

Sharing best practices on integrative approach to rare diseases in different countries



Hellas (Greece)

Vasilios Berdoukas
Thalassaemia Unit
1st Department of Paediatrics
University of Athens
“Aghia Sophia” Children’s Hospital

Strategies for rare diseases



- ***A rare disease is defined as one which affects 1 in 2000 citizens of the country according to EURORDIS definitions.***
- ***In Greece the laws etc. are governed by the EURORDIS regulations.***
- ***In Greece, the carrier state for β -thalassaemia is present in 8% of the population and the severe disease affects approximately 3,500 patients. The number of patients with Sickle Cell syndromes is less than 500.***
- ***Comprehensive care for thalassaemia is available for all patients. Transfusions and chelation therapy are government guaranteed.***



Funding of actions, regarding rare diseases



Funding Processes for thalassaemia and sickle cell disease:

- *Prenatal screening – Health Department through Central and Regional Thalassaemia Centres as well as from University Centres*
- *Neonatal screening: Not for thalassaemia and sickle cell disease*
- *Diagnostic genetic services: Health Department, University Departments and private services (user pay).*
- *Treatment (clinical management): Health Department, Health Funds and Department of Social Welfare.*
- *There are 641 Orphan drugs + all theoretically available in the country and paid for usually by the patient's Health fund. For thalassaemia only Exjade (Deferasirox) and Sildenafil are available as orphan drugs, the former for iron overload and the latter for pulmonary hypertension.*
- *Rehabilitation: Health Department and Private sector (user pay), rarely needed for thalassaemia*
- *Social care: Department of Social Welfare*

Provision of information for rare diseases



- *Availability of website-based information – International and local websites available for thalassaemia*
- *These websites offer help lines*
- *Availability of an official list of RDs: Yes – this includes thalassaemia and sickle cell disease.*

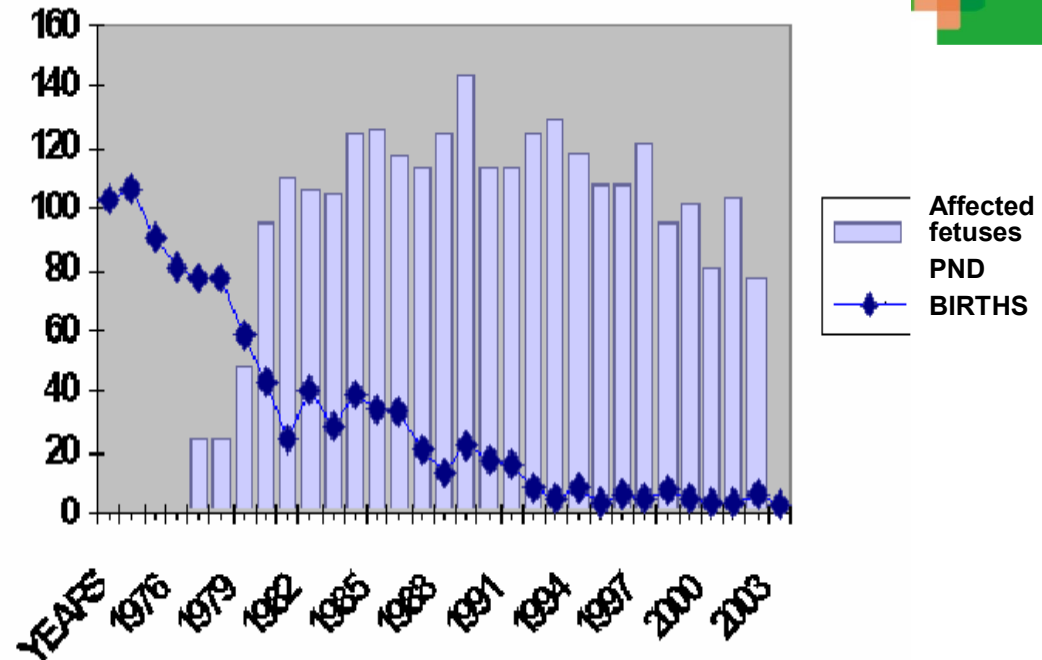
Research on rare diseases

- *Availability of research programme for RDs – only individually based at hospital centres but includes participation in multicentre clinical trials.*
- *Please, provide information on major sources:*
 - *Hospital and University based, supported by the Department of Health, University Medical Schools and pharmaceutical industry.*



Prevention

- **Folic acid: Given for women planning to become pregnant.**
- **Prenatal screening: Effective programme for haemoglobinopathies including diagnostic genetic testing.**
- **Neonatal screening: not for haemoglobinopathies.**
- **Delay of diagnosis – rare**
- **Other preventive practices? For other rare genetic diseases prevention is usually offered after an index case is identified.**



Effectiveness of Greek Prevention Program
Annual PND of Affected fetuses and births of affected infants
National Thalassemia Prevention Center (A. Loutradi, person. com.)

Empowerment of patients organisations



- *Four Groups only registered in Greece for Rare diseases.*
- *For Thalassaemia there is the EOTHA group which is the federation organisation for all the patient organisations both in large cities and regional areas.*
- *All organisations have active representation and consultation. There are numerous information conferences organised annually for patients and their supporters both by the Federal body and by the individual societies.*
- *Recently an European organisation for rare anaemias, based in Greece, has been established (EORA) with a website and general information activities.*

Specialised social services



- *Respite Care Services: rarely needed for haemoglobinopathies*
- *Therapeutic Recreational Programmes: As most patients with thalassaemia are now adults such activities are not usually undertaken. Patient support groups at conferences include significant interaction and recreational activities.*
- *Services aimed at the integration of patients in daily life. Social tourism support (Department of Social Welfare), tax reduction (income tax reduction, duty free vehicles and no payment of annual registration fees), disability allowance according to % of disability and early pensions. In public departments there is obligatory employment of a percentage of patients for all disabilities.*
 - *An essential component of integration is the provision of services that minimise the patients time spent in hospitals as well as providing services that do not interfere with the ability to work and need the patient to take time off work.*