

Presidential Address

by *Helena Kääriäinen, President of the ESHG*

Another interesting year!

In ESHG we do not talk about year 2014 or 2015 but “the year between Milan and Glasgow”. This is because the most important yearly achievement of ESHG is its scientific conference which seems to be even more exiting year after year.



So, what has happened in ESHG between Milan and Glasgow? ESHG tries to follow political discussion related to genetics in Europe and react when needed. This year the confusing wordings of the EU draft IVD regulation have continued to create some worry. The Society feels that even though genetic counselling in relation to genetic testing is, depending on the testing situation, an important part of the process, it should not be regulated on EU level. Different EU (and European) countries have different health care structures where the task of pretest genetic counselling may be performed by different professionals, including medical geneticists, genetic counsellors, genetic nurses or even other professionals trained for certain testing situations. ESHG does not feel that this part of health care should be regulated on EU level as, in general, organising health care is a national issue.

Another EU draft with problematic wordings is the data protection regulation. ESHG feels that research is such an important tool in promoting our aim to improve the health of European people, that the regulations should be really carefully thought to allow sufficient flexibility. Of course, the right to privacy and autonomy are important values but so is solidarity and wide participation in medical research. ESHG wants to be involved in this discussion as, for instance, research using registries and biobank samples requires wider and longer lasting consents than clinical trials or some other types of research.

These are future EU regulations but often the EU rules are reflected in legislation all over Europe and even globally, so all of us should follow these discussions, get involved with patients and citizens including politicians in our own countries and try to understand each other’s views and explain those of ours.

Yet another important development between Milan and Glasgow was signing the Memorandum of Understanding between Eurordis and ESHG. Eurordis is the umbrella organisation of European rare diseases national organisations. It has grown to be a very strong and respected player in the field of rare diseases which continue to be one of the main interests of our Society. Please visit the Eurordis Booth in the Exhibition in Glasgow to learn more about their interesting work.

The role of ESHG in trying to create workable regulations and practices and form collaborations in our field seems to highly interest the membership. There have been more candidates for ESHG Board Membership than ever and also some Committees, especially PPPC, have so many enquires for committee membership that all cannot be accepted. Hopefully you ESHG members present at Glasgow will join the Membership Meeting and tell your views and wishes so that ESHG can focus, between Glasgow and Barcelona, on the things that you prioritise.

Helena Kääriäinen
President of the ESHG

Secretary General’s report

by *Gunnar Houge, Secretary General of the ESHG*

After eight years in the ESHG executive board (the “Exec”), the last two terms as general secretary, it is time for a change if the Board agrees. The Exec is, as the name implies, running our day-to-day matters, but it is not the general secretary that really keeps wheels in motion – it is our executive officer, Jerome del Picchia. The Exec can be regarded as his council. All significant ESHG matters are discussed among Exec members before decisions are made. If matters are considered to be of major political or economical importance, the Board is also consulted. Otherwise, the full Board is mainly the watchdog of the Exec, and they should keep a keen eye on what the Exec is doing and deciding. In addition, they are encouraged to make suggestions for future activities, and



to give feedback on policy documents and guidelines. If the Exec steers the Society in unacceptable or irresponsible directions, the Board must take action.

In practice, the Exec has two parts; a “political part” (the deputy president, the current president and the vice president) and a not so dynamic “administrative part” (the executive officer, the deputy gen sec, the gen sec, and the treasurer). Administrative officer terms are for 3 years and renewable. I think it is an advantage to have a mixture of “new and old” (or “long-term memory and new ideas”) in the Exec, as long as everybody is open-minded and attentive to the opinion of others. During my eight years I cannot remember a single occasion when the Exec did not achieve consensus on important matters. We are especially grateful to Andrew Read, who has been a wise and prudent (but not too prudent) treasurer for years, and the head of the SPC before that. He is also the head of the Annual Meetings Committee (AMC), which picks future venues for annual meetings in collaboration with Jantie de Roos, our long-standing conference sales manager.

ESHG activities have changed significantly during the last eight years. By far, the annual meetings are still our main activity, and the SPC is a committee that cannot fail without severe consequences. My thanks to Brunhilde Wirth that once again has held everybody “in the ear” and made SPC work so efficient and enjoyable. In addition, the European Board of Medical Genetics (EBMG) has been established as a legally independent certification body, but still with tight links to ESHG. We are most pleased that they are doing so well under the leadership of Heather Skirton, and now they appear to become economically independent as well. A major new development is all the applications (> 250) for the first round of CLG certification that the clinical laboratory geneticist committee has received, chaired by Thomas Liehr. It is also likely that the EU Professional Qualifications Directive will be operative from next year, which may give many of these EBMG certificates better legal standing. Somewhat more in the moulding face is the Eurogentest committee, which is a continuation of Eurogentest after EU support ended. They will mainly deal with service quality issues through Quality, Training, Professional Guidelines and Dissemination subcommittees. A major new development is the restructuring of the Education Committee (EC) where our off-going Vice President, Han Brunner, has agreed to be the chair. In addition to traditional tasks like arranging the DNA day (this year again lead by Christophe Cordier), the new EC will update and maintain a course portfolio that should cover most major areas of human genetics. This portfolio should reflect the needs of EBMG and also make it easier for national health care systems to educate specialists in clinical/medical genetics, genetic counselling and clinical laboratory genetics. Everybody does not have to invent the wheel, and for small nations (like Norway) this is especially important. It is the

wish of ESHG that our courses will become a compulsory part of many national education schemes.

Finally, I want to thank the whole SPC for once again putting together such an excellent program – and I hope to see you all at the party!

Gunnar Houge
Secretary General of the ESHG

A personal welcome by the programme chair

By Brunhilde Wirth, Chair of the SPC

On behalf of the ESHG Program Committee I welcome you to the ESHG conference in Glasgow. Based on the 2217 submitted abstracts, we expect again a large number of attendees at the meeting. A highly attractive program will hopefully enable you to particularly enjoy this meeting, to foster your scientific work and to find sufficient opportunities to discuss science and develop new ideas. I hope you will meet many old collaborators and friends and find new ones. Enjoy the meeting overall and the city of Glasgow!

Activities of the Scientific Programme Committee

The Scientific Programme Committee (SPC) for 2014-2015 was composed of twenty regular SPC members: Brunhilde Wirth (chair, D), Paul de Bakker (NL), Yanick Crow (F), Helen Dollfus (F), Erik Iwarsson (SE), David FitzPatrick (GB), Daniel Grinberg (ES), Genuardi Maurizio (I), Jose (Machado) (PT), Giovanni Neri (I), William Newman (UK), Minna Nyström (FL), Francesc Palau (E), Anita Rauch (CH), Samueli Ripatti (F), Peter Robinson (D), Joris Veltman (NL), Joris Vermeesch (BE), Xavier Jeunmaitre (F), Kristel Van Steen (BE), four local SPC members from UK: Dominic McMullen, Emma Woodward, Pia Ostergaard, Mark Longmuir and three observers of the ESHG board: Martina Cornel (NL), Gunnar Houge (N) and Karin Writzl (SI). In addition Tara Clancy (UK) from EMPAG joint this year SPC.

The SPC met twice to organize the ESHG conference in Glasgow 2015: in July 2014 in Amsterdam to decide on the plenary sessions and symposia and in Vienna at the VMA offices in March 2015 to select the abstracts for oral presentations and posters.

Based on the restructured topics the assignment of the abstracts, the evaluation and decision for concurrent sessions appeared more straightforward. All 2217 abstracts have been on-line scored by 3-11 evaluators including SPC members and a total of 74 external reviewers and ESHG board members, who have been proposed by the SPC members as experts for the various topics. I would like to thank all reviewers for their fantastic work and commit-

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ment. The ESHG will particularly acknowledge the contribution of all external reviewers by giving them a discount of 30% on the registration fee.

Based on topics and scores, 144 (17.1%) abstracts submitted with the preference for an oral presentation were selected for the 24 concurrent sessions including one plenary highlight session. The meeting has been extended to Saturday morning and a block of 6 additional concurrent sessions have been added. Among the oral presenters, 68 were Young Investigator Candidates (at least 1 in almost every session), reflecting the high level of contribution of young scientists to this program. From the 1638 poster presenters, 38 Young Investigator Candidates were selected for the best poster award and will be marked with an ESHG Rosette. 359 abstracts with a score <1,75 will be “published only” and 74 abstracts were rejections either due to very low quality (score < 1.0) or multiple submissions of the same first author.

After the Glasgow conference, the SPC shall have to say goodbye to Paul de Bakker, David Fitzpatrick, Giovanni Neri and Anita Rauch. We thank them for their excellent work and their dedication to making the meeting better.

2015 Meeting Highlights

After the opening ceremony, the meeting will start with the first plenary session including three local stars: Wendy Bickmore, Matthew Hurles and Andrew Jackson. The “What’s new?” session will follow with the most exciting new findings selected from submitted abstracts.

The meeting will include 22 concurrent symposia, which will address topics of new insights and challenges from next generation sequencing in gene discovery, new mechanism underlying human disease, functional studies and underlying pathomechanism of various human disease groups, cancer genetics, clinical and social implications of genomics and the implementation of the new technologies in genetic testing, modelling in statistical genetics and analysis of complex datasets, challenges in clinical genetics and genetic counselling. Our initiative of building bridges between the ESHG and the ASHG will continue with a common symposium on Sunday on “Genetic testing in Children”.

Four educational sessions will start already before the opening session on Saturday morning and more educational sessions will continue throughout the meeting. These include: CRISPR-Cas9, From Genes to Networks, Translational Cancer Genetics, Care for Rare Diseases, Automating



Clinical Genetics, My vision on Genomic Medicine, Imprinting-related Disorders, Palliative Care of Genetic Conditions and Mutation Prediction Tools.

On Tuesday, we will have an interactive debate on „Should all geneticists have their genome sequenced?“ chaired by Martina Cornel and Joris Veltman with Wendy Bickmore, (UK), Carsten Bergmann (D), Kate Bushby (UK), Heidi Howard, (SE) and Gijs Santen (NL).

The meeting will conclude with our distinguished speaker of the Mendel lecture Thomas Südhof (USA), who won the Nobel Prize in Physiology or Medicine in 2013 and who will talk about “The neurexin enigma - from synapse formation to schizophrenia”. The ESHG award will be given to Svante Pääbo from the Max-Planck Institute of Anthropology in Leipzig, Germany, in recognition of his groundbreaking work on paleontology and understanding human evolution. The ESHG Educational Award will be given to Heather Skirton for her extraordinary contribution to the educational offers through the ESHG and the EGF.

The meeting will end with awarding the Best Young Investigators: There will be 8 awards selected from oral presentations (4 ESHG, 1x Isabelle Oberlé, 1x Lodewijk Sandkuijl, 1x Vienna Medical Academy Award, 1 x Mia Neri Award) and 2x for the best posters selected from the category of “best posters” (abstracts scored >3.8 and not included in the concurrent sessions).

I wish you a fruitful, informative and enjoyable meeting in Glasgow.

Brunhilde Wirth
Chair of the Scientific Program Committee 2015
Institute of Human Genetics,
University of Cologne, Germany

Report from the Public and Professional Policy Committee 2014-2015

By Martina Cornel, Chair of the PPPC

The Professional and Public Policy Committee aims:

To identify and discuss the ethical, social and policy issues related to human genetics and its application in research, clinical practice and laboratory genetic services.



- To be informed of various research projects, conferences and events, as well as policy initiatives and actions relating to those issues
- To inform and stimulate the discussion around these issues at meetings
- To address these issues and provide guidance through background documents, policy statements, recommendations or other publications
- To participate in the public debate around these issues
- To inform, interact with and provide advice to national and international policy makers

Whole-genome sequencing in newborn screening? A statement on the continued importance of targeted approaches in newborn screening programmes.

The advent and refinement of sequencing technologies has resulted in a decrease in both the cost and time needed to generate data on the entire sequence of the human genome. This has increased the accessibility of using whole-genome sequencing and whole-exome sequencing approaches for analysis in both the research and clinical contexts. The expectation is that more services based on these and other high-throughput technologies will become available to patients and the wider population. Some authors predict that sequencing will be performed once in a lifetime, namely, shortly after birth. The Public and Professional Policy Committee of the European Society of Human Genetics, the Human Genome Organisation Committee on Ethics, Law and Society, the PHG Foundation and the P3G International Paediatric Platform address in a joint publication the important issues and challenges surrounding the potential use of sequencing technologies in publicly funded newborn screening (NBS) programmes. The statement presents the relevant issues and culminates in a set of recommendations to help inform and guide scientists and clinicians, as well as policy makers regarding the necessary considerations for the use of genome sequencing technologies and approaches in NBS programmes. The primary objective of NBS should be the targeted analysis and identification of gene variants conferring a high risk of preventable or treatable conditions, for which treatment has to start in the newborn period or in early childhood.

Howard HC et al. *European Journal of Human Genetics* advance online publication, 28 January 2015; doi:10.1038/ejhg.2014.289.

Non invasive prenatal testing (NIPT)

As a “building bridges” activity, two of the world’s largest professional societies of human geneticists have issued a joint position statement on the promise and challenges of non-invasive prenatal testing (NIPT), a new procedure to test blood drawn from pregnant mothers for Down syn-

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drome and other chromosomal disorders in the fetus. The document addresses the current scope of and likely future improvements in NIPT technology, ways it may best fit with existing prenatal screening tools and protocols, options and priorities in its implementation, and associated social and ethical issues. By virtue of its greater accuracy and safety with respect to prenatal screening for common autosomal aneuploidies, NIPT has the potential of helping the practice better achieve its aim of facilitating autonomous reproductive choices, provided that balanced pretest information and non-directive counseling are available as part of the screening offer. Crucial elements are the quality of the screening process as a whole (including non-laboratory aspects such as information and counseling), education of professionals, systematic evaluation of all aspects of prenatal screening, development of better evaluation tools in the light of the aim of the practice, accountability to all stakeholders including children born from screened pregnancies and persons living with the conditions targeted in prenatal screening and promotion of equity of access. The statement, drafted by the Social Issues Committee of the American Society of Human Genetics (ASHG) and the Public and Professional Policy Committee of the European Society of Human Genetics (ESHG), has been posted on the ESHG website for membership consultation from 10 October 2014 until 15 November 2014, and sent to the ASHG and ESHG Boards to elicit further comments. It was published online March 18 in the European Journal of Human Genetics:

W Dondorp et al. (pub 2015 Mar 18). Non-invasive prenatal testing for aneuploidy and beyond: Challenges of responsible innovation in prenatal screening. *European Journal of Human Genetics*. DOI: 10.1038/ejhg.2015.57

Responsible implementation of expanded carrier screening

Carrier screening for recessive diseases is the detection of carrier status in persons who do not have an a priori in-

creased risk of being a carrier based on their or their partners' personal or family disease history. Expanded carrier screening is the detection of a greater number of recessive conditions than previously possible. Carrier screening aims to facilitate informed reproductive decision-making by identifying couples at risk of having an affected child. In the last decades, carrier screening was typically performed for relatively common, recessive disorders associated with significant morbidity and reduced life-expectancy. New genetic testing technologies enable the extension of screening to multiple conditions, genes or mutations with at a faster turnaround time for lower costs. Those expanded carrier screening panels that have been introduced to date have been advertised and sold to health care professionals and the public on a commercial basis. The ESHG Public and Professional Policy Committee, in collaboration with experts drafted a background document and Recommendations regarding the selection of disorders, timing, information and counselling, and informed consent. This document discusses the challenges that expanded carrier screening might pose in the context of the lessons learnt from decades of population-based carrier screening and in the context of existing screening criteria. It aims to contribute to the public and professional discussion and better clinical and laboratory practice guidelines.

This draft has been posted on the ESHG website and sent to the ESHG membership and external experts to solicit comments and further suggestions until March 15, 2015. After integration of the comments the document will be sent to the ESHG Board for approval.

ESHG-PPPC Course: Genetics in health care: Practice and Policies

The members of ESHG's Public and Professional Policy Committee shared their expertise with participants on 12-13 February 2015 in Istanbul in this interactive course. Approximately 30 participants from Turkey and the imme-



diate region had a diverse background: clinical geneticists, laboratory geneticists, genetic counsellors, and policy makers interested in genetics and its integration in health care. The aim of this course was to reflect on ways to improve the integration of genetics in health care in an efficient way: How can we make use of genetic testing, optimize referral and trace family members in case of, for instance, cancer and cardiac disorders? How can we improve counseling skills (personal contact, use of telecommunication and internet). How to creatively use scarce resources and prepare for further demand (establish centers of expertise and data bases). What are the social and ethical issues that need to be taken into account (such as autonomy and access) given recent European guidelines on genetic testing and screening? ESHG provided scholarships for 11 participants.

Prioritization in genetic testing

Already during the summer of 2013 a draft text on prioritization in genetic testing was posted on the ESHG website for consultation. Comments were integrated and the final text was approved by the Board of ESHG in March 2014. Genetic testing provides multiple benefits to patients and relatives, both from a clinical and a non-clinical perspective. While the potential to use genetic tests in health care is increasing fast, many experience difficulties in the capacity of the staff available and in the budgets. Medical benefit, health need and costs have to be taken into account when resources are too limited to fund all beneficial genetic testing services. The final document on "Points to consider for prioritizing in clinical genetics services" appeared in *Eur J Hum Genet.* 2014 Sep 24. doi: 10.1038/ejhg.2014.190.

Members of the PPPC in 2014-2015 were Pascal Borry, Martina Cornel (Chair), Florence Fellmann, Francesca Forzano, Heidi Howard, Hülya Kayserili, Christine Patch, Borut Peterlin, Dragica Radojkovic, Emmanuelle Rial-Sebbag, Wolf Rogowski, Maria Soller and Lisbeth Tranebjaerg, supported by Carla van El.

Report from the European Board of Medical Genetics - 2014/2015

by Heather Skirton, Chair of the EBMG

In the past year we have been able to build upon strong foundations. It was announced in Milan that the EBMG had been constituted as a legal entity. Our first General Assembly was held



during the Milan meeting and a Board was appointed. Professional Branch Board Chairs are Thomas Liehr and Isabel Carreira (Clinical Laboratory Geneticists), Heather Skirton and Milena Paneque (Genetic Nurses and Counsellors) and Ulf Kristoffersson (Medical).

The main highlight this year was the launch of the Clinical Laboratory Geneticists (CLG) Professional Branch launched the registration system for CLGs. As two branches are now offering registration, we are working closely with Jerome del Picchia to prepare an online submission system, which will expedite the application and assessment process. We hope that this will be available for registrants applying in 2015 (applications open from **August 1st**).

One problematic issue for those wishing to register as CLGs or genetic counsellors is the availability of relevant educational courses in some countries. The EBMG is working alongside the ESHG to help potential registrants to access the training they need to develop the necessary competences for registration.

One decision that was made by the EBMG General Assembly in 2014 was that practitioners can only be registered in the main branch of their profession. We have had numerous applications from medical doctors wishing to register as either laboratory geneticists or genetic counsellors. However, the EBMG has taken the decision that, while it is acknowledged that doctors may perform genetic counseling or work in a laboratory setting, each profession has its own specific requirements and dual registration is not appropriate.

Clinical Laboratory Geneticist Branch

The CLG branch has been extremely busy this year. The web page was constructed and the application process was planned. Previously, work had been done on categorising the training and assessment systems for CLGs in each country in Europe. Group 1 countries were defined as those where a recognised training and assessment process was available. In 2014, the registration system was opened for applicants from group 1 countries. In this first round of 271 applications, 259 were approved for European registration as a CLG.

We have now European registered CLGs from Austria (16), Belgium (52), Czech Republic (1), Finland (2), Germany (49), Hungary (3), Italy (5), Poland (3), Portugal (30), Sweden (1), Switzerland (19) and The Netherlands (78). Some of these CLGs work in Greece, UK or other countries.

Even though CLGs from Macedonia, Lithuania and Slovenia were eligible, there were no applications from those countries.

As from August 2015, applicants from all European countries can apply. The process will vary according to the applicant's country, as assessment of those applicants from countries where there is no current formal training or

formal assessment process will need to be more stringent than for Group 1 countries.

Go to <https://www.eshg.org/clg.0.html> for details on (1) the European registration process for CLGs, (2) a list of European registered CLGs, (3) a list of European countries and their alignment in groups 1 to 3, (4) courses suited for CLG (on-going) education and (5) the history of the board.

Genetic Nurse and Counsellor Branch

The registration process was run for a second year. We received a total of 28 notifications of intention to register. Most applicants applied using the grandfather clause, which still open as a registration route till 2018. From the initial 28 intentions, 25 were assessed as eligible and 17 full applications were received. In addition, we assessed three applications from the previous year that were re-submitted after working on minor or major changes.

The members of the NGC Professional Branch Board met for final agreement on the evaluation of all applications and as a result there are 20 new registered nurses or genetic counsellors. We congratulate those professionals who have successfully registered this year. They are:

Aurélie Ayme (Switzerland); Caroline Benjamin (United Kingdom); Inga Bjørnevoll (Norway); Mar Borregán (Spain); Carla Bruzzone (Italy); Karen Copeland (Switzerland); Christophe Cordier (France); Türem Delikurt (Cyprus); Abby Grant (Norway); Silvia Iglesias Casals (Spain); Rosie O'Shea (Ireland); Milena Paneque (Portugal); Sara Pabalados (United Kingdom); Rebecka Pestoff (Sweden); Mónica Salinas (Spain); Janice Scott (United Kingdom); Ares Solanes (Spain); Vigdis Stefansdottir (Iceland); Siv Lisbeth Tønder (Norway); Ángela Velasco (Spain).

Currently only practitioners registered with the GCRB (UK) and American Board of Genetic Counsellors can seek European registration under the national registration clause, but the Division is actively working to extend this to other international systems (e.g. Australian, Canadian and South African Board).

We have received enquiries about approving other MSc courses and have streamlined our application for approval. In addition, the registration process has been improved by provision of more clear information and instructions on the website and the development of a process for dealing with a complaint about a registered practitioner or person applying for registration.

The high quality of applications has been very satisfying and the number of applicants from a wide range of countries leads us to believe that there is real value for the profession in this registration system for genetic nurses and counsellors in Europe.

Medical Branch

The EBMG section for medical doctors is working alongside the UEMS Section for Clinical Genetics. This enables

effort in setting and ensuring standards to be maximised for the profession. The UEMS section has commissioned the EBMG to update the curriculum and suggest a syllabus for training of residents in clinical genetics. The members of the section syllabus group for 2014-2015 are: Ulf Kristofersson, Lund (chair); Bela Melegh, Pécs; Peter Turnpenny, Exeter; Alessandra Renieri, Siena; Beata Lipska, Gdansk; and Feliciano Ramos (ESHG observer), Zaragoza.

The updated curriculum will be presented at the UEMS membership meeting on June 6 at the ESHG meeting in Glasgow, and, if endorsed, brought to both the UEMS sections and boards meeting, and the ESHG board and membership during the autumn for endorsement. These documents will serve as a base for a European examination in medical genetics. The syllabus is expected to be finalised during the coming year, and the first examinations may be held in 2017.

Thanks

We sincerely thank the ESHG Executive for continuing encouragement and advice. As ever, we could not carry out the work without the immense practical support we receive from Jerome del Picchia and the staff of the VMA.

Finally, the members of each branch contribute considerable amounts of their time on an entirely voluntary and unpaid basis and the success of the registration schemes is due to their diligence and generosity. I thank them most sincerely.

Heather Skirton
Chair
European Board of Medical Genetics

ESHG educational activities

By Han Brunner

The importance of education in genetics in Europe

Human and Medical geneticists throughout the world are involved in teaching and educating modern human and medical genetics and genomics.

Educational efforts can be aimed at the general public, at medical and biomedical students, at professionals in training for a genetic specialty, at genetic specialists as part of continuing education, and at other healthcare professionals.

Existing efforts:

Each target audience has its own needs, and ESHG already



has activities with an educational perspective. These include:

- Educational sessions at the annual ESHG conference a part of continuing professional education
- ESHG course in Bertinoro and elsewhere for young professional in Genetics, and genetic counsellors
- DNA day essay contest involving school children throughout Europe.
- One-day School event associated with the annual meeting organized by the local hosts

New developments in 2015:

The ESHG executive board proposes to establish a new Education Committee that will consist of 4 working groups, each tasked with overseeing one of these activities. This will be decided at the ESHG board meeting during our annual meeting in Glasgow.

During the last year, the Society has significantly expanded its course portfolio. ESHG has a long tradition of providing courses that aim at young professionals in all areas and subspecialties of Human Genetics.

A number of courses are run at Bertinoro in Italy. These include annual courses in Medical genetics, and in Next Generation Sequencing, and bi-annual courses on Genetic Counseling, and on Ophthalmic genetics.

The dysmorphology course in Manchester has also become something of a tradition.

New additions are planned for 2015 – 2017 notably courses on cardiac genetics, hereditary cancer, prenatal and pre-implantation genetics, and statistical genetics.

ESHG will provide fellowships to especially enable students from low-income countries to attend, in addition to those from more privileged countries who can pay their way.

With these new additions, a concerted program of professional continued education is taking shape.

Further plans will be explored by the new Education Committee after Glasgow.

On behalf of the ESHG executive Board

Han Brunner
ESHG vice-president

Obituary

On 8 December 2014 Franca Dagna Bricarelli passed away at the age of 73 years and the European community of geneticists lost one a main figure in medical genetics and a dear friend.

An obituary by her colleague Paola Granmatico can be found at www.eshg.org/news.0.html

EuroGentest-Quality Committee

By Ros Hastings, Chair of the GSQC

The Genetic Services Quality Committee (GSQC) meets bi-annually. The Committee is informally referred to as the Quality Committee and its aims are to:-



- Identify gaps in quality issues within diagnostic genetic testing services;
- Identify where there can be harmonisation between the biochemical genetic, cytogenetic and molecular genetic disciplines;
- Commission and approve new documents relating to quality in genetic testing;
- Give recommendations for those countries where no guidance is currently available.

Six areas of need relating to quality issues in the genetics community have been given priority and working groups with the committee will take them forward.

- Changing landscape of genetic testing
- Laboratory performance in EQA
- Genetic Counselling
- Best practice guidelines
- Rare Variants
- Newborn Screening

Following the satellite meeting and workshop on External Quality Assessment of Genetic Counselling in Paris in 2013 and in response to requests from the European Genetic Counselling community, the ESHG Quality Committee Genetic Counselling Working Group organised and developed, in collaboration with CEQAS, an educational pilot EQA for Genetic Counselling. The pilot EQA provided four multi-stage case scenarios (one each for dysmorphology, oncogenetics, monogenic disorder and cardiovascular genetics) with each case based on a real scenario as presented to the clinical geneticist who compiled the case. Out of a total of twenty-two enrolled centres, 16 centres, from 11 countries participated in the educational pilot EQA. Participants were asked to also complete a questionnaire to evaluate the outcome of the pilot. The results of the educational pilot EQA will be presented at the CEQAS Participants' Meeting on 7th June 2015 and as an oral presentation during session C-14 'Challenges in Genetic Counselling' on 8th June 2015 at the ESHG annual conference in Glasgow.

The GSQC provides a governance structure for the four

European EQA schemes [CEQAS - Cytogenetics, CF Network - Molecular Genetics, EMQN - Molecular Genetics and ERNDIM - Biochemical Genetics] and reviews their annual management reports. There are still some EQAs across the genetic EQA providers where >10% of laboratories had a critical analytical error. Training workshops have been instigated and a successful workshop on reporting with 74 participants was held in 2014 at the ESHG annual conference in Milan.

Due to the integration of EuroGentest into the ESHG, there has been a restructuring of the GSQC. There are now four sub-committees which will share the remit of the original Genetic Services Quality Committee and feed back to the EuroGentest committee which will have a management and coordination role. The four sub-committees are: Quality Sub-Committee (QSC); Professional Guidelines Sub-Committee (PGSC); Training Sub-Committee (TSC) and Dissemination Sub-Committee (DSC). Committee members will be appointed for at least three years.

The Quality Sub-Committee (QSC) will continue to work on issues that affect the quality and competence of diagnostic genetic laboratories and genetic counselling clinics. The chief aims and objectives of the QSC are to:-

- Identify gaps in quality issues within diagnostic and clinical genetic testing services;
- Identify where there can be harmonisation within genetics Quality Management (QM) and service provision;
- Give recommendations for those countries where no guidance is currently available.
- Promote harmonisation with EQA providers to help to reduce poor performance in genetics;
- Review management and governance of EQA providers;
- Establish educational EQAs for genetic counselling;
- Define quality issue relating to rare genetic diseases covered by Rare Diseases Topic Advisory Group;
- Address quality issues in new-born screening programs;
- Explore quality issues relating to International databases of genetic variants;
- Review ISO standards or other standards with reference to genetic testing laboratory needs and propose changes to the ISO committee or representatives.

Help us to help you further by letting us know of any quality issues that need addressing. Please submit your suggestions to any member of the new Quality Sub-Committee: Ros Hastings (Chair), Conny van Ravenswaaij-Arts, David Barton, Brian Fowler, Victor Kožich, Thomy de Ravel, Marta Rodriguez de Alba, Els Dequeker (Advisory).

A list of the Committee Members and a synopsis of the meetings are also available on the ESHG website.

EJHG Highlights of 2014

By Gertjan van Ommen, Editor in chief, EJHG

Impact Factor ups and downs

In the previous years EJHG's Impact Factor has been (very) slightly decreasing, from 4.4 in 2012 and 4.32 in 2013 to 4.23 in 2014. EJHG now ranks 37/164 in 'Genetics and Heredity' and 76/291 journals in 'Biochemistry and Molecular Biology'. However, a preliminary citation analysis suggests that EJHG's IF may go up again, as the citations of 2014 papers are slightly better than those to 2013 papers. No warranties, it's not over till the fat lady sings - but she has been singing late recently: last year the IF data arrived only in July.



Submission surge – increased rejection rate

The major surge of submissions in 2012 and 2013 has gone on in 2014. There was an increase from 771 in 2012 to 876 in 2013 and we rounded the 1000 in 2014! This has caused our processing time to rise. Our decision time for the 55% of manuscripts rejected without review lies around 14 days. The average time to first decision for reviewed manuscripts is now 65 days. Our overall acceptance rate has remained constant at about 35%. After acceptance, typically papers are published online in 25 work days. However the surge in submissions has caused a too long delay for the actual print publishing, around 9 months now. Fortunately, the submission trend seems to be curbed: in the first quarter of 2015 we saw 15% fewer submissions than the same period in 2014. To further reduce shelf life – and increase our Impact Factor! - we aim to increase the editorial rejection rate to 65%.

New Section Editors

To assist with the work load, we have welcomed several new Section Editors: Nicole Soranzo (Cambridge), Cecilia Lingren (Oxford) and Jenny Barrett (Leeds) for Statistical Genetics and Maurizio Genuardi (Florence) for Cancer Genetics

Web visibility

The journal website has been viewed nearly 450,000 times in 2014 – an increase of 7% compared to 2013 (419,675 page views). This equates to an average of 4,996 page views per day. Full-text articles amassed more than 315,000 views, compared to 287,206 at the beginning of 2014. The highest proportion of visitors to the website

came from those located in the US (31%), followed by the United Kingdom (9%), China (6%) and Canada (4%). Direct traffic to the website accounted for 20% of views, followed by the referring domain Google.com (18%) and by PubMed (11%).

Electronic Table of Contents (e-Alerts): The average number of individuals receiving e-Alerts is currently approximately 60,000 per month.

EJHG Award

As every year, EJHG, and Nature Publishing Group, jointly offer a junior authors' high-citation award. This is given to the top-3 articles published in 2013, with citation counted in the 12 months following after publication. The 1st prize includes a € 500 award and positions 1-3 receive one year free ESHG membership, including an online EJHG subscription, and free registration for that year's or next year's meeting. This year we have two ex aequo winners who will both get a first prize! The winners are:

1. Bradley N Smith, Stephen Newhouse, Aleksey Shatunov, Caroline Vance, Simon Topp, Lauren Johnson et al. The C9ORF72 expansion mutation is a common cause of ALS+/-FTD in Europe and has a single founder. *Eur J Hum Genet.* 2013 Jan;21(1):102-8. doi: 10.1038/ejhg.2012.98. Epub 2012 Jun 13.
19 Citations

1. Charlotte Andreassen, Jonas B Nielsen, Lena Refsgaard, Anders G Holst, Alex H Christensen, Laura Andreassen et al. New population-based exome data are questioning the pathogenicity of previously cardiomyopathy-associated genetic variants. *Eur J Hum Genet.* 2013 Sep;21(9):918-28. doi: 10.1038/ejhg.2012.283. Epub 2013 Jan 9.
19 Citations

2. Linda Broer, Veryan Codd, Dale R Nyholt, Joris Deelen, Massimo Mangino, Gonneke Williamsen et al. Meta-analysis of telomere length in 19 713 subjects reveals high heritability, stronger maternal inheritance and a paternal age effect. *Eur J Hum Genet.* 2013 Oct;21(10):1163-8. doi: 10.1038/ejhg.2012.303. Epub 2013 Jan 16.
18 Citations

The first authors will be given this award in the final ceremony of the Glasgow meeting, to be handed by Nature Publishing Groups' managing editor for EJHG, Sohini Pavlovic.

Immediacy score

An important impact predictor turns out to be the 'immediacy score' of papers: citations gained in the same year of publication. Indeed, the #1 winner of this year's EJHG award was already mentioned as a contender in this report last year. Similarly, the paper "Panel-based next generation sequencing as a reliable and efficient technique to detect mutations in unselected patients with retinal dystrophies" by Glöckle et al., published in EJHG of January 2014, is a strong candidate for next year's award: it received 13 cita-

tions already by December that year.

New Partnership between ESHG & EURORDIS

By Eva Bearryman, EURORDIS

The ESHG and EURORDIS, the European Organisation for Rare Diseases (booth #112), have entered into a new partnership. Both organisations will work together to promote or develop awareness of rare genetic diseases, research for rare diseases and access to information.

The two organisations will align and support one another on a range of activities including conferences (including the annual ESHG Conference and the European Conference on Rare Diseases & Orphan Products, the latter of which is organised by EURORDIS and partners), policy actions and initiatives of common interest.

EURORDIS is a patient-driven alliance representing 678 rare disease patient organisations in 63 countries. The organisation's mission is to build a strong pan-European rare disease community and to be the voice of the 30 million people living with a rare disease in Europe.

EURORDIS seeks to improve the quality of life of rare disease patients and reduce the impact of rare diseases on the lives of patients and families by supporting research and medicines development, advocating at the European level, facilitating networking between patient groups and raising awareness, among many other activities.

Patients' representatives have an increasingly present voice in all aspects of drug development from fundamental research through regulatory processes and to health technology assessment. Although major advances have been made in raising awareness and increasing funding for rare disease research, important challenges remain in terms of best use of resources, coordinating efforts and improving policy. EURORDIS and other rare disease patients' organisations are continually developing initiatives, from providing funding to being directly involved in research projects, at the national and European level to promote research into rare diseases.

EURORDIS is a partner in a range of genetic research projects including RD-Connect (an FP7 infrastructure project that links databases, registries and biobanks used in rare disease research into a central resource for researchers worldwide) and the Genetics Clinic of the Future Project (GCOF), part of the H2020 Framework Programme, which aims to map the opportunities and challenges that sur-

NEWSLETTER PARTNERSHIP BETWEEN ESHG & EURORDIS

round the clinical implementation of new genome technologies. Through the GCOF Project, the hope is to be able to understand how to harness the potential of these technologies for healthcare while still respecting fundamental ethical and regulatory frameworks.

E-Rare-3 is a H2020 project that aims to pursue and expand activities that accelerate the development of new diagnostics and therapeutics for rare disease patients. E-Rare-3 launches annual calls to fund research that addresses research gaps. EURORDIS participates in this project to open the possibility for patients' organisations to foster their engagement in funding of rare disease research at a transnational level. Within this project, EURORDIS will coordinate a network of patient organisations that fund research to develop an innovative funding scheme with national funders.

The International Rare Diseases Research Consortium (IRDiRC) brings together researchers and organisations involved in rare disease research and aims to achieve two main objectives by the year 2020 to deliver (1) 200 new therapies for rare diseases and (2) means to diagnose most rare diseases. Yann Le Cam, EURORDIS Chief Executive Officer, chairs the IRDiRC Therapies Scientific Committee.

EURORDIS and partners also organise the European Conference on Rare Diseases & Orphan Products, the 8th edi-

tion of which will take place in Edinburgh 26 – 28 May, 2016. The Conference provides the latest updates on the rare disease environment and covers research, development of new treatments, healthcare, social care, information, public health and more. The call for posters for the Conference will open in September 2015 at www.rare-diseases.eu.



EURORDIS is a non-governmental patient-driven alliance of patient organisations and individuals active in the field of rare diseases, dedicated to improving the quality of life of all people living with rare diseases in Europe.
www.eurordis.org



Transforming Medicine Through Responsible Data Sharing

By the Steering Committee of the Global Alliance for Genomics and Health

The combination of genomic and clinical data holds great promise for facilitating the discovery of disease mechanisms and the development of new therapeutics. But in many cases, making such advances requires access to datasets that are larger than any single laboratory or institution can collect on its own. Interoperable protocols and methods for smoothly accessing and exchanging data would allow many more members of the research community to use them in innovative ways, thereby advancing biomedical science.

The Global Alliance for Genomics and Health (GA4GH) was founded in 2013 to enable such responsible data sharing to advance research and human health. We aim to do this by convening key stakeholders, assessing current policies and procedures for data-sharing, and, when necessary, developing new interoperable technical methods and harmonized regulatory frameworks and approaches. We hope these efforts will help shape the emerging ecosystem of genomic and clinical data generators and users in ways that will serve biomedical research as a whole.

GA4GH began as a collection of 70 institutions committed to this cause. In two years, we have grown to include more than 300 member organizations across more than 30 countries. Global leaders such as the National Institutes of Health, GlaxoSmithKline, and Google have come together across the biomedical, technical, and clinical sectors. Within this large, diverse partnership, smaller working groups have convened to take on the most pressing issues and develop products. A series of exemplar projects are demonstrating value by putting these products into immediate practice.

Storing, aggregating, and harmonizing genomic data

Due to ever-improving genomic technology, petabytes of data are amassing at decreasing cost in both the clinic and the laboratory. These data must be rendered interoperable for structures to efficiently and effectively store, aggregate, and integrate them. A wide range of barriers and challenges exists, from incompatible file formats to conflicting international protocols.

To overcome these challenges, the GA4GH Data Working Group (DWG) is developing internationally relevant bioinformatics file formats and application programming interfaces (APIs) for the exchange of genomic sequencing data.

While the DWG does provide oversight for integrating existing file formats, these formats are disadvantaged in a globally distributed, large-scale, and increasingly cloud-based environment. Thus, the DWG's main priority is to develop new data models, APIs, and reference implementations specifically designed for the unique needs of scalable, distributed, genomic data sharing.

The DWG has released the GA4GH Genomics API, currently v0.5.1, which allows for the interoperable exchange of genomic information across multiple organizations on multiple platforms. The freely-available, open standard for interoperability is available as a web service that can be integrated into various types of genomics-related software and infrastructures.

Establishing appropriate regulatory and ethical protocols

For data sharing to have broad impact, the biomedical community must adopt a new structure that will incentivize participation among scientists, clinicians, and research subjects. But we must first overcome many regulatory and ethical challenges, including a myriad of complex and often conflicting regulations. International guidelines and ethical frameworks must be established to ensure that the transition to a more open system is responsible, transparent, and inclusive.

Toward that end, the Regulatory and Ethics Working Group (REWG) of the GA4GH seeks to harmonize policies, procedures, standards and codes of conduct for the storage, analysis and sharing of genomic and clinical data; develop forward-looking consent and privacy procedures that responsibly engage patients and researchers; and develop best practices in governance and transparency of data repositories.

In 2014, the REWG developed the Framework for Responsible Sharing of Genomic and Health-Related Data, which relies on human rights, especially the UN's Universal Declaration of Human Rights, for "actionable" guidance and inspiration. The Framework has as its foundations the right of all citizens to benefit from scientific progress, and the rights of data producers and users to be recognized for their contributions to research, balanced by the rights of those who donate their data. Other REWG teams are developing policies and tools for harmonizing genomic and health-related data sharing practices across international boundaries.

Connecting genomic data to the clinic

Human genomic data is often only as useful as the phenotypic data attached to it. Unless we can effectively bridge the gap between genomic data collected for research and phenotypic data collected in the clinic, the potential of both efforts will be vastly under-realized. The Clinical Working Group (CWG) was established to help prevent that fate.

The CWG is developing standardized approaches for repre-

senting phenotypic information and linking it to genotypic data, specifically in the areas of rare genetic disorders, cancer, complex traits, and infectious diseases. To date, it has created catalogs of data sharing activities around the globe. These catalogs are intended as resources for researchers and clinicians as they attempt to work together across diverse national and institutional boundaries.

Navigating issues of privacy and security

If individuals do not feel safe in sharing genomic and clinical data broadly, the potential benefits described above will be irrelevant. We need protocols for both dictating and securing various levels of privacy that are interoperable across systems and regions. These protocols must also be flexible, such that they ensure participant autonomy while also making it possible for consent to be transferred across organizations, updated based on new information, or withdrawn.

Several industries—including healthcare, business, and finance—have already established standards and technologies for protecting the privacy of individuals while simultaneously ensuring the confidentiality, integrity, and availability of large datasets. The Security Working Group (SWG) takes inspiration from these existing sectors and, when possible and appropriate, reuses technologies and standards already in operation. We also anticipate benefiting from the industry-leading security structures of the commercial cloud, where data can be most easily stored.

The SWG has developed a security infrastructure in which it recommends specific policy and technology for stakeholders in the GA4GH ecosystem.

Demonstrating value through real-world activities

A series of cross-cutting demonstration projects utilize the GA4GH Working Groups and the resources they create to exhibit the transformational possibilities of genomic data sharing. These projects aim to ensure relevance by aligning with the most pressing needs within the community. The three current demonstration projects are Matchmaker Exchange, BRCA Challenge, and the Beacon Project. Matchmaker Exchange is a federated network of rare disease datasets, which allows researchers around the world to locate data on rare phenotypes or genotypes of interest. BRCA Challenge (see companion piece in this newsletter) is an international collaboration to improve our understanding of the genetic basis of breast cancer. It aims to produce a definitive and freely accessible database of variants in the breast cancer genes BRCA1 and BRCA2 pooled from around the world. The Beacon Project is a technically simple public web service that tells users whether or not a participating dataset contains any genomes with a particular base at a specific point on the genome. The simple “yes” or “no” response is designed to ensure patient privacy while still encouraging data sharing and genomic research.

Driving Progress for Responsible Data Sharing

As genomic and clinical data accrue at unprecedented speed and volume, the world faces an inflection point: Genomic data will only improve health and medicine through a cultural shift toward data sharing. But this demands effective stewardship and broad participation across the scientific, regulatory, and patient communities.

Nearly two years after our inception, we are encouraged to see real engagement among our ever-growing membership, which will convene in Leiden in June 2015. At our third plenary meeting members will hear about progress made on our key projects and updates on the GA4GH as an organization, and will have the opportunity to provide input on our forward-looking goals and next steps.

But our efforts demand even broader participation. Increased engagement among individuals and organizations across diverse sectors and geographies is at the forefront of our goals for the coming years. For we believe that the great promise of genomic medicine will only succeed if the global community recognizes the value and urgency of shared data.

The BRCA Challenge: Improving health through data sharing

By Dr. Rachel Liao, the Coordinator for the Clinical Working Group and the BRCA Challenge at the Global Alliance for Genomics and Health

and

Professor Sir John Burn, Professor of Clinical Genetics, Newcastle University

Introduction

BRCA1 and BRCA2 are well known to the scientific community for their crucial roles in maintaining DNA damage response pathways in cells. Since the discovery of their association with hereditary breast and ovarian cancer more than twenty years ago, they have also become of great interest to clinicians, particularly as the use of diagnostic sequencing has increased.

Indeed, recent technological advances and regulatory decisions have led to an explosion of diagnostic sequencing of these two genes. Due to the combination of their large size (BRCA1’s coding sequence alone is more than 5kb and BRCA2’s coding sequence is 10kb) and high level of polymorphic variation, sequencing of these genes frequently results in identification of a variant of uncertain significance (VUS), the implications of which are entirely

unknown to the clinician, the patient, and the medical and research community at large. Not infrequently, this uncertainty leads to drastic surgical treatment decisions and increased unnecessary mental anguish.

Interpretations of VUSs emerge as recurring variants are identified and collected, along with the medical and family histories of cancer patients, which lead to more informed treatment decisions and superior patient outcomes. However, variants from around the world are currently stored in siloed databases, which are unique to their institutions and are generally not able to incorporate information on variants in other databases because of technical incompatibility and privacy concerns. This balkanization stymies potential interpretations that could be identified if the databases were pooled.

This is a conundrum—more sequencing of BRCA1 and BRCA2 means that more variants will be available for interpretive analyses of pathogenicity, yet large collections of variants are fundamentally unable to communicate in order for researchers to perform such health-benefitting analyses. Therefore, we propose a solution: to develop strategies to share data across databases that are aggregating variants and interpretations of pathogenicity, using standards and methodology approved by the Global Alliance for Genomics and Health (see companion piece in this newsletter) to ensure safety, privacy, proper regulatory oversight, and scientific rigor, for the purpose of improving variant interpretation and easing the uncertainty of treatment decisions made because of a VUS.

The BRCA Challenge of the Global Alliance for Genomics and Health

The BRCA Challenge, born out of these issues, has a stated mission: to improve the care of patients at risk of monogenic disease using global data sharing and collaboration in the analysis of BRCA1 and BRCA2. We propose to accomplish this goal using the following strategies:

1. All members of the BRCA Challenge agree to share BRCA1 and BRCA2 variants publicly
2. The BRCA Challenge creates an environment for collaborative variant curation with access to evidence, including phenotypes, family history, genetic data, and functional studies
3. The BRCA Challenge helps develop curated lists of variants that have been interpreted by expert consensus and will enable accurate clinical care
4. The BRCA Challenge addresses the social, ethical, and legal challenges to global data sharing

The BRCA Challenge is co-chaired by Dr. Sir John Burn of Newcastle University and Dr. Stephen Chanock of the National Cancer Institute (US), who lead a steering committee of experts in the field who are committed to appropriate data sharing for the benefit of patient care. They, along with members of working groups, have begun to tackle

several key issues associated with the strategy above, including:

- Ethics, regulation, and patient advocacy concerns
- Evidence gathering standards to streamline technical needs
- Classified variant database identification and aggregation
- Interpretative rigor to resolve classification disputes and classify VUSs

The BRCA Challenge has received support and commitment to share from large databases and BRCA interpretation organizations including ClinVar, LOVD, and ENIGMA. In the near future we hope to welcome more global databases, and to solicit, deposit, and classify variants not yet in the public domain.

Conclusions

The explosion of genomic sequencing demonstrates a profound need for high-quality data sharing strategies in all contexts, and the particular burden from hereditary breast and ovarian cancer is clear, even as more sequencing increasingly leads to uncertain prognosis. The BRCA Challenge rises to meet this international need where sequencing, regulation, and clinical care come together on the important issue of hereditary breast and ovarian cancer risk. We hope that this initiative will set the stage for similar data sharing and collective interpretation across the whole “Human Variome”.

This is the moment in time to have a profound impact on patient health and well-being, and we must get to work to accomplish it.

Report of the Executive Officer

By Jérôme del Picchia, Executive Officer of the ESHG

Time and again

With a regular attendance of around 3,000 delegates, the ESHG conferences have become a quite complex and elaborate meeting over the years and a society with over 2,500 members has entirely different needs than one with 1,000. When writing these articles, it is difficult to stay on top of the multitude of details and innovations we try to implement together with my colleagues *Kristina Libova* and *Mirjam Uebelhör* from the conference department and *Andrea Robinson* from the association department. There are quite a few obvious answers to the question “What’s new, actually...?”, at least for



us, behind the scenes. One very visible addition would be the live streaming of the plenary hall lectures into the exhibition hall, which will equally serve as "overflow area". In case a lecture room is overcrowded, our AV technicians can flip a few switches and transmit the presentation images and sound of this room directly into an area with an additional 150 seats in the exhibition.

A new session format (at least for ESHG conferences) was born around a dinner table last year in Milan in a discussion with the ESHG Board Members *Han Brunner*, *Milan Macek Jr.*, *Brunhilde Wirth* and *Martina Cornel*. A TEDx format workshop: "A case that changed my life as geneticist", a session about storytelling in genetics. We have invited professionals in medical genetics to submit a short video in which they describe an event or moment when genetics made a difference and had a profound impact on how they see the field of Medical Genetics. Five submitters were chosen to tell their story. I believe that this is really worth a look: Saturday, June 6, 10.30 hrs in Hall 1.

While resting or studying in the seating area on the far end of the poster exhibition, you may notice the "phone & tablet charging towers" with cables and connectors fitting most smart phones of today; a small but convenient novelty these days. As of Monday morning, most of the laptops used for on site registration will be converted in "check-in and print your boarding pass" stations with a printer. Feel free to drop by.

A very interesting novelty was put forward and eventually realised by my colleague *Andrea Robinson* of the membership department. Knowing that most membership renewals arrive in close proximity to registration deadlines before the annual meeting, and that these are sometimes over a weekend, our membership often had to experience a certain delay, which inevitably occurred when many hundreds of renewals needed to be processed in just a day or two. Andrea, in cooperation with our IT department, developed the ESHG online membership renewal and at the same time included improved security features for online credit card payments via an external Payment Service Provider. I must say that, while being rather cautious at the beginning (these things often take time to work adequately), the result clearly convinced me that this was the right step to take at this stage. Not only that you are able to renew, pay your fee and get a confirmation instantly, you are also able to edit your contact details at any time without having to contact the ESHG office (which you obviously still can do if you prefer so). In case you wonder what the cost-value ratio of such an improvement is: it paid off in the first year. The administration costs of 2014 decreased by more than the investment, while there was an almost 40% increase in number of members in 2014.

The Offspring

Besides these mostly administrative changes, I was very impressed with the substantial developments that the ESHG, its committees and their "offspring" have been going through in the last 12 months, namely the successful marriage of *EuroGentest* and the *ESHG Quality Committee*. The EuroGentest meeting on the Friday pre-

ceding the ESHG meeting will be a regular satellite in the future. The tremendous success of the *European Board of Medical Genetics* who started an apparently longed for registration process for Clinical Laboratory Geneticists and Genetic Counsellors and Genetic Nurses was clearly another highlight of the year 2014 ESHG was involved in. The next round for registrations will start in August 2015 on the EBMG website www.ebmgeu. Last but not least, I have to mention the recently started reformation of the *ESHG Education Committee* now focusing, among others, on developing a new ESHG course portfolio of which you will see the first results very soon, while continuing the very successful cooperation with the European School of Genetic Medicine in Bertinoro. You will be able to read about most of these developments in the articles by my co-authors in this Newsletter.

Allegiance

I sincerely hope that this 15th Annual Meeting of the ESHG I had the privilege to organise together with my colleagues of the *Vienna Medical Academy* (starting with the ICHG 2001 in Vienna) will continue the successful history of the recent ESHG meetings and that you will be able to enjoy the great scientific programme and also have the time to get a glimpse of the fascinating city of Glasgow.

I certainly look forward to seeing you at the CCIB in beautiful Barcelona in May 2016 for the joint meeting of the ESHG and the European Meeting on Psychosocial Aspects of Genetics!

Centre de Convencions Internacional de Barcelona

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Invitation to the

Annual Membership Meeting 2015

At the EUROPEAN HUMAN GENETICS CONFERENCE 2015

Sunday, June 7, 2015 at 7.00 – 8.00 p.m.

Room: Gala

ESHG Conference venue: SECC, Glasgow, Scotland, United Kingdom

AGENDA

1. Opening by the President of the Society, Professor Helena Kääriäinen
2. Activity of the Society 2014-2015
3. Financial Report of the Society 2014
4. Discharge of the Board Members for the year 2014-2015
5. Results of election for President-Elect
6. Results of election for Board Members
7. Membership fees 2016
8. Site of future European Human Genetics Conferences
9. Budget proposal 2016
10. Major policy questions proposed by Board
11. Future activities

Please find the minutes of the last membership meeting in Milan 2015 in the restricted area:

<https://www.eshg.org/members.0.html>