President Address
by Helena Kääriäinen, President of the ESHG

Dear ESHG members, dear colleagues,

ESHG membership consists of human geneticists with a wide range of backgrounds and interests. The biggest groups among the membership are clinical geneticists and molecular researchers, followed by laboratory geneticists, cytogeneticists, genetic counselors and many more. While many of us are devoted to science, most of us share the practical goal of improving the health and life quality of individuals with different genetic diseases. We also represent different countries with their different cultures. The youngest ones among us are newcomers in the field while the oldest still remain interested in genetics in spite of having retired already years ago.

All this contributes to the fact that the membership of ESHG forms a strong, multifaceted and innovative society, the richness of which is best reflected in our excellent scientific conferences. But in addition there is less visible work done in the many Committees to promote the values of the Society as well as the role of genetics in health care and in the society at large.

What is new in ESHG this year? The enthusiastic work to clarify and promote the roles of genetics professionals has led to the birth of an independent body, European Board of Medical Genetics (EBMG) chaired by Heather Skirton. On the other hand, a former “independent” player in European genetics namely Eurogentest has found a way to join with ESHG enforcing the Society’s capacity in dealing with quality issues in genetic services. ESHG is formally represented in the EU Commission Expert Group of Rare Diseases as our Vice President Han Brunner has been elected to represent ESHG in that group. We have been and continue to be active in influencing the developments in EU Regulations relating to human genetics, especially the discussions around IVD Directive. Recently we have started a process aiming at closer collaboration with rare disease patient organizations via EURORDIS. These examples illustrate how dynamic times ESHG is living.

At the same time, human genetics is moving ahead with a speed faster than ever before. We are witnessing new applications of genetics coming to patients care, disease prevention and life in general. We can be proud and enthusiastic when being part of these developments but we also have to be alert to recognize and react if we foresee unwanted developments. I believe that ESHG can promote the European values like equality, tolerance and human rights in the genetics revolution.

ESHG is the sum of its members. As the President, I invite you all to share your ideas with the Board Members or directly the Executive Board including the three Presidents: President-Elect Feliciano Ramos from Zaragoza in Spain, Vice-President Han Brunner from Nijmegen in the Netherlands and myself from Helsinki in (the genetically very interesting) Finland. Feel free to contact any of us!

Waiting for to meet with all of you in the next European Human Genetics Conference in Glasgow in June 2015!

Helena Kääriäinen
President of the ESHG

Updates on “EBMG Professional Branch Board for Clinical Laboratory Geneticists” – Open for applications

By Thomas Liehr, Chair Clinical Laboratory Geneticists Professional Branch

The European Board of Medical Genetics was established by the ESHG in 2012 and is now a legal entity in its own right. It is legally independent of the ESHG but connected through ESHG nomination of some members of the EBMG Board.

The EBMG Professional Branch Board for Clinical Laboratory Geneticists pursues the target that the professional titled “Clinical Laboratory geneticist (CLG)” working with laboratory diagnosis will be a recognized profession in all European...
countries. It is hoped that eventually we will have a specialist profession for CLGs in Europe, under European governance. However, the EU Professional Qualifications Directive (PQD) is not yet in force, and in 2012 the ESHG decided to install the EBMG, which has the goal to issue the European title ‘Clinical Laboratory Geneticist’ (CLG) from the EBMG (prior to any EU directive). As well as giving CLGs the opportunity to have their competence formally assessed and recognized at European level, we believe that having a voluntary system in place will support our call for the profession to be formalized by the EU at a later time. As soon as the PQD is in force, the EBMG will work to request the EU to accept the CLG-title as the EU-approved one.

In 2011 and 2012 the CLG committee collected data from all European countries concerning the national CLG-titles and the 41 countries listed on https://www.eshg.org/407.0.html were categorized into three groups now

+ group 1: countries which have a national CLG title/ education scheme which fulfills at least 36/38 of the knockout criteria.

+ group 2: countries which have a national CLG title/ education scheme which fulfill less than 36/38 of the knockout criteria.

+ group 3: countries which have no national CLG title.

Now we are proud and happy to announce that we opened the possibility to apply for the CLG title for group 1 countries mid of August on our web page. https://www.eshg.org/407.0.html, i.e. for following specialists from:

- Austria: Fachhumangenetiker/Fachhumangenetikerin (ÖGH)
- Belgium: Medical genetic laboratory supervisor
- Czech Republic: Clinical Bioanalytician in Clinical Genetics
- Finland: Clinical Molecular Geneticist
- Germany: Fachhumangenetiker/Fachhumangenetikerin (GfH)
- Hungary: Molecular Genetic Diagnostics (MD)/Molecular Biology Diagnostics (PhD)
- Italy: Genetica Medica
- Lithuania: Medical geneticist (laboratory)
- Macedonia: Clinical Laboratory Genetics
- Netherlands: Clinical Laboratory Geneticist
- Poland: Laboratory Medical Genetics
- Portugal: Técnico Superior de Saúde, ramo de Genética
- Slovenia: Medical Genetic Laboratory Programme
- Sweden: Sjukhusgenetiker/”Hospital geneticist”
- Switzerland: Specialist for genetic laboratory medicine FAMH.

We stress that we will be introducing systems for registration of CLGs working in Group 2 or 3 countries within the next year.

We encourage **CLGs in Group 1 countries** to apply to obtain European recognition. If you think you could be eligible for a CLG title please visit: https://www.eshg.org/407.0.html and for more information https://www.eshg.org/587.0.html. Applications (using the forms on the website), should be sent by email to me (Thomas.Liehr@med.uni-jena.de) or Prof. Isabel Carreira (carreiraim@gmail.com).

Thomas Liehr, Jena University Hospital, Friedrich Schiller University, Institute of Human Genetics, Kollegiengasse 10, D-07743 Jena, Germany

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**The new ESHG Board Members**

**Angus CLARKE, Cardiff, United Kingdom**

Angus studied genetics at Cambridge and then clinical medicine at Oxford in the 1970s. After full registration in 1980, he worked in general medicine and then paediatrics for several years. He obtained a research position with Professor Peter Harper in Cardiff in 1985-6, studying clinical and genetic aspects of X-linked hypohidrotic ectodermal dysplasia. He then worked in Newcastle upon Tyne in both paediatric neurology and clinical genetics, where he developed interests in Duchenne muscular dystrophy and Rett syndrome. He then returned to Cardiff in 1989 and has worked there since then, with clinics in general genetics, paediatric genetics, muscle disease, Rett syndrome and Huntington’s Disease.

He established a BSc course in genetics for medical students in Cardiff in 1990 and, in 2000, the second MSc course in Genetic Counselling in the UK. He has a strong interest in the social and ethical aspects of advances in human genetics and of genetic services, including newborn and other genetic screening programmes. He contributes to policy debates and discussions, for example through his role on the (now defunct) Human Genetics Commission. He supports patient organisations in his areas of interest, including Rett UK and the UK Ectodermal Dysplasia Society, and he has published (edited and/or authored) seven books.

Angus wishes to play a full part in the life of ESHG and the activity of the Board, especially in relation to the professional challenges now facing medical genetics services in Europe: (i) the introduction of next generation sequencing into diagnostic practice, (ii) supporting clinical geneticists and genetic counsellors to work together to manage the counselling and family issues that arise in our work, especially as genetics comes to play a broader role across the whole of medicine, and (iii) developing effective pathways of care for patients with rare
diseases across Europe, despite the great differences in medical practice and access to health care across the continent.

Francesca FORZANO, Genova, Italy

I am a Consultant in Clinical Genetics in Genova, Italy, and I trained in Genova and in London, UK. My main interests are congenital malformations, dysmorphology, skeletal dysplasias and prenatal diagnosis, as well as the ethical, legal and social issues intrinsically connected to this exciting discipline.

As a board member I would have the chance to work in a stimulating environment shaped by excellent colleagues with different professional and cultural background, and I’d hope to bring my experience in clinical care, through which I got insight into needs of patients and health professionals, my enthusiasm for the amazing and fast developments of our wonderful discipline and a ponderous approach in respect to the translation of these developments into the medical practice.

I am already an active member of the ESHG, being a member of the PPPC since 2008.

I am co-Director with prof Heather Skirton of the EGF Course on Genetic Counselling since 2009.

I am also an active member of the Italian National Society of Human Genetics (SIGU); currently a member of the Committee for the Quality Certification of Genetic Services, I chaired the Committee for the Quality Certification of the Clinical Genetics Units in 2012-2013.

Dijana PLASESKA-KARANFILSKA, Skopje, Macedonia

Employed at the Research Centre for Genetic Engineering and Biotechnology “Georgi D Efremov”, within the Macedonian Academy of Sciences and Arts, holding the position of Full Professor and Head of laboratories. Participating in lecturing molecular subjects for both undergraduate and graduate studies at the University “Ss. Cyril and Methodius”, Skopje, Macedonia. MD degree received from the Medical Faculty in Skopje and PhD from the Medical Faculty in Maastricht, the Netherlands, following defending thesis in the field of hemoglobinopathies. Continuously studying different inherited diseases and contributing to the translation of a number of molecular genetic tests to clinical practice in Macedonia. Coordinator of an EU FP7 REGPOT project, which has strengthened the national research capacities in the fields of genomics and proteomics. One of the founders of the Macedonian Society of Human Genetics, as well as of the initiators for the specialization in Clinical Laboratory Genetics in Macedonia. Permanent active participant in the Balkan Human Genetics Meetings and Editor of the Balkan Journal of Medical Genetics. Regular attendee of the ESHG conferences and representative of Macedonia at the Meetings of National Societies. It is to be expected that being a member of ESHG board will represent an instigation and contribution to the endorsement and implementation of European standards in the field of human genetics in the Balkan countries.

Isabella CECCHERINI, Genova, Italy

After receiving a Biological Science degree and a PhD in Genetics at the University of Pisa, and following a one year post-doc in the Medical Research Council in Brighton, I moved to Genova in 1988, where I became interested in the study of inherited rare diseases. Since then, I have been working in the Medical Genetics Lab of the Gaslini Institute, managing research projects, mainly focused on neurocristopathies and, more recently, on autoinflammatory disorders, often within international collaborations and Consortia. I have also been performing molecular tests for the molecular diagnosis of several inherited diseases. Finally, university teaching and mentoring PhD students has always represented a continuous incentive to improve. I am very pleased to have had the chance of participating in the ESHG Board, and I hope I will be able to promote and support the scientific and educational ESHG values, also interpreting requests and needs from Southern Europe.

Dr Inga PROKOPENKO, London, United Kingdom

I am a senior Lecturer in Human Genomics at Imperial College London, UK. I received my higher education in Ukraine, trained at the University of Pavia in Italy, worked in R&D at GlaxoSmithKline in Italy and have done research at the Wellcome Trust Centre for Human Genetics, University of Oxford, UK. The main area of my research lies in
human genomics and statistical genetics. In recent years, my focus has been on the genomics of type 2 diabetes, variability of glycaemic traits in non-diabetic individuals, early growth and multi-phenotype effects. Having worked in large international collaborations, I enjoy interaction with experts in various scientific fields, as well as training students of different levels. In coming years, I foresee huge potential in extension of the interaction of the ESHG network of researchers to Eastern European countries. I hope to be able to contribute to furthering the aims of the ESHG through expanding the statistical expertise of current junior scientists.

Spain finally has the specialty of Clinical Genetics officially recognized

by Juan C. Cigudosa, President of the Spanish Society of Human Genetics (AEGH) and Feliciano Ramos, President-elect of the ESHG

On August 6, 2014, the Spain’s Official Gazette published the Royal Decree announcing the creation of the new specialty “Genética Clínica” (Clinical Genetics) in the National Health Care System. This date will remain as a milestone in the history of counsellors of Human Genetics in Spain and in parallel in the history of the Spanish Society of Human Genetics (AEGH) because, after more than three decades of struggle and after many setbacks (mostly political) this goal had been achieved.

During all these years the various Boards of Directors, joined by the appointed “Genetics Specialty Committee” during the last decade, have worked tirelessly and selflessly to achieve the recognition of Genetics as an independent specialty in our Health Care System. We can not name here every one of the individuals that were part of those “negotiating” teams, but we want to highlight here their tenacity. We also recognize the unconditional support of many of the members of our Society; we reached our goal. Thank you all, from the first to the last!

One of the greatest difficulties we encountered in this long journey was to listen to the legitimate aspirations of the different professional groups represented in the AEGH, biologists, physicians, pharmacists, chemists and others. Today, those difficulties are overcome thanks to the dialogue and consensus of all partners involved, each of which accepted to foster the common interest over their legitimate particular and/or professional interests. In the end all got aligned in the same direction and we passed the “finish line” together. This is a great responsibility for the future, and we must continue working together.

Until August 6th, Spain was one of the few EU countries (together with e.g. Belgium, Greece, Ireland) where the specialty of Clinical/Medical Genetics has not been officially recognized. This was despite the fact that we had a large number of professionals working in the field in the majority of main hospitals throughout the country. Many of them were either “self-made experts” (of course in positive sense), while some of them obtained their specialist qualification in other European countries with recognized training programs in clinical/medical genetics. The clinical and scientific activity of Spanish geneticists, despite the lack of official recognition as a medical specialty, has been broadly recognized by our international colleagues and institutions all over the world. Currently, more than 1,000 professionals, grouped by the AEGH, provide clinical genetic services, work in diagnostic laboratories, and are involved in research. Majority of our colleagues publish and participate in European scientific congresses. Now we have joined the “big family” and we ought to set up a training program in accordance with the best practice in Europe, and beyond.

At the EU level, the ESHG is also working through the UEMS, eu and EBMG on the modernisation and recognition of the specialty within the European Community, including the recognition of the three professional areas that configure our specialty: clinic, laboratory, including genetic nurses and counsellors. This is the multidisciplinary future of our field of interest! Spain wants to be part of it, not as a spectator but rather as a qualified contributor.

We want to publicly and sincerely thank the ESHG for the support we have received from the beginning of this endeavour. There are too many names to mention among all European colleagues who supported us directly or indirectly, but we wish to mention the several past Presidents and Executive Board members and the Chairmen and members of ESHG Committees. A special mention goes to Prof. Milan Macek, under whose ESHG Presidency, Genetics was included in the European Professional Qualifications Directive (www.eshg.org/111.0.html). This fact marked a definitive argument that finally convinced the Spanish health care authorities to go ahead with the recognition of our specialty in Spain, albeit almost three years later.

Finally, we are aware that this has been only the first step in a long road that lies ahead of us. We expect to successfully
complete it in two or three years from now when the first residents of Clinical Genetics begin their training as specialists in the different areas, at the accredited hospitals of our country. We are confident that through hard work and with the help of all of you, we will succeed!

Juan C. Cigudosa
President of the Spanish Society of Human Genetics (AEGH)

Feliciano J. Ramos
President-elect of the European Society of Human Genetics (ESHG)
Former President of the AEGH

A personal invitation to ESHG 2015 in Glasgow
By David FitzPatrick, Edinburgh, Member of the Scientific Programme Committee

When I heard that the ESHG meeting was going to be in Glasgow in 2015 I was very excited: the ESHG has always been my favourite Human Genetics meeting and Glasgow is one of my favourite cities. For those of you have never been to Glasgow you are in for a treat. Glasgow is a fun city and probably the most friendly place you will ever visit; you will find it difficult to walk down any street in Glasgow without a stranger talking to you. When this happens and you may find the famous Glasgow accent rather difficult to understand - but, fear not, everyone becomes much easier to understand after you have had a few drinks (a “bevvie”) in one of the MANY local pubs or bars. There is also a cultured side to Glasgow with the excellent Kelvingrove Art Gallery and the amazing Burrell collection. From Glasgow is also easy to access the beautiful lochs, mountains, islands and coastline of Scotland. And, although most Glaswegians refused even to recognise it’s existence, you can also get to Edinburgh on the train in less than one hour. So everyone in UK Human Genetics is looking forward to welcoming you to Glasgow in 2015 for what is going to be another tremendous ESHG meeting. It does very occasionally rain in Glasgow so bring a waterproof coat.

David FitzPatrick
MRC Human Genetics Unit, University of Edinburgh

From Milan to Glasgow 2015
By Jérôme del Picchia, Executive Officer of the ESHG

I am glad to say, that the last ESHG annual meeting in Milan has been a very successful one. 3,000 participants from 86 countries represent the second best attendance in history. Over 130 exhibiting companies and institutions made the exhibition hall the vibrant heart of the meeting.

Scientific Method

The scientific sessions offered several premieres in 2014. On one hand, the ESHG-ASHG “Building Bridges Session, towards Finding global agreement on topical discussions in genetics” was the first out of a hopefully long and successful series of joint ESHG-ASHG sessions. These will be continued at the ASHG Annual meeting in San Diego and the next ESHG meeting in Glasgow.

It was also the first time in recent history, that this joint Plenary session was held as interactive debate. The audi-
ence was able to vote on questions discussed by the panel. This voting system was also a novelty at our meetings, the audience could make use of a tool, which is accessible from all WIFI capable mobile devices, no matter if cell phone, tablet or laptop. Several workshops equally used this system to get feedback of the attendees.

The third major addition to our meeting was clearly the Live-webcast of the Tuesday plenaries (interactive debate, Mendel and ESHG Award lectures). All sessions are still available for online viewing on the website www.eshg.org/eshg2014.

**Lessons**

Unfortunately not everything worked as flawlessly as the improvements above. Two major issues kept us organisers quite unhappy. The quality of the lunch boxes on the first two days, which were unfortunately the result of a translation-based misunderstanding at the caterer. Unfortunately the Sunday and the holiday on Monday did not help in sorting this out quickly. This should not reproduce in Glasgow translation-wise. Also it has been decided to offer several alternatives for lunch at the meeting, allowing you to make a choice. The options will be announced on the website.

The second issue was obviously the WIFI. Unfortunately the high standard hardware on the surface was not continued “below the surface”. The high number of devices searching to connect to the system uncovered the “bottleneck” in the configuration beyond the visible access points. The regrettable result was not only the frustrating attempts to connect but also the slow (if at all) updating of the conference app. A pity, as it is designed to work offline, still it has to be downloaded at least once, an impossible task using the Milan WIFI. Like many other apps, when downloading the “ESHG conference app”, only the “operating shell” is loaded from the App Store. When opened the first time, the app checks for content updates and will download the actual sessions, abstracts, plans etc.

The lessons we take from this problem are numerous. The 2015 app will be launched earlier, allowing most attendees to download everything “at home”. Our technical manager is being included as early as possible in the negotiations with the future conference centers, so that we are able to check all hard and software configurations well in advance.

Last but not least, we have learned to set a different “in deep” standard with the venues, as WIFI today is so much more than a “nice to have” item.

On a side note, when witnessing the situation at our meeting, the management of the conference center in Milan has immediately decided to rebuild the entire WIFI infrastructure in order to be able to cover meetings over a certain size. We hope to be able to see proof of this investment, should we return to Milan in a few years.
You are cordially invited...

As you may have seen, we have recently opened the 2015 website at www.eshg.org/eshg2015. Online registration is available and abstract submission is possible until February 13, 2015. Check the impressive list of speakers (over 95% are confirmed as per September 2014), including the Mendel Lecturer Thomas Südhof (Nobel Laureate 2013) and ESHG Award Lecturer Svante Pääbo.

The new schedule of Milan has proven to be a successful format, so the opening day (Saturday) will start again at 10.30 hrs with educational sessions and workshops and concludes with a series of concurrent sessions from submitted papers. Also the exhibition is open already on Saturday morning, and closes on Monday evening.

We will also continue the webcast of the Tuesday plenaries, including the interactive debate: “Should all geneticists have their genome sequenced?”, as well as the “ESHG-ASHG Building Bridges” Session: “Genetic testing in Children” and the joint session with the European Society of Cardiology (ESC) “From rare to common variants in cardiovascular diseases”.

In terms of logistics and accessibility, please remember that you may fly in not only via Glasgow airport, but also via Edinburgh, followed by a short train ride to Glasgow. More information will be available on the website.

On a very personal note, I must say that I was genuinely amazed by how my expectations of the city were exceeded and how enchanting Glasgow and its people are. I have learned that Glasgow offers a blend of internationally-acclaimed museums and galleries, stunning architecture, vibrant nightlife, fantastic shopping and a diverse array of restaurants and bars. SPC member Mark Longmuir tells me that Glasgow is unsurprisingly one of the “Lonely Planets top ten cities to visit”.

So I can only seriously recommend marking your calendars with the date of ESHG 2015: June 6-9, 2015, where the ESHG, in conjunction with the BSGM will hold its 48th meeting. See you there!

Jerome del Pichia
Executive Officer of the ESHG

Courses in collaboration with the European School of Genetic Medicine (ESGM)

The European Society of Human Genetics has developed a partnership with the European School of Genetic Medicine, in order to promote advanced training in human-genetic sciences and preventive medicine in Europe. The European School of Genetic Medicine organizes courses in the charming venues of Bertinoro (Italy). These courses began in 1988 with the first course in Medical Genetics of the European School of Genetic Medicine conducted by Prof. Victor A. McKusick. For detailed information about the School visit www.eurogene.org

ESGM-ESHG Courses - 2015 - First Announcements:

- 4th Course on Next Generation Sequencing
  May 13-16, 2015
- 28th Course on Medical Genetics
  May 17-21, 2015

Selected Meetings from the ESHG Conference Calendar

See www.eshg.org/101.0.html for the full list.

2014
Epigenomics of Common Diseases
Cambridge, UK, October 28-31, 2014
https://registration.hinxton.wellcome.ac.uk/display_info.asp?id=441
**EMBO Conference: From Functional Genomics to Systems Biology**  
Heidelberg, Germany, November 8 - 11, 2014  
www.embl.de/training/events/2014/OMX14-01

**Chromatin Structure and Function**  
Hinxton, UK, November 10 - 14, 2014  
www.wellcome.ac.uk/Education-resources/Courses-and-conferences/Advanced-Courses-and-Scientific-Conferences/Advanced-Courses/wtp055912.htm

**NGS 2014 Sheffield: From Research to the Clinic**  
Sheffield, UK, November 11 - 12, 2014  
https://biotexcel.com/event/ngs-2014-sheffield

**The 4th Global Cancer Genomics Consortium Symposium**  
Kyoto, Japan, November 14 - 15, 2014  
www.octr-institute.org/gcgc/2014/

**NGS 2014 Nordic**  
Odense, Denmark, November 20, 2014  
www.biotexcel.com/event/ngs-2014-nordic

**2015 Genomics and Clinical Microbiology**  
Cambridge, United Kingdom, January 18 - 23, 2015  
www.wellcome.ac.uk/Education-resources/Courses-and-conferences/Advanced-Courses-and-Scientific-Conferences/Advanced-Courses/WTVM051628.htm

**Human Genome Meeting 2015**  
Kuala Lumpur, Malaysia, March 14 - 15, 2015  
www.hugo-hgm.org

**Workshop: Epigenetics as the meeting point between nature and nurture**  
Uppsala, Sweden, March 19 - 20, 2015  
www.crb.uu.se/epigenetics

**Proteomic Forum 2015**  
Berlin, Germany, March 22 - 25, 2015  
www.proteomic-forum.de

**Development of the Enteric Nervous System: Cells, Signals, Genes and Therapy**  
Rotterdam, the Netherlands, April 19 - 22, 2015  
www.ens-development-meeting.com

**Advances in Next Gen Sequencing - ANGS 2015**  
Online Event, May 5 - 6, 2015  
http://goo.gl/ePFHic

**EMBO Conference Series: Chromatin and Epigenetics**  
Heidelberg, Germany, May 6 - 10, 2015  
www.embl.de/training/events/2015/CHR15-01

**Personalized Medicine**  
Cambridge, UK, May 12 - 13, 2015  
www.regonline.co.uk/banking2015

**Biobanking 2015**  
London, UK, May 19, 2015  
www.regonline.co.uk/banking2015

**Advances in qPCR & dPCR**  
Singapore, May 21 - 22, 2015  

**The European Human Genetics Conference 2015**  
Glasgow, Scotland, UK, June 6 - 9, 2015  
www.eshg.org/eshg2015

**Stem cells: from basic research to bioprocessing**  
London, UK, June 9, 2015  
www.regonline.co.uk/stem2015

**EMBL Conference: Protein Synthesis and Translational Control**  
Heidelberg, Germany, September 9 - 13, 2015  
www.embl.de/training/events/2015/TRC15-01

**EMBL Conference: Cancer Genomics**  
Heidelberg, Germany, November 1 - 4, 2015  
www.embl.de/training/events/2015/CAN15-01

**12th World Congress of Perinatal Medicine**  
Madrid, Spain, November 3 - 6, 2015  
www.wcpm2015.com

**International Congress of Human Genetics 2016**  
Kyoto, Japan, April 3 - 7, 2016  
www.ichg2016.org