

Association of patients with blood diseases Slovenia
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Third National Conference on RARE DISEASES

BRDO CONGRESS CENTRE, SLOVENIA
FEBRUARY 28, 2017



The 3rd National Conference on Rare Diseases was held in Slovenia on the Rare Disease Day 2017. The conference was organized by the Ministry of Health of the Republic of Slovenia and the Association of Rare Diseases Slovenia and held under the honorary patronage of Slovenian president, Mr. Borut Pahor.

The meeting was attended by the members of twelve organizations and brought together rare disease patients, representatives of the Ministry of Health, representatives of the Clinical Institute of Medical Genetics of University Medical Centre Ljubljana, professional medical staff, a representative of the Health Insurance Institute, representatives of governmental and non-governmental organizations, representatives of pharmaceutical companies, representatives of the University of Ljubljana, as well as the media.

Health Minister Milojka Kolar Celarc has highlighted the concern of the Ministry for rare diseases and announced an even more active implementation of the Work plan in this field, which will be supported by an active working group, composed of medical experts and patient representatives. She expressed a belief that in order to achieve our common goal - a satisfied patient that is an active and equal member of society - we need an integrated system of treatment of rare diseases, which can only be achieved through close cooperation and understanding of difficulty of the task ahead.

Following the opening speech by representatives of the Ministry of Health and the president of the Association of Rare Diseases Slovenia, the first part of the conference consisted of lectures thematically dubbed "Living with a rare disease". The main topics of the lectures were the following: classification of rare diseases, the achievements in the field of rare diseases in Slovenia during the period in-between the conferences, activities in the field of rare diseases in Slovenia and public health aspects on how to diagnose rare diseases as soon as possible.

Three young patients with different rare diseases also presented their life stories together with their doctors at the conference. They spoke about how to live with a rare disease, and shared with the participants their problems, fears and the immense will to live.

The second part of the conference was marked by two interesting round tables, both touching upon very current topics, namely the "Psychological support for patients with a rare disease" and the "Medical-ethical aspects of rare diseases".



Roundtable patients with rare diseases

The conference was closed by the panel discussion, which highlighted the importance of the linked chain "Patient - Physician - Researcher". Speakers agreed with the opinion, stated by the president of the Association for Rare Diseases Slovenia, prof. Jože Faganel, who pointed out that "the engine of progress has always been the thought of not coming to terms with the disease, not letting the hopelessness overcome us, the need of seeking the hidden cause, which could only then be prevented." With this in mind, and together with numerous participants, he firmly endorsed the slogan of the Rare Disease Day 2017 - **"With research, possibilities are limitless."**

On the occasion of the conference, the Association of Patients with Blood Diseases Slovenia presented the book entitled "Rare Diseases February 28, 2017 - 7th edition", which was available to all conference participants. The book's forewords were written by the Slovene Minister of Health, Ms. Milojka Kolar Celarc, and the President and Vice-President of the Association for Rare Diseases Slovenia, prof. Jože Faganel and Ms. Majda Slapar.

In the central part of the publication, mag. Julijan Naskov, dr. med. draws readers attention to the value of medical innovations. This is followed by a contribution on rare diseases as cases of congenital metabolic diseases by doc. dr. Mojca Žerjav Tanšek, dr. med., doc. dr. Urh Grošelj, dr. med., and prof. dr. Tadej Battelino, dr. med. from the Clinical Department of Endocrinology, Diabetes and Metabolic Diseases, Division of Paediatrics, University Medical Centre Ljubljana. Rare diseases in the field of child neurology are presented by Valentina Prevc, Karmen Zrnc, asist. dr. Maja Jekovec-Vrhovšek, dr. med., and prof. dr. David Neubauer, dr. med. from the Faculty of Medicine, University

of Ljubljana and Clinical Department for Children, Youth and Development Neurology, Division of Paediatrics, University Medical Centre Ljubljana. In the book, Head of the Clinical Department of Hematology at the University Medical Centre Ljubljana, prof. dr. Peter Černelč, dr. med. also introduces the reader to the availability of drugs for treatment of less common diseases in the field of clinical hematology in Slovenia. The field of rare cancers is later covered by dr. Branko Zakotnik, dr. med. from the Institute of Oncology Ljubljana. Specialist of dermatoneurology from Dermatoneurologic Clinic, University Medical Centre Ljubljana, assoc. prof. dr. Mateja Dolenc Voljč, dr. med. presents the problem of epidermolysis bullosa. The



Participants of the 3th National conference on rare diseases at Brdo Congress Centre, Slovenia

psychological support and "Patient - Patient" counseling is described in the publication by Ms. Majda Slapar, the president of the Society of patients with blood diseases Slovenia.

The second part of the book defines and describes rare diseases, presents drugs for the treatment of rare diseases - orphan drug products, research on rare diseases, their determinants in Europe and the way forward. Information about research projects in Europe supporting the collaboration between organizations in the field of rare diseases, as well as the list of international meetings on rare diseases in 2017 are also given in this chapter. Appendix 1 contains a list of different medicinal products to treat rare diseases, and Appendix 2 contains a list of all collaborating associations, which bring together rare disease patients.