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

Republic of Macedonia

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Strategies for rare diseases

- ***According to the New RDs program introduced in RM rare diseases are defined as less than 1:20 000***

- ***Many rare diseases are covered by specific programs:***
  - *GH deficiency*
  - *Familial colon carcinoma*
  - *Cystic fibrosis*
  - *Rare malignant diseases*
  - *Congenital heart malformations*
  - *Immunodeficiencies*

Program for treatment of rare diseases
April, 2009

M Gausher	1:50 000-100 000
Hereditary angioedema	1: 5 000
Multiple myeloma	5:100 000
Phenylketonuria	1:20 000-1:100 000
Myelodysplastic syndrome ?	
Acromegaly	1:15 000
Gatrointestinal stromal tumors GIST	14.5:1 000 000
Gastroenteropancreatic neuroendocrine tumors	2.5-5:100 000
Mucopolysacharidosis	1:25 000 (1:150 000)
Hereditary tyrosinemia	1:100 000
Galactosemia	1:260 00-1:500 000
Hereditary fructose intolerance	1:20 000

Funding of actions, regarding rare diseases

How different issues, concerning RDs are funded:

- *Prenatal screening*
- *Neonatal screening*
- *Diagnostic genetic services-Health fund, scientific projects*
  MANU, Pediatric Clinic
- *Treatment (clinical management)-Health fund*
- *Orphan drugs- Health fund, donations*
  Mucopolysacharidoses
  M. Gausher-donation
- *Rehabilitation – Health fund*
  Center for physical rehabilitation, nonspecific
  Center for care and early intervention in children with delayed development
- *Social care - Project , Humanitarian aid*
  Day care centers for children with different causes of mental impairment including syndromes

Provision of information for rare diseases

- *Availability of website-based information web-site "pediatric health"*
- ***Availability of an official list of RDs***

Preparation in process

Research on rare diseases

- *Availability of research program for RDs*
  Scientific projects - international collaboration

  OI
  Cistinuria
  Tubulopathies
  Neonatal diabetes
  Other

Prevention

- *Folic acid*
- *Prenatal screening*
  - *Major malformations, compulsory ultrasound check up*
- *Neonatal screening*
- *Delay of diagnosis*
  - *Training of doctors*
- *Other preventive practices*
  - *Prevention of fractures in OI by therapy with bisphosphonates*
  - *Prenatal diagnosis of CAH for early treatment*
  - *Prenatal diagnosis of chromosomal abnormalities*

Specialised social services

- *Therapeutic Recreational Programmes*
- *Services aimed at the integration of patients in daily life*

Empowerment of patients organisations

- ***Support to the activities of patient organisations***
  - ***Cystic fibrosis***
  - ***GH deficient children***
  - ***Initiative for RDs parent organization***
- ***Representation and consultation of patient organisations***

Republic of Macedonia

Experience

size of the country	-
centralized care	+

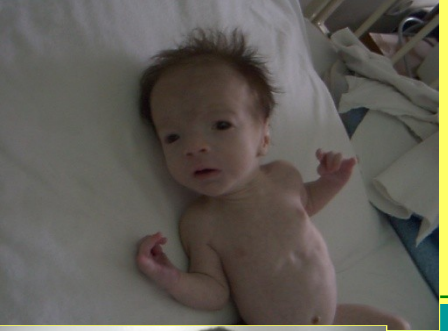
Diagnostic tools

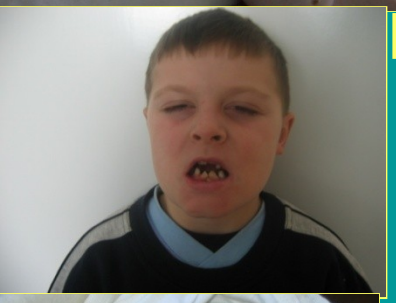
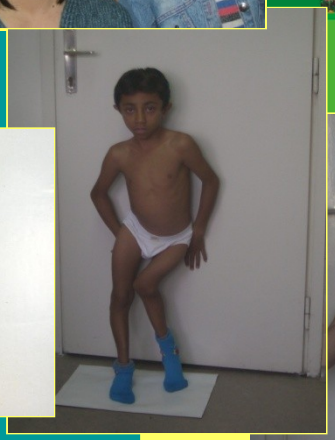
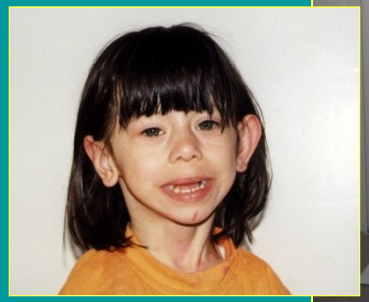
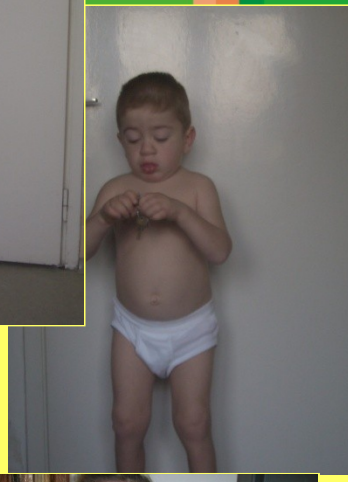
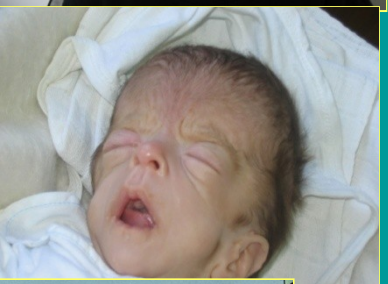
biochemistry	-
molecular diagnostics	+

Therapy

medication (CAH, storage diseases)	
devices	-
surgery	±
specialized programmes	±

Dysmorphology unit
Paediatric Clinic



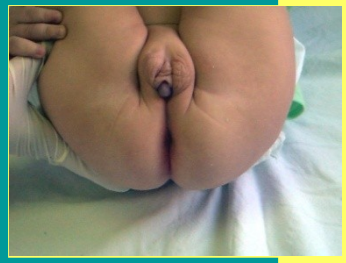
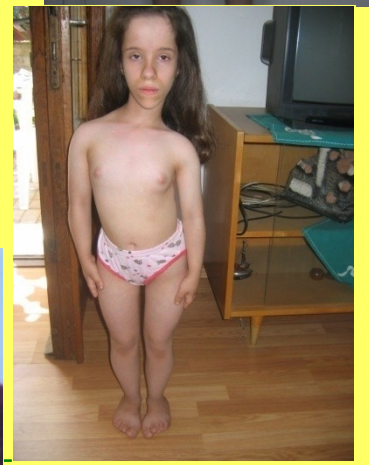
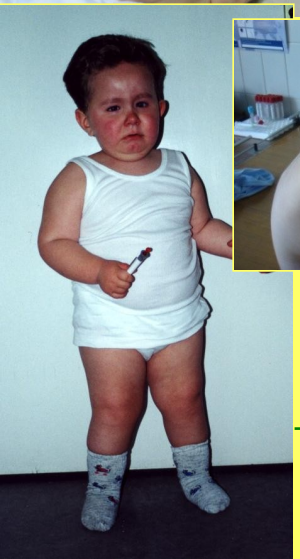







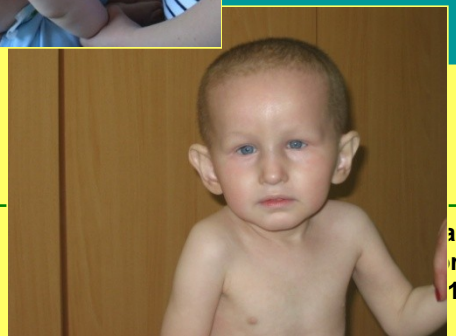
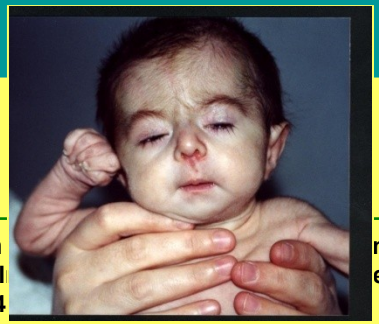
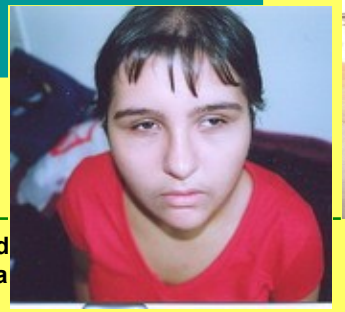
Detection of rare syndroms

- Williams
- Prader Willi
- VCF syndrome

Detection of extremely rare syndroms

- Cohen syndrom
- Alstrom syndrom
- Syndromic obesity
- Sexual ambiguities

In collaboration with

EUROPLAN
European Project for Rare Diseases National Plans Development

Expertise

Paediatric Clinic - diagnosis and treatment

Congenital adrenal hyperplasia (Serbia, Montenegro, Kosovo)
Hemoglobinopathies
Cystic fibrosis
Osteogenesis imperfecta
Rare multimalformative syndroms
Hyperinsulinemic hypoglycemias
Sexual ambiguities
Rare tubulopathies
Immunodeficiencies
Rare neurologic disorders

MANU-Molecular diagnosis

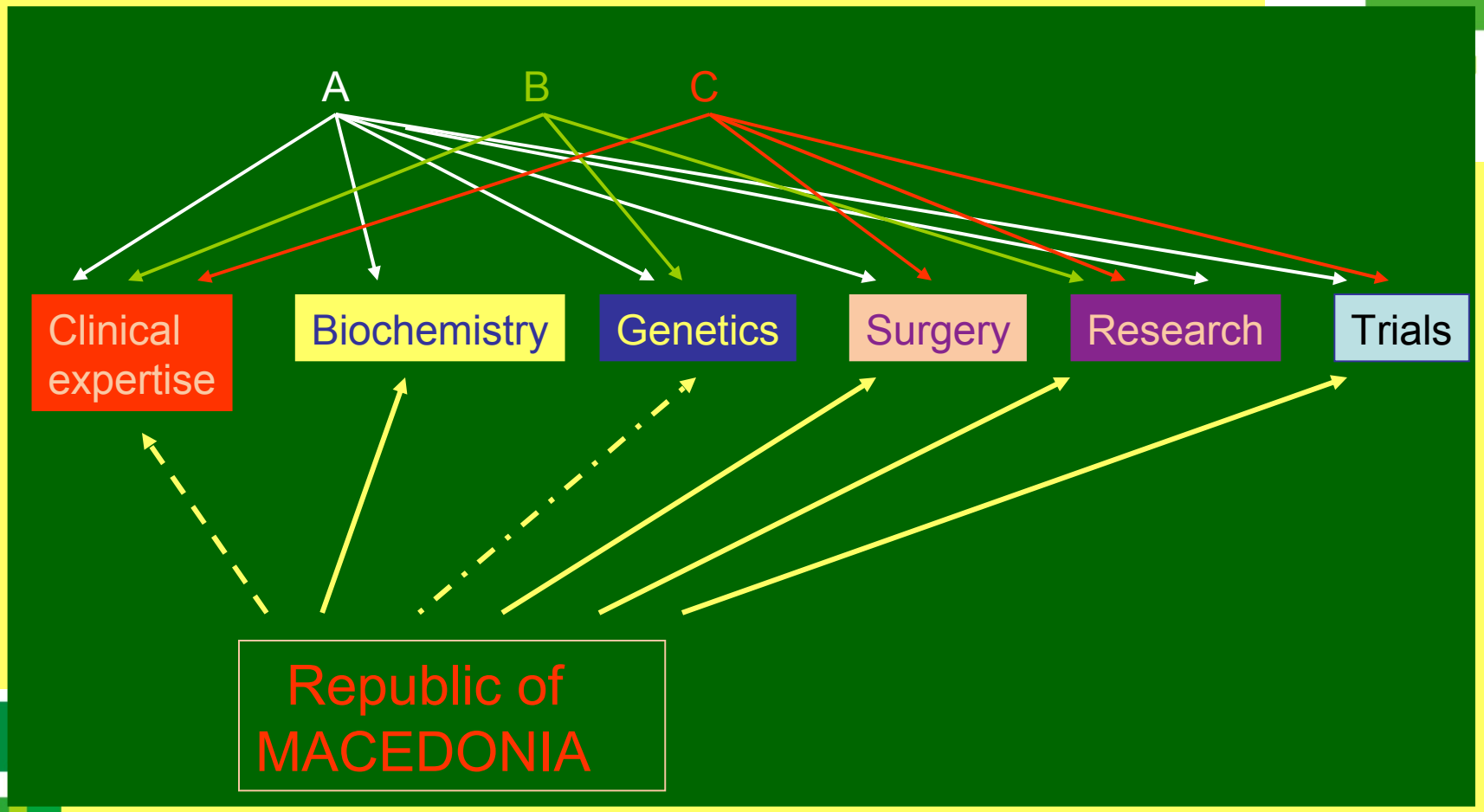
Cystic fibrosis
Hemoglobinopathies
Haemophilia
Haemochromatosis
Fragile X Syndrome
Male infertility
Cystinuria
Muscular dystrophy
Spinocerebelar ataxia
Spinal muscular
Hungtington disease
Androgen insensitivity

Faculty of Farmacy

Familial Colon Carcinoma

SUGGESTIONS

Networking within the region



Challenges

- To cover rare or extremely rare diseases?
- To cover “promising” patients?
- How to set up priorities (diagnosis, treatment or rehabilitation)?
- How to provide justice with limited financial resources?
- How to present “cost-benefit” issues?
- How to create centers of excellence, and how to transfer patients