MÆDICA - a Journal of Clinical Medicine

REPORTS FROM MAJOR CONGRESSES

Rare Disease Day – at a glance

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Rare Disease Day is an annual event, on the last day of February, coordinated by EURORDIS (European Organisation for Rare Diseases) at the international level and Alliances of Patient Organisations at the national level and, for this, hundreds of patient groups and their partners organise activities to rise awareness on rare diseases. This year's theme focused on the importance of rare disease research around the moto " Patients and Researchers: Partners for Life". A disease is defined as rare in Europe when it affects less than 1 in 2000 citizens. There are between 6000 and 8000 rare diseases, 80% of them have identified genetic origins and 75% of rare diseases affect children.

For the second year Mother and Child Care Institute IOMC "Prof. Dr. Alfred Rusescu" from Bucharest joined in the celebration of Rare Disease Day. "Rare diseases – beyond appearances" was called the symposium organized by Research Department of the Institute.

In line with this year's theme doctors, specialists or residents, researchers, patient organizations were invited to present their experience in diagnosing of rare diseases. Our guests from Pediatric Neurology, "Alexandru Obregia" Psychiatric Clinical Hospital, "Mina Minovici" National Institute of Legal Medicine, "Francisc I. Rainer "Anthropology Institute, "Filantropia" Hospital, "Sf. Imparati Constantin si Elena" Ilfov District Hospital were invited to become Rare Disease Day partners.

All participants tried to bring attention to rare diseases and promoted collaboration among specialists from different medical fields. It was with great success. Presentation of cases with genetic diseases dominated audience, bringing further evidence that rare diseases are mostly genetic diseases, monogenic or chromosomal disorders.

Wiliams Syndrome Association represented by its president stressed the idea of multidisciplinarity

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for diagnosis and treatment of a patient with a rare disorder.

The clinical presentations had very interesting topics: patients with multiple congenital anomalies, metabolic disorders, primary immunodeficiencies, congenital ophtalmic conditions, rare heart defects, chromosomal anomalies. It also outlined the idea of prenatal prevention of some rare diseases. New techniques for diagnosis have been presented, based on cases encountered in medical practice. Listing of free information sources for rare disease has been a great help to all.

The scientific program provided also research findings on a wide variety of clinical issues. Overall, the deliberations at the meeting sensitized participants to the large spectrum of rare disorders. The most important idea of our symposium was that "rare diseases are rare, but patients with a rare disease are many".

We thank everyone for making this event happen. ❑