Pediatricians caring for children with Rare Diseases in Europe, fall 2018

Portoroz, Slovenia 27 September, the Slovenian pediatrics society pays attention to rare diseases at their annual congress. Reimbursement of the **expensive orphan drugs** is a subject of debate in each EU member state. Leading to frustration and anxiety of the patients and their families, who hope for a cure. Slovenia is at the top of the European member states meeting the needs of its citizens. Children with spinal muscular dystrophy in Slovenia get the novel drug Nusinersen. While for example in the Netherlands there is an upper age limit for the novel drug therapy. Inequalities in reimbursements are difficult to understand. How do you explain to a Dutch girl of 13 years with a lung function of about 30% due to spinal muscular dystrophy that she is too old for the new treatment ?

Utrecht, The Netherlands 31 October, the kick-off meeting of a revision of the Dutch Down syndrome guideline with the GRADE method. The third edition is an initiative of the Down syndrome multidisciplinary working group. Issues as screening for feasible thyroid function test and best support for the week ankle joint will be reviewed. The **awareness of need of social support** of families is rising. The birth of a child with special needs seem to put constraint of caregivers, the parents. With over 20 years' experience the Dutch pediatric lead Down syndrome working group hopes to contribute to better health and care children with Down syndrome around the world.

Hannover Zoo, Germany 3 November, Families with Shwachman Diamond Syndrome travelled over 500 kilometers distance to learn and meet other families. Exchanges on **daily live experiences** is one of the most important for people living with a rare condition. How to deal with obvious symptoms as failure grow and fatty smelling stools, not recognized by your doctor? Specialist pediatrician and dietician answered many questions as on when to take the pancreas enzymes, the need of hematological checks. Still much is unknown. International registries can improve care management.

Gdansk, Poland 10 th -11th November. Families and clinicians gathered to talk about supporting care of patients and with rare diseases, Duchenne muscular dystrophy and other types of muscular dystrophies. For most individuals with a rare condition a **multidisciplinary** approachis essential. Health will improve when data are collected during lifetime. The cardiologist is performing an ECHO measuring the cardiac ejection fraction and introduces medications to prevent cardiomyopathy and tachycardia. The pulmonologist controls the lung capacity to adequately provide respiratory rehabilitation if needed. The endocrinologist wants to follow child's growth, puberty and bone density. When new medicine evolve their effect should be measured with objective data, such as motor function expressed by e.g. distance to walk. An innovative IT application integrates care offered by the multidisciplinary hospital team with care at home to provide individualized medicine. Person-centred care is realised when patients and their families themselves are facilitated to collect and get insight in their personal data.
Question is arising who will provide the sustainable finances for the implementation of personalized medicine.

Jolanta Wierzba, paediatrician and co-founder of EAP Rare Disease WG, Ewa Pilarska –

neurologist, Joanna Kwiatkowska and Jaroslaw Meyer- Szary pediatric cardiologist, Joanna Minkowska – endocrinologist, Eliza Wasilewska pulmonologist, Joanna Jablonska- Brudlo – specialist in physical medicine and rehabilitation, Agnieszka Sidorkiewicz- Szlagatys – gastroenterologist, Arkadiusz Manski – psychologist, Sylwia Małgorzewicz and Edyta Wernio – specialists in nutrition and dieticians, Liesbeth Siderius, paediatrician EAP Rare Disease Katarzyna Witkowska co-ordinator of Rare Disease Centre Juliusz Chorążewicz – ophthalmologist

November 27th 2018