

PERSPECTIVE

# The annual meeting 1988–2017

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*European Journal of Human Genetics* (2017) 25, S35–S36; doi:10.1038/ejhg.2017.150

I well remember the 1988 Cardiff meeting as my first introduction to European Society of Human Genetics (ESHG). It was a friendly meeting of about 100 geneticists, including prominent scientists such as Jean de Grouchy, John Edwards, and Peter Harper. The conference also hosted budding young talents such as Andrew Wilkie. There was a single session, and the small number of attendees provided for ample opportunities to make new acquaintances. In retrospect, Cardiff was an important meeting in which the ESHG membership approved a change towards the creation of a more modern society and meeting. Unfortunately, I was too young to realise the importance of the event, and I took the afternoon off with new-made scientific friends to explore the local countryside and pubs.

The ESHG meeting has grown steadily ever since, hitting 1000 attendees around the year 2000, and then expanding to 3500 in Barcelona in the year 2016. This has led to a growth from 2002 to 2016 from 9 to 24 concurrent sessions with more than 200 oral presentations, from 7 to 18 workshops, and from 4 to 25 company satellites. There were >150 commercial exhibitors in Barcelona with >500 representatives. One innovation from 2009 has been the Educational track which runs parallel to Invited symposia. First proposed by Ulf Kristofferson, this has been a particularly successful innovation that has done much to keep the ESHG annual meeting attractive for those practising primarily in medical genetics.

The EMPAG European Meeting on Psychosocial Aspects has been around for more than 25 years. They decided to combine their biannual meeting with that of the ESHG starting in 2002 in Strasbourg. This has been a particularly propitious arrangement for both communities. It is always striking to see how much cross-over happens with ESHG attendees visiting EMPAG sessions, and *vice versa*.

The Society has had an annual award for many years. It started in 1992 as the Baschiroto award, and then became the ESHG award from 2003 onwards. This award is the main scientific award that the Society makes. The award winners are all genetic scientists who contributed to the growth of human and Medical Genetics as a scientific discipline in Europe. There is something that the ESHG leadership may wish to remedy, however. Strikingly, while four of the first eight winners were women (Lore Zech, Mary Lyon, Leena Peltonen, and Pat Jacobs), gender equality has not been maintained. With only Veronica van Heyningen receiving this distinction in 2006, the ESHG award has seen a worrying lack of female winners over the last 15 years. There is hope that after Edith Heard in 2017, the trend has now been reversed.

A constant feature of the ESHG meetings has been its emphasis on welcoming young scientists. The society provides numerous fellowships to young investigators based in the less affluent countries in Europe's East and South. It also has presented prizes for the best presentations by young investigators since 2002. Quality (and age) are the only factors

that the YIA jury takes into account here. Strikingly, Florence Molinari, Louise Harewood, Marjolein Willemsen, and Marjolijn Jongmans each won a Young Investigator Award on two separate occasions.

The scientific quality of the ESHG conference has been rigorously maintained over the years by the Scientific Programme Committee, which is composed of geneticists from all over Europe. Scientific excellence is the only criterion, and the meeting's organisers have avoided any influence of sponsors on the content of the meeting apart from the company satellites. Longstanding conference organiser Jerome del Picchia and his staff at VMA, working with Jantje de Roos from Rose international—recently assisted by her daughter Flora—make sure that the exhibition remains an integral part of the meeting.

Several features of the ESHG conference have set it apart from other meetings in Medical and Human Genetics, and probably from other European Societies. There has been a strong emphasis on inclusion, and on making the meeting truly European. One way of stimulating inclusion is to create opportunities for participants from countries in Southern and Eastern Europe to attend the meeting. ESHG has had a steadily growing fellowship system for the last 15 years, at least. This provides free access to the meeting, as well as some support for travel and accommodation. Each year, the best young scientists from less developed countries can attend and become a part of this international community. Andrew Read, during his stint as chairman of the Scientific Programme Committee, developed guidelines that have been largely observed by subsequent SPC chairs. These have sometimes been referred to as 'the Read rules'. These rules arise from the realisation that the same scientific effort produces more results in some countries than others, and so within reason ESHG is prepared to accept posters that show modest new data (but not no data!) from countries where any result is an achievement. The SPC also understands that many students need the acceptance of their poster to qualify for any support in their own countries to attend the meeting. This concept has sometimes been summarised as: 'As long as the work is not totally mad or unethical a poster shall be accepted'. This policy is one reason why the meeting has a relatively large poster exhibition. With increasing pressure on space for posters, the bar has been raised somewhat in the last few years, but inclusion is still the dominant concept. The recent inclusion of electronic posters is a particularly elegant solution which allows our meeting to grow even further without the need for going to ever larger venues.

A particular success for the last 20 years has been the dysmorphology workshop. This started in Genoa 1997 with Robin Winter and Dian Donnai running a small session with about 50 people and an 'expert panel' that was asked to sit in the front row. It was quite a challenge, with slides being put in a carousel. As soon as technology allowed, the London Dysmorphology Database was included in the

session, necessitating double projection which provided another challenge for the conference centre. Gradually the number of attendees increased as well as the number of cases to be presented and there have been two workshops at the conference for the last ten years with attendance sometimes reaching 400 people per session. Current facilitators are Dian Donnai, Jill Clayton-Smith, and Sophia Douzgou. The great things about the dysmorphology sessions are that (1) delegates from anywhere get to present and often the best prepared cases come from doctors in the most economically deprived countries; (2) colleagues from big centres often offer to do investigations for those from poorer countries—it is like a 'dating agency' since publications have resulted from these connections; (3) it has a real educational aspect since doctors new to dysmorphology get to see how more experienced colleagues formulate a differential diagnosis because new diagnostic techniques are discussed.

Finally, an integral part of the annual conference has been the conference party. This is always a lively affair; a special mention goes to the Amsterdam 2001 meeting for the drag queens impersonating the Supremes, and to the Barcelona 2008, and the Gothenburg 2009 parties that saw particularly lively bands and dancing as well as good food and the celebration of new and old friendships. When I contrast the 1987 Cardiff and 2017 Copenhagen meetings, what has remained is an exceptionally congenial and relaxed atmosphere, balancing clinical, technological and basic science progress. At the same time, the content and quality have grown as much as the overall size of the meeting. And what joy to come back to Copenhagen after 50 years and see 3200 attendees from 92 different countries! ESHG has become a truly international European Society. Surely, had they known this 50 years ago, those who started our society would have been very proud.



**Henri Gerrit (Han) Brunner** (MD, PhD), Han Brunner trained as a clinical geneticist at Nijmegen University. In 1998 he was appointed full professor and head of the department of Human Genetics at Nijmegen University Hospital. In 2014 he was also appointed chairman of the Department of Clinical Genetics at Maastricht University Medical Center, in the Netherlands. He was previously elected member of the board of directors of the Dutch, European (president in 2014–2015) and of the American Societies of Human Genetics. Han Brunner was elected member of the Royal Netherlands Academy of Arts and Sciences in 2013, and of the Academia Europea in 2012. He is a Knight in the Order of the Dutch Lion since 2013. He is a co-winner of the King Faisal International Prize in Medicine 2016, with Joris Veltman. He was awarded the 2017 Carter medal of the British Clinical Genetics Society.

Han Brunner pursues the scientific understanding of the connections between clinical and molecular features of rare diseases, including applications to patient care. He has pioneered the discovery of a large number of disease genes, and the application of cutting-edge genomic technologies (genomic microarrays, exome sequencing, and whole genome sequencing) to understand genetic diseases. Much of this work is on neurodevelopmental conditions such as intellectual disability and abnormal behavior. His work has contributed to our understanding of intellectual disability by showing that in non-consanguineous populations, its major cause lies in spontaneous new mutations.