

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

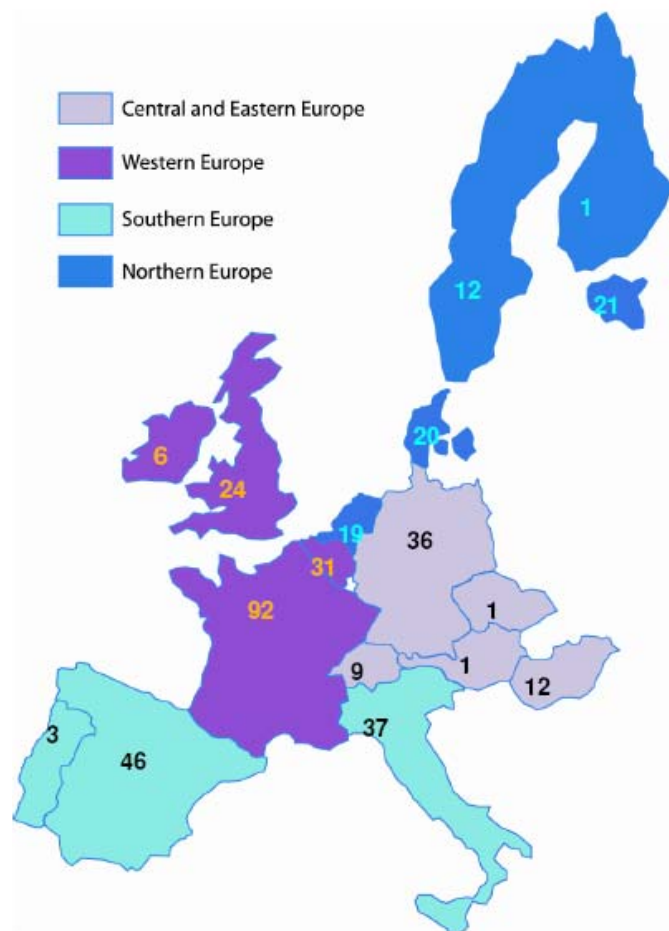
Dr. Rumen Stefanov, MD, PhD

Information Centre for Rare Diseases and Orphan Drugs (ICRDOD)

Bulgarian Association for Promotion of Education and Science

[www.raredis.org](http://www.raredis.org)

# Information Services: a European survey



**EURORDIS**  
European Organisation  
for Rare Diseases

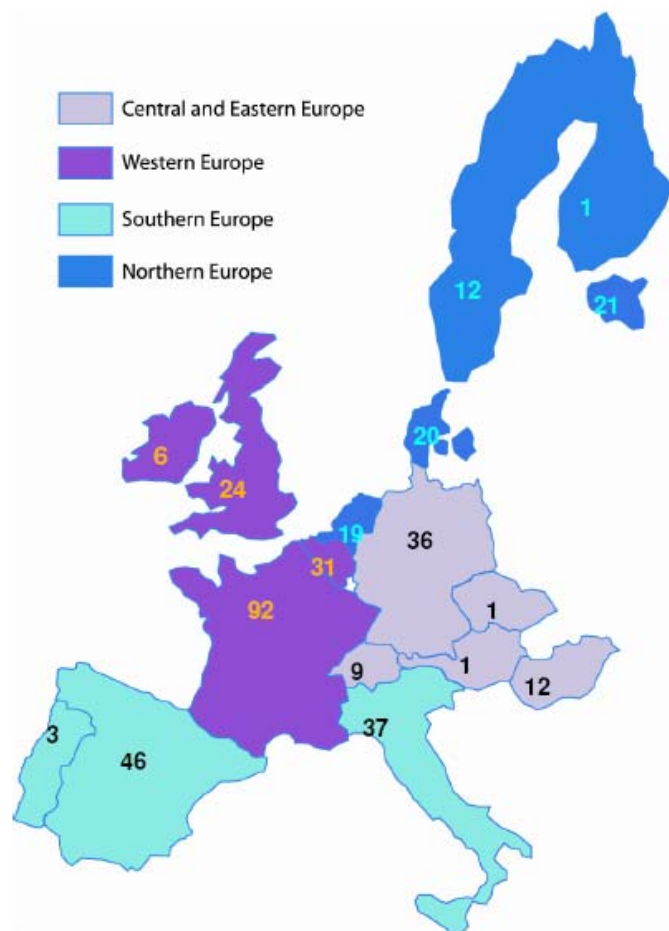
## Analysis of a questionnaire to 372 Information services on rare diseases

- 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](http://www.eurordis.org)

# Information Services: a European survey

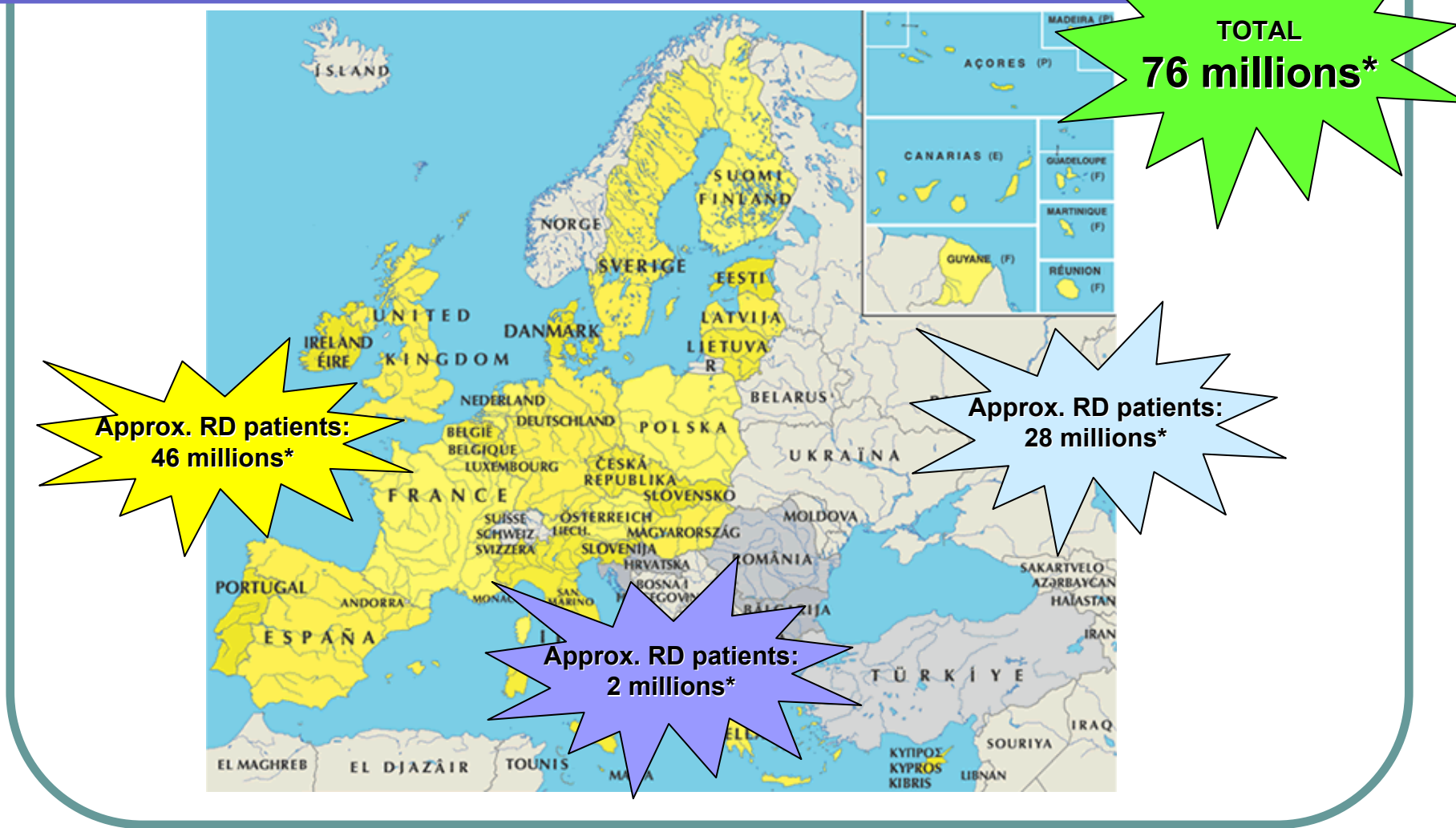


## Key results:

- 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](http://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](http://www.wrongdiagnosis.com/r/rare_diseases/stats-country.htm)

## The Number of Specialized Centers for Rare Diseases in Europe is Extremely Limited

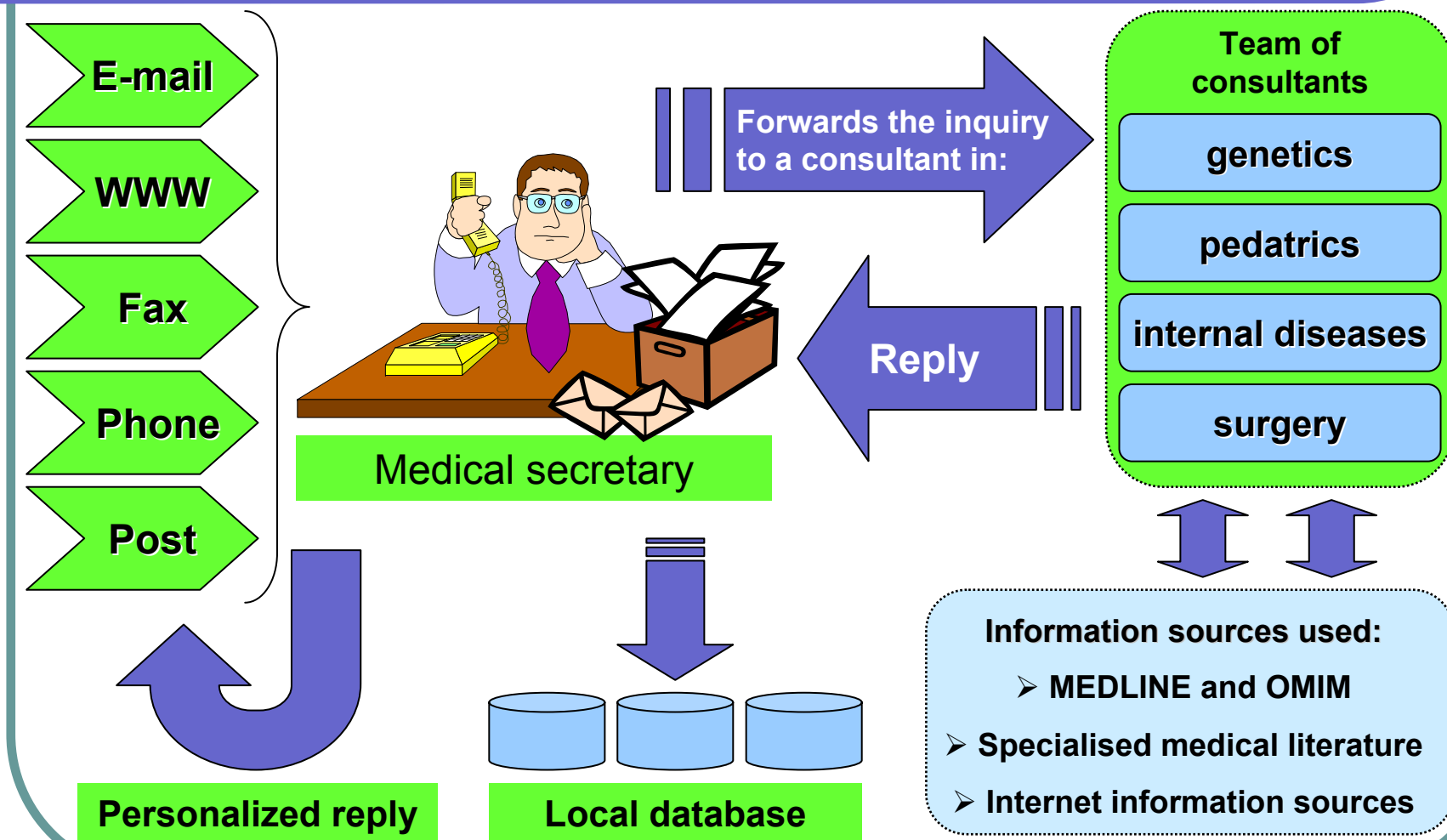
However, their **impact on the public health** is enormous:

- *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

# Organization of Work



## An example of reply to a medical professional - I

**DISORDER:** Parry–Romberg syndrome (PRS)

**SYNONIMS:** 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:** Recomendated.



## An example of reply to a medical professional - II

**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

**URL OMIM:** <http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?cmd=entry&id=141300>

**ICD 10:** L94.8

**PATIENT ASSOCIATIONS:**

<http://www.geocities.com/rombergs/>

<http://www.geocities.com/HotSprings/1018/index.html>

**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:

1. Adebajo, A. O.; Crisp, A. J.; Nicholls, A.; Hazleman, B. L. : Localized scleroderma and hemiatrophy in association with antibodies to double-stranded DNA. Postgrad. Med. J. 68: 216-218, 1992. PubMed ID : 1589383.
2. Bandello F, Rosa N, Ghisolfi F, Sebastiani A. New findings in the Parry-Romberg syndrome: a case report. Eur J Ophthalmol. 2002 Nov-Dec;12(6):556-8. PMID: 12510729.
3. Franceschetti, A.; Koenig, H. : L'importance du facteur heredo-degeneratif dans l'hemiatrophie faciale progressive (Romberg). Etude des complications oculaires dans ce syndrome. J. Genet. Hum. 1: 27-64, 1952. PubMed ID : 12981240
4. Hensch, E.; Romberg, H. M. : Klinische Ergebnisse. Berlin: A. Forstner (pub.) 1846. Pp. 75-81.
5. Klingmann, T. : Facial hemiatrophy. J.A.M.A. 49: 1888-1891, 1907.
6. Lakhani, P. K.; David, T. J. Progressive hemifacial atrophy with scleroderma and ipsilateral limb wasting (Parry-Romberg syndrome). J. Royal Soc. Med. 77: 138-139, 1984.
7. Lerner, A. J.; Bennison, D. P. Some observations on the aetiology of progressive hemifacial atrophy ('Parry-Romberg syndrome'). (Letter) J. Neurol. Neurosurg. Psychiat. 56: 1035-1039, 1993. PubMed ID : 8410030.
8. Lewkonja, R. M.; Lowry, R. B. Progressive hemifacial atrophy (Parry-Romberg syndrome) report with review of genetics and nosology. Am. J. Med. Genet. 14: 385-390, 1983. PubMed ID : 6601461
9. Parry, C. H. Collections from the unpublished medical writings of the late Caleb Hillier Parry. Vol. I. London: Underwoods (pub.) 1825. Pp. 478 only.
10. Pichiecchio A, Uggetti C, Grazia Egitto M, Zappoli F. Parry-Romberg syndrome with migraine and intracranial aneurysm. Neurology. 2002 Aug 27;59(4):606-8; discussion 481. PMID: 12196658.
11. Stone J. Parry-Romberg syndrome: a global survey of 205 patients using the Internet. Neurology. 2003 Sep 9;61(5):674-6. PMID: 12963760.
12. Walsh, F. B. Facial hemiatrophy: report of 2 cases. Am. J. Ophthal. 22: 1-10, 1939.
13. Wartenberg, R. Progressive facial hemiatrophy. Arch. Neurol. Psychiat. 54: 75-96, 1945.

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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 screenshot displays the homepage of the Information Centre for Rare Diseases and Orphan Drugs (www.raredis.org). The header features the organization's logo and name in both English and Bulgarian, along with its affiliation as an associate member of EURORDIS. A central banner promotes the 1st Eastern European Conference on Rare Diseases and Orphan Drugs, held in Plovdiv, Bulgaria, in May 2005. The main content area is divided into sections for 'RARE DISEASES', 'FAMILIAL FORM OF CUSHING'S SYNDROME', and 'NEWS'. Each section includes a brief description of a new disease profile added to the database, such as Pulmonary alveolar microlithiasis (PAM) and the familial form of Cushing's syndrome. The right sidebar contains a search bar, advanced search options, and logos of supporting organizations, including the Republic of Bulgaria Ministry of Health and the Clinical Research Centre for Rare Diseases. A poll at the bottom right asks visitors for their opinion on the site.

**INFORMATION CENTRE FOR RARE DISEASES AND ORPHAN DRUGS**  
**ИНФОРМАЦИОНЕН ЦЕНТЪР ЗА РЕДКИ БОЛЕСТИ И ЛЕКАРСТВА СИРАЦИ**  
Associate member of European Organisation for Rare Diseases - EURORDIS  
www.erordis.org

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Register now!  
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2 user(s) are online (2 user(s) are browsing **fiàèè**)  
Members: 0  
Guests: 2  
more...  
**New Members**

**The 1-st Eastern European Conference on Rare Diseases and Orphan Drugs**  
**Promotion of Healthcare and Research on Rare Diseases and Orphan Drugs in Eastern European Countries**  
27 May 2005, Plovdiv, Bulgaria  
**Register Now!**

— [dropdown] 10 [dropdown] [Go!]

**RARE DISEASES : PULMONARY ALVEOLAR MICROLITHIASIS**  
*Posted by informer on 2005/4/19 22:41:18 (1 reads)*

A new disease profile is added to our database - **Pulmonary alveolar microlithiasis (PAM)**. It is a rare disease, characterized by intra-alveolar calcium deposits. The aetiology of the disease is still unknown. In most cases patients have mild clinical symptoms, contrasting with the severe radiographic appearance: this is a typical feature that should raise the suspicion of PAM. However, a certain degree of dyspnoea with a productive cough may occur together with a sporadic haemoptysis and thoracic pains. To receive an e-mail with more information about the disease completely free of charge, please click [here](#) to send us a request. The profile was written by our consultant in internal diseases - Dr. Nikolay Botushanov, MD.

**RARE DISEASES : FAMILIAL FORM OF CUSHING`S SYNDROME**  
*Posted by informer on 2005/3/29 15:03:17 (18 reads)*

A new disease profile is added to our database - **the familial form of Cushing`s syndrome**. It is characterized histologically by small to normal-sized glands with cortical micronodules (average 2 to 3 mm). About half of the cases are sporadic. The reminders occur in association with a variety of other abnormalities, including myxomatous masses of the heart, skin or breast; blue nevi and other endocrine disorders. To receive an e-mail with more information about the disease completely free of charge, please click [here](#) to send us a request. The profile was written by our consultant in internal diseases - Dr. Nikolay Botushanov, MD.

**NEWS : COLLABORATION WITH ASSOCIATION "GEN"**

**Search**  
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REPUBLIC OF BULGARIA  
MINISTRY OF HEALTH

Clinical Research Centre for Rare Diseases  
"Aldo e Cele Dacco",  
Mario Negri Institute, Italy

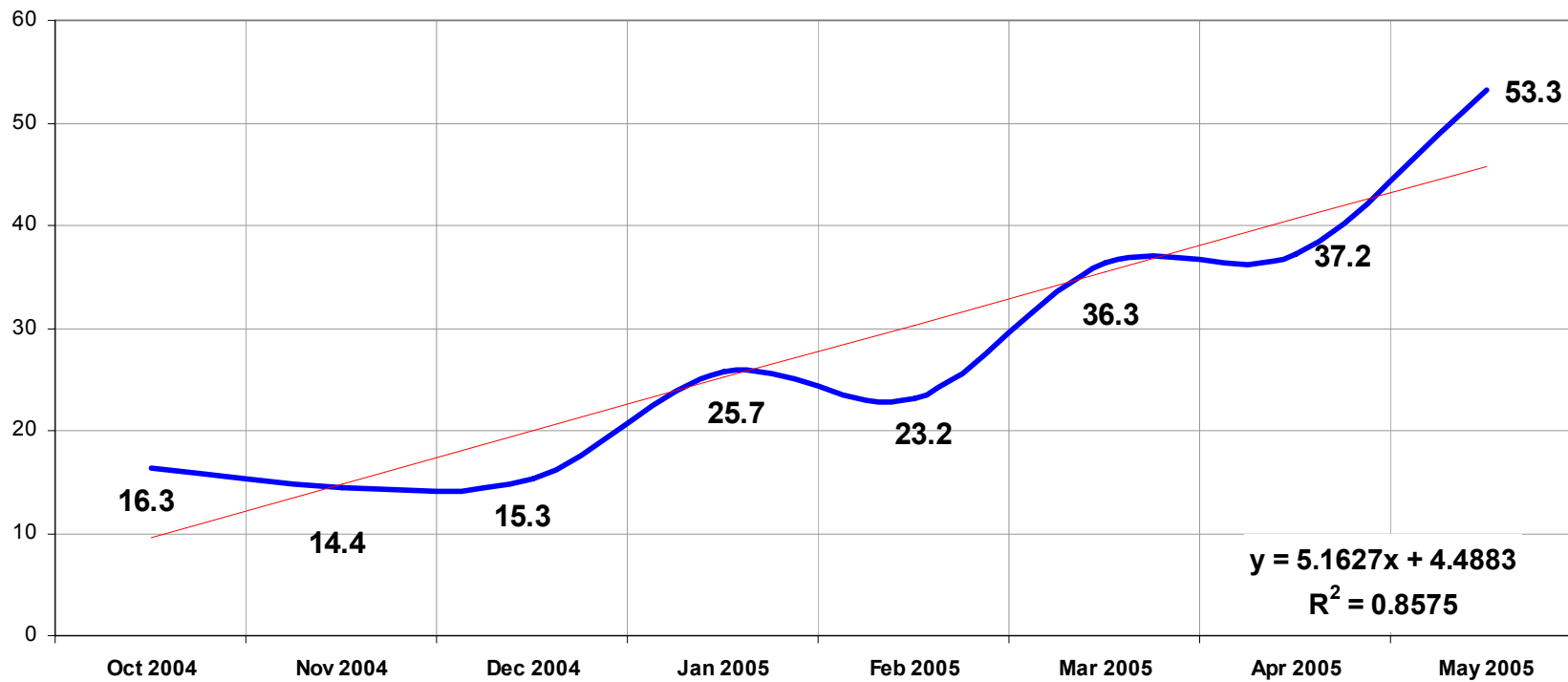
Българско дружество по неврология  
БЪЛГАРСКО ДРУЖЕСТВО ПО НЕВРОЛОГИЯ

БЪЛГАРСКО ОРТОПЕДО-ТРАВМАТОЛОГИЧНО ДРУЖЕСТВО

СЕКЦИЯ ПО ДИАЛИЗА  
НАУЧНО ДРУЖЕСТВО ПО НЕФРОЛОГИЯ, ХЕМОДИАЛИЗА И ТРАНСПЛАНТАЦИЯ

**Polls**  
**What do you think about this site?**  
 Excellent  
 Cool

# The Internet portal – Unique Visitors per Day



## Activities for Nov 2004 – April 2005

- Building and running the [Internet service](http://www.raredis.org) – [www.raredis.org](http://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
- **The First Eastern European Conference** on rare diseases and orphan drugs – **27 May 2005 г.**



# Established Contacts, Support and Collaboration

