2013 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE

STATE OF THE ART OF RARE DISEASE ACTIVITIES IN CYPRUS

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.

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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
g - 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).

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 2012. 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 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 2012
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

Each part contains the following 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 Cyprus. 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.
RARE DISEASE ACTIVITIES IN CYPRUS

Definition of a rare disease
Stakeholders in Cyprus accept the European Orphan Drug Regulation definition of a prevalence of no more than 5 in 10,000 individuals.

National plan/strategy for rare diseases and related actions
The Cyprus National Strategic Plan for Rare Diseases (CNSPRD) was established following the European Council’s Recommendation that each Member State should develop a national plan or strategy on rare diseases, preferably by the end of 2013. The final version of the CNSPRD was developed by a national steering committee for rare diseases, which consisted of Ministry of Health officials, experts in various fields relating to rare diseases as well as patient representatives, following a public consultation (the second) with local stakeholders including patient’s representatives, in March 2012. The CNSPRD was approved by the Council of Ministers of the Republic of Cyprus in November 2012.

The main objective of the National Strategic plan is to ensure that patients with rare diseases will have access to high quality care (diagnostics, treatments as well as rehabilitation for those living with the disease). The CNSPRD is based on the following 5 pillars:

(a) Prevention – Early Diagnosis
(b) Treatment and Management
(c) Palliative Care / Social Inclusion / Support
(d) Registries/Epidemiology
(e) Research

Following the approval of the CNSPRD, the National Committee for Rare Diseases was appointed by the Council of Ministers with the task of implementing as well as monitoring the progress of the plan. In addition, the National Committee for Rare Diseases is responsible for defining a number of priority actions with objectives and follow-up mechanisms.

Centres of expertise
Currently, no officially designated centres of expertise for rare diseases exist in Cyprus since official guidelines and procedures are yet to be established at the administrative level. Nevertheless, a number of institutions are currently functioning at the level of centres of expertise. Among these are the Cyprus Institute of Neurology and Genetics, the Makarios Hospital for Mother and Child and its specialised clinics, the Cyprus Thalassemia Centre, the Centre for the Study of Hematological Malignancies, the Bank of Cyprus Oncology Centre as well as a number of specialised clinics and departments at the Nicosia General Hospital and other hospitals. The Cyprus Institute of Neurology and Genetics operates as a centre of research, treatment and management for various rare neurological and genetic conditions. The Clinical Genetics Clinic, located both at the Cyprus Institute of Neurology and Genetics and the Archbishop Makarios III Hospital, is involved in the management of over 3500 patients and their families living with or at risk of a genetic condition in Cyprus. The Archbishop Makarios III Hospital for Mother and Child, in Nicosia, is the main referral hospital for children and adolescents where most young patients with rare diseases are referred for diagnosis and management. Several specialised clinics in this hospital operate as referral clinics for rare diseases by specialty such as, paediatric endocrinology/nephrology/cardiology/ neurology/ pulmonology/ infectious diseases, etc. The Cyprus Thalassaemia Centre is the main centre for screening (premarital), counselling and management of thalassaemia on the island. The Centre is based in Nicosia but also holds special clinics for the management and care of patients with hemoglobinopathies in all other public hospitals on the island. The newly established Centre for the Study of Hematological Malignancies operates as a centre of research and diagnosis of various rare haematological malignancies while management is offered by the Haematology clinics of the Nicosia and Limassol General Hospitals. The Bank of Cyprus Oncology Centre and the Oncology department of the Nicosia General Hospital are the main referral centres for the diagnosis, management and treatment of rare cancer syndromes. Several other departments and specialised clinics serve as referral centres for rare disorders including but not limited

1 http://www.moh.gov.cy/MOH/MOH.nsf/All/CD61A0712284C04422579DC0023AF8A/$file/Strategic%20Plan%20Rare%20Diseases.pdf
to rare haematological and congenital heart disorders, cardiomyopathies, etc. The majority of these clinics are based at the Nicosia General Hospital.

The procedure for officially designating Centres of Expertise for rare diseases in Cyprus is currently under discussion.

**Registries**

There is currently no designation process for rare disease registries in Cyprus, but this will be one of the considerations of the National Plan for Rare Diseases. Several registries have been formed by physicians and scientists at various specialised clinics and laboratories. Also a few patient organisations have their own registries based on their members. Cyprus participates in the EUROCARE CF European registry.

**Neonatal screening policy**

There are nationwide schemes for neonatal screening, which include screening for phenylketonuria and congenital hypothyroidism. Also a nationwide screening for congenital hearing deficit exists. An advisory committee has been established by the Minister of Health with the task of addressing the current situation of newborn screening in Cyprus and to evaluate the new emerging needs and possible expansion of the offered screening program. The committee has drafted a report summarizing their findings and highlighting Cyprus’s future needs. The report is currently under review by the Ministry of Health officials.

**Genetic testing**

Genetic testing is available for many genetic disorders. This includes conventional and molecular cytogenetics, screening for metabolic disorders, neurogenetics, genetic testing for inherited cancers as well as for other predisposition genes, thalassemia molecular diagnostics, screening for other haematological genetic disorders and many others. In summary, diagnostic tests are available in Cyprus for around 220 genes and an estimated of 110 rare diseases².

**National alliances of patient organisations and patient representation**

In June 2010, the Cyprus Alliance for Rare Disorders (CARD)³ was established with the aim of uniting the voices of all patients with rare diseases at a national level. The principal goals of the Alliance are the following: to lend support to the national rare disorders programme; to support the efforts of rare disease patients for improvement of prevention, medical treatment, as well as social and other services related to each of the rare disorders to improve the health and quality of life rare diseases patients; to provide support and continuous education to the patients and their families concerning the latest developments in medicine and research; and to raise awareness regarding rare diseases in the Cypriot society. Now legally registered, the Cyprus Rare Disease Alliance is becoming Cyprus’ representative for patients in health organisations and institutions at European and international level. Furthermore many other patient organisations exist, representing (but not exclusively) rare disease patients with a disability such as vision deficit or hearing loss, patients with mental retardation, patients with Down syndrome, patients with congenital heart disorders, etc.

**Sources of information on rare diseases and national help lines**

**Orphanet activities in Cyprus**

Since 2004 there is a dedicated Orphanet team in Cyprus, currently hosted by the Archbishop Makarios III Medical Centre Clinical Genetics Department. This team was designated in 2010 as the Orphanet National team for Cyprus by the Medical and Public Health Services of Cyprus. The Orphanet Cyprus 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 Cyprus for entry into the Orphanet database. A national website of Orphanet Cyprus⁴ is also operational aiming to provide information on local activities in the field of rare diseases.

**Official information centre for rare diseases**

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

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² Source: Cyprus Institute of Neurology and Genetics

³ http://www.thalassaemia.org.cy/cyprus_alliance.html

⁴ http://www.orpha.net/national/CY-EL/index/homepage/
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Help line
Currently, no rare disease help line is operational in Cyprus.

Other sources of information
The Cyprus Institute of Neurology and Genetics is the main source of information for several neurological and other genetic disorders. Information on rare diseases is also published by Gene Net Cyprus\(^1\), a project that aimed to create a bicomunal network for genetic diseases bringing together health professionals patients and families. The project has produced trilingual leaflets on genetic conditions in Cyprus: 6 leaflets were published in English, Turkish and Greek. All these documents are available on the Gene Net website which provides links to Orphanet. Furthermore, the Thalassemia National Centre is the main source of information for haemoglobinopathies. In addition, several specialised clinics and organisations, which operate in Cyprus are functioning as sources of information for specific disorders.

Best practice clinical guidelines
Internationally accepted best clinical practice guidelines are being used in Cyprus as reference documents.

Training and education initiatives
Activities have been organised in this field with a training/education angle, including conferences, courses and lectures within main and teaching hospitals, especially the paediatric department of the Makarios Hospital and the Cyprus Institute of Neurology and Genetics, and also within meetings of local scientific societies. These included teaching lectures and presentations on rare genetic syndromes and rare metabolic disorders. The Cyprus Institute of Neurology and Genetics serves also as a satellite centre to the EGF courses which include several activities on rare genetic disorders.

National rare disease events in 2012
The Ministry of Health organised a seminar on 1 March 2012 to honour Rare Disease Day. The seminar was addressed by the Minister of Health and was attended by many health professionals, scientists and stakeholders in the field of rare diseases as well as patients and their representatives. During the seminar, members of the National Steering Committee, presented the first draft of the National Strategic Plan for Rare Diseases, followed by discussion and public consultation. Other rare disease-related events in 2012 included the Inherited Neurological Disorders Seminar organised by the Cyprus Society of Human Genetics and the Neurological Society of Cyprus (2 February 2012, Nicosia), the Familial Cancer Management Seminar organised by the Cyprus Oncological Society and the Department of EM / Molecular Pathology of the Cyprus Institute of Neurology and Genetics (22 March 2012), the Galactosemia seminar organised by the Pediatric Clinic of the Makarios III Hospital and the Biochemical Genetics Department of the Cyprus Institute of Neurology and Genetics (29 March 2012), the 8th Postgraduate Pediatrics Conference (which included topics on rare metabolic diseases and rare genetic diseases) organised by the Pediatric Clinic of the Makarios III Hospital in April 2012. Additionally, several events took place combining scientific seminars and public awareness such as the Scleroderma International Day.

Hosted rare disease events in 2012
Cyprus hosted a number of rare disease related events including: the 3rd Pan-European Conference on Haemoglobinopathies and Rare Anaemias organised by the Thalassaemia International Federation in October 2012, the XIXth NeuroMediterannée Conference, organised by the Cyprus Institute of Neurology and Genetics, the Cyprus Neurological Society and the NeuroMediterranée Society in November 2012, the 3rd International Conference of the Cyprus Society of Human Genetics in November 2012, and the 3rd Thalassemia Conference organised by the Cyprus Institute of Neurology and Genetics and the Cyprus Thalassaemia Centre in November 2012. Furthermore, experts in the management and treatment of various rare diseases presented the latest advancements in their fields of expertise as part of general or more specialised medical conferences.

Research activities and E-Rare partnership
National research activities
There are no dedicated research funds for rare diseases in Cyprus. In general funding opportunities are offered by the Cyprus Research Promotion Foundation and the Cyprus Institute of Neurology and Genetics. In addition,
Telethon is organised by the Cyprus Institute of Neurology and Genetics (CING) to support scientific research, including research on rare diseases.

**Participation in European research projects**
Cyprus participates, or has participated, in European rare disease research projects including: EUROPEAN LEUKEMIA NET, Ithanet, LEISHMED and MYELINET.

**E-Rare**
Cyprus is currently not a member of E-Rare and does not participate in their calls.

**IRDiRC**
Cyprus is not currently a committed member of the IRDiRC.

**Orphan drugs**
**Orphan drug committee**
No specific activity reported.

**Orphan drug incentives**
No specific activity reported.

**Orphan drug market availability situation**
No specific information was reported on the orphan drugs marketed in Cyprus. Several orphan drugs have been requested through the Department of Pharmaceutical Services of the Ministry of Health and they were approved for use, i.e. Enzyme Replacement Treatment namely for Gaucher, Maroteaux–Lamy and Pompe disease patients. Other orphan drugs were also requested.

**Orphan drug pricing policy**
No specific activity reported.

**Orphan drug reimbursement policy**
No specific activity reported.

**Other initiatives to improve access to orphan drugs**
Reimbursement is available for the compassionate use of orphan drugs.

**Other therapies for rare diseases**
No specific activity reported.

**Orphan devices**
No specific activity reported.

**Specialised social services**
Social services for patients suffering of disabilities as a result of rare disorders are in place. The legislation is not specific to rare diseases but concerns the nature of the disability.
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DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2012 IN CYPRUS

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LIST OF CONTRIBUTIONS

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Contributions in 2011
Lily Cannon (Cyprus Alliance for Rare Disorders)
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Contributions in 2012
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Cyprus Alliance for Rare Disorders
The National Committee for Rare Diseases agreed on this updated report.

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SELECTED BIBLIOGRAPHY AND SOURCES

- Cyprus National Strategic Plan for Rare Diseases
  http://www.moh.gov.cy/MOH/MOH.nsf/All/CD61A07312284C0A422579DC0023AF8A/$file/Strategic%20Plan%20Rare%20Diseases.pdf
- Gene Net Cyprus
  http://www.genenet.org.cy/English/index.htm
- Orphanet Cyprus national website
  http://www.orpha.net/national/CY-EL/index/homepage/
- Cyprus Alliance for Rare Disorders
  http://www.thalassaemia.org.cy/cyprus_alliance.html

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.

All websites and documents were last accessed in May 2013. A more detailed list of sources is available in the full report: