RARE DISEASE IN SOUTH-EASTERN EUROPE,
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Under the auspices of the European Academy of Paediatrics, the first Rare Disease in South-Eastern Europe (SEE) meeting was held on November 15–17 2012, at the Macedonian Academy of Sciences and Arts, Skopje (MASA). This was a manifestation in honour of the 45 years since its establishment, under the guidance of the G.D. Efremov Research Centre for Genetic Engineering and Bio-technology. The Macedonian Chamber of Doctors granted 20 CME credit points for the meeting.

The meeting started with talks on the importance of rare diseases in general (Liesbeth Siderius, The Netherlands, John Dodge, UK) and in Macedonia (Zoran Gucev, Velibor Tasic, Dijana Plasevska-Karanfilska, Momir Polenakovic). The importance of having registries for RDs, team work, additional education and international collaboration was stressed as being especially important. All speakers, as well as the NGO patient organization (Slavica Stojanovska, Vesna Stojmenska) underlined the need for comprehensive societal efforts in tackling this emerging social and medical problem. Surgical problems regarding the ear and hearing were finely exposed in the presentation by Ilija Filipce (Macedonia).

At least 80% of RDs are genetic in their origin. Anna Tylki-Szymańska (Poland) spoke about Lysosomal acid lipase deficiency, Wolman disease and cholesteryl ester storage disease, while Agnieszka Jurecka (Poland) introduced the audience to the natural history of mucopolysaccharidosis type VI. Especially insightful presentations were those describing the genetics of abnormal vertebral segmentation and the Notch signalling pathway (Peter Turnpenny, UK) and...
the overgrowth syndromes in their interesting and exciting relationship to human cancer (Between Mendel and cancer – PI3K/AKT-related overgrowth syndromes, Robert Sample, UK).

The genetics and the clinical features of the pituitary were addressed by several prominent scientists. The genetic causes of IGF-I deficiency and insensitivity were presented by Jan-Maarten Wit (The Netherlands), while the defects within pituitary growth factors and their phenotypes in infancy and adolescence was brilliantly presented by Roland Pfaeffle (Germany). In addition, the fine aspects of the septo-optic dysplasia were presented by Liljana Saranac (Serbia). The ever growing body of genetic insights was enriched by the fundamental findings in epigenetics (genomic imprinting anomalies) presented by Yves Le Bouc (France) with his talk on the Beckwith-Wiedemann overgrowth syndrome (BWS) and the Russell-Silver (RSS) syndrome.

As, rather unfortunately, patients with Gaucher’s disease (GD) are not treated in Macedonia, Gaucher disease was a separate section at the RD meeting at MASA. GD as a model disorder for targeted therapy (Patrik Deegan, UK) and the therapeutic options in Gaucher’s disease (Deborah Elstein, Israel) introduced the public to the basic and the newest available therapeutic options.

Regional collaboration was especially stressed in the talk on the laboratory diagnosis of lysosomal storage diseases in Bulgaria, by Ivanka Sinegirska from Sofia, Bulgaria. Arunas Valiulis (Lithuania) talked about the EU Leonardo Project, Building a Network of Management of Alpha-1 Antitrypsin Deficiency in Central / Eastern Europe, further pointing towards the need for international and especially European collaboration on the particular form of RD.

As the first patient(s) with Fabry’s disease have been discovered in Macedonia the talk on Fabry enzyme replacement therapy (ERT) (Patrik Deegan, UK) was especially insightful. The molecular diagnosis and treatment of Wilson's disease is always a difficult clinical situation, as stressed by Georgios Laudianos, Italy.

Insights on primary hyperoxaluria type III were given by Yaacov Frishberg (Israel), especially interesting were the presentations on atypical haemolytic uremic syndrome (Zoltan Prohaska, Hungary), hypouricaemia (Ivan Sebesta, Prague, Czech Republic; Dganit Dinour, Israel; and Velibor Tasic, Macedonia). Alport's Disease (Gordana Petrujevska, Macedonia), Autism (Nada Pop-Jordanova, Macedonia), progressive familial and benign recurrent intrahepatic cholestasis (Viktorija Chaloska-Ivanova, Macedonia) and a comprehensive survey on patients with primary immune deficiency diseases in Macedonia (Kristina Mironska, Macedonia) were also presented. Several groups have also contributed with their posters (27) on different RDs.
The meeting was attended by 128 delegates from 21 countries. In addition, there were conclusions and proposed modifications for further progress in the development of diagnosis and treatment of RDs.

The meeting and its organization were rated by delegates and lecturers as an exceptional scientific success. Of particular importance also is the fact of the intensive collaboration of SEE countries and SEE and the EU countries. It was also an opportunity to reconsider the state of the organizational, educational and scientific integration of SEE and the EU on the rather specific field of RDs.

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