

May 18th 2018: 13.15- 14.45 Sofia, Bulgaria
Spring Meeting European Academy Paediatrics
Rare Diseases/ Ethics Working Group
Session chair: Liesbeth Siderius

Rare Diseases in Lower and Middle Economies

"Combating inequalities of child health: specific problems and solutions in lower economies of European countries, towards a global strategy".



Introduction Rare Diseases in Bulgaria

Professor Rumen Stefanov, MD, PhD

Professor of Social medicine and Public Health,
Medical University of Plovdiv, Bulgaria
Director, Institute for Rare Diseases, Bulgaria

Bulgaria has a national plan for rare disease. Most EU member states have developed rare disease plans. The plans differ per member state. The national plan needs political and social support, support of the medical community and support of the national patient alliance for rare diseases.

Expert Rare diseases Medical centers in Bulgaria are participating in European Reference Networks.

In Bulgaria studies are performed on access to orphan drugs.

Diagnostics and Therapies in Macedonia

Velibor Tasic, Macedonia, RD WG co-founder

In Macedonia diagnostic facilities for accurate diagnosis of rare diseases are limited due to lack of the comprehensive neonatal screening program, sophisticated metabolic studies and minimal financial coverage for the genetic tests. A few experienced pediatricians manage to describe new rare diseases entities with the help of a large network of colleagues in Europe and North America performing metabolic studies and DNA investigations for them.

In the meantime the government has stopped the national screening program on PKU. Few patients have access to orphan drugs through the national fund for rare diseases.

Screening program to detect rare diseases in Bulgaria

Professor Iva Stoeva, Bulgaria

In Bulgaria neonatal screening is performed on PKU, and congenital hypothyroidism since the 90's, and since 2010 also for congenital adrenal hyperplasia. Most dried blood card arrive < 11 days, but not all, due to different reasons. An unsolved question is the availability of Hydrocortisone in the country, as well as the pending introduction of additional diseases in the neonatal screening.

Gaps in the education and teaching of rare diseases

Professor Violeta Iotova, Bulgaria

Bulgaria is participating in ERN's. Recent Survey showed gaps in training at specific age groups (neonates, adolescents) and in specific areas (transition). The representatives of the ENDO-ERN collaborate with general pediatricians in the regions. Boosting rare diseases educations at different levels (medical schools, trainees in pediatrics, GPs, patients, general public, etc.) is an immediate task. Pediatric specialties curriculum is an important tool as well as on-line and easy accessible platforms. Suggestions on how to collaborate with EAP are welcomed.

Pediatric Rare Diseases in Sri Lanka

Professor S P Lamabadusuriya, Rare Forum, Colombo, Sri Lanka

The Sri Lankan Pediatric Society has established a Rare Forum. Pediatricians face numerous challenges in diagnosing and managing rare diseases due to the lack of experience and resources. The aim of the Rare Forum is to improve clinical diagnosis, investigation, management and reporting of rare diseases in Sri Lanka.

Read more in the first newsletter of the Forum, available at EAP website.

May 25th 2018

References:

December 2017,

World Bank and WHO report reveals that " currently, 800 million people spend at least 10 percent of their household budgets on health expenses for themselves, a sick child or other family member. For almost 100 million people these expenses are high enough to push them into extreme poverty".

<http://www.worldbank.org/en/news/press-release/2017/12/13/world-bank-who-half-world-lacks-access-to-essential-health-services-100-million-still-pushed-into-extreme-poverty-because-of-health-expenses>

March 2018

Unicef report : Progress for every child in Sustainable Development Goals (SDG) era

https://data.unicef.org/resources/progress-for-every-child-2018/?utm_source=email&utm_medium=publications-newsletter&utm_campaign=SDGs

Ensure healthy lives and promote well-being for all at all ages

Internationally comparable data and global targets