

**2012 REPORT ON THE STATE OF THE ART
OF RARE DISEASE ACTIVITIES IN EUROPE
OF THE
EUROPEAN UNION COMMITTEE OF EXPERTS
ON RARE DISEASES**



**STATE OF THE ART OF RARE DISEASE ACTIVITIES IN
HUNGARY**

This work was financed by the EUCERD Joint Action: Working for Rare Diseases N° 2011 22 01

This document has been produced by the Scientific Secretariat of the European Union Committee of Experts on Rare Diseases (EUCERD, formerly the European Commission's Rare Diseases Task Force) through the EUCERD Joint Action: Working for Rare Diseases (N° 2011 22 01, Coordinator: Kate Bushby, University of Newcastle, United Kingdom), within the European Union's Second Programme of Community Action in the Field of Health.

More information on the European Union Committee of Experts on Rare Diseases can be found at www.eucerd.eu.

Disclaimer:

The findings and conclusions in this report are those of the contributors and validating authorities, who are responsible for the contents; the findings and conclusions do not necessarily represent the views of the European Commission or national health authorities in Europe. Therefore, no statement in this report should be construed as an official position of the European Commission or a national health authority.

Copyright information:

The "2012 Report on the State of the Art of Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases" is copyrighted by the European Union Committee of Experts on Rare Diseases (EUCERD). This product and its contents may be used and incorporated into other* materials on the condition that the contents are not changed in any way (including covers and front matter) and that no fee is charged by the reproducer of the product or its contents for their use. The product may not be sold for profit or incorporated into any profit-making venture without the expressed written permission of EUCERD. Specifically:

- 1) When the document is reprinted, it must be reprinted in its entirety without any changes.
- 2) When parts of the documents are used or quoted, the following citation should be used.

*Note: The "2012 Report on the State of the Art of Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases" contains material copyrighted by others. For material noted as copyrighted by others, the user must obtain permission from the copyright holders identified in the document.

To quote this document:

Aymé S., Rodwell C., eds., "2012 Report on the State of the Art of Rare Disease Activities in Europe of the European Union Committee of Experts on Rare Diseases", July 2012.

ISBN : 978-92-79-25355-3

DOI : 10.2772/50554

<http://www.eucerd.eu/wp-content/uploads/2012/09/2012ReportStateofArtRDActivitiesHU.pdf>

©European Union, 2012

ACRONYMS

General

CAT - Committee for Advanced Therapies at EMA
CHMP - Committee for Medicinal Products for Human Use at EMA
COMP - Committee on Orphan Medicinal Products at EMA
DG - Directorate General
DG Enterprise - European Commission Directorate General Enterprise and Industry
DG Research - European Commission Directorate General Research
DG Sanco - European Commission Directorate General Health and Consumers
EC - European Commission
ECRD - European Conference on Rare Diseases
EEA - European Economic Area (Iceland, Switzerland, Norway)
EMA - European Medicines Agency
ERN - European reference network
EU - European Union
EUCERD - European Union Committee of Experts on Rare Diseases
EUROCAT - European surveillance of congenital anomalies
EUROPLAN - European Project for Rare Diseases National Plans Development
EURORDIS - European Organisation for Rare Diseases
FDA - US Food and Drug Administration
HLG - High Level Group for Health Services and Medical Care
HTA - Health Technology Assessment
IRDiRC – International Rare Diseases Research Consortium
JA - Joint Action
MA - Market Authorisation
MoH - Ministry of Health
MS - Member State
NBS - New born screening
NCA - National Competent Authorities
NHS - National Health System
PDCO - Paediatric Committee at EMA
RDTF - EC Rare Disease Task Force
WG - Working Group
WHO - World Health Organization

Pilot European Reference Networks

Dyscerne - European network of centres of expertise for dysmorphology
ECORN-CF - European centres of reference network for cystic fibrosis
Paediatric Hodgkin Lymphoma Network - Europe-wide organisation of quality controlled treatment
NEUROPED - European network of reference for rare paediatric neurological diseases
EUROHISTIONET - A reference network for Langerhans cell histiocytosis and associated syndrome in EU)
TAG - Together Against Genodermatoses – improving healthcare and social support for patients and families affected by severe genodermatoses
PAAIR - Patients' Association and Alpha-1 International Registry Network
EPNET - European Porphyrin Network - providing better healthcare for patients and their families
EN-RBD -European Network of Rare Bleeding Disorders
CARE-NMD -Dissemination and Implementation of the Standards of Care for Duchenne Muscular Dystrophy in Europe project
ENERCA - European network for rare and congenital anaemia – Stage 3

GENERAL INTRODUCTION TO THE REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE OF THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

The 2012 Report on the State of the Art of Rare Disease Activities in Europe was produced by the Scientific Secretariat of the European Union Committee of Experts on Rare Diseases (EUCERD), through the EUCERD Joint Action: Working for Rare Diseases (N° 2011 22 01), which covers a three year period (March 2012 – February 2015).

The report aims to provide an informative and descriptive overview of rare disease activities at European Union (EU) and Member State (MS) level in the field of rare diseases and orphan medicinal products up to the end of 2011. A range of stakeholders in each Member State/country have been consulted during the elaboration of the report, which has been validated as an accurate representation of activities at national level, to the best of their knowledge, by the Member State/country representatives of the European Union Committee of Experts on Rare Diseases. The reader, however, should bear in mind that the information provided is not exhaustive and is not an official position of either the European Commission, its Agencies or national health authorities.

The report is split into five parts:

Part I: Overview of rare disease activities in Europe

Part II: Key developments in the field of rare diseases in 2011

Part III: European Commission activities in the field of rare diseases

Part IV: European Medicines Agency activities and other European activities in the field of rare diseases

Part V: Activities in EU Member States and other European countries in the field of rare diseases

Each part contains a description of the methodology, sources and validation process of the entire report, and concludes with a selected bibliography and list of persons having contributed to the report.

The present document contains the information from Parts II and V of the report concerning Hungary. A list of contributors to the report and selected sources are in annex of this document. For more information about the elaboration and validation procedure for the report, please refer to the general introduction of the main report¹.

¹ <http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArRDActivities.pdf>

RARE DISEASE ACTIVITIES IN HUNGARY

Definition of a rare disease

Stakeholders in Hungary accept the European Regulation on Orphan Medicinal Products definition of a prevalence of no more than 5 in 10'000 individuals.

National plan/strategy for rare diseases and related actions

A decree of the Ministry of Health established the National Rare Disease Centre (NRDC) in Hungary on 11 November 2008 as a part of the National Centre for Healthcare Audit and Inspection (OSZMK) by modifying its foundation deed. The National Centre for Healthcare Audit and Inspection was a part of the National Public Health Institute, and was empowered to investigate quality related issues in health care, public health consequences of health care operation, and to initiate interventions if needed. Due to the restructuring of the national public health institutions, OSZMK was disbanded on 30 April 2011. The National Institute for Health Development (OEFI) became the new host organisation, which is subordinated to the Chief Medical Officer and is a part of the central public health institutions. The continuation of the NRDC operation has been ensured by the modification of the foundation deed of OEFI.

The NRDC network participates in preparation of recommendations for Governmental health authorities in the following ways:

- It elaborates its own data collecting technology and co-operates with other stakeholders in order to obtain rare diseases related data and to prepare these indicators;
- It defines public health indicators for rare diseases;
- It initiates the elaboration of rare diseases guidelines and carries out the audit projects;
- It maintains the national database of rare diseases specialised health care providers;
- It contributes to the assignment of national centres of expertise and their participation in european networks;
- It facilitates the establishment and operation of the quality management programs for the hungarian rare diseases laboratories;
- It facilitates the application of e-health in rare disease related care;
- It initiates the rare diseases teaching programs launching in the universities;
- It participates in the work of national agencies responsible for orphan medicinal product and orphan medical device legislation;
- It supports the improvement of the availability of special social services for rare disease patients;
- It supports the effective primary preventive program;
- It evaluates the efficacy of the rare diseases screening programs;
- It facilitates the rare diseases research projects, the international co-operations;
- It contributes to the development of collaboration between governmental bodies, providers and patient organisations;
- It supports the hungarian participation in the european rare diseases projects;
- It initiates programs, which contribute to the improvement of the perception of rare diseases among the general public;
- It co-ordinates the elaboration and monitoring of national policy on rare diseases;
- It reports on the hungarian achievements regularly.

The NRDC is supported by an advisory group; the member experts were appointed by the Chief Medical Officer. Its members are from the four medical universities (the Hungarian Medical Universities' representatives to the national advisory group are nominated by the deans), governmental institutions, and patient organisations. This group has a key advisory function of strategic planning, but does not have influence and control on the implementation of the decisions made.

The NRDC is assembling a National Plan Organising Committee by supplementing the current expert committee with representatives of sectors such as government and industry. The Ministry is designating a competent, responsible Head of the expert committee, authorised to make decisions, to lead the development of the National Plan.

The former IT centre facilities are under reconstruction for the systematic analysis of the hospital and outpatient discharge records of rare diseases patients (for rare diseases which have their own ICD10 code), as well as laboratories, research programmes and patient groups.

The NRDC also works with the National Rare Disease Research Coordination Centre established in 2009 under the umbrella of OSZMK, National Public Health Institute, and the University of Pécs. This unit operates under the monetary support of the University of Pécs.

The NRDC cooperates with the National Ministerial Board for Clinical Genetics, and with the officials responsible for rare diseases policy at the Ministry of Health, and at the new National Institute for Quality and Organisational Development in Healthcare and Medicines (GYEMSZI). Project based collaboration has been established with universities' internal rare disease coordination units, sociological centres (for studying sociological characteristics of the patient groups), the National Foundation for Disabled Persons (multi-sector conference organisation), the National Centre for Statistics (for studying the mortality trends of rare diseases), and the Hungarian Federation of People with Rare and Congenital Diseases (HUFERDIS).

At the Europlan Hungarian national conference on rare diseases², organised by HUFERDIS on 18-19 October 2010 in Budapest, it was suggested that the issue of rare diseases should be adapted into the present, on-going reorganisation of the health care and social care system. The second Hungarian Europlan Conference on Rare Diseases³ (16-17 November 2011) was organised by HUFERDIS, together with the Ministry of National Resources, in Budapest, without European grant support. All stakeholders were present during the two day conference aimed at defining further concrete steps to develop a national action plan for rare diseases.

In 2011, the Ministry of Health appointed a National Coordinator and the National Plan Organising Committee (including the advisory board of the National Rare Diseases Centre and the patient groups) started to develop the National Plan with his leadership to develop a rare disease plan and to elaborate the plan. At the end of 2011, the main content of the plan was finalised and an expert meeting was held to finalise the chapters. Expert opinion will be sought by the end of March 2012, and it is hoped by end of June 2012 the plan will be ready to be included in the negotiations of the national budget at the start of August 2012.

Centres of expertise

There are currently no officially approved centres of expertise in Hungary, although around eight are informally recognised. There are four university centres with expertise in the field of rare diseases and diagnostic and therapeutic facilities: Budapest, Szeged, Pécs and Debrecen. In Hungary, a committee on the treatment of rare conditions has been set up within the Scientific Health Council (*Egészségügyi Tudományos Tanács*). It ensures, *inter alia*, that people suffering from such conditions receive adequate care in all cases. People suffering from rare conditions in Hungary are registered at the various treatment centres.

Two main factors are to be considered for the designation of Hungarian national centres of expertise: the presence of equipment for diagnosis, and personal expertise of the medical professionals in the centre. In Hungary, the need for 5 rare disease centres playing a coordinating role has been identified. The 4 existing medical universities could play this role, but it has to be assured that the adequate expertise is provided in these centres. Healthcare pathways will be considered as will interdisciplinary, which should be a key feature of the designation. In the National Plan for Rare Diseases, therefore, the strategy will be to designate the four medical universities as centres of expertise due to the existing structure of the health system by speciality and the prominence and reputation of the medical university in terms of research, amongst other disciplines. There are expert groups outside of the medical universities who respect the criteria, but the ways of involving these groups into the existing structures needs to be examined.

NRDC initiated a collaboration with the National Health Insurance Fund for the listing and transparent accreditation of centres of expertise, hospitals, and laboratories working in the field of rare diseases taking into account existing resources and their concentration, as well as eliminating parallelism and formalising existing informal relations and determining patients' pathways. The research project final report is expected to be published in 2012.

The NRDC has also initiated an open registry concerning the activities of centres of care and expertise, including the activities of consultants and laboratories requiring accreditation.

Pilot European Reference Networks

Hungary participates, or has participated, in the following European Reference Networks for rare diseases: EPNET/EPI and Care-NMD.

²http://download.EURORDIS.org/europlan/2_EUROPLAN_Guidance_Documents_for_the_National_Conference/final_report_%20hungary_europlan.pdf

³<http://europlan.rirosz.hu/>

Registries

The National Register of Congenital Anomalies (VRONY) operates country according to the EUROCAT protocol. The OSZMK has initiated the establishment of an overall register for rare diseases. Currently, the clinical centres of rare diseases maintain registries of cared patients: these registries do not report their cases to a national data collecting system, and their registration methodology is developed according to the local need of care management and to the research requirements. All of these registries are in line with the Hungarian laws on genetic data handling and on the personal data protection. Hungary contributes to European Registries such as TREAT-NMD, EUROCAT and EUROCARE CF.

Neonatal screening policy

A compulsory, government-financed newborn screening program covering the whole country has been running since 1975, and after introduction of tandem mass spectrometry screening in 2007, now includes 26 diseases, amongst which phenylketonuria, hypothyroidism, galactosaemia and biotinidase deficiency which belong to the classic core. Two centres are responsible for the operation of this nationwide network.

A HURO-euro programme started in May 2011 on the "Newborn screening and molecular genetic diagnosis of rare diseases: developing a Euro-regional infrastructure and cooperation". The University of Szeged is the project leader, and the Clinic de Urgență pentru Copii "Luis Țurcanu", (Timișoara) and Universitatea de Vest "Vasile Goldis" (Arad) are the Romanian partner institutions.

Genetic testing

The Genetic Professional College established in 2004 a protocol adopted by the Ministry of Healthcare entitled *Genetic Consultation*, which defines the conditions necessary for supplying the laboratory background, the infrastructure and the personal/operational costs for genetic diagnosis. Around 20% of laboratories have at least one diagnostic test validated by an external quality control scheme. The National Centre for Healthcare Audit and Inspection has also initiated an open registry including laboratories requiring accreditation.

Genetic diagnostic testing abroad is available through an application process to the National Health Insurance Fund and in many instances the Fund reimburses the costs.

Diagnostic tests are registered as available in Hungary for 57 genes and an estimated 77 diseases in the Orphanet database⁴.

National alliances of patient organisations and patient representation

The Hungarian Federation of People with Rare and Congenital Diseases (HUFERDIS) is the national alliance of 38 rare disease patient organisations in Hungary, affiliated with EURORDIS. HUFERDIS is currently encouraging the creation of a Hungarian Rehabilitation Centre for Rare Disease Patients. HUFERDIS represents rare diseases patients in the Hungarian Expert Committee of Rare Diseases (transforming to National Plan Organising Committee), the Council of National Alliances (CNA) of EURORDIS, and at the EUCERD.

Patient organisations provide information and act as contact points for rare disease patients and organise conferences. HUFERDIS organised an Expert Committee to help the National Plan Organising Committee in the development of National Plan, and participates in the accreditation of centres of expertise, the determination of guidelines, and in the therapeutic education and care programs, medical and social care training etc.

Non-medical services for rare disease patients are currently available at local level or by non-profit organisations. Patient organisations are partly supported by the '1% Law' which allows taxpayers to transfer 1% of their previous year's taxable income to a non-profit organisation (which may be a patient organisation), without loss of income. Grants from the National Civil Fund are also available to patient organisations. There is no regular, direct governmental support for rare disease self help groups, but there are many indirect governmental financing mechanisms: 25% of the civil budget is from governmental sources. HUFERDIS does not receive nominative state support such as that received by other umbrella patient organisations in Hungary.

Following a collaboration established between HUFERDIS, NRDC and the Hungarian Orphanet team, the EURORDISCare 2 and 3 surveys were carried out in Hungary. HUFERDIS takes part in the several international projects including Europlan, POLKA, BURQOL-RD, Rare Disease Days, etc. To foster the opinion of patient representatives on future European policies for rare diseases, or to collect their views on existing ones, HUFERDIS participated on the European POLKA project coordinated by EURORDIS. Hungary won the POLKA competition of EU Member states with the management of HUFERDIS.

⁴ Information extracted from the Orphanet database (September 2011).

Sources of information on rare diseases and national help lines

Orphanet activities in Hungary

Since 2004 there is a dedicated Orphanet team in Hungary, currently hosted by the University of Pécs. After its establishment, the NRDC was designated as the official Orphanet team for Hungary in 2010 by the Ministry of Health. This team is in charge of collecting data on rare disease related services (specialised clinics, medical laboratories, ongoing research, registries, clinical trials and patient organisations) in their country for entry into the Orphanet database. The Orphanet website is widely used by professionals.

Official information centre for rare diseases

There is currently no official information centre for rare diseases in Hungary apart from Orphanet.

Helpline

There is currently no rare disease specific helpline in Hungary. Many patient organisations provide support by telephone.

Other sources of information

Some websites, maintained by the government (www.gyemszi.hu, www.oefi.hu/aboutus.htm), have limited information concerning rare diseases. Scientific societies (www.mhgt.hu), non-governmental expert groups (www.betegmagzat.hu) and market-based organisations (www.webdoki.hu) have web based services for patients. The only other significant rare disease-specific website is the homepage of HUFERDIS (www.rirosz.hu), which was renovated during 2011. Several member associations of HUFERDIS have also detailed specific websites for a given rare disease.

Good practice guidelines

Guidelines related to rare diseases have been produced by the Ministry of Health and are available including: autism spectrum, cystic fibrosis, diagnosis of the inherited metabolic diseases, genetic counselling, haemophilia, investigation of familial clustering of anomalies, investigation of multiple congenital anomalies, Legg-Calve-Perthes disease (Perthes disease), multiple sclerosis, myasthenia gravis, Osgood-Schlatter disease, prenatal screening of Down syndrome, Scheuermann disease, systemic lupus erythematosus, Tibial hemimelia, clubfoot. Hungary supports the participation of local experts in the development of international guidelines that should help diagnosis and care of rare diseases patients at national level. Some guidelines have been developed in collaboration with patient organisations and specialised clinics, in line with the new European guidelines (e.g. Williams syndrome). One of the missing guidelines in the field of rare diseases was a national protocol for the communication of a diagnosis: another expert team of HUFERDIS has thus developed a new rare disease protocol to properly communicate a diagnosis.

Training and education initiatives

The education provided to health professionals currently includes information about the existence of rare diseases and the resources available for their care. This includes medical training in fields relevant to the diagnosis of rare diseases (genetics, oncology, immunology, neurology, paediatrics), further education for young doctors and scientists working in the field of rare diseases, and exchange and sharing of expertise between centres of expertise in the country.

Annual courses on rare diseases for graduates and postgraduates have been held at the Debrecen University, Department of Rare Diseases, since 2003 (40-100 participants). Rare diseases are also discussed at the Days of Internal Medicine of Debrecen (100-150 participants) which have been held seven times to date. The Department of Medical Genetics at the University of Pécs organises since 2009 a 3-day clinical genetics course covering among others the diagnosis and management of selected rare diseases; the meeting designated for specialists in the field as well as for family practitioners. The Semmelweis University also organises courses even for patients such as the "Molecular Medicine for Everybody". Regular conferences are organised on the area of rare diseases by the Hungarian Society of Personalised Medicine or by Industry, like the Personalised Healthcare Days of Roche.

The Epidemiology of Rare Diseases has been accepted as research area by the Health Sciences Doctoral School of University of Debrecen. The students are involved in the folic acid supplementation, prenatal screening, patient pathway and diagnostic delay investigations.

National rare disease events in 2011

HUFERDIS, the Hungarian rare disease alliance, organised the Hungarian Rare Disease Day as an informal central event of the EU supported by the Hungarian presidency⁵ on 26 of February 2011 in Budapest. The event was attended by a range of stakeholders from Hungary and Europe and gave insights into actions at Hungarian and European level in the field of rare diseases. Many parallel programmes were organised: an expert conference, poster section, games and handcrafting for children, entertainment programmes, “Rare Beauties” Art Exhibition, concerts, press conference, all-day exhibition of the HUFERDIS member associations. During the day Dr. Ildikó Horváth, Head of Department for Health Politics, State Secretariat for Healthcare, Ministry of National Resources, and her colleague Ildiko Szy presented the enacted clinical guidelines for rare diseases. More than 800 people, several journalists and TV teams participated at the event. Many interviews were given. These all guarantee that awareness about rare diseases is developing continuously.

The Orphanet Hungary team and the NRDC are also involved in Rare Disease Day events organised by HUFERDIS.

The second Hungarian Europlan Conference on Rare Diseases⁶ (16-17 November 2011) was organised by HUFERDIS, together with the Ministry of National Resources, in Budapest, without European grant support. All stakeholders were present during the two day conference aimed at defining further concrete steps to develop a national action plan for rare diseases. 125 participants took part in the two day event including experts, patients and representatives of government and the Industry. Participants at the conference monitored a number of main priorities: the creation of a Committee to lead the development of a national plan, the accreditation of centres of expertise for rare diseases, the inclusion of rare diseases in health care and social care systems (currently under reorganisation), the organisation of external quality control of accredited institutions, the provision of information on rare diseases in Hungarian, the participation of Hungary in EU projects, the organisation of awareness campaigns, and the organisation of a body to maintain a rare disease information helpline.

Other events included a “Prenatal Diagnosis of Down Syndrome: How Best to Deliver the News” was organised in Debrecen by the local patient organisations, the Debrecen University’s departments and the NRDC. A working group meeting was held in Budapest for the partners involved in the audit for the prenatal screening and diagnosis of Down syndrome organised by NRDC and VRONY. A symposium on “Multidisciplinary care for Rare Disease” was organised in Pecs by the National Rare Disease Research Coordinating Centre and NRDC.

Hosted rare disease events in 2011

The Rare Disease Day of 2011 became the official event of the Hungarian Presidency of the European Union, after the initiation of HUFERDIS. Several European experts gave presentations with simultaneous Hungarian/English translation and live online broadcasting of the conference on the internet, organised by HUFERDIS. The opening speech was given by prof. Dr. Miklós Réthelyi, Hungarian Minister of National Resources.

Research activities and E-Rare partnership

National research activities

Governmental research funds for rare diseases are available from the Hungarian Scientific Research Fund.

The Ministry of Health announces its health related research grants through the Scientific Health Council (ETT), Department of Research Coordination every three years. In the last evaluated period (2004-2006) €3 million went to support research grants. The summary report of the 2009-2011 program evaluation will be available in 2012. In these programs, rare diseases were not one of the priority areas, but many rare diseases related grants were financed (e.g. governmental supported the project on the periconceptual folate status and on attitude towards different supplement programs).

A multidisciplinary centre had been established in the Semmelweis University (Budapest) on the rare neurological disorders. The centre organises its work according to the principals published in the Communication from the European Commission on Rare Diseases. The centre has a patient registry, a diagnostic department, a multidisciplinary care providing network, research projects, and a teaching program⁷. To ensure the scientific expertise for NRDC, the general director of the National Centre for Healthcare Audit and Improvement, the rector of Pecs University, and the head of the Department of Medical Genetics signed

⁵ <http://sites.rirosz.hu/rbv/ritka-nap-2011/programme-in-english>

⁶ <http://europlan.rirosz.hu/euroterv-ii-konferencia-1/az-elhangzott-eloadasok>

⁷ <http://www.molneur.eoldal.hu/cikkek/english>

the detailed agreement which established the National Rare Disease Research Coordinating Centre on the 21 April 2009; this Centre is still embedded into the Department of Medical Genetics of University of Pécs. The Medical Faculty, Faculty of Health Sciences and the Faculty of Special Pedagogy are involved in this cooperative project. The experts employed by these faculties come from the fields of medicine, paramedicine, social services and education. The new working environment is expected to improve the Hungarian teams' ability to contribute to the work of European organisations.

All Hungarian Medical Faculties have started to establish their own coordinating centres to harmonise their rare diseases related activities, including research.

The IT centre of the NRDC elaborated the on-line registration system for health care providers, laboratories, research programs and patient groups related to rare diseases. This data collection is in line with the Orphanet data collection standards. The system has been launched and the primary database will be used to contribute to the Orphanet database.

Participation in European research projects

Hungary participates, or has participated, in European rare disease research projects including: BNE, EURO BONET, EURO GENTEST, EUROPEAN LEUKEMIA NET, EUROSCA, EUROWILSON, GENESKIN, NMD-CHIP, TREAT-NMD, SCRIN-SILICO, BBMRI and SIOPEN-R-NET.

E-Rare

Hungary is full partner of E-Rare-2 via the National Rare Disease Research Coordinating Centre at University of Pécs.

IRDiRC

Hungarian funding agencies are not currently committed members of the IRDiRC.

Orphan medicinal products⁸

The holders of marketing authorisations for orphan medicinal products (or their representatives in Hungary) cooperate with the medical profession and the OEP (The National Health Insurance Fund - *Országos Egészségbiztosítási Pénztár*).

Orphan medicinal product committee

There is no committee for orphan medicinal products in Hungary

Orphan medicinal product incentives

No specific activity reported.

Orphan medicinal product market availability situation

At the beginning of 2011 in Hungary, 69 of the 74 orphan medicinal products were available according to the Department of Rare Diseases in University of Debrecen. This institution implemented in Hungary the survey of the Rare Disease National Alliances & EURORDIS on Patients' Access to Orphan Medicinal Products in Europe. Most of orphan medicinal products are available via centres assigned by the National Health Insurance Fund.

The orphan medicinal products on the market in Hungary are: Afinitor™ (Everolimus), Aldurazyme™ (Laronidase), Arzerra™ (Ofatumumab), Atriance™ (Nelarabine), Busilvex™ (Busulfan (Intravenous use), Carbaglu™ (N-carbamyl-L-glutamic acid), Cayston™ (Aztreonam lizinát (inhalációs alkalmazásra), Ceplene™ (Hisztamin-dihidroklorid), Cystadane™ (Betaine anhydrous), Diacomit™ (stiripentol), Dudopa™ (Levodopa/Carbidopa (Gasztroenterális alkalmazás)), Elapraxe™ (iduronate-2-sulfatase), Evoltra™ (clofarabine), Exjade™ (deferasirox), Fabrazyme™ (α-Galactosidase A), Firazyr™ (icatibant acetate), Firdapse™ (amifampridine), Gliolan™ (5-aminolevulinsav hidroklorid), Glivec™ (Imatinib mesilate), Ilaris™ (Az IgG1/K osztályba tartozó humán IL 1β elleni rekombináns humán monoklonális antitest), Increlex™ (Mecasermin rinfabát), Inovelon™ (Rufinamid), Ixiaro™ (Tiszított, inaktivált japán encephalitis SA14-4-2 vírus vakcina), Kuvan™ (Tetrahidrobiopterin), Litak™ (cladribine), Lysodren™ (mitotane), Mepact™ (Muramil Tripeptid Foszfátidil Etanolamin), Mozobil™ (plerixafor), Myozyme™ (Recombinant human acid α-glucosidase), Naglazyme™ (N-acetilgalaktózamin-4-sulfatáz), Nexavar™ (Sorafenib tosylate), Nexavar™ (sorafenib tosylate), Nplate™ (Rekombináns megakaryocita képződést serkentő fehérje), Nymusa™ (caffeine citrate), Onsenal™

⁸ This section has been written using the Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products. 2005 Revision (p15)

(Celecoxib) , Orfadin™ (Nitisinone), Pedeas™ (Ibuprofen), Photobarr™ (Porfimerum nátrium(fotodinámiás kezeléscéljára), Prialt™ (α-Galactosidase A), Replagal™ (α-Galactosidase A), Revatio™ (Sildenafil citrate), Revlimid™ (Lenalidomid), Revolade™ (Eltrombopag olamin), Savene™ (Dexrazoxane), Siklos™ (Hydroxyurea, Soliris™ (Eculizumab), Somavert™ (pegvisomant), Sprycel™ (Dasatinib), Tasigna™ (Nilotinib), Tasigna™ (Nilotinib), Tepadina™ (Thiotepa), Thalidomide Celgene™ (Thalidomide), Thelin™ (Sitaxentan nátrium), Torisel™ (temsirolimus), Torisel™ (Temsirolium) , Tracleer™ (Bosentan), Tracleer™ (Bosentan) , Trisenox™ (Arsenic trioxide), Ventavis™ (Iloprost), Vidaza™ (Azacitidin) , Vidaza™ (azacitidine), Volibris™ (Ambrisentan), Votrient (Patorma)™ (Pazopanib hidroklorid), Wilzin™ (Zinc acetate dihydrate), Xagrid™ (Anagrelide Hydrochloride), Yondelis™ (Ecteinascidin 743), Yondelis™ (Trabectedin), Zavesca™ (miglustat) , Zavesca™ (Miglusztát).

Orphan medicinal product pricing policy

The OEP does not have a direct impact on pricing.

Orphan medicinal product reimbursement policy

According to the *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal product*⁹, "the Hungarian Government promotes the use of orphan medicinal products for specific patients by means of special financial arrangements".

"Some rare conditions (such as Fabry disease or adult-type chronic myeloid leukaemia - CML), the National Health Insurance Fund (OEP) provides standard price-support for the medicinal products in a predetermined manner. In this case the patient's contribution is negligible or 0%. In other cases, support for the orphan medicinal products imported for patients' treatment can be provided on application under a special equity procedure laid down by law. The OEP pays the price-support for the necessary medicinal products from earmarked resources in the outpatients' equity fund"¹⁰. In most cases, support is only available via discretionary claims. There is a yearly budget for such claims managed by the OEP. The discretionary procedure takes into account the financial situation of the claimant. In 2009, 289 patients had their discretionary claims accepted. Around 13 rare diseases receive support within the framework of discretionary claims. 33 orphan medicinal products are 100% reimbursed in Hungary. The re-regulation of pharmaceutical reimbursement inclusion decisions started in 2011.

Other initiatives to improve access to orphan medicinal products

Off-label use is possible, provided that the benefits of the drugs for a certain disease are certified, but the process is highly bureaucratic.

Specialised social services

There are good and high quality programs in the field of early development and respite care which support patients and their families. However, these programmes do not cover the whole country. Legislation exists on care, training, integration, work help for special needs children (i.e. extra home care), however these initiatives are not available to all rare disease patients. The change of this legislation has started in favour of rare disease patients. There are measures in place to support patients who need to travel inland to access health care through an assessment of needs by the Health Insurance Fund.

The Ministry of Human Resources started to work together with HUFERDIS for a project establishing the National Habilitation, Development and Service Centre of Rare Disorders to help the social integration of rare disease patients. Several health care institutions started to change care profile during the reorganisation within the Semmelweis Plan. Some of them will be able to offer more rehabilitation and social care.

HUFERDIS joined the therapeutic recreational programs of a member association (Hungarian Williams Syndrome Association) and organised programmes for capacity building and training, networking, awareness raising, exchange of information and best practices, during a special development family camp and after.

⁹ *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products. 2005 Revision (p15)*

¹⁰ *Inventory of Community and Member States' incentive measures to aid the research, marketing, development and availability of orphan medicinal products. 2005 Revision (p15)*

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN HUNGARY

National plan/strategy for rare diseases and related actions

In 2011, the Ministry of Health appointed a National Coordinator and the National Plan Organising Committee (including the advisory board of the National Rare Diseases Centre and the patient groups) started to develop the National Plan with his leadership to develop a rare disease plan and to elaborate the plan.. At the end of 2011, the main content of the plan was finalised and an expert meeting was held to finalise the chapters. Expert opinion will be sought by the end of March 2012, and it is hoped by end of June 2012 the plan will be ready to be included in the negotiations of the national budget at the start of August 2012.

The National Institute for Health Development (OEFI) became the new host organisation of the National Rare Disease Centre (NRDC) in May 2011. The OEFI is subordinated to the Chief Medical Officer and is a part of the central public health institutions. The continuation of the NRDC operation has been ensured by the modification of the foundation deed of OEFI.

Neonatal screening policy

A HURO-euro programme started in May 2011 on the "Newborn screening and molecular genetic diagnosis of rare diseases: developing a Euro-regional infrastructure and cooperation". The University of Szeged is the project leader, and the Clinic de Urgență pentru Copii "Luis Țurcanu", (Timișoara) and Universitatea de Vest "Vasile Goldis" (Arad) are the Romanian partner institutions.

Training and education initiatives

The Epidemiology of Rare Diseases has been accepted as research area by the Health Sciences Doctoral School of University of Debrecen. The students are involved in the folic acid supplementation, prenatal screening, patient pathway and diagnostic delay investigations.

National rare disease events in 2011

HUFERDIS, the Hungarian rare disease alliance, organised the Hungarian Rare Disease Day as an informal central event of the EU supported by the Hungarian presidency¹¹ on 26 of February 2011 in Budapest. The event was attended by a range of stakeholders from Hungary and Europe and gave insights into actions at Hungarian and European level in the field of rare diseases. Many parallel programmes were organised: an expert conference, poster section, games and handcrafting for children, entertainment programmes, "Rare Beauties" Art Exhibition, concerts, press conference, all-day exhibition of the HUFERDIS member associations. During the day Dr. Ildikó Horváth, Head of Department for Health Politics, State Secretariat for Healthcare, Ministry of National Resources, and her colleague Ildiko Szy presented the enacted clinical guidelines for rare diseases. More than 800 people, several journalists and TV teams participated at the event. Many interviews were given. These all guarantee that awareness about rare diseases is developing continuously.

The second Hungarian Europlan Conference on Rare Diseases¹² (16-17 November 2011) was organised by HUFERDIS, together with the Ministry of National Resources, in Budapest, without European grant support. All stakeholders were present during the two day conference aimed at defining further concrete steps to develop a national action plan for rare diseases. 125 participants took part in the two day event including experts, patients and representatives of government and the Industry. Participants at the conference monitored a number of main priorities: the creation of a Committee to lead the development of a national plan, the accreditation of centres of expertise for rare diseases, the inclusion of rare diseases in health care and social care systems (currently under reorganisation), the organisation of external quality control of accredited institutions, the provision of information on rare diseases in Hungarian, the participation of Hungary in EU projects, the organisation of awareness campaigns, and the organisation of a body to maintain a rare disease information helpline.

¹¹ <http://sites.rirosz.hu/rbv/ritka-nap-2011/programme-in-english>

¹² <http://europlan.rirosz.hu/euroterv-ii-konferencia-1/az-elhangzott-eloadasok>

Other events included a symposium on “Prenatal Diagnosis of Down Syndrome: How Best to Deliver the News” was organised in Debrecen by the local patient organisations, the Debrecen University’s departments and the NRDC. A working group meeting was held in Budapest for the partners involved in the audit for the prenatal screening and diagnosis of Down syndrome organised by NRDC and VRONY. A symposium on “Multidisciplinary care for Rare Disease” was organised in Pecs by the National Rare Disease Research Coordinating Centre and NRDC.

Research activities and E-Rare partnership

IRDiRC

Hungarian funding agencies are not currently committed members of the IRDiRC.

LIST OF CONTRIBUTIONS¹³

Contributions in 2010

Janos Sandor (*Orphanet Hungary, University of Debrecen, Department of Biostatistics and Epidemiology*)
Gabor Pogany (*HUFERDIS*)

Contributions in 2011

Janos Sandor (*Orphanet Hungary, University of Debrecen, Department of Biostatistics and Epidemiology*)
Gabor Pogany (*HUFERDIS*)
Béla Melegh, (*Department of Medical Genetics, University of Pécs*)
Gyorgy Kosztolanyi (*National Coordinator of the Hungarian National Plan*)

Contributions in 2012

Janos Sandor (*Orphanet Hungary, University of Debrecen, Department of Biostatistics and Epidemiology*)
Gabor Pogany (*HUFERDIS*)

Validated by: Janos Sandor (*EUCERD Representative Hungary, University of Debrecen, Department of Biostatistics and Epidemiology*)

SELECTED BIBLIOGRAPHY AND SOURCES¹⁴

- National Surveillance Centre for Congenital Anomalies and Rare Diseases
<http://193.225.50.35/ujhonlap/control.php?PAGE=RBK>
- HUFERDIS
www.rirosz.hu
- Orphanet Hungary national website
<http://www.orpha.net/national/HU-HU/index/homepage/>
- Europlan Hungarian National Conference Final Report
http://download.EURORDIS.org/europlan/2_EUROPLAN_Guidance_Documents_for_the_National_Conference/final_report_%20hungary_europlan.pdf
- Hungarian National Conference resources
<http://europlan.rirosz.hu/euroterv-ii-konferencia-1/az-elhangzott-eloadasok>

¹³ The contributors and validators of the report have contributed information which is accurate to the best of their knowledge. However, readers should take note that the contents of this report are illustrative and not exhaustive.

¹⁴ All websites and documents were last accessed in May 2012. A more detailed list of sources is available in the full report:
<http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf>