'Early childhood disabilities, rare diseases and guidelines; a rare-best practices session'.
6th Congress of the European Academy of Paediatric Societies (EAPS) in Genève
Friday 24th October 2016 chairs: Arunas Valiulis and Liesbeth Siderius

Bettina Schwethelm PhD, MPH *Young Child Health and Development Specialist*
UNICEF Regional Office for CEE/CIS; United Nations Children's Fund (UNICEF) Geneva, Switzerland
Young children with disabilities are among the most socially excluded children in the 21 countries of CEE/CIS (Central, Eastern Europe & the Commonwealth of Independent States) region. Such infants and young children are often placed in residential care and do not benefit from social inclusion, including mainstream education. To promote the early identification of young children with disabilities and/or at-risk for developmental difficulties, UNICEF is prioritizing two areas with government partners and other stakeholders:

- Reform of maternal and child home visiting systems and capacity building of home visiting professionals (often nurses) in more than a dozen countries to ensure that home visits address issues related to comprehensive young child health, development, and and wellbeing and identify young children at risk; and
- The introduction of developmental pediatrics, family-centered services in line with ICF-CY, and the use of basic validated tools to monitor and assess child development.

As UNICEF, is looking at referral pathways and capacities at the primary care level, a partnership with professional associations, such as EAP can contribute importantly to building the capacities of practitioners in early childhood identification and intervention.

Erik and Marian de Graaf – *Founders of Stichting Downsyndroom (SDS) and pioneers advocating for persons with Down syndrome in Netherlands, Europe and worldwide for more than 30 years, Wanneperveen, Netherlands*
From learning to live with the diagnosis, via early intervention to family involvement in guideline development, a lifelong experience.
The experiences after the birth of their own son in 1984 led to the establishment of the Dutch Down Syndrome Foundation (SDS) in 1988. The authors' search for information across borders lead, via the SDS, to the introduction of early intervention programs for parents, the first syndrome specific guideline and ditto medical teams, support for inclusive education and valuable scientific research. Finally, the authors, based on their experience, address other parents with suggestions on how to unite and initiate their own syndrome specific organizations and in such a way contribute to person-centered health care. They stress that much can be done already even without financial support.

Professor **David Neubauer**, MD, PhD *Department of Child, Adolescent & Developmental Neurology University Children's Hospital, Ljubljana, Slovenia* Rare neurological diseases in Slovenia , how guidelines support care provision in smaller countries.
Presented first some guidelines of RBP project and IRDiRC guidelines and than talked about the frequency of RND in Slovenian children and evaluate the burden of RND in respect to QoL. 100 children with RND at University Children's Hospital in Ljubljana were included and evaluated for their level of health condition by 5-level scale. Strength and Difficulties Questionnaire (SDQ) was used for evaluation of QoL. Parents were also asked whether they preferred home care to hospital care.
The prevalence of RND in Slovenia is similar to that in Europe. The average level of damage of children with RND is 2.08. The participants in the research who suffer from neurometabolic diseases have the highest level of damage ($p<0.010$) compared to those with all other RND. We have not confirmed that QoL of children with RND is low. In 66.7% parents prefer home care.
In conclusion:
These are the first epidemiological data of some of the RND in Slovenia that can be compared to European data. Neurometabolic diseases cause the highest level of disability, but the higher level of

disability does not mean the lower quality of life. Parents wish for less in-patient hospital care and more hospital at home.

Liesbeth Siderius, MD, *Youth Health Care GGD IJsselland, Kampen, Netherlands and EAP representative in RareBestPractice Project and*
Karen Ritchie, *Healthcare Improvement Scotland (Scottish Intercollegiate Guidelines Network) Glasgow, Scotland, partner in RareBestpractice Project* RareBestPractice Guidelines as an aid to harmonize child health care in Europe.
The Rare best practice project aims to collect best practice guidelines and identify rare disease research needs. The ultimate goal is facilitate timely, effective and efficient translation of research into patient oriented clinical and public health practice.
Over 200 rare best practice guidelines are identified and systematically appraised with the AGREE tool.
The EU commission Expert Group on Rare Diseases recommends a multidisciplinary, holistic, continuous, person-centered and participative care to people with rare diseases and full realization of their fundamental human rights. The EAP advocates for international IT standards in open vendor independent IT networks to facilitate person-centered , participative care.

November 2016