EUROPLAN - NATIONAL CONFERENCES

Final Report of the conference in ITALY

COUNTRY	ITALY
Date and place of the national conference	11/13 November 2010 Firenze Centro Formazione Montedomini Via Dei Malcontenti, 6
Website	www.uniamo.org
Promoter	UNIAMO F.I.M.R. onlus
Steering Committee	1. AIFA – Italian agency for medicine 2. BIO ETHICAL CENTRE CLINIC AND Governance OF HEALTH - IRCCS Cà Granda INSTITUTION 3. CERISMAS – Centre of research and study of health management. 4. FARMINDUSTRIA 5. Federazione UNIAMO F.I.M.R. Onlus 6. Federsanità-Anci 7. FIMMG – Italian Doctors of General Medicine confederation. 8. FIMP - Italian confederation of paediatricians. 9. FISM – Italian confederation Medical scientific societies. 10. MINISTRY OF PUBLIC WELFARE 11. Ministry of Labour and Social Policy 12. ORPHANET ITALY 13. SIGU – Italian Society Human Genetic. 14. SIMG – Italian Society of General Medicine. 15. SIMGePeD – Italian Society of Paediatric Genetic disease and congenital Disabilities. 16. TASK FORCE – now Committee of Experts of rare disease of European Union. 17. TELETHON 18. INTER – REGIONAL UNION FOR RARE DISEASE – Co-ordination health committee conference of Regions and Autonomous Provinces 19. SINODE' s.r.l.
Names and list of Workshops	**n. 1 Methodology and Governance of a National Plan / Strategy (NP)** **n. 2 Definition, codification and inventorying of RD** **n. 3 research on RD** **n.4 Standards of care for RDs - Centres of Expertise (CoE)/ European Reference Networks (ERN)** **n. 5 Patient Empowerment and Specialised Services** **n. 6 orphan drugs and accessibility to treatments**
Chairs and Rapporteurs of Workshops **n. 1 governance** **n. 2 code – training and information** **n. 3 research** **n. 4 networks, centres of experience** **n. 5 empowerment** **n.6 orphan drugs and accessibility to treatments**	**Chairs** / **rapporteurs** Marco Sessa – Giuglietta Cafiero Renza Barbon – Maria Marcheselli Fabrizio Seidita – Elisa Grella Michele Del Zotti – Maria Pia Sozio Annalisa Scopinaro – Antonella Esposito Loredana Nasta – Gabriele Bona
Enclosures (programme, list of participants etc.)	Conference program Members of work groups Participants' list

I. General Information

In N**ovember 2009** the former Minister for labour, public health and social policy On.le Maurizio Sacconi was informed about the organization, planned for the year 2010, of the National Conference within EUROPLAN project with the necessity of setting up a multidisciplinary steering committee (SC).
With the change of the institutional disposition of the Public Health Ministry, in **February 2010,** on the occasion of the RDD, the Minister of Public Health Prof. Ferruccio Fazio, designated as his representative Prof. Bruno Dallapiccola, former Scientific Director of Mendel Institute, and now director of the Children Hospital "Bambino Gesù" in Rome, supported by two technicians of the Health Ministry, Dr Giovanni Ascone, member of the General Directorate of Prevention , and Dr Maria Elena Congiu member of the General Directorate of Planning. That represented the starting point of the organization of the SC.

II. Main report

In **March 2010,** a first strategic meeting was developed in Rome (Barbon Galluppi, Bellagambi e Dallapiccola) whose intent was of identifying the corporations that could take part in the Steering Committee and the date of the first official meeting.
Then the invitation letters were sent, and on the 13th April 2010, the first meeting took place, by the Public Health Ministry in Rome, representatives of some of the invited corporations took part in it.
A letter of intent was written by UNIAMO F.I.M.R. onlus and signed by the others corporations of the SC.
Then monthly meetings took place (May 5th / June 7th / July 14th and September 8th, 2010).
From October 12th, 2010 a Public Consultation was activated for a wider involvement of different stakeholders, whose outcomes were elaborated and presented during the Conference as enclosure of the final document.
In order to get significant outcomes from the discussion, it was decided that the six working groups must include all the different stakeholders (patients, health professionals, institutions, industry and their representatives).
The **SC** decided to assign both the roles – Chair and Rapporteur – to **patients or patients'relatives.** Therefore patients'representatives in the 6 groups ended up to be **18** (6 Chairs, 6 Rapporteurs and 6 members), identified by UNIAMO FIMR with very precise criteria legitimated by the SC during the meeting that took place on September 11th, 2010.

EURORDIS Advisor, Simona Bellagambi, co-ordinated the Chairs in the management of groups.

In order to go on with the work a "virtual office" was developed – inside this space all the documents of the project EUROPLAN were posted, either general (for example: Council Recommendation ; the EUROPLAN Recommendation) or specific ones divided into subjects (for example: ad hoc open questions and the final document on "**good practices"**). In addition every stakeholder posted relevant documents to contribute to the discussion on the specific subjects of the WG.

Following the change of representation of the Health Committee in the State-Regions Conference, the Technical Board for Rare Diseases formed by several regional representatives decided belatedly to make a contribution in a consistent manner. They had their own "space" in the virtual office and they also took part in the final working groups in Florence after two meetings held in Rome

Transversal Members for all Groups: CERISMAS and SINODE' s.r.l.

STATE OF THE ART

In order to contextualise the government system of health policy for rare disease currently being developed in Italy, we must say that Italy has a public and universalistic healthcare system and moreover the institutional authority for health matter is constitutionally divided into two levels, the Central Government and the Regions.
The central level guarantees the right to health, and makes an index of the treatments due to Italian citizens and to foreigners regularly living in Italy LEA (Essential levels of primary care), while the regional one is in charge of the planning and of the organization of services, in order to guarantee the effective output of these treatments. The Regions have also the possibility to expand the levels of the national primary care with further treatments that can be made at total expense of the Regions.

The provision that started the enactment and implementation of specific health policies for people with RD, is the Ministerial Decree n. 279/2001 that:

- Guaranteed specific rights to Rare Disease patients,
- Established an official list of Rare Disease for which these rights are payable ;
- Defined the burden of the National Health Service and, as from November 2001 of the Regions, to identify in a formal way the Centres of Reference where RD patients can and must refer in order to have a diagnosis and care;
- Established a monitoring system organised in both a local level and a more concise national stream to support national strategies and LEA in the hands of the National Register at the ISS (National Health Institute)

As a follow up of that Decree either the Government or the Regions have enacted many acts that have, step by step, made the following Government system (see enclosure n 1 and 2)
The Regions, with the State/Regions agreement made in 2007 took up the responsibility to build within their health systems an organization of functions and services dedicated to Rare Diseases, in that way they have created a regional network managed by a Coordination Centre. The Ministry bears the burden of implementation and management of national policies, supported in this by technical-institutional bodies such as the National Institute of Health (ISS), the Italian Pharmaceutical Agency (AIFA). The link between the national and the regional level has to be made by the State/Regions Conference and advised for the technical aspects by a State/Regions permanent technical Board (see enclosure n. 1 and 2).This Board includes all the representatives of the Regions and of the Health Ministry and, where competent, the representatives of the National Register - ISS (National Health Institute) and AIFA (Italian Pharmaceutical Agency) The patient Associations' representatives and of the Scientific Societies are not involved.

MAIN THEMES

Theme 1 - Methodology and Governance of a National Plan / Strategy (NP)

Sub-Themes

1. Mapping exercise before developing a National Plan
2. Development and structure of a National Plan / Strategy
3. Governance of a National Plan
4. Monitoring the National Plan
5. Sustainability of the National Plan

Monitoring of a National Plan

STRENGTHS

1. The course of the past 10 years has increased the general awareness on Rare Diseases at the level of institutions, public and private technical organizations, associations of patients
2. A higher and dedicated level of care is given to RD patients, including also some treatments.
3. Priority procedures used to value and to authorise medicines compared to those without the European certification of orphan drugs, and moreover procedures used to value and authorise the use, "off-label" for rare therapeutic indications paid by the SSN (National Healthcare Service).
4. The establishment of the State/Regions technical Board created the opportunity of integration between the different institutional levels and the achievement of relevant results.

a. The creation of networks of centres of reference for Rare Disease that is an integrating part of the functions and systems of the regional healthcare services (for example: palliative care, rehabilitation treatments, primary care, and domiciliary assistance etc.);
b. The start of actions of national co-ordination on research concerning RD and their treatment;
c. The definition of a list of Rare for which recognise special rights;
d. The development of agreements and the improvement of actions of inter-regional co-operation whose aim is the planning of interventions and actions in the field of Rare Diseases, either by free regional initiatives, or by the stimulation of Health Ministry through aimed funding ;
e. The development of a monitoring system that supports the regional networks of care.

CRITICAL ASPECT

1. Absence of the representatives of patients and Scientific-societies at the inter-institutional Boards.
2. The quality of treatments and the organization of healthcare are not homogeneous on the national territory. Differences depend upon the regional health systems, and upon the measures enacted by each administration.
3. The difficulty to translate into reality the daily care as defined by the national health policy, with a consequent different perception felt by patients, on the provision and quality of the service.
4. The scarce awareness of patients and their families about their rights and how to go to see them guaranteed.
5. The list of rare diseases in not complete and not regularly updated, that's why some patients affected by rare diseases not included in the list, don't have the possibility to achieve their rights.
6. The distance between centres of reference and the residence of the patient carries a high social cost for the continued transfer: there is also an inverse relationship between the quality of care and the distance from the center of care.

GOALS

1. **Identify one** National Committee that could give voice to all actors involved (patients, the Ministry of Health, the Ministry of Education, University and Research, the Ministry of Labour and Social Policies, and other ministries ; Regions, the AIFA (Italian Pharmaceutical Agency), the ISS, Scientific Societies ..) in order to ensure the widest possible involvement , and outlines the strategies to be implemented in assistance, research, protection and social promotion, training and information, in rare diseases
2. **Promote** all the actions of agreement or co-operation amongst Regions aiming at the establishment of areas of inter-regional intervention gradually homogeneous in care provision .

The offer must be efficient with respect to the necessary level of experience concentration and expertise, adequately comprehensive and integrated in order to allow the real access to care to all people in need and a comprehensive care which is not only medical care but also social-care.

3. **Adopt** a measure of integration of the strategies and action plans in place or to be developed, consisting of a National Plan on Rare Diseases, adjusted to take account of the regulatory environment, institutional and organizational structure of our country.

Therefore the National Plan will:

- Refer constantly to the overall environment on the health and social policies, education, etc.:
- Provide for balanced action in support of all parties involved in the policies of the Plan
- Respect the principle of equity as defined and guaranteed to all citizens
- Include economic resources devoted to ensure the ability to implement what expected

4. **Use** technological solutions to support the sharing of clinical information (for example: telemedicine), in order to reduce patient mobility while making available the expertise at the local level (decentralised care centres).

5. **Provide** the testing and enforcement of new administrative instruments to detect and quantify the cost-benefit of the health professional long distance consulting.

6. **Make sure** that innovation requested by RD patients' healthcare is always within a context of security and of proven efficacy, in the protection of patients.

7. **Provide** a guarantee fund of the current and incremental expenses, linked to the provision of very expensive treatments and diagnosis, avoiding the risk that this fund falls within the set of fuzzy health care expense and its constraints.

8. **Use** the regional monitoring systems in an integrated way (including regional Registers that have the function to support the provision of care) and the national ones (involving the national Register) as fact-finding elements to direct government policies and actions and of evaluation of the system.

9. **Address** aspects of care, innovation, research and organization for the RD following a two-pronged approach that considers both the dimension of the disease and the health-social care needs in the following way:

- The individual patient care will basically focus on the profile of needs, which may be common in patients with different diseases and different in patients with the same disease

- The organization, the reference centres, the innovation and the research content, should be aggregated around themes relevant to both dimensions of the disease and care needs

<u>Theme 2 – Definition, codification and inventory of rare disease</u>

Under themes

1. Definition of Rare Disease
2. Classification and traceability of rare diseases in the national health system.
3. Inventories, registers and lists.

CODIFICATION

STATE OF THE ART

In Italy the official EU definition of rare disease is adopted: a clinical condition with a prevalence of no more than 5 per 10,000 people . In Italy the current codification of RD is ICD9 CM (DM 279/2001).

STRENGHTS

The National Health Institute (ISS) and Veneto Region take part in the European project in order to improve RD

codification and classification in ICD11. Furthermore the ISS is the coordinator of an Italian working group on the revision of ICD9 CM in collaboration with the European group. Some corporations use either ICD9 CM classification or ICD10 and OMIN (genetic classification).

CRITICAL ASPECTS

Different codifications are used: ICD9-CM for SDO (Hospital discharge records), and ICD10 for deaths (ISTAT).
The Orphanet ICD10 and OMIM are not yet used.

GOALS

It's necessary to introduce in Italy the most up-dated codification of R.D. in line with Europe.

RECOMMENDATION

- To keep in Italy the current definition of RD as suggested by Europlan and apply in the European Countries, the ICD disease codification and ICF functional assessment for RD

THE RARE DISEASE REGISTER

STATE OF THE ART

The Ministerial Decree (DM 279/2001) set up the NATIONAL REGISTER OF RARE DISEASES by the National Health Institute

The State-Regions Agreement of 2002 envisaged the establishment of regional or interregional registries, resulting from cooperation between regions, which serve to organise and assist its residents. These registers feed, through debt-defined information from a subsequent State-Regions Agreement of 2007, the National Register of Rare Diseases at the ISS

STRENGHTS

The Ministerial Decree DM 279/2001 is the only legislation in Europe with the attached list of RD .

All Regions have taken over this decree

CRITICAL ASPECTS

The implementation of the rules of the Ministerial Decree 279/2001 is not homogeneous on the national territory. Not yet been implemented.

- Monitoring of the codification system;
- Updating of RD (as required by rule every three years, although planned and implemented in the technical investigation) resulting in inequality between patients and underestimation of the epidemiological data of RD

GOALS

It is necessary:

- Make operating the RD Registers in all Regions according to the best models.

- Set up monitoring actions[1] of the Regional Registers and provide for sanctions against the Regions that don't apply the rule.
- Set up, throughout the National Territory, regional Boards for the monitoring of the registers and surveillance of the RDs.

2.1 Information and training

1. How to improve information on available care for RDs in general, for different audiences
2. How to improve access to quality information on RDs
3. How to ensure adequate training of healthcare professionals on RDs

INFORMATION

STATE OF THE ART

The Conference of State in July 2010 shows the information as the most critical point for Rare Diseases, although there are a lot of information and awareness initiatives promoted by different actors. Orphanet Italy is active, led by www.orpha.net.

The Health Ministry has opened a space for RD on its web site www.salute.gov.it and the National Health Institute has been appointed as technical body- National Centre for Rare Disease www.iss.it/cnmr, that issues the supplement of the News-letter "Rare disease and orphan medicines" and in 2008 started the toll free helpline for RD 800896949. The State Regions agreement State-Regions in 2007 gave to the Regions the task to inform either by telephone lines or by dedicated counters. Some regions have established information centres with specific RD help-line related and interacting with the network of regional / inter-regional Centre of reference, the network of local services and other institutions involved in the care of patients, and websites, also by searching for signs and symptoms in suspected MR. Since 2008 the "indispensible aiuto" has been established by UNIAMO (Italian Alliance for Rare Diseases) www.malatirari.it .There are many initiatives to raise awareness.[2].

STRENGTHS

The current initiatives in Italy guarantee a first level of information to public society, patients and health operators on RD and on different levels of the social health system (enclosure 3 with a map of the helps-lines).

CRITICAL ASPECTS

The information system on RD is not homogeneous on the national territory and not co-ordinated.
The quality of the information is not always validated.

[1] The Governance WG report that those actions have been activated

[2] Among the initiatives to raise awareness we underline the RDD organised by the associations of patients, coordinated by UNIAMO, throughout Italy with the support of Farmindustria ,the Italian Guidebook of RD Associations issued in 2006 and the following one issued in 2008/2009, with Orphanet/Farmindustria, and Uniamo, and the support of Farmindustria; - The yearbook ORPHANET-Italy on R.D. supported by Farmindustria; - Realization of two spots and a cartoon on R.D. by Farmindustria.

There has been little information initiatives aimed at the prevention and health promotion of people affected by RD.

GOALS
It is necessary to:

- Introduce minimal requirements for information and verification system;
- provide the training of the staff ;
- Make a periodic map of all available information sources in every Region;
- Identify quality control systems of the information in the web ;
- Give to the desired National Committee, as requested in the working group of the Governance, the coordination, evaluation of information needs for all subjects, the standardization of information, the monitoring and evaluation of the effectiveness of information services

TRAINING

STATE OF THE ART

In Italy the training is provided by the following:
- base training (University)
- master training (University, Regions)
- ECM (educational follow up of medicine) for all sanitary staff organised at national and regional level
- ECM for GPs, Paediatricians and specialists managed by region and professional associations

The State-Regions Agreement of July 2010 notes substantial difficulties for GPs / Paediatricians or specialist to interpret a complex symptom and to formulate a diagnostic suspicion, leading to delays in diagnosis and therapy and, consequently, a system should provide efficient support to physicians

STRENGHTS

- Existence of an organization and a system of training within which you can enter the specific knowledge of rare diseases.
- Tangible willingness to cooperate in the development of joint educational projects between associations, central and regional institutions, medical and pharmaceutical industry categories

Amongst the current initiatives, we can point out, the following ones as very good examples:

- The three-year training project “Knowing to care - Conoscere per assistere “aimed at GPs / Paediatricians, the result of joint planning between different actors (FIMG, FIMP, SIP, and SIMGePeD SIGU and supported by Farmindustria) focused on the suspected diagnosis, quality of care and transition from paediatric age to adult age
- The second level Master in Rare Diseases organised by the University of Turin;
- The project “Rare diseases from the monitoring to the training” (ISS CNMR-financed by the Ministry of Health)
- The project “Orphan drugs and the access to treatment for RD”;
- RDs are present in the pre-graduate training, post-graduate courses at the Optional Integrated Degree Course of Medicine and School of Specialization courses at the University of Padua

CRITICAL ASPECTS

There is limited interest on the specific issue of RD and this affects its non-inclusion in the curricula at all levels of training, including university education, despite some examples of good practice. In addition, GPs, Paediatricians as well as some specialists have little knowledge and competence in the RD.

GOALS

- To include the issue of RD as a matter of priority for the training of the Interregional Technical Board of Continuing Training and therefore include it in the Training plans of Local Health Agency (AA.SS.LL.), Hospitals, Research Institutes;
- To include training programs on RD at all levels of the system and for all health-social operators.
- To activate systems for evaluating the quality and effectiveness of training, with outcome indicators.
- To promote the training of RD patients and their representatives finalised to the acquisition of knowledge to participate in concertation tables
- It is desirable the participation of patients in the planning phase of information interventions on RD.

Theme 3 – Research on Rare Disease

Sub-Themes

1. Mapping of existing research resources, infrastructures and programmes for RDs
2. Needs and priorities for research in the field of RDs
3. Fostering interest and participation of national laboratories and researchers, patients and patient organisations in RD research projects
4. Sustainability of research on RD
5. EU collaboration on research on RD

STATE OF THE ART

The Italian researchers' ability to produce competitive scientific results on RD is attested by the number of publications and the values provided by bibliometric indicators. This result is all the more significant, if related to the limited availability of dedicated funds. In fact, according to a study of CERM (2009) Italy accounted for more than 10% of all scientific publications on the subject

STRENGHTS

There is a good attitude to research on RD.

There is a consolidated ability of networking at international level.

CRITICAL ASPECTS

- There is no guarantee of the continued commitment of public and private agencies in funding for research projects
- No guarantee on the timing of the funds;
- The results are not always evaluated according to the internationally shared criteria;
- Lack of a central system for connections, funding and performance monitoring

GOALS

- Promote multidisciplinary and translational research;
- All areas of research need attention, we don't identify any specific priority but it is necessary to strengthen the less developed areas (clinical, public health and social)
- Address the resources on objectives shared by the patients, centres of expertise and scientific excellences;
- Improve research on rare diseases also has important effects on the knowledge of common diseases;
- Develop strategies to disseminate the results and transfer them in clinical practice;
- Promote national and supranational aggregations

In order to promote research that can respond to the instances of RD patients in clinical, biomedical, public health and social research, the recommendations are as follows:

RECOMMENDATIONS

Bearing in mind the social dimension of the RD problem, it is necessary:
To bind some of the funds provided by the Ministry of Health, other relevant Ministries and the Regions to be allocated to this research and ensure the principle of subsidiarity of the State towards funding agencies and performers of scientific research
To promote a system of financial management to the RD fed constantly by punctual and clear funds, inspired by the best practices of research evaluation based on merit and supported by a system of ex post monitoring of results
To introduce a tax credit (automatic procedure) for investment in research on orphan drugs and RD
To map existing resources, infrastructure, and funded research projects dedicated to RD and strengthening them where necessary (to model and enhance the work of RD Platform);
To promote the creation of infrastructures (e.g. technology transfer offices, centres of clinical trials, tissue banks and biobanks) to facilitate technology transfer of research results;
To promote positive interaction for groups of diseases, through the support of associations, to coordinate activities and case studies,
To promote the development of a collaborative model between the main protagonists of the path of research on RD: patients, doctors, researchers, industry, public institutions and private research funding agencies;
To promote the scientific coordination between regions, in the interests of RD patients

Theme 4-Standards of care for RDs - Centres of Expertise (CoE)/ European Reference Networks (ERN)

Sub-Themes

1. Identification of national or regional CoE all through the national territory by 2013

2. Sustainability of CoE
3. Participation in ERN
4. How to shorten the route to diagnosis
5. How to offer suitable care and organise adequate healthcare pathways for RD patients
6. How to ensure in CoE multidisciplinary approaches and integration between medical and social levels
7. How to evaluate CoE

CENTRES OF EXPERTISE

Preliminary remarks

With reference to the features of a Centre of Expertise , the working group has confirmed what reported in the document drawn up in 2005 by RDTF, now EU CERD.

STATE OF THE ART

The Ministerial Decree 279/01 establishes the National network for rare diseases, formed by the seats identified by the Regions. The State-Regions agreement in 2002 decrees the creation of a Coordination Centre (see enclosures n. 1 and 2).

STRENGTHS

- Experience gained by certain Regions in defining the Reference Centres and based on objective parameters.

CRITICAL ASPECTS

- The reference centres not always are in compliance with the definition provided by RDTF Centre of Expertise
- Heterogeneity with respect to the territory
- Shortage of information regarding the access

GOALS

To ensure the Centres of Expertise (CeO) be formed by experts in the specific RD or groups of RD, as for the clinical and research aspects, it is advisable to identify and use a series of common parameters shared by the different stakeholders, among them:
- the appropriate structural facilities, the adequate equipments and human resources,
- the use of information systems for the patients registration and for the management of a care path
- the promotion and participation of the specialists in working groups on national and international research projects,
- promotion and systematic participation in dedicated training events,
- creation of a transitional path from the paediatric age to the adult age,

- performance validation by the patients,
- scientific publications on index-linked journals.

In compliance with the RDTF indications, **to appoint a CoE (Expertise Centre)** it is necessary:

- promoting the accreditation process for the Expertise Centres (as defined by the RDTF), through the definition of shared criteria among the different stakeholders;
- a periodic CoE assessment based on clinic results and patient satisfaction using criteria to be shared with the patients associations;
- Promoting the CoEs by homogeneous groups of diseases. For specific pathologies with peculiar features, regarding for example a particular rarity, complexity or concerning the specific care, the CoEs can rely upon the competences of high specialised centres, also through telemedicine/tele consulting;
- the over-regional or regional distribution according to the pathology predominant incidence;
- facilitating the CoEs access to the rare disease patient;
- the use of information platforms to functionally connect the centres with the other structures and services involved in the continuity care of the patients;

The structures suitable to become CoE are those in possession of the requirements mentioned in the preliminary remarks and capable of satisfying the patient's needs widening the assessment of the functional areas (functional assessment) and the transfer to the territory. Taking care of rare disease patients is a complex activity involving both horizontal and vertical networks. It is correct to create CoEs by homogeneous groups of rare diseases in order to guarantee a differential diagnosis and the global management of the health care.

Recommendations
Having considered the situation of our Country, it is not advisable to create new structures but, rather, to identify new Expertise Centres within the existing networks and to arrange them rationally in order to improve and optimise the existing resources

It is essential to create accredited and monitored CoEs for the correct equipment and human resources allocation, taking into account their attraction capacity in order to guarantee their activity over time.

It is recommended the activation of multidisciplinary teams, if possible within the same CoEs, through proper methods and adequate financing and incentives.

PARTICIPATION IN THE EUROPEAN REFERENCE NETWORKS

STATE OF THE ART

National and European networks are already existing.

STRENGTHS

The present experiences gained by the networks are considered real strengths.

CRITICAL ASPECTS

Nowadays just few networks are active; most of them are focused on the basic biological research.

So far only one-time financing has been allocated.

GOALS

It is deemed necessary to:

- Promote the participation to the European networks as indicator of the CoE activity;
- favour the official acknowledgment of the existing networks by the single Member States (for example IPINET for Primary Immune Deficiencies in Italy) and the sharing of a platform for network creation, assessment, financing and dissemination;
- foster the use of the best technological solutions existing in order to forward patients' clinical data and biological samples and not the same patients

In order to create a proactive cooperation among national or international **experts**, it also to be hoped:
- the activation, by the European and International scientific societies, of forums and coordinated initiatives on the rare diseases; the collection and optimization of information exchanges on occasion of already planned congresses and international meetings.

SPEEDING UP THE ACCESS TO DIAGNOSIS

STATE OF THE ART

There is census of the diagnostic laboratories by S.I.G.U. Italian Genetic Society, completely acknowledged by Orphanet Italy. Orphanet (in cooperation with Eurogentest) carried out a census on a series of Italian diagnostic laboratories dealing with RD.

STRENGTHS

The experiences gained in some territories (registries).

CRITICAL ASPECTS

- Lack of knowledge coordination and exchange of among the different ties of the service networks;
- Lack of homogeneity throughout the territory;
- Too many rare disease patients have to transfer for diagnosis and treatment.

GOALS

It is deemed necessary to disseminate knowledge and competences through:
- information dissemination on the existing networks also thanks to specific validated websites and databases (e.g. Ipinet);
- training and information of GPs, Paediatrician and National Health Services specialists;
- connection among the network ties of also through information tools (e.g. electronic health file) in order to reduce the patient mobility and to improve the intervention integration;

- Use and widening of the existing databases (Orphanet);
To map, connect on the network and support the national laboratories it is advisable to:
-promote, in agreement with and through the support of the Regions, additional national census of laboratories and tests carried out with a special attention to the genetic ones. The participation to the census shall be mandatory and the results shall be validated at national level by the local institutions (Regions, Ministry of Health and High Health Institute);
- maintain and network new laboratories through the existing databases, especially Orphanet;
- carry out a rationalization of the applicable tests thus supporting the laboratories offering tests for ultra rare diseases and favouring economies of scale.

SCREENING AND GENETIC TESTS

STATE OF THE ART

Within the national framework there are consolidated procedures to send biological and genetic samples. There are also the Guidelines 2008 concerning the wider screening (developed by Società Italiana Studio Malattie Metaboliche Ereditarie (Italian Society for the Study of Hereditary Metabolic Diseases) and by the Società Italiana Screening Neonatali (Italian Society for the Neonatal Screening)).

STRENGTHS

- The Law 104 dated 5th February 1992 has introduced, on the national territory, the mandatory screening for three pathologies: phenilchetonuria, cystic fibrosis and congenital hypothyroidism.

GOALS

- With reference to the "Wider Screening", it is advisable that every single region indicate the different methods used to implement the "widened screening" (with the BP format).
To support the development of European guidelines on the diagnostic tests and to foster the population screening, the dissemination and implementation of the diagnostic test guidelines already existing at European level (in particular in Eurogentest) is strongly suggested.

It is necessary a dedicated funding to develop the guidelines for specific diseases.

At European level it would be advisable to define a common price list for sample costs, transportation and acceptance.

CONNECTION AMONG COES AND SOCIO-HEALTH CARE AT LOCAL LEVEL

CRITICAL ASPECTS

Lack of coordination regarding the interventions.
Insufficient comprehensive patient care at local level.
The social dimension is not contemplated.

GOALS

It is deemed necessary to:

- Promote a connection among CoEs network, hospitals, primary care network, rehabilitation services and the urgency-emergency networks;
- Develop and promote the electronic health file;
- Favour multidimensional and multi-professional assessment processes concerning both the health care and the social aspects (such as examples of good practices already applied, see enclosure 5[3]).

4.1. Orphan Drugs

8. Future of OD
9. Access of RD patients to orphan drugs Pricing and Reimbursement
10. Compassionate use and temporary approval of orphan drugs. Off label use

STATE OF THE ART

Since the prices in Italy are among the lowest in Europe, often the pharmaceutical companies, following up the marketing authorization by EMA, prefer negotiating the selling price in other European countries first and then in Italy. One of the critical points for the market availability of orphan drugs, is the long time that elapses from when the company required to AIFA (Italian Pharmaceutical Agency) pricing and reimbursement and the closing of the negotiation. AIFA has developed a new mechanism for price negotiation, based on the increased number of patients actually treated compared to those assumed to be treated. This in order to reduce and facilitate the access to particularly costly drugs destined to a few number of people.

Pricing and reimbursement are National prerogatives while **the definition of the Therapeutic Plan (note State-Regions Conference) falls within the specific competence of the CoEs.** The provision and administration of treatment can also be provided by the structures in the area of residence of the patient, in consultation with the Reference Centre prescribing

The Law 326/2003 provides that the 50% resources merged into the fund (5% of the promotional expenses of the Pharmaceutical **Industries**) shall be destined to the Agency (AIFA) for the Creation of a National Fund for the use of ODs or drugs for RD, paid by the National Health Service. Such drugs, while waiting for the marketing, represent a hope for treatment for particularly severe pathologies. There is a codified path to access this Fund.

Italy has already developed a conditioned pay-back consisting of three items: cost sharing, risk sharing, and payment by result[4].

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[3] Enclosure 5, requires for best practices in the different Regions: Rare Diseases Centre for Trentino Region, PDTA for Lombardia region, and the on-line health file for the Veneto region…

4 a) Cost Sharing: discount on the price of the first cycles extended to all the eligible patients. The drug is administered for the first therapeutic cycles after which the clinical efficacy is assessed. For respondent patients the treatment continues, while for non -respondent patients the treatment is suspended; b) Risk Sharing: discount on the price of the first cycles for non respondent patients at first assessment. After the initial therapeutic cycle a post treatment assessment is carried out. For the respondent patients the treatment continues and will be refunded by the National Health Service (SSN) while for the non-respondent patients the treatment will be suspended and the AIC holder shall pay part of the therapy; c) Payment by results: total refund by the pharmaceutical company for the first cycles of non respondent patient at the first reassessment. After the initial therapeutic cycles a post treatment assessment is carried out. For respondent patients the treatment continues and will be paid-back by the SSN, while for the non-respondent patient the treatment will be suspended with pay-back for the non effective treatment.

In Italy the global compassionate use is ruled by the Ministerial Decree dated 8th May 2003. In addition, the national legislation, L. 648, gives the possibility to use, at the National Health System (SSN) expense

- drugs with non registered indications,
- drugs marketed in other States but not in Italy,
- Drugs under experimentation that may be used outside the experimentation if there isn't a valid therapeutic alternative.

It is then possible to enforce the same law to prescribe off-label drugs. The off-label drugs prescription for the single patient is ruled, at National level, by the Law 94/98. Under the direct responsibility of the physician it is possible to prescribe the drug. The prescription is supported by scientific evidence on effectiveness and safety and it is necessary the patient's informed consent.

The list of drugs contemplated by the Law 648 and allocable for the total account of the National Health System is published by AIFA and periodically integrated.

At present the OD National Registry, managed by the National Health Institute (Istituto Superiore di Sanità), does not permit to trace back the number of patients under treatment.

In Italy there are three main phases between the marketing authorization by EMA and the final availability of the orphan drug on the national market.

1. Time elapsing between EMA authorization and the presentation of pricing and reimbursement request to AIFA(Italian Pharmaceutical Agency)
2. time elapsing between such request and the definition of price and reimbursement class
3. Drug availability on the market.

There may also be different times of access to care available to patients in the different regions, since regions can evaluate whether to include an orphan drug within its Therapeutic Regional Guides (ProntuariTerapeuticiRegionali) and Therapeutic Hospital Regional Guides(ProntuariTerapeuticiOspedalieriRegionali) .

In Italy, at present, 42 out of the 62 orphan drugs approved with centralised procedure by EMA (data updated on October 2010) are available on the market and their cost, for the therapeutic indication, is fully paid by the national Health System. Of the remaining 20 orphan drugs approved by EMA, 10 have a pending request at AIFA (Agenzia Italiana del Farmaco) by the companies in charge of pricing and reimbursement. and their assessment is ongoing. As for the remaining 10 orphan drugs, the pharmaceutical companies didn't ask for the marketing authorization in Italy.

STRENGTHS

- In Italy, orphan drugs and other innovative treatments for the treatment of RD patients are guaranteed because included in the National LEA (Essential Assistance Levels). All the orphan drugs currently marketed in Italy are completely refunded by the National Health System (SSN).

- Regulation for the compassionate use, as provided by for Ministerial Decree dated 8th May 2003.

- So far all the Regions have guaranteed their RD patients extra-LEA treatments via formal and informal integration procedures to the National LEA.

- In Italy there is a good availability of orphan drugs, 42 of the 62 orphan drugs approved by EMA are marketed in our country. AIFA also contributes through various means to further improve the accessibility and availability of drugs intended to treat rare diseases: providing, at total charge of the National Health Service, the drugs included in the Law 648/96 and off-label drugs with established use ,based on the data of the scientific literature, including the extension of permitted claims, the financing of independent research, the application of reimbursement models of conditioning eligibility for potentially innovative drugs

- Protocols for the treatments of rare disease patients have already been developed also within the framework of an interregional cooperation concerning the orphan drugs, the creation of dedicated regional databases, and the development of extra-LEA treatments essential for the patients and granted for free by some Regions (using their own funds).

CRITICAL ASPECTS

- Long waiting times to access the treatment, due to the methods to apply for pricing, reimbursement and marketing authorization.
- The Regions may decide to delay the marketing of an orphan drug included in the PTOR, because of budgets limits or for different reasons regarding the health policies thus generating a mismatch in the drugs availability among patients in the different Regions.
- Difficulties to access the AIFA fund, as provided for by art. 48, Law 326/2003. Such difficulty is probably due to the lack of information about the access to the above-mentioned fund.
- Need of price review processes as part of the post marketing surveillance in case of enlargement of the indication of the actual clinical use of the orphan drug
- At present the OD National Register does not permit to trace back the number of patients under treatment in the Italian Reference Centres prescribing and dispensing such drugs. These data are however available through the regional system of monitoring of the pharmaceutical output, it is to define a specific information debt between regional and national levels, as occurred with the National Register
- It is deemed necessary to define that the CoEs, identified by the Regions, are entitled to the monitoring and prescription of ODs. Moreover, according to the European definition, they are committed to develop a therapeutic-care plan.

GOALS

It has been pointed out:
- the need for a new negotiating system regarding the OD price on the basis of the number of patients treated (see AIFA State of the Art)
- the importance of a post marketing monitoring system to assess the real orphan drug clinical efficacy and safety and to carry out a post marketing cost adjustment based on what actually allocated by the Regions.
- the need to standardise prescription, allocation and administration methods among the different Regions. Such goal can be achieved through discussion on occasion of interregional technical meetings on RD and through the possible involvement of AIFA.
- the need to improve the access methods to the AIFA funds as provided by for art.48 of the Law 326/2003.
- the need to develop a method for the creation of therapeutic protocols by groups of rare pathologies also within new territorial, regional and interregional contexts; the development of a new context in order to broaden the benefits provided by law, the identification of the essential and indispensable treatments for rare diseases and/or their complications including not only galenical preparations, nutritional supplements and health products in general but also dermatological products, medical products, etc. Such activities are

aimed at guaranteeing a uniform access to the currently available treatments by those patients resident in different surrounding areas.

Topic 5 – Patient and social services empowerment

Sub-Themes

1. Involvement of patients and their representatives in decision-making processes in the field of RDs
2. Support to the activities performed by patient organisations
3. Specialised social services: Respite Care Services; Therapeutic Recreational Programmes; Services aimed at the integration of patients in daily life
4. Help Lines

STATE OF THE ART

In the National Health Plan 2010 – 2012 the patient's empowerment[5] is considered only for the control of the services allocation (customer satisfaction and civic audit), while the National Health Plan 2006 – 2008 was much richer in contents, with implications leading to a wider and longer strategic vision.
In certain Regions the implementation resolution for the Establishment of the Rare Diseases Coordination also provides patients and operators information and training.
In some Regions operative round tables for RDs have been created. They involve all the stakeholders, patients included.
In order to support the activities of the patients' organizations, state and regional calls for funding and tax reliefs have been proposed
The Law 266/2001 provides for the creation of service centres (CESV) to coordinate and facilitate the voluntary organizations activities.
In some Regions a "special" rehabilitation for RD, regardless of the standard consulting activities, has been developed. In Trentino Alto-Adige, a region with a particular form of autonomy under special statute, it has been created the socio-health director profile.
There are numerous help lines centres, also supported by the institutions.

STRENGTHS

Some first and second level associations and institutional agencies are committed in different empowerment initiatives (see enclosure 6[6]). As examples of GOOD PRACTICES it is useful to remember:

- Informed consent governance, participating model of good clinical practice, Fondazione IRCCS Ca' Granda;
- "Dado Magico" initiatives: training course on the patients' empowerment on the institutional languages, UNIAMO FIMR;
- "Momo initiative: the empowerment makes the difference", UNIAMO FIMR;
- The initiative "empowerment for caregivers "- Prader-Willi;

5 The expression patient empowerment is often used but actually the empowerment is a continuous improving process involving all the stakeholders: each one contributes with his/her personal expertise, knowledge and profile in order to share knowledge, decisions and a new competence (created together) to meet the challenges of the global care.
5 National examples of the Regional coordination centres, Telethon – Bacheca delle Associazioni, TVMR, POLO PROVINCIALE PER LE MALATTIE RARE, etc.

- Progetto "Il Codice di Atlantide: promoting a RD research culture" empowerment on research, selection and financing processes , UNIAMO FIMR ;
- Also in Italy there is a good practice for the recreational therapeutic care carried out by Dynamo Camp in Tuscany.

CRITICAL ASPECTS

- Lack of patient involvement in the pathways he/she is part of, from the action planning to its implementation;
- The patient is not considered an actor within the framework of a participatory medicine (usually he is the object and not the subject).
- No mapping of needs, especially the cross-cutting ones.
- The local projects, also the praiseworthy ones, are fragmented, left at the good will of the people involved, not structured and not in network.
- Reduction of funds devoted to social and socio-health initiatives
- There aren't health care services suitable to face the functional complexity both for patients in severe conditions and for those with medium-mild disabilities and in general for the most vulnerable social categories affected by RDs (complexity within the complexity).
- No family assistance, including psychological assistance.
- Waiting lists for habilitation and rehabilitation are very long (up to years).
- The habilitation therapy for the pathologies presenting mental problems is missing or insufficient.
- No transfer of expertise to the personnel dedicated to the socio-health assistance.
- Need to include the RD experts in the medical-legal Commissions.

GOALS

Assigning to the assumed National Committee, proposed by the Governance WG, the identification of shared criteria for the social cooperative operators' training.

As for the support to the patient organizations' activities it is advisable:

Information guidance on research projects, associations and the expert centres;

- A logistic and information support through preferential term (e.g.: granting of premises, computers, etc.).

As for the patient/association empowerment it is advisable:

- Including the patient and/or his/her family members in all the decision-making processes with the sharing of the different languages;

• The recognition of training undertaken by the patient and / or his family, with different instruments (ECM, reimbursement of expenses - per diem? Work permits ad hoc)

• The recognition of the fact that education leads to virtuous circles that fall in cascade, as well as on patients and associations belonging to the entire system

- Promoting memoranda of understanding/ projects with a pool of patients/associations representing common interests.

As for the specialised care services, it is desirable:

- The systematic identification of needs through a peer to peer discussion;
- Shared and multidisciplinary care (socio-health);

- The patient participation in the control and accreditation process of private and contracted social services.

As for the Help-lines, it would be advisable:

- The creation of regional free-toll numbers linked to the national and European free-toll numbers;
- The monitoring of the information needs as requirements for the creation of a proper help-line.

Horizontal topics

Topic 6 – Sustainability

National framework

From the PSN[7]National Health Plan analysis emerges that the central administration commitment to support the RD initiatives has been constant and explicit, with several decision-making process levels and different programmatic lines developed over the last ten years.

Nevertheless there has never been a clear indication regarding the central administration policy on RD system sustainability[8]. From analysis of the National Health Plan (PSN) is evident the intention and the will of the public administration to give a proper financial support to rare diseases without any further specification. It is important to underline that Health National Plan, due to its own nature, can't fix the regulations in detail nor planning the single expense items because such task falls within the regional authorities[9]'competences[10].

Presently the main financing sources for rare diseases are the followings:

- *ISS National Health Institute – supports the activities of the National centre for rare diseases established at the ISS and of the national council (Consulta). The first of these bodies is a research, consulting and documentation agency for rare diseases and orphan drugs, aimed at creating RD prevention, treatment and surveillance and in charge of the coordination of regional activities and registers. The council is the body in charge of the relationships with the associations. Its task is strengthening relations and synergies among the organizations safeguarding the rare disease network in our Country, strongly believing that their role is fundamental to give guidance to the*

[7] See enclosures n. 1 and 2

[8] The concept of sustainability entails different dimensions, the economical, social, ethical, cultural, etc. For this reason the sustainability assessment of an action or a project primarily implies finding a balance among all these dimensions. Such balance refers to the system of values of a community generating the perceived priorities.

[9] Regional context**.** Following then with the Analysis of the last Regional (Social) Health Plans enforced, it is observed, with different aspects, the detailed analysis concerning the areas of interventions but none of them, with regard to four Italian regions (Lombardia, Emilia-Romagna, Veneto and Toscana) contains a clear and analytic indication as for the rare disease system financing mechanisms. Also in this case it is to be pointed out that such document are health programming documents for which (in most of the cases) special budgets are fixed for every the single element identified.

[10] Regional context**.** Following then with the Analysis of the last Regional (Social) Health Plans enforced, it is observed, with different aspects, the detailed analysis concerning the areas of interventions but none of them, with regard to four Italian regions (Lombardia, Emilia-Romagna, Veneto and Toscana) contains a clear and analytic indication as for the rare disease system financing mechanisms. Also in this case it is to be pointed out that such document are health programming documents for which (in most of the cases) special budgets are fixed for every the single element identified.

patients and their families within the National Health Service and to identify priorities for the public policies agenda. The Council only has an advisory function and works without portfolio;

- *The Ministry of Health/of University and Research – allocates funding concerning the current research and aimed at supporting the research activity that, directly or indirectly, in some special cases, involves also the rare disease field;*
- *State/Region Conference – has been involved in the rare disease and signed:*
    - *The agreement among Government, Regions and the special statute Provinces of Trento and Bolzano regarding the proposal advanced by the Ministry of Labour, Health and Social Policy for an health research announcement for 2008 destined to rare disease reaching a total amount of € 8.000.000 (of which € 5.000.000 as provided for by art. 1, par. 813 Financial Law 2007,€ 3.000.000 coming from the AIFA fund, Legislative Decree n. 269 dated 30/9/2003 converted into the Law 24/11/2003 n . 236 Art. 48 par. 19 letter b);*
    - *The agreement among Government, Regions and special statute Provinces of Trento and Bolzano regarding the proposal advanced by the Ministry of Labour, Health and Social Policy pf guidelines for the correct use of the bound resources by the special stature Region and Provinces as provided by for art. 1, par. 34 and 34bis, Law dated 23th December 1996, n. 662, in order to implement the primary and nationally important objectives for the year 2010. In particular "Rare Diseases: for the year 2010, 20 million Euros has been introduced";*
- *The Ministry of Labour and Social Policy – directly or indirectly, supports rare diseases by allocating funds to rare disease associations, according to project-based announcements, thus backing social promotion (APS) or voluntary organizations (law 383/2000, art. 12, par. 3);*
- *The regional autonomies – through its own funds destined to the expenses and budget items for the social and socio-health activities considered top priorities of Regional Socio-Health Plans for a specific period;*
- *Other funds come from the activities supporting the research carried out by the specific associations within the pharmacology field rather than fund raising activities carried out by the rare disease patients associations at public and/or private agencies, from legacies and donations by legal or natural persons* and from gifts collected during the campaign to raise public awareness.

Summary

Based on the Council Recommendations on the sustainability of the rare disease system, it is possible to state that nowadays the sustainability topic is widespread and there is a high level of awareness at national, regional and local level. Presently, given the implementation of a federal model within the health field, the ability of funding rare disease initiatives with an effective impact in the patients' daily life, is referred to the Regions as body enforcing its own and over commissioned guidelines.

Within this framework it is useful to observe that so far the main sources of financing for rare disease are public (structural financing). Most of the times the sources of financing resulting from the private participation, important but residual with respect to the global scope, are destined to specific investments (ad hoc financings).

Within the present context it is observed a very poor use of the financing sources, according to auxiliary logics capable of actively involving and investing with responsibility the knowledge and relational heritage of all the network stakeholders. This is probably imputable to the lack of a stable comparison and of a systematic coordination of the RD financing system involving all the network actors. From this viewpoint, it is possible to stress that this project has fostered a careful consideration for the mutual knowledge of the people involved in the network.

GOALS

As for the rare disease sustainability, the new horizon for the near future shall be closely connected to the creation of a social value. It is necessary to plan, project, implement, monitor and assess policies that

permit a clear definition of tasks, assessment criteria and consequent responsibilities[11]. Focusing the attention on such element means acknowledging of the global need of rare disease patient and developing an integrated approach that, in terms of sustainability, (in its widest meaning), may guarantee and added value both for the individual and the Society.

With respect to priorities regarding system sustainability and financing, the need for a future plan and a better coordination of the sources (at least in the public context) is evident; regarding the RD field in order to develop an homogeneous and coordinated policy, regarding RD priority policies (expertise centres, socio-health networks, research, registries, orphan drugs, etc.). A technical economic coordination to support the policies implementation, priority investment guidelines, identification of the proper periods to develop investments and financing policies at every institutional level is essential. Such context should represent all the people involved in the rare disease system and should be on equal terms, advisory and proactive. It is desirable that the shared and financed contexts and projects be transversally systematic with the focus on the rare disease patient in order to generate an immediate and desirable organizational and health care effect to give real assistance and economic and social benefits to the rare disease patients.

Taking into account the above considerations, it is manifest the need to consider the sustainability not only economically but also socially. A pilot study on a sample of 392 families belonging to 10 Rare Disease Associations – conducted for the first time by the Istituto per gli Affari Sociali (IAS), in cooperation with Federazione Italiana Malattie Rare Uniamo FIMR Onlus, Orphanet-Italia and Farmindustria shows that the economic sustainability is a important problem but also the social and environmental sustainability are priority aspects for the RD patient. The social cost is considerably more significant than the "actual" costs supported to assist such patients. It is necessary to create a synergy and the socio-health conditions to sensibly improve the life quality of a rare disease patient.

Consequently it is possible to opt for the proper use of the available financing in the RD field by accepting, and supporting the request of the reference stakeholder as aware carrier of assistential needs.

It is also important to guarantee the proper resources to support long-term shared investments, (long-term sustainability) to implement structured development policies on the most important topics among the rare disease stakeholder. The idea is sharing a long-term development strategy without meeting the contingent or regular needs but programming local investments to cover the present expenses thus creating a progressive, structured and synergic rare disease system.

It is advisable to define a structured fund to support RD initiatives and policies with dedicated expenses items (registers, expertise centres, training, information, orphan drugs, etc.).

It is also desirable to develop a financing system for RD to enhance the importance of virtuous partnerships among the public administration and the private social sector in order to support agreed elements of the system. It is useful to consider an economic/fiscal system to provide incentives to the private system committed in the direct and indirect support to RD, in particular in the research and rare disease associations filed.

Much has been done, with more or less awareness, but much could be done by recognizing the social values of the already existing initiatives, by supporting the knowledge heritage developed by rare disease system stakeholders in synergy with the system actors.

Issue 7 – Gathering expertise at European level

CENTRES OF EXPERTISE IN OUR COUNTRY AND WITH THE OTHER COUNTRIES

[11] The sustainability assessment depends on the moment in which the possible benefits manifest with respect to the moment in which the financial and social costs must be paid. The present situation is always considered more than the future events.

STATE OF ART

Multidisciplinarity is not only the expression of different expertise and points of view, but also the integration of different organisational, economic and ethical dimensions. With respect to this last point, the integration of the point of view and values of the patient and their family members are important.

Centres of reference mainly refer to scientific literature and scientific evidence (EBM), guidelines and recommendations.

In Italy there is the National Guideline System (*Sistema Nazionale Linee Guida* or SNLG) co-ordinated by the *Istituto Superiore di Sanità* - National Institute of Health and the *Agenzia per i Servizi Sanitari Regionali* - Office for Regional Health Services and within which some guidelines, consensus, etc. have been carried out (www.snlg-iss. en).

Using a much needed clarification that highlights the difference between guidelines and recommendations as a starting point, in Italy the situation is the following:

- the CNMR(National Centre for RD) of the I.S.S. is engaged in researching, documenting and studying the implementation of new guidelines for rare diseases using the DELPHI method in collaboration with the SNLG, other research national and international organisations and several structures within the *Sistema Sanitario Nazionale* - National Health System (university hospitals, polyclinics, IRCCS, etc.), patient associates.
- To date, three guidelines have been made (Alternating Hemiplegia, Down Syndrome, considered a rare disease in Italy), one is currently being published (hereditary epidermolysis bullosa) and three are being developed (aniridia, hereditary multiple exostosis and tuberous sclerosis).

In various regions, diagnostic and therapeutic approaches have been defined to which it was believed wise to add aspects of assistance (PDTA). This activity is recognised as one of the tasks of regional co-ordination and is being undertaken in collaboration with inter-regional reference centres. In some instances these protocols have been the basis for specific regulatory actions for the recognition of enforceable patient rights in addition to the LEA (e.g., pharmaceuticals, parapharmaceuticals, dietary, etc.).

GOALS

The question that remains to be answered is how to get to provide concrete responses for all rare diseases that are awaiting a definite diagnosis as well as a fair and consistent treatment, regardless of where it is provided.

The mechanism that ensures the exchange of expertise is knowledge sharing through networks for diseases that can look to the network of associations in Europe and beyond for inspiration and should become the benchmark for the world of science that deals with that disease.

It would be useful to include some key guidelines that will help the doctor move towards the suspected diagnosis of a rare disease in information about patients held in computer databases used by general practitioners and paediatricians in their offices, including through the National Network of rare diseases.

Conclusion of the Final Report

In addition to the reports on each issue the Final Report includes:

- A general evaluation of the usefulness of EUROPLAN recommendations and EUROPLAN indicators for the furthering of a national strategy in the country;
- The transferability of EUROPLAN recommendations in Italy especially in the context of "sustainable governance".

Attachments

Attachments 1 and 2 - Legislation
Attachment 3 - Help Lines

III. Document Details

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