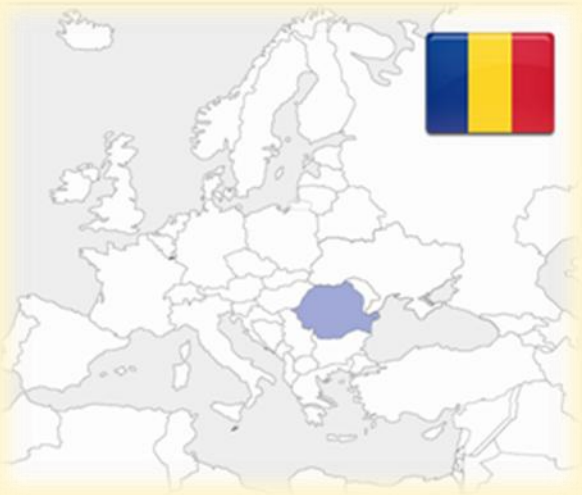
2014 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE



STATE OF THE ART OF RARE DISEASE ACTIVITIES IN ROMANIA

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The European Union Committee of Experts on Rare Diseases (EUCERD) was established in 2009 and its mandate ended in 2013. It is replaced from 2014 by the Commission Expert Group on Rare Diseases. The EUCERD Joint Action continues to support the activities of the new Expert Group until 2015.

More information on the activities of the former European Union Committee of Experts on Rare Diseases and the EUCERD Joint Action can be found at www.eucerd.eu.

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ACRONYMS

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
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

GENERAL INTRODUCTION

This document 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).

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The present 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 2013. 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 Commission Expert Group on Rare Diseases. The reader, however, should bear in mind that the information provided is not exhaustive and is not an official position of the European Commission, its Agencies or national health authorities.

The report is split into six parts:

Part I: Overview of rare disease activities in Europe
Part II: Key developments in the field of rare diseases in 2013
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
Part VI: Activities at National level in each EU Member State and other European countries in the field of rare diseases

Parts I – V also include a description of the methodology, sources and validation process of the entire report, and 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 Romania. 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 report.

Each year, there are around 15 000 downloads of the different sections of the report combined.

RARE DISEASE ACTIVITIES IN ROMANIA

Definition of a rare disease

In Romania, stakeholders accept the EU definition of a rare disease (RD) laid down in Regulation EC n° 141/2000 on Orphan Medicinal Products, the Directive 2011/24/EU on Cross Border Healthcare as well as in the Council Recommendation on an action in the field of rare diseases of 8 June 2009. According to the EU definition a RD is defined as life-threatening or chronically debilitating condition that afflicts fewer than 5 in 10 000 persons in the general population.

National plan/strategy for rare diseases and related actions

Since February 2008, there is an official RD status: RD are recognised by stakeholders in the field as a priority for health care in Romania. A partnership agreement was signed by the Romanian Ministry of Health and the country's National Alliance for Rare Diseases (RONARD) aimed at guiding and structuring the actions for the elaboration of a National Plan for RD.

Between 2008 – 2010, RONARD and representatives of Ministries of Health, Education, and Labour, as well as the National Medicine Agency, the Authority of People with Disabilities and the Child Welfare Authority worked together on rare diseases in a comprehensive manner, with different topics ranging from the assessment of the current situation of RD in Romania to the increase knowledge of the epidemiology of RD, recognition of the specificity of RD, the development of information for patients, healthcare professionals and the public, the improvement of access to medical care, timely and appropriate diagnosis, better treatment and access to required drugs, the identification of the specific needs of assistance for people with RD and the definition of institutional and legal framework in the field of RD.

On the debate and consultation process involving RONARD and stakeholders, several objectives and priority actions have been identified as relevant for the development of a draft of the NPRD launched in 2010. The draft was posted on the Romanian Prader Willi Association website for online public comments and reactions. Moreover, the draft of NPRD was sent to European Commission in order to harmonise and share the strategic vision in accordance with the recommendations delineated by the Council of EU Recommendation on action in the field of RD. The plan was structured based on general objectives, such as:

1. Develop an institutional framework;
2. Develop services for the diagnosis, treatment, rehabilitation and prophylaxis of RD;
3. Improve access to the appropriate medication and technology;
4. Improve access to the information in the field of RD;
5. Develop human resources;
6. Stimulate the research in the field of RD;
7. Empowerment of patient organizations;
8. Develop national and European partnerships in the field of RD.

By the end of 2012 the inclusion of the NPRD in the National Public Health Strategy was the next step concerning the implementation and monitoring of the plan by the end of 2013. At the end of 2013, the National Council for RD (NCRD) was created by the Ministry of Health (MoH) through a Ministerial Order (1215/2013).

In 2013 RONARD continued its active work in the development of the NPRD. The main activities of the RONARD in 2013 were the following:

- RONARD organised the Rare Diseases Day (RDD) Campaign and during the National Conference for Rare Diseases on 2nd of March 2013, at the end of the RDD campaign it was signed a new partnership agreement[1] with Ministry of Health Romania to create the institutional framework for the implementation of the National Plan for Rare Diseases (National Council for Rare Diseases). It is an interdisciplinary scientific body without a legal personality, working as an advisor of the Ministry of Health, providing specialised expertise both from its own team, as well as from specialised teams in different areas, developing criteria for designating centres of expertise in the management of rare diseases, defining evaluation process and identify the Centre of Expertise in Romania, communicating with RONARD and other institutional partners and define priority actions to ensure continuity of care for patients with rare diseases: information, diagnosis, treatment or specific therapies, counselling and patient and family education, training and integration specialists in the community;

[1] http://www.ms.gov.ro/documente/ordin%20boli%20rare_823_1638.pdf

- The second Europlan National Conference[2] was organised under the auspices of Ministry of Health of Romania in Bucharest, on 24-25 May 2013. The Conference aimed at facilitating dialogue, participation and involvement of all stakeholders in the field of rare diseases in Romania (patients, professionals, authorities, politicians, industry, media, etc.) in order to address solutions that need to be taken to update the proposals of the National Plan for Rare Diseases (NPRD).
- A Resolution of the Europlan Conference has been agreed by all the stakeholders involved in rare diseases field in Romania around the main objectives and future activities for rare diseases in 2013 and the following NPRD for the period: 2014-2020.

The economic context had a negative impact on public funds allocated to NPRD in 2013.

Rare cancers are mentioned in the NPRD in Romania and future actions will be included for the period 2014-2020.

Emergency cards have not yet been developed in Romania.

Centres of expertise

So far a number of centres of expertise are functioning in Romania, but they are not officially recognised / labelled as centres of expertise. Expertise has been developed around the medical Universities and National Institutes for Health and currently many rare diseases are diagnosed, treated and followed-up.

NCRD (National Council for Rare Diseases) started to develop a policy concerning Centres of Expertise for RD. A procedure for the designation and evaluation of centres of expertise is under development using the EUCERD Recommendations on Quality Criteria for Centres of Expertise adapted to the situation in Romania. NCRD will select the criteria, define the policy in the country and organise the national competition for expertise centres. The number of total national/regional centres of expertise is still debateable based on population size and geographic distribution. Following the competition the Centres of Expertise designated will participate in the future European Reference Network.

At the moment, expert medical care is provided by many different centres all over the country, including: National Institutes of Oncology (Trestioreanu – Bucharest and Chiricuta - Cluj Napoca), Institute of Cerebrovascular diseases (Bucharest), Heart Institute (Cluj), National Institute of Endocrinology (Parhon - Bucharest), National Institute for Mother and Child (Alfred Rusescu - Bucharest), National Institute for Research and Development Victor Babes (Bucharest), Fundeni Clinical Institute (Bucharest), and many others. In addition, in major Romanian medical centres (Bucharest, Iasi, Constanta, Cluj, Timisoara, Targu Mures) there is a programme in place for the diagnosis of foetal anomalies which also aims to improve healthcare during pregnancy and diagnosis of possible genetic disorders.

A network for Pulmonary Hypertension (PHT) has been composed, including the following ~~following~~ institutes: Institute of Cardiovascular Diseases "CC Iliescu"(Bucharest), Pneumology Hospital (Iasi), Hospital "Victor Babes" (Timisoara), Heart Institute (Cluj) Institute of Cardiovascular Diseases and Transplantation - Pediatric Cardiology Clinic (Targu Mures), Children's Emergency Hospital "Louis Turcanu (Timisoara), Heart Centre - Cardiovascular Surgery Clinic (Cluj), Department of Paediatric Cardiology (Bucharest).

Since 2011 a Pilot Reference Centre for Rare Diseases “NoRo” was opened in Zalau, made possible through the project "Norwegian - Romanian (NoRo) Partnership for Progress in Rare Diseases" (2009- 2011) with financial support from the Norwegian Cooperation Programme for sustainable economic development in Romania. The centre offers information concerning RD and through the helpline they refer patients to the specialists involved in the field.

Registries

Since 2013 NCRD deals with the issue of a national registry for RD but no public financial resources have been allocated so far. At the moment, NCRD selects the common data elements for Rd databases and extends the work by ensuring that the dataset are defined in the same way, using the same standards and same terms. Also, NCRD is considering appropriate EU standardised databases in order to find how to harmonise, share and exchange information. In addition, NCRD establishes baseline measures for data safety and protection. National registries and databases can be use to plan and manage services in the field of RD.

So far some registries are in place managed by Academia, clinicians or patient organisations (Romanian biliary atresia registry and Romanian cystic fibrosis patient registry, both having national coverage). There are more patients’ registries in the field of RD but they do not fulfil all the requirements for a registry (National Registry of Haemophilia, the National Registry of Primary Immunodeficiency, the National Registry of

[2] http://www.eurordis.org/sites/default/files/flags/finalreport-romania.pdf

Infant Diabetes Mellitus, the National Registry of Thalassemia, the National Registry for Pulmonary Hypertension, the National Registry for Hyperparathyroidism, the National Registry for Acromegaly, and the National Registry of Neuromuscular Diseases; The National Registry of congenital Hypothyroidism and National Registry of Neuroendocrine tumours are still under development).

Romania contributes to the following European registries: EBAR (European Biliary Atresia Registry), EUROCARE CF (European Cystic Fibrosis Registry) and EUTOS (European Treatment and Outcome Study for Chronic Myeloid Leukaemia).

Neonatal screening policy

In 2013, according to the national health policy a mandatory newborn screening program was available for two diseases, phenylketonuria (PKU) and congenital hypothyroidism (CHT), with the goal of screening all infants born in the country. The screening is performed in 4 public medical centres throughout the country (Bucharest, Iasi, Cluj-Napoca and Timisoara). The current health policy improved the management of screening for PKU and CHT at local level, reaching 80% coverage for newborns.

In the last two years, efforts were made via the Health Programme at local level in order to generalise and improve the organisation of neonatal screening for these two disorders, as well as to introduce screening for other (rare) diseases for which some form of treatment is available. Newborn screening tests for other diseases are provided by private clinics / laboratories at full cost or can be carried out in the framework of research programmes (e.g., hearing loss).

However, despite the screening programme, some tests are not available nationwide due to logistic and resource problems and no additional RD have been added to the panel of diseases tested in 2013.

Also, in 2013, Romania continued to join in efforts aimed for the preparation of European guidelines on diagnostic tests or population screening (EUNENBS: European Network of Experts on Newborn Screening) respecting national decisions and competences.

Genetic testing

Genetic testing in Romania is available through public or private Medical Genetic Clinics but it is not covered in the National Programme for Rare Diseases. Usually, genetic testing is performed in University Medical Centres (Bucharest, Cluj, Craiova, Iasi, Oradea, Targu-Mures and Timisoara). Physicians specialising in Medical Genetics only are allowed to provide genetic counselling and pre- and postnatal testing.

Services include molecular and cytogenetic analysis such as sexual chromatin, conventional karyotype and interphase and metaphase FISH techniques, and DNA tests (MLPA, QF-PCR, arrayCGH, targeted sequencing). Genetic testing is carried out before birth (via amniocentesis or chorionic villus samples and, recently, non-invasive prenatal testing using cffDNA in maternal blood for the detection of fetal chromosomal abnormality) and after birth.

The current policy for prenatal screening and diagnosis in Romania includes a national programme for all pregnant women over age 35 at conception offering tests free of charge on a limited budget. Biochemical screening, ultrasound and CVS/amniocentesis are performed in 6 public medical centres throughout the country. Prenatal screening/diagnosis is also offered to all pregnant women independently of maternal age with costs eligible to be covered by national health insurance. At this time, some prenatal testing procedures are invasive and performed for the most common clinically significant foetal aneuploidies. Prenatal diagnosis is also used to determine whether a foetus has a rare monogenic disorder. Usually, for a foetus at increased risk for rare monogenic diseases, CVS/amniocentesis and DNA isolation are performed in many public or private clinics. Then the DNA samples are sent abroad for molecular diagnosis of rare monogenic diseases (sequencing for entire gene or selected exons).

All genetic laboratories, public or private, are accreditated at national level according to SR EN ISO 15189 :2013 (international standard for medical laboratories).

Patients are referred for genetic testing by a physician (i.e. obstetrician, paediatrician, medical geneticist, haematologist and oncologist). Usually the results of genetic tests are interpreted by a medical geneticist who can also offer the genetic counselling. In Romania the health insurance does not cover the costs of genetic tests. But some genetic tests are free of charge for children who are enrolled in national health programme for birth defects. Other times, the patients could be enrolled in research programmes or non-profit humanitarian programmes, so that genetic tests are available for free.

Romania, as is the case of other European countries, cannot provide genetic tests for all disorders: other specific tests unavailable nationally are available abroad. Form S2 for Health Care Abroad/E112 ~~offers~~ is

used in these cases. Diagnostic tests are registered as available in Romania for 31 genes and an estimated 55 diseases in the Orphanet database[3].

There are no national practice guidelines for genetic testing yet, but guidelines are in progress. Professional organisations (Romanian Society of Medical Genetics) and other NGOs are working to complete this task using their experience and European recommendations.

National alliances of patient organisations and patient representation

RONARD – Romanian National Alliance for RD is the main organiser of the activities in the field of RD in Romania. The activity of the National Alliance is not supported by the National Authorities.

At the end of 2013, the National Council for RD was created by the Ministry of Health (MoH) through a Ministerial Order (1215/2013) and 3 representatives of the RONARD are full members in the executive committee of the council. The council, as an advisory group for MoH in the field of RD is playing an important support role in the consultation and implementation of the NPRD.

Sources of information on rare diseases and national help lines

Orphanet activity in Romania

Since 2004 there is a dedicated Orphanet team in Romania, currently hosted by "Gr T Popa" University of Medicine and Pharmacy, Iasi. This team was designated as the official Orphanet team for Romania by the Ministry of Health in 2010. 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. An Orphanet country site was launched in 2012.

There is no provision to fund our Orphanet national team within the NPRD. The Orphanet Romania Team has an active representative in the National Committee for Rare Diseases. The Orphanet Romania Team organised an event for Rare Disease Day in Iasi and also, have presented Orphanet in different meetings (at local and national level).

Official information centre for rare diseases

The Romanian Prader Willi Association has established a centre for information for rare genetic disorders in 2005 which is accredited by the Ministry of Work for its activity. RONARD and the Romanian Association for Rare Cancers are also accredited for providing information and counseling for patients with RD and rare cancers in Romania. Their activity is funded through projects. Through the NoRo project a virtual platform for rare diseases has been developed: www.edubolirare.ro, both for information and authorised training sessions for different professionals, including personal assistants.

The NoRo Centre for RD has videoconference facilities which can be used for provision of information, counseling and training of patients, parents and professionals.

In October 2013, RONARD has also launched an online platform for the development of organisational capacity of patients' organisations in Romania: www.aspac.ro .

Help line

The Romanian Prader Willi Association – RPWA – manages the NoRo helpline (080 080 1111). In 2011 the helpline was improved by introducing Orphacodes in the call information management software. In addition, a caller profile analysis was carried out, together with the other members of the European Network of Help Lines for Rare Diseases. Legal attestation has been granted assuring that the service operates according to the Romanian legislation of data protection and privacy. The helpline is part of the European Network of Helplines organised by Eurordis, together with the helpline provided by the Romanian Association for Myasthenia Gravis. At the end of 2013, RPWA has initiated a restructuration of the NoRo HelpLine in order to develop the network of professionals that support the activity at national level.

Other sources of information on rare diseases

The site http://bolirare.ro/ provides some information on rare and genetic diseases, in the Romanian language.

The Romanian Prader Willi Association also produces the publication "Rare People and Rare Diseases" for patients and the public.

Leaflets with information concerning major genetic disorders are available for patients and parents in some Medical Genetics Centres all over the country.

Other sources of information include lectures by specialists in the field of RD.

[3] Data extracted from Orphanet (January 2014)

Guidelines

Before 2013, clinical practice guidelines have been produced for every medical specialty including RD but several of them are at national level (e.g., diagnosis and follow-up in Oncology, Pulmonary Hypertension or Hereditary Angioedema).

In Romania the following translated or elaborated RD guidelines were launched in 2013:

- The Romanian Prader Willi Association translated and printed "A medical alert for Prader Willi Syndrome";
- In cooperation with Romanian Society for Human Genetics and many other professionals a book entitled "Medical Alert in Rare Genetic Diseases, an emergency guideline for 18 rare diseases clusters" was re-published. The book's first edition (including 98 different RD) was elaborated during the implementation of NoRo project.

Training and education initiatives

In Romania, Universities of Medicine and Pharmacy all over the country included in their curricula relevant topics on RD incorporating them in the basic medical training for the undergraduate students. In addition, RD are included in optional/ facultative lectures covering rare diseases for medical students, as well as post graduate lectures on Medical/ Clinical Genetics are organised in major university centres.

The Romanian Society of Medical Genetics provided continued education for medical doctors and other health professionals, organising training courses every year from 2007 until now.

The Romanian Prader Willi Association has developed the www.edubolirare.ro, a platform for rare diseases, and the training courses are adapted to the needs of different professional working in the field of rare diseases. The training courses are authorised and accredited by the Ministry of Education and Ministry of Work but, also, by National Physicians' Collegium when the training is addressed to medical doctors.

In November 2013 the 2nd rare diseases training course for medical journalists in Romania and also a course for parents and personal assistants was debuted in December 2013.

Other training courses have been provided through our workshops organised in different conferences during 2013.

National rare disease events in 2013

The Romanian National Alliance for Rare Diseases has marked Rare Disease Day with many events since 2008, with the support of Eurordis and the Romanian Society for Human Genetics. The alliance coordinates efforts and collects the information about the campaign events organised by the member organisations. To mark Rare Disease Day a march for rare diseases took place on 28 February 2013 in Zalau and a workshop on European Reference Networks and Centres of Expertise in Romania for rare diseases, organised by the National Alliance for Rare Diseases Romania, was also organised the week before on 21 February 2013. In addition to these events the first National Conference on Rare Diseases was organised on 2 March 2013 in partnership with the Romanian Society of Medical Genetics and the National Alliance for Rare Diseases with the aim of bringing together different professionals in the field to exchange information on rare diseases.

The Romanian Europlan conference[4] was held on 24-25 May 2013 in Bucharest to facilitate an open dialogue between all stakeholders (patients, professionals, authorities, politicians, industry, media). This event was organised by ANBRaRo under the patronage of the Ministry of Health. The conference rendered support from the Ministry of Health of Romania, who are motivated to carry forth the National Plan for Rare Diseases in the near future. In addition to updating the national plan for Romania, the conference also discussed establishing relevant procedures for assessing the Centres of Expertise as well as finalising the procedure for appointing the National Committee for Rare Diseases and working groups. The process of reimbursement of orphan drugs in Romania was also analysed and alternative strategies to facilitate access to orphan medication were examined. A push towards rare disease research and the identification of possible sources of funding were considered. In conclusion, the outlook for adoption of the long-awaited National Plan for Rare Diseases in Romania looks promising.

In addition a Campaign for rare cancers and CML was organised in September 2013.

[4] http://www.eurordis.org/sites/default/files/flags/finalreport-romania.pdf

Hosted rare disease events in 2013
No reported events.

Research activities and E-Rare partnership
National research activities
There is no specific research programme for RD in Romania. Research projects aimed to RD are included in the same group with other topics research projects. In 2013 there were few calls for research projects but not rare disease specific. There are currently no other fund-raising initiatives for RD research in Romania.

Participation in European research projects
Romanian team(s) participate/participated in 1 FP7 rare disease related project.

E-Rare
Romania joined the E-Rare consortium in 2012.

IRDiRC
Romanian funding agencies have not yet committed financing to the IRDiRC.

Orphan medicinal products
Orphan medicinal product committee
No specific activity reported.

Orphan medicinal product incentives
No specific activity reported.

Orphan medicinal product availability
From 1 January 2007, date at which Romania became an EU Member State, all medicinal products were required to obtain new authorisations according to EU standards: this created significant delay in the importation of certain orphan medicinal products. As a consequence of the creation of the National Plan for Rare Diseases, the Ministry of Public Health enlarged coverage of orphan medicinal products from July 2008 onwards in their health programme.

The list of orphan medicinal products available/commercialised in Romania and free of charge through the National Health Programme is available on the website of the Romanian National Medicines' Agency[5] and includes: Aldurazyme, Atriance, Busilvex, Carbaglu, Cystadane, Diacomit, Elaprase, Evoltra, Exjade, Fabrazyme, Firazyr, Gliolan, Glivec, Increlex, Inovelon, Litak, Lysodren, Myozyme, Naglazyme, Nexavar, Onsenal, Orfadin, Pedea, PhotoBarr, Prialt, Replagal, Revatio, Revlimid, Savene, Siklos, Soliris, Somavert, Sprycel, Sutent, Tasigna, Thalidomide Pharmion, Thelin, Torisel, Tracleer, Trisenox, Ventavis, Volibris, Wilzin, Xagrid, Xyrem, Yondelis, Zavesca.

Orphan medicinal product pricing policy
There is national debate for regulation in this area.

Orphan medicinal product reimbursement policy
The National Programme for Rare Diseases provides for the reimbursement of 47 orphan medicinal products in Romania.

Other initiatives to improve access to orphan medicinal products
In Romania there are several ways of accessing orphan medicinal products via Order N° 962/2006 for approval of the application of art. 699, paragraph (1) of Law N° 95/2006 including: compassionate use of drugs for a certain patient (in the case where the drug already has marketing authorisation); compassionate use of drugs for a group of patients with an invalidating disease, either chronic or serious, or a disease considered to be life-

[5] http://www.anm.ro/_/Lista%20medicamentelor%20orfane%20valide%20in%20Romania.xls

threatening (the provision of a centrally authorised product); the off-label use of drugs. However, the public payer will not always reimburse the orphan medicinal product provided.

Many companies (i.e. Genzyme, Novartis) use humanitarian programs, sponsorship or partnership with medical societies in order to provide the orphan medicinal products, like Tasigna, Evoltra, Fabryzyme, Myozyme, and others. This way was a successful manner to give access of the patients to some orphan medicinal products. Other ways include the enrolment of patients in clinical trials for orphan medicinal products. Doctors have to prescribe the compassionate / off-label drugs and follow-up the patients.

Other therapies for rare diseases

The NoRo Centre offers to patients with rare diseases access to therapies such as: medical evaluation, groups of support, psychological counseling, behaviour therapy, speech therapy, physical therapy, massage, sensorial therapy, hydro - kinetic – electric therapy, ergo-therapy, weight management, educational and occupational therapy. The NoRo Centre is accredited for specialised social services by the Ministry of Work and also for medical services from Ministry of Health; it is authorised for training by Ministry of Education and for research capacity by National Agency for Scientific Research. It is a resource centre and could be part of the patients' pathway and network of the future centres of expertise in Romania, ensuring continuity of care while implementing quality standards of services. It is the main goal of ExpertRARE – a project developed by Romanian Prader Willi Association and co-funded by a grant from Switzerland through the Swiss Contribution to the enlarged European Union.

Orphan devices

No information reported yet.

Specialised social services

The NoRo Centre was established by Romanian Prader Willi Assoctian in 2011 with Norwegian funding through Norwegian Cooperation Program and it is working for 1 year and a half, supported partly by the local and national authorities. The service includes training courses, information and guidance services, and provision of information about social services, documentation and research. Daily support therapies, medical and psychological consultations are also provided by NoRo centre. This service also aims to create a bridge between patients/families and all the stakeholders involved in patient care, such as medical services, rehabilitation and Therapeutic services, social services and social support authorities, education professionals and other professionals directly working with RD patients.

It is funded by the Local Council Zalau and County Council Salaj and other projects for the development of the services. It is mentioned on the map Specialized Social Services developed by Eurordis in the EUCERD Joint Action for Rare Diseases (www.eurordis.org; www.eucerd.eu;). The guidelines for specialised social services produced in the Joint Action started to be translated and will be available on RONARD website in June 2014.

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2013 IN ROMANIA

National plan/strategy for rare diseases and related actions

By the end of 2012 the inclusion of the National Plan for Rare Diseases in the National Public Health Strategy was the next step concerning the implementation and monitoring of the plan by the end of 2013. At the end of 2013, the National Council for RD (NCRD) was created by the Ministry of Health (MoH) through a Ministerial Order (1215/2013).

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- The second Europlan National Conference[7] was organised under the auspices of Ministry of Health of Romania in Bucharest, on 24-25 May 2013. The Conference aimed at facilitating dialogue, participation and involvement of all stakeholders in the field of rare diseases in Romania (patients, professionals, authorities, politicians, industry, media, etc.) in order to address solutions that need to be taken to update the proposals of the National Plan for Rare Diseases (NPRD).
- A Resolution of the Europlan Conference has been agreed by all the stakeholders involved in rare diseases field in Romania around the main objectives and future activities for rare diseases in 2013 and the following NPRD for the period: 2014-2020.

Due to the complexity of political and socio-economic situation at the national level, no budget has been allocated for the NPRD in 2013 but it is expected financial support through budgetary integration on MoH.

Rare cancers are mentioned in the NPRD in Romania and future actions will be included for the period 2014-2020.

Centres of expertise

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Registries

Since 2013 NCRD deals with the issue of a national registry for RD but no public financial resources have been allocated so far. At the moment, NCRD selects the common data elements for Rd databases and extends the work by ensuring that the dataset are defined in the same way, using the same standards and same terms. Also, NCRD is considering appropriate EU standardised databases in order to find how to harmonise, share and exchange information. In addition, NCRD establishes baseline measures for data safety and protection. National registries and databases can be use to plan and manage services in the field of RD.

[6] http://www.ms.gov.ro/documente/ordin%20boli%20rare_823_1638.pdf
[7] http://www.eurordis.org/sites/default/files/flags/finalreport-romania.pdf

Neonatal screening policy

In 2013, according to the national health policy a mandatory newborn screening program was available for two diseases, phenylketonuria (PKU) and congenital hypothyroidism (CHT), with the goal of screening all infants born in the country. The screening is performed in 4 public medical centres throughout the country (Bucharest, Iasi, Cluj-Napoca and Timisoara). The current health policy improved the management of screening for PKU and CHT at local level, reaching 80% coverage for newborns.

In the last two years, efforts were made via the Health Programme at local level in order to generalise and improve the organisation of neonatal screening for these two disorders, as well as to introduce screening for other (rare) diseases for which some form of treatment is available. Newborn screening tests for other diseases are provided by private clinics / laboratories at full cost or can be carried out in the framework of research programmes (e.g., hearing loss).

However, despite the screening programme, some tests are not available nationwide due to logistic and resource problems and no additional RD have been added to the panel of diseases tested in 2013.

Also, in 2013, Romania continued to join in efforts aimed for the preparation of European guidelines on diagnostic tests or population screening (EUNENBS: European Network of Experts on Newborn Screening) respecting national decisions and competences.

National alliances of patient organisations and patient representation

At the end of 2013, the National Council for RD (NCRD) was created by the Ministry of Health (MoH) through a Ministerial Order (1215/2013) and 3 representatives of the RONARD are full members in the executive committee of the council. The council, as an advisory group for MoH in the field of RD is playing an important support role in the consultation and implementation of the NPRD.

Sources of information on rare diseases and national help lines

Official information centre for rare diseases

In October 2013, RONARD has also launched an online platform for the development of organisational capacity of patients' organisations in Romania: www.aspac.ro.

Help line

At the end of 2013, The Romanian Prader Willi Association has initiated a restructuration of the NoRo HelpLine in order to develop the network of professionals that support the activity at national level.

Guidelines

Before 2013, clinical practice guidelines have been produced for every medical specialty including RD but several of them are at national level (e.g., diagnosis and follow-up in Oncology, Pulmonary Hypertension or Hereditary Angioedema).

In Romania the following translated or elaborated RD guidelines were launched in 2013:

- The Romanian Prader Willi Association translated and printed "A medical alert for Prader Willi Syndrome";
- In cooperation with Romanian Society for Human Genetics and many other professionals a book entitled "Medical Alert in Rare Genetic Diseases, an emergency guideline for 18 rare diseases clusters" was re-published. The book's first edition (including 98 different RD) was elaborated during the implementation of NoRo project.

Training and education initiatives

In November 2013 the 2nd rare diseases training course for medical journalists in Romania and also a course for parents and personal assistants was debuted in December 2013.

Other training courses have been provided through our workshops organised in different conferences during 2013.

National rare disease events in 2013

The Romanian National Alliance for Rare Diseases has marked Rare Disease Day with many events since 2008, with the support of Eurordis and the Romanian Society for Human Genetics. The alliance coordinates efforts and collects the information about the campaign events organised by the member organisations. To mark Rare Disease Day a march for rare diseases took place on 28 February 2013 in Zalau and a workshop on European

Reference Networks and Centres of Expertise in Romania for rare diseases, organised by the National Alliance for Rare Diseases Romania, was also organised the week before on 21 February 2013. In addition to these events the first National Conference on Rare Diseases was organised on 2 March 2013 in partnership with the Romanian Society of Medical Genetics and the National Alliance for Rare Diseases with the aim of bringing together different professionals in the field to exchange information on rare diseases.

The Romanian Europlan conference[8] was held on 24-25 May 2013 in Bucharest to facilitate an open dialogue between all stakeholders (patients, professionals, authorities, politicians, industry, media). This event was organised by ANBRaRo under the patronage of the Ministry of Health. The conference rendered support from the Ministry of Health of Romania, who are motivated to carry forth the National Plan for Rare Diseases in the near future. In addition to updating the national plan for Romania, the conference also discussed establishing relevant procedures for assessing the Centres of Expertise as well as finalising the procedure for appointing the National Committee for Rare Diseases and working groups. The process of reimbursement of orphan drugs in Romania was also analysed and alternative strategies to facilitate access to orphan medication were examined. A push towards rare disease research and the identification of possible sources of funding were considered. In conclusion, the outlook for adoption of the long-awaited National Plan for Rare Diseases in Romania looks promising.

In addition a Campaign for rare cancers and CML was organised in September 2013.

Other therapies for rare diseases

The NoRo Centre offers to patients with rare diseases access to therapies such as: medical evaluation, groups of support, psychological counseling, behaviour therapy, speech therapy, physical therapy, massage, sensorial therapy, hydro - kinetic – electric therapy, ergo-therapy, weight management, educational and occupational therapy. The NoRo Centre is accredited for specialised social services by the Ministry of Work and also for medical services from Ministry of Health; it is authorised for training by Ministry of Education and for research capacity by National Agency for Scientific Research. It is a resource centre and could be part of the patients' pathway and network of the future centres of expertise in Romania, ensuring continuity of care while implementing quality standards of services. It is the main goal of ExpertRARE – a project developed by Romanian Prader Willi Association and co-funded by a grant from Switzerland through the Swiss Contribution to the enlarged European Union.

[8] http://www.eurordis.org/sites/default/files/flags/finalreport-romania.pdf

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[9] 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.

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