ESHG

THE EUROPEAN SOCIETY OF HUMAN GENETICS

NEWSLETTER

No. 29 - September 2016

Presidential Address

by Olaf Riess, President of the ESHG

Dear colleagues,

The last 5 years have been particularly exciting for human geneticists and for the thousands of patients who can now receive a definitive diagnosis. Many of these are truly novel as the altered genes were not previously known to be involved in disease pathways. The broad implication of next generation sequencing technology across Europe, starting in research, has now developed as an integrated part of routine diagnostics basically in all European countries. The demands are enormous: besides technology, genome analytics has to be implemented in each human genetic facility, and the implications of dealing with additional or clinically unclear genetic information and its ethical dimension have to be explained to our patients or their relatives. All this is nothing really new for human geneticists, however, as these findings are part of basically each diagnostic report; however, other clinicians are clearly feeling uncertain in the face of this new dimension. The challenges are enormous - but worth while for the benefit of the patients - the consequences are multiple.

One of these consequences is the steady growth of involvement of genome analytics in routine diagnostics. This is of great relevance for our society in several ways. We have to integrate bioinformaticians and people with skills in genome analytics as an integral part of our society and of our society meetings, and we have to adapt our own ESHG training portfolio, but also the general specialist training of human geneticists accordingly.

Another consequence of exome or genome diagnostics will be more complex clinical-genetic reports. Although it is obvious that we need standardized bioinformatic algorithms to evaluate NGS data and standards to harmonize diagnostic reports (e.g. Matthijs et al. 2016), our focus should also be to make reports understandable for other medical specialists, or to train other specialists in reading complex genetic reports. If we want to be integrated basically in each medical specialty, we human geneticists and we as society have to provide teaching courses on „next generation genetics“ at meetings of other societies, and we have to invite other medical disciplines to our meetings. We are encouraging our Scientific Program Committee (thanks to all of you, you are fantastic setting up year after year such a great program!) to make this a strategic discussion for future conferences.

Whole exome or even whole genome sequencing in about fifty percent or more of all patients with presumed genetic condition does not reveal the genetic cause today. First publications to unravel the remaining 50% include RNA sequencing and proteomic studies paving the way towards system medicine. In particularly scientists deciphering the

The next big move based on genetic data and using pathway analysis data will be the intense development of treatment. The tools for large scale and affordable compound screening have been developed. Genetics has to contribute by pathway identification and readout design. Knowledge of genetic modifiers may help in this process for some diseases. Easy access to European research infrastructures and directed European funding could encourage established and young scientists to move into this laborious field.

Dear All, members of the society or „soulmates“,

Human genetics has never been as fascinating as now, but also the challenges have never been as big. Clearly, the Society needs open discussion about where to develop, whom to integrate and how. Human genetics as a medical field is growing rapidly and needs further hands to extend into other fields. We welcome you: experienced and newcomers, human geneticists or other experts in other fields. Human genetics profits from diversity and we need the input of each single one of you to remain an integrated specialty in medicine.

We scientists in Europe welcome the European Union and of course also European research funding. As exciting as genetic research is right now, the political situation is also difficult. Most European countries are on a good way to master the economic crises, however, the stream of refugees and the decision of the British people to leave the European Union will not be without consequences for science and how we used to work together in research. I, as a former citizen of East Germany and now full avowed European, encourage our society to welcome other genetic societies worldwide, in particularly from less developed countries, a process that was already initiated by our former ESHG president, Feliciano J. Ramos. We encourage scientists from developing countries to present their data at our annual meetings and we encourage European scientists to integrate in the scientific discussion and projects. We all have to gain so much from collaborations! And this goes also for the British and European governments for the Brexit negotiations. We will deeply miss the British

expertise in European networks and hope for the sense of proportion for both sides to retain Britain in the global European research infrastructure. Whenever YOU have the chance to discuss science with politicians, please raise awareness that science worldwide will benefit from working together in teams. European research funding is a success story!

In our society, of course, our British colleagues will play a prominent role, most recent evidence is the selection of Christine Patch as the President-elect of the ESHG: Welcome and thanks, Christine, for joining the executive board!

As an open society what we want is integration: of other nations and of other expertise. This includes open discussion on policy of the ESHG, on the focus of our excellent and steadily growing annual conference (please mark your calendar for the next highlight, the 50th anniversary meeting in Copenhagen at the end of May 2017!), on the work of our board and of the numerous committees (thank you all for the great enthusiasm and the time and the effort you put into our society!), but also on future developments of our society. We welcome all of you to participate in the process of shaping our society for the future.

Matthijs et al. Guidelines for diagnostic next-generation sequencing. Eur J Hum Genet 2016; 24:2-5

European genetic research in a post Brexit world

By Sir John Burn, former President and Member of the ESHG

The decision by a majority of British people to leave the European Union sent shock waves across the world, no more so than in European Genetics centres. There had been dire warnings about the impact of a withdrawal on research funding in the UK but these should be seen in the context of a hard fought political battle when both sides were attempting to maximise their position.

Like my fellow Brits and fellow Europeans, I have strong feelings on the subject and have thought long and hard about the reasons for the decision and its implications. This is not the place to speculate on what was and what might have been. The practicality is that we have a Conservative Party in power with a new leader who has pledged to carry out the wishes of the electorate, not least because the members of the Conservative Party are strongly in favour of Brexit. The majority of elected members of parliament and the majority of the members of the House of Lords were in favour of the "remain" camp. There is a remote possibility that a second referendum is called or that there is a snap election which is won by the Labour party who, in coalition with the Scottish National Party, decides to ignore the decision of the first referendum. Both options are in the "flying pigs" file of likelihood. There is a legal battle brewing over whether Article 50 can be actioned without a vote in parliament since the referendum was only advisory. This might slow the process but it is unlikely that it will stop it completely.

There is now a focus on what "Brexit" means in practice. "Hard Brexit" means leaving the single market and imposing strict migration totals. This would impact on EU migrants wishing to settle in the UK though there is almost certainly a need to accept existing residents as there would otherwise be a huge pressure to repatriate the hundreds of thousands of retired British people in Spain and France. Ironically, this argues in favour of any current plans for scientists to move as they would probably end up with "dual status" if a new regulation on movement were to be introduced.

There is intense lobbying to avoid "Hard" and opt for "Soft Brexit" which will mean we stay in the single market. Given that apparently the UK is the biggest European market for expensive Bavarian cars there will be pressure from that quarter to find a solution to the unfettered flow of people in to London and its surrounding districts. One option I have read about is to switch to a free movement of labour which means people must have a job to go to before arriving, something the Swiss have tried I understand. Again, this would not impact significantly on European students and scientists travelling into the UK and vice versa.

Whichever Brexit is adopted, there is a strong case for our continued membership of the European research system. The net value to the UK economy is reported to be greater than the cost of membership and the UK Life Sciences lobby is strong. I believe that EU research would be weakened by loss of UK partners and urge everyone to work to retain and strengthen existing ties.

No decision can or will be taken, however, until the headline position is clearer. The greatest threat now is for a planning blight to inhibit applications from the UK or an undeclared bias to enter into the refereeing process of existing applications. I hope this won't happen and expect a keen eye to be kept on the awards statistics. The fact is that nothing has changed yet and it is up to the whole European scientific and medical community to set an example by continuing to work in harmony as we have done for decades. To quote a current favourite retro poster,"keep calm and carry on".

New Kuwaiti law will severely affect international collaborations

By Martina Cornel, Chair of the ESHG PPPC

After a terrorist attack the Kuwait Government introduced a law mandating compulsory collection of DNA samples from its entire population (citizens, legal, and illegal residents), as well as foreign visitors. The law will also apply to tourists and visiting scientists, and will impose a one year

prison term and a fine on those who refuse to provide samples. This makes Kuwait the first country in the world to legislate for the mandatory collection of human DNA sam-

ples. Several important organisations have opposed this law and called upon the Kuwait government to amend it[1].

The President of the European Society of Human Genetics and the chair of the Professional and Public Policy Committee have sent a letter dated 31 August 2016 to His Highness Sheikh Jaber Mubarak Al-Hamad Al-Sabah and to The Council of Ministers of the State of Kuwait asking them to reconsider this law and to amend it so that human DNA is collected for legal purposes only from individuals suspected of having committed serious crimes. This would be in line with The European Convention on Human Rights, which in ARTICLE 8 on the Right to respect for private and family life states that:

1. Everyone has the right to respect for his private and family life, his home and his correspondence.

2. There shall be no interference by a public authority with the exercise of this right except such as is in accordance with the law and is necessary in a democratic society in the interests of national security, public safety or the economic wellbeing of the country, for the prevention of disorder or crime, for the protection of health or morals, or for the protection of the rights and freedoms of others.

As the professional organisation bringing together human geneticists across Europe we are in agreement with the UN Human Rights Committee that such a law is disproportionate and violates the right to privacy. We understand that the current government is building the collection of human DNA samples for the protection of the Kuwaiti citizens. However, the simple existence of such a resource could be dangerous in the future, for example, if hacked or in the event of a régime change.

Fears voiced in the media include discrimination based on the attribution of ancestry, whether correct or wrongful. While many countries consider that a child's rights are determined by the country of birth, fears are that DNA testing might lead to exclusion of persons not considered belonging to the country.

An additional concern is the potential effect of the compulsory testing of all visitors, including scientists. We believe that this is likely to lead to the isolation of Kuwaiti research institutions, as visiting scientists may refuse to give samples and therefore will not attend valuable scientific conferences in Kuwait, for example. We consider that the global challenges in human health and demography can best be addressed by all industrialised countries in a collaborative way. We see this new law as a major threat to joint actions in the field of genomic health that involve national European genetic societies.

[1]*U.N. rights panel urges Kuwait to amend broad DNA testing law http://www.reuters.com/article/us-kuwait-security-un-idUSKCN0ZV1VY*

GeneWatch UK PR: Expansions of police DNA databases worldwide urgently need human rights safeguards http://www.genewatch.org/article.shtml?als[cid]=566699&als[itemid]=576234

A Report on ESHG 2016 in Barcelona and an Outlook to the 50th Anniversary Meeting in Copenhagen 2017

by Joris Veltman, Chair of the Scientific Programme Committee

#ESHG2016

The 49th annual meeting of our society was organized in the beautiful city of Barcelona, together with the European Meeting on Psychosocial Aspects of Genetics. This location guaranteed a large number of attendees and made it easy to invite top speakers from around the world. More than 3,500 attendees indeed made it to Barcelona, representing a record attendance at ESHG meetings, together with over 160 exhibiting companies and institutions and their over 500 representatives. These attendees and representatives were treated to outstanding presentations by 72 invited speakers as well as 150 selected oral presentations, interacted in workshops and discussed their science at traditional as well as electronic posters, the last one being a novelty this year.

The scientific program committee promoted interactions online by appointing so-called Twitter-chairs at some sessions. While not all attendees may have appreciated this, more than 5000 Tweets were sent at the meeting and #ESHG2016 even became a trending topic online! The scientific program contained too many highlights to mention, including the Mendel lecture by *Adrian Bird*, the ESHG award lecture by Stefan Mundlos and visionary presentations by *Marie-Claire King* and *Jean-Louis Mandel*. Most impressive, however, may have been the contribution of many of our young scientists, the best of which won ESHG presentation and poster awards.

1967-2017 - The 50th Anniversary Meeting

The 2016 Awardees will be invited to co-chair a session at next year's 50th Anniversary meeting of the ESHG in Copenhagen and contribute to making this meeting "the best ever", a worthy challenge! During this next meeting we will celebrate the past, present and future of Human Genetics in Europe and beyond. As teasers we can already announce that the Mendel lecture will be given by *George Church* from Harvard University, an eminent and thought-provoking genomics scientist, not to be missed. In addition, *Edith Heard* will give the ESHG award 2017 lecture which she receives in recognition of her groundbreaking research in the field of epigenetics and developmental biology.

Most important to the success of the meeting and our society, however, will again be your contribution and sparkling attendance, at the many symposia and workshops, poster sessions, the commercial exhibition and last but not least on the dance floor during our famous ESHG party!

So make sure to mark your agenda: ESHG 2017, May 27-30, Copenhagen, Denmark

Abstract deadline: Friday, February 10, 2017

Finally, on behalf of the entire scientific programme committee and staff in Vienna I want to thank *Brunhilde Wirth* for her enormous contribution and leadership in shaping the scientific programme of the ESHG for so many years. Brunhilde, your efficiency is impressive and your enthusiasm is contagious. You will be missed dearly!

Regards

Joris Veltman
Chair ESHG Scientific Programme Committee

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.

Presentation of the new ESHG Board Members

Marta Bertoli, United Kingdom

I'm a Clinical Geneticist working in Newcastle-upon-Tyne, UK. My special interests are dysmorphology and congenital malformations, prenatal diagnosis, muscle diseases and hereditary cancer syndromes. I trained as a Clinical Geneticist in Rome (University of Tor Vergata) and Paris (Hôpital Necker Enfants Malades), and I spent one year of my undergraduate training in Spain (Universidad de Salamanca). Before moving to Newcastle, where I had the opportunity to broaden my experience in muscle diseases and early phase clinical trials, I worked for 5 years as a consultant in Clinical genetics in Rome (Ospedale San Pietro Fatebenefratelli). During my career I have been involved in teaching to undergraduate and postgraduate medical students and to a broad range of other professionals.

I consider ESHG a key player in improving clinical practice standards through basic and translational research promotion and oversight, as well as by educating the public and scientific communities. As a ESHG board member, I intend to apply my European-wide clinical experience and my commitment to the rapidly-evolving field of human genetics, to contribute addressing related ethical and cultural challenges in order to ultimately provide patients with the best, up-to-date, and equitable service possible.

Olaf Bodamer, United States

I view myself at the nexus of Clinical Genetics and Translational Science where "bench meets the bedside" following training as pediatrician and clinical geneticist at prestigious institutions on both sides of the Atlantic. The experience of working with different, sometimes contradictory health care systems has been instrumental in providing visionary leadership to the Division of Pediatric Genetics at the University of Vienna Medical School, to the Austrian Newborn Screening Program, to the newly founded Division of Clinical and Translational Genetics at the University of Miami and more recently as Clinical Chief to the Division of Genetics and Genomics at Boston Children's Hospital/Harvard Medical School. My professional time is divided between providing clinical care to patients with complex genetic disorders and inborn errors of metabolism, translational and clinical research, teaching and administration. My research is centered around utilization of systems biology approaches in connection with multiple OMICs (genome, transcriptome, methylome, microbiome and others) first to study the longitudinal aspects of pregnancy relative to outcome (PMAP, Precision Medicine and Prematurity study) and secondly to identify the molecular underpinnings of selected monogenic disorders relative to therapeutic targets (PMAT, Precision Medicine and Therapies study).

As ESHG board member I look forward to leverage my international experience and expertise to benefit the society's goals at a broader scale. In particular, I am interested in a) promoting applied, translational research through close collaboration between industry, academia, patient advocacy groups and professional societies; b) education of health care professionals and the public on the utility of genomic medicine; c) development of consensus guidelines for best practice in Medical Genetics with regards to diagnostics, management and therapies.

Ellen Heitzer, Austria

After receiving a Biological Sciences degree at the Karl Franzens University of Graz, Austria, I wanted to specialize in medical research and therefore did my PhD at the Department of Dermatology in the field of cancer research. After a two year post-doc at the Division of Oncology, I switched to the Institute of Human Genetics in 2010 and after a 5-year training, I became a European registered Clinical Laboratory Geneticist (ErCLG). Since our institute is part of the Medical University of Graz, Austria, I am involved in teaching and supervising diploma and PhD students, routine diagnostics, and research. With respect to diagnostics, my main focus is hereditary cancer syndromes. In the last few years, I have been actively involved in the implementation and accreditation of Next Generation Sequencing at our institute. As far as research is concerned, I am working on liquid biopsies, i.e. the analysis of cell-free circulating tumor DNA and circulating tumor cells. We have developed a variety of tools that enable tight non-invasive tumor monitoring during the entire course of the disease, which is of utmost interest in the era of personalized medicine.

Since I have always been interested in discussions and decision-making processes that involve my profession, I hope to be able to contribute to furthering the aims of the ESHG. In view of the rapid changes occurring in genomics, it is crucial to establish standards and guidelines as to how clinical practice should proceed when genetic information is involved in order to make a substantial contribution to the community and professional awareness of genomics. In addition, I think that in the era of NGS, genetics comes to play a broader role across all of medicine, and collaborations with and support of clinical geneticists and genetic counsellors is becoming more and more important. This can only happen within the framework of professional associations like the ESHG.

Philipos Patsalis, Cyprus

Professor Philippos Patsalis studied Human Genetics at the City University of New York, USA. He received post-doctoral training at the Cornell Medical Center at New York University as well as at the Memorial-Sloan Kettering Cancer Center and Columbia University in New York, USA.

For the last 25 years, Professor Patsalis worked in the field of human genetics, and has served the community and society as an active academic, researcher, and administrator from different posts. He founded and directed the Cytogenetics and Genomics Department at the Cyprus Institute of Neurology and Genetics (CING) for 10 years, and served as the Chief Executive Director of the Institute for another 10 years pursuing scientific research and providing diagnostic services. He founded the Cyprus School of Molecular Medicine, where he fulfilled two roles as Professor as well as Provost providing postgraduate education and training. Professor Patsalis is currently Distinguished Professor at the Cyprus Institute of Neurology and Genetics and the Cyprus School of Molecular Medicine. He is a founding member and President of the Cyprus Society of Human Genetics. He previously served as an elected board member of the ESHG, and was a member of several National and European committees, boards, and councils. He also had the honour of serving for one and half years as the Minister of Health of the Republic of Cyprus.

Professor Philippos Patsalis has stated that he joined the board to offer his services and support to the ESHG providing his knowledge and experience to promote research and services in human genetics. He recognizes the importance to provide education and training to professionals, the need to develop opinions, guidelines, and policies on key issues in research and society, and has actively contributed towards the development of National and EU regulations on human genetic testing. He is determined and eager to devote his time and energy to work closely with board members of the ESHG to serve scientists, people, and humanity.

Report from the First course basics in human genetic diagnostics – A course for Clinical Laboratory Geneticists (CLGs) in education

Nicosia, Cyprus, June 20-24, 2016
by Hasmik Mkrtchyan, Head of cytogenetic diagnostics laboratory GeneTechnology LLC, Moscow, Russia

The first course "Basics in human genetics diagnostics - A course for CLGs in education" took place in the Cyprus Institute of Neurology and Genetics in Nicosia (CING) and was organized by Drs. Thomas Liehr (Jena, Germany), Leonidas A. Phylactou and Carolina Sismani (both Nicosia, Cyprus). The course was enabled in first place by a generous sponsoring of the European Society of Human Genetics (ESHG), being interested in a sound education of Clinical Laboratory Geneticists (CLGs) in all (European) countries. Also CLGs coming from countries without a national education scheme for CLGs need such courses to be eligible for the European registered CLG title (ErCLG – see https://www.eshg.org/clg.0.html). Besides, the support from Carl-Zeiss Jena, DAKO – an Agilent Technologies Company, Illumina, MetaSystem, MRC-Hollan, Oxford Gene Technology and Cy-

tocell and ZytoVision GmbH and the fees of the overall 30 participants enabled the realization of this five day course. Nonetheless, the financial frame would have been still extremely dense, if CING would not have provided a lecture hall free of charge, and if not all speakers just claimed their expenses and did not ask for being paid. Thanks for all these efforts to all involved parties!

Before talking about the course itself, I would like to thank again CING for hospitality and well organization of everything around the course – main work was done here by Carolina Sismani with support of Leonidas A. Phylactou and his CING team. The course schedule developed together by Thomas Liehr, Leonidas A. Phylactou and Carolina Sismani, was dedicating each of the five days to a special field. Every day was divided to four major parts: two of them were lecturers about theoretical basics of certain topics; one part was a workshop with discussion of possible situation of case analysis using genetic diagnostic tools; each day ended with a written test, where participants were asked to answer to several questions concerning day topics. In 4/5 tests participants needed to have >50% of the available points, which was managed by all of us.

Day 1 was devoted to the basics of human genetics, the history of genetic diagnostics from karyotyping to NGS, and the inheritance patterns. Here, the participants got a chance not only to listen about genetic counselling and syndromology, but also due to a workshop to empathize themselves in the place of counselor and a patient, construct a family pedigree and perform own risk assessments.

Day 2 was devoted to cytogenetics from history to application in diagnostics and research. During the workshop participants were asked to do a karyotype and to pass a quest about ICSN nomenclature.

Day 3 was about molecular cytogenetics from FISH to array-CGH, with introduction to main approaches in these fields and discussion of possible simple and complex cases results. During a workshop on array CGH, case-results were discussed. Lecturers about basics of molecular genetics, knowledge of monogenic syndromes and PCR techniques completed this day.

The presentation of PCR techniques and their possible applications in diagnostics was continued in the day 4. Knowledge about MPLA and related approaches was given to the participants with several case presentations and deep analysis and discussion of the results.

Day 5 was the last one and was very intense. We heard about sequencing techniques, NGS and their application, non-invasive prenatal diagnostics, biochemistry genetics and metabolic disorders, basics of epigenetics and gene therapy, futures of diagnostics using proteomics knowledge. After a very interesting presentation made by Helena Kääriänen about future of human genetics services we all could exchange and discuss about this topic from the view of the different countries being represented by the participants, i.e. Armenia, Bulgaria, Canada, Cyprus, Finland, Greece, Italy, Latvia, Romania, Russia, Sweden, Ukraine, and USA.

Although we had an intense course and tight scheduled of lectures and workshops, There was also always well planned breaks and thus time for inter-participant and lecturer conversations; and this is most important for communication with other scientists and colleagues.

In conclusion I would like to thankful all organizers for this course that provided a comprehensive introduction to genetic diagnostic tools that maybe helpful for beginner CLGs to choice his/her own future specialty and for advanced researchers to refresh knowledge and keep update.

Report on the AEGH-ESHG Schoolday in Barcelona, May 20-21, 2016

by the AEGH Education Committee

A Schoolday Workshop was celebrated in Barcelona, in CAIXA Forum Museum, during the 20th and 21th of May. The event was organized by the Education Committee of the Spanish Association of Human Genetics.

We organized several workshops and informative speeches in order to teach the students about the new progress in genetic techniques and the use of the clinical genetics. Almost one hundred students from Catalonia and a small group from Palma de Mallorca have participated to the event during these two days.

On the first day, the students have attended an informative speech "Genetics in Art, History and in our times" and they could discover the presence of the genetic diseases not only in the hospitals but also in art and history. After that speech they participated in several workshops and learnt about genetic diseases, different diagnostic techniques, inheritance in families, forensic genetic and genomics in the future. They learnt how to identify a murderer, how to view the chromosomes under the microscope and how to make a genealogic tree and genetic counseling.

On the second day, Dr. Manel Esteller gave a lecture about Epigenetics in CAIXA Forum Museum for the students. There was an interesting discussion concerning environmental influence in epigenetics and how epigenetics could change our expectance life. The youngest visitors (children under 5-6 years old) made a double helix DNA with balloons in CAIXA Forum Museum. After the conference, the students were invited to visit the European Congress of Human Genetics. They could talk to the genetic investigators who explained their posters, discussing genetic techniques to approach different diseases and learning about genes, exomes, proteins, expression regulation, bioinformatics, etc. The students asked about the genetics degree, research, laboratory training and employment opportunities in clinical genetics. The students also visited many commercial stands where they could discover the last innovative equipments and enjoyed a lot with all the presents they got from each stand.

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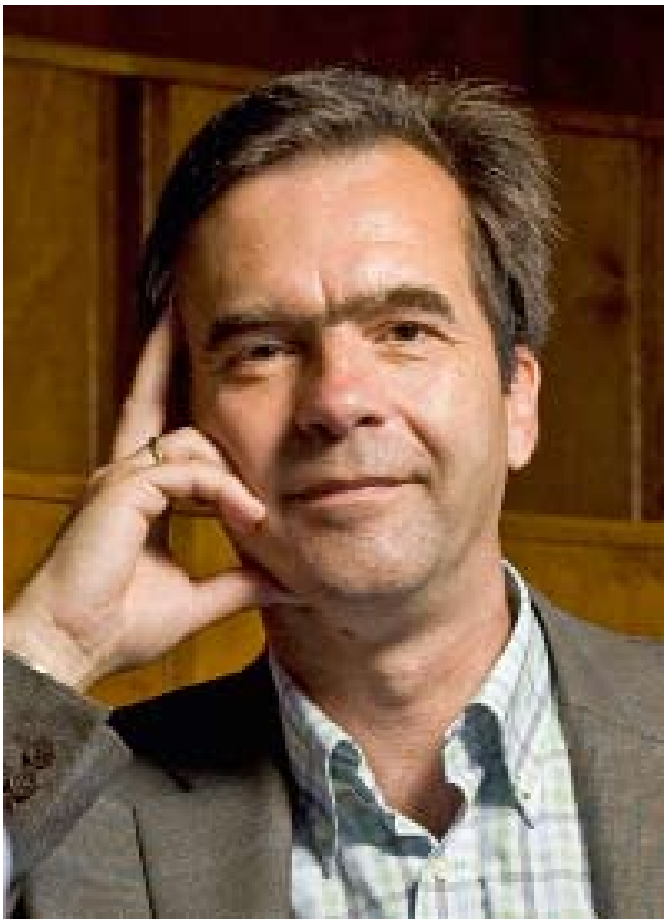
Editors: Karin Writzl, Jerome del Picchia

c/o Vienna Medical Academy, Alser Strasse 4
1090 Vienna, Austria, office@eshg.org, www.eshg.org

ESHG 2016 - Distinguished Speaker Interviews: Stefan Mundlos, ESHG Award Lecturer 2016



Stefan Mundlos is director of the Institute for Medical and Human Genetics at the Charité and group leader at the Max Planck Institute for Molecular Genetics in Berlin, Germany. He will be giving the ESHG Award lecture on Tuesday, May 24, 2016 at 14:15 hrs. He talked to Mary Rice about his life and work.

Stefan Mundlos is clearly happy with what he does. He'll retire when he is forced to, not before, he says. And if you ask him if there's anything he regrets in his career, the only thing he can come up with is not starting his scientific work earlier. "I started my scientific career fairly late, when I was in my early thirties. Although I did some research during my time as a paediatrician, it was relatively little."

Mundlos' interest in science began in the forests and ponds near his home. "As a boy I was always interested in nature and the secrets it holds. I was interested in reptiles, amphibians and fish – I caught them in the ponds in our neighbourhood and kept them and watched how they developed, how they caught prey and so on. I was certainly always very interested in the way nature presents itself. We lived in a suburb of a larger town, but were right next to the forest, and so I spent a lot of time exploring nature there, looking at animals and trying to work out why they behaved the way they did."

This interest led him to medical school in Göttingen, where they had an exchange programme with the University of California leading to a year spent in a San Diego laboratory working on a breath test for pancreatic function in rats. He finished his studies in Marburg and Heidelberg before beginning his training in paediatrics. A specialism in paediatrics holds a particularly happy memory. "When I started my training we had a very sick new-born child with life threatening diarrhoea. Through trial and error, I managed to work out what the problem was. The child had an intolerance of the sugars found in milk [glucose and galactose], and once we removed these and substituted with fructose, she was completely fine. This was an important and very rewarding experience for me, both on a professional and a personal level."

Another highlight, once he had moved into science, was the discovery of the disease gene for cleidocranial dysplasia. "The gene mutated in cleidocranial dysplasia turned out to be a transcription factor essential for bone formation and chondrocyte differentiation," he says. "This finding was a breakthrough in the bone field, once again showing how the identification of rare disease mutations can provide major insights into biology."

More recently, his group has been working on gene regulation and the non-coding genome. "I think that the interpretation of the non-coding genome, which after all makes up more than 98% of our genome, holds one of the major future challenges in human genetics,",he says. The effect of structural variations on gene regulation has been another major interest of his group. A highlight of the more recent work has been the discovery that structural variations (deletions, duplications, inversions) can interfere with the 3D folding of chromatin, thereby influencing gene regulation.

Of course he has concerns for the future. He is not alone in bemoaning the importance attached to high impact publications. "The publication of an important paper in a low impact vs a high impact paper can ruin a career. This is getting out of hand, and it has a big effect, especially for young people."

Outside work, Mundlos still enjoys nature. "I very much like my garden and I enjoy gardening. I grow my own vegetables. I also like to work with wood, and I build furniture. And I listen to a lot of music – being in Berlin, there's a wide variety of classical music and jazz to be enjoyed at concerts.

He'll be talking to the conference about the importance of genome folding. "These are very exciting new developments; how our genome is folded in a three dimensional way and its importance for gene regulation. I want to open eyes to a part of the genome that has not been well studied, the non-coding part of the genome. Most studies have focused on finding the genes, finding mutations in genes, but the big question now is "What is the rest doing?"

It seems that Mundlos' insatiable quest for new knowledge will continue to shed new light for a long time to come on the

way in which changes in the genome cause diseases.

ESHG 2016 - Distinguished Speaker Interviews: Sir Adrian Bird, Mendel Lecturer 2016

Professor Sir Adrian Bird is Buchanan Professor of Genetics at the University of Edinburgh in Scotland. He will be giving the Mendel Lecture on Tuesday, May 24, 2016 at 13:30 hrs. He talked to Mary Rice about his life and work.

DNA has fascinated Adrian Bird since he was in his teens. "Like many children of my generation, I grew up interested in growing frog spawn and watching birds, etc. But it was a black and white TV programme about science that really caught my attention. On Sunday mornings, scientists like John Kendrew or Asher Korner would stand behind a lectern and talk about proteins or DNA. If there were illustrations, I don't remember them – it was just a man giving a lecture aimed at the general public. I have no idea why I started watching those programmes, but they appealed to me."

No-one in Adrian's family had been a scientist before. His father was a public relations officer and journalist, and most of his relatives work in publishing or journalism. "When I retire – and I don't know yet when that will be – I'd quite like to write, though that sounds a bit boringly obvious, like a Miss World candidate saying she wants to travel," he laughs. "But I do have quite strong opinions about certain aspects of science and indeed biology that I would like to write down. Sometimes I think I should take a sabbatical to do that, but I am not sure that I ever will."

His strong opinions look to be well-founded. "My research has focused on DNA methylation and its associated binding factor MeCP2. We found that the MeCP2 protein binds specifically to methylated CpG sites and we now know that disruption of this interaction caused Rett syndrome. At that time Rett syndrome was considered to be a neurodevelopmental disorder, the implication being that the condition was irreversible. We decided to make a mouse model of the syndrome to try to understand things better.

"We then put back the gene in an adult mouse that was terminally ill, and it got better. In other words, we showed that this wasn't strictly a neurodevelopmental disorder – though people still call it that. That was really exciting, because it was completely the opposite of what everyone thought would happen. Even we expected that, once you had this disorder, it would probably be too late to do anything about it. But that turned out to be wrong."

The fact that there is, as yet, no effective treatment for humans with Rett syndrome is frustrating to him. "There's a huge amount of interest in labs around the world in trying to get therapy to work. Our work arguably had an impact beyond

Rett syndrome in the sense that people are now conscious that a neurodevelopmental disorder they are working on might end up being curable. So it's not considered crazy to consider trying to cure autism or other profound neurological disorders, although in most cases reversibility has not been demonstrated.

"For Rett syndrome itself, there are several clinical trials under way now that people realise that you can reverse this condition rather than having to catch it before it starts, but no therapy is yet imminent.

"I'm not a mouse geneticist, even though I'm a professor of genetics – I'm really a biochemist. It's somehow exhilarating that we got into an area where genetics and neuroscience were the dominant approaches and were able to show that some prevailing views were incorrect.

"But working out how MeCP2 works turns out to be rather more difficult to disentangle than we had hoped. It looks as if it affects a lot of genes subtly rather than affecting a few genes in a switch-like manner. The trouble with subtle effects is that they're quite difficult to study. But that's what we're doing and I think things are steadily getting clearer with time."

Although Adrian hopes he'll continue working for a long time to come, he has had some thoughts about retirement. "In addition to writing, I could spend more time cooking – I find it therapeutic. I enjoy watching football, like cycling, and used to run in a hill-running club. Unfortunately I've lost the cartilage in my knee so I can't do that anymore. A regret about the state of science today is that not enough people work on knees! It's particularly disappointing that stem cell scientists haven't yet managed to grow me new knee cartilage."

More serious concerns about the state of science today include the hegemony of certain journals. "I do think that science is pretty fashion-conscious at the moment. I would like to see a literature that more accurately reflects the scientific process, rather than the cross between science and journalism that we sometimes appear to have. I don't want to sound cynical

though, because there's fantastic science going on now, and technologically it's never been better. But it's somewhat frustrating when unfounded studies making flashy claims get published."

He will talk to the conference about his work on Rett syndrome. "We're manipulating the MeCP2 gene to see if the parts that we thought were important really are. I'll also discuss reversibility, because this still needs explaining at the molecular level."

Even though Adrian describes himself as 'no spring chicken', it looks as he'll be intriguing us with his strong opinions for some time yet.

ESHG Conference Calendar

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

EMBO|EMBL Symposium: The Complex Life of mRNA
Heidelberg, Germany, October 5, 2016
www.embo-embl-symposia.org/symposia/2016/EES16-08/

Circulating Biomarkers 2016
Dundee, UK, October 12-13, 2016
https://biotexcel.com/event/circulating-biomarkers-2016/

Computational RNA Biology
Hinxton, Cambridge, UK, October 17-19, 2016
https://coursesandconferences.wellcomegenomecampus.org/events/item.aspx?e=58

Annual Meeting - American Society of Human Genetics 2016
Vancouver, Canada, October 18-22, 2016
http://www.ashg.org/

Chromatin, Replication and Chromosomal Stability 2016
Copenhagen, Denmark, October 19-21, 2016
http://www.abcam.com/events/chromatin-replication-and-chromosomal-stability-conference

Marfan Syndrome and Beyond: An update on aortopathy diagnosis and management
London, UK, October 21, 2016
http://www.vantagepointmedical.com/event

Next Generation Sequencing Bioinformatics
Hinxton, United Kingdom, October 23-29, 2016
http://atnd.it/48418-0

Variant Effect Prediction Training Course
Heraklion, Crete, Greece, October 31 - Nov 3, 2016
http://vep.variome.org/

Epigenomics of Common Diseases
Hinxton, Cambridge, UK, November 1-4, 2016
https://coursesandconferences.wellcomegenomecampus.org/events/item.aspx?e=594

NGS 2016 Nordic
Lund, Sweden, November 8-9, 2016
https://biotexcel.com/event/ngs-2016-nordic/

EMBO Conference: From Functional Genomics to Systems Biology
Heidelberg, Germany, November 12, 2016
www.embl.de/training/events/2016/OMX16-01/index.html

NGS 2016 Benelux
Brussels, Belgium, November 14-17, 2016
https://biotexcel.com/event/ngs-2016-benelux/

3rd Middle East Molecular Biology Congress and Exhibition
November 14-17, 2016 - Doha, Qatar
http://membs.org/membs/congress

18th EMBL PhD Symposium: Life by Numbers: Towards Quantitative Biology
Heidelberg, Germany, November 17, 2016
http://phdsymposium.embl.org/

First Announcement of the second Course: Basics in Human Genetics Diagnostics - A Course for CLGs* in Education

A course Supported by ESHG

Dates: 5 days t.b.d.;
Monday, 8:30 hrs to Friday 18:30 hrs

Location: t.b.d. Athens, Greece

Main organizers: Thomas Liehr (Jena, Germany),
Hellenic Association of Medical Geneticists

Target Audience: *Clinical Laboratory Geneticists

Registration: is planned to be open from 15. January 2017

Costs: EUR 500
(including tea, coffee, 5 lunches and 2 dinners, abstract book and USB-drive);
ESHG fellowships are available to cover tuition and partial travel fees.

Travel and stay: to be organized by the participants themselves

Preliminary Programme:

- Each day starts at 8:30 hrs and finishes around 18:30 hrs;
- Two coffee and one lunch breaks are scheduled each day;
- The afternoons will contain a practical part;
- At the end of each day a written exam will be taken;
- A certificate will be issued at the end of the course (in case of successful participation).

Topics:
- Basics of Human Genetics and Genetic Counselling
- Cytogenetics

- Molecular Cytogenetics and Molecular Genetics I

- Molecular Genetics II

- Molecular Genetics III, Biochemical Genetics and future directions of the field

ESHG

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EUROPEAN HUMAN GENETICS CONFERENCE 2017

Bella Center | Copenhagen - Denmark | May 27 - 30

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