4th Baltic Paediatric Congres May 2019

CHALLENGES IN PAEDIATRICS: FROM RARE TO CARE

The child with rare disease in primary care

United Nations Sustainable Development Goals, Primary Care, New Diagnostics

The United Nations NGO Committee for Rare Diseases has associated 6 out of the 17 sustainable development goals as specifically relevant to be achieved for people with a rare condition *# 1 End Poverty in all its forms everywhere, # 3 Ensure healthy lives and promote well-being for all at all ages, # 4: Ensure inclusive and equitable quality education and promote lifelong learning opportunities for all, # 5: Achieve gender equality and empower all women and girls, #10: Reduce inequality within and among countries, #17: Revitalize the global partnership for sustainable development.*

How do we, pediatricians, act in line with the SDG? Manage the child with genetic diagnosis without suitable clinical diagnosis or management guideline. Support children at special school appropriately and help understand their learning problem? How can we support the mothers of a child with a rare condition? Facilitate their life as a woman as well as a professional, and reduce her societal isolation?

Duchenne's disease: Model of interdisciplinary approach and networking

At the Medical University of Gdansk over 90 children with Duchenne muscular dystrophy are managed by a multidisciplinary team. The team is following international guidelines and is facilitated by a digital network to provide care at home. Over the years the team has built a strong relationship with the patient organization.

Familial Mediterranean fever in children

In Armenia Familial Mediterranean Fever (FMF) is a more common rare condition. At the Arabkir Medical Centre - Institute of Child and Adolescent Health a special clinic is dedicated to the care of children with FMF which currently serves 3500 < children until age of 18. A model of follow up care for these children has been developed including regular check-ups and providing treatment with Colchicin. An essential part of care is psychologic support aimed at families and children. Meanwhile, some families with FMF are stigmatized and try to avoid involving in care local health care providers, because of fears that neighbors and community members will know about the disease.

7 years old child with Coffin-Siris syndrome: First case in Baltic countries

The search for the diagnosis started when the child presented with dysmorphic features and a global developmental delay. The child was referred to the clinical geneticist and chromosome studies where performed, no abnormalities. Because of hepato- and splenomegaly a mucopolysaccharidose was considered, metabolic studies where normal. At the age of 3, Coffin-Siris syndrome was diagnosed. With this diagnosis a care management can be provided.

Survey of attitude of primary care doctors towards follow-up of cancer survivors

In 1981 a pediatric hemato-/oncology center was established. Since 2002 bone marrow transplantations are performed in Lithuania. Recently a study was performed among primary care doctors on their awareness and follow up on survivors of childhood cancer.