ESHG

THE EUROPEAN SOCIETY OF HUMAN GENETICS

NEWSLETTER

No. 33 - November 2018

Presidential Address

by Gunnar Houge, President of the ESHG

Dear ESHG membership,

It is an honour with obligations to be your president, and hopefully some of the aims stated at the membership meeting in Milan can be achieved before I step down next year. These are exciting days for human genetics with new diagnostic tools and treatment options added to our clinical repertoire. These are also exciting days for ESHG because of the planned transfer of the organisation from Belgium to Austria, as our executive officer, Jerome del Picchia, has informed you about. Hopefully the economical and legal practicalities of this rather complicated transfer will be sorted out before the end of my presidency.

I like the new ESHG webpage, developed by the VMA team after important input from the Board members Isabella Ceccherini and Francesca Forzano. You should all get acquainted with our excellent educational resources, e.g. the conference webcast archive, where many educational talks can be found, and the educational resources overview made by Edward Tobias from Glasgow University (www.EuroGems.org). Our education committee, led by Han Brunner, now has a course portfolio that covers major themes in human genetics, and this can be expanded even further – which is being planned. Our courses get excellent reviews from the participants, which tells me that the people responsible for the different courses are doing things right, and use our fellowship money in the best way possible. We should all be grateful for their efforts! The goal is to make a course portfolio that covers the need for courses that the three branches of the European Board of Medical Genetics may have (www.ebmg.eu).

ESHG has also created a "task force" that will work on how to improve the variant classification system, e.g. how to handle the "VUS problem". The task force has a mixture of PhDs (Hans Scheffer representing Eurogentest, Gert Matthijs representing Eurogentest, Johan den Dunnen representing LOVD and HGVS , Nicole de Leeuw representing molecular and structural genomics) and MDs (Olaf Reiss well connected to ERNs, Helen Firth from DECIPHER and TGMI, and me, representing the ESHG Executive committee). We also think about making a "reference group" to test and comment whatever we should propose. Hopefully we will have a suggestion ready to present at our annual meeting in Gothenburg next June. See you all there!

Gunnar Houge
ESHG President

European Reference Networks

by Dr. Holm Graessner, Coordinator of the European Reference Network for Rare Neurological Diseases

Update on European Reference Networks

Approximately 30 million patients in Europe have one of the ~5.000–8.000 known rare diseases. These patients often do not receive the care they need or they have a substantial delay from diagnosis to treatment. In March 2017, twenty-four European Reference Networks (ERNs) were launched with the aim to improve the care for these patients through cross border healthcare, in a way that the medical knowledge and expertise travels across the borders, rather than the patients and the experts. These networks are composed of healthcare providers that have been approved by the EU member states as rare disease expert centers. More than 300 hospitals and 900 highly specialised teams from 25 European member states are participating in the approved ERNs. It is expected that through the ERNs, European patients with a rare disease get access to expert care more often and more quickly so that existing healthcare inequalities across the EU are leveled, and that research and guideline development will be accelerated resulting in improved diagnostics and therapies.

The current state of rare diseases in Europe

Health systems in the European Union (EU) aim to provide high-quality, cost-effective care. Across Europe there is substantial variation in health care for patients with a rare disease, especially considering prognosis, quality of life, healthcare costs, available guidelines and patient information. For rare and complex diseases, this is a challenge due to the fragmented knowledge about these diseases and missing accessible expertise [2, 3]. Hence, patients with a rare disease often experience severe problems regarding access to care, diagnosis, continuous care and treatment.

A rare disease is defined as a condition that affects few-

er than 1 in 2,000 people. It is estimated that 6–8% of all people someday in their lives will face a rare disease [2]. In the EU there are approximately 30 million people who experience daily suffering from one of the 5000 to 8000 known rare diseases. It has been estimated that 80% of all rare disease have a genetically determined. Given that about 50% of all rare disease patients have not got a confirmed molecular diagnosis yet this means that about 12 million patients with a rare disease do not know the molecular cause of their disease.

The objective of European Reference Networks

European reference networks (ERNs) are virtual networks of medical experts from all over Europe. ERNs are focused on rare or complex diseases that require highly specialized treatments and a combined effort of expert knowledge and resources. The ERNs aim to improve the care of patients with a rare disease [4, 5]. The European Commission Expert Group on Rare Diseases, in collaboration with EURORDIS, a European alliance of rare disease patient organisations, has identified themes in which all rare diseases will be classified. In March 2017, 24 ERNs were appointed to these themes by the Board of Member States of the European Union, which oversees the ERNs [4, 6].

The procedures and criteria for the establishment of an ERN and the selection of members are regulated in EU legislation. To become a member of an ERN, a hospital or healthcare institute needs national endorsement and must meet a number of general requirements set by the European Commission (such as recognition by the national government) and a number of theme-specific criteria for each ERN (such as minimum number of patients and demonstrable facts related to expertise).

Services of European Reference Networks

Mainly, ERNs provide two services that are (i) expert knowledge pooling, generation and dissemination as well as (ii) virtual consultation.

In order to facilitate virtual consultation on complex or rare diseases and conditions on the basis of individual patients, the European Commission has set up a secure e-health platform. The Clinical Patient Management System (CPMS) provides the workflow for the discussion of cases of rare disease patients in multidisciplinary teams. The CPMS has been available for consultations from end of 2017 on.

Pooling, generation and dissemination of expert knowledge for example on care standards plays a central role in the success of the networks and it is of critical importance that structures and processes exist that allow exchanging medical expertise within and between the – cross-border – networks. [7]

The implementation of these services, however, poses a number of issues such as the currently missing integration of ERNs in the national healthcare systems including referral pathways from national healthcare to ERNs, missing standards of cross-border healthcare as well as the lack of sufficient and sustainable funding. A joint effort of ERNs, patient organisations and EU member states is underway to tackle these issues and find workable solutions for them.

Future of European Reference Networks

Based on the structure, expertise and the provided services, ERNs as healthcare infrastructures may have the potential to significantly change the fashion how healthcare for rare disease patients is being provided in the European Union in the near to medium future.

1. Vos JR, Giepmans L, Röhl C, Geverink N, Hoogerbrugge N. (2018) Boosting care and knowledge about hereditary cancer: European Reference Network on Genetic Tumour Risk Syndromes. Fam Cancer. 2018 Oct 9. doi: 10.1007/s10689-018-0110-6. [Epub ahead of print]

Note: Part of the information presented in this article is based on this open access publication.

2. Puiu M, Dan D (2010) Rare diseases, from European resolutions and recommendations to actual measures and strategies. Maedica 5(2):128–131

3. European Union (2009) Publication sheet C151 of the Europese Union. ISSN 1725–24744. European Commission Rare disease policy. https://ec.europa.eu/health/non_communicable_diseases/rare_diseases_en. 5 Oct 2018

5. Eurordis. European Patient Advocacy Groups (ePAGs). http://www.eurordis.org/content/epags. Accessed 5 Oct 2018

6. Wijnen R, Anzelewicz SM, Petersen C, Czauderna P (2017) European Reference Networks: share, care, and cure-future or dream? Eur J Pediatric Surg 27(5):388–394

7. European Commision (2017) European reference networks. Working for patients with rare, low-prevalence and complex disease. https://ec.europa.eu/health/sites/health/files/ern/docs/2017_brochure_en.pdf

8. Heon-Klin V (2017) European Reference networks for rare diseases: what is the conceptual framework? Orphanet journal of rare diseases 12(1):137.

Presentation of the new ESHG Officers and Board Members

Josef Gecz, Australia
Board Member

My career spans three decades of competitive research in genomics and molecular biology of childhood onset neurological disorders across three countries, Slovakia, France and since 1994 in Australia. I am a scientist, Australian National Health and Medical Research Council

Senior Principal Research Fellow and Channel 7 Children's Research Foundation Inaugural Chair for the Prevention of Childhood Disability at the University of Adelaide and South Australian Health and Medical Research Institute. My main expertise is in childhood onset neurodevelopmental disease gene discovery, disease mechanisms and personalised medicine, namely in intellectual disabilities, epilepsies, autisms and cerebral palsies. My professional career was inspired at one of the 1st European Schools of Medical Genetics in Sestri Levante in 1990 and I was only delighted to be Faculty at several of these from 2010.

My research is patient centric and focuses on the application of genomics for precision diagnosis to drive best possible health outcomes for children living with disabilities. I am passionate about equal access to genomic technologies for all children, world-wide.

Aleš Maver, Slovenia
Board member

I have been working in the field of human genetics since 2002 with the primary focus on complex disease genetics research, bioinformatics and diagnostics of monogenic disorders. After obtaining a medical degree in 2011, I worked on the identification of rare genetic variation in familial multiple sclerosis using whole exome sequencing as a part of my PhD work. Since then, I have been directing my efforts towards translating next-generation sequencing into routine health care for patients in Slovenia and neighbouring countries. To achieve this, I participated in the establishment of the Centre for Mendelian Genomics in 2013, which now acts as a hub offering state-of-the-art technology and bioinformatics for diagnosing monogenic conditions and attempts to integrate with worldwide data exchange initiatives. Nowadays, my primary interests include identification of novel genes for monogenic disorders, improving genetic variant interpretation on the genome scale and finally, promoting and facilitating data sharing in my home country and across Europe. I am honoured to have been elected as an ESHG Board Member and will be happy to contribute to the efforts of the Society in the aforementioned fields.

Rebecka Pestoff, Sweden
Board Member

I am very interested in promoting the clinical specialty of human genetics and in particular the genetic counselling profession. I am on the board of the Swedish societies for genetic counsellors (www.sfgv.n.nu) and of human genetics and genomics (www.sfmg.se) and work actively in the field of clinical genetics and genetic counselling, since I achieved my MSc in genetic counselling from Manchester University in 2009. I am also very interested in research in the field of professional development for genetic counsellors and have successfully published several articles. Currently I am also undertaking a PhD, part-time, that focuses on the area of video-consultations in order to improve access and equality of genetic counselling to all patients in need of it.

I am deeply honored to be on the board of the ESHG, and will do my best to fulfill my commitments in the best of ways. I will work toward improvement in the field of clinical genetics and genetic counselling, especially focusing on countries where the profession is still under development.

I am looking forward to learn, meet and work together with the committee toward improving our field of interest to become even better and reach those who can benefit from it.

Sérgio B. Sousa, Portugal
Board Member

Genetics has always fascinated me. In 2009, when I completed my training as a Medical Geneticist, next generation sequencing was tickling our toes and I decided to spend some time doing research. My PhD at UCL Institute of Child Health, London, focused on identifying novel genes for rare unsolved syndromes.

Since 2015, I have been back to clinical work at the Paediatric Hospital of Coimbra. My main interests are intellectual disability syndromes, dysmorphology and skeletal dysplasias. I have also been involved in implementing a local multidisciplinary genomics interpretation team. Additionally, I love to teach and spend a significant portion of my week with medical students and clinical genetics trainees.

Having studied and worked in several countries in Europe, I have a good perception of regional asymmetries and of the value of scientific societies and networks, such as ERNs, in countering these through sharing knowledge and experience. I am honoured to be part of the ESHG board and will do my best to contribute to its goals.

ESHG Newsletter No. 33, November 2018
Editors: Karin Writzl, Jerome del Picchia

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

Birute Tumiene, Lithuania Board Member

Ever increasing pace and spread of genetic revolution and genomics into new areas, as exemplified by recently created European Reference Networks (ERNs), and across Europe, including EU-13 countries, strengthens genetic community and emphasizes its importance in many fields of medicine and research. The need for implementation and usage of genomic achievements is acknowledged at the highest EU political levels, as shown by agendas of European Health Forum, Gastein, or a recently EC-organized Digital Day 2018 with an initiative „Towards access to at least 1 Million Sequenced Genomes in the European Union" signed by 13 Ministers of Health. However, these developments result in increased responsibilities and commitment of genetic community, represented by ESHG in Europe, to disseminate high standards and best practices across countries, specialties and fields, including development of professional competencies, bioethical and data privacy requirements among multiple other relevant issues. By taking responsibilities in the Board of ESHG, I am ready to commit myself to further development of public and professional policies of ESHG and dissemination of these policies towards novel professional communities, as ERNs. Besides, special consideration of historical and cultural specificities of EU-13 countries is of importance for their further successful and wide-spread inclusion into ESHG activities.

Report on the establishment of the specialties of Laboratory and Medical Genetics in Greece

by Aspasia Tsezou, Prof. Medical Genetics, University of Thessaly, Faculty of Medicine, President HAMG

The Hellenic Association of Medical Geneticists (HAMG) is pleased to announce that the long-term efforts for the establishment of the specialties of Laboratory and Medical Genetics in Greece finally came to a successful end.

The reform on all medical specialties was initiated by the Greek Ministry of Health in 2016 and included the formation of 14-member "working groups" from the Academia, Public Health Sector and Private sector for each already established medical specialty as well as for the few ones (Laboratory and Medical Genetics specialties were among them). The task of the "working groups" was to reform or construct (in the case of new specialties) the Educational Program, logbooks, evaluate the Educational Centers, establish rules for the final examination in order to attain each specialty. After a long period of meetings and consultations with a Special Council of Ministry of Health we managed to construct the 5-year educational program and all the other requirements for the Laboratory Genetics specialty.

Finally, the decision of the Minister of Health regarding the establishment of the 2 Genetics specialties (Laboratory Genetics" and "Medical Genetics") was published in the Official Journal (20th September 2018, # 4138).

Regarding the specialty of Laboratory Genetics, a Degree of Biology or Medicine is required and the duration of training is 5 years (48 months in Laboratory Genetics and 12 months in Medical Genetics).

We are confident that the establishment of the 2 new Genetics specialties, which will closely collaborate, will significantly contribute to the improvement of all genetics services in Greece.

Distinguished Speaker Interview with Matthew Hurles, ESHG Award Lecturer 2018

by Mary Rice, ESHG Press Officer

When Matthew Hurles isn't in his lab trying to identify rare genetic diseases, he may sometimes be found cycling up a mountain. He's clearly someone who likes a challenge.

His interest in science started in his mid-teenage years. The books of Stephen Jay Gould sparked an interest in evolution and started him thinking about the role of science in deciphering it. Encouraged by his mother, a biochemistry teacher, he studied biochemistry at Oxford. "The course was general and the genetics component very slight, but for me that was the most interesting part. So for longer projects, both as a undergraduate and for my PHD, I sought out things that were in that evolutionary space."

His interest in the genetics of evolution led naturally to his current work in the understanding of human genetic variation, its clinical impact, and how the understanding of the genetic causes of rare disorders and their biological mechanisms can provide insights into human development.

For the last seven years he has led the UK's Deciphering Developmental Disorders Project, which aims to use the latest genomic technologies to diagnose chidren where a genetic disorder is suspected, but where the tests available to the National Health Service have not so far been able to arrive at a diagnosis. "Over the last seven years we've been able to work with about 13,000 families and provide to the clinical teams that care for them what we think are likely diagnoses that they can work up and confirm", he says.

Matthew Hurles, ESHG Award Lecturer 2018

This work is clearly rewarding, but Hurles has a few qualms too. "I would have liked to see our work enter routine clinical service more quickly than it has, but when you are engaging with an entire healthcare system it seems that delays are inevitable. And the Genomics England programme is now taking up the baton of transforming the system through the better use of genomic data. However, the work that we're doing has made me more aware of the ways in which, as a society, the support that we give families who look after disabled children is woefully short of what they should receive. I find that kind of societal unfairness to be deeply frustrating."

"But on the other side of the coin, I'm very happy to have been able to find a place within science, which is something that fascinates me at an intellectual level, where there is also a real demonstrable benefit to people – especially to vulnerable people in our community, people with rare genetic disorders. It's a real privilege to be able to follow your own interests and do things that you're curious about but that also have outputs that are extremely meaningful for people on a personal level."

If he hadn't been become a scientist, there was a time when Hurles would have liiked to play cricket professionally. "But I realise now that it probably wouldn't have been a wise choice, even had I been good enough." Now he likes the idea of designing gardens even though, he says: "It sound very middle-aged !" As this suggests, retirement is a long way off, and he will decide what he wants to do at a later stage. "There are two types of retirement amongst scientists. Some retire and continue essentially as before, and some you never see again. I don't yet know which category I will fit into."

Gardening and cycling remains big interests, so he will have plenty to do if he decides to quit science for good in twenty years or so. "Every few years friends and I go to the Alps and cycle up some of the classic climbs from the Tour de France. It's very beautiful and quite different from the landscape around Cambridge ! And every gardener will tell you that if they spent more time in the garden it would look better than it does currently."

Hurles has been telling the conference about what he and colleagues have learned during their time with the Deciphering Developmental Disorders Project. "We've acquired a lot of new knowledge about the genetic architecture of these children, who have disorders that are essentially genetic and that cause their severe developmental problems. We've been able to generate different types of genetic data and interrogate it and thereby identify the relative contributions of dfferent classes of variations, as well as defining many new genetic disorders that can now be diagnosed around the world. This has required bringing together a clinical engagement with the latest technologies and the computational approaches required to marry all the data together. It's something that we are proud of, and I am personally happy to have been able to use the science that I find so fascinating in a way that can assist people who are really in need of help."

Distinguished Speaker Interview with Emanuelle Charpentier, Mendel Lecturer 2018

by Mary Rice, ESHG Press Officer

Although she was very interested in the natural and human sciences while at school, Emmanuelle Charpentier didn't consciously think at that time that she might become a microbiologist one day – or so she believed. Yet, when she mentioned her plans to join the Institut Pasteur for her Masters, her mother told her that, aged 12, she had come home from school and said that she would work at the Pasteur one day. "I have no recollection of having said that; but my biology teacher must have talked about something that made me think of the possibility of becoming a microbiologist in the future," she says.

The support of her parents has always been important to her, and particularly so when she left France to pursue her career as a postdoc in the United States. "Being far away from your home in a completely different work culture is not always easy, but I could always talk to them and they helped me expand my mindset. That was a valuable experience and I believe that it also made me a better scientist."

Emanuelle Charpentier, Mendel Lecturer 2018

After five years in the States, she returned to Europe and worked in Austria, Sweden and Germany, where she has been at the Max Planck Institute in Berlin since 2015. The discovery of CRISPR-Cas9 gene editing technology thrust her into the spotlight. "It is truly an experience that has shaped my life as a scientist in a way that I could never have imagined, and I feel very honoured that it has had such an impact on the scientific community. Although there were already many tools for gene surgery, CRISPR-Cas9 has proved to be more precise, easier to use, more efficient and more versatile."

Whereas most technologies take some time to be adopted widely, "thousands of labs around the world are already working hard to further develop the technology," says Charpentier. "I am thrilled about the prospect that one day, the CRISPR discovery may be used to treat serious genetic diseases in humans. CRISPR Therapeutics, the company I co-founded with Rodger Novak and Shaun Foy, has recently filed its first application for clinical trials for a CRISPR-based gene therapy against genetic blood disorders, such as ß-thalassemia and sickle cell disease."

Charpentier still has plenty of challenges to face, both in her work and more widely. "Advocating for basic science, and for microbiology in particular, is not easy. Understanding the basic workings of nature is definitely something that drives me as a scientist in my research, and I'm sure that goes for many colleagues too. But unfortunately, we all have to fight for funding for basic science. The discovery of the CRISPR-Cas9 technology shows clearly that pure basic science can lead to a major breakthrough with practical applications. There are no old or obsolete topics ; one can make interesting findings in many research fields."

If Charpentier hadn't be a scientist, she might have been a ballet dancer or a performer in the arts. "I imagined doing this when I was a child." She would also have enjoyed being an athlete, and still tries to find time for sports.

"It's a way for me to achieve equilibrium after long hours at work. I try to spend time on the track or swimming whenever I can, but with my busy schedule, I no longer have the time for the cultural and artistic life that I used to enjoy."

Retirement is still a long way off, and "once a scientist, always a scientist," she says. But she does sometimes think about projects outside the lab. "The impact of the CRISPR-Cas9 technology has meant that I have had the privilege of meeting many people who have initiated innovative and exciting projects that have the aim of supporting scientists in their work, engaging society, and increasing the visibility of research among the public. I find that very inspiring."

In her talk, Emmanuelle Charpentier intends to share the history surrounding the discovery of the CRISPR-Cas9 gene editing technology with her geneticist colleagues. "As a microbiologist, I have always been interested in the fundamental mechanisms of infection and immunity in bacteria, and this is how I identified the CRISPR-Cas9 mechanism. Its versatility and simplicity has an immense potential for the treatment of serious genetic disease, but it also comes with challenges and responsibilities for scientists, particularly when it comes to editing the human germline, the subject of huge debate throughout Europe and more widely. I am already looking forward to the discussions!"

Student Interviews from the SIGU-ESHG Schoolday in Milan, June 16-19, 2018

by Mary Rice, ESHG Press Officer

Chiara Latini and Luca Sportelli, both aged 17 and studying at the Liceo Scientifica Leonardo da Vinci, Milan, were among students attending a special schools workshop at this year's conference. The workshop included plenty of hands-on activities; a new experience for both of them.

"I've done some very different things, and it's been quite unlike the formal lessons we have in Italy. We don't much use the hands-on approach that is more common in the US or in Britain. Italian science teaching tends to be very academic and not practical, which is helpful because you gain a lot of knowledge and you understand in depth how things work. But I was missing the practical side, and now I have it !" said Chiara.

"Even if genetics isn't my favourite area of science, I really enjoyed it because I learned something new and learning something new is always important and useful. It was a practical experience where we had to something more than just taking notes and listening – I had to try to do something that I didn't know how to do and that's why it's been really useful for me," Luca added.

Luca's ambition is to be an astronaut. "I'd like to go where no-one has been before. I want to study aerospatial engineering and work for NASA – that's my dream!" Maybe this seems a long way from genetics, but he sees the bigger scientific picture too. "It's the future for humanity because if we want to go on to become better in any way you have to understand science in order to evolve. Science can show humanity how to grow and to become better."

Chiara's ambition is to study medicine. "I've always been very interested in understanding why things happen – why somebody has a particular pathology and why, maybe, their child has not. It's understanding humans and how we are all so similar but different at the same time. I know that studying medicine is going be very hard and time-consuming, but it's the future. If we want to cure illnesses that are destroying people, like Alzheimer's and Parkinson's, I want to able to contribute. And genetics is where everything begins."

Beatrice Zanini, from the Liceo Scientifico Statale Nicoloso da Recco, Recco, near Genoa, was one of the organisers of the day. "In 2008 we started to involve students, schools and teachers in a lot of initiatives. Genetics is part of the Italian curriculum and we wanted to include more schools so I contacted Genoa University. During my doctorate there I took part in a number of teaching experiences in the lab with students from schools in Genoa and elsewhere in Liguria. Based on that experience, we were able to propose more topics to schools and improve the genetic knowledge of teachers and students."

"The schools day at ESHG is organised each year by the local national society and sometimes it's difficult, because it's not an official session and it's extra work. The national society is free to organise the day as it wishes, and therefore the format can vary considerably from one year to the next. I hope, though, that it will continue along the same lines, with workshops and hands-on activities. We get excellent feedback from this kind of event and, if I were asked to advise next year's organisers, this is what I'd suggest."

Report on the 3rd Course of Basics in human genetic diagnostics

by Burac Nadejda, first year resident in Medical Genetics, Chisinau, Moldova

The 3rd Course of "Basics in human genetic diagnostics – A course for Clinical Laboratory Geneticists (CLGs) in education", Zagreb, Croatia, September 03-07, 2018.

This year, the 3rd Course of Basics in human genetic diagnostics – A course for Clinical Laboratory Geneticists (CLGs) in education took place in Croatian Institute for Brain Research from Zagreb, Croatia and was organized by Thomas Liehr (Jena, Germany), Martina Rinčić (Zagreb, Croatia) and Feodora Stipoljev (Zagreb, Croatia). The main sponsors (European Society of Human Genetics, ADS BIOTEC, Agilent Technologies, Carl Zeiss, ZytoVision) provided support for the course. The participants that attended the course were from different 17 countries, i.e. 48 people coming from Bosnia and Herzegovina, Croatia, Finland, Germany, Greece, Kosovo, Libya, Macedonia, Moldova, Morocco, Netherlands, Romania, Serbia, Spain, Thailand, Turkey, and UK. Each day we had lectures from the morning (9 AM), in the afternoon we performed some workshops, and at the end of each day (~5 PM) we had a written test. All questions from the tests were covered during lectures and we had all lectures from the beginning of the course on a USB-drive. During breaks we had the opportunity to know each other, to discuss some questions with lecturers and participants regarding topics from the course. The course was not only about learning, also we enjoyed our time in Zagreb, in the second and last day we had an awesome dinner together in two different local restaurants, and in the third day we had a very informative and entertaining guided tour through the city. Despite of our busy schedule we had great time learning, and we were updated with the latest news and topics of genetics.

In the first day of the course we discussed basics of genetics, Mendelian and non-Mendelian inheritance; also were covered some interesting topics such as syndromology, genetic diseases and basics of genetic counseling. During workshop we had the opportunity to learn how to do own risk-calculations analyzing pedigrees. Also, we met Nicole Fleischer from FDNA (https://www.face2gene.com/fdna/), who presented how to use Face2Gene application, which is great help for clinicians to provide potential diagnoses for dysmorphic children. During the second day we have got familiar with cytogenetics. Anja Weise and Thomas Liehr (Jena, Germany) covered topics such chromosomal syndromes and databases, classical and banding cytogenetics, human chromosomes, and at the end of that day we had a very interesting workshop: we identified human chromosomes on a metaphase picture and performed a karyogram.

The third day was about molecular cytogenetics and molecular genetics. We discussed fluorescence in situ hybridization (FISH-) technique, types and applications, comparative genomic hybridization (CGH) and array-CGH, gene therapy, PCR techniques and applications. The fourth day was dedicated to molecular genetics, were we got insights into molecular techniques such as multiplex ligation-dependent probe amplification (MLPA), Sanger- and next generation sequencing (NGS). During a workshop some Sanger sequencing cases were exemplified. On the last day of the 5 day course topics presented were very heterogeneous and included actual topics such as NIPT (Non Invasive Prenatal Testing), cancer genetics, inborn errors of metabolism, neurogenetics, proteomics, and future directions of the field. Also, we had the opportunity to provide written evaluation of the course and have an open discussion with the course organizers how course could be developed on future.

This course gathered specialists of all ages, those who were experienced had the opportunity to refresh their knowledge, and those who were at the beginning of their career had a unique chance to learn many new things and to see what future perspectives in genetics are. I am very grateful to the organizers who have done tremendous work for this course, to all lecturers who have come up with useful and well-structured information, and of course to all participants who have shared their own experiences – these were besides already mentioned organizers also from Croatia Miljenko Katunarić, Irena Drmić Hofman, Romana Gjergja Juraški, Gordan Lauc, Snježana Židovec Lepej, Kristian Vlahoviček, Oliver Vugrek and Tamara Žigman, and from Slovenia Ales Maver, Borut Peterlin, Gorazd Rudolf and Marija Volk. I would like to thank especially for a fellowship, which was extremely useful for enabling my participation – thanks to support by ESHG overall 17 of the 48 participants could be sponsored by fellowships covering parts of the course fees. I hope very much in the future to meet participants and lecturers at other courses or conferences and to collaborate with each other. Being at the beginning of the way in clinical genetics, I left this course, even though it was at some points a busy and challenging program at some points, very inspired and motivated.

Picture:

Group-photo with organizers Martina Rinčić (second from left, row 2), Thomas Liehr (first from left, row 1) and Feodora Stipoljev (second from left, row 1) in front of the venue, the Croatian Institute for Brain Research, Zagreb.

EJHG Appoints Social Media Editor

As of december 2018, the EJHG will have a Social Media Editor, dr Alisdair McNeill. Alisdair is a Consultant Clinical Academic at the University of Sheffield and Sheffield Children's Hospital NHS Foundation Trust, has been guest editor for a special edition of Brain Sciences and has extensive experience as a genomics educator, leading an Msc module on genomic counselling at Sheffield University and teaching multiple undergraduate lectures. His research focuses on identifying novel genetic brain disorders and characterising the prodromal phase of neurodegeneration in genetically predisposed cohorts. He has an active profile on twitter (@am_sheffgenet), with around 1300 followers, successfully used twitter to publicise study days he organised, recruit research participants and build an academic network, and uses Kudos (https://www.growkudos.com/about/researchers) to publicise his research papers. He will , amongst others, develop the Twitter and Facebook presence of the Journal, highlighting published and in-press EJHG papers, develop an EJHG Blog page for authors, and a Youtube EJHG section subsuming the EJHG-tube material, rendering it more visible and facilitating the submission process.