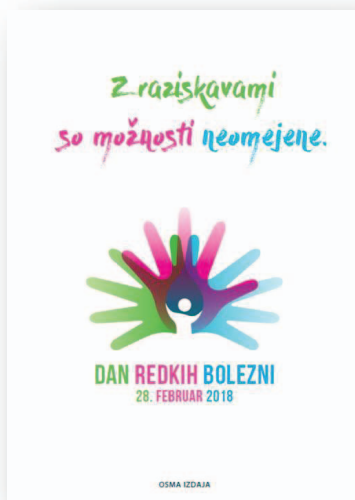


Association of Patients with Blood Diseases Slovenia
published updated edition of »Rare diseases –
February 28th 2018, 8th edition«



Jože Faganel
President of the Slovenian rare disease association
<https://zrbs.si/>

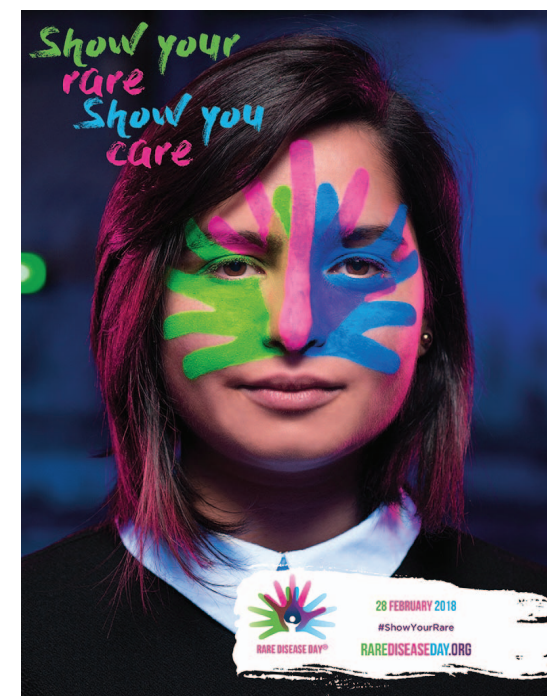


REPUBLIKA SLOVENIJA
MINISTRSTVO ZA ZDRAVJE



Fourth National Conference on Rare Diseases

BRDO CONGRESS CENTRE, SLOVENIA
FEBRUARY 28, 2018



The event was sponsored by:



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with Blood Diseases Slovenia
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Participants of the 4th National conference on rare diseases at Brdo Congress Centre, Slovenia

The fourth National Conference on Rare Diseases was held in Slovenia on the Rare Disease Day 2018. The conference was co-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 numerous patient organizations, representatives of the national 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.

Minister of Health **Mrs. Milojka Kolar Celarc** pointed out in her introductory address that activities for improved healthcare for patients with rare diseases are also one of the guidelines of the Resolution on the national healthcare plan 2016-2025 "Together for the Health Society". She highlighted the successfully implemented project of the National Contact Point for Rare Diseases. The goal of this project of the Ministry of Health is to connect institutions, professionals, patients with rare diseases and their relatives. At the same time, it provides patients and experts with access to quality information on the treatment of rare diseases in Slovenia. The Minister stated that Slovenia also has a great interest in joining the European Reference Networks, which provide access to state-of-the-art health services to every individual, regardless of where they live in the European Union. She shared the information that nine Slovenian healthcare providers applied successfully in nine different reference networks.

Following the introductory addresses by the President and Vice-President of the Association for Rare Diseases of Slovenia, Prof. Jože Faganel and Mrs. Majda Slapar, lecturers from the Clinical Institute of Medical Genetics and the University Pediatric Clinic UKC Ljubljana, the National Institute of Public Health and the Health Insurance Institute of Slovenia presented the following achievements and dilemmas from the period since the last year's National Conference on Rare Diseases: reference networks, national contact points, the register of rare and nervous-muscular diseases, and the financing of the treatment of rare diseases in Slovenia.

In the second part of the conference, lecturers from the University Pediatric Clinic UKC Ljubljana, the Idrija Psychiatric Hospital, the Oncology Institute of Ljubljana, the Scientific Research Center of Koper and the Medical Ethics Committee of the Republic of Slovenia discussed the integrated treatment of congenital rare pulmonary diseases, rare diseases in psychiatry, diagnosing congenital diseases, the inclusion of patients in research, the philosophical view of ethical dilemmas, the role of the national ethics commission in addressing rare diseases, as well as the legislation and modern therapeutic options for rare diseases.

After the discussion, an example of good practice Debra Croatia was presented, followed by a round table, which was moderated by Prof. Jože Faganel and attended by doctors, as well as patients with Gaucher's disease, myodysplastic syndrome, hereditary angioedema, and transthyretin amyloid polyneuropathy (TTRFAP).

The Association of Patients with Blood Diseases Slovenia has been paving the way for a public dialogue on rare diseases by organizing conferences and publishing Conference Proceedings for many years. On the occasion of the conference, the Association of Patients with Blood Diseases Slovenia presented the book entitled "Rare Diseases February 28, 2018 - Eighth Edition", which was available to all conference participants; it is also available in electronic form from the Association's website (www.drustvo-bkb.si). The book's forewords were written by the Slovene Minister of Health, Mrs. Milojka Kolar Celarc, and the Vice-President of the Association for Rare Diseases of Slovenia, Mrs. Majda Slapar. This is followed by the interview with the President of the Association for Rare Diseases of Slovenia, Prof. Jože Faganel.



Minister of Health Mrs. Milojka Kolar Celarc and the Vice-President of the Association for Rare Diseases Slovenia, Mrs. Majda Slapar

The central part of the publication presents the program of the national contact point for rare diseases and the current level of development of the register of rare diseases and the register of children with neuromuscular diseases, as well as the solutions to their overall treatment. The booklet also presents rare diseases in psychiatry, the problems of early diagnosis of congenital diseases, the dilemma of involving patients with rare cancers in research, as well as the ethical aspects of rare diseases. The booklet also presents rare diseases in psychiatry, the problems of early diagnosis of congenital diseases, the dilemma of involving patients with rare cancers in research, as well as the ethical aspects of rare diseases. Hereditary angioedema, amyloidosis, disseminated plasmocyte and non-hodgkin's lymphoma are diseases that are presented separately in the Proceedings. The Appendix of the book defines and describes rare diseases, presents drugs for the treatment of rare diseases - orphan drugs, rare disease research and projects, and international meetings. The Appendix also contains a list of different medicinal products to treat rare diseases, as well as the presentation of all collaborating associations - members of the Association for Rare Diseases of Slovenia.

The fourth National Conference on Rare Diseases firmly endorsed the slogan of the Rare Disease Day 2018 - "**Show your rare. Show you care.**", thus giving a promising message to over 30 millions of patients with rare diseases across Europe, as estimated by EURORDIS.