



**CYPRUS**

**EUROPLAN NATIONAL CONFERENCE**

*in the framework of the EU Joint Action RD-ACTION*

***Nicosia, 09 March 2018***

**FINAL REPORT**



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# FOREWORD

The EUROPLAN national conferences or workshops are organised in many European countries as part of a coordinated and joint European effort to foster the development of comprehensive National Plans or Strategies for Rare Diseases addressing the unmet needs of patients living with a rare disease in Europe.

These National Plans and Strategies are intended to implement concrete national measures in key areas from research to codification of rare diseases, diagnosis, care and treatments as well as adapted social services for rare disease patients while integrating EU policies.

The EUROPLAN national conferences/ workshops are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS–Rare Diseases Europe. **Rare Disease National Alliances and Patient Organisations have a crucial role to shape the national policies for rare diseases.**

The strength of EUROPLAN national conference/ workshop lies in its shared philosophy and format:

- **Patient-led:** National Alliances are in the best position to address patients' needs;
- **Multi-stakeholders:** National Alliances ensure to invite all stakeholders involved for a broad debate;
- **Integrating both the national and European approach to rare disease policy;**
- **Being part of an overarching European action** (project or Joint Action) that provides the legitimacy and the framework for the organisation of EUROPLAN national conferences/workshops;
- **Helping national authorities adhere to the obligations stemming from the Council Recommendation of 8 June 2009 on an action in the field of rare diseases.**

Since 2008, National Alliances and EURORDIS have been involved in promoting the adoption and implementation of National Plans and Strategies for rare diseases. Altogether, 41 EUROPLAN national conferences took place in the framework of the first EUROPLAN project (2008-2011) and the EU Joint Action of the European Committee of Experts on Rare Diseases – EUCERD - (2012-2015).

Within RD-ACTION (2015-2018), the second EU Joint Action for rare diseases, National Alliances and EURORDIS continue to get involved in a coordinated European effort to advocate for and promote integrated national policy measures that have an impact on the lives of people living with rare diseases.

The EUROPLAN national conferences or workshops taking place within RD-ACTION focus on specific themes identified by the National Alliances as the most pressing priorities to tackle with national authorities. These thematic priorities are addressed in sessions where all the stakeholders discuss relevant measures to be taken or ways to sustain the full implementation of already approved measures.

Each National Alliance prepares a final report on the national workshop, based on a common format such as the one that follows.

## GENERAL INFORMATION

<b>Country</b>	Cyprus
<b>National Alliance (Organiser)</b>	Cyprus Alliance for Rare Disorders
<b>Date &amp; place of the national workshop/conference</b>	09 March 2018, Semeli Hotel, Nicosia
<b>Website</b>	<a href="http://www.raredisorderscyprus.com">www.raredisorderscyprus.com</a>
<b>Members of the Steering Committee</b>	See attached programme ( <i>under the title: 'Contributors'</i> )
<b>List of Themes addressed</b>	<ol style="list-style-type: none"><li>1. Prevention and early diagnosis</li><li>2. Diagnosis and Treatment</li><li>3. Supportive Care and Social Welfare</li><li>4. Registries</li><li>5. Research</li></ol>
<b>Annexes :</b>	<ol style="list-style-type: none"><li>I. Programme in English</li><li>II. List of Participants (by stakeholders' categories)</li></ol>

## FINAL REPORT

### I. Introduction/ Plenary session

The Cyprus National EUROPLAN Conference was organized in the framework of the EU Joint Action RD-ACTION. The Conference was held on 09 March 2018, at Semeli Hotel in Nicosia. It was attended by 77 persons, across a broad multi-stakeholder spectrum including the Ministry of Health, Ministry of Labour & Social Insurance, Health Insurance Organisation, members of the European Parliament, representatives of parliamentary parties, Office of the European Parliament in Cyprus, Commissioner for NGOs, Cyprus Medical Association, the major private and public universities, directors, medical specialists and other healthcare professionals from the public hospitals and private sector, pharmaceutical companies, learned societies, patients/parents associations, and specialised healthcare centres (e.g. Cyprus Institute of Neurology & Genetics, Thalassaemia Centres, Karaïskakio Foundation - Cyprus Bone Marrow Donor Registry, Center for Preventive Paediatrics etc). In addition the Conference was attended by Mr Yann Le Cam, EURORDIS CEO.

### Country Status

Cyprus is an island state with a relatively small population size, approx. 854,800 (2016 Census). As such medical expertise on rare disorders (hereafter RD) is concentrated in a few individuals and

widely dispersed across health-providing centres, some being more specialised in some areas than others.

A National Strategy for Rare Disorders (hereafter NS) was adopted by the Council of Ministers in November 2012, focusing on the following pillars:

1. Prevention and early diagnosis
2. Diagnosis and Treatment
3. Supportive Care and Social Welfare
4. Registries
5. Research

It is noted that the NS included an extensive description of the current (in 2012) status quo in each of the pillars, as well as suggested actions for improvement of these. However, no time or financial frameworks were set for the implementation of any actions.

A multidisciplinary National Committee for Rare Disorders (hereafter NCRD) was set up for a 5 year term (2013 – 17), comprised of medical specialists and health professionals working in various fields, a representative of the Department of Social Integration at the Ministry of Labour & Social Insurance and patient/parent association representatives. As outlined in the Terms of Reference provided to each participating individual, the Committee was mandated to :

- Advise the Minister of Health on the development of policies for RDs;
- Develop an Action Plan based on the NS, including methods of monitoring its implementation and;
- Ensure the cooperation and collaboration of all stakeholders in the promotion of this joint effort.

The mandate of the members of the NCRD expired in late 2017 and nominations for a new composition of the Committee were recommended to the Minister of Health and Council of Ministers for ratification. Delays due to the Presidential elections in January 2018 as well as other procedural challenges relating to the Committee's hierarchy pushed the appointment of the new Committee to March 2018.

The organisation of the National EUROPLAN Conference at the present time, provided an excellent opportunity to ascertain the actions implemented by the Committee since 2013 and to identify weaknesses and challenges. Indeed, the conclusions and suggested actions (see below) of the Conference will be submitted to the Chairperson of the new NCRD (i.e the Director-General of the Ministry of Health), and will provide the foundations of its work for its term.

Moreover, two other factors led the Cyprus Alliance for Rare Disorders (C.A.R.D.) to seize the opportunity at the present moment to organise this Conference. These are :

1. The official implication of the patient perspective in all issues promoted at the legislative level (relevant law of 2016)

2. The imminent implementation of the legislation for the creation of a National Health System, which will fundamentally change the scene of healthcare in Cyprus for all citizens but RD patients in particular.

### **National EUROPLAN Conference Themes**

A total of five (5) workshops, each on separate themes took place. These were selected to reflect the pillars of the NS, considering that an important objective of the Conference was to gauge the current situation of each pillar, compared to 2012 when the NS was adopted. In addition, each theme encompassed a number of sub-themes (as outlined below).

1. Theme/Workshop 1: Prevention and early diagnosis (Sub-themes: Community Awareness, Centres of Expertise)
2. Theme/Workshop 2: Diagnosis and Treatment (Sub-themes: Centres of Expertise, Access to Orphan Drugs, European Reference Networks, new National Healthcare System, Pricing & Reimbursement)
3. Theme/Workshop 3: Supportive Care and Social Welfare (Sub-themes: Social Policies, Rehabilitation, Respite Care)
4. Theme/Workshop 4: Registries (Sub-themes: e-health, codification)
5. Theme/Workshop 5: Research (Sub-themes: Clinical Trial Design & Participatio, Pricing & Reimbursement)

### **Aims of the National EUROPLAN Conference**

In this context, the organisation of a National EUROPLAN Conference in Cyprus aimed to:

- Revisit the policy and public service issues concerning RDs since the NS had been adopted in 2012;
- Ascertain the activities of the NCRD in the implementation of the NS and identify challenges ;
- Inform a wide range of stakeholders on the status quo regarding RDs;
- Highlight the integration of RD in the National Health System, including the e-health record, registries, research, drugs etc;
- Focus on specific areas that require significant developments in Cyprus, such as development of Indicators, Social Services, Centres of Expertise & European Reference Networks.

The Conference reflected these aims both in the plenary session and in the workshop discussions.

The Workshops were extremely productive and contained lively discussions in all areas. Following the Opening Session, there were 5 consecutive Workshops to cover the topics/themes mentioned on page 3 of this report. All participants took part in all Workshops. Experts in each field and patient advocates acted as Coordinators for each Workshop. C.A.R.D. Staff and Volunteers acted as Rapporteurs.

## **II. Themes**

### **THEME 1: PREVENTION AND EARLY DIAGNOSIS**

#### **WORKSHOP FORMAT**

The Workshop was initiated by presentations by the Coordinators (Dr Michael Angastiniotis and Dr Adonis Ioannides) on Prevention and Early Detection, providing background content for the discussions which followed.

The discussion, facilitated by the Coordinators, was completed with the statements of two patient advocates (Mrs Katerina Margaroni and Mr Marios Vakanas) each providing their perspective on the Workshop theme.

#### **INTRODUCTION**

Prevention constitutes a cornerstone in the planning of public health services . It concerns the prevention of the expression of the disease itself, or its complications. Public health in Cyprus has shown improvements during the last few years and healthcare provided is at adequate levels.

All prevention programs through the years have focused on selected common disorders, or various inherited diseases that constituted a major issue for public health and required immediate actions, such as thalassaemia. Such programs require further strengthening, encompassing RD and development into national prevention programs.

Since 2010 (when the NS was prepared) primary prevention has been strengthened through the following actions:

- I. Dissemination of information by medical and professional associations, research foundations and the Ministry of Health, through the publication of special reports and targeted campaigns for the distribution of leaflets, particularly during the designated internationally observed Days of specific diseases
- II. Constant references, briefings and various programs or shows in the mass media
- III. Activities of patient groups, thus strengthening the patients' voice and promoting their involvement at decision making centres
- IV. The significant increase in access to information, especially online, in addition to printed material
- V. The financial support of special education and awareness campaigns by national competent authorities or academic institutions at the national or European level
- VI. The promotion, information and education of numerous resolutions regulations and recommendations regarding RD, as required by the European institutions
- VII. Activities relating to the education of the general public and healthcare professionals for the inclusion of prevention programs in national strategies concerning specific diseases and/or disability as per the recommendations of the United Nations and the World Health Organisation.

### **Existing programs at the national level**

Provision of Health Education classes in schools with the participation of the School Health Service of the Ministry of Health.

Some programs exist both in the public and private sector but without any national coordination or scientific monitoring of results which is crucial in order to assess the necessity for modifications or strengthening of the programs.

### **Preventative medical examinations / Population screening programs**

Even though a long term national practice has not been implemented, the medical and scientific advances of recent years have enabled the upgrading of several areas. Despite the fact that there is still a huge need for research for the thousands of RD that remain without diagnosis and subsequently without treatment, any research conducted must be utilized to the utmost for the benefit of citizens and national prevention.

#### **1. Preventative Medical Examinations**

- Application and adoption of good clinical practice during the 7 defined and regular paediatric examinations required during the post-natal period and in childhood, both in the public and private sector
- Further development of several paediatric specialisations, especially at Arch. Makarios III Hospital (hereafter NAM III), which is the Reference Centre for paediatric cases (rare and non-rare) with referrals by paediatricians from across Cyprus
- Creation of health education programs in schools with a preventive paediatric examination especially for detecting diseases relating to cardiology, ophthalmology, auditory, skeletal deformities etc.

#### **2. Population screening programs**

- Counselling services are generally provided by the public sector, particularly by the Genetics Clinic of NAM III and by the Cyprus Institute of Neurology and Genetics (hereafter CING), whilst there appears to be a limitation in the number of genetic counsellors working throughout Cyprus either in the public or private sector
- The first and only national program that involves counselling, screening and prenatal diagnosis is that of thalassaemia, as well as testing for chromosomal or other congenital abnormalities which are provided to all pregnant women during their first trimester of pregnancy.

### **Newborn screening programs**

The National program that is available for all newborns all over Cyprus covers:

- Phenylketonuria (PKU)
- Congenital Hypothyroidism
- Auditory check –up

This test has been conducted since 1990's from the Preventive Paediatrics Centre. In addition, the Biochemical Lab at CING has been established for the support of timely and accurate screening of

RD , particularly for metabolic disorders. This is the Reference Laboratory with a holistic approach providing services nationally.

Periodically pilot RD screening programs have been implemented which target populations in specific geographical locations. Examples include:

- I. Duchenne Muscular Dystrophy
- II. Friedrich`s Ataxia
- III. TTR – FAP
- IV. Sandhoff`s disease
- V. GM 1
- VI. Cystic Fibrosis
- VII. Huntington`s disease

The high quality of clinical and research laboratories was highlighted during the discussion, despite the lack of quality assurance or official accreditation. Furthermore, the dramatic scientific advances in the area of diagnosis which has occurred over the last decade was also underlined. This progress has further enabled the timely and accurate diagnosis of many genetic and other RD today.

The following comparative table was presented during the discussion showing Neonatal Screening Examinations across a number of countries.

Country	Metabolic	Endocrinological	Cystic Fibrosis	Hemoglobinopathies	Auditory
Cyprus (2)	1 disorder	hypothyroidism			yes
U.K. (10)	7 disorders	hypothyroidism	Yes	Yes	yes
France (5)	1 disorder	hypothyroidism adrenal hyperplasia	Yes	Yes	Yes
Greece (4)	2 disorders	hypothyroidism adrenal hyperplasia		Hemolytic anemia	
Malta (2)		hypothyroidism		Yes	
Luxemburg (5)	5 disorder	hypothyroidism adrenal hyperplasia		Yes	
Latvia (1)	1 disorder	hypothyroidism			
Austria (48)	45 disorders	hypothyroidism adrenal hyperplasia	Yes		Yes

Source: Therrell et al Current status of neonatal screening worldwide. Science Direct 2015



The need for expanding the neonatal screening program was highlighted, so that it includes the diagnosis of RD . At the same time co-ordinated programs that can cover the general population and also be financially viable must be created. An extensive reference to the work of the Committee for the Investigation of Programs of Pre-natal Screening was also discussed. The Committee investigated this subject for 3 years but none of its suggestions have been adopted yet. Their report was recently submitted to the Ministry of Health, with the contribution of Preventive Paediatrics Centre and projected budgetary requirements. The response of the national health authorities is pending.

## **CONCLUSION**

1. There is a lack of a national focal point for the provision of information on prevention programs and raising awareness
2. There is an urgent need for a thorough identification of all information programs currently in existence in Cyprus provided by all Ministries and other relevant authorities and the development of a well coordinated strategy for the provision of information about RD to the general public
3. Creation of a sub-committee within the NCRD, and the assumption of a formal collaboration with the Cyprus Medical Association, Professional Medical Associations and Academic Institutions for the development of a specific program for the continuous education of healthcare professionals
4. Integration of information and education about RD in Medical Schools.
5. Re-evaluation of the suggestions of the Committee for the Investigation of Programs of Pre-natal Screening
6. Continuation of the efforts to define National Reference Centres and their networking with their respective European Reference Networks to facilitate the timely detection of RD where difficulties are encountered is necessary. Timely and accurate diagnosis will always remain an important issue in small populations like Cyprus, which can be dealt with through official, mainly European, networks
7. Promotion of programs for the training and certification of genetic counsellors in order to strengthen the counselling services of the Paediatric and Gynaecological Departments, CING and all other relevant departments of the public and private healthcare service providers
8. Families with severe genetic or inherited disorders should be financially supported via specific legislation for pre-natal diagnosis and in vitro fertilization
9. Counselling should be provided to couples before conception, as part of primary health services including the assessment of possible outcomes, wherever necessary.

## **SUGGESTED ACTIONS**

1. Creation of a sub-committee of the NCRD with the objective to identify the specialised centres and laboratories, either of the public or private sector, including the suggestions of the Committee for the Investigation of Programs of Pre-natal Screening. Thereafter the NCRD will submit these to the Minister of Health.
2. Conduct a further discussion with the Health Insurance Organisation (HIO), considering that prevention and screening programs will be included in the primary health services of the new National Health System, and as such the HIO will be responsible for their implementation and allocation of budget.
3. The registration of all RD in Cyprus is a necessity in order for the planning and services development to be effective and in order to save resources.
4. The NCRD to undertake the development of a strategy for the continuous information flow to the general public regarding RD .

## **THEME 2 – DIAGNOSIS & TREATMENT**

### **WORKSHOP FORMAT**

The Workshop was initiated by presentations by the Coordinators (Dr Georgios A. Tanteles, Dr Panayiota Protopapa and Mr Andreas Papaconstantinou) on Diagnosis and Treatment, providing background content for the discussions which followed.

The discussion, facilitated by the Coordinators, was completed with the statements of two patient advocates (Mr Demetris Genethlis and Mrs Maria Ioannou) each providing their perspective on the Workshop theme.

### **INTRODUCTION**

Timely and accurate diagnosis of any RD is difficult not only on account of the vast number of RD which exist across multiple medical disciplines, thus requiring a multitude of approaches for their management, but also due to the fact that they are not appropriately documented. In Cyprus, as in other countries, several diagnostic services and treatments are offered by various medical centres.

In addition to the Paediatric Department of NAM III Hospital, that has been evolved into a Reference Centre, the General Hospitals across Cyprus and particularly the Nicosia General Hospital offers various diagnostic services. There is no cohesive infrastructure for specific departments/ clinics to deal with RD, therefore the diagnostic approach, investigation and confirmation of diagnosis are dealt with in the context of the relevant clinics (e.g. cardiology, gastroenterology etc) which also deal with more common diseases. In addition many other clinics, hospitals and clinical laboratories offer related services.

Moreover there are some clinics that address issues relating to the diagnosis and treatment of adult RD patients. For example :

- Inherited cardiovascular Diseases Study Unit – Located at the General Hospitals of Nicosia and Larnaca. The Unit handles disorders such as cardiomyopathies, sudden cardiac death etc.
- Haemorrhagic Diseases Clinic. Located in Nicosia General Hospital within the Haematology Department.
- Thalassaemia Centre – Located in NAM III Hospital and the General Hospitals of Larnaca, Limassol, Paphos and Famagusta. These are affiliated with the Thalassaemia Molecular Laboratory at CING.
- Oncology Department – Located in Nicosia General Hospital and Bank of Cyprus Oncology Centre. In addition to diagnosis and treatment of patients with various forms of cancer, rare malignancies and rare complications are diagnosed and treated at these Centres. Recently a new privately owned Oncology Centre has been developed in Limassol, that provides high quality services for patients with various forms of cancers. Negotiations with the Ministry of Health are ongoing so that the Centre can provide contracted services.
- CING – Founded in 1990 in Nicosia, and it is an NGO that is jointly supported by government funding and various research programs and donations. Its main objective is to develop and offer high quality medical and laboratory services and to develop innovative research and training. For the accomplishment of CING's goals various clinics have been established. The Genetics Clinic (that works in collaborations with NAM III) handles patients with various rare neurological issues. CING's laboratory departments offer diagnostic screening on a wide range of RD such as metabolic disorders, chromosomal anomalies etc.
- Preventive Paediatrics Centre – It is located in Limassol and it offers a Preventive Natal Screening Program that tests newborns for Phenylketonuria (PKU) and Hypothyroidism. Blood samples are collected from Paediatricians from all over Cyprus and sent to the Centre's Biochemical Laboratory for testing. If indications of metabolic disorders occur, the Paediatrician refers the newborn for further investigation. Regular Biochemical checkups of a disorder's progress and treatment effectiveness are also provided by the Centre. Other services are provided such as mental deficiency evaluation program and all children are monitored by medical specialists (in collaboration with NAM III).
- Karaiskakio Foundation – It is located in Nicosia and through the work of Molecular Haematology and Immunogenetics Laboratory offers support in the investigation of patients with Haematological diseases. The new Haematological Malignancies Study Centre contains a specialised diagnostic and research unit for rare haematological disorders.

### **Diagnosis & Treatment services**

In Cyprus the diagnosis and treatment of RD is mainly provided by the Public Hospitals, Cyprus Institute of Neurology and Genetics, Preventive Paediatrics Centre, Bank of Cyprus Oncology Centre, Karaiskakio Foundation and the Molecular and Medical Genetics Laboratories of the University of Cyprus.

In the private sector there is a limited range of services provided due to the fact that isolated medical specialties and diagnostic centres are involved in the diagnosis of RD.

A distinctive reference about the Paediatric Clinic of NAM III was made regarding the hospital's growth and development in the various medical sub – specialties such as genetics, endocrinology, nephrology, cardiology, oncology, neurology, pneumonology, gastroenterology and hepatology. One of the most important services that the Genetics Clinic provides is Genetic Counselling. The

referrals for genetic evaluation and genetic counselling have significantly increased and so the clinic and its services must be strengthened.

A vast range of diagnostic services, mainly for adults are provided by public hospitals. The diagnostic approach, investigation and confirmation of diagnosis are set within the framework of the several clinics daily function. Considering the lack of awareness and education regarding RD's and the absence of structured specialised clinics in RD's, timely diagnosis, monitoring and treatment is difficult. The challenges of individual RD patients is enormous - diagnosis can take from several months to years and subsequently receipt of appropriate treatment is delayed with severe, and sometimes irreversible, consequences in some cases.

Perhaps the most complete unit that offers a holistic treatment, including prevention, diagnosis and clinical management is the Thalassaemia Centre, even though the management of oncology patients on BOC Oncology Centre, and CING's neuropathy clinic are in the process of completing the design of their holistic programs.

The implementation of laboratory techniques is tied to these development, and will provide diagnoses with unlimited potential and whenever possible assessment of the clinical outcome of a disorder. Even though there is a vast spectrum of laboratory interventions of this kind, especially at CING and Karaiskakio Foundation, beyond the purely research area, they haven't been explored or integrated in the national guidelines. In addition a coordinated process for sending samples to reference laboratories with official collaborations between the Ministry of Health and the private sector is not available.

The necessity to promote clinical genetics and genetic counselling was emphasised. The 3 genetics clinics that operate in Cyprus collaborate, exchanging knowledge and experience and the opinions of specialists from Reference Centres abroad are requested wherever necessary. To date, no official collaborations with centres from abroad exist, except for paediatric hepatological / gastenterological diseases. This makes treatment difficult considering the limited expertise of medical specialists in Cyprus due to the small number of cases that each will encounter.

There is of course a large number of patients with undiagnosed and unknown disorders for which research must be strengthened and academic – research centres and pharmaceutical industries must be mobilised for the search of innovative treatments and drugs for effective treatment.

The Health Insurance Organisations (HIO) representative confirmed that matters of treatment for chronic and rare disease patients will be included at the National Health Plan and the implementation of electronic patient health records will be a huge step towards the appropriate management of RD. A detailed documentation of the needs of rare disease patients, regarding diagnosis, consumables, medical equipment and drugs will enable their inclusion in the National Health Plan.

At the same time this documentation will help the identification of weaknesses and needs in order to promote required infrastructures or policies.

## **Access to drugs**

Orphan and innovative drugs are vital for RD patients due to the chronic and often degenerative nature of RDs. Timely access to these drugs is essential. Unfortunately the cost of the drugs in many cases is very high and bureaucracy, needed for their approval (either on an individual basis or for inclusion in the public prescription drugs list) extends the waiting time especially when it comes to new and innovative drugs. For that matter to be addressed, new and effective mechanisms, within the National Health Plan, must be developed for the timely inclusion of new drugs to the public prescription drugs list as well as the appropriate pricing of those drugs.

Within the National Health Plan a Consulting Committee will be appointed by the Council of Ministers in order to consult the HIOs Board of Directors in matters of :

- The inclusion or the removal of drugs from the National Health Plan`s catalogue of approved drugs
- The granting of expensive pharmaceutical products or products that might have doubtful results.

After consulting with the committee, the Board of Directors of HIO will be responsible for the final decision. Applications for the inclusion of drugs on the catalogue must be submitted by the marketing authorisation holder or distributor, or in exceptional cases, and under specific prerequisites, treating physicians. The HIO is obligated to reach a decision within 90 days, thus ensuring timely access to the drug(s). In addition, the Council of Ministers will appoint a Review Committee, that will consult with the HIO in cases of challenges to the Board's decisions concerning drug approvals. The HIO Board of Directors will also appoint a Drug Reimbursement Committee, that will consult the HIO in matters of drug reimbursement.

Patient representatives were reassured that all pharmaceutical products that are included today in the public prescription drugs list but also those currently available in the Cyprus market will be included in the National Health System. In addition to that patients can choose to use a different medicine than the one provided, but HIO will only be able to reimburse at the level of the lowest drug price.

Other categories of products that are not considered to be pharmaceutical but are used for treatment of patients need to be listed and brought in attention of the HIO for investigation as to whether or not they can be listed in the catalogue of free pharmaceutical products that are provided to patients.

The huge cost of orphan and innovative drugs will be a challenge for National Health System budget which is based on the thinking to keep expenses for health at current levels. The expenditure of 7% of the GDP of Cyprus to healthcare is small in comparison to what other countries spend on health, but the success of a health system does not just depend on the amount spent. For example the USA spends 17% of GDP on health but has 25 million uninsured citizens. In the meantime Estonia spends just 7% of their GDP but has a quite successful system. In order for the system to work, all needs must be taken into consideration and the available budget must be used wisely.

There is no assurance that a separate fund for orphan drugs will be available within the National Health System even though there are several examples that can be followed such as:

- In Italy if a drug is proved to be successful the state is obligated by the law to provide it to patients.
- In the UK NICE sets an upper financial limit to what can be covered and cannot surpass it
- In the UK different budgets are available for drugs that are not included in the NHS. This provides flexibility, which is very important in the case of RDs.

In every case the National Health System must protect each person against the unbearable cost of treatment, especially in the case of chronic and RDs, whilst at the same time safeguard vulnerable groups for diagnosis and treatment.

One of the biggest challenges in area of health is undoubtedly the constant need to increase the budget for health. A special provision exists within the National Health System which states that the budget can be increased from time to time and always in consideration of the country's economy.

All patient groups have a role and a strong voice through their participation in the discussions, consultations and the decision making.

## CONCLUSION

1. The knowledge and the education of health professionals regarding RD diagnosis and treatment is limited.
2. There is a lack of reference points for RD's, both in the public and the private sector, especially for adult RD patients.
3. The Genetics Clinic of NAM III is the Reference Centre for all paediatric clinics, both in public and private sector. The hospital is understaffed, the architectural infrastructure is limited and hence the quality of treatment received by the children is jeopardized. The lack of coordination of multi-disciplinary groups for various RDs results in the everyday suffering of the patients. The public sector must collaborate with private sector until National Health System is established for the alleviation of these issues. Furthermore, where the possibility of collaborating with an expert in Cyprus is not possible, then collaborations with Reference Centres abroad should be pursued.
4. Even though great improvement has been achieved in matters of pharmaceutical products, many patients still deal with the following:
  - Lack of clinical guidelines due to the limited knowledge by healthcare professionals and lack of data
  - Expensive drugs
  - Drugs are not included in protocols (this can be due to the bureaucratic delay in updating existing international guidelines, as new evidence is included)
  - Drugs are not included in the list of approved drugs
  - Access to clinical trials
5. There are still many things to be resolved, such as:
  - Inclusion of the management of RDs in the National Health System

- The strengthening of existing Reference Centres and the designation of new Reference Centres and their networking with European Reference Networks, wherever possible
- Establishment of official specific agreements with other Reference Centres for RDs that fall into the European Reference Network in areas where Cyprus does not have enough expertise

6. Cross Border Healthcare has not been properly implemented yet

## **SUGGESTED ACTIONS**

1. A National Registry for RDs must be implemented immediately in order to better understand the demographics of the various different RDs, their management, the centres and divisions with expertise, so that focal points on a National Level for, the benefit of patients, can be appointed.
2. Provide continuous education opportunities for healthcare professionals.
3. Strengthen and modernise the Genetics Clinic of NAM III, in order for it to form the information, referral and holistic approach centre for newborn genetic RDs.
4. Network with Reference Centres abroad for areas where there is a lack of expertise.
5. Promotion of various departments and clinics as hubs or Reference Centres.
6. Formation of a scientific committee for the preparation of medical guidelines based on European guidelines on RDs.
7. Creation of new and update old guidelines according to the latest scientific developments in the area of pharmaceutical products.
8. Active involvement of NCRD in the tasks of HIO, Organisation of Public Health Services and other.
9. Expedite the implementation of the legislations regarding rehabilitation, National Drug Organisation, University Clinics etc and create a network of services for the holistic treatment of RDs.
10. Implementation of National Health System according to the timeframes that have been set.
11. Establishment of a national regulatory authority to deal with the pricing of new innovative drugs, along the lines of the European Medicines Agency.
12. Promotion of mechanisms to ensure the smooth transition of patients from paediatric clinics to adult clinics once they have completed adolescence. In this context, further training of adult specialists should be pursued.

### **THEME 3 – SUPPORTIVE CARE, SOCIAL WELFARE & INCLUSION**

#### **WORKSHOP FORMAT**

The Workshop was initiated by presentations by the Coordinators (Dr Myrto Azina, Dr Androulla Eleftheriou and Mrs Marina Pavlou) on Supportive Care, Social Welfare and Integration, providing background content for the discussions which followed.

The discussion, facilitated by the Coordinators, was completed with the statements of two patient advocates (Mrs Anna Zannetou and Mrs Maria Kyriacou) each providing their perspective on the Workshop theme.

#### **INTRODUCTION**

The vast majority of RDs require specialised and continuous supportive care. RDs are their greatest majority heterogeneous, complex, progressive and in most cases degenerative diseases, thus resulting in delay in appropriate diagnosis and provision of treatment in a timely fashion. Subsequently, supportive care either for the alleviation of psychosomatic symptoms or management of psycho-social issues both of the patient and their family is a necessity. In fact, all services need to be adequately staffed and the providers to have professional expertise in dealing with complex RDs. In countries with small populations, however, like Cyprus, the accumulation of knowledge on all fields is impossible.

In Cyprus services such as physical therapy, occupational therapy, speech therapy, nutrition and dietetics are offered by the public and private sector and also from various NGOs.

The shortage of staff identified in the public sector as well as the lack of expertise in many areas, has serious consequences on the daily life of patients. In addition to that, if a patient receives such services from the private sector, which may have a greater expertise, the out of pocket expenses are severe and often unbearable.

Provision of home nursing services is provided by the public sector community nursing services for specific geographic locations. However, there are plans to extend this service to cover patients all over Cyprus.

The Mental Health Services via the regional Psychiatric Departments, General Hospitals, NAM III Hospital and several local health centres provide services for patients with mental health issues with the assistance of psychologists and trained nurses.

Furthermore, the Pain Management Centre at the Nicosia General Hospital and the Bank of Cyprus Oncology Centre, offer palliative care to patients. Likewise, the 'Arodafnousa' Centre offers similar services, to inpatients admitted.

The School Health Services constitutes the connecting link between families and children with the Ministries of Health and, Education and Culture. Unfortunately, the identification of specific issues relating to speech, hearing, vision, mobility, mental capability etc that RD patients face is significantly limited.



### **Available services**

The difficulty in the provision of supportive care for patients with RDs and their documentation lies in their heterogeneity and rareness. Thus the services offered by various public or private sector departments as well as NGOs have a huge role to play.

The biggest gap is identified in the field of rehabilitation, given the fact that centres and services are limited. The existing centres are Arodafnousa, Melathron Centre, the Paraplegic Department of the Nicosia General Hospital and various institutions such as Eleousa and Anemoni. Meanwhile some nursing homes and public hospitals have been converted to recovery centres without however any strengthening or upgrading to their infrastructure. Coordinated supportive care services are provided in a semi-coordinated fashion but with great inadequacies in some cases.

The importance of utilization of the whole spectrum of rehabilitation services for the appropriate and holistic treatment of almost all RD has only recently been acknowledged and thus the appropriate attention in building services and infrastructures has previously not been provided. Hence, many patient NGO, through funding from various Ministries and organisations, provide rehabilitation programs or services targeted mainly at children, adolescents and young adults.

#### **i. Ministry of Health**

The Ministry of Health issues the Hospital Identity Card that provides free hospitalisation for recipients of Social Welfare benefits and to people with annual income that is less than 37,754.93. For specific disorders the Hospital Identity Card Identity is provided regardless the patients income.

#### **ii. Ministry of Labour, Welfare and Social Insurance (MLSI)**

The MLSI provides the following monetary benefits (independent of income and age criteria):

- Severe Motor Disability Benefit Scheme
- Care Allowance Scheme for Paraplegic Persons
- Care Allowance Scheme for Quadriplegic Persons
- Mobility Benefit
- Special Allowance for Blind Persons
- Disability Pension

Other benefits include:

- Allowance to persons with disability for the provision of a car
- Scheme for the provision of financial assistance for the purchase of a wheelchair
- Scheme for wheelchair loans to persons with motor disability
- Financial assistance scheme for the provision of technical means, instruments and other aids
- Scheme for the management / provision of technical means, instruments and other aids
- Disability parking card scheme (Blue Badge)

Vocational Training:

- Scheme for the creation and operation of small units for self-employment purposes for persons with disabilities

- Scheme for the vocational training of persons with disabilities
- The employment of persons with disabilities in the wider public sector (up to 10%)
- Scheme providing incentives for employing individuals with disabilities
- Scheme providing incentives for employing individuals with chronic diseases

## **CONCLUSION**

In Cyprus, numerous rehabilitation and supportive care services are provided in a scattered way without central coordination. However, the main objective is the development of services that allow the reintegration of patients at the societal and occupational levels. There are some supportive care services that contribute towards improving the quality of life of patients, but there is a lack of services that cover the holistic management of RD patients.

## **SUGGESTED ACTIONS**

### **1. Social Welfare**

- i. Preventive measures for the limitation of genetic diseases, e.g. via the subsidisation of in vitro fertilization and expansion of the prenatal and preventive new-born screening.
- ii. Adoption of the European definition of Disability.
- iii. Raise awareness of the general public about RDs and diversity.
- iv. Creation of policies by the Ministry of Education to promote the inclusion of patients with RDs in all school activities, thus decreasing stigmatisation and isolation.
- v. State policies to be developed by taking into account the distinctive needs of persons with RDs.
- vi. Improve government social policy – the provision of financial benefits does not solve all problems.
- vii. Creation of services based on the needs of patients', e.g. physical therapy, speech therapy and occupational therapy as part of specialised programs.
- viii. Involvement of experts in an array of disciplines for strategic planning of services at the decision making centres.
- ix. Creation of rehabilitation centres for persons with RDs.
- x. Improvement of the current retirement policy for medical reasons of the MLSI.
- xi. Achievement of simplification of procedures, reduction of bureaucracy and improvement of the medical assessment process at the MLSI.

## **2. Social Inclusion**

- i. Resolution of the accessibility issues to public buildings for persons with disabilities.
- ii. Creation of social programs with the involvement of the local government.
- iii. Encouragement of patients to use the internet for creating networks with patients from other countries as a means to share knowledge and experiences, thus reducing marginalisation and isolation.
- iv. Strengthening and extension of targeted and specific policies for the promotion of employment of persons with RDs.
- v. Re-evaluation of the current criteria for employment of persons with disabilities in the wider public sector.
- vi. Provision of workplace facilitation (e.g. accessibility) for persons with RDs.
- vii. Establishment of flexible forms of employment for persons with RDs.
- viii. Legislative means for implementation of support mechanisms in the workplace for persons with disabilities (e.g. bullying, lack of respect etc).
- ix. Development of a legislation for the mandatory employment of an occupational physician for each workplace.
- x. Specific reference in the legislation concerning extended periods of sick leave for RD patients, in cases where they are required to be absent for receiving diagnosis or treatment abroad.
- xi. Facilitation of patient attendants (e.g. respite care services etc).

## **3. Supportive Care**

- i. Creation of programmes by MLSI in collaboration with the Ministry of Education and the Ministry of Health and the various Universities for the continuous and specialised training in matter of RDs to the following groups (students and professionals):
  - Social workers
  - Nurses and carers
  - Physical therapists
  - Occupational therapists
  - Speech therapists
  - Psychologists
  - Carers

- a. Supportive care is one of the most important issues that needs to be addressed, and thus emphasis needs to be given to the following:
- Carer profession needs to be officially established through the creation of a registration council that will provide official permits to those who fulfil the legal requirements to practice the profession.
  - Incentives need to be provided so that the profession becomes a career choice and not a necessity.
  - Carers need to be properly trained and knowledgeable regarding the disease, its peculiarities and the appropriate approach required for its care. The training of carers should be provided by academic institutions.
  - The training of carers should include:
    - Patient and family psychology
    - Effective communication channels with the patient
    - Problem solving methods
    - Coping mechanisms for avoidance of burn-out or identification
  - Carers need to be supervised on a regular basis aiming to safeguard the quality of services provided taking into account psychological debriefing, resolution of challenging work environments etc.
  - Carers supervision must be conducted by competent and accredited social workers.
  - Several services need to be defined including:
    - Home day care (e.g. self-sufficiency care , house cleaning and food preparation)
    - Transportation care: Further support for the main carer for transporting the patient from the wheelchair to the bed or the bathroom
    - Nursing: Provision of nursing services for the treatment of wounds, blood tests, gastrostomies etc. The home nursing services need to be geographically expanded.
    - Respite Care for Carers and Family members.
    - Social Support Care (e.g. transportation of the patient to the hospital or even for recreational purposes)

For the implementation of the program a dedicated unit needs to be created that will deal with only this issue. The implementation could be assigned to the Department for Social Inclusion of Persons with Disabilities given that this Department handles disability issues, in collaboration with the Ministry of Health. Also the Department for Social Inclusion of Persons with Disabilities must undertake the whole project of Supportive Care. The department that the unit is going to be assigned to, needs to be staffed by specialised and trained personnel in the intricacies of specific groups of diseases. This requires the continuous education of the personnel, including social workers.

The Ministry of Health in collaboration with the Department for Social Inclusion of Persons with Disabilities will be in charge for the implementation of community nursing.

A necessity for the operation of this program is the collaboration of all professionals and Reference Centres in order to achieve the multidisciplinary approach required.

The suggestion for application and supervision of the program by Social Workers is based on European and International Standards, where the Social Worker evaluates based on his knowledge the appropriate course of action for the patient and his/her family.

- ii. The creation of a Committee that will be composed by the Ministry of Health, the Ministry of Labour, Welfare and Social Insurance and the Ministry of Education and Culture. The Committee's objective will be the monitoring of all the Social Workers and to report to the General Directors of those Ministries.
- iii. Subsequently, the Ministry of Finance will have to approve funding for adequate staffing of all units. The proposal for the funding must be provided by the Committee of the 3 Ministries that will also have to list all needs regarding human resources and level of expertise.

#### **THEME 4 – REGISTRIES**

##### **WORKSHOP FORMAT**

The Workshop was initiated by presentations by the Coordinators (Dr Eleni Zamba Papanicolaou and Prof. Christos Schizas) on RD Registry, providing background content for the discussions which followed.

The discussion, facilitated by the Coordinators, was completed with the statements of two patient advocates (Mr Miltos Miltiadous and Mr Philippos Akamas) each providing their perspective on the Workshop theme.

##### **INTRODUCTION**

There exist a number of registries of various disorders maintained by doctors or clinics of the public or private sector and many of them contain RD cases. Some examples include:

- Genetic disorders registry at CING and NAM III Hospital
- Congenital Heart Defects registry at Nicosia General Hospital
- Research Centre for Molecular Medicine registry at the Medical School of the University of Cyprus
- Haematological diseases registry at Nicosia General Hospital
- Paediatric gastroenterology and hepatology registry at Engomi Medical Centre (part of NAM III Hospital)
- Viral hepatitis registry at Nicosia and Limassol General Hospitals

Some other registries are maintained by private centres and clinics, used by the department or the scientists that handle them. Nonetheless, these individual registries cannot form a sufficient basis

for complete epidemiological reports and for monitoring of the disorders. For those purposes the creation of a national registry is essential.

### **e-health**

The necessity of promotion for the electronic health records and the digitalisation of patients data was emphasised. This can contribute substantially to the coordination of the multidisciplinary approach that is needed for patients with RDs. The software that will be used by HIO for the needs of the National Health System will register all persons that have received treatment in the public sector, including information on diagnosis and treatment.

### **Codification**

Specific items relating to RDs must be included in the software such as coding mechanisms (ICD11 / Orpha code). This will enable the collection of epidemiological and clinical information about RDs on a national level. Thus the size, geographical orientation and other data can be assessed. Furthermore, it will allow the identification of specialised centres and departments, participation in clinical trials and research programs and contribute to the proper planning of preventative programs and other services.

Some examples of pilot registries were provided:

- Huntington`s disease: In Cyprus 19 families and 58 patients (only 38 are alive at the present point) have been recorded. The prevalence is estimated at 4.64 persons for 100,000 persons of the population, which is consistent with the prevalence numbers that occur in Western European populations. This can lead us to the safe conclusion that Huntington`s disease is European oriented. This research and documentation led to the conclusion that the highest prevalence is located in the Famagusta – Larnaca area.
- Amyotrophic Lateral Sclerosis (ALS) : 179 patients have been tracked down in the Greek – Cypriot population and only 7 of them had a family history of the disease. The prevalence is estimated at 7.9 persons per 100,000 of the population that is consistent with the prevalence numbers of the Western European populations. According to the study the average life expectancy of patients with ALS is 58.6 years and there is no specific geographical orientation for the disease in Cyprus.
- Friedrich`s Ataxia: The study revealed that there is a high prevalence of the disease in the province of Paphos and especially at the villages of Kathikas and Arodes. Those findings initiated the formation of a specialised prevention program that covered the population of those areas by gene screening of all young couples.

The possibility of the aforementioned pilot registries has been enabled through the interest and funds of research and academic centres. Unfortunately, no research has been accomplished by the public sector.

## **CONCLUSION**

There are major weaknesses in the collection of data and inter-connectivity of the various registries. There is also a huge delay (since 2012) in the development of a specific national registry for RDs, which was one of the main objectives of the NCRD.

## **SUGGESTED ACTIONS**

1. The Health Monitoring Unit of the Ministry of Health and the Data Processing Department in collaboration with the Medical Professionals and the NCRD must document all the necessary information for the e-registration of RDs and their inclusion in the software that will be used by HIO. It is also essential that the Ministry of Health, the Ministry of Labour, Welfare and Social Inclusion and the Ministry of Education and Culture, release a circular for the amalgamation of all registries of the public sector and on a voluntary basis of those in the private sector. An immediate discussion between the Data Processing Department and the HIO must be arranged with the objective of setting the provisions for the inclusion of RDs registry in HIO's software.
2. Various informative and training programs must be initiated by using all available communication routes for the diffusion of information about clinical studies and especially the modification of legislations for conducting clinical trials.

## **THEME 5 – RESEARCH**

### **WORKSHOP FORMAT**

The Workshop was initiated by presentations by the Coordinators (Dr Paschalis Nicolaou and Dr Theodoros Kyriakides) on Research, providing background content for the discussions which followed.

The discussion, facilitated by the Coordinators, was completed with the statements of two patient advocates (Mr Panikos Voskos and Mrs Maria Charalambous) each providing their perspective on the Workshop theme.

### **INTRODUCTION**

The value of research is undeniable. Without research, progress cannot be achieved. It is due to research that new diagnostic tests, new treatments and better preventative and therapeutic programs can be developed. Today's research is tomorrow's healthcare services. Furthermore, research for RD often can provide new knowledge that leads to better understanding of diseases (both common and rare).

### **Research in Cyprus**

In Cyprus only the 0, 5% of the country's GDP is spent on research programs whilst the EU's average is 2%.

At this point 30 research groups in Cyprus are active and have published more than 400 papers for the main research centres i.e. the CING, the University of Cyprus and the Cyprus University of Technology.

The research portfolio of the Karaiskakeio Foundation can be used as an example. The Foundation maintains the national bone marrow registry, conducts research on immunology, paediatric oncology and haematology, in addition to undertaking a drug repurposing program for rare types of cancer. However, the Foundation meets various challenges due to limited funding availability.

Currently nearly 300 research programs are being conducted in Cyprus, in various fields, with a total funding estimated to be approx. 10,000,000 euros.

### **Value of international consortia**

Participation of patients in research programs is considered pivotal for both patients and the scientific community. Nevertheless, opportunities to join research programs are limited in Cyprus (due to small population and limited funding) and are difficult to locate due to the lack of a central coordinating body to provide information. This means that each interested patient or family has to locate research programs on their own. Most can be found abroad and participation is difficult since there is no state funding for their participation.

Through the participation in research programs (clinical trials or other) patients can achieve a better understanding of their disease, its etiology, its course and clinical outcome as well as impact on their health and quality of life.

In addition, research programs with participants from more than 1 country can provide:

- The exchange of knowledge and experience between medical professionals
- The networking between Reference Centres
- The establishment of a national or a regional patient registry, which can be of great significance in collecting information for the further understanding of a diseases course and development but also for the policy making on a national level.

An analysis of the participation of Cypriot patients in an international clinical trial for a pharmaceutical product for the Transthyretin Related Familial Amyloid Polyneuropathy (TTR-FAP), conducted by CING was presented. Cypriot patients were included in this clinical trial due to the fact that Cyprus has the fourth highest prevalence for TTR-FAP worldwide.

The value of finding new pharmaceutical products for RDs is unmeasurable. For example for TTR-FAP a year of pharmaceutical treatment costs around 120,000 euros per patient, while the cost of liver transplant costs 100,000 euros. Concerns were voiced regarding accessibility to such drugs upon the completion of clinical trials, given the high pricing of innovative products.

Furthermore, the use of off label drugs should be investigated and the development of compassionate use programs for patients that could benefit from certain drugs should be encouraged.

The European Reference Networks have already started to register patients, and subsequently this can lead to the increase of interest by pharmaceutical industries for conducting clinical trials as numbers of patients, and geographical orientations will be available. In the meantime, CING has received funding for the creation of a congenital anomalies registry.



Research results must be utilized by the national authorities for the purposes of policy making, with the contribution of citizens/ patients / family members that might have knowledge and expertise in specific areas.

All channels of community awareness should be used including the Volunteerism Commissioner and Patients Advocate Commissioner.

## **SUGGESTED ACTIONS**

1. Increase of the existing funding for research
2. Promotion for the development of a department that will coordinate research and clinical trials on a national level according to the standards of the European Registry of Clinical Trials

## **III. Conclusions**

### **THEME 1: PREVENTION AND EARLY DIAGNOSIS**

The currently available / running information programs provided by all Ministries and other relevant authorities need to be identified, leading to the development of coordinated strategy for prevention and awareness raising purposes. A sub-committee of the NCRD could undertake this, thus acting as the national focal point for the provision of this information. In addition, focus needs to be placed on the provision of education to healthcare professionals, either in the context of continuous professional education and/or academic courses. Furthermore, the efforts to define laboratory centres of expertise should continue in order to facilitate timely and accurate diagnosis, utilizing the expertise gathered by the European Reference Networks where necessary and strengthening the services offered by genetic counsellors.

### **THEME 2 – DIAGNOSIS & TREATMENT**

Healthcare professionals encounter few cases of each RD on account of the small population of Cyprus. Hence, a National Registry for RD must be implemented immediately in order to better understand the demographics of the various different RDs. This will provide a stepping stone for the identification of gaps and weaknesses, and enable targeted service planning (e.g. development of specific centres of expertise, networking with European Reference Networks etc). Alternative modes of clinical management / monitoring should be implemented in cases where there is limited expertise and reference hubs cannot be created (e.g. agreements with centres of expertise from abroad). Moreover, the imminent changes due to the implementation of the National Health System should be clarified, especially concerning the access to orphan drugs (taking into account pricing and reimbursement issues), regular updating of clinical guidelines to reflect current international research, transition from childhood/adolescent clinics to adult ones, improvement of the implementation of the Cross Border Healthcare directive etc. The active involvement of the NCRD in the organisations responsible for the National Health System should be pursued.

### **THEME 3 – SUPPORTIVE CARE, SOCIAL WELFARE & INCLUSION**

In Cyprus, numerous rehabilitation and supportive care services are provided in a scattered way without central coordination. However, the main objective is the development of services that allow the reintegration of patients at the societal and occupational levels. There are some supportive care services that contribute towards improving the quality of life of patients, but there is a lack of

services that cover the holistic management of RD patients. Specific policies targetting the improvement of social welfare services, social inclusion and supportive care programs should be pursued, including those under the responsibility of the Ministry of Health, Labour and Social Insurance, and Education and Culture. Relevant budgetary allocations should be incorporated by the Ministry of Finance.

#### **THEME 4 – REGISTRIES**

There are major weaknesses in the collection of data and inter-connectivity of the various registries. There is also a huge delay (since 2012) in the development of a specific national registry for RD, which was one of the main objectives of the NCRD. Hence, the implementation of the e-health record and codification systems under the National Health System is crucial to the identification of RD and enable suitable planning by the responsible Ministries and authorities.

#### **THEME 5 – RESEARCH**

Although a number of research consortia are active in Cyprus conducting research on RD, nonetheless the level of funding provided is limited. Moreover, due to the small number of patients it is difficult for researchers to conduct independent studies and hence patients interested in participating in trials need to identify relevant research groups abroad. Therefore, an improved coordination mechanism matching trials/research groups to patient participants should be developed.

## ANNEXE I – Programme

**NATIONAL WORKSHOP**  
**EUROPLAN**  
**RD-ACTION**  
**RARE DISEASE DAY**  
**2018**

EUROPLAN  
European Project for Rare Diseases Research and Development

RD-ACTION  
RESEARCH AND POLICIES FOR RARE DISEASES

Co-funded by  
the Health Programme  
of the European Union

**FRIDAY, 9 MARCH 2018**  
**• SEMELI HOTEL, NICOSIA • 08:00 - 17:00**

**ORGANISER:**

**UNDER THE AUSPICES OF:**

**SUPPORTERS:**

With support from Bank of Cyprus  
Volunteers

With support from the European Parliament  
Information Office Cyprus

# PROGRAMME

## OPENING SESSION

08:00-08:30 Registrations

08:30-09:00 Welcome Address:

- Dr Androulla Eleftheriou, President, Cyprus Alliance for Rare Disorders
- Mr. Yann Le Cam, Chief Executive Officer, EURORDIS
- Mr. Neoklis Sylikiotis, Member of the European Parliament
- Mrs. Christina Flourentzou-Kakouri, representing Mrs. Zeta Emilianidou, Minister of Labour, Welfare and Social Insurance
- Dr Christina Yiannaki, Permanent Secretary, Ministry of Health & President, National Rare Disease Committee

## EUROPLAN / RD-ACTION NATIONAL CONFERENCE

### Cyprus Strategic Plan for Rare Diseases: Toward the Future

09:00-10:15 **Pillar 1: Prevention & Early Detection**

Coordinators: Dr Michael Angastiniotis, Dr Adonis Ioannides

Patient Representatives: Mrs. Katerina Margaroni, Mr. Marios Vakanas

09:00-09:15 Presentations | 09:15-10:10 Discussion | 10:10-10:15 Conclusions

10:15-10:30 **Coffee Break**

10:30-11:45 **Pillar 2: Diagnosis & Treatment**

Coordinators: Dr Georgios A. Tanteles, Dr Panayiota Protopapa, Mr. Andreas Papaconstantinou

Patient Representatives: Mr. Demetris Genethlis, Mrs. Maria Ioannou

10:30-11:00 Presentations | 11:00-11:40 Discussion | 11:40-11:45 Conclusions

11:45-13:00 **Pillar 5: Research**

Coordinators: Dr Paschalis Nicolaou, Dr Theodoros Kyriakides

Patient Representatives: Mr. Panikos Voskos, Mrs. Maria Charalambous

11:45-12:00 Presentations | 12:00-12:55 Discussion | 12:55-13:00 Conclusions

13:00-14:00 **Light Meal**

14:00-15:15 **Pillar 4: Rare Diseases Registry**

Coordinators: Dr Eleni Zamba Papanicolaou, Prof. Christos Schizas

Patient Representatives: Mr. Miltos Miliadous, Mr. Philippos Akamas

14:00-14:15 Presentations | 14:15-15:10 Discussion | 15:10-15:15 Conclusions

15:15-16:30 **Pillar 3: Supportive Care, Social Welfare & Integration**

Coordinators: Dr Myrto Azina, Dr Androulla Eleftheriou, Mrs. Marina Pavlou

Patient Representatives: Mrs. Anna Zannetou, Mrs. Maria Kyriacou

15:15-15:30 Presentations | 15:30-16:25 Discussion | 16:25-16:30 Conclusions

16:30-16:45 **Conclusions** - Dr Androulla Eleftheriou

16:45-17:00 **Next Steps** - Dr Christina Yiannaki

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RARE  
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CARE





# CONTRIBUTORS

## SPEAKERS

- Dr Michael Angastiniotis, Thalassaemia International Federation
- Dr Adonis Ioannides, University of Nicosia
- Dr Georgios A. Tanteles, Cyprus Institute of Neurology and Genetics
- Mr. Andreas Papaconstantinou, Health Insurance Organisation
- Dr Panayiota Protopapa, Archbishop Makarios III Hospital
- Dr Paschalis Nicolaou, Cyprus Institute of Neurology and Genetics
- Dr Theodoros Kyriakides, Cyprus Institute of Neurology and Genetics
- Dr Eleni Zamba Papanicolaou, Cyprus Institute of Neurology and Genetics
- Prof. Dr Christos Schizas, University of Cyprus
- Dr Myrto Azina, Ministry of Health
- Dr Androulla Eleftheriou, Cyprus Alliance for Rare Disorders
- Mrs. Marina Pavlou, Cyprus Institute of Neurology and Genetics

## PATIENT REPRESENTATIVES

- Mrs. Katerina Margaroni, Huntington's Disease Association - Cyprus
- Mr. Marios Vakanas, Association of Inherited Metabolic Diseases "Aspida Zois" - Cyprus
- Mr. Demetris Genethlis, TTR FAP Association of Cyprus
- Mrs. Maria Ioannou, Children with Liver Diseases – George Psaras – Round Table
- Mr. Panikos Voskos, Muscular Dystrophy Association Cyprus (MDA Cyprus)
- Mrs. Maria Charalambous, Cyprus Primary Immunodeficiencies Association and Friends
- Mr. Miltos Miltioudou, Pancyprian Thalassaemia Association
- Mr. Philippos Akamas, Cyprus Liver Patient and Friends Association "Prometheus"
- Mrs. Anna Zannetou, Myasthenia Gravis Association Cyprus
- Mrs. Maria Kyriacou, Cyprus Brain Tumour Association

## MODERATOR

- Mrs. Eleni Antoniou, Thalassaemia International Federation

## RAPPORTEURS

- Mr. Spyros Polyviou, Cyprus Alliance for Rare Disorders
- Mrs. Lily Cannon, Thalassaemia International Federation

## OVERALL COORDINATION

- Dr Androulla Eleftheriou, Cyprus Alliance for Rare Disorders



## ANNEXE II - Participants List

NAME	SURNAME	CAPACITY	ORGANISATION	STAKEHOLDER GROUP
Christos	Schizas		University of Cyprus	Academic/Researcher
Leonidas	Phylactou	Medical Director	Cyprus Institute of Neurology & Genetics	Academic/Researcher
Pascalis	Nicolaou	Researcher	Cyprus Institute of Neurology & Genetics	Academic/Researcher
Pavlos	Costea	Director	Karaiskakeio Foundation	Academic/Researcher
Popi	Kanari	President	Karaiskakeio Foundation	Academic/Researcher
Adonis	Ioannides	Geneticist	University of Nicosia	Clinician/GP
Costas	Michaelides	Neurologist	American Heart Centre	Clinician/GP
Eleni	Zamba Papanicolaou	Neurologist	Cyprus Institute of Neurology & Genetics	Clinician/GP
Eliza	Markidou	Head	Nutrition Dept, Arch. Makarios III Hospital	Clinician/GP
Georgia	Tsiappa	Nutritionist	Nutrition Dept, Arch. Makarios III Hospital	Clinician/GP
Georgios	Tantele	Geneticist	Cyprus Institute of Neurology & Genetics	Clinician/GP
Nicos	Makrides	Paediatrician		Clinician/GP
Panayiota	Protopapa	Paediatric Gastenterologist	Liver & Gastenterology Clinic, Arch. Makarios III Hospital	Clinician/GP
Theodore	Kyriakides	Neurologist	Cyprus Institute of Neurology & Genetics	Clinician/GP
Andreas	Kettis	Head	Office of the European Parliament in Cyprus	European Body
Neoclis	Sylikiotis	Member	European Parliament	European Body
Antonis	Louroutziatis	General Manager	Genesis Pharma	Industry
Eria	Nicolaou		MSD	Industry

Kyriakos	Mikelli	General Manager	Pfizer	Industry
Maria	Simiakaki	Executive Director	Sanofi	Industry
Maria	Nicolaou	Patient Advocacy Head	GlaxoSmithKline	Industry
Poly	Toumpa	Government & Public Affairs Offices	Novartis	Industry
Sofronis	Savva		Genzyme	Industry
Tania	Kyriakides	Sales Manager	Genesis Pharma	Industry
Despoina	Kaimi	President	Cyprus Occupational Therapist Association	Medical/Learned Society
Maria	Christopoulou		Cyprus Association of Speech Therapists	Medical/Learned Society
Soteris	Koumas	President	Cyprus Union Government Doctors	Medical/Learned Society
Maria	Kalli	Psychologist		Other Healthcare Professional
Marina	Pavlou	Social Worker	Cyprus Institute of Neurology & Genetics	Other Healthcare Professional
Marina	Kleanthous	Head, Thalassaemia Molecular Laboratory	Cyprus Institute of Neurology & Genetics	Other Healthcare Professional
Marios	Filaktides		Cyprus Institute of Neurology & Genetics	Other Healthcare Professional
Marios	Vogiazianos		Centre of Preventative Paediatrics	Other Healthcare Professional
Marinos	Sizopoulos	President	EDEK	Parliamentary Party
Androulla	Eleftheriou	President	Cyprus Alliance for Rare Disorders	Patient Representative
Anna	Zannetos	President	Myasthenia Gravis Association	Patient Representative
Charis	Charalambous	RD Patient Spouse	Association of patients with Primary Immunodeficiencies	Patient Representative
Christina	Papamiltiadous	President	Cystic Fibrosis Association	Patient Representative

Constantinos	Ierides	Secretary	Elpidoforos Association for Persons with Psychiatric Disorders	Patient Representative
Dimitris	Genethlis	President	Cyprus Amyloidosis Association	Patient Representative
Eleni	Farmaka	RD Patient	Myasthenia Gravis Association	Patient Representative
Epifania	Constantinou	Member	Muscular Dystrophy Association	Patient Representative
Giannoula	Koulla	Secretary	Association for Liver Disease Patients 'Promitheas'	Patient Representative
Irene	Patsalidou	Member	Anti-Rheumatism Association	Patient Representative
Katerina	Margaroni	Administrator	Cyprus Huntington Disease Association	Patient Representative
Katia	Kyriakou	President	Monadika Hamogela	Patient Representative
Kyriakos	Karatzias	Member	Myasthenia Gravis Association	Patient Representative
Margarita	Georgiou	RD patient parent	Association of patients with Primary Immunodeficiencies	Patient Representative
Maria	Ioannou	President	Association of Paediatric Liver Disease Patients	Patient Representative
Maria	Charalambous	President	Association of patients with Primary Immunodeficiencies	Patient Representative
Maria	Kyriakou	Administrator	Muscular Dystrophy Association	Patient Representative
Maria	Nicou	Member	Muscular Dystrophy Association	Patient Representative
Maria	Kyriakou	Member	Cyprus Brain Tumor Association	Patient Representative
Marios	Vakanas	President	Association for Hereditary Metabolic Diseases	Patient Representative
Marios	Dimitriou	Member	Myasthenia Gravis Association	Patient Representative



Marios	Kouloumas	President	PanCyprian Federation of Patient Associations	Patient Representative
Melina	Kammenou	RD patient parent		Patient Representative
Michael	Angastiniotis	Paediatrician	Thalassaemia International Federation	Patient Representative
Miltos	Miltiadous	President	Cyprus Thalassaemia Association	Patient Representative
Nicolas	Philipou	Executive Director	PanCyprian Association of Cancer Patients and Friends	Patient Representative
Niki	Kameri	Member	Monadika Hamogela	Patient Representative
Pambos	Papadopoulos	President	Muscular Dystrophy Association	Patient Representative
Panicos	Voskos		Muscular Dystrophy Association	Patient Representative
Panos	Englezos	President	Thalassaemia International Federation	Patient Representative
Pantelitsa	Vakanas	Member	Association for Hereditary Metabolic Diseases	Patient Representative
Philippos	Akamas	President	Association for Liver Disease Patients 'Promitheas'	Patient Representative
Popi	Eleftheriadou		Haemophilia Patients Association	Patient Representative
Soteris	Zannetos	RD Patient Spouse	Myasthenia Gravis Association	Patient Representative
Stalo	Kyriakidou	Administrator	Monadika Hamogela	Patient Representative
Xenofon	Ioannides	President	Elpidoforos Association for Persons with Psychiatric Disorders	Patient Representative
Yann	Le Cam	CEO	EURORDIS	Patient Representative
Andreas	Papaconstantinou	Acting Director	Health Insurance Organisation	Public Administration

Christina	Giannaki	Director General	Ministry of Health	Public Administration
Christina	Flourentzou	Director	Dept. Social Inclusion for Persons with Disability	Public Administration
Christodoulos	Kaisis	Director	Arch. Makarios III Hospital	Public Administration
Giannis	Giannaki	Commissioner	Volunteerism and NGOs	Public Administration
Irene	Papatheodoulou	President	Inter-Ministerial Committee for Rare Diseases, Ministry of Labour & Social Insurance	Public Administration
Myrto	Azina	Officer	Ministry of Health	Public Administration