

Rare diseases - common problems in Bulgaria

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- Rare diseases pose a great challenge to our society
 - “Rare diseases” - a prevalence of fewer than 5 per 10,000 in the community.
 - Rare diseases affect 6-10% of the population
 - EURORDIS estimates that there exist between 5,000 and 7,000 distinct rare diseases
 - At least 80% of them have identified genetic origins

Strategy for rare diseases in Bulgaria

- Project of National Program for Rare Diseases 2007-2011- November 2006
- March 2008- the Healthcare Committee at the Bulgarian National Assembly started discussion on issues concerning prevention, diagnosis, treatment and rehabilitation of people with rare diseases in Bulgaria
- September 2008- Mr. E. Zhelev, Minister of Health, has approved the National Programme for Rare Diseases (2009-2013)

Actions, regarding rare diseases

- Diagnostic genetic services- 1971
- Neonatal screening
 - PKU- 1978
 - Hypothyreoidism- 1993
- Prenatal screening- 1982

Treatment (clinical management)

- The Orphan Drug Act of 1983
- Expert panels
- Ministry of Health cover the treatment of 10 rare diseases- cystic fibrosis, MPS I, II, VI, M.Pompe, GH deficiency, PWS, arterial hypertension, acromegaly and thalassemia.
- Problems to solve-
 - to improve the treatment control
 - to optimise the treatment algorithms
 - to evaluate the treatment effects
 - to ensure the best treatment for our patients.

Provision of information for rare diseases

- Information Centre for Rare Diseases and Orphan Drugs (ICRDOD)
- National network of information-reference centers for rare diseases.
 - To disseminate and implement of the NCCRD's programs
 - They will be a regional unit of the National Register for Rare Diseases
 - A place for assistance and training of general practitioners.

Prevention

- Extending the available neonatal screenings for congenital hyperthyroidism and phenylketonuria and the first and second trimester screening for Down syndrome.
- Introduction of screening practices for 21 alpha hydroxylase deficit
- Training of staff for the needs of the medical genetic consultations
- **Late diagnoses**
 - lack of information
 - lack of appropriate medical training
 - difficulties in accessing care

Patients organisations

- 1st National Conference of Patients with Rare Diseases - 14 April 2007 in Sofia.
- 20 patients organizations
- The main purpose of the National Alliance is to be a link between the people with rare diseases and the representatives of the social and healthcare system.
- The Alliance works for the protection of one of the principal human rights – the right of timely and equal medical care.

Specialised social services

- Integrated approach for prevention, diagnostics, treatment, and social integration of patients with rare diseases
- Services for people suffering from rare diseases and their families:
 - temporary care
 - information and counseling services
 - improving the financial situation

Thank you!



gene therapy