

Rare Diseases Day in Romania

Vasilica PLAIASU, MD^a; Michaela NANU, MD, PhD^b

^aGenetics Department, IOMC "Alfred Rusescu", Bucharest, Romania

^bSenior Researcher, IOMC "Alfred Rusescu", Bucharest, Romania



Series of meetings and activities took place in February in the context of World Rare Disease Day. Mother and Child' Care Institute "Prof. Dr. Alfred Rusescu" (IOMC) has agreed to promote this event and hosted a one-day Symposium "Pediatrics Rare Diseases" which took place on February 26th, 2009, in Bucharest and focused on rare diseases. The conference was attended by 100 health professionals from IOMC, National Legal Medicine Institute "Mina Minovici", Al. Obreja Hospital, Colentina Hospital, Anthropology Institute "Francisc I. Reiner". This was the primary annual meeting of our institution that allowed maintaining and enhancing programs for patients with rare diseases and their families.

IOMC considers that one of its most important missions is to promote information about rare disorders to doctors and patients. The conference was opened with remarks from prof.dr.Adrian Georgescu, who had come to underscore the needs of people with rare dis-

eases.

For chronic and rare severe diseases, care is not restricted to medical treatment. Patients and their families require other types of care

throughout their entire lives – information and support via associations. Rare disease patients often feel isolated by their condition. The president of Williams's Syndrome Association underlined the impact on the relationships between health professional and patients.

The objectives of the meeting were to improve prevention, diagnosis and access to health care services for rare disease patient. A novelty of this symposium was the presentation of case reports from daily practice. All speakers encouraged medical professionals to contribute to patients' care in an interdisciplinary team.

A rare disease is generally considered to have a prevalence of less than 5 per 10,000 persons in the European Union. The last day of February has been designated as worldwide "Rare Disease Day" to call attention to rare disorders.

Address for correspondence:

Vasilica Plaiasu, MD, IOMC "Alfred Rusescu", 120 Lacul Tei Blvd., Bucharest, Romania
email address: vasilica.plaiasu@gmail.com

The European Organisation for Rare Diseases (EURORDIS) represents more than 300 rare disease organisations in 34 different countries, covering more than 1,000 rare diseases. It is therefore the voice of the 30 million patients affected by rare diseases throughout Europe. Many rare diseases are genetic in nature and almost all rare conditions are incurable.

Activities undertaken in the meeting included:

- presenting of rare pediatrics cases
- assure access to quality laboratory testing
- promote collaboration, cooperation and community involvement
- raise public awareness
- rare diseases have to identified as a public health issue in Romania.

This educational effort was intended to help patient obtain earlier diagnosis and speedy referrals to appropriate sources of assistance. Rare diseases often go undiagnosed or misdiagnosed for long periods. The inability of doctors to interpret patient symptoms and features correctly

appears to be a major impediment to prompt diagnosis and treatment. A doctor may become aware of new information that could be important in diagnosing a patient as time goes on.

It was a conference where medical professionals from different institutions took opportunity to exchange their experience and discuss problems they face in providing healthcare to their patients.

It is clear from this conference that IOMC is deeply committed in improving the human health and we believe by working together that we can diagnose and treat a lot of rare diseases.

Some of recommendations made by commenters can be integrated into needs assessment: enhanced interaction and communication between pediatric clinicians and development of a network of children's hospitals with expertise in rare pediatric diseases and conditions.

The hope is that Rare Diseases Day will increase awareness of rare diseases and the need for effective treatments or cures.

