Subject:	Rare diseases		
Study	General Medicine	Study Period:	Winter time
Evaluation:	Graduated	Subject Type:	Elective
Content:	1 h practical/seminar / week		Total 14 hours

## Department: Department of Paediatrics and Adolescent Medicine UPJŠ FM

Week	Practical Exercises
1.	<ul> <li>Introduction into the Subject Rare Diseases</li> <li>Newborn screening, diagnostics tools</li> </ul>
2.	<ul> <li>Therapy, new treatment modalities</li> <li>National and international registers, Orphanet project</li> <li>Patients organisations, social issues</li> </ul>
3.	- Inborn errors of metabolism
4.	- Endocrine rare diseases - Neurological rare diseases
5.	<ul> <li>Cystic fibrosis</li> <li>Metabolic diseases of the liver</li> </ul>
6.	- Rare genetic syndromes - Rare immunodeficiencies
7.	- Rare kidney diseases - Rare malignancies