THE EUROPEAN SOCIETY OF HUMAN GENETICS

NEWSLETTER

No. 31 - October 2017

# Presidential Address

***by Christine Patch, President of the ESHG***

Dear colleagues,

It is an enormous privilege to be president of this Society in these exciting (and challenging times). It was an even greater privilege to take up the baton at the occasion of the meeting in Copenhagen which celebrated 50 years since the inception of the European Society for Human Genetics (ESHG). This meeting gave an opportunity to reflect on the past, consider the present and examine the future with all its uncertainties.

The current aims of the Society as stated in its statutes are:

to promote research in basic and applied human and medical genetics, to ensure high standards in clinical practice and to facilitate contacts between all persons who share these aims, particularly those working in Europe. The Society will encourage and seek to integrate research and its translation into clinical benefits and professional and public education in all areas

The strength of the European Society for Human Genetics is in collaboration and the multidisciplinary nature of its members. In the theme of reflecting briefly on the past I take the liberty of commenting on my own history in clinical genetics originally as a genetic nurses and subsequently, as the profession of genetic counselling developed across Europe, as a genetic counsellor.

When I started working in clinical genetics in the late 1980's there were limited genetic tests; cytogeneticists used microscopes to examine karyotypes, PCR had just been developed and there were no molecular tests in routine use apart from family analysis using linked markers and mutation analysis for a small number of known mutations such as deletions in the Duchenne Muscular Dystrophy gene. We also spent a considerable amount of time doing 'back of the envelope' Bayes calculations incorporating information such as the new mutation rate, biochemical assays and family structures.

My work was talking with patients and families and obtaining information from them to assist the clinical geneticist in making a diagnosis and genetic risk assessment. This might be to assess the risk of another child with a condition or their own risk of developing the same condition as a family member, but often with little to offer as an intervention. I spent considerable time liaising with other health professionals, helping the families to manage their genetic

diagnosis. The laboratory scientists, the medical doctors and the nurses, genetic counsellors, psychologists and others worked as teams but there were very few genetic departments and few health professionals were exposed on a regular basis to the reality of families with rare syndromes and inherited disorders. Much of the work was communication and conversation about genetics.

There followed a wave of technological development and gradually most of the Mendelian disorders were first mapped and then molecularly characterised. There were more choices available for patients and families and increased understanding of the aetiology of many of the rare syndromes and disorders that we encountered. However medical genetics was still a small speciality, many clinicians did not see the relevance of it to their practice and treatments for patients and families were still not in sight. The practice of genetic counsellors became more autonomous and more focused on decisions about testing and the choices that may be available.

At this current time we have entered another transformation period driven by technology. It is unclear exactly how genomic medicine will impact on health care, patients and families, but it is clear that transformation is underway. The promise is that the technological advances will mean speedier diagnoses, increased understanding of the biological mechanisms underpinning many genetic disorders, lead to treatments and be a platform for the delivery of personalised medicine. There are well recognised challenges but undoubtedly there will be an expansion of genomics across the whole of health care with many more health professionals incorporating it into their practice.

In order for this new 'genomics conversation' to happen it is imperative that the interdisciplinary nature of the ESHG and its focus on the linkage between research in basic and applied science in the field of human genetics and translation to clinical benefit continues. Professor Olaf Reis in

his address in the newsletter as President of the Society last year noted some of the challenges that will need to be met to realise the promise of these developments. In addition there is the necessity of being an open and inclusive professional Society with an eye to the future, encouraging younger scientists and clinicians to work for the aims of the Society and to continue the progress that has been made.

Alongside the scientific and technical developments we also have witnessed the start of a transformation in patients from recipients of the science and care to active participants in setting the agenda of the development of scientific priorities and the way care is delivered. At a European level Eurordis, an alliance of rare disease patient organisations is a powerful advocate in enabling the patients' voice to be heard and to influence policy. The developments in genomics is today delivering more and speedier genetic diagnoses and in time there will be more treatments developed out of the new knowledge. The integration of the science with the medical applications requires partnership and a conversation with patients and their families.

I have had the privilege over the years to interact with rare disease patient organisations and am struck by their professionalism and understandable passion. I paraphrase sentiments that have been expressed to me in conversations. The evolution of genetics to genomics has been partly based on working with people who did not have a choice about the fact that they have a genetic disorder in their family. The only way in which those who do not have a choice can be given choice is through the concerted effort of professionals applying their knowledge and expertise to creating the opportunity for those who do not have the freedom to choose, to exercise their options to escape from the impact of, what currently are, mostly intractable diseases. This will happen through the application of new knowledge to the resolution of problems for families who otherwise will continue to experience the consequences of these disorders.

In the excitement of the increasing knowledge and technological advances we must not forget that what we will be delivering is complex, difficult to interpret, sometimes uncertain information that has significant consequences for patients, families and their medical teams. A speedier diagnosis, while positive, is the first step on what still may be a long journey of adjustment. We cannot focus just on the diagnosis but must also put our energies into working with patients and families to develop ways of supporting them in integrating that diagnosis into their future lives and improving the choices they or their relatives may face.

As we work together to solve the challenges of interpretation, new understandings of the biological mechanism of genetic disorders, potential pathways for treatment or the ability to offer more choices we must continue to work with patients and their families. We must also continue as the European Society for Human Genetics to consider the implications of genomics for the wider society and most importantly continue to work together collaboratively towards the aims of the ESHG.

In this 51st year of the ESHG, it is necessary to look forward to the future while taking the lessons from the past. I am of course a UK citizen and have been challenged by the uncertainty of developments in the UK relationship to the European Union. The European Society for Human Genetics however is a society of Europe and is inclusive of members linked to all European countries. In these years with the turmoil and pull towards separation rather than integration I reiterate the sentiments of the previous president of the ESHG that we as a professional society should welcome members from across Europe and beyond, we should continue to work towards collaboration with scientists and colleagues across the world and, with mutual respect for colleagues from all disciplines, continue to support the aims of the Society.

The ESHG would not function without the board, numerous committees and working groups. My thanks goes to all of them. I particularly thank Gunnar Houge the current president elect for taking up the baton at the next scientific meeting. I would encourage any of you that wishes to contribute to the work of the Society to explore the website and express an interest. Our next meeting is in Milan, June 16th to 19th, and is jointly with the European Meeting on the Psychosocial Aspects of Genetics (EMPAG) I hope that as many of you as possible are there and are able to meet old friends and colleagues and perhaps make new ones. Whatever happens in the meanwhile I am sure that when we meet in Italy we will be continuing the conversations.

Christine Patch

## EJHG Tube

**European Journal of Human Genetics** invites you to include a video presentation with your submission to the journal as part of our new initiative **EJHG-Tube**. The video presentation should be included as supplementary material and is a unique way for authors to present the information in their paper and further enhance the visibility of their work by sharing on social media. Through this video authors can convey their findings without the constraints of the written word, plus provide a new and enhanced user experience for readers of the journal.

We accept the following files: .mov, .mpg, .mp3 and mp4. Please see EJHG-Tube at http://www.nature.com/ejhg/videos for our current video presentations and also refer to the journal's Guide to Authors for details on how to submit yours.

# Personal Statement of the President-elect

***By Gunnar Houge, President-elect of the ESHG***

I am honoured to be your president-elect. After 9 years of service as ESHG deputy secretary general or secretary general, I know the organisation quite well. I aim to help building ESHG even stronger, especially regarding the organisations role in education and personalized/precision medicine.

One focus will be close collaboration with the ESHG Education Committee and the independent European Board of Medical Genetics (EBMG) to establish an ESHG course portfolio covering all aspects of human genetics, addressing the educational needs of the three branches of EBMG (genetic counsellors/nurses, clinical laboratory geneticists and medical specialists). This will help small countries to achieve the same professional standards as larger countries with more internal resources, and even more important: European courses will form bridges between professionals in Europe. It was such a course that kicked me into European genetics in the late nineties.

Because my education is medical but my background is molecular, I have a strong heart for all the major groups of professionals that regard ESHG as their professional home. ESHG should always encourage teamwork and collaboration, also with other specialties. We should, however, take a leading role in the implementation of precision medicine into everyday clinical work. Otherwise, the potential for doing more harm than good is definitely there. Finally, ESHG should continue to support of the International Federation of Human Genetic Societies, making it into a viable world-wide umbrella for all human genetic societies.

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# Presentation of the new ESHG Officers and Board Members

**Carla Oliveira, Portugal**
**Deputy Secretary General**

When I was five I wanted to be an Astronomer, and at 7 a Medical doctor. In the end I became a Biochemist that wished to be a Scientist. I have worked in Human Genetics more than 20 years, first as a PhD student in Cambridge - UK, then as a Postdoc in Vancouver - Canada, and in 2011, I became Group Leader at Ipatimup and Professor at the Medical Faculty in Porto – Portugal. I am a Consultant for cancer-associated syndromes at Ipatimup Diagnostics and Founder/CEO of a Bioinformatics Start-up Company Bioinf2Bio. Recently, I engaged with the ENR-GENTURIS on "Genetic tumour risk syndromes", where I serve as National Representative for Portugal and expert in Hereditary Diffuse Gastric Cancer.

I dedicated my scientific life to the study of gastric cancer, a disease that afflicts 1 million people/year worldwide, and I am particularly interested in understanding germline mechanisms that cause hereditary forms of this disease. I have been a Founding Member of the International Gastric Cancer Linkage Consortium (IGCLC) since 1999 and have worked with scientists, medical doctors and patients worldwide, aiming to improve diagnosis and management of this disease. I became a Member of the ESHG SPC in 2010 and had a wonderful experience in this Society. I never failed an ESHG annual meeting since then. I wish to thank the opportunity to return to the ESHG - SPC and serve as Deputy Secretary General of ESHG. This is for me a privilege. I have accepted the challenge, first because I trust this Society and its Leaders, then because this is a unique opportunity to be involved and be part of a changing world where the words "Genetics" and "DNA" appear virtually associated with all subjects, from agriculture to artificial intelligence. My dream is to be part a future where knowledge and technology help to prevent genetic diseases.

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### Christian Gilissen, The Netherlands
### Board member

Christian Gilissen studied computer sciences at the Radboud University Nijmegen and obtained his PhD at the department of Human genetics of the Radboud University Medical Hospital in Nijmegen, the Netherlands, on the topic of "Disease gene identification through Next Generation Sequencing". He currently works in the same department as an associate professor in genome bioinformatics on the development and application of bioinformatics methods to improve the interpretation of genome variation. In particular, he has been interested in identifying the genetic causes that underlie intellectual disabilities using exome and genome sequencing data. He holds a part-time position in the division of genome diagnostics where he coordinates the bioinformatics for the analyses of exome and genome sequencing data for patient diagnosis. By serving on the ESHG board I hope to contribute to the better involvement of younger ESHG members to the society.

### Kinga Hadzsiev, Hungary
### Board Member

Kinga Hadzsiev MD, PhD assistant professor, deputy director in the Department of Medical Genetics at the Medical School of University Pécs, Hungary. Graduated at the Medical Faculty to the University of Pécs in 1992. Since then, board examinations have been taken as follows: pediatrics, neonatology, clinical genetics and child neurology. As a vice director she is responsible for the patient care unit in the Department of Medical Genetics to the Pécs University. Being a well-trained expert in management of rare disease patients the department has been designated in 2015 as one of the four National, Rare Disease Expert Center in Hungary. The issue of the PhD thesis was the genotype -phenotype association in rare diseases. Every year more than 200 Hungarian and 300 international /German and English speaking/ students graduate the medical faculty being trained in the institution for Medical Genetics and Rare disease management. Post gradual training is also part of her daily routine work in the Department. The main field of interest is brain development disorders and patients with mental retardation-dysmorphic syndromes.

### Bart Loeys, Blegium
### Board Member

Since 2005, I am working as a clinical geneticist with a strong translational research interest in Belgium and the Netherlands. I have a joined appointment in the Center for Medical Genetics, Antwerp, Belgium and the Department of Human Genetics from the Radboud University Medical Center in Nijmegen, The Netherlands. My main research interest is the genetic basis of cardiovascular disease with strong focus on aneurysmal disorders. I have contributed for many years to the European School of Genetic Medicine and for the last two years, in collaboration with Bill Newman from Manchester, I have organized two ESHG training courses on cardiogenetics. I really enjoy bringing knowledge on human genetics to the next generation of young geneticist and would like to help build a growing ESHG in this direction. I feel a strong ESHG is important with regards to European Policy Making in the era of personalized precision medicine and I would like to contribute to these processes.

### Hild Van ESCH, Belgium
### Board Member

After receiving my medical degree, I combined a training in pediatrics with a PhD in human genetics at KULeuven, Belgium. Subsequently, I went for a post-doc at Institut Cochin in Paris. Since 2003, I work as a clinical geneticist at the Center for Human Genetics in Leuven, where I cover a broad field of medical genetics and patient care, with specific interest in intellectual disability and syndromology. My academic research is mainly focused on the genetics of cognition, with a strong love for the X chromosome.
The advances in genomics technologies in the last decade has revolutionized human genetics and patient care. Individual genome sequencing holds the promise to contribute to better diagnosis, counseling, treatment and eventually prevention of diseases and overall health improvement. I am pleased to contribute towards the goals of the ESHG, especially in the field of clinical genetics and the translation of genomics for the benefit of the patients.

**Reiner A. Veitia, France**
**Board Member**

I have been working on human genetic disorders since my PhD thesis, which I received from Pasteur Institute and Paris-Diderot University, in 1998. I was working then on the molecular genetics of various Disorders of Sex Development. In 2000, I was appointed as a tenured Assistant Professor at Paris-Diderot University and started a project to better understand the genetic bases of ovarian function. In 2005, I was promoted to Full Professor of Genetics in the same University, up to becoming Exceptional Class Professor in 2016. Since 2005, I run a research team focused on the genetics of primary ovarian insufficiency and non-epithelail ovarian malignancies. I have also tried to find mechanistic explanations for genetic dominance. I am a member of Academia Europaea and Corresponding Member of the French National Academy of Medicine. In short, I am an active researcher in human molecular genetics, also involved in scientific publishing, as the Editor-in-Chief of Clinical Genetics. Being a member of the ESHG board is a great personal and scientific experience. I am proud and delighted to be at the service of the ESHG and I hope to efficiently contribute to the achievement of its missions.

# 20 years of Orphanet, the rare disease and orphan drug database

***By Sylvie Maiella, Orphanet***

This year marks the 20th anniversary of Orphanet, the rare disease and orphan drug database. Orphanet (www.orpha.net) is a unique resource, gathering and improving knowledge on rare diseases so as to contribute to the better diagnosis, care and treatment of patients with rare diseases. Orphanet aims to provide high-quality information on rare diseases, and ensure equal access to knowledge for all stakeholders. Orphanet also maintains the Orphanet rare disease nomenclature (ORPHA number), essential in improving the visibility of rare diseases in health and research information systems.

Orphanet was founded by Dr. Ségolène Aymé, former Chair of the ESHG, in France at the INSERM (French National Institute for Health and Medical Research) in 1997. The initiative became a European endeavour from 2000, supported by grants from the European Commission: Orphanet has gradually grown to a Consortium of 40 countries, within Europe and across the globe.

Orphanet works towards meeting three main goals:

1) Improve the visibility of rare diseases in the fields of healthcare and research by maintaining the Orphanet rare disease nomenclature (ORPHA numbers), thus providing a common language to understand each other across the rare disease field. In a global community, we need to understand each other, although we may not speak the same language. A stable nomenclature, cross-referenced with other international terminologies is therefore essential. In order to improve the visibility of rare diseases in information systems, Orphanet has developed, and maintains, a unique, multi-lingual nomenclature of rare diseases, around which the rest of our relational database is structured. Each disease is assigned a unique ORPHA number: integrating this nomenclature in health and research information systems is essential in ensuring that rare diseases are visible and that different system scan work together. This nomenclature is aligned with other terminologies: OMIM, ICD-10, SNOMED-CT, MedDRA, UMLS, MeSH, GARD. This cross-referencing is a key step towards the interoperability of databases.

2) Provide high-quality information on rare diseases and expertise, ensuring equal access to knowledge for all stakeholders, so as to orientate users and actors in the field in the mass of information online. Rare diseases patients are scattered across the globe, as are rare disease experts. Orphanet provides visibility to experts and for patients by providing access to a catalogue of expert services in 40 countries by disease, such as centres of expertise, laboratories and diagnostic tests, patient organisations, research projects and clinical trials. This data promotes networking, tackles isolation and helps foster appropriate referrals. Orphanet draws on the expertise of professionals from across the world to provide scientific data on rare diseases (gene-disease relationships, epidemiology, phenotypic features, functional consequences of diseases, etc.). In addition, Orphanet produces an encyclopaedia of rare diseases, progressively translated into the 7 languages of the database (English, French, Spanish, Italian, German, Dutch, Portuguese) with texts also currently available in Polish, Greek,


*Figure 1 : Screen capture of the new look Orphanet website*

Slovak, Finnish and Russian, freely available online. Orphanet integrates and provides access to high quality information produced around the world, such as clinical practice guidelines and information geared to the general public.

3) Contribute to generating knowledge on rare diseases by piecing together the parts of the puzzle to better understand rare diseases. To develop and curate the scientific data in the Orphanet database, Orphanet works with experts from around the globe, from health care professionals and researchers, to patient representatives and professionals from the medical -social sector. The wealth of data in Orphanet and the way this data is structured allows additional knowledge to be generated, helping to piece together data that at times can resemble pieces of an irresolvable puzzle. Integration of this data adds value and renders it interpretable. Orphanet provides standards for rare disease identification, notably via the Orphanet nomenclature, an essential key for interoperability. Orphanet provides integrated, re-usable data essential for research on the www.orphadata.org platform and as a structured vocabulary for rare diseases, the Orphanet Ontology of Rare Diseases (ORDO). These resources contribute to improving the interoperability of data on rare diseases across the globe and across the fields of health care and research. They are being integrated in several bioinformatics projects and infrastructures around the world in order to improve diagnosis and treatment. Orphanet is committed to networking with partners across the globe in order to help contribute to generating new knowledge on rare diseases.

The integral role played by Orphanet in the research and care spheres has led to its recognition as an IRDiRC Recognised Resource, and integration in the French node of ELIXIR, a European Research Infrastructure Consortium uniting Europe's leading life science organisations. Orphanet and the ORPHA nomenclature are also cited as key resources in every European legislative text on rare diseases and as key measures in many national plans/strategies for rare diseases.


*Figure 2 : OrphaNews newsletter*

To mark the 20th anniversary of this key resource, the Orphanet website (www.orpha.net) is undergoing a complete makeover, starting with the look and feel of the site. The new site is designed be easier to navigate and read, with simplified search options and a responsive design. The Orphanet team hopes that the new format will make the data in Orphanet easier to find for its different audiences. Further evolutions to improve the way data is searched and displayed will follow later in the year. Users' feedback will be sought in the next annual Orphanet satisfaction survey.

The OrphaNews (http://international.orphanews.org/home.html) newsletter has also undergone a facelift, with new search functionalities and an easier to read format. This freely available, twice-monthly electronic newsletter presenting an overview of scientific and political news about rare diseases and orphan drugs. Be sure to subscribe to keep up to date on the latest news in the field (http://international.orphanews.org/subscribe.html).

In addition, Orphanet launched at the start of 2017 the new Orphanet Knowledge Management System (OKM). This interactive website, developed in partnership with the Garvan Institute, within the RD-Action Joint Action on rare diseases, enables rare disease experts to browse the content of scientific data managed by Orphanet and contribute to their quality, consistency and comprehensiveness. This platform will allow rare diseases experts to discuss in order to validate and curate Orphanet data in a transparent and traceable way. This tool will also mobilise the expertise of the European Reference Networks (ERNs) for rare diseases in order to improve the scientific content of Orphanet.

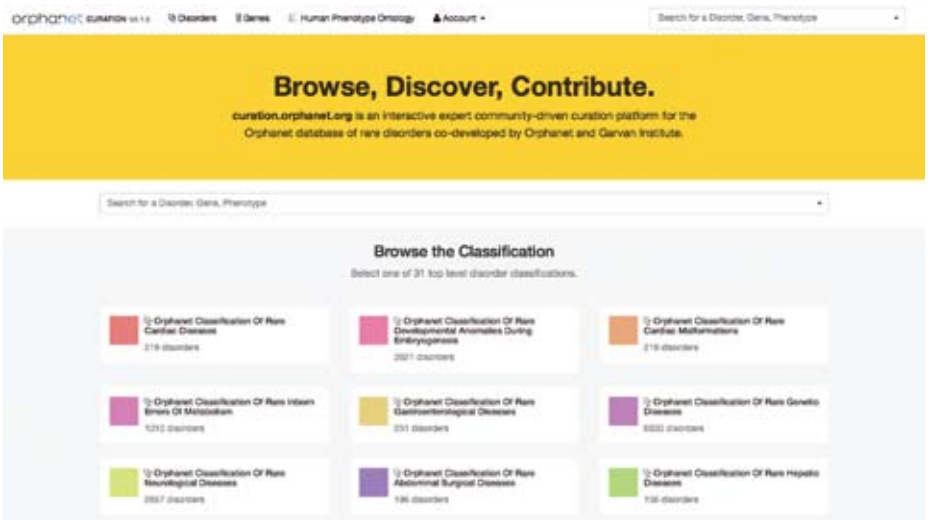



*Figure 3 : Orphanet Management System*

# Student Interviews from the DSMG-ESHG Schoolday in Copenhagen, May 27-30, 2017

***by Mary Rice, ESHG Press Officer***

For the first time, ESHG, in cooperation with the Danish Society of Medical Genetics,arranged for students from a local school to attend the conference. The day's visit was arranged by ESHG Board and SPC member, Zeynep Tümer, and the students were accompanied by Søren Søgard, their teacher of biology, chemistry and biotechnology.
Astrid Hotha le Fevre and Kristoffer Kristiansen attend the Rysensteen Gymnasium (high school) in Copenhagen. They spoke to Mary Rice about their experience.

***How old are you and what are you studying ?***
Kristoffer : I'm 18 and I go to a high school where we have a biotech line.
Astrid : I'm 17 and I'm studying biotechnology and mathematics as well as doing ancient history.

***When did you decide to specialise in this way ?***
Astrid : I've always been inspired to do science because my parents are in science. I think that in the Danish school system choosing science opens up a lot of opportunities. So if you are already interested in the subject, it's a good way to go.
Kristoffer : Much the same for me. My father's a doctor. I think natural science is nice and it's easier than a lot of other subjects – and I like it !

***Is it your first time at a scientific conference ?***
Both: Yes, very first time.

***What did you like about it the most ?***
Astrid: That I felt like I was taken seriously. As a young student, our teacher gives us material to read, we have to study and take notes, but when you meet scientists who work in the field every day they're so into it and they're so knowledgeable that you can ask them anything. Their passion shines through and they explain to you. It's much more specific than what we learn at school and it's amazing to talk to people who are so involved in what they do. You don't really get that at school.

Kristoffer : I like that you get to see all the new stuff, so you get to see the new technology and what's being developed. You can see that it's possible to do this and this, because until it's published it's a bit like fairy tales – you learn at school that it's not possible and then you see these new techniques and realise that it can be. I really enjoyed that side of it.

***What did you like the least ?***
Astrid : There were things I didn't understand, for example the abbreviations, for example the GWAS. They're reminders that these people work with it every day and, being professionals, they understand. For me, if I don't understand something, I have to know. Other than that, I liked everything.
Kristoffer : I didn't understand everything, but many people were good at explaining what they meant and going down to a lower level for our sakes. Some of them were not real people persons so they had trouble communicating and explaining at a lower level, so that was a bit of a drawback.

***What do you want to specialise in ?***
Astrid : I want to specialise in beer ! The fermentation of yeast etc, and insulin in diabetes. So definitely microbiology.

Kristoffer : I'm not sure yet. Maybe I'll be a doctor like my father. I like helping people, and contact with people, and using science in a practical way. I'd like to be a specialist rather than a general doctor, though I'm not sure in which specialism. My dad is a cardiologist, but I'm not sure as yet.

***Has coming here reinforced your desire to go into science ?***
Astrid : Yes, definitely. It's inspired me, because every time I saw a poster I thought : « I've got to do that ! Stem cells, there's a whole world there…… Yeast and brewing…… You know we talk a lot about PCR and DNA and RNA, but it's so important for students to get to experience something real that's relevant to real life, because it inspires you so much.

Kristoffer : You know the theories and you know the techniques used but it's nice to see these being used in real life to actually achieve something, whereas we just learn about it in a theoretical sense. The experiments we do are like, for example, can you taste this ? Yes, you can. But know we can see it genetically and to see it being used in real research where it can solve something that you couldn't just conclude yourself is impressive. And I also like to see the new technology. I'm impressed by how quickly things are going, like CRISPR. I saw an amazing PCR machine, for example.

***How could this day have been improved for you ?***
Astrid : I would like to have done some homework. We were given abstracts for the talks we went to, but I would have preferred a bit more information beforehand so that some of the terms would have been easier to understand. Other than that it's been so cool ! And people have been so nice. People would ask : « What do you do ? » and I'd reply

that I was a student. « Oh, what university ? » and I'd say : «Even younger ! » And they'd reply : « It's amazing that you know so much ». That was a nice reaction, and it's good that people want to talk to you.

Kristoffer: It would have been good if people whose talks we attended knew that we were coming. Of course we wouldn't expect them to totally change what they said, but maybe they could have helped us by talking to us beforehand, or something like that in order to ensure that we understood a bit more. There are terms that we didn't understand, things that we haven't learned, and when a piece of the puzzle is missing it's hard to understand the whole thing. We went to some good talks where we understood because they were more general, but the more specific subjects were quite hard to understand because we didn't have sufficient information.

***And the view from Søren Søgard, their teacher :***
How do you think the students found today ?
I think they found it very interesting. They had a good time seeing different things like the talks, some of which were good for them and some too difficult, and they enjoyed just seeing how a scientific conference works.

***How do you think it could have been done better for them ?***
If we'd known about the visit a little bit earlier we could have planned some kind of homework, but I'm not sure how necessary that is because I think it's interesting for them just to see how it works.

***Would you take them to other scientific conferences is they're invited ?***
Definitely Yes. It's a very good opportunity for them, and also for me.

# ESHG 2017 - Distinguished Speaker Interview: Edith Heard, ESHG Award Lecturer 2017

***Edith Heard is the Head of Genetics and Developmental Biology Unit, Institut Curie and Professor at the Collège de France. She will be giving the ESHG Award lecture on Tuesday, May 30, 2017 at 14:15 hrs. She talked to Mary Rice about her life and work.***

Born into a bilingual Greek/English family, and, now working in Paris, Edith Heard says that she learned to be adaptable at an early age. « I was brought up in central London amidst lots of heated Greek political discussions with members of my mother's family who were staying with us in exile – it was the era of the Greek colonels – while my father was quietly engineering in his garage downstairs.

« So I would often hide and read a book. This meant that I learned to concentrate wherever I was, and also to think about two different topics in two different languages more or less

simultaneously, » she says.

Heard's father was an electrical engineer. « For him science was physics or engineering ; he didn't consider biology to be real science. My mother inspired me to care a lot about how things work, and also about people. » Her interest in biology didn't begin until she was at Cambridge, having previously been more attracted to mathematics. « I realised that I was fascinated by all the unknowns in biology, and the buzz that was there in the mid 1980s when molecular biology, genetics and developmental biology were exploding was very exciting. I was lucky enough to come across many great and inspiring scientists in Cambridge. »

As a post doc, she started working in the field of X-chromosome inactivation. « I did not realise at the time how lucky I was to be working on on such a beautiful biological problem that opened up so many questions and fields. Working on X inactivation means that one can work on development, gene regulation, chromatin, non-coding RNAs, and chromosome biology. It has meant that my team and I have explored many different disciplines and it keeps us curious and happy ! »

Among the discoveries of which Heard is proudest is the insight obtained by looking at early mouse embryos, where the team uncovered the highly dynamic process of X inactivation, with a wave of silencing, followed by reactivation and then silencing again. « This was unexpected, and an unexpected example of in vivo reprogramming. »

Another high point was the discovery of topologically associated domains, or TADs, in collaboration with Job Dekker. « Elephege Nora, a PhD student in our lab, used chromosome conformation capture to explore the X-inaction centre, and we stumbled across these sub-megabase scale domains of chromosome interaction. This totally changed our way of

thinking about the locus we were interested in, and also had many repercussions in terms of our understanding of chromosome structure and gene regulatory landscapes in general. »

Like so many others, the current state of science support worries her. « The perception – and related funding – that biology must always be related to human health in some way, is disturbing. It seems that, in the last decade or so, curiosity-driven research is much less supported than previously – except for the ERC's grants which are a blessing for European research. Often, however, research has to be focused on improving the human condition, or else applicable commercially in order to attract funding. Things will turn around, though – I am sure we will realise (yet again) that it is only curiosity-driven research that can lead to discoveries that will be applicable to human health. »

Even so, Heard feels she is lucky to have ended up as a scientist. « I almost became a musician and I used to think that I would have liked to be a historian. But working in science is a truly fasinating job, and I also enjoy watching the emergence of young scientists and seeing the leaps in understanding that are happening in biology. »

A downside to this fascination, perhaps, is that she has little time for other interests. « I like music and art, and I love to read. Although I have no concrete plans for retirement at present, I don't want it to be too late. And when it comes I would definitely like to do more of these things, as well as to watch my family evolve, write a book, spend more time in the Mediterranean where I have family roots, and maybe try to help the world in some way. Like many people, at present I am watching the news and worrying about our future…….. »

The subject of Heard's lecture will be her lab's work on trying to understand one of the most fundamental questions in biology : how do you shut down genes and how do you turn them back on again in a developmental context using the inactive X chromosome ? « Our work on chromosome organisation has led us to some exciting new avenues and we are now exploring the process of X inactivation in the context of chromosome dynamics. Is chromosome folding into TADs a cause or a consequence of gene activation, and when and how does this happen in a chromosomal context ? »

Answering this key question takes time. Such is her curiousity for further knowledge that it looks as though an early retirement is unlikely to appeal to Edith Heard.

## Report of the Executive Officer

***by Jerome del Picchia, Executive Officer of the ESHG***

It was a pleasure to welcome over 3,200 delegates at the Anniversary Conference in Copenhagen in May for an exceptional week, in terms of scientific content, venue (which seemed to be appreciated by the delegates) and wheater, knowing that just a couple of days before the conference temperatures were more than chilly.

On this occasion we were able to gather 15 past ESHG presidents for a group picture. *FLTR, back: S. Anotonarakis, M. Macek Jr., A. Metspalu, H. Brunner, O. Rieß, Sir J. Burn, S. Lyon-*

*net, F. Ramos, M. Pembrey. Front: V. van Heyningen, J. Schmidtke, C. Patch, D. Donnai, H. Kääriäinen, GJ. van Ommen.*

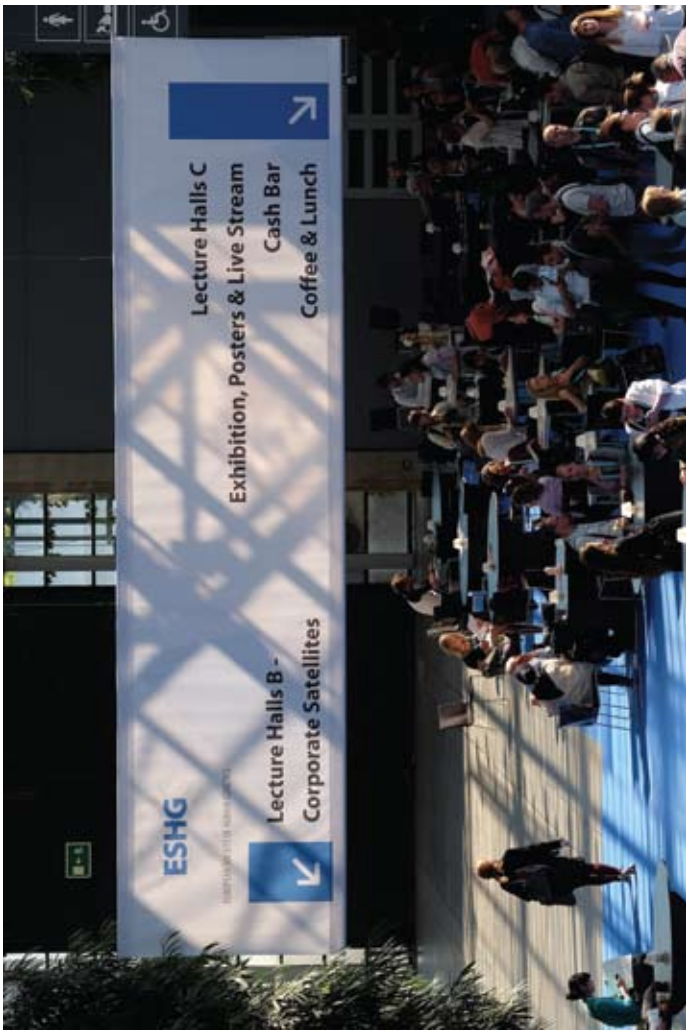
The SPC once again tailored a great scientific programme which gathered around 100 invited speakers over 200 platform presenattions from submitted abstracts and over 1800 Posters (traditional and electronic) in over 180 sessions.

While the Bella Center was able to show that it is a very professional, reliable and attractive venue for a conference, the general price level in Copenhagen has proven to be rather prohibitive for ESHG meetings in the near future, but we might be back for the 100th Anniversary Meeting.

What's new in 2018?

The ESHG 2018 will once again be held as a joint meeting with European Meeting on Psychosocial Aspects of Genetics and will have a number of joint sessions with EMPAG. The ESHG has further fostered a cooperation with other societies by organising joint symposia, among others with the ASHG, the European Society of Neurology and the European Society of Gynecology.

The successful introduction of electronic posters will be continued and expanded in 2018 and the SPC has also scheduled a number of new interactive formats in workshops as well as new "Genomic Quiz" engaging the audience. We are equally proud to announce a workshop on "Career development for Scientific Millenials: How to present - How to networkt - How to enhanec your career" specially focused on young (and not so young) investigators, organised by a media professional, Roy Shepard, a former BBC anchorman on Saturday, June 16.




Some of the highlights will obviously be the Mendel Lecture by Enmanuelle Charpentier and the ESHG Award Lecture by Matthew Hurles on Tuesday, June 19, as well as the Leena Peltonen Award Lecture on Complex Genetics in the Opening Plenary Session on Saturday, June 16.

The website http://2018.eshg.org is now open for abstract submission and registration. You will be able to find the current programme as well as the list of confirmed speakers as well as a number of relevant information.

Remember to reserve your accommodation early, the Milan Men's Fashion Week will take place at the same time, good news the *fashionistas* among you.

We look forward to seeing you in Milan in June!

Jerome del Picchia
Executive Officer of the ESHG

## ESHG Conference Calendar

***More meetings on www.eshg.org/633.0.html***

**November 2017**
**Pre-implantation Genetic Diagnosis: the who, the what, the why and the how**
Guy's Hospital, London, United Kingdom, November 3-4, 2017
http://www.guysandstthomasevents.co.uk/pre-implantation-genetic-diagnosis-who-what-why-how

**EMBL Conference: Cancer Genomics**
Heidelberg, Germany, November 5, 2017
http://www.embl.de/training/events/2017/CAN17-01/index.html

**Variant Effect Prediction Training COURSE**
Prague, Czech Republic, November 6 - 8, 2017
http://vep.variome.org/

**NGS Data Analysis Workshop - Genomic Medicine 2017 Nordic**
Lund, Sweden, November 7, 2017
https://biotexcel.com/event/genomic-medicine-2017-nordic/#workshop

**Genomic Medicine 2017 Nordic Conference**
Lund, Sweden, November 8-9, 2017
https://biotexcel.com/event/genomic-medicine-2017-nordic

**Target Validation using Genomics and Informatics**
Hinxton, Cambridge, UK, December 6-8, 2017
https://coursesandconferences.wellcomegenomecampus.org/events/item.aspx?e=673

**9th Annual Next Generation Sequencing & Clinical Diagnostics Congress**
London UK, November 9-10, 2017
http://www.nextgenerationsequencing-congress.com

# Spring Course in Hereditary Cancer Genetics

**24th – 27th of April 2018**
**University Residential Centre of Bertinoro, Italy**

*This course aims at delivering up-to-date knowledge on hereditary cancer to clinical and molecular geneticists in training or certified.*

*It creates the best opportunity for interaction and discussion with experts from all over Europe, in the fabulous environment of Bertinoro, the headquarters of the ESHG sponsored courses.*

*The faculty combines experts from many fields of cancer genetics known for their didactic skills. Participants are encouraged to present a clinical or genetic case in a Poster format for on-site discussion. Prizes will be awarded for best presentations.*

**Director of the course:** N Hoogerbrugge (NL)

**Organizing Committee:** C Oliveira (PT), H Høberg-Vetti (NO), E Holinski-Feder (DE), together with J Bazzoli (IT) and D Turchetti (IT)

**21** Interactive plenary lectures
**5** Concurrent workshops in small groups
**2** Poster discussion sessions and **2** Quiz sessions

## Teachers and lectures

| | | |
|---|---|---|
| **Prof. Maurizio Genuardi** | Italy | The heritability of cancer |
| **Dr. Marjolijn Ligtenberg** | The Netherlands | The genetic mechanisms of cancer |
| **Prof. Rolf Sijmons** | The Netherlands | NGS in familial cancer genetics: panels, exomes and genomes |
| **Dr. Marc Tischkowitz** | United Kingdom | Hereditary Breast and Ovarian cancer |
| **Dr. Ulf Kristoffersson** | Sweden | Risk assessment and clinical management |
| **Prof. Thierry Frebourg** | France | Li-Fraumeni syndrome |
| **Prof. Gareth Evans** | United Kingdom | Neurocutaneus tumour syndromes |
| **Prof. Eamonn Maher** | United Kingdom | Renal cancer & VHL |
| **Prof. Evelin Schröck** | Germany | Pheochromocytoma/paraganglioma & MEN |
| **Dr. Elke Holinski-Feder** | Germany | Lynch syndrome |
| **Prof. Stefan Aretz** | The Netherlands | Polyposis & other colon diseases |
| **Prof. Shirley Hodgson** | United Kingdom | PTEN and Peutz-Jeghers syndrome |
| **Dr. Ignacio Blanco** | Spain | Moderate risk genes: testing and clinical management |
| **Dr. Hildegunn Høberg-Vetti** | Norway | Familial melanoma & familial pancreatic cancer |
| **Prof. Carla Oliveira** | Portugal | Hereditary diffuse gastric cancer |
| **Dr. Judit Balmaña** | Spain | Germline mutations as a therapeutic target in cancer |
| **Dr. Svetlana Bajalica Lagercrantz** | Sweden | Prophylactic mastectomy & oophorectomy |
| **Prof. Bruce Poppe** | Belgium | Prophylactic colectomy & gastrectomy |
| **Patient representative** | Europe | Prophylactic gastrectomy – a personal experience |
| **Prof. Matthias Kloor** | Germany | Chemoprevention and vaccines for hereditary colorectal cancer |
| **Prof. Jan Lubinski** | Poland | Chemoprevention for hereditary breast cancer |

**REGISTRATION FEE: 750 €**
Tuition, course material, lunches, coffee breaks, dinners, transportation and accommodation in double room
**ESHG FELLOWSHIPS ARE AVAILABLE: Deadline for applying:** 1st February 2018 at 18:00 (CET)

*More information on the full program, registration forms and deadlines, fellowship applications, accomodation and venue, will be soon available at: https://www.eshg.org/courses.0.html and www.ceub.it. For information and contacts: Jessica Bazzoli jbazzoli@ceub.it Tel: +39 0543 446500 Fax: +39 0543 446557.*

ESHG

EUROPEAN SOCIETY OF HUMAN GENETICS

*1967 - 2018 | 51st Meeting*

# EUROPEAN HUMAN GENETICS CONFERENCE 2018

**MiCo | Milan - Italy | June 16 - 19**

## EUROPEAN SOCIETY OF HUMAN GENETICS

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