The Information Centre for Rare Diseases and Orphan Drugs as a Promoter of Research of Rare Diseases

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www.raredis.org
Analysis of a questionnaire to 372 information services on rare diseases

- From September to December 2003
- A questionnaire sent to 1402 information services in 20 EU countries
- A survey of organisations, their needs, sources, tools, services and expectations

EURORDIS PARD III: A PROJECT SUPPORTED BY THE RARE DISEASES PROGRAMME OF DIRECTORATE C OF THE EUROPEAN COMMISSION, AND ASSOCIATION FRANÇAISE CONTRE LES MYOPATHIES

Source: EURORDIS. Complete report available from www.eurordis.org
Information Services: a European survey

Key results:
- 372 respondents from 18 countries
- 62% dealing with a single rare disease
- 5% (~18) dealing with >50 diseases
- 26% with national funding
- only 2% (~7) with EC funding

Source: EURORDIS. Complete report available from www.eurordis.org
Estimated Prevalence of Rare Diseases in Europe

Extrapolations based on the assumption of 9.19% prevalence rate of rare diseases.
Source: http://www.wrongdiagnosis.com/r/rare_diseases/stats-country.htm
The Number of Specialized Centers for Rare Diseases in Europe is Extremely Limited

However, their impact on the public health is enormous:

- **First**, they provide patients, relatives and doctors with up-to-date high-quality information about the conditions of interest.

- **Second**, they systemize and create databases of hospital settings, genetic and clinical laboratories and prominent medical teams, occupied with a given rare disease research or study.

- **Third**, they provide evidences about the epidemiology of rare diseases.

- **Fourth**, these centres serve as a bridge between the patients with rare diseases and the public health care systems by advocating for the basic human right to receive contemporary and equal medical care.
Information Centre for Rare Diseases and Orphan Drugs (ICRDOD)

- **Unique for Eastern Europe** (40% of the European population)
- Operates in **Bulgarian and English languages** (projected to increase the number of languages with Russian and Greek)
- Provides **free information** to patients and medical professionals with educational purposes
- Builds **databases** of doctors, associations, clinical centers and clinical trials
- Provides data on the **epidemiology** of rare diseases
- **Workshops, conferences and research** on rare diseases
- Increases the **public awareness** on rare diseases
- **Lobbying** for adequate national health policy for rare diseases

[www.raredis.org](http://www.raredis.org)
Organization of Work

Forwards the inquiry to a consultant in:
- genetics
- pediatrics
- internal diseases
- surgery

Information sources used:
- MEDLINE and OMIM
- Specialised medical literature
- Internet information sources

Medical secretary

Personalized reply

Local database
DISORDER: Parry–Romberg syndrome (PRS)

SYNONYMS: progressive hemifacial atrophy

DEFINITION: This syndrome is characterized with slowly progressive atrophy of the soft tissues of essentially half the face, accompanied usually by contralateral Jacksonian epilepsy, trigeminal neuralgia, and changes in the eyes and hair. It was first described by Parry (1825) and Romberg (1846) and it has been known for at least 2000 years. Because of its rarity, the literature on Parry–Romberg syndrome (PRS) largely consists of case reports. There is a prevalence of affected women towards men (M/F = 2/3).

CAUSES: They are not known yet. Autoimmune factors have been suggested but their role is not proved as yet. There is no data to support a direct heritability. A study which set up an internet interview among 205 patients with Parry–Romberg syndrome found that none of them has affected relatives, 3% report one relative with a pronounced face asymmetry but without a hemifacial atrophy (Stone, 2003). There was suggested a primary involvement of connective tissue in this disease (Bandello F at al., 2002). Some authors hypothesize that it could be related to a neural crest migration disorder, from which both fronto-nasal mass and cranial vessels take origin (Pichiecchio A at al., 2002).

GENETIC COUNSELING: Recommended.
AVAILABLE THERAPIES: At the moment there is no reliable therapy which would influence the progress of the disease. A reconstructive surgery is suggested for the advanced atrophy cases. Surgical intervention has to be planned in advance and performed at least two years after the disease becomes stable. There is a disease acceleration during or immediately after pregnancy.

EXPERIMENTAL THERAPIES: N/A

OMIM code: 141300


ICD 10: L94.8

PATIENT ASSOCIATIONS:
http://www.geocities.com/rombergs/

SPECIALIZED CLINICS: The surgical treatment is done in specialized clinics for plastic surgery. Accompanying symptoms are treated in the neurology, endocrinology or rheumatology departments as needed.
An example of reply to a medical professional - III

REFERENCE:

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DATE OF LAST UPDATE: 31.10.2004

DISCLAIMER: The information in this reply is provided completely free of charge for educational purposes and should not be used for self diagnosis and treatment. In the case of a health problem, please contact your doctor!
The Internet Portal: www.raredis.org
The Internet portal – Unique Visitors per Day

Activities for Nov 2004 – April 2005

- Building and running the Internet service – www.raredis.org
- Building team of consultants
- Creating databases of medical professionals, associations and clinical settings
- Established contacts with major national and international scientific and patient associations
- Deposited an official proposal for establishment of National program for rare diseases and orphan drugs at the Bulgarian Ministry of Health
- Organized a workshop on Hereditary Angioedema (11 March 2005) together with the Foundation for the Prevention and Treatment of Fatal Angioedematous Disease, Hungary
- Established and equipped office of the Information Centre
Results from the Workshop on Hereditary Angioedema - 11 March 2005

- Created initiative group for improvement of diagnosis, treatment and healthcare of HAE patients in Bulgaria
- **Establishment of HAE center in Plovdiv**, referring for all Bulgarian HAE patients.
- **Specialized training for two clinicians in Hungary.**
- Dr. Farkas and Varga will provide immunological diagnostic tests for HAE to the Department of immunology at the Medical University of Plovdiv.
- **Increasing the awareness and knowledge about HAE** among medical specialists and patients in Bulgaria – papers, conferences.
- **Genetic counseling** to HAE patients
- **Initiatives for reimbursement** of diagnosis and treatment of HAE patients.
Plan for 2005

- Including rare diseases as a priority area in the National Health Strategy of Bulgaria
- Lobbying for reimbursement of the prophylaxis, diagnosis and treatment of all rare diseases
- Working towards adequate legislation, regarding easier access and registration of orphan drugs
- Close collaboration and integration with similar European organizations and services
Established Contacts, Support and Collaboration

Bulgarian Ministry of Health
Bulgarian Society of Neurology
Bulgarian Society of Cardiology
Bulgarian Society of Endocrinology

Bulgarian Society of Human Genetics

Bulgarian Orthopedic-Traumatology Society

Central & Eastern European Genetic Network

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