

Presidential Address

by Stanislas Lyonnet, President of the ESHG

Gen-Y

“Each generation has its own unique genetic make-up and Gen-Y is no exception”. I came across this statement from the CEO of the Institute of Leadership and Management, and it prompted me to ask myself: what is Gen-Y (generation Y)? The first thoughts that sprung to mind were a lobby of over-achieving males, or a group of evolutionary geneticists worrying about the selection pressure reducing the size of the Y chromosome and its gene number, predicting the catastrophic scenario of its disappearance 1.5 million years from now and crying: “Darwin! Look what you’ve done!” Nothing of the sort. Neither was it a club of people asking never-ending questions, nor a next-next-next generation sequencing procedure.



I was eventually taught that, more simply, it is our residents, interns, post-docs, PhD students, and other postgraduate students that make up Gen-Y. The rationale is actually fairly trivial: they belong to generation Y because they follow generation X, the classical baby-boomers, who currently head our Departments and services, run the labs, and in short, are Gen-Y’s bosses.

And by the way, the idea that generations themselves might have their own genetic make-up - despite not contradicting Mendel’s laws - could lead one to consider gene-environment interactions and, even more challengingly, some generalised epigenetic mechanism at work.

The phenotype of Gen-Y syndrome is defined by consistent features: a strong desire, or even a need to be continually connected, the usage of its own language, the treatment of authority with a good handful of scepticism, gaining enjoyment by embracing a variety of subcultures, being both expressively and digitally creative, having a craving for instant information, reading by skimming, and being easily bored. Well, if so, I am afraid that all geneticists, whatever their ages, belong to the Y generation!

Anyway, what Gen-X might wish to say to the Gen-Y is that they are living in a great era for genetics. So many concepts, novel or rediscovered, and so many open questions to which we would love to know the answers: is heritability really missing, or are we missing the point? How might our genomes ‘get personal’? What impact will next generation sequencing have in medicine and public health? What will remain of GWASes: visionary steps towards facilitating the deconstruction of complex modes of inheritance and individual risk assessment, or the mere conveyors of high P-values combined with useless low relative risk? Is epistasis a biologically addressable question or just a nice word? And, above all, what will remain of genetic science and practice once all rare disease genes and most of the variants involved in common disorders have been identified?

Gen-Y is indeed witnessing an exciting time in genetics: the histone code is being unravelled, we are entering a brave new RNA world, and we are shedding light upon non-coding genomic matter which will not remain dark for much longer. We are using patients’ cancer genomes in order to develop novel treatments, and human genetic variation is now the overwhelming rule rather than the exception.

One more thing Gen-X might wish to say to their Gen-Y protégés is that, besides research, genetics has not always been so fun, especially for Gen-W, who had to remain alert to the possibility of the misuse of their science. And things can change very rapidly in either direction. For instance, our society had to react energetically last year, to the unethical intention to use genetics in order to stir up hatred and divisions. As is often the case, the science behind this attempt suffered from basic flaws; but this is no reason to “baisser la garde”.

Perhaps those risks can be lessened if the ‘human’ in Human Genetics is remembered and respected; this might be our real “Treasury of human inheritance”. Our European heritage is thriving, and yet equally, it demands of us our utmost attention, in order to ensure that our genetics remains “human”. That effort, particularly visible in the organisation and scientific programmes of our conferences, was recently acknowledged by a nice editorial in *Nature Genetics* (“Can we all just get along?” *Nat Genet* 2012, 44, 833) stating: “We commend the organizers of this year’s ESHG meeting for anticipating the integrated future of genomic medicine and for organizing their meeting so as to showcase the usefulness of each strand of genetic research to the others”. This is largely due to the long-standing contribution of one of our former presidents, Pr. Dian DONNAI, and our colleague Jill CLAYTON-SMITH, in organising the now world famous “Unknown” diagnoses ses-

sions. It also confirms our role in paving the road for future successes in genetics, and in maintaining high standards of professional integrity for genetics as a both a scientific field and as a medical specialty. By being open to discussion and keen to question, this also explains why, from time to time, the ESHG might not necessarily align with our contemporary societies. This is the case regarding the recommendation issued by our Public and Professional Policy Committee (PPPC) regarding clinical exome and genome sequences, as what the ESHG proposes is in some ways a more prudent approach than the one suggested by the American College of Medical Genetics and Genomics, and I recommend reading about this in our recent EJHG issue.

Therefore, in order to guarantee that within genetics, we strive not only for goals in science but also for goals for mankind, and, furthermore, to avoid the situation whereby geneticists become merely remote, old-fashioned manipulators of concepts, I would risk suggesting the following:

- i) Whenever possible, we should regroup and integrate services in all dimensions: for example, clinical genetics and other services may be included alongside genomic centres, thus taking into account the various multidisciplinary components of genetic services, including counsellors and genetics specialist nurses. Even if each centre does not unify all of these dimensions in equal parts, it is important to strive for an appropriate balance.
- ii) To respect and reinforce the continuum between clinical work and the biological components of genetics: chromosomal and molecular genetics, epidemiology and, now, genomics. The essence of our discipline's diversity is exemplified by the bench-to-bedside continuum, and should prompt centres to search for, and utilise complementary expertises.
- iii) To be part of therapeutic projects and programmes as often as possible, thus remaining congruent with other academic disciplines; we should open doors, rather than be protective of whatever knowledge we possess, of discoveries that we make, or of research fields in which we are involved. Genetics cannot be disentangled from its natural partners, in particular; neurology, paediatrics, oncology, cardiology, and internal medicine.
- iv) To teach, educate, and travel as often as possible. The alliance between hospital and research activity is a great asset that enriches the training of students and residents.

I hope that if Gen-Y are ever feeling frustrated by the progress of their research, by having so many forms to fill in, so many consents to gain, and by the dialogue between themselves and ethics committees, which can sometimes reach the point of unethically slowing down research, the points that I mentioned might help them to continue to fight for our 'raison d'être': to help and safeguard the wellbeing of patients suffer-

ing from genetic diseases, and their families. I also hope that this might help to convince us that the deluge of "big data" and NGS analyses should increase the impact of genetics, rather than act as a swansong at the end of a golden era.

The organisation of ESHG has become very professional, and thanks are due to the following people: our general secretary, Gunnar HOUGE, our former and now deputy general secretary, Helena KÄÄRIÄINEN, our executive officer, Jerome DEL PICCHIA, and his crew in Vienna, the Vienna Medical Academy (VMA) – because of these people, a number of steps have been undertaken to consolidate the structure of the Society, which will allow it to continue to contribute to the vibrancy of genetics, as a research field, as well as its clinical, laboratory and health systems correlates.

These actions include the establishment of the European Board of Medical Genetics (EBMG), created from the remaining ad hoc committees. In line with the recognition of medical genetics as a European speciality and the drafting of a European curriculum, we can now envisage the use of the EBMG to offer certification to our members - clinical geneticists, and to counsellors as well.

Along these lines, because traditionally - and nearly universally - several professional components comprise medical genetics, Heather SKIRTON, chair of the EBMG after Jörg SCHMIDTKE, our last president and founder of EBMG, created a new European registration system for genetic nurses and counsellors. This is a very significant step for our society, and we hope that the general awareness of that new system among the European genetics community will guarantee its success.

We have also decided to facilitate a possible grouping of EuroGentest activities under the umbrella of the Society. This is quite an endeavour and, whilst we have to be cautious regarding possible investments in personnel, it is quite obvious that having EuroGentest under our auspices would strengthen the visibility of the Society in the field of laboratory genetics.

With such orientation, I believe that our society will reach a good political balance. Diversity and international representation is present in the many facets of our society: it originates from our membership, yet is also present within the society board, within the executive committee acting under the control of the board for any matter of interest, and our four historical committees. Furthermore, the Society will provide and make use of two multidisciplinary arms: the leadership of professionals, undertaken by the EBMG, which facilitates the work of clinical geneticists, counsellors, nurses and laboratory geneticists, and, on the biological side, the society will help the accreditation and quality control of our laboratories and their investigations (hopefully on the EuroGentest model).

Another important move was to open the Society to individuals or groups of individuals from non-European countries.

In these times of great economical, and therefore societal tension, we felt that this was an important move that would grant access to several developing countries that might consider genetics as unreachable. Now, the affiliate membership status has been abandoned, and non-Europeans can hold regular ESHG membership as well as board positions, albeit in a limited proportion. We feel that this might keep the doors open to a number of countries looking at us with both interest and sometimes envy, in particular those in the Mediterranean and Middle East areas. Yet of course, this step should not divert attention from populations here in Europe, who are themselves underserved as far as genetics services are concerned.

Besides the structural changes implemented last year, I would like to endorse the importance of our education and training programmes that acted so often as the spark that lit Gen-X's passion for genetics. For various reasons, mostly related to the financial crisis across Europe, the European School of Medical Genetics needed strong support from the Society in order to survive. We felt that this exceptional situation, needed not only understanding and financial effort, but was also a remarkable opportunity to reaffirm the interest of the Society in endorsing and supporting at least two courses run by the School. As part of its role in education, the ESHG will therefore increase its participation in providing courses in genetic medicine: courses that are based in Europe, are internationally recognised, with a high-standard and widespread faculty, on a yearly basis, with an excellent academic level, that evolve as fast as our discipline does. At least, two courses have been maintained: the European Course of Medical Genetics and a Genetic Counselling course. Supporting a third, possibly in human genomics, involving the interpretation and translational aspects of next generation sequencing is also an aim of the Society. Our efforts to stabilise and, hopefully, to increase the impact of the European School of Genetic Medicine/ESHG courses will involve an increase in the attribution of fellowships, the construction and evaluation of programmes with ESHG representatives, the advertisement of the courses and, hopefully, their inclusion in the national training cursus of our younger colleagues. Since this focus on education is a long-standing one, we decided with our next president, Professor Han BRUNNER, to work towards these goals over at least two years of presidency, and I am sure that Han will maintain these objectives, and hopefully extend them to other aspects of education and training. I wish him luck not only for that, but also for the successful presidency that we all anticipate.

To promote genetics research, from Europe and elsewhere, and to ensure high-standards in clinical genetics practices, we need a healthy, productive and high-level journal. This is what GertJan VAN OMMEN and his team of section editors has achieved, maintaining the journal in a very high-ranking position in the tough competition of genetics publications. We are very much indebted to him for this great success.

Finally, I would like to thank all of the committees who are so active in the Society, including the ESHG Education Committee, in particular for organising and refreshing the DNA day contest, our treasurer Andrew READ who kindly accepted the challenge of pursuing that great and difficult task, the Genetic Service Quality Committee chaired by Ross HASTINGS, and especially, our very active PPPC, chaired by Martina CORNEL. They all have done a lot of extremely good work, which merits our warmest thanks.

Last but not the least, I warmly welcome you to the 2013 annual conference of the European Society of Human Genetics (ESHG) in Paris!

The number of submitted abstracts reached a record high of approximately 2,700. This might be partly related to our attractive city(!) but, more importantly, it reflects the dynamics and health of our Society. In particular, the number of abstracts from non-European countries has almost doubled this year. Such an important meeting, where we expect at least the number of colleagues present in Amsterdam and Nuremberg (2,500) requires the efforts, commitment and work of highly motivated colleagues. Because science comes first, I warmly thank the scientific programme committee chaired by Brunhilde WIRTH, who proposed a fantastic programme for invited sessions and symposia, as well as selected oral communications covering all fields of genetics. I also acknowledge, with many thanks, Rose international and Vienna Medical Academy for expertly taking care of the commercial, industrial and practical aspects of our meeting, essential elements which we need in order to focus our attention on the translation of scientific progress to the best clinical care.

In addition to his crucial role in keeping our finances stable and wealthy, Pr. Andrew READ also chairs the Annual Meetings Committee and, together with our executive officer, had the great task of deciding upon the plans and locations for our future conferences. It is my great pleasure to confirm that we will meet in Milan in 2014, in Glasgow in 2015, and furthermore, I am happy to announce that we have reached a positive decision regarding our 2016 meeting, which will be held in Istanbul. After so many years of efforts from our Turkish colleagues, and despite a few questions to be resolved, not only Istanbul should be a very appealing destination but, most importantly, Turkey has a large human genetics community who is eager to bring our meeting to their country. This will be an important move for us. One year after that, we will travel to Copenhagen to celebrate the 50th anniversary of the first ESHG meeting, in the location where it was first held in 1967.

Well, with so many colleagues actively promoting genetics and communicating their science whenever possible, across Europe and beyond, I truly believe that we will be able to deliver fantastic promises for generation Z.

Stanislas Lyonnet

Secretary General's Address

by Gunnar Houge, Secretary General of the ESHG

Human genetics is teamwork. The division of our professional branch EBMG into MDs, lab professionals and genetic counsellors reflects this fact. The teamwork nature of our profession makes human genetics one of the most rewarding of all medical disciplines – on a scientific, clinical and personal level. This spring I have spent Down Under on sabbatical in a non-MD research environment on basic genomics, focusing on the RNA world of the genome and genome structure. Such knowledge will surely be needed to solve the challenges of future clinical genetics, especially when the present day disease gene bonanza subsides. Multidisciplinarity and an open mind will characterize the successful. ESHG is a society welcoming multiple professions as members, not limited to the ones with professional ties to EBMG. You just have to have your heart in human genetics. Together we can break borders, both professionally and geographically. Have a good meeting both on the professional and private level in the enchanting city of Paris!



fessional certification process. This is essential to avoid conflict of interest and maintain professional integrity and credibility.

The objectives of the EBMG are to develop systems of certification and/or registration for professionals working in genetic healthcare in Europe. In practice, this means designing, organising and managing schemes for three divisions of practitioners:

- Clinical laboratory geneticists working with laboratory diagnosis of human genetic disorders (Scientist Division, Chair Dr Thomas Liehr)
- Genetic nurses and genetic counsellors (Counsellor Division, Co-Chairs Dr Milena Paneque and Prof. Heather Skirton).
- Clinical geneticists (Medical Division, Chair Prof. Ulf Kristoffersson).

Following the formation of the EBMG in June, 2012, Chairs were appointed to lead each divisional committee and to recruit a group of core members to their respective committees. The EBMG is guided by a Steering Group, comprising the Chair, the three Divisional Chairs and two representatives of the ESHG Board (in the first year, Joerg Schmidtke and Helena Kaariainen).

Terms of reference were prepared and presented to the ESHG Exec Board for approval. Board organisational structure and objectives are all available in full on the EBMG webpage of the ESHG website [<https://www.eshg.org/413.0.html>].

The Steering Group meets monthly via webinar to report on progress and to plan future activity. These meetings also enable the Chairs to be fully aware of developments and progress in the other Divisions and to support each other. The activities undertaken in each of the three Divisions are reported below.

Scientist Division

The European core curriculum for clinical laboratory geneticists had been developed by the previous Ad Hoc Committee. An extensive survey has now been conducted to investigate training and education of scientists, including the extent to which the specialisation schemes for scientists in each European country working in Clinical Genetics Diagnostics conform to the European core curriculum. This involved contacting over 150 individuals in 43 countries and information has been returned by colleagues from 41 countries. The results were varied, with 46% of countries having guidelines for training of Clinical Laboratory Geneticists. However, 42% had no formal guidelines, confirming the need for a European system. Of the 21 national training programmes assessed, the majority indicated 70% or more compliance with the European core curriculum.

A meeting of the Division is planned for July in Dublin. At this meeting a system for forming a European register of Clinical Laboratory Geneticists will be discussed.

European Board of Medical Genetics Report, May 2013

by Heather Skirton, Chair of the EBMG

As many of you will know, during the last annual scientific meeting in Nuremberg the ESHG Board decided to form the European Board of Medical Genetics (EBMG), replacing three previously existing Ad Hoc Accreditation Committees. The overarching aim of the EBMG is to establish common standards of training and practice for professionals involved in providing specialist clinical genetic services to patients and at-risk families in Europe. While the EBMG exists under the auspices of the ESHG and is responsible to the ESHG Board, it operates as an autonomous body with regard to the pro-



Counsellor Division

There has been until now no European approach to the education of genetic counsellors and genetic nurses. Initially, we used a Delphi study to develop a core curriculum (for Master's level courses) for genetic counsellors and genetic nurses (Skirton et al, 2013). Work was undertaken to map the curricula of the seven current Master's programmes in genetic counselling to the European core curriculum. We have up till the present time been able to confirm that four of the courses are consistent with the core curriculum.

The Division also met in February to finalise the European registration process for genetic nurses and counsellors: this is being launched at the 2013 Paris meeting. At present, there is a registration system operating in only one European country, meaning the majority of genetic nurses and counsellors have no formal way of demonstrating their competence. We conducted a short survey of European genetic nurses and counsellors to assess interest in this scheme and have strong indications that it is welcomed by practitioners. As of June 2013, practitioners can apply for European registration (all details and application forms are available on the EBMG website).

Medical Division

In medical genetics we have had a European curriculum for some years, while medical genetics was also recently recognised as a European speciality. This means that once a medical doctor has satisfied the requirements to be a specialist in medical genetics in one country this is recognised in all other EU countries. The only additional requirement for a medical specialist to work in another country is that he or she must register according to the local national regulation. However, there is no formal European organisation responsible for providing quality control on how individual doctors can manage. Further, for continuing professional education in genetics for medical specialist doctors there is a need and the EBMG can fulfil this role.

In order to achieve this, it was felt necessary to establish Clinical Genetics as a section of the Union of Medical Doctors, UEMS. This is an EU umbrella organisation focusing on training and specialisation for medical doctors. As a recognised speciality we were able to apply to form a Section for Clinical Genetics. This process has been followed and our application to form a Clinical Genetics section was accepted on April 20 at the Council meeting of UEMS.

The Council of the section consists of 2 members from each UEMS national medical member organisation. A list of these is available at www.uems.eu. From the Council, a board will be elected. A inaugural meeting to organise the Section will take place during the ESHG Paris conference at 11.00 hrs on Monday June 10. The meeting is open, but only delegates have voting rights.

As for most medical professional specialist organisations

an Education and Training Board is a joint project. The intention is that the EBMG Medical Genetics Division will function in this way, with input from both the EBMG and the UEMS to organise post-certification training for medical geneticists to ensure competence is maintained and to offer a European Board examination.

Conclusions

At the end of our first year, we have made very real steps towards a European system for all three professions working in the field of medical genetics. We await further information regarding the European directive on professional activity, but anticipate that the work done already will facilitate formal establishment of European specialities in genetic counselling and clinical laboratory genetics, when the route to achieve that becomes clear.

Our progress is due to the hard work of the Divisional Chairs and their committee members, and I thank them sincerely for their efforts. I was greatly honoured to be asked to Chair the Board and it has been a privilege to lead such a committed group. Our progress has also been dependant on the very real support provided by Jerome del Picchia and the ESHG Executive Board. We look forward to further progress in the coming year.

Report from the Public and Professional Policy Committee 2012-2013

by Martina Cornel, Chair of the PPPC

After consultation and of the membership and several workshops, in May 2013 the Public and Professional Policy Committee (PPPC) of the ESHG Recommendations on **whole genome sequencing (WGS)** were published, including:



1. Where possible, targeted sequencing or analysis is preferable to avoid unsolicited findings or findings that cannot be interpreted.
2. The use of genome-wide arrays or WGA requires a justification in terms of necessity (the need to solve a clinical problem) and proportionality (the balance of benefits and drawbacks for the patient).
3. A protocol has to be in place to give guidance on the reporting of unsolicited findings. If the detec-

tion of an unsolicited genetic variant is indicative of serious health problems (either in the person tested or his or her close relatives) that allow for treatment or prevention, in principle a health care professional should report such genetic variants.

4. Guidelines for informed consent regarding diagnostic testing need to be developed. Patients' claims to a right not to know do not automatically override professional responsibilities when the patient's own health or that of his or her close relatives are at stake.
5. A sustained effort at genetic education of health care professionals is required at various levels: in primary care and in specialized care.
6. Genetic experts should engage in discussing new developments in genetics and raise public awareness. Enhancing genetic literacy will help to involve wider society in this debate.

Prioritization in genetic testing

The new theme of prioritization in genetic testing was discussed at a joint workshop in Munich, 28-28 November 2012, organized by Wolf Rogowski and colleagues. It will first of all be followed up by publications in the framework of EUROGENEST. Members of PPPC are following up the theme of availability and equal accessibility of genetic testing in Europe.

Private databases vs. sharing information

A different issue was the use of clinical data by Myriad®. On the one hand Myriad® has patents on BRCA genes in the USA. Apart from that, they built a private database with clinical data, to provide better insight in variants of unknown significance (VUS). The business model raises questions, since many clinicians and molecular geneticists share data in open access databases, which would provide companies with a private database the advantage of both. A press release on behalf of ESHG was issued 31 October 2012: "We are very concerned that such important data is being withheld from those who most need it. By not sharing their data on the VUS obtained from individuals undergoing BRCA1/2 testing, where Myriad is the sole commercial provider of a test in the US, geneticists have been unable to develop the up-to-date algorithms that are necessary to best interpret the effects of genetic variants".

Members of the PPPC in 2012-2013 were Pascal Borry, Anne Cambon-Thomsen, Martina Cornel (Chair), Florence Fellmann, Francesca Forzano, Shirley Hodgson, Heidi Howard, Hülya Kayserili, Christine Patch, Borut Peterlin, Wolf Rogowski, Jorge Sequeiros, Maria Soller, Aad Tibben and Lisbeth Tranebjaerg, supported by Carla van El.

Report of the ESHG Scientific Programme Committee

by *Brunhilde Wirth, Chair of the SPC*

A personal welcome from the program chair

On behalf of the ESHG Program Committee I welcome you to the ESHG conference in Paris. Based on the 2700 submitted abstracts, which represents an increase of 35% compared to previous years, we expect the largest attendance at an ESHG meeting ever. Based on a very attractive program I hope that this year the meeting will be a highlight again, but also that you will find sufficient opportunities to meet old collaborators or find new ones, have fruitful discussions and enjoy the meeting overall.

Activities of the Scientific Programme Committee

The Scientific Programme Committee (SPC) for 2012-2013 was composed of seventeen regular SPC members: Brunhilde Wirth (chair, D), Corinne Antignac (F), Paul de Bakker (NL), Jeffrey Barrett (GB), Alexis Brice (F), The-Hung Bui (S), David FitzPatrick (GB), Daniel Grinberg (ES), Genuardi Maurizio (I), Giovanni Neri (I), Minna Nystrom (FL) Francesc Palau (E), Anita Rauch (CH), Peter Robinson (D), Carla Oliveria (P), Joris Veltman (NL), Joris Vermeesch (BE), three local SPC members from France: Helen Dollfus, Dominique Stoppa-Lyonne, Damien Sanlaville and two observers of the ESHG board Martina Cornel (NL) and Gunnar Houge (N).

The SPC met twice to organize the Paris 2013 ESHG conference: in July 2012 in Paris to decide on the plenary sessions and symposia and in Vienna at the VMA offices in March 2013 to select the abstracts for oral presentations and posters.

For the first time the SPC restructured all topics taking into account the new development in the field of human genetics. The SPC had the impression that the new system was very well received by the scientist submitting abstracts and for the SPC the selection of abstracts and composition of concurrent sessions was easier and more meaningful. The number of submitted abstracts was 2700. All abstracts have been on-line scored by 3-11 evaluators including SPC members, ESHG board members and 62 external reviewers, 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 commitment.

Based on topics and scores, 126 (4.6%) abstracts were selected for the 20 concurrent sessions and one plenary session (best abstracts). Among the oral presenters, 60 were Young Investigator Candidates (at least 1 in every session), reflecting the high level of contribution of young scientists to this program.

Of the remaining abstracts, 48 were selected as best post-

ers and will be marked with an ESHG Rosette. They are qualified for the poster prize. 535 abstracts with a score <5 will be “published abstracts” and 130 abstracts were rejections either due to bad quality (score < 3.0) or multiple submissions of the same first author.

After the Paris conference, the SPC shall have to say goodbye to Carla Oliveria, Corinne Antignac and The-Hung Bui. We thank them for their work and their dedication to making the meeting better.

2013 Meeting Highlights

The meeting will start with the first plenary session including three local stars: *Edith Heard*, *Alain Prochiantz* and *Jean-Laurent Casanova*. The “What’s new?” session will follow with the most exciting new findings selected from submitted abstracts.

The meeting will include 16 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.

The “educational sessions” throughout the program were very well attended in the past years, so that the committee decided to continue with 9 educational symposia this year. These include: Performance and future of next generation

sequencing (what’s next?); Prenatal and Preimplantation Genetic Screening; Tweeting about Genomics; Cancer risk in developmental syndromes; Epilepsies; Retinal dystrophies: news and views; How to get published in the European Journal of Human Genetics (EJHG); NGS in the clinic, and Where do we come from?

On Tuesday, a third plenary session on “Large Scale Cohorts Studies to Identify Novel Highly Penetrant Genetic Disease Causing Variants” with *Matt Hurler* (UK), *Nicholas Katsanis* (USA) and *Jane Kaye* (UK) will present us the implementation of NGS into the clinic, its use for identification of developmental disorders followed by functional analysis and the ethical challenges.

The meeting will conclude with our distinguished speaker of the Mendel lecture *Huda Zoghbi* (USA) who will talk about Rett syndrome and *MECP2* Disorders: From the Clinic to Genes and Neurobiology. This year, the ESHG award 2013 will be awarded to *Felix Mitelman* from Stockholm, Sweden, in recognition of his groundbreaking work in the development of cancer cytogenetics and development of the Mitelman Database of Chromosome Aberrations and Gene Fusions in Cancer, used everyday by many cytogeneticists worldwide.

I hope that all of you will enjoy the meeting in Paris

Prof. Dr. Brunhilde Wirth
2013 Scientific Committee Program Chair
Institute of Human Genetics
University of Cologne, Germany



Report of the ESHG Genetic Services Quality Committee

by Ros Hastings, Chair of the GSQC

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



The Genetic Services Quality Committee (GSQC) meets biannually. 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

The application of the new genome-wide genetic technologies, whole genome arrays and whole exome/genome sequencing has had an impact on genetic laboratories (Cytogenetic and Molecular Genetics) and clinical/medical genetic services. The GSQC together with the PPPC has published a consultation document on 'Whole genome sequencing and analysis and the challenges for health care professionals: recommendations of the European Society of Human Genetics'.

The GSQC has also prepared reporting guidelines giving 'recommendations for reporting results of diagnostic genetic testing' applicable for all diagnostic genetic laboratories (Biochemical, Cytogenetic and Molecular Genetics). These recommendations are due to be published soon.

The following guidelines have also been prepared by other

collaborating groups. These are now available for:

Specific Constitutional Cytogenetic guidelines (a supplement to the General Cytogenetic guidelines) and the Cytogenetic analysis of acquired disorders guidelines (available at <http://www.e-c-a.eu/EN/>)

Molecular Genetics: Osteogenesis Imperfecta and Huntington's Disease (available at www.emqn.org/emqn/Best+Practice).

The GSQC provides a governance structure for the four European EQA schemes [CEQA - Cytogenetics, CF Network - Molecular Genetics, EMQN - Molecular Genetics and ERNDIM - Biochemical Genetics] and reviews their annual management reports. The GSQC has formed a working group (in collaboration with Eurogentest 2) to discuss unsatisfactory performance identified by the EQA schemes in genetic testing.

The GSQC recognizes there is a need for training workshops 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) will be held on 7th June as a satellite ESHG meeting. Laboratories participating in External Quality Assessment have been alerted to this meeting through the European EQA providers.

The GSQC has completed the survey on Quality Assessment in Genetic Counselling. The survey included questions on whether there are any National QA Schemes for genetic counselling, the nature of these schemes if they exist, whether there is perceived need for European Genetic Counselling EQA and whether Clinical Geneticists are willing to participate in a pilot study. A half day satellite meeting and workshop on External Quality Assessment of Genetic Counselling will be held during the Paris ESHG 2013 Conference on Saturday 8th June from 9am until 12.45. The meeting will give feedback on the survey results, discuss quality assurance systems currently available and discuss a way forward.

Finally, the GSQC is organizing a third satellite meeting on Tuesday 11th June to discuss 'How to Reach European Consensus on reporting Unsolicited Findings and Unknown Variation.' This is 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.

Henry Ford is quoted as saying 'Quality means doing it right when no one is looking' and 'You can't build a reputation on what you are going to do'. Help us to help you 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.

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

Report of the EJHG Editor in chief

By Gertjan van Ommen, Editor in chief, EJHG

New nomenclature and database policy

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. While in the past genetic data often were imprecise due to modest resolution of the technology, today the reverse is almost true. DNA data have become precise to the base pair. Indeed, more information is often obtained than one knows how to interpret. In this situation, the least which Journals, databases and other data sources can do to counteract confusion, is to be more stringent in nomenclature, in order to avoid ambiguity and degradation of the quality of high precision data. EJHG has decided to adapt its editorial policy, changing it from stimulating authors to follow existing nomenclature, to actively assess and sanction the nomenclature compliance of manuscripts to be published.



A second field where genetics journals may assist, is in the improvement of the availability of annotation. It is well known that much data does not make it into the published literature, but also much of the data which does make it into the literature, is not deposited in databases, while very suitable databases do exist. In time this leads to loss of information or of the traceability thereof. To address this issue, EJHG will also assess the data presented in manuscripts for their deposition in the appropriate databases. As of the April 2013 issue the instructions to authors will reflect these more stringent policies which we expect to increasingly become common policy in biomedical sciences.

EJHG Highlights of 2012

- Last year, EJHG saw a quite spectacular increase in Impact Factor, from 3.56 to 4.38 (22 %!), causing us to rise 15 places in the ranking, from 45/144 to 30/156. This position was consolidated in 2012, with a slight increase to IF 4.4 (while most comparable journals showed a small decrease).
- The Clinical Utility Gene Cards, a joint activity with Eurogentest, edited by Professor Joerg Schmidtke and run on a daily basis by Dr Anna Dierking, are another strong asset of , with 25 CUGC published in 2012. The

abstracts are published in the journal (in blocks of three to a page), and the full data are published online. We have already published the first updates.

- Increasing attention is paid to 'immediacy': citations gained in the same year of publication. Typically, papers published in January and February win hands down. Indeed, this year, this top 10 is headed by 'Identification and functional analysis of novel THAP1 mutations' by Lohmann et al. in the February issue, gaining 8 citations. However there are two exceptions: the Gilissen et al. review on exome sequencing strategies in March 2012, with 7 citations, and Borry et al.: the policy paper on 'Legislation on DTC testing in seven European countries' in July 2012, already gaining 6 citations.

Another key component of our portfolio, the Practical Genetics items, remains very popular, with 5 places in our top 10 most accessed online papers. But strikingly, this year even 3 articles figure in this top 10, and # 1 is the review on exome sequencing strategies by Gilissen et al. mentioned above.

- The 'Open Access' format introduced in 2010, by which authors pay a market-conform advance fee for making their paper freely available, has doubled to 23 open access papers in 2012 (11 last year).

Section editors sought (still!).

Our campaign, last year, for more section editors has paid off: Yanick Crow (UK, Clinical Genetics), Eva Klopocki (DE, Clinical Genetics), Christine Patch (UK, Genetic Services), Peter Robinson (DE, Molecular genetics/bioinformatics), and Martina Witsch-Baumgartner (OS, Molecular Genetics) have joined our ranks. In the wake of the recruitments, we have reduced the final decision time from 28 to 24 days. Still we hope to spread the work over more people, so we are still 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..). Notably the field of cancer genetics could use extra hands. IF YOU HAVE AN INTEREST COME SEE US AT THE BOOTH

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 2010 and cited in the 12 months following after publication. The 1st prize includes a € 500 award and places 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:

1st prize: "Genome-wide association study confirms extant PD risk loci among the Dutch", Simon-Sanchez, J; van Hilten, JJ; van de Warrenburg *et al.* with 34 citations in the first year.

2nd prize: "von Hippel-Lindau disease: A clinical and scientific review", Maher ER; Neumann, HPH; Richard, S, with 27 citations.

3rd prize: "A major Y-chromosome haplogroup R1b Holocene era founder effect in Central and Western Europe", Myres, NM; Rootsi, S; Lin, AA; *et al.*, with 26 citations.

Report of the Executive Officer

By Jérôme del Picchia, Executive Officer of the ESHG

Evolution: The ESHG Conference App 2.0

It was a leap of faith, when in 2011, the Annual Meetings Committee agreed to let us explore the possibility of producing an "ESHG smartphone and tablet App" which would combine programme as well as abstracts and could (at one stage) be used with convenience, carrying less weight than the printed programme book, despite of now being separated from the abstracts.

Today a multitude of companies offer conference apps, some highly sophisticated, some not, and some just focused on social networking, hence hardly useful as what we would consider a "conference app". At the time, it was the beginning of conference-app developing and we opted for having it custom-made for our meeting, specifically using the resources we already had in terms of abstract handling and data export.

What seemed especially important, and is widely unavailable with commercially available generic programs, is that we are in control of the backend: content updates, push messages, etc. Whenever a room change becomes necessary, this can be corrected in the app and communicated instantly. Also, we planned the realisation in a way, which would make it possible to use the app in the coming years, making only a minimum of amendments and improvements necessary.

Version 1.0 was ambitious and looking back, there are

things we would have solved differently today. Given not only the feedback of the more than 40% of participants who downloaded the app eventually, but also looking at other conference apps that came out after ESHG's, who were solving quite a few things the way we did earlier, we knew that we should continue this path.

So the aim for version 2.0 was to improve visuals, making navigation even more intuitive, and adding new content.

We now have a special focus on exhibitors, which can be browsed alphabetically or by subject in the product index. We have more extensive possibilities of adding items related to the venue and its surroundings.

Of course we kept the "What's on now" button which will take you to the programme items currently "live", the possibility of taking notes, emailing or printing them, of adding talks to your personal agenda, etc...

We have also added an "Amazon-style like-rating", which is intended to show us the relevance of the specific topic to your practice, in view of planning the scientific programme of future ESHG conferences. Obviously we hope for your input. Needless to say that results are totally anonymised, kept confidential both to speakers and general public, as our primary aim is, as I said above, the relevance of the topic, rather than personal likes and dislikes of the presenters.

Where will we go from here?

Whenever I was discussing the future with providers of apps, and more explicitly (scientific) conference apps, developers and conference professionals, we agreed on one point: One day, may be not so far away, a tablet might be fit to replace pretty much everything that is contained in your conference bag.

To some extent tablets can (at least on a temporary basis) replace a laptop already today, when visiting a meeting. Your emails, agenda, internet browser, contacts, camera, social media apps, gaming, boarding passes, train tickets, your favourite newspaper... all there.

On top of combining all these items, which we consider today being some of the most common (necessary) things to have with us when traveling, tablets have the right format to read conference programmes, abstracts, and floor plans. And, let's face it, smart phones, are incredibly versatile and handy, but the screen is just too small to be really useful for serious "conferencing".

So why not, on a medium term (and as soon as we have batteries available, that will not leave us helpless during the second coffee break of the day) seeing the "how-ever-they-might-look-like"-tablets of the future to be also the conference programme, the abstracts, the poster-, floor- and exhibitor plan, lunch vouchers or even badge, all in one?

I really believe that this is a possible future for an item,



which I considered a “nice to look at, but completely superfluous” *zeitgeist* icon not so long ago.

Paper doesn't blush, so you will be able to remind me of this in a few years from now, should this vision of my profession turn out to be completely wrong. Talking about visions of the future, one of the greatest visionaries of our time, Gene Roddenberry, the creator of the Star Trek universe, equipped his starship crews with tablets already decades ahead of Steve Jobs. I am curious to see...

Enjoy the meeting and Paris.

Now in your App Store: ESHG 2013 Congress App

for iOS:



for Android:



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

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. The European School of Genetic Medicine organizes courses in the charming venues of Bertinoro (Italy). These courses began in 1988 with the first course in Medical Genetics of the European School of Genetic Medicine directed by Prof. Victor A. McKusick. For detailed information about the School visit:

www.eurogene.org

Plan to attend the next ESGM-ESHG Courses in Bertinoro:

Course in Eye Genetics:
October 13-16, 2013

27th Course on Medical Genetics:
May 11-15, 2014

3rd Course on Next Generation Sequencing:
May 16-19, 2014

ESHG Fellowships are available!



Participants of the NGS Course 2013



EUROPEAN SOCIETY OF HUMAN GENETICS

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Invitation to the

Annual Membership Meeting 2013

At the EUROPEAN HUMAN GENETICS CONFERENCE 2013

Sunday, June 8, 2013 at 19.00 – 20.00 hrs.

Room "Room Maillot"

Palais des Congrès, 2 Place de la Porte Maillot, 75017 Paris, France

AGENDA

1. Opening by the President of the Society, Professor Stanislas Lyonnet
2. Activity of the Society 2012-2013
3. Financial Report of the Society 2012
4. Discharge of the Board Members for the year 2012-2013
5. Opening by the new President of the Society, Professor Han Brunner
6. Results of election for President-Elect
7. Results of election for Board Members
8. Membership fees 2014
9. Site of future European Human Genetics Conferences
10. Budget proposal 2014
11. Major policy questions proposed by Board
12. Future activities

Please find the minutes of the last membership meeting in Nuremberg 2012 in the restricted area:
<https://www.eshg.org/39.0.html>

**Renew your ESHG membership!
Visit our booth 460 on level 1**