State of the Art of Rare Disease - Activities in EU Member States and Other European Countries

Cyprus Report

Definition of a Rare Disease

Cyprus has adopted the European Commission definition of a rare disease (i.e. a disease affecting no more than 5 per 10,000 people, as per Regulation (EC) No 141/2000). The National Strategic Plan espouses this definition.

Status Quo of any National Plan of Strategy for Rare Diseases

Cyprus has had a National Strategic Plan for Rare Diseases since late 2012.

The draft of the 'Cyprus National Strategic Plan for Rare Diseases' (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, and patient representatives. A public consultation (the second of its kind) supported the finalisation of the draft, which involved local stakeholders (including patients' representatives). **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 (encompassing diagnostics, treatments, and also rehabilitation for those living with the disease).

Content and Funding:

The CNSPRD is based on the following 5 pillars:

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

There is no dedicated funding associated with the CNSPRD and its activities; instead, funding is incorporated into the general budget.

Implementation and Monitoring:

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. The Committee met for the first time in January 2013 and continues to meet - this body is multistakeholder in composition (healthcare professionals, physicians, pharmacists, therapists, patient organisation representatives, volunteering organisation representatives, researchers, public health authorities and information technology scientists) and can be described as 'functioning' (although meetings are held irregularly). The members of the committee are active within different pillars of the national strategy such as public awareness, education, and the investigation of ways to promote the rights of patients within different fields, i.e. social inclusion. Work is also being conducted through subcommittees and devoted subgroups. Although rare cancers are included in the CNSPRD, they are also addressed by the 'National Plan against Cancer'; therefore, collaboration was ensured between the National Committee for Rare Diseases and the National Committee for Cancer. Currently there is an effort to introduce indicators which will evaluate the progress and implementation of the National Strategy.

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Achievements of the Strategic Plan to-date

In 2013, the actions defined in the the CNSPRD were prioritised, and those requiring little or no budget (such as training initiatives and public awareness-raising activities) were put into action. In recent years, workshops, lectures, seminars and interviews -addressing the general public, university students, health-care professionals and other relevant groups- were organised. In addition, during 2013 the National Committee for Rare Diseases addressed the three relevant ministries, namely the Ministries of Health, Education & Culture, and Labour & Social Insurance, in order to establish a dialogue with authorities on important policy matters. Topics discussed included the necessity of introducing a helpline, as well as the introduction of a special medical card and an alert card for patients.

The major successes of the National Strategic Plan to-date may be summarised as follows:

- Increased public awareness of rare diseases
- Patient empowerment
- The provision of education through professional bodies
- The inclusion of rare diseases into the political agenda

Importantly, the CNSPRD addresses the issue of coding rare diseases in health information systems.

Organisation of Rare Disease Health and Social Care (Centres of Expertise; ERNs; Integrated Care and Social Support)

Centres of Expertise

There is an official policy for the designation of Centres of Expertise (CEs) for rare diseases in Cyprus. This is a national policy which has been fully implemented (in the sense that Centres have actually been designated) and the criteria used are in accordance with the EUCERD criteria.

2 Centres of Expertise officially comply with the National Policy (equating to 1.71 Centres per million inhabitants). Both of these CEs adhere to the EUCERD criteria. The Centres reportedly ensure a holistic approach to care, i.e. they liaise with social services.

(In addition to the Officially-designated CEs -i.e. Archbishop Makarios III Hospital and the Cyprus Institute of Neurology and Genetics- expertise in rare diseases is available via the Cyprus Thalassemia Centre, the Centre for the Study of Haematological Malignancies, and the Bank of Cyprus Oncology Centre, as well as through a number of specialised clinics and departments at the Nicosia General Hospital and other hospitals (see the 2014 SoA Report)).

European Reference Networks (ERNs)

Cyprus has a formal process in place for the endorsement of Health Care Providers (HCPs) who wish to participate as either members or coordinators of a European Reference Network. At present there are 2 HCPs participating as full members of 2 ERNs in Cyprus (see https://ec.europa.eu/health/ern_en)

Member HCP	European Reference Network (ERNs)
Archbishop Makarios III Hospital	ERN EuroBloodNet
Cyprus Institute of Neurology and Genetics	ERN ITHACA

Integration of Rare Diseases into Social Policies and Services

Specific actions exist to enable people with rare diseases to gain meaningful access to *general* social/disability programmes in Cyprus. A multidisciplinary/interministerial working group for rare diseases, which includes

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representatives from the Ministry for Social Affairs/Services, acts as a mechanism to facilitate multidisciplinary, holistic, continuous care provision. There are currently several relevant proposals for actions within the National Strategic Plan to ensure further social integration for people affected by rare diseases.

Rare Disease Registration

There are neither national nor regional-level registries for rare diseases in Cyprus, and there are no specific laws or regulations in place to govern rare disease registration. However, a variety of disease-specific registries exist: clinician-led registries are dedicated to rare genetic syndromes and diseases, rare neurological conditions, and thalassemia syndromes; researcher-led registries exist for rare anaemias; and some registries are patient-led, such as the congenital cardiac anomaly registry.

Genetic Testing and Neonatal Screening

Currently there is no national legislation to govern genetic testing in Cyprus; however, testing is governed by the Bioethics Convention. The Orphanet Cyprus site maintains a list of reference laboratories providing genetic testing, and this is updated annually. **According to the information in the Orphanet database, at the end of 2016 Cypriot laboratories offered tests for 70 genes and 96 rare diseases** (excluding panels of genes).

There are no specific provisions for the reimbursement of testing costs, although many tests are covered by the Public Health sector. There is a process in place to enable genetic testing abroad via the 'Cross-Border Healthcare Directive', following an evaluation by the appropriate medical committee within the Ministry of Health.

A **Neonatal screening programme** exists as part of the public health programme in Cyprus: screening is offered free-of-charge to all newborns, for 2 rare diseases at present (Congenital Hypothyroidism and Phenylketonuria). In addition, all newborns are screened for congenital Hearing Deficits.

In June 2013 a National Committee for Neonatal Screening was appointed by the Ministry of Health, with the task of revisiting the current practices, discussing the possible need for expansion of the current policy and providing a plan of action in the field. The committee was multidisciplinary and consisted of experts as well as stakeholder representatives. The advisory committee prepared a report summarising its findings and recommending the establishment of a National Committee. This report was accepted and endorsed by the Ministry of Health.

Clinical Practice Guidelines and Training Activities

Clinical Practice Guidelines (CPGs)

Cyprus has not produced any CPGs for rare disease at the national level. There is currently no national policy in place for the development or implementation of CPGs; however, there *is* a policy for the adoption of CPGs in Cyprus.

Training and Education

There are several initiatives in place in Cyprus for rare disease-related training and education. These include:

- Tutoring and lectures aimed at medical students, professionals and the general public;

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- The organisation of rare disease-related workshops and conferences;
- TV and Radio programmes designed to increase awareness of rare diseases

In recent years, the National Committee for Rare Diseases contacted all University schools offering education in fields relevant to rare disorders (such as Medicine, Nursing, Physiotherapy, Speech therapy, Biology etc.), requesting the incorporation of rare disorders into their teaching curriculum. Collaboration with National Medical Associations and scientific societies has also been established, to support education and training.

Information Resources for Rare Diseases

Orphanet Activities

Cyprus has an operational national Orphanet team, hosted by the Clinical Genetics Clinic at the Archbishop Makarios III Hospital (a hospital within the Public Health Service). The institution is a partner in RD-ACTION; however, there is no specific funding via the National Strategy (or otherwise) to support the activities of the Orphanet team. The Orphanet Cyprus team produces certain documents in the Cypriot national language, and has managed to collect data regarding patient organisations, healthcare professionals, clinical laboratories, genetic tests, and research activities, whilst also providing lectures on the activities of the Orphanet team and on rare diseases in general.

Official/Unofficial Information Centres

Both specialised clinics and patient organisations are currently fulfilling the role of *unofficial* information centres for rare disease in Cyprus.

National Helplines

There is no helpline in place dedicated to rare diseases in Cyprus; however, there is strong interest in setting-up such a helpline in future, both from the National Committee for Rare Diseases and from patient associations.

Rare Disease Research Activities

Specific rare disease research projects are funded from the *general* research budget in Cyprus: there has been no policy decision to allocate a portion of the national research budget specifically to rare disease-related research. As of late 2016, various proposals were being evaluated, concerning social or socio-economic-focussed research into rare diseases (including one MCS proposal).
At present, Cyprus participates to neither E-Rare nor IRDiRC; however, there is substantial interest in participating to IRDiRC and it is hoped that future participation will be possible.

National Alliance of Patient Organisations and Patient Representation

In June 2010, the Cyprus Alliance for Rare Disorders (CARD) 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 as follows:

- to lend support to the national rare disease programme;
- to support the efforts of patients to improve prevention and medical treatment of rare diseases, as well as social and other services related to rare diseases, in order to improve the health and quality of life of patients;
- to provide support and continuous education to patients and their families concerning the latest developments in medicine and research;

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- to raise awareness of rare diseases in Cypriot society.

The main achievements of CARD since 2014 include patient empowerment, raising awareness of rare diseases, playing a role in healthcare reform projects, supporting patients, and meaningful engagement in the CNSPRD.

Orphan Medicinal Products (OMPs)

The precise number of OMPs with an EU Marketing Authorisation available in Cyprus is unknown. OMPs are requested for patients via the authorities. There are measures in place to facilitate access to OMPs for patients; notably, a named-patient supply is possible (meaning doctors can request a supply of a medicine directly from the manufacturer, for use by a patient under their immediate responsibility).

Rare Disease Day

Rare Disease Day was celebrated in 2014, 2015 and 2016, through various activities such as open day discussions, festivals, video productions and school sports awareness activities. Lectures within conferences and public dialogue events have also taken place.

Main achievements since 2014:

Notable achievements in the last few years include the following:

- Interaction with senior-ranking political figures -including the President of the Republic of Cyprus, Members of Parliament, and other officials- to put rare diseases onto the political agenda.
- Increased Public and Professional awareness of rare diseases.

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