



ITALY

EUROPLAN NATIONAL CONFERENCE

in the framework of the EU Joint Action RD-ACTION

Rome, 9-10 February 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, 30 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

Country	Italy
National Alliance (Organiser)	UNIAMO Federazione Italiana Malattie Rare onlus
Date & place of the national workshop/conference	9 th – 10 th February 2018 Bambino Gesù Children’s Hospital Rome
Website	www.uniamo.org
Members of the Steering Committee	UNIAMO Board of Directors SIMONA BELLAGAMBI ROMANO ASTOLFO Sinodè
List of Themes addressed	<ul style="list-style-type: none"> • ERN – European Reference Networks • PDTA – Diagnostic and Therapeutic Care Pathways • Integration of Rare Diseases in Social Policies and Services
Annexes :	I. Programme in English II. List of Participants

FINAL REPORT

I. Introduction/ Plenary session

The opening speeches of the plenary session (Mrs. Tommasina Iorno, President of UNIAMO FIMR Onlus; Prof. Bruno Dallapiccola, Scientific Director at Bambino Gesù Children’s Hospital; Prof. Paola Facchin, Coordinator of the Interregional Table for Rare Diseases of the Conference of Regions and Autonomous Provinces) have all stressed the **need for early and adequate updating of national and regional plans** in the fields of rare diseases, to be carried out through the **new National Plan for Rare Diseases for the period 2017 – 2020**. Dr.ssa Simona Bellagambi, EURORDIS - EUROPLAN Advisor gave a presentation on the EUROPLAN Project, the State of Art report on rare diseases and RD-ACTION National Workshops.

Within this framework, three working groups have been created and dedicated to:

1. ERNs – European Reference Networks
2. PDTAs – Diagnostic and Therapeutic Care Pathways
3. Integration of Rare Diseases in Social Policies and Services

II. Themes

THEME 1: ERNs

Less than a year after the launch of ERNs, it is still not possible to appreciate a significant impact on the lives of persons suffering from rare diseases, but expectations remain high. Actually, ERNs represent a quite new pathway, which will be further developed in the medium and long term. The main benefit for patients with rare diseases will come from the link that will be created between ERNs and National and Regional Networks for RDs. In Italy, the launch of ERNs was characterized by some weaknesses, like the delayed setting up of the National Coordinating and Monitoring Body for the development of ERNs (this national coordinating and monitoring body for the development of ERNs is specific to Italy and was defined by MD of 27 July 2015), the failure to involve in this body representatives of patients with rare diseases and of some Regions and the lack of uniformity in the implementation of the criteria set out in the early stages of Centres' identification. To this shall be added the heterogeneity in the implementation of the activities of ERNs and the relevant timing and, in particular, in the involvement of representatives of patients with rare diseases. Two ERNs have Italian coordinators (ERN-Bond, Dr Luca Sangiorgi, Ern-ReConnect, Prof Marta Mosca). In the EU, Italy has the highest number of Centres which have joined ERNs: 189 centres belonging altogether to 23 ERNs (out of 24 ERNs). A management burden (for Centres that are not ERN coordinators) was observed, which is not covered by any support to operating costs. Finally, a lack of dialogue between the many Italian Centres belonging to the same ERN was noticed, as well as the need to disseminate information among all healthcare professionals concerning ERNs' aims and activities.

In this regard, the long-awaited National Plan for Rare Diseases 2017 – 2020 should namely:

- **Harmonize and enhance the role of reference centres/Italian hospitals belonging to ERNs with the work carried out in national and regional care networks**, dedicated both to rare diseases and care/social issues of general interest.
- **Set up a regional/national governance system** of the fields of activity of reference centres/hospitals belonging to ERNs to support interaction and integration actions and tools, such as registries, guidelines and diagnostic therapeutic and care pathways, enabling the full functioning of the National Coordinating and Monitoring Body for the development of ERNs (MD of 27 July 2015, art. 13, paragraph 2 of LD no. 38 of 4 March 2014), in which **it is fundamental to provide for the involvement of patient representatives.**
- **Devise function-based funding systems and/or systems based on specific services (e.g.: remote consultancy)**, that are relevant for reference centres/hospitals institutionally belonging to the National/Regional RD Network and also members of ERNs, through the implementation of the State-Regions Agreement of January 2015 on remote consultancy and management (being the Italian Health System heavily regionalized, agreement as this one between the two levels, are needed to guarantee that Regions will have funds to support specific functions or services).
- **Carry out organization and management tests** to refocus the internal organization of hospitals, hosting the Centres that belong to ERNs. The aim is to facilitate the interdisciplinary and multi-professional management of patients with rare diseases, **thus creating the RD functional platform** that will serve as a single physical location for access to care. On this RD functional platform all the professionals of Operational Units (OUs)

belonging to the same Centre – even if activated at different hospitals (e.g.: dedicated to children or adults) – can assess the patient’s needs and define a **single and integrated management plan**. To this end, it is also necessary to **experimentally re-organize hospital budgets**, where the RD Platform exists, **according to the rare disease patient’s pathway, and not to the services provided by each single OU**. Such an organizational and methodological change is needed to refocus the system for the care of patients with rare diseases, according to their complex care needs, in an integrated way; its extension to the whole national and regional healthcare system is much advisable.

- Reduce the number of undiagnosed persons who may be potentially affected by rare diseases; reduce the time needed to reach a diagnosis through an **adequate and appropriate access to diagnosis infrastructures**, for example by **including next-generation sequencing techniques in LEAs** (essential levels of care). In Italy, neonatal screening was mandatory throughout the country for 3 diseases: phenylketonuria, congenital hypothyroidism and cystic fibrosis. Since January 2017, with the new LEAs, the total number is around 40 with the extension to all newborns of enlarged neonatal screening for hereditary metabolic diseases.

THEME 2: PDTA – Diagnostic and Therapeutic Care Pathways

Diagnostic and Therapeutic Care Pathways (PDTAs) are a clinical management tool used to devise the best care pathway to meet specific healthcare needs, according to recognized recommendations (guidelines, scientific consensus, etc.) and adapted to the local context. PDTAs aim at enhancing the integration between healthcare professionals, reducing clinical variability, contributing to the dissemination of evidence-based medicine, using resources in a reasonable way and allowing for the assessment of the services provided through specific indicators.

Despite the indications already present in the NPRD (National Plan for Rare Diseases) 2013-2016 and many other reference documents, the dissemination of PDTAs in the field of rare diseases is progressing very slowly. Furthermore, PDTAs very often do not fully meet the purpose for which they were designed; this is due to the lack of awareness by the social and healthcare system on what PDTAs are and how they differ from two other tools: scientific guidelines and Individualized Care Plans (ICPs).

A PDTA plays a key role for people suffering from rare diseases because **it contains the list of services that can be claimed by the patient in the regional context, at the healthcare, social and care levels**. This is a tool that:

- encourages the appropriateness of organization, diagnosis and prescription;
- ensures the connection between the nodes of the national/regional RD network and all the services involved, by supporting the multidisciplinary approach of the global management;
- shows clear and transparent pathways to guarantee the rights that can be enforced within a specific regional context;
- ensures the continuity of care (transition medicine, continuity between different care settings);
- defines the responsibilities of the different actors;
- optimizes resources.

Therefore, PDTAs shall be inspired by guidelines containing all the scientific knowledge acquired. On the other hand, ICPs have to be defined by the Reference Centre, identifying in the territory where the RD patient lives, the reference point that is responsible for its implementation.

According to the features of such tool, the working group agrees that the PDTA shall:

- have at least a regional role, with possible interregional cooperations;
- belong to the Region;
- be dependent on the place and time of implementation (need for updates);
- contain the definition of the actor providing specific services in that context.

The first critical issue is due to the fact that, by their very nature, rare diseases often lack specific guidelines PDTAs should be based on. In such case, the drafting process will be inspired by good clinical practice recommendations.

The second critical issue depends on the large number of diseases that could potentially be included in PDTAs. For this reason, the Group deems that PDTAs for RDs can be drawn up not only by disease (when the numbers allow so) and/or by groups of rare diseases, but mainly by specific care needs (that can be common to different conditions), according to which the organization pathway shall be conceived and assured in the life context of the person affected by a rare disease, including social services and rehabilitation.

The following actions shall be integrated in the new NPRD:

- **Drafting of PDTAs:** the PDTA will assess the available resources, building an ideal path to develop the **reference pathway within the context**, i.e. the best space-time sequence possible for the activities to be carried out in a specific context of organization and resources. In the PDTA setting-up process, a key role is played by the exploitation of the competence and experience of the patient – through the involvement of the associations that are most representative of RD patients at the regional level;
- **Implementation of PDTAs and relevant monitoring actions:** information and training for all the actors of the system; monitoring of the process and outcome indicators; methods of intervention in case of non-compliance with the standards; periodic review.

THEME 3: Integration of Rare Diseases in Social Policies and Services

The theme of rare diseases in social policies and services in Italy is very complex due to the differences in the provision of social and care services at the regional level, that are magnified by the difficult – and often lacking – integration between social services, territorial healthcare services and Reference Centres for rare diseases.

Furthermore, social policies were most notably absent in the NPRD 2013-2016. In fact, in the programming document, no mention is to be found on social care interventions; this contrasts greatly with the central role of the social dimension in the everyday life of patients affected by rare diseases and their families. Social protection, integration into the school system, employment and social inclusion were not taken into account. In this regard, the NPRD 2017-2020 shall stress the need for the definition of Essential Levels of Social Care, as well as:

- Encourage the integration of rare diseases in social policies and services through a **stronger cooperation between healthcare and social services**, that shall recognize the Multi-dimensional Assessment Unit as the place where the social and healthcare integration and the integrated management of the RD patient take place, **enhancing the contribution of the Reference Centre for rare diseases in the functional assessment** of the patient. The Multidimensional Evaluation Unit (UVM) is established by a team of specialists composed by

various professionals of the clinical, health and social area whose function is to determine the care need of the patient with RD with regard to health, social and psychological issues;

- Encourage the training of healthcare professionals and social workers on social protection mechanisms for patients with RD, supplying specific training courses. The same training should be offered also to RD patients and their families, to further disseminate awareness of the rights they are entitled to;
- Support a **stronger cooperation between INPS** (Italian National Social Security Institute) **commissions for the recognition of invalidity/assessment of the handicap and Reference Centres** for a stronger social protection of RD patients, focusing on the assessment of their care needs;
- Support the **requalification of the functional evaluation of the Individualized Education Program (IEP) at school**, conceived as a care and integration action by professionals and based on the functional assessment carried out by the Reference Centre, with special attention to the capacity to translate the IEP into concrete actions;
- Help overcome the bureaucratic barriers to allow patients **to take medicines and to ensure the provision of other care services (e.g.: aspiration) at school**;
- Ensure, also through the **implementing decrees** of LD no. 151/2015, a more adequate system for the **access to the labour market by RD patients**, considering the functional assessment of the person affected by a rare disease and providing resources dedicated to the assistance to the RD patient in the first working period;
- **Increase the number of tools needed to support the independence of the RD patient**, also through the so-called “after us” fund, tax benefits on wealth funds extended to medium-mild disabilities and tax disregard for financial investments for RD patients.

III. Conclusions

Considering the limits in terms of time and issues considered, that did not allow for the discussion of many other factors of major importance for RD patients (such as the access to medicines and research), the Conference stressed the urgent need for a new National Plan for Rare Diseases 2018-2020 aimed at encouraging early diagnosis and a better quality of life for RD patients and their families. The new NPRD shall strengthen the healthcare issue through the “networking” of resources in terms of:

- *Relationship between OUs and Reference Centres of the national /Regional Network belonging to ERNs*, through innovative organizational solutions such as the “RD functional platform” for an interdisciplinary and multiprofessional management of RD patients and the introduction of function-based RD funding;
- *Relationship between Centres and between Centres and territorial (social and healthcare) services*, through the harmonization and enhancement of the role of Centres belonging to

ERNs in relation to the specific national and regional institutional and organizational set-up of care networks for RD patients, to be implemented also through remote consultancy;

- *Relationship between different services, involved in the care provided to the RD patient, through the development of Diagnostic and Therapeutic Care Pathways.*

To meet such objectives, the new NPRD must include a **dedicated funding**, with a special attention to the **development of professionals' knowledge and skills on new technologies and infrastructures**, that are necessary to keep the public healthcare system abreast of the times.

At the same time, the government action must coordinate and integrate the administrative functions that shall meet the overall needs of the RD patient, overcoming today's disjunction between healthcare, social, working, welfare (and many other) actions. Therefore, **the new planning must absolutely integrate social and healthcare issues, to encourage a comprehensive management of RD patients and ensure their actual social inclusion.**

Finally, the new NPRD shall **strengthen the assessment issue**, through the definition of dedicated indicators aimed at monitoring and evaluating the degree of achievement of the expected results.

ANNEXE 1

PROGRAMME
RD-ACTION NATIONAL CONFERENCE
Bambino Gesù Children's Hospital AUDITORIUM
Rome, 9th – 10th February 2018

Friday, 9th February 2018

- **10:15 a.m.: Participants' registration**

Plenary session open to the members of working groups; the project and the state of the art will be presented, together with the activities planned for the two-day meeting and its objectives

- **10:30 a.m.: Welcome speech by Prof. Dallapiccola, Mrs. Iorno (UNIAMO President) and Presentation National RD-ACTION National Workshops Mrs. Bellagambi (EURORDIS EUROPLAN Advisor)**
- **10:50 a.m.: RD ACTION Project – Mrs. Facchin – Veneto Region Coordinating Centre**
- **11:20 a.m.: Presentation of the working groups**
- **11:45 a.m. – 01:00 p.m.:**
Start of works by the thematic working groups
The following issues will be addressed:
 - ❖ ERNs (European Reference Networks) Reti di riferimento Europee – Chair Mr. Flavio Minelli
 - ❖ PDTAs (Diagnostic and Therapeutic Care Pathways) – Chair Mrs. Renza Barbon
 - ❖ Integration of Rare Diseases in Social Policies and Social Services – Chair Mrs. Riccarda Scaringella
- **1:00 p.m. – 2:00 p.m.: Lunch**
- **2:00 p.m. – 6:00 p.m.**
Continuation of works; production of slides showing the results

Saturday 9th February 2018

- **09:00 a.m. – 10:0 a.m.**
Conclusion of works by the working groups – if needed
- **10:15 a.m. – 11:15 a.m.**
Speech by Dr R.Migliorini INPS Medical Examiner General Coordination
Plenary session, open to the public – presentation of the results of the working groups
- **11:30 a.m. – 1:00 p.m.**
Discussion and delivery of key messages
- **1:00 p.m.: Conclusion**

ANNEXE 2 List of Participants

Working Group on ERNs

- Ales Claudio – Representative of the ERN GENTURIS
- Astolfo Romano – Sinodè Srl
- Coviello Domenico – Italian Society of Human Genetics
- Dallapiccola Bruno – Bambino Gesù Children’s Hospital
- Facchin Paola – Interregional Round Table on Rare Diseases
- Marinello Diana – PM ERN ReCONNET
- Marino Valentina - Pfizer
- Minelli Flavio – Representative of ePAG ERN Skin
- Mostaccioli Stefania – Representative of the ePAGS ERN GENTURIS
- Torreri Paola – Italian National Institute of Health

Working Group on PDTAs

- Barbon Renza – UNIAMO Italian Federation for Rare Diseases Onlus
- Borgia Rosa – CARD Italia
- Calmasini Michela – ULSS 3 Serenissima
- Cifani Andrea – Pfizer
- Ciofi Degli Atti Marta – Bambino Gesù Children’s Hospital
- Da Riol Rosalia – Coordinating Centre for Rare Diseases of Friuli Venezia Giulia Region
- Genuardi Maurizio – Italian Society of Human Genetics
- Kodra Ylka – Italian National Institute of Health
- Macrì Francesco – Federation of Italian Medical and Scientific Societies
- Marinelli Pietro – UNIAMO Italian Federation for Rare Diseases Onlus
- Mazzucato Monica – Interregional Round Table on Rare Diseases
- Piccinocchi Gaetano – Italian Society of General Medicine
- Porchia Stefania – Sinodè Srl
- Vaccarotto Manuela – UNIAMO Italian Federation for Rare Diseases Onlus
- Verna Roberto – Federation of Italian Medical and Scientific Societies

Working Group on Social Services

- Bona Gabriele - Association for Rheumatic Diseases in Childhood Onlus
- Calderone Maria Cinzia – Sicilian Association IRIS M.E.M. Onlus
- Celesti Lucia – Bambino Gesù Children’s Hospital
- Ciampa Serena – Orphanet Italia
- Conte Maria Stella – Bambino Gesù Children’s Hospital
- De Santis Marta – Italian National Institute of Health
- Ghirardini Alessandro – AGENAS
- Iorno Tommasina – President of UNIAMO Italian Federation for Rare Diseases Onlus
- Manea Silvia – Interregional Round Table on Rare Diseases
- Scaringella Riccarda – Association Network for Rare Diseases Onlus, Network AMARE Puglia Region
- Scopinaro Annalisa – Williams Syndrome Association, Toscana Forum