Presidential Address
by Han Brunner, President of the ESHG

The ESHG is coming to Milan for its 48th Annual conference. Italy has always been one of the more active countries on the European scene. Three of our past presidents have come from Italy, and a large number of scientists and clinical geneticists actively participate in our meeting each year. We are welcomed by the Italian Society of Human Genetics and local host Antonio Amoroso.

The meeting continues to grow in the number of attendees, and in its content. This is due to the continued activities of the conference organizers at the Vienna Medical Academy, to Rose International who organize our exhibition, and of course to the excellent work of the Scientific program committee under the leadership of Brunhilde Wirth.

This year, the meeting has a plenary debate on how we deal with the incidental findings that may come with various forms of next generation genetic analysis. This session is the first of a series of sessions that are jointly developed between ESHG and ASHG under the title “building bridges”, and that will be held at the annual meetings of the two societies. The boards of ESHG and ASHG have decided that they will develop further connections and foster exchange between the two societies.

We see many new developments in Human Genetics, both scientifically and in the practice of medical genetics. The game-changer of this year clearly is the wide-spread implementation of NIPT in many European countries. We shall hear about experiences from several countries at the meeting.

The European Society of Human Genetics (ESHG) has developed a partnership with the European School of Genetic Medicine (ESGM), in order to promote advanced training in human-medical genetics and preventive medicine in Europe. These courses began in 1988 with the first course in Medical Genetics of the European School of Genetic Medicine directed by Prof. Victor A. McKusick and organized by Professor Giovanni Romeo. Courses in 2014 include Genetic Counselling, Next Generation Sequencing, Medical Genetics, and Eye Genetics. ESHG provides a number of fellowships aiming to allow students from less privileged parts of Europe to attend these courses, and to connect with modern genetics.

ESHG needs to play an active role in fostering responsible, informed, and innovative applications of genetics and genomics to healthcare within Europe. This takes many shapes. In the past year we have seen a number of documents prepared by the Publications and Public Policy Committee, for instance on the use of Whole Genome Sequencing in Neonatal Screening, and on Whole Genome Sequencing in Health Care. Currently, ESHG takes an active stance in the political discussions in Europe on a proposed EU directive on in vitro devices which has an added amendment on genetic counselling which the society finds is burdensome, bad for patients, unworkable, and according to legal opinion the EU doesn’t have the legal authority to enact.

As another example, the ESHG has decided to incorporate a number of EUROGENTEST activities, and to merge these with those of the Genetic Services and Quality Committee. Through the European Board of Medical Genetics, the ESHG aims to develop systems of certification and/or re-certification for professionals working in genetic healthcare in Europe. This includes Clinical laboratory geneticists working with laboratory diagnosis of human genetic disorders, genetic nurses and genetic counsellors, and clinical geneticists. A specialist section for Clinical Genetics was created this year in UEMS (Union for European Medical Specialists). An important milestone was reached this year: The European registration process for genetic counsellors and genetic nurses is now starting, and applicants can visit the ESHG website for information on the process. All these activities signal ESHGs commitment to maintain and improve quality standards on genetic care across Europe.

More than 40 presidents of national societies will meet at the Milan conference to discuss the current developments in human genetics across Europe and to find topics that we can develop together within ESHG.

The European Journal of Human Genetics has established itself among the leading scientific journals in the field of human genetics under the inspiring leadership of Gertjan van Ommen. The current impact factor of EJHG (4.32) attests to its relevance to the wider community of human genetics. The journal received more than 800 submissions in 2013, of which roughly half are from Europe with the other being distributed across all continents. Because of the continued
popularity with authors, only the best 1/3 could be accepted after careful review. The journal is well-read with more than a million online article views from more than 200 countries in addition to the regular print readership. Taken together we see that ESHG has come a long way in providing a platform for outstanding science in the field of human genetics. The task ahead is to keep doing this while at the same time engaging actively in the many discussion on the implementation of all this new knowledge into our health care systems and society at large. I believe that we should all consider ourselves lucky to be able to participate in all these developments, whether they are purely scientific, applied or of a societal or regulatory nature. ESHG aims to be the focal point for these developments now as well as in the future.

Han Brunner
President of the ESHG

Secretary General’s report
by Gunnar Houge, Secretary General of the ESHG

The annual conference is the major task of ESHG. There are three major players in this arrangement: The Scientific Planning Committee (SPC) that makes the program, the Vienna Medical Academy (VMA) that organises the conference, and Rose International that deals with sponsors and exhibitors. Without a well functioning SPC ESHG would be in trouble, also financially. Even though these conferences aren’t planned to return a profit, we can easily be hit by a big loss if the quality isn’t good and people do not find attendance worthwhile. We are therefore most grateful to Brunhilde Wirth and the SPC for yet another great scientific meeting and to the VMA staff for making it all run so smoothly.

We want ESHG to be remembered not only for good scientific and educational conferences, but also for good parties. The conference parties are therefore subsidised by us to make them affordable for a major target group: the younger people in our society. We want our conferences to be of special value for the young, and the party is such a nice place for them to easily meet and make new friends.

As you can see in this Newsletter, ESHG is not only the annual meeting. We are a society of different professionals that need each other, reflected by the three branches of our new independent legal body: the European Board of Medical Genetics (EBMG). We are also a society promoting quality, reflected by our Genetic Services and Quality Committee and our adoption of EuroGentest. We are also a society for education in genetics, mainly on the professional level but also on the society level through the DNA day, arranged by our Educational Committee in collaboration with ASHG.

I addition, you may have noticed that the traditional European School of Medical Genetics courses in Italy are now a part of the ESHG course portfolio, and still sponsored by ESHG fellowships. Three successful courses were arranged in 2013, and three more will take place in 2014. In addition, the educational tracks that run through our annual meeting, an idea put forward by Ulf Kristofferson some years ago, has turned out to be most successful, drawing large audiences of people needing an updated within the ever-increasing field of human genetics. Enjoy Milano, the good science and the chance to meet colleagues and get new connections! After all, human genetics is all about people.

Gunnar Houge
Secretary General of the ESHG

Report from the European Board of Medical Genetics - May 2014
by Heather Skirton, Chair of the EBMG

The EBMG was formed in June 2012, with the aim of establishing professional standards of education, training and practice in human and medical genetics and genetic counselling. It is working to achieve this by developing and administering systems of certification and/or re-certification for professionals working as specialists in genetic healthcare in Europe.

The EBMG was formed under the auspices of the ESHG and has been responsible to the ESHG Board and membership. However, as it has become clearer that the EBMG needs to be fully autonomous, it was decided at the ESHG Council meeting in Paris in 2013 that the EBMG should be established as a legal entity separate from the ESHG. This will provide greater legitimacy for the Board and those registered by the Board. This process is now being undertaken, statutes have been agreed and an application for legal sta-
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tus has been submitted in Austria by the Executive Officer of the ESHG. We hope to have confirmation of the success of the application by the time we meet in Milan on 5th June.

While this is an important step forward for the EBMG, we were intent on retaining substantive links with the ESHG. The statutes therefore state that while the EBMG is an independent body for the purposes of setting professional standards and operation of registration processes, to ensure the work of the EBMG remains current and informed by professional practice, the EBMG will be affiliated with the European Society of Human Genetics. For example, the Executive Board of the ESHG will propose three ESHG members to serve on the EBMG Executive, as a tangible way to ensure those links remain strong.

During the year, we have made significant progress in all three divisions.

Genetic Nurse and Genetic Counsellor Division
Co-Chairs: Heather Skirton and Milena Paneque

The registration process was launched at the Paris ESHG meeting, one year ago. We had 31 applications for registration, of these 29 were eligible to proceed with a full application. We received 22 applications and the divisional members met to assess these. As a result, 18 genetic counsellors are registered, a further 4 have been advised that some additional work is required. The website has been updated and we already have a number of enquiries for registration next year. In summary, we feel that the process has been initially successful and the number of applications indicates that registration is valued by genetic nurses and counsellors in Europe.

We congratulate those genetic counsellors who have successfully registered this year. They are:

- Nina Bosch (Spain)
- Fanny Coron (France)
- Irene Ferroce (Italy)
- Anniken Hamang (Norway)
- Debby Lambert (Ireland)
- Anita Matadeen (UK, NL)
- Ramona Moldovan (Romania)
- Christine Patch (UK)
- Heather Skirton (UK)
- Irene Valenzuela (Spain)
- Margareta van Mourik (UK)
- Alana Ward (Ireland)
- Cecile Zordan (France)

In addition, the following counsellors have been provisionally registered pending minor additional information: Elin Eriksen (Norway), Emmanuelle Haquet (France), Barbara Girerd (France), Laetitia Monteil (France), Audrey Vichier (France).

We have also assessed and approved several courses as suitable for training of genetic counsellors. These are Master’s degree in Genetic Counselling courses offered at the following universities: University-Aix, Marseilles II, France; University of Porto, Portugal; Babes-Bolyai University, Romania; University of Barcelona, Spain; Cardiff University, UK; Manchester University, UK.

Medical Geneticist Division
Chair: Ulf Kristoffersson

A great deal of progress has been made with respect to the Medical Division. This Division is different to the other two, as medical genetics is already acknowledged as a specialty in Europe. It was decided that the aims of the division would best be achieved by forming a section of the UEMS that would work alongside the Medical division of the EBMG.

On November 28, 2013, a board for the new Section for Clinical Genetics of UEMS has been elected. Ulf Kristoffersson was elected president by the 13 countries represented around the table, while Kristiina Aitomäki (Finland) was elected secretary and Helen Kingston (UK) treasurer. Vice presidents are Feliciano Ramos, Spain; Bela Melegh, Hungary; Milan Macek, Czech Republic; and Andre Reis Germany. According to the statutes of UEMS a junior doctor nominated by the European Junior Doctors association will be associated with the board.

As we now formally are the professional medical organisations voice on training and education in Europe we have to seek formal agreement with other bodies involved in these matters, and it is obvious that the most important is the ESHG and the EBMG. We are seeking to have a strong relationship with ESHG and EBMG and have appointed Milan Macek as our liaison officer to ESHG.

One of the most important issues we have is to present a syllabus for the European curriculum to become a specialist in clinical genetics, and this is also a task for the EBMG-MD section. Report submitted

Clinical Laboratory Geneticist (CLG) Division
Co-Chairs: Thomas Liehr and Isabel Carreira

The Division has been Chaired by Thomas Liehr, however Isabel Carreira has recently agreed to support the Division as a Co-Chair. Members of the CLG Division met in June 2013 in Dublin to discuss and finalise the curriculum for clinical laboratory geneticists. An assessment has been made of 1) those countries where there is a national system for training and registering CLGs, 2) those countries where there is a training program but no registration, and 3) those countries where no structured training programme exists. The results indicate the complexity of the situation in Europe and will form the basis for introduction for the registration system.
Establishment of a European registration system is in progress. Documentation and guidance is currently being produced and we expect that this will be introduced within the next year. Many thanks to Bela Melegh, Jacqueline Schoumans and Isabell Marques Carreira, who attended the Dublin meeting, and to others who contributed comments. However, this is a large task requiring much work and more support is needed. If there are laboratory geneticists who would be keen to be involved in this Divisional work, please do contact Thomas or Isabel.

Finally, my sincere thanks to all those contributing to this work: the Divisional Chairs, members of Divisional working groups and all those who have already supported the system through applications. In addition, this work would not be possible without the huge support from the ESHG Executive Committee and from the Executive Officer, Jerome del Picchia, who has worked very hard on preparing and translating the statutes for the submission for legal status and in maintaining the website.

Heather Skirton
EBMG Chair

Report from the Public and Professional Policy Committee 2013-2014

By Martina Cornel, Chair of the PPPC

After publication of the ESHG Recommendations on whole genome sequencing (WGS) (EJHG 2013;21:580–584) the issues of incidental findings and return of results were discussed in two Science letters (2013;341:958-9 and 2014;343: 968-9). Where possible, whole genome analysis should be restricted to those genome regions linked to the patient’s indications, and wider testing needs to be justified in terms of necessity. Adding additional targets to a diagnostic test would be a violation of this. At the same time, in the case of unsolicited findings, the patient’s right not to know may sometimes have to be secondary to clinical geneticists’ professional responsibilities. The patient may not have foreseen a specific finding and in some cases the physician will have a moral duty to warn close relatives. Pending further debate, a cautious approach continues to be warranted.

The second contribution responded to a suggestion to give the patient access to his raw sequence data. On behalf of PPPC it was argued that actively handing out data that we know are not fully reliable is at odds with the responsibility of health care professionals. Furthermore, the goals of research and health care should not be conflated. Discovery is the goal of research. Patients should not be turned into research subjects without their consent.

Prioritization in genetic testing

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” will appear in the EJHG.

Whole genome sequencing for newborn screening

Together with the HUGO Ethics Committee and the P3G Pediatric Platform the PPPC developed draft recommendations on the use of next generation sequencing for newborn screening. Some predict that the use of this technology will change the current practice of medicine and public health by enabling more accurate, sophisticated and cost-effective genetic testing. Today, many newborn screening programmes worldwide identify treatable conditions, many of which are genetic, including phenylketonuria, using laboratory techniques looking at proteins, metabolites and enzyme activity. Some authors predict that the earliest application of NGS might be in these state newborn screening programmes. The membership of ESHG was invited to comment to the draft recommendations in February 2014. According to the draft, the primary justification for performing neonatal screening should be in the health interests of the child. Sequencing and analysis should target treatable or preventable conditions that usually present in childhood. Publication of the final version is foreseen in the summer of 2014.

Members of the PPPC in 2013-2014 were Pascal Borry, Anne Cambon-Thomsen, Martina Cornel (Chair), Florence Fellmann, Francesca Forzano, Shirley Hodgson, Heidi Howard, Hülya Kayserili, Christine Patch, Borut Peterlin, Dragica Radojkovic, Wolf Rogowski, Maria Soller, Aad Tiben and Lisbeth Tranebjaerg, supported by Carla van El. Emmanuelle Rial-Sebbag was invited as an observer.

Martina Cornel
Chair of the PPPC
A personal welcome by the programme chair
By Brunhilde Wirth, Chair of the SPC

On behalf of the ESHG Program Committee, it is my pleasure to welcome you all to the ESHG and EMPAG conference in Milan. Based on the 2555 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 long-time collaborators and find new ones. Enjoy the meeting and Milan!

Activities of the Scientific Programme Committee

The Scientific Programme Committee (SPC) for 2013-2014 was composed of twenty regular SPC members: Brunhilde Wirth (chair, D), Paul de Bakker (NL), Jeffrey Barrett (GB), Alexis Brice (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 Nystrom (FL), Francesco Palau (E), Anita Rauch (CH), Samueli Ripatti (F), Peter Robinson (D), Joris Veltman (NL), Joris Vermeesch (BE), Xavier Jeunmaitre (F); three local SPC members from Italy: Antonio Amoroso, Lidia Larizza and Marco Seri; and three observers of the ESHG board: Martina Cornel (NL), Gunnar Houge (N) and Karin Writzl (SLO). In addition, Tara Clancy (UK), Francesca Forzano (I) and Elisabetta Razzaboni (I)

The SPC met twice to organize the ESHG conference for Milan 2014: in July 2013 in Milan to decide on the plenary sessions and symposia and in Vienna at the VMA offices in March 2014 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 2555 abstracts have been scored on-line by 3-11 evaluators including 27 SPC members, 22 ESHG board members and 49 external reviewers, who have been proposed by SPC members as experts for the various topics. I would like to thank all reviewers for their fantastic work and commitment. For the first time, the ESHG will specifically acknowledge the contribution of all external reviewers by granting them a discount of 30% on the registration fee.

Based on topics and scores, 138 (15.7%) abstracts submitted with the preference for an oral presentation were selected for the 23 concurrent sessions including one plenary highlight session. To make this possible the meeting has been extended to Saturday morning and a block of 6 additional concurrent sessions has been added. Among the oral presenters, 57 were Young Investigator Candidates (at
least 1 in almost every session), reflecting the high level of contribution by young scientists to this program. From the 1524 poster presenters, 39 Young Investigator Candidates were selected for the best poster award and will be marked with an ESHG Rosette. 661 abstracts with a score <2.70 will be “published only” and 113 abstracts were rejections either due to very low quality (score < 2.0) or multiple submissions of the same first author.

After the Milan conference, the SPC shall have to say goodbye to Alexis Brice and Jeffrey Barrett. We thank them for their work and their dedication to making the meeting better.

**2014 Meeting Highlights**

After the opening ceremony, the meeting will start with the first plenary session including three local stars: Marco Tartaglia, Elena Cattaneo and Luigi Naldini. The “What’s new?” session will follow with the most exciting new findings selected from submitted abstracts.

The meeting will include 22 concurrent symposia addressing topics of new insights and challenges from next generation sequencing in gene discovery, new mechanisms underlying human disease, functional studies and underlying pathomechanisms 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.

Four “educational sessions” will start already before the opening session on Saturday morning and more educational sessions will continue throughout the meeting. These include: The platelets planet: from diagnosis to therapy of inherited thrombocytopenias; Genetic prediction scores in common diseases: are they of any value? What’s new in Next Generation Sequencing? DNA repair and genomic instability; Mosaicism in human disease; How to be successful in rare disease gene identification; From mutation identification to therapy; and Current developments in legal aspects of genetics: Untangling the law and what it means for you.

On Tuesday, a first initiative to build bridges between ESHG and ASHG will start with an interactive debate on finding global agreements on topical issues. “What IF... (Incidental Findings)”, chaired by Han Brunner with Angus Clarke (UK), Martina Cornel (NL), Robert Green (US), Stephen Kingsmore (US), Marjolijn Kriek (NL) and Arnold Munnich (F) will discuss the challenges of NGS diagnostics and how the human genetic societies cope with them.

The meeting will conclude with our distinguished speaker of the Mendel lecture Mario Capecchi (USA) who will talk about “Gene Targeting into the 21st Century: Mouse Models of Human Diseases from Cancer to Neuropsychiatric Disorders”. This year, the ESHG award 2014 will be conferred to Michael Stratton from Sanger Center, UK, in recognition of his groundbreaking work on understanding cancer genetics.

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

Prof. Dr. Brunhilde Wirth
2014 Scientific Committee Program Chair
Institute of Human Genetics
University of Cologne, Germany
European EQA schemes [CEQA - Cytogenetics, CF Network - Molecular Genetics, EMQN - Molecular Genetics and ERNDIM - Biochemical Genetics] and reviews their annual management reports. The Quality Committee is concerned that there are still some genetic EQAs where poor performance is >10% of the participants. Consequently the GSQC organised a training workshop to assist laboratories with the use of international nomenclature and the interpretation of the results of genetic testing. A workshop on the interpretation of diagnostic genetic results (biochemical, cytogenetic and molecular genetics) was held as a satellite meeting at the ESHG conference in June 2013. More than 80 participants attended the training workshop. Feedback showed that the participants found the workshop helpful and would like to do it again. Due to the success of the workshop, it is anticipated that a similar workshop will be held again at the ESHG conference.

The GSQC organised a half-day satellite meeting and workshop on External Quality Assessment of Genetic Counselling during the Paris ESHG 2013 Conference in June 2013. The meeting provided feedback on the survey results, discussed the quality assurance systems currently available and discussed how to progress towards the establishment of EQA of Genetic Counselling. Fifteen countries have expressed a willingness to participate in the pilot EQA which is due to take place in 2014.

The GSQC organized a further satellite meeting at the ESHG conference in June 2013, to discuss ‘How to Reach European Consensus on reporting Unsolicited Findings and Unknown Variation.’ This was a closed meeting with invited experts to reach a consensus about key recommendations on whether, what, to whom, and how much genomic information should be disclosed to participants (NGS research), patients (NGS clinical testing), their families and/or their referring physicians. There were 18 participants, representing 12 countries. The first part of the meeting focused on defining IF (incidental finding) and UV (unknown variation) and the second part focused on how to reach a consensus on reporting IF or UV in the context of clinical NGS sequencing. A meeting report has been written and is due to be published in the near future.

Help us to help you further by letting us know of any quality issues that need addressing. Please submit any suggestions to the Chair of the Quality Committee Ros.Hastings@ouh.nhs.uk.

Committee Members: David Barton; Mireille Claustres; ElsDequeker; Brian Fowler; Ros Hastings (Chair); Jane Hehir-Kwa, Viktor Kozich; Konstantin Miller; Cor Oosterwijk; Borut Peterlin; Conny van Ravenswaaij-Arts and Uwe Zimmerman.

A synopsis of the meetings are also available on the ESHG website.

**EJHG Highlights of 2013**

*By Gert Jan van Ommen, Editor in chief, EJHG*

- In the last few years EJHG’s Impact Factor is consolidating: 4.38 in 2011, 4.4 in 2012, and 4.32 in 2013. Our current position in the Genetics and Heredity category is 34/161, and 71/290 amongst Biochemistry and Molecular Biology.

- The Clinical Utility Gene Cards, a joint activity with EuroGentest, edited by Professor Joerg Schmidtke and coordinated by Dr Anna Dierking, keep being a popular item, both with readers and submitters. In 2013, 11 new CUGC were published and 9 updates of previously published ones, and both science and services move on.

- In 2013 we published two extra issues as a supplement. One is a new ESHG recommendation: Whole genome sequencing in health care: Recommendations of the European Society of Human Genetics (EJHG 21-S1, June 2013). The other is a fully revised update of a previous joint publication of ESHG and ESHRE: Current issues in medically assisted reproduction and genetics in Europe: Research, clinical practice, ethics, legal issues and policy (EJHG 21-S2, November 2013).

- An important impact predictor turns out to be the ‘immediacy index’ 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 “The C9ORF72 expansion mutation is a common cause of ALS+/−FTD in Europe and has a single founder” by Smith et al. and published in EJHG of January 2013, is a strong candidate for next year’s award: it received 13 citations already by December that year.

**Copy flow**

In 2013, we have been hit by a major surge of submissions: 871, against 776 in 2012, an increase of 12%. Still, our decision time for manuscripts rejected without review is short: 10-12 days. The average time to first decision for manuscripts one out for review is 59 days (the median is 31 days). Our acceptance rate has remained constant at about 35%. Of the 65% rejected papers, as much as 51% are rejected without review. For aspiring authors, the good news is here that of the manuscripts going out for review ~70% are accepted. So once you are under review, you have a better than 2/3 chance to make it – i.e. pay good attention to the reviewers’ suggestions!
Section Editors

To address the surge of submissions and spread the editorship work over more people, we keep looking for experienced mid-career scientists and clinicians interested in advancing their field and their society, at the cost of a little community service (and with the benefit of keeping abreast of new developments.). Last year we welcomed Thierry Frebourg (FR) to join us as SE for Cancer Genetics. IF YOU HAVE AN INTEREST COME SEE US AT THE BOOTH.

Nomenclature and database policy

Starting 2013, EJHG decided to adapt its editorial policy and join the ranks of Genetics Journals requiring mutation nomenclature compliance and deposition of the data presented in manuscripts in publicly accessible databases. As of then, our instructions to authors reflect these more stringent policies, which we expect to increasingly become common policy in biomedical sciences of manuscripts to be published. With the advent of Next Generation Sequencing, basic, translational and clinical scientists in the biomedical field are confronted with a data deluge of unprecedented proportions. Indeed, more information is often obtained than one knows how to interpret. Thus, journals need to become more stringent in nomenclature and public annotation, to counteract ambiguity and degradation of the quality of high precision data and stimulate interoperability. We are subjecting manuscripts not only to the typical peer review for content and quality, but also assessed on compliance with HGVS nomenclature and accessibility of the data through public databases. Further, and in reply to a frequent first question of authors, we should clarify that making data available on institutional or personally-held websites is not sufficient. Not only does this preclude broad variant-based queries, but in under five years, 40% of such urls are lost or no longer contain the data. Hence, we require data deposition in stable, commonly used databases, like HGMD, LOVD and dbSNP. Now, a year later, it has been our experience that while in over 95% of the manuscripts indeed require adaptations of this nature, the large majority of authors are very cooperative and support the rationale of this policy.

EJHG Award

As every year, EJHG, together with Nature Publishing Group, offers a junior authors’ high-citation award. This is given to the top-3 papers published in 2012 and cited in the 12 months following after publication. The 1st prize includes a 500€ award and positions 1-3 receive one year free ESHG membership + online EJHG subscription, and free registration for that year’s or next year’s meeting.

The winners are:

1 “Disease gene identification strategies for exome sequencing”, by Gilissen, C; Hoischen, A; Brunner, HG; Veltman, JA - EJHG 2012 20/5, with 45 citations in the first year.

2 “1000 Genomes-based imputation identifies novel and refined associations for the Wellcome Trust Case Control Consortium phase 1 Data”, by Huang, J; Ellinghaus, D; Franke, A; Howie, B; Li, Y - EJHG 2012 20/7 with 24 citations in the first year.

3 “High prevalence of genetic variants previously associated with LQT syndrome in new exome data” Refsgaard, L; Holst, AG; Sadjadieh, G; Haunso, S; Nielsen, JB; Olesen, MS - EJHG 2012 20/8 with 22 citations in the first year.

ESHG Education Committee Report

By Tayfun Ozcelik, Chair of the EC

The main activity of the Education Committee of ESHG in 2013 has been the organization of the “Seventh Annual European DNA Day Essay Contest for High School Students”. In October, at the annual meeting of the American Society of Human Genetics, representatives from the education committees of ESHG, ASHG and HGSA (Human Genetics Society of Australasia) met to formulate the following DNA-DAY 2014 - ESSAY CONTEST question: Complex traits, such as blood pressure, height, cardiovascular disease, or autism, are the combined result of multiple genes and the environment. For ONE complex human trait of your choosing, identify and explain the contributions of at least one genetic factor AND one environmental factor. How does this interplay lead to a phenotype? Keep in mind that the environment may include nutrition, psychological elements, and other non-genetic factors.

This year 17 European countries submitted 163 essays. The evaluation was made involving 35 experts in the field. And to the delight of the European Society of Human Genetics 4 winners and 13 honourable mentions were selected:

1st Place
Glielmo Finotti,
Teacher: Manuela Granella
IIS Ferrari, Este, Italy.

2nd Place
Rebecca Nunn,
Teacher: Jonathan Avon
The North Halifax Grammar School, Halifax, United Kingdom

3rd Place - ex aequo
Margot C.M. Legal,
Teacher: Paule Bernaud
Lycée Sainte Marie, Cholet, France
EuroGentest integrates with the European Society of Human Genetics

By Gert Matthijs, on behalf of the EuroGentest Team

...to join forces in supporting the genetics community to achieve the highest quality in genetic testing and services!

EuroGentest plays a leading role in promoting quality and harmonization of clinical and laboratory genetic services in Europe. EuroGentest has used the financial support from the European Commission to achieve this goal. It started as a Network of Excellence, coordinated by Prof. Jean-Jacques Cassiman in 2005. In 2011, EuroGentest became a Coordination Action, still supported by the European Commission. Thus, over nearly a decade, EuroGentest has been able to raise awareness, train and bring people together, issue guidelines and documents, influence policy and build a strong reputation. However, from the standpoint of the European Commission, initiatives such as EuroGentest have to become self-sustainable at the end of the granting period. This sounds fine for research projects and technological endeavors. This is not evident for a network that has generously served the community and offered a lot of free services to geneticists, lab specialists, patients and the public. Hence, new ways to support EuroGentest have to be explored.

Over the years, EuroGentest has always been liaising with the European Society of Human Genetics (ESHG) and their activities became more and more entangled. Actually, most people who played an active role in EuroGentest were active members of the society, and vice versa. Hence, it is obvious that our journey has to continue along those lines. Together, we have greatly benefitted from the support from the European Commission, to improve the quality in genetic testing, and to increase the impact of the genetics community on European and national policies in genetics and rare diseases.

So after the funding by the European Commission for EuroGentest ended in December 2013, it was the logical next step to join forces and to integrate EuroGentest into the ESHG. This integration has become a fact in January 2014! It will allow a continuation of the key activities of EuroGentest regarding quality in genetic testing:

- Organize training for professionals (lab specialists and genetic counselors) including the further exploration of the online E-courses
- Support the development of guidelines
- Provide patients and their family with correct and clear information on genetic testing
- Give expert advise to policy makers regarding genetic testing and provide this information to the public
- Liaise with other organizations and projects, and with the industry, that have an interest in genetic testing.

Our focus will thus remain on helping the genetics laboratories in achieving and maintaining good quality in genetic testing, and to promote the best possible organization of genetic services thereby strengthening the role of the ESHG in the provision of optimal clinical genetic services.

Additionally EuroGentest wants to create a network of genetic laboratories and institutes. The aim is to be able to represent the genetic services, and together with the ESHG, be able to lobby and defend the interest of the clinical and laboratory geneticists and genetic counsellors. More detailed information on this will be available on due time.

The EuroGentest website will remain available and will be updated at regular times with useful information on our activities, news and events: www.eurogentest.org. Additionally we plan to organize an annual workshop on quality in genetic testing prior to the ESHG annual conference. The first workshop is jointly organized with ESHG and with 3-Gb-Test, a European project on whole genome diagnostics. The workshop will take place May 30th, 2014 in Milan (Italy).

We look forward to see you in Milan. And we sincerely hope that your laboratory and institute will join the EuroGentest network, as soon as it will be founded.

Gert Matthijs
On behalf of the EuroGentest team
2014 marks the year after a record attendance at the European Human Genetics Conference in Paris. Our congress has now grown into what can seriously be called a “big meeting” with over 3,200 active participants and 150 exhibiting companies, making the ESHG conference the second biggest Human Genetics meeting in the world.

While we are now facing the problem of a continuously decreasing number of venues that will satisfy our needs and provide a setting where compromises on our “preferred way” can be kept at a minimum, we also see a growing responsibility towards resources, our environment and sustainable conference material. It felt timely to change a few items that we have been using over the past years, with the challenge to keep a high standard for our delegates.

Your 2014 conference bag is made from all natural “fair-trade” cotton and juco, manufactured by a South African non-for-profit association (www.township.co.za), hand-made by women from autonomous worker-owned sewing co-operatives in the townships of Cape Town, supporting around 50 women and their families. We would like to thank our bag sponsor, for having gone the “extra mile” by raising an additional amount and making it possible to introduce these bags in 2014.

Your pen is made from recycled PET-bottles, your bag- and lunch vouchers are now laminated and can be re-used at the next meetings.

In general, we have taken care to make a number of items reusable for future meetings without you probably even noticing it.

The ESHG stopped printing an abstract book 3 years ago. This 500+ pages publication totalised around 5-6 tons of ink and paper each year. The online programme planner as well as the conference app, which we continue to improve over the years, have been adopted by our participants. The convenience in using these tools as preparation ahead of the meeting is a quasi standard today, however the fringe benefits of reducing the number of produced items seems less obvious, but definitely contributes to the whole.

On a smaller scale, our new registration and online payment systems allow us to save several thousand printouts every year. Using online surveys and onsite registration systems instead of paper contributes as well.

While the “paperless conference” is not yet a reality, we have been able to reduce the amount of paper produced for our meeting by over 50% compared to 4-5 years ago, money the ESHG can now spend on other conference budget items.

Becoming the Executive Officer of the ESHG has moved me further away from the front line organising team of the conference, Ms. Mirjam Uebelhör and Ms. Kristina Libova, whom I wish to thank for their excellent work in the last years. I stay connected via the “science part”, dealing with SPC matters, their meetings and the abstract management, which always was my favourite task as a Professional Congress Organiser. So I am witnessing first hand the tremendous work done by the SPC in shaping the science of the meeting. The number of sessions and abstracts has almost tripled in the past 14 years and so has the work involved. The growing number of attendees makes me believe that we are moving in the right direction, not least thanks to a number of outstanding SPC chairs in the last decade.

As you could notice, 2014 also marks a change in the schedule of the meeting. The opening day (Saturday) already starts at 10.30 hrs with educational sessions and workshops and concludes with a series of concurrent sessions from submitted papers. We hope that by reducing the number of parallel sessions while increasing the number of talks will contribute to your experience.

As you will have noted, the exhibition is open already on Saturday morning, and closes on Monday evening.

Provided the new schedule proves successful, it will be continued at the next meeting in Glasgow in 2015. On a 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.

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, or as @eshgsociety would say today: #eshg15 rocks in #Glasgow.

I look forward to seeing you at the SECC in 2015. Until then, enjoy Milan 2014!
Functional Genomics and Systems Biology
Hinxton, Cambridge, June 18-27, 2014
www.wellcome.ac.uk/Education-resources/Courses-and-conferences/Advanced-Courses-and-Scientific-Conferences/Advanced-Courses/WTX026850.htm

9th Biennial International 22Q11.2 Deletion Syndrome Meeting
Mallorca, Balearic Islands, Spain, June 19-22, 2014
www.22q11mallorca.com/

A Spectrum of Perspectives: Native Peoples and Genetic Research
Washington, DC, USA, June 23, 2014

Alzheimer’s Disease: Biomarker Discovery and Assay Development
London, UK, Monday, 23 June 2014
www.alzheimersdiseasecongress2014.com

Analysis of Next Generation Sequence Data Course For Complex and Mendelian Traits
Berlin, Germany, June 23-27, 2014

UPCP 2014 – The 3rd International Congress on Personalized Medicine
Prague, Czech Republic, June 26-29, 2014
http://2014.upcp.org/

8th Annual Skeletal Dysplasia Diagnosis Course
Lausanne, Switzerland, July 7-11, 2014
www.skeldys.org

Biostatistics-2014
Baltimore, MD, USA, July 14-16, 2014

NGS Workshop Sheffield 2014
Sheffield, UK, July 16, 2014
conference.biotexcel.com/ngs-workshop-2014/

Human Genome Analysis: Genetic Analysis of Multifactorial Diseases
Hinxton, Cambridge, July 23-29, 2014
www.wellcome.ac.uk/Education-resources/Courses-and-conferences/Advanced-Courses-and-Scientific-Conferences/Advanced-Courses/WTX026851.htm

International Clinical Cardiovascular Genetics Conference 2014
Brisbane Convention and Exhibition Centre, August 6-9, 2014
www.iccc2014.com/

Leena Peltonen school of Human Genomics
Cambridge, UK, August 17-21, 2014
https://registration.hinxton.wellcome.ac.uk/display_info.asp?id=414

55th Annual Short Course on Medical and Experimental Mammalian Genetics
Bar Harbor, ME, United States of America, July 20- August 2, 2014
http://courses.jax.org/2014/55th-short-course.html

FEBS-EMBO 2014
Palais des Congrès de Paris - France, August 30 - September 4, 2014
www.fesb-embo2014.org/

Respiratory Genetics 2014
Nottingham, United Kingdom, September 3-4, 2014
www.nottingham.ac.uk/respiratorygenetics2014

25th European Dysmorphology Meeting
Le Bischenberg, France, September 10-12, 2014
www.eurodysmorph.org

2014 Golden Helix Summer School - “Pharmacogenomics and Genomic Medicine - Bridging research and the clinic”
Aegina Island, Greece, September 11-15, 2014
www.goldenhelix.org/index.php/education/golden-helix-conferences/
Invitation to the

Annual Membership Meeting 2014

At the EUROPEAN HUMAN GENETICS CONFERENCE 2014

Sunday, June 1, 2014 at 7.00 – 8.00 p.m.
Room “Amber 3/4”
MiCo - Milano Congressi, Gate 2 - South Wing, Viale Eginardo, 20149 Milan, Italy

AGENDA

1. Opening by the President of the Society, Professor Han Brunner
4. Discharge of the Board Members for the year 2013-2014
5. Opening by the new President of the Society, Professor Helena Kääriäinen
6. Results of election for President-Elect
7. Results of election for Board Members
8. Membership fees 2015
9. Site of future European Human Genetics Conferences
10. Budget proposal 2015
11. Policy questions proposed by Board
12. Future activities

Please find the minutes of the last membership meeting in Paris 2014 in the restricted area: https://www.eshg.org/members.0.html