REPORT ON THE EUROPEAN HUMAN GENETICS CONFERENCE, 21–24 MAY 2016, BARCELONA, SPAIN

Urszula Lechowicz, Agnieszka Pollak, Monika Oldak

Department of Genetics, Institute of Physiology and Pathology of Hearing, Warsaw, Poland

Corresponding author: Urszula Lechowicz, Department of Genetics, World Hearing Center of the Institute of Physiology and Pathology of Hearing, Mokra 17 Str., Kajetany, 05-830 Nadarzyn, e-mail: u.lechowicz@ifps.org.pl

The 49th European Society of Human Genetics (ESHG) Conference took place in Barcelona, Spain between 21st and 24th May 2016, organized in cooperation with the Spanish Association of Human Genetics. The ESHG Conference is the most important annual meeting for researchers and clinicians in the area of human genetics. It provides a unique occasion to learn of recent discoveries in the field. This year the conference hosted over 3,500 attendees and offered an opportunity to listen to more than 215 oral presentations, 18 workshops, and 8 educational sessions. Additionally, over 150 companies exhibited their range of products and services.

By tradition, the Conference starts and finishes with Plenary Sessions, which are particularly interesting, picking up on current issues for the benefit of all participants. The sessions are presented by speakers who have been honored with prestigious awards in biology and medicine. This year's Mendel Lecturer was Professor Sir Adrian Bird from the University of Edinburgh, Scotland. He is holder of the Gabor Medal (1999), Louis-Jeantet Prize for Medicine (1999), Gairdner Foundation International Award (2011), Royal Society GlaxoSmithKline Prize (2012), Thomson Reuters Citation Laureate (2013), BBVA Foundation Frontiers of Knowledge Award (2013), and Shaw Prize (2016). In 2013 Professor Bird was a contender for the Nobel Prize for his fundamental discoveries in DNA methylation and gene expression. His research focuses on DNA methylation mechanisms in animal cells, especially regulation pathways of the mammalian genome via epigenetic modifications and their role in diseases such as Rett syndrome. He discovered that the MeCP2 protein binds specifically to methylated CpG sites and has proved that failure of this interaction leads to Rett syndrome.

The ESHG Award Lecturer 2016 was Professor Stefan Mundlos, Director of the Institute for Medical and Human Genetics at the Charite, Berlin, and group leader at the Max Planck Institute for Molecular Genetics, who gave a fascinating speech about gene regulation and the noncoding genome. He emphasized that the interpretation of non-coding regions, which comprise more than 98% of the human genome, is the main challenge in human genetics. His research is focused on the effects of structural variations on gene regulation, particularly on the finding that structural aberrations such as deletions, duplications, or inversions may affect the 3D folding of chromatin.

During the Conference participants could choose between many extremely interesting concurrent educational sessions, interactive workshops, corporate satellites, or comprehensive poster session comprising more than 1500 presentations – which is the scientific core of the conference. Usually there are at least 6 parallel sessions to choose from. During Concurrent Symposia, invited lecturers speak about new results on a given topic, offer reflections, or compare data with other researches. The main topics of this year's Concurrent Symposia included: understanding the functional effects of genomic variants, the importance of cell-free nucleic acids, sensory disorders, long distance regulation of transcription and translation, epigenetic reprogramming, and therapy in rare diseases. During Concurrent Sessions, speakers are chosen from among the authors of abstracts submitted to the conference, meaning the most outstanding work gets to be discussed. The following topics were presented: (i) molecular mechanisms underlying intellectual disability, predisposition to cancer, neurogenetics, and disorders of the neuromuscular, metabolic, ocular, and cardiovascular systems; (ii) functional genomics; and (iii) bioinformatics and data sharing and mining. Moreover, a wide range of workshops took place, during which discussions included the analysis of next generation sequencing data, interpretation and classification of DNA variants, use of data bases, and dysmorphology.

Because of the sheer size of the ESHG assembly, it is not easy to navigate among all the lectures. To up-date all participants, a free ESHG 2016 Congress App was provided to both iOS and Android users. This interactive guide made it easy to find talks or sessions; what is more, it gave a short outline about the speaker, their work, and contact details, and allowed feedback to be given on talks or sessions.

The most newsworthy aspects of human genetics presented to the ESHG conference were the increasing importance of analysing the "gene desert" (noncoding DNA); new genome editing tools with the CRISPR/Cas9 technology; development of long reads in next generation sequencing; and development of diagnostic algorithms for better identification of rare genetic syndromes.

We presented five posters to the conference, all of which made a favourable impression, leading to many exciting and inspirational discussions. The posters were:

"Distinct clinical and radiological phenotype associated with *POU3F4* mutations" (Pollak A., Lechowicz U., Kedra A., Stawinski P., Rydzanicz M., Mrowka M., Skarzynski P., Furmanek M., Skarzynski H., Ploski R., Oldak M.)

"*TMPRSS3* mutation as a cause of non-syndromic hearing impairment among Polish hearing loss patients" (Lechowicz U., Pollak A., Podgorska A., Stawinski P., Oldak M., Skarzynski H., Ploski R.)

"Fuchs endothelial corneal dystrophy: strong association with rs613872 not paralleled by changes in corneal endothelial TCF4 mRNA level" (Ozieblo D., Ruszkowska E., Udziela M., Binczyk E., Sciezynska A., Ploski R., Szaflik J.P., Oldak M)

"Technically difficult, diagnostically important: exon ORF15 of the *RPGR* gene in retinitis pigmentosa" (Oldak M., Ruszkowska E., Siwiec S., Pollak A., Stawinski P., Szulborski K., Szaflik J.P.)

"Deep next generation sequencing of the whole mitochondrial genome in Polish patients with aminoglycoside-induced hearing impairment" (Rydzanicz M., Pollak A., Lechowicz U., Stawinski P., Wrobel M., Wojsyk-Banaszak I., Ploski R.).

As always, the 2016 ESHG Conference was remarkable, and it remains a "should be there" event for us human geneticists.