hope of sparking new research initiatives or collaborations in the rare disease field. The ECRD is on 26-28 May and registration is required, to find out more please visit rare-diseases.eu.

If you are interested in any of these events, or would like more information about Rare Disease Day, please write to us at rarediseaseday@eurordis.org.

Lara Chappell,
Communications Director, EURORDIS

ESHG Course Programme 2016

The ESHG has expanded its course portfolio. More information on course programme, downloads etc. can be found at www.eshg.org/courses.0.html

The following courses are scheduled for 2016:

Manchester Dysmorphology Course
Manchester, UK, April 19–21, 2016
Course Organisers: Jill Clayton-Smith, Dian Donnai, Sofia Douzgou, Sid Banka

Spring Course in hereditary cancer genetics
Bertinoro, IT, April 10–13
Course Organisers: N. Hoogerbrugge & C. Oliveira

Preconception, Preimplantation & Prenatal Testing
Maastricht, NL, April 13–15
Course Organisers: E. Coonen

European Course on Cardiogenetics
Manchester, UK, April
Course Organisers: B. Newman & B. Loeys

Basic & Advanced Course in Genetic Counseling
Bertinoro, IT, April 28–May 3
Course Organiser: H. Skirton

Next Generation Sequencing
Bertinoro, May 4–7
Course Organiser: J. Veltman

Medical Genetics
Bertinoro, IT, May 8–12
Course Organisers: H. Brunner & B.Wirth

Basics in Human Genetic Diagnostics
Nicosia, CY, June 20–25
Course Organiser: T. Liehr

Introduction to the statistical analysis of GWAS
London, UK, July 4–8
Course Organisers: I. Prokopenko

Clinical Cytogenetics
Goldrain, IT, August 27–September 3
Course Organiser: A. Schinzel

Short-Report of the Executive Officer

by Jérôme del Picchia, Executive Officer of the ESHG

New Ground

The SPC, in consultation with the Professional Congress Organisers of the Vienna Medical Academy and Rose International, has implemented quite a number of changes for this year's **European Human Genetics Conference, May 21-24, 2016 in Barcelona, Spain**. So, what's new in 2016? Quite a few things actually.

Tutorial on Abstract Submission on January 19, 2016

On January 19, at 17.00 hrs CET, Professor Heather Skirton will do a short tutorial online, on writing an abstract, and preparing a poster or spoken presentation. This is aimed at those who may not have extensive experience in submitting an abstract to a scientific meeting. Please see the ESHG 2016 website for more information (www.eshg.org/eshg2016) The tutorial will be available as web cast after this date.

New registration hours

For the first time the registration desks open already on Friday, May 20, from 15.00 - 18.00 hrs. So in case you are already in Barcelona, pass by and avoid queuing on Saturday morning.

New Poster Times and Presentation Formats

The SPC has decided to double the number of poster discussions ('Poster Viewing with Authors').

Posters will now be discussed in **4 groups from 10.15 to 11.15 hrs and 16.45 to 17.45 hrs both on Sunday and Monday**.

Posters will remain on display for the entire meeting (Saturday - Monday). We hope that this will facilitate interaction at the posters and increase convenience for the presenters.

The ESHG meetings now have the following **3 types of acceptance for submissions**:

- **Spoken presentation** in a concurrent or plenary session
- **Poster presentation** in one of the poster sessions
- **Electronic Poster presentation** in the electronic poster session

The decision on the final allocation will be based on the peer reviewing process. We hope that the new presentation format of 'Electronic Poster' (replacing 'Accepted for Publication') will facilitate participation for authors.

New rules for publication

Attention: note that from this year, the presentation at the meeting will be the condition for the publication of the abstract in the electronic supplement of the European Journal of Human Genetics.

Young Investigators in Focus

Two new workshops ('W02 . Career development and funding opportunities for young investigators' on Saturday and 'W09. How to get published' on Sunday) aim directly at young investigators attending the conference.

You might also be interested to know, that the Scientific Programme Committee decided to have at least **30% of its members aged under 40 years**.

Another novelty: Post-doc Young Investigator Award Winners of the last meeting will be invited to **co-chair a session** at the next conference.

Also a number of **new travel fellowships** have been created. There are now three categories, all based on abstract acceptance and the reviewing process:

- Conference Fellowships for a restricted number of European Countries
- Conference Fellowships for non-European Countries
- Conference Fellowships of Excellence (no country restrictions)

A fourth category are the "National Fellowships", at the discretion of the National Human Genetics Societies.

More web casts and streaming

In addition to the Plenary Sessions on Tuesday and the Building Bridges Session (joint with the ASHG), the ESHG will webcast all **Educational Sessions** and have them available for streaming after the conference.

More interaction with the audience - #eshg16

You may have noted an increase in the number of sessions using interactive means (e.g. online voting). We are again encouraging this feature in 2016 and will also open the discussion in selected sessions to questions asked **via twitter**.

We really hope that these changes and innovations will contribute to the fantastic programme and the illustrious speakers, who accepted to speak at the meeting and most definitely look forward to welcoming you to the meeting in Barcelona in May.

There is still a little time to submit your abstracts, by the way. **Deadline is February 12, 2016!**

The website www.eshg.org/eshg2016 is constantly updated with all details.

See you in Barcelona.

Jerome del Picchia
ESHG Executive Officer

PS: Remember to renew your ESHG membership (online) at www.eshg.org/join0.0.html

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