6th NATIONAL CONFERENCE TO MARK RARE DISEASES DAY

Tuesday 3rd March 2020
Venue: Congress Centre Brdo
Session hall: SPLENDENS

9:00 – 9.30 Registration

A. INTRODUCTION

Prof. Jože Faganel, Association for Rare Diseases of Slovenia

9:50 – 10:10 European Commission and rare diseases
Mag. Tanja Fajon, MEP

10:10 – 10:30 European reference networks and project RARE 2030
Doc. dr. Mojca Žerjav Tanšek, dr. med., University Children's Hospital, University Medical Centre Ljubljana

10:30 – 10:40 Presentation of the European Reference Network (ERN) for all patients with one of the rare genetic tumor risk syndromes: ERN GENTURIS
Doc. dr. sc., B, Mateja Krajc, dr. med., Institute of Oncology Ljubljana

10:40 – 10:50 National Register of Tested Persons of Families with Hereditary Cancer
Simona Hotujec, mag. manag. kak., Institute of Oncology Ljubljana

B. TREATMENT OF RARE DISEASES IN SLOVENIA

10:50 – 11:10 Slovenian health insurance system and rare diseases
Marjan Sušelj, univ. dipl. psih., General Manager of Health Insurance Institute of Slovenia

11:10 – 11:30 Health Council of the Republic of Slovenia and rare diseases
Akad. prof. dr. Franc Strle, dr. med.

11:30 – 11:50 Medication from the invention to the finished, protected product
Prof. dr. Lovro Stanovnik, dr. med.

11:50 – 12:10 Break
12:10 – 12:30  **Regulatory mechanisms: the path from product to patient**  
Nada Irgolič, mag. farm., spec.

12:30 – 12:50  **Pharmaceutical Ethics**  
Prof. dr. Mitja Kos, mag. farm., Faculty of Pharmacy, University of Ljubljana

12:50 – 13:10  **Ethical considerations from the perspective of compliance patients**  
Doc. dr. Božidar Voljč, dr. med., Predsednik Komisije RS za medicinsko etiko

**C. INSIGHT INTO LESSER KNOWN RARE DISEASES**

13:10 – 13:30  **Hereditary angioedema**  
Doc. dr. Mihaela Zidarn, dr. med., Hospital Golnik

13:30 – 13:50  **Rare diseases in the oral cavity**  
Prof. dr. Matjaž Rode, dr. dent. med.

13:50 – 14:10  **Langerhans histiocytosis**  
Mihela Šajn, dr. med., University Medical Centre Ljubljana

14:10 – 14:30  **Gaucher disease**  
Prof. dr. Samo Zver, dr. med., University Medical Centre Ljubljana

14:30 – 14:50  **Achondroplasia**  
Doc. dr. Primož Kotnik, dr. med., University Children’s Hospital, University Medical Centre Ljubljana

**D. LIVING WITH RARE DISEASE**

14:50 – 15:10  **Experience of the values of life with Amyotrophic lateral sclerosis**  
Doc. dr. Blaž Kortinik, dr. med., University Medical Centre Ljubljana  
Damijan Škrbina, Muscular Dystrophy Association of Slovenia

15:10 – 15:30  **Concluding remarks**  
Prof. Jože Faganel, president of the Association for Rare Diseases of Slovenia

15:30 – 16:30  **Lunch and informal socializing**