

Subject: <b>Rare Diseases</b>	Subject type: <b>elective</b>
Study year: 5 <sup>th</sup>	Content: <b>winter time</b>
Study program: <b>General Medicine</b>	

## **Aim of the course**

Elective subject Rare Diseases for student of study program General Medicine content from practical lessons. The aim is to teach students the basics of knowledge about Rare diseases. This course provides an introduction to rare diseases, their screening, diagnosis and treatment in general. National and transnational registers, as well as, social issues. Students will learn about the characteristics, clinical picture, diagnostics treatment and prognosis of the most common rare diseases – inherited metabolic disorders, endocrine diseases, cystic fibrosis, neurological diseases with emphasis on the pediatric patient.

**Education:** 14 hours practical lessons

**Assessment:** oral test

## **Syllabus**

- Introduction into the topic of Rare Diseases
- Newborn screening, diagnostics tools
- Therapy, new treatment modalities
- National and international registers, orphannet project
- Patients organisations, social issues
- Inborn errors of metabolism
- Endocrine rare diseases
- Neurological rare diseases
- Cystic fibrosis
- Metabolic diseases of the liver
- Rare genetic syndromes
- Rare immunodeficiencies
- Rare kidney diseases
- Rare malignancies