EUCERD
European Union Committee of Experts on Rare Diseases

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 CYPRUS

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

EUCERD European Union Committee of Experts on Rare Diseases
2011-22-01
EUCERD Joint Action

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.

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/2012/09/2012ReportStateofArtRDActivitiesCY.pdf

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 Porphyria 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 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[1].

[1] http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf

RARE DISEASE ACTIVITIES IN CYPRUS

Definition of a rare disease

Stakeholders in Cyprus accept the European Regulation on Orphan Medicinal Products definition of a prevalence of no more than 5 in 10000 individuals.

National plan/strategy for rare diseases and related actions

The first draft of the National Strategic plan for rare diseases was prepared by the National Steering Committee for rare diseases following numerous meetings and hard work by all involved parties. The National Steering Committee for rare diseases consists of Ministry of Health officials, experts in different fields related to rare diseases and patient representatives.

Initially, in early 2011 a document was prepared by the National Steering committee which consisted of the following three parts: a) a detailed description of the current situation and practices in the field of rare diseases in Cyprus, b) an extensive analysis of the five chapters to be addressed in the strategic plan, namely prevention, diagnosis, treatment and management, palliative care - social inclusion – supportive measures, registries and research and c) a proposed action plan.

In June 2011 a two-day workshop was organised by this committee with the support and under the auspices of the Ministry of Health. During this workshop, local stakeholders from all areas and fields relevant to rare diseases participated in brainstorming sessions, commented and shared their experiences on the current situation in Cyprus and forwarded their suggestions regarding the development of a National Strategic plan to the members of the National Steering committee. The leader of the Europlan project was invited to address this meeting with a lecture and to share experiences: different working groups reviewed the current situation and the proposed actions for designing a national plan of action for rare diseases. The workshop was a success in the sense that it achieved extensive discussions and active involvement of all participants and ended with a strong conclusive report of strong interest and commitment for active involvement of many, in the future steps for achieving progress and agreement on a National plan of action for rare diseases in Cyprus. The National Steering Committee undertook the responsibility of reviewing the conclusions, comments and suggestions of the workshop in order to enable those that hold the general consensus in the strategic plan. The final draft of the strategic plan will be reviewed once again by the local stakeholders in a second workshop which will take place on the 28 of February 2012 before being finalised and submitted to the Ministerial Council for its final approval. The overall goal is to proceed to the finalising of the proposed strategic plan and its approval by the Council of Ministers within 2012.

Centres of expertise

There are currently no officially designated centres of expertise for rare diseases in Cyprus as there are no criteria or procedures determined yet at the administrative level. Nevertheless there are several institutions which are already functioning at the level of centres of expertise.

The Cyprus Institute of Neurology and Genetics offers diagnosis, treatment and management for various rare neurological and other genetic disorders and has a strong research output. Many research projects on rare disorders have been completed and others are in progress. These include studies on epidemiology, biochemical and molecular characterisation, pathogenetic mechanisms and novel therapies. The Institute is considered a centre of excellence in the fields of neurology and genetics and acts as a referral and training centre for neighbouring countries.

The Archbishop Makarios III Hospital for Mother and Child 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, immunodeficiencies etc. The Clinical Genetics Clinic operating both at Makarios Hospital and the Cyprus Institute of Neurology and Genetics is involved in the management of a large number of patients and their families living with or at risk of a genetic condition.

The Cyprus Thalassaemia Centre is the main centre for screening (premarital), counselling and management of thalassaemia, sickle cell disease and other rare anaemias on the island. The Centre was designated as a WHO collaborating centre in 1985 because of its expertise in community awareness, prevention strategies and educational activities for both patients and medical specialists. 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.

Several specialised clinics and departments in Nicosia General Hospital and other hospitals serve as referral centres for disorders such as rare haematological diseases, rare heart, rheumatologic and immunological disorders etc.

The Center for the Study of Haematological Malignancies was established recently with the aim of offering lifesaving diagnostic and prognostic information which help guide therapeutic decisions and monitoring treatment of rare haematological disorders. The creation of this newly established Center will provide the opportunity for the enhancement of hemato-oncology services in Cyprus and will facilitate the execution of high-impact research in the field of rare hematological malignancies. This centre is housed together with the Karaiskakio Foundation which has the highest per capita ratio of bone marrow donors in the world.

The procedure for officially designating centres of expertise for rare diseases in Cyprus is included as part of the National Strategic plan for Rare Diseases. Hence it is expected that once the strategic plan is approved centres of expertise for rare diseases in Cyprus will be officially designated.

Pilot European Reference Networks

Cyprus participates, or has participated, in the following European Reference Networks for rare diseases: Network of centres of expertise for dysmorphology (Dyscerne),, European Network for rare and congenital anaemias (ENERCA) and Together Against Genodermatoses (TAG).

Registries

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. Recently, in 2011, 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.

Genetic testing

Genetic testing is available for many rare genetic disorders. This includes chromosomal abnormalities, metabolic disorders, neurogenetic disorders, thalassaemia and other rare hemoglobinopathies, heritable cancers and many others. Genetic testing is available in Cyprus for around 200 genes and an estimated of 100 rare diseases.

National alliances of patient organisations and patient representation

In June 2010, the Cyprus Alliance for Rare Disorders (CARD[2]) 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 announced recently by the Ministry of Health; 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 of 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. In 2011, CARD expanded their awareness programme through the organisation of several educational and informative events. These events had as a focus increasing knowledge about the challenges that rare disease patients face and stress the need for a national plan for rare disorders.

In addition to CARD, numerous patient organisations are active and support and represent patients with rare diseases as well as aim to raise public awareness and improve quality of life of their members.

[2] http://www.thalassaemia.org.cy/cyprus_alliance.html

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 Genetic Department. This team was designated in 2010 as the Orphanet national team for Cyprus by the Medical and Public Health Services of Cyprus. 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. Since December 2011, the national website of Orphanet Cyprus[3] is operational aiming to provide information on local activities in the field of rare diseases.

Official information centre for rare diseases

Officially there is no information centre for rare diseases apart from Orphanet in Cyprus.

Help line

Currently, there is no dedicated rare disease help line in Cyprus.

Other sources of information

The Cyprus Institute of Neurology and Genetics is a source of information for several neurological and genetic disorders. Information on rare diseases is also published by Gene Net Cyprus, a project that aims to create a bicommunal 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[4] () which provides links to Orphanet. Furthermore, the Thalassemia National Centre is the source of information for haemoglobinopathies. Several specialised clinics and organisations are functioning as sources of information for specific disorders.

Good practice 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, such as 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. Furthermore, training and education sessions were organised within meetings of local scientific societies. These included teaching lectures and presentations on rare genetic syndromes, metabolic disorders and rare liver diseases in childhood. In addition, the Cyprus Institute of Neurology and Genetics serves also as a satellite centre to the European Genetics Foundation[5] courses which include several activities on rare genetic disorders. The Cyprus Thalassaemia Centre serves as a satellite centre to ESH courses through ENERCA project.

National rare disease events in 2011

The Cyprus Society of Human Genetics organised a seminar on 24 February 2011 to honour Rare Disease Day. The seminar was addressed by representatives of the Ministry of Health and CARD and was attended by many health professionals, scientists and stakeholders in the field of rare diseases as well as patients and their representatives. In addition, CARD organised a Press Conference on 28 February 2011, in honour of Rare Disease Day, to highlight issues concerning rare diseases in Cyprus.

Hosted rare disease events in 2011

Cyprus hosted a number of rare disease-related events in 2011. These include the Pancyprian Scientific Conference on Rare Diseases organised by CARD in March 2011, the "Together Against Genodermatoses, TAG" International Seminar organized by the Clinical Genetics Department of the Cyprus Institute of Neurology and Genetics and the Makarios III Hospital in November 2011 and the 2nd Thalassemia Conference organized by the Cyprus Institute of Neurology and Genetics and the *Pancyprian Antianemic Association* in December 2011. Furthermore, as part of specialized medical conferences experts in the management and treatment of GIST,

[3] http://www.orpha.net/national/CY-EL/index/homepage/
[4] http://www.genenet.org.cy
[5] www.eurogene.org

myelodysplastic syndromes and other rare diseases presented the latest advancements in their fields of expertise.

Research activities and E-Rare partnership

National research activities

Funding opportunities for rare disease research (without being specifically ear-marked for this purpose) are offered by the Cyprus Research Promotion Foundation as well as European and International organizations (FP7, Muscular Dystrophy Association, USA and other patient organizations). Telethon is an international charitable institution which is organised by the Cyprus Institute of Neurology and Genetics (CING) to support scientific research on neuromuscular diseases. A large proportion of net revenue (approximately 30%) from the Telethon is allocated to the Association for Patients with Muscular Dystrophy and the rest supports specific research projects conducted at the Institute. The selection of these investigations is made with the help of an independent international scientific committee.

Scientific community in Cyprus is engaged into research activities aiming towards establishing new therapies for a number of rare disorders, in particular gene therapy for rare hemoglobinopathies and neurological disorders. Furthermore, a novel test for non-invasive prenatal diagnosis for Down syndrome which only uses a small amount of blood from the pregnant women was developed and is currently being validated.

Participation in European research projects

Cyprus participates, or has participated, in European rare disease research projects including: EUROPEAN LEUKEMIA NET, Ithanet, LEISHMED, MYELINET, ENERCA and Chain of Trust.

E-Rare

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

IRDiRC

Funding agencies in Cyprus are not yet committed members of the IRDiRC.

Orphan medicinal products

Orphan medicinal product committee

At present there is no orphan medicinal product committee in Cyprus but a dedicated pharmaceutical officer deals with all matters relating to orphan medicinal products in collaboration with treating physicians and Ministry of Health officials.

Orphan medicinal product incentives

No specific activity reported.

Orphan medicinal product market availability situation

No specific information was reported on the orphan medicinal products marketed in Cyprus. Several orphan medicinal products 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 and Pompe disease patients. Other orphan medicinal products were also requested.

Orphan medicinal product pricing policy

No specific activity reported.

Orphan medicinal product reimbursement policy

The Medical Health Services of the Republic of Cyprus cover the cost of orphan medicinal products administered to patients provided that their use is approved by the Pharmaceutical Committee.

Other initiatives to improve access to orphan medicinal products

Reimbursement is available for the compassionate use of orphan medicinal products.

Orphan devices

The Medical Health Services of the Republic of Cyprus cover the cost of orphan devices provided to patients in cases where their use is approved by the relevant Medical Committee.

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.

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN CYPRUS

National plan/strategy for rare diseases and related actions
The first draft of the National Strategic plan for rare diseases was prepared by the National Steering Committee for rare diseases following numerous meetings and hard work by all involved parties. The National Steering Committee for rare diseases consists of Ministry of Health officials, experts in different fields related to rare diseases and patient representatives.

Initially, in early 2011 a document was prepared by the National Steering committee which consisted of the following three parts: a) a detailed description of the current situation and practices in the field of rare diseases in Cyprus, b) an extensive analysis of the five chapters to be addressed in the strategic plan, namely prevention, diagnosis, treatment and management, palliative care - social inclusion – supportive measures, registries and research and c) a proposed action plan.

In June 2011 a two-day workshop was organised by this committee with the support and under the auspices of the Ministry of Health. During this workshop, local stakeholders from all areas and fields relevant to rare diseases participated in brainstorming sessions, commented and shared their experiences on the current situation in Cyprus and forwarded their suggestions regarding the development of a National Strategic plan to the members of the National Steering committee. The leader of the Europlan project was invited to address this meeting with a lecture and to share experiences: different working groups reviewed the current situation and the proposed actions for designing a national plan of action for rare diseases. The workshop was a success in the sense that it achieved extensive discussions and active involvement of all participants and ended with a strong conclusive report of strong interest and commitment for active involvement of many, in the future steps for achieving progress and agreement on a National plan of action for rare diseases in Cyprus. The National Steering Committee undertook the responsibility of reviewing the conclusions, comments and suggestions of the workshop in order to enable those that hold the general consensus in the strategic plan. The final draft of the strategic plan will be reviewed once again by the local stakeholders in a second workshop which will take place on the 28 of February 2012 before being finalised and submitted to the Ministerial Council for its final approval. The overall goal is to proceed to the finalising of the proposed strategic plan and its approval by the Council of Ministers within 2012.

Centres of expertise
The procedure for officially designating centres of expertise for rare diseases in Cyprus is included as part of the National Strategic plan for Rare Diseases. Hence it is expected that once the strategic plan is approved centres of expertise for rare diseases in Cyprus will be officially designated.

Neonatal screening policy
In 2011, 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.

National alliances of patient organisations and patient representadtion
In 2011, the Cyprus Alliance for Rare Disorders (CARD) expanded its rare disease awareness programme through the organisation of several educational and informative events. These events had as a focus increasing knowledge about the challenges that rare disease patients face and stress the need for a national plan for rare disorders.

National rare disease events in 2011

The Cyprus Society of Human Genetics organised a seminar on 24 February 2011 to mark Rare Disease Day. The seminar was addressed by representatives of the Ministry of Health and CARD and was attended by many health professionals, scientists and stakeholders in the field of rare diseases as well as patients and their representatives. In addition, CARD organised a Press Conference on 28 February 2011, in honour of Rare Disease Day, to highlight issues concerning rare diseases in Cyprus.

Research activities and E-Rare partnership

IRDiRC

Funding agencies in Cyprus are not yet committed members of the IRDiRC.

LIST OF CONTRIBUTIONS[6]

Contributions in 2010
Violetta Anastasiadou and Turem Delikurt *(Orphanet Cyprus, Archbishop Makarios III Medical Centre Clinical Genetics Department)*

Contributions in 2011
Lily Cannon *(Cyprus Alliance for Rare Disorders)*
Violetta Anastasiadou *(Orphanet Cyprus, Ministry of Health – Medical Public Health Services)*

Contributions in 2012
Violetta Anastasiadou *(Orphanet Cyprus, Ministry of Health – Medical Public Health Services)*
Cyprus Alliance for Rare Disorders

Validated by: Violetta Anastasiadou (EUCERD Representative Cyprus, Ministry of Health – Medical Public Health Services)

SELECTED BIBLIOGRAPHY AND SOURCES[7]

- Gene Net Cyprus
  http://www.genenet.org.cy/
- 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

[6] 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.
[7] 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