EUROPLAN
European Project for Rare Diseases National Plans Development
Coordinated by the Italian National Centre for Rare Diseases
Italian National Institute of Health
2012-2015

ALAN asbl

EURORDIS
Rare Diseases Europe

LUXEMBOURG

EUROPLAN NATIONAL CONFERENCE

FINAL REPORT

19-20 November 2013, Luxembourg

EUCERD
Joint Action

FOREWORD

The EUROPLAN National conferences are aimed at fostering the development of a comprehensive National Plan or Strategy 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 are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS – the European Organisation for Rare Diseases. For this purpose, EURORDIS nominated 10 EURORDIS-EUROPLAN Advisors - all being from a National Alliance - specifically in charge of advising two to three National Alliances.

EUROPLAN National conferences share the same philosophy, objectives, format and content guidelines. They involve all stakeholders relevant for developing a plan/strategy for rare diseases. According to the national situation of each country and its most pressing needs, the content can be adjusted.

During the period 2008-2011, a first set of 15 EUROPLAN National Conferences were organised within the European project EUROPLAN. Following the success of these conferences, a second round of up to 24 EUROPLAN National Conferences is taking place in the broader context of the Joint Action of the European Committee of Experts on Rare Diseases (EUCERD) over the period March 2012 until August 2015.

The EUROPLAN National Conferences present the European rare disease policies as well as the EUCERD Recommendations adopted between 2010 and 2013. They are organised around common themes based on the Recommendation of the Council of the European Union on an action in the field of rare diseases:

1. Methodology and Governance of a National Plan;

2. Definition, codification and inventorying of RD; Information and Training;

3. Research on RD;

4. Care - Centres of Expertise / European Reference Networks/Cross Border Health Care;

5. Orphan Drugs;

6. Social Services for RD.

The themes "Patient Empowerment", "Gathering expertise at the European level" and "Sustainability" are transversal along the conference.

I. GENERAL INFORMATION

Country	**Luxembourg**
Date&Place of the National conference Plenary Session Workshops	19th November 2013 : Mamerschlass – MAMER 20th November 2013 : Maison d'Accueil des Soeurs Franciscaines – LUXEMBOURG
Organiser	ALAN (Association Luxembourgeoise d'aide pour les personnes Atteintes de maladies Neuro musculaires et de maladies rares)
Website	www.alan.lu
Members of the Steering Committee	• Dr Anna Chioti- CRP- CIEC Head of Unit • Gaby Damjanovic – Past President ALAN • Mrs Gwenn Crohin- Social worker- ALAN • Mrs Gaby Turmes-Assistant Manager ALAN • Mrs Shirley Feider – President ALAN • Dr Jos Even - Vice President ALAN • Prof. Karin Dahan - LNS – Head of Genetic Counselling Unit • Dr Marguerite Leches - CHL Reeducation and Rehabilitation • Dr Yolande Wagener - Ministry of Health – Head of Division • Dr Michel Hoffmann - Neurologist ChDN • Dr Fernand Pauly – CHL Pediatry and Reeducation and Rehabilitation
European Commission – **DG for Health and Consumers**	Michael Hübel – Head of Unit "Programme Management and Diseases" Antoni Montserrat Policy Officer for Cancer and Rare Diseases Jaroslaw Waligora Policy Officer for Rare Diseases
Eurordis – Europlan Advisor	Lily Cannon
Eurordis	Yann Le Cam - Chief Executive Officer Valentina Bottarelli – EU Public Affairs Senior Advisor Flaminia Macchia - EU Public Affairs Director Ariane Weinmann - EU Public Affairs Manager

I. GENERAL INFORMATION, cont.

National Centre for Rare Diseases ISS (Italy)	Dr Domenica Taruscio – Director
Eucerd Joint Action coordinating team	Prof. Kate Bushby, Institute of Genetic Medecine – Newcastle; Vice-Chair EUCERD and leader of the EUCERD Joint Action Dr Antonio Atalaia - Neurologist Institute of Genetic Medecine - Newcastle
Name and List of Workshops	
Workshop A	Health care systems supporting people with rare diseases (social security benefits concerning diagnosis, genetic testing, treatments/orphan drugs)
Workshop B	Creation of a rare diseases' platform
Workshop C	Creation of a registry – codification – research
Workshop D	Social services for rare diseases
Chairs and rapporteurs of workshops	
Workshop A	Prof. Karin Dahan – Dr Yolande Wagener
Workshop B	Dr Michel Hoffmann – Dr Fernand Pauly
Workshop C	Dr Anna Chioti – Dr Jos Even
Workshop D	Mrs Gwenn Crohin – Dr Marguerite Leches
Annexes	- Programme - List of participants in the workshops

II. INTRODUCTION

The EUROPLAN conference in Luxembourg was organised by ALAN, with the support of the Ministry of Health and Social Security and under the auspices of the Ministry of Family and Integration and the Ministry of Higher Education and Research.

Plenary session - 19th November 2013 -

The conference started with a plenary session which brought together 140 people: medical professionals and organisations, representatives of all ministries and national institutions working in the field of rare diseases, laboratories, social organisations, associations for rare diseases and patients themselves.

Gilles Roth, the Mayor of Mamer, where ALAN has its seat welcomed the participants. He explained the social engagement of the town of Mamer where not only ALAN but also other associations have their seat: an institution for mentally disabled people (Ligue HMC), the autism foundation and SIPO which takes care of children with early development problems. He paid tribute to all the people who defend patients' right and look after them: the patient' associations and all the volunteers.

The conference was then introduced by ALAN's President, Shirley Feider. After acknowledging and thanking the participants, she explained the change of context since ALAN and EURORDIS took charge of the organisation of EUROPLAN conference. She briefly described the current political situation; Luxembourg being in the process of anticipated election which would imply changes at the government and ministries level. She reconfirmed the EUROPLAN conference objectives which remained the same; carrying the hopes and expectations of all the patients, their family, the voluntary workers, and the community of all the professionals involved in the field of rare diseases. She underlined one of the expected outcomes of the conference in the short term: the development of a National Plan on Rare Diseases in Luxembourg.

During the introduction session, the mother of a 20 years old young man with Leigh syndrome and a man living with Marfan syndrome, shared their experiences. They showed appreciation for some aspects of the healthcare system of Luxembourg and supervision. However these 2 testimonies pointed out the complexity of the situation that patients have with rare diseases. They described their ups and downs in their daily life. They also explained the difficulties they encounter while dealing with the existing health care system in the first case (reimbursement problems) and in trying to maintain a professional activity in the second case (stigmatisation, exclusion).

Unfortunately, Mars Di Bartolomeo, the Health and Social Security Minister couldn’t attend because discussions for the formation of a new government were held at the same time as the plenary session. Dr Yolande Wagener, Head of Division at the Ministry for Health, represented him. She underlined the meaning of Rare Diseases in Luxembourg, the difficulties encountered by patients, doctors and all those involved. With a population of around 500 000 inhabitants, approximately 30 000 people are affected by rare diseases which makes it a major public health issue.

She pointed out that rare diseases complex symptomatology is often unknown to the professionals which leads to extremely long delays for diagnosis. She insisted on the necessity of adopting a multidisciplinary approach which has already been implemented in other domains such as cancer.

She recalled that Luxembourg has been involved in all European initiatives since the beginning and that it is not in its infancy stage. The healthcare system is efficient and many initiatives have already been taken which should help to develop a National Plan in Luxembourg.

She then paid tribute to all the people who fought over the years to raise awareness on rare diseases, and who, thanks to their involvement, made them more visible, namely: EURORDIS team, DG Sanco, ORPHANET, ALAN and all the other patient associations. Their combined efforts have led to the recognition of orphan drugs, the promotion of patients' rights and a better access to treatments. It also leads to the creation of alliances. She gave a special thanks to ALAN and to all the professionals who helped to organise the conference.

Michael Hübel, Head of the Unit « Programme Management and Diseases » from DG for Health and Consumers in the European Commission based in Luxembourg, presented the role of European institutions in structuring European Rare Diseases networks (Orphanet, Europlan, Epirare...) and creating consultative structures with patients and other stakeholders (EUCERD). He insisted on the importance of the European cooperation and its significant influence on the efficiency of the national policies concerning rare diseases as well as on the development of good practices. He gave also the example of the European Reference networks, which soon will be established to help structure healthcare pathways. Rare diseases representing a crucial public health issue, the national plan on RD is the key building block for the definition and implementation of actions aiming to improve the situation of the patients. It is also one of the best instruments to raise awareness.

Yann Le Cam, Chief Executive Officer of EURORDIS, closed the introductory session. He reminded the audience that one of the starting points for the European policy on rare diseases was the European conference which took place in Luxembourg in 2005. A public consultation followed which gave way to European Council Recommendation and the definition of a European Policy on Rare Diseases by the European Commission. Luxembourg must be an example of the way it takes care of its vulnerable citizens.

He pointed out the specific situation of Luxembourg as a small country having to work closely with the neighbouring countries in order to obtain a critical mass and to benefit from relevant expertise and resources. Luxembourg needs to seize various potential opportunities, namely; the European directive on cross border healthcare, the creation of European reference networks and the pilot European network on Health Technology Assessment (EUnetHTA). He underlined the necessity to make financial resources available. However he warned not to focus excessively on budgeting issues at the expense of developing a long term approach and a sustainable system.

Workshops - 20 November 2013

The second day of the conference took place at a conference centre in Luxembourg. The mayor of Luxembourg, Xavier Bettel (who has become Luxembourg's Prime Minister in December) gave an introductory speech. He mentioned the engagement of Luxembourg City in supporting disabled people. He insisted on the importance of considering rare diseases not only as a public health priority but also as part of a program aiming to build an inclusive society.

Dr Yolande Wagener gave an overview on the history of the care and treatment of patients with rare diseases in Luxembourg.

Luxembourg has always been involved and always supported European initiatives, for example, RD task force replaced later on by EUCERD.

In 2005 Luxembourg was President of the European Council and organised the European Conference on rare diseases which had a very positive impact. It enabled the professionals to become more familiar with each other. A panel of experts was then created to reflect on the issue of rare diseases in Luxembourg (MARA Group). It allowed ALAN to enlarge its scope (ALAN represents more than 100 rare diseases). The singularity of RD became recognised and as a result many patients were recognised. The first priority became the care of these new patients before considering the elaboration of a strategy. The difficulty today lies in the fact that no resources have been made available to sustain the actions that had been engaged, which should be solved thanks to the elaboration of a National Plan.

Antoni Montserrat, Policy Officer for Rare Diseases (DG for Health and Consumers) stressed the importance of adopting a co-ordinated European approach. He pointed out that some actions would be impossible without a European framework of cooperation as created by the European Commission. As Luxembourg is a small country with a reduced number of patients and resources, the participation to these initiatives is crucial.

He gave a brief overview of the on-going projects. European reference networks will become concrete in 2014. They will bring together specialists centres across Europe and help to disseminate information and expertise. The new version of the ICD (International Codification of Diseases) will improve classification and codification of rare diseases (today only 240 rare diseases are codified). The IRDiRC (International Rare Diseases Research Consortium) will support development of 200 new therapies in international cooperation with USA, Canada and Australia (and recently also China). He insisted on the necessity of reinforcing the role of patients associations.

This EUROPLAN conference gives Luxembourg the power to develop and implement a national plan, as many European states have already done. He indicated that in 2015, Luxembourg will again be President of the European Council. He recalled how decisive the 2005 European conference had been in the change concerning RD. Thus 2015 should be considered as an opportunity.

Lily Cannon who was EURORDIS Advisor for the preparation of the EUROPLAN Conference in Luxembourg recalled its main objectives and presented the themes of the workshops to follow. She gave a brief description of their content and she presented the chairs in charge.

All the workshops gathered the professionals concerned (medical field, healthcare, social care, research...), and included patient associations' representatives.

Doctor Fernand Pauly then took charge of the coordination of the session. At the end of the day, a "questions and answers" session, moderated by Cécile Hemmen, followed the presentation of the workshops conclusions and Doctor Fernand Pauly closed the conference before giving the floor to ALAN's President, Shirley Feider, who on behalf of both the patients and patiens associations, thanked all the participants.

III. MAIN REPORT - WORKSHOPS

6 main themes were discussed in the workshops.

- Methodology – Governance and monitoring of the national plan
- Definition – Codification and inventorying of rare diseases - Creation of a registry
- Research
- Care for rare diseases - Creation of a platform for rare diseases
- Health care systems supporting people with rare diseases (social security benefits concerning diagnosis, genetic testing, treatments/orphan drugs)
- Social services for rare diseases

1. Methodology – Governance and monitoring of the national plan

There is no national plan or strategy concerning rare diseases in Luxembourg. The structuring of a national plan/strategy in Luxembourg was considered as a transversal theme which should be discussed in each of the four workshops, on the basis of the same set of questions:

1. How to develop a national plan or strategy to address the healthcare and social needs of patients with rare diseases in Luxembourg: what steps and actions should the workshop recommend to advance the development of a national plan/strategy (the process)?
2. How to make sure that the participation of patients is envisaged in the development and implementation of the National Plan?

Background and actions put in place

In Luxembourg, the people and professionals concerned by rare diseases in the 70's initially worked in an isolated way. They became progressively more and more involved in European initiatives. Patient associations were created in the 90's; today Luxembourg has approximately 20 patient associations.

The European Conference on Rare Diseases, which took place on the 21-22 June 2005 in Luxembourg, gave impetus to the consideration of rare diseases as a public health issue and helped to appraise the situation of rare diseases in Luxembourg

Apart from the well-known problems and obstacles encountered by patients, it also helped to highlight the particular features of Luxembourg, as a small country very much dependent on cross border cooperation, namely:

- the absence of a university hospital with centres of expertise specialised in the field of rare diseases;
- clinicians who studied abroad and who maintain special ties with centres of expertise, in France, Belgium, Germany, Austria...;
- no national institution to provide coordinated information, guidance, psychological and social support;
- demography with more than 35% of foreigners with specific linguistic and cultural needs.

A multidisciplinary national group was created, MARA Group (Groupe de Travail Maladies rares) which was supported by the Ministry of Health and Social Care.

It conducted a survey on rare diseases in 2007 with the support of the patients associations. Its purpose was to appraise patients' needs and its results were published in 2011.
The conclusions of the survey helped to define actions to improve the existing situation. They were meant as part of a National Plan. The structuring of this National Plan has been put on hold essentially due to lack of resources even though some improvements were implemented separately.

Workshops conclusions

The structuring of a National Plan on rare diseases for Luxembourg is considered as a priority. There is a large consensus on its objectives: to help patients with a rare disease to have a better life, to improve equal access and availability of prevention, diagnosis and treatment and to participate in related European programs. The conference was considered to be a first step in a process which will lead to the National Plan in the proceeding year.
The empowerment of patients through the support of patient associations was debated. In each workshop the participants proposed the creation of an Alliance in Luxembourg which would be a key stakeholder in the development of the national plan. Each Association would keep its specific characteristics while pursuing the same goal. The Alliance would also promote the exchange and pooling of information and resources. It would have a stronger voice at European level (amongst other in the CNA – Council of National Alliances – EURORDIS).
In addition, the need for specific resources, budget and coordination skills was emphasised.
(cf. details of the workshops proposals which would serve as a basis for the national plan in the following pages of this document).

Government's commitments – December 2013

The anticipated election process in Luxembourg was concluded in December and the new government was put in place with effect from 3 December 2013. The new government parties in Luxembourg structured a « coalition agreement », published on the 3 December 2013. A chapter addresses the rights of patients (p. 173, p. 174). It is summarized below.

“In order to protect the patients’ rights and interests in case of incidents or damages, the agreement plans to create a compensation fund which will also specify the handling of therapeutic hazards. This fund will be monitored in connection with the systems in place in the neighbouring countries.

The government will attach more importance to the collaboration with patient associations, with the purpose of implementing prevention campaigns and to also fine-tune modalities and healthcare paths for some specific diseases.

A National Plan on Rare Diseases will be structured together with national and international organisations (such as Eurordis) on the following aspects:

° treatment and healthcare system

° creation of a platform for rare diseases

° creation of a rare diseases registry

° social services

The objectives will be to address patients’ expectations and to allow them to make well-grounded choices on healthcare, care providers and treatment.

The patients' bill of rights in cross border healthcare (transposition of the directive 2011/24/UE-European Parliament) will be adopted by the new deputy chamber after seeking advice from the Council of State. The government will take responsibility for chronic diseases".

2. Definition, codification and inventorying of RD - Creation of a registry

2.1. Definition

Luxembourg uses the definition of "rare diseases" as adopted in the European Union (see European rules on orphan medicinal products —141/2000/EC), namely a disease affecting not more than one person in 2000.
5-7 % of the population is concerned, which means that in Luxembourg at least 30 000 people may be affected.

2.2. Codification of RD and traceability in national health system

The survey carried out in 2007 under the supervision of MARA group "the situation of people with rare diseases in the Grand Duchy of Luxembourg", has enabled a first progress report to be produced.
In Luxembourg today there is no approach defined at national level for the collection and recording of data on rare diseases. They are not easily coded or traceable in the health system. There is no comprehensive list of rare diseases.

2.3. Registries and data bases

In the existing healthcare system, the situations are managed on a case-by-case basis. Therefore, the creation of a registry is an essential step to foster improved care and treatments of rare diseases.
It requires systematic collection of data, allows a better knowledge of rare diseases and thereby provide better responses to the challenges raised by the prevention and care requirements.

The national cancer registry has recently been launched in Luxembourg. The project was entrusted to the CRP (public research centre) by the Ministry of Health. All stakeholders locally are involved (laboratories, hospitals, doctors, ministries). This experience could contribute to the definition of a methodology applicable to the building of a registry at national level.

<u>Structuring of a registry for rare diseases in Luxembourg</u>
Three approaches can be considered:
° the creation of a national registry;
° the participation in international registries to access data for clearly identified diseases;
° the participation in international registries, to access those data for a specific treatment;
The creation of a national registry should be favoured.

Objectives of a national registry

° collect clinical and epidemiological data and develop knowledge on rare diseases
° initiate clinical and epidemiological research projects
° enable voluntary patients to participate in those research projects
° dispose of relevant data to identify patients who could benefit from new treatments (orphan drugs)
° ensure a long term follow up of patients affected by a newly discovered disease and of patients having been treated with new therapies.

The creation of a focal point should be considered. It would help to orientate the patients, to facilitate their access to new treatments and their participation in international research projects.

Data collection

The "shared care file "(electronic shared healthcare record) under the e Health project (see section 3.1 for more details on the project), will be the main source of information. It will be necessary to define the relevant information which are to be extracted; a "minimum set of common data elements" to ensure standardisation of data. Furthermore, patients should be able to communicate data directly.

EPIRARE, the European Platform for Rare diseases Registries, launched the project to develop this standard and define the data which should appear as "common data set", the legal aspects, etc. The inter-operability of registers is highlighted.

It would therefore seem appropriate to wait for the outcome of this project before launching the registry of rare diseases in Luxembourg.

A legal and ethical framework will have to be defined and serve as a basis for data collection.

What coding system to adopt?

ICD10 is used in Luxembourg. It is a complex system that, in addition, fails to adequately codify rare diseases.

The new system ICD11 will include rare diseases. It is currently being prepared and will be submitted to the National Assembly of the WHO for approval and implementation in 2015. ORPHANET (in which Luxembourg is a party) is involved in this process of classification of rare diseases in the new system. Today ORPHANET uses a codification system: Orpha Code.

Measures will have to be anticipated to be able to quickly implement the new system, once it has been adopted.

In January 2013 the Health Directorate (Direction de la Santé) has started to identify rare diseases in Luxembourg, using Orpha Code.

2.4. Training healthcare professionals to recognise and code RD

The fact that codification is completed by different persons carries a risk of interpretation and threatens the homogeneity of data. In order for professionals to appropriately encode, proper training needs to be addressed.

Orphanet/Inserm within the framework of the EUCERD Joint Action will organise a workshop on Orpha Code in March 2014, which will include presentations of countries who are currently in the process of implementing the Orpha Code.

3. Research on RD

Research is carried out at two levels:
- national projects conducted in Luxembourg laboratories involved in international networks;
- international projects with Luxembourg samples.

Clinical and epidemiological research raises the issue of the access to personal data.

3.1. RD Research programs – EU and International collaboration

Most organisations in Luxembourg are associated with European organisations and participate in joint projects. They are grouped within the PMC (Personalised Medicine Consortium of Luxembourg). This consortium ensures coordination in research projects requiring important multidisciplinary teams.

LNS (Laboratoire National de la Santé) is a public institution under the supervision of the Ministry of Health. It is in charge of a public health mission on epidemiology, hygiene, involvement in health policy and human medicine. It is responsible for developing analytical and scientific expertise concerning prevention, diagnosis, monitoring and surveillance of diseases. The laboratory is involved in European and international networks and projects.

IBBL (Integrated Bio Bank of Luxembourg) supports medical research which includes the collection, storage and analysis of biological samples. IBBL works together with ISBER (International Society for Biological Environmental Repositories) and takes an active part in its working groups, in particular those concerning pre-analytical validations. In the next future, IBBL may adopt the role of observer in the network BBMRI (Bio banking and Molecular Research Infrastructure) which brings together more than 225 organisations, mainly bio banks, from 30 countries.

LSCB (Luxembourg Centre for System Biomedicine) belongs to Luxembourg University. It establishes the link between the biological systems and medical research including bioinformatics. The centre is linked to EATRIS (European Infrastructure for Translational Medicine) which transforms the research results generated by laboratories into new approaches to diagnosis and treatment. LCSB recently launched a research project on Batten disease.

The Luxembourg national eHealth Agency (Agence Nationale eSanté) created in 2011 is part of the Economic Interest Grouping "Healthnet": a telematic platform created in 2005 for the healthcare sector which provides a secure IT network for sharing and exchanging health data. The agency's work is supervised by a body made up of the Ministry of health and social security, the national insurance fund (CNS), representatives of service providers and patients representatives.
It manages the eHealth platform which allows for exchanging data and sharing services (http://www.esante.lu). The platform provides health professionals with secure tools to ensure continuity of care for the patients.
The first services offered by the platform are the healthcare professionals and patients' directories, a secure messaging system and a space designed for sharing information about the patients (electronic shared healthcare record – "DSP: Dossier de Soins Partagé »).
The eHealth Agency also fosters community of practices by providing an infrastructure which takes the Luxembourgish healthcare context into account. It promotes cooperation based on a European

interoperability framework within cross border areas (“Grande Région”) and the European member states.

CRP Santé (Public Research Centre) conducts various research projects within CIEC (Clinical and Epidemiological Centre). The CIEC is part of the European network ECRIN (European Clinical Research Infrastructure Network). This network organises academic research. It covers three areas of research: medical devices, nutrition and rare diseases. CIEC is also associated to EUPATI (European Patients Academy on Therapeutic Information). This organisation aims to communicate reliable information to patients, in the field of medical research, in order to enable them to participate in therapeutic innovation.

European and international initiatives to consider in the future:

E-RARE: Consortium of organisations involved in research funding

IRDiRC: International Rare Diseases Research Consortium, which now works on two main objectives for the 2020: setting up 200 new therapies for rare diseases, and diagnosis for the majority of rare diseases

Research projects undertaken at European level by JRC (Joint Research Centre Ispra – I taly) in particular in the field of biotechnology.

DG for Health and Consumers (European Commission)

EUCERD: European Union Committee of Experts on Rare Diseases

Contact: Y Wagener Ministry of Health

EURORDIS Council of National Alliances (CNA) and Council of European Federations (CEF)

Contact: Shirley Feider ALAN

COMP: Committee from Orphan Medicinal Products) within EMA (European Medicines Agency)

Contact: Henri Metz, Ministry of Health

MARA group should play an active role in the establishment of a registry and research.

3.2. Legal, ethical aspects

The registry of rare diseases raises the sensitive issue of data protection. The law of 2 August 2002 lays down the legal framework on the protection of individuals with regard to the processing of personal data. A National Commission for Data Protection (CNPD) was created which also extends to the protection of privacy in the electronic communications sector.
The formal framework for the creation of a registry and the participation in research projects must comply with this framework, or complete it, if necessary.

3.3. Patients’ role

Patient associations play an essential part in informing, supporting and accompanying the patients having a rare disease. The creation of an Alliance would contribute to the patients’ involvement not only in the creation of a registry but also in research programs.

3.4. Pending questions

The following questions will have to be addressed:
- who will manage the registry?
- in which legal framework should it be established and maintained?
- how can it be made sustainable knowing that a register implies significant costs?

Several ministries may be involved (ministry of health and social security, ministry of family and integration, ministry of higher education and research). A budget should be made available.

4. Care for rare diseases. Creation of a platform for rare diseases

Given the size of Luxembourg and the lack of possibilities in terms of research, there is no centre of expertise as defined at European level. However, there are experts for certain rare diseases and several doctors participate in European or international research projects.
The University of Luxembourg is also engaged in this field for the forthcoming years.
The creation of a platform for rare diseases which is accessible to everyone is the relevant option for Luxembourg.

It should be emphasised that a meeting was held at the Ministry of Health, prior to the Conference EUROPLAN. The Health Directorate intends to make an active contribution to the development of a rare disease strategy and to the establishment of a rare diseases platform.

Scope and functioning of a RD platform

Objectives
° to provide support, information, advice and care to the patients and their families, orientate them
° to provide support to all professionals involved in healthcare, who most of the time, work in isolation.
° to contribute to identify clearly existing resources: national experts, international experts and centres of expertise in neighbouring countries or other countries for extremely rare diseases.
° to provide medical consultations (accelerating research diagnosis, optimising care) and social support.
° to organise training for the professionals.

This platform is to be considered the 'focal access point' to various services, grouped in the same place. This pooling of resources based on a multidisciplinary approach, will help to optimise as well as enrich the knowledge and the skills in the pool. Secondary prevention, which reduces the duration of evolution of the disease (screening, early stage of processing) will be enhanced.
It will operate on the basis of multidisciplinary care with a list of diseases so that all speak the same language.
It will constitute a single gateway, accessible and known to patients, their families and to the clinicians and provide them with an overview of existing expertise.
It will define a strategy for collaborating with the European Networks of expertise and assess the quality of the subcontractors' services while ensuring sustainability.
The size of Luxembourg can be considered an asset here: privileged relationships already exist between the parties concerned.

Conditions of implementation

Communication and coordination between the various players is a prerequisite for effectiveness. The more complex the disease, the more it requires a concerted action of different stakeholders. This applies to the diagnostic phase and for long-term care.

Medical training on “rare diseases” has to be organised in particular for generalists and paediatricians professionals who are often the first point of contact with patients affected by rare diseases. It can take quite a lengthy time to obtain a diagnosis due to the uncertainty of the professionals where to refer their patients to. The coordination becomes more complex if the patients have previously met several specialists over time.

The patient's file created by the general practitioner (“médecin référent”), will influence the quality of care that will be implemented afterwards. A specific model must be defined for patients affected by rare diseases.

Recourse to external experts will be orchestrated within the platform. Today patients are sent abroad at the initiative of the doctors who use their personal network of experts.

The use of telemedicine/conferences will bring together several experts who will liaise on a single case at the same time as it facilitates local care.

Patients will be represented in the platform. The creation of an alliance in Luxembourg should be encouraged. It will represent all patients affected by rare diseases and not only those who are members of an association. It will participate in decisions and actions of the platform with more weight than if associations were represented individually.

Specific healthcare paths will be structured on national and international levels, to guarantee the continuity in the implementation of clinical protocols.

Exchange of information must be organised between the parties in order to be efficient and to ensure safety. They must be aware of what has been carried out, the available resources and the availability in terms of capacity. A national document should be structured with all relevant information.

The platform will be easily accessible through a rare diseases hotline and a single e-mail address.
Communication towards patients and their families will have to be adapted in all areas related to care.

Link with ministries and bodies of administration:

The platform will allow the administrative duties be conducted and it will be the correspondent vis-à-vis official bodies. Given the complexity related to the number of institutions concerned, interlocutors, procedures, compartmentalization, this is an important challenge.
Simplification is needed particularly in medical records. The administrative language must be adapted and “translated” into understandable terms.

The designation of knowledgeable contact persons in the organisation of medical control (a doctor and pharmacist) will help to ease the reimbursement process.
Reimbursement is a sensitive issue. The costs of treatments are important, genetic tests are increasingly being requested. The creation of a special fund could be envisaged. A model remains to be found which would be viable and preserve the spirit of solidarity.

The platform will have to use a unique coding system; rare diseases have to be codified according to the international system
The Public Health Authority will have to make means available for the development of this platform. A group of experts should be created to proceed with the project.

5. Health care systems supporting people with RD (social security benefits related to genetic testing, diagnosis, treatments/orphan medicinal products)

The health system in Luxembourg does not provide specific framework for rare diseases and even less for unusual situations. Medical and psychological care depends very much on the medical experience of the general practitioners and/or on the psychosocial support provided by patient associations; often by ALAN. Decisions are often made on a 'case by case' basis with the support and goodwill of all the people concerned.

5.1. Biotechnology – Diagnosis - Genetic testing

Advances in technology have greatly improved the diagnosis possibilities in the field of rare genetic diseases, thanks to the availability of an effective tool, shared knowledge on their history and evolution (Orphanet, GeneReviews), access to clinical expertise and, for some diseases, therapeutic monitoring.
However, the description of a genetic abnormality called "causal" or "damaging" in a patient, not only concerns the clinician but also the health system:
- will the other family members be tested?
- shall the genetic mutation be searched in relatives who are clinically healthy but may develop the genetic diseases if it can be transmitted (recessive inheritance with high risk of transmission for siblings, dominant or related to X chromosome and its intergenerational concerns)?

These questions concern what geneticists call the pre-symptomatic diagnosis in the field of curative medicine: the detection of a genetic disease before the appearance of clinical symptoms. They also raise the issue of criteria and procedures for making decisions as well as the application of adequate arrangements to provide healthcare while ensuring equal access.

Access to genetic tests is not clearly defined. Currently the majority of genetic tests prescribed to patients living in Luxembourg are carried out in laboratories abroad (neighbouring countries) at the request of hospital clinicians (paediatricians, obstetricians, neurologists…) or other clinicians.
Furthermore the skills and clinical expertise of national or international laboratories are not always known.

The quality of prescriptions and diagnostic hypothesis (clinical, biological) have been discussed with the prospect of clarifying the process as well as the reimbursement of genetic analysis for the patient.
The reimbursement request is submitted for approval to the medical control institution (Contrôle médical) which belongs to the ministry of social security. In this matter, the doctors having to assess the request want to have access to the complete file with all relevant explanations.

5.1.1. Screening:
As regards to the national prenatal screening, a program is in place which ensures excellent coverage and exemplary quality, not only technically but also in terms of cooperation between LNS (Laboratoire National de Santé) and all the maternities, to ensure a follow up of children tested positive.
However, in case of anomaly detected during the ultrasound exam, there is no multidisciplinary medical structure. Cooperation is efficient between obstetricians and paediatricians at the CHL (Centre Hospitalier de Luxembourg). It is problematic with the genetic service of LNS (Laboratoire National de la Santé) because of its location (situated in other premises) and due to the fact that there is a lack of clinical expertise (no medical geneticist specialised in dysmorphia and syndromology in foetal stage).
Interdisciplinary discussions are necessary in the monitoring of high-risk pregnancies and could be a key element in the decision leading to therapeutic abortion. For the time being, it is not required to submit a “file for concertation” to an ethic committee, prior to a therapeutic interruption of pregnancy (ITG). Furthermore there is no record for malformations and intrauterine deaths.
Fetopathology expertise exists abroad, not in Luxembourg. There is no designated centre for foetal pathology.

5.1.2. Difficulties encountered by and with laboratories:
When receiving a prescription, the staff in the laboratories do not always have the skills required for an accurate analysis.
Laboratories work in collaboration with their peers abroad; these collaborations are often more concentrated on old links than on expertise.
The collaboration on a national level is limited: there is no harmonisation of tests procedures, no common quality criteria, no cutpoints…
The lack of multidisciplinarity is a problem.
The assessment of the quality of the laboratories, their skills (in particular in the field of foetal pathology) is not based on a structured approach (no accreditation).
Cross border cooperation presents difficulties in particular on returns and deadlines and also because of the language used for communication (German or French).
The tests' costs are high, today they are charged on the operating costs of the hospital.
The prenatal genetic diagnosis laboratory is located outside the hospital premises

The clinicians propose that the samples should be collected by a central laboratory which would transfer and receive results complying with high quality standard, within a reasonable period of time.

5.1.3. The ethical dimension

Bioethics issues arise in parallel to progress on biotechnology in particular the access to genetic material in pre-natal stage and during life.

Pre-implantation diagnosis:

The genetic diagnosis on embryos, applied in a limited number of centres of expertise in Europe to detect genetic disorders and chromosomal abnormalities, is used in conditions other than prenatal screening and raises other issues especially ethical issues.

The National Commission on Ethics has delivered a reasoned and favourable opinion for this diagnosis procedure. A favourable opinion was also given to in vitro fertilisation for homosexual couples but as of yet, nothing is formalised on this subject at the level of the CNS (national health insurance fund) or the institution in charge of medical control (Contrôle médical). There is no legal framework.

A national reflection should be conducted on the status of pre-symptomatic tests in the diagnosis process. This need was discussed in April 2013 within the Scientific Council which should now give its opinion.

5.1.4. Reimbursement, role of the Medical Control institution and CNS

The Medical Control institution wants to apply the fairest system. At the same time, it has to comply with the Article 23 paragraph 1 which implies “to provide the best indication for the patient according to the therapeutic impact and also to the generated costs”.

Two possibilities should be examined to improve the situation:

- harmonisation of procedures;
- accessibility of the patients files for doctors working for the Medical Control institution. These files provide all necessary information to allow for a relevant opinion. In all cases one should get the opinion of a doctor with the relevant expertise.

Legislation must be reviewed (laws, regulations, National Health Insurance Fund’ status).

Access to genetic testing should be fair, and the same for all:

- the CNS taking in charge the diagnoses;
- a solution should be found to cover the costs of genetic testing for screening purpose;
- a convention will be defined for pre-symptomatic tests including a reflection on the prescription and the chain of subcontractors (quality, cost, deadlines).

The Swiss model could be transposed with the creation of a list of rare diseases and the setup of a fund to deal with the situations which are not covered elsewhere.

The Ministry of Health is willing to participate in the definition of a framework with all the stakeholders concerned.

5.2. Access to treatments - Orphan Drugs

Today the reimbursement of orphan medicinal products is not guaranteed. It is based on two lists:
- a “positive list” of drugs distributed by pharmacies and open to all doctors who can prescribe;
- a list of specific medicinal products under hospital control.

The drugs prescribed in ambulatory context should be included in the positive list.
As far as orphan drugs are concerned, prior approval is often required to allow for total reimbursement (protocole therapeutique).
Drugs which are used within the hospital (H) or prescribed in the hospital (D) are charged on the operating costs of the hospital and do not require a decision for their reimbursement. In theory, they are rapidly available but the patient has to go to the pharmacy of the hospital to get them.
The procedure is as follows: doctors prescribing the drugs must submit a request with supporting arguments for the drug to be registered on the positive list.
The “health technology assessment” procedures and drugs reimbursement procedures are not harmonised on a European level contrary to those requesting to put a new drug on the market.

‘Off label use’ of medicinal products (which does not correspond to the official authorised use) are not reimbursed. There is no legal disposition for drugs prescribed in this context.
There is no legal framework either concerning the anticipated access to drugs which are in the process of registration on the positive list (Medical need) or in the process of obtaining an authorisation to be put on the market or in the process of obtaining a new therapeutic indication out of the clinical testing program (compassionate use).
An unofficial procedure is in place in the Division of Pharmacy and drugs to handle those situations.

There are no reimbursement problems for medicinal products already available in Belgium.
It is more complicated if a new molecule has not yet obtained the European recognition (EMA) or if it has not received the authorisation to be put on Belgium market. In certain situation, the approval allows to order the drug in Germany but at a very expensive price.
A solution should be found for the “off label” use of drugs, with the prior agreement of the Division of Pharmacy.
The question of the quality of prescription and the access to prescription will have to be answered ; who should prescribe? What should be his profile?

5.3. The Creation of a centre of expertise/rare disease platform

A rare disease platform could contribute to solve some of the difficulties mentioned. It should be based on the principle of multidisciplinarity. It would offer a unique access point to the patients, their families and the clinicians. It would give an overview of the existing skills. It should define a strategy for national interdisciplinarity and for collaboration with European expertise networks and use a unique system of codification (e.g. Orphacode). It would approve subcontractors based on a quality assessment.

5.4. What strategy to develop - proposals

- structure recommendations on diagnosis and care;
- launch a national reflection on solidarity and intersectoral cooperation
- reinforce and structure multidisciplinarity and collaboration between the partners concerned (optimise inter-professional communication);
- set up a rare diseases network (criteria, healthcare, and healthcare paths, psychosocial support,genetic centre);
- expand Orphanet Luxembourg (catalogue of skills, codification and registry);
- establish a professional platform (centre of expertise), a hotline for the patients;
- define a legal framework for the issues related to genetics;
- define a concept and procedures for all situations who are not covered by the healthcare criteria in use today by the medical control institution (Contrôle médical) and the national health insurance fund (CNS);
- update the existing nomenclatures;
- set up a panel of national and international experts in order to support all the requests which are excluded from the existing framework and/or exceptional;
- structure the collaboration between all the partners concerned in order to ensure healthcare continuity, quality of diagnosis and treatment, equal access, communication pathways, reimbursement;
- reinforce and structure collaboration with centres of expertise abroad.

6. Social Services for Rare Diseases

There is no specific national programme to assist patients affected by rare diseases and their families in Luxembourg. They benefit from existing measures, in particular those applying to disability. However, these measures do not address the special nature of rare diseases and the specific needs of the patients. There is no centre which would bring together all medical expertise, information and social support. Actions are not coordinated and information on rare diseases is scattered and not easily accessible.

6.1. Social resources and services for people with disabilities

6.1.1. The situation of children affected by rare diseases and their families

The care of a child may require a 24 hours presence. There is no support for parents coping with their child at home or any night assistance available.
However, “Stëftung Hëllef Doheem” (institution offering social and medical services for patients at home), has put in place on 1 March 2009 an experimental project which aims to offer a 24 hours coverage. This project entitled “Nuetswaach” is placed under the authority of the long term care insurance and funded by the Ministry of Family.

There is no dedicated childcare organisation which could help the family in case a child “at risk” cannot go to a nursery for example. Financial support is provided to keep children in existing institutions, but not if the child is being cared for at home, by a parent.
The Ministry of Family encourages the child-care establishments (“MR: maisons relais”), to welcome children with a rare disease. Children have access to the MR in their municipality and they can keep

contact with other children. These MR may benefit from additional resources from the Ministry of Family ("additional hours").
(*Maisons relais are public establishments, approved by the State; they host children during the day and the working time of parents and offer educational and recreational activities).*

Guichet.lu (Administrative guide of the Luxembourg State) provides information on custody of children in general: the principles, the rights and the existing infrastructures.

Due to the lack of expertise, patients are often sent abroad for diagnosis and treatment, mainly in neighbouring countries (Germany, France and Belgium). When the parents accompany their child abroad, the question of care of the siblings who stays in Luxembourg arises. Furthermore there is nothing organised to assist the parents with their stay or to provide them with psychological support. Language problems may arise for both children and their parents.

The family may become destabilised; often one of the parents needs to stop working to care for the child. Sometimes, following a long parental leave, the parent concerned loses her/his job and becomes unemployed. Marital problems can arise resulting in a spouse leaving the family home. Psychological support with a professional qualified is not easily found and there are long waiting lists. Funding can also be a problem.

6.1.2. Patient associations 'support
Various associations take care of children having a rare disease. Following are a two examples.

The Foundation Kriibsskrank Kanner offers support for children till the age of 18 and their family, when the child is affected by cancer or a serious and rare illness implying vital risk. In this last case, the approval of the foundation's medical committee is required.
Its scope of action includes:
° Administrative, legislative and financial assistance (organisation of special leave with the doctor, communication with the employer, payment of all the invoices directly related to the pathology);
° Psychosocial support in Luxembourg and Brussels; the main centre for treatment;
° Logistical support (arranging transportation, housing care of the siblings who are staying at home)
° Medical support based on an agreement with "Kannerklinik" (Children's Hospital) which offers homecare;
° Housekeeping and home maintenance according to the hygiene rules imposed by the pathology
° School support to siblings staying at home in the absence of the parent;
° School support to the sick child after his return in case of academic regression;
° Organisation of recreational activities to take the family out of its isolation and reconcile the sick child with the joy of living.

The foundation has put three houses available to families:
° The Children House in Strassen, offering school-related activities as well as extra-curricular and recreational activities;
° Losch House in Brussels also called 'Parents house, offering lodging to the parents while their child is hospitalized in Brussels;
° An apartment in Westende, available to the families when they need rest.

The parents association for mentally disabled children (APEMH) created the "Incluso Service". It is geared towards professionals offering educational support and care for children and their parents. It offers extra-curricular activities for children and information, training, coaching, advice and assistance, coordination, networking for professionals. Incluso also assesses the development for children and their specific needs.

6.1.3. Institutional measures

Even though various measures are offered within the existing legal and institutional framework, there is no global approach.
The parent of children affected by cancer may benefit from a special leave for family reasons for one or two years – and this applies to both parents (this should be considered as a right and not as an exceptional favour).
The national health insurance fund (CNS) offers various benefits, but procedures are long and complicated.
Sometimes school health services play the role of "case manager" because the families are often lost in the procedures and the various parties involved.
The National Office for Childhood (ONE) created in 2008 is in charge of preventing and handling the situations of psychological and social distress. Today it addresses more social issues than medical problems and the waiting delays are very long.
CPI services (Coordinateur de Projet d'Intervention) coordinate projects, orientate and assess the various measures put in place for a child, its family or a young adult. There are 3 CPI services in Luxembourg which bring together many stakeholders in the psycho-social field.

There is no coordination between all existing possibilities dispersed among various ministries and institutions (Ministry of Health, Ministry of Family, National Office for Childhood "ONE", CPI, etc.).

There is no continuity in the care provided throughout the lifecycle, from child to adult.

6.1.4. Proposals to improve child care and families' support:

- Ensure child care and assistance 24 hours a day;
- Provide financial support in case of home care;
- Create shelters for sick children to enable parents to care for their other children;
- Grant special leave for family reasons, on the basis of an acquired right and not as favour granted in an exceptional situation;
- Increase the offers of school-related activities as well as extra-curricular activities for children with special needs;
- Meet psychological and occupational therapy needs;
- Reconsider the role of both the school doctor, referrals and the social assistant of the school, (they could act as co-ordinator);
- Redefine the role of the national childhood office (ONE) at the Ministry for Family Affairs taking into account children with rare diseases.

6.2. Access to information

The information is not easily accessible. The fact of being informed depends very much on personal initiative.
Even though the national website “Guichet.lu” provides a lot of information and forms, there is no centralised information service on the different subjects (medical, social, professional, etc.). For example, there is a lack of information on how to apply for disability benefits, pension, how to fill in the tax return form...

A Guide for disability is edited by Info Handicap (Federation gathering 53 Associations for the disabled), but there is no specific guide for rare diseases.

There is also insufficient exchange of information on the different diseases and pathologies between the professionals.

6.3. Policies to integrate people living with rare diseases into professional life

Patients face many obstacles. In some cases, the doctors do not react immediately which can prevent early diagnosis and detection of the disease.

The medical certificate gives a diagnosis, but without indicating the person’s limitation and the status of “disabled person”, it is not sufficiently precise. Moreover, certain persons concerned do not want to be “classified” under the status of disabled employee. The patient is often isolated even though there are likely to be other persons in the same situation. The disease is often invisible, which may cause prejudice.

The global approach is rather administrative. Obtaining the status “worker with reduced capacity” involves lengthy procedures and many people give up before completing them. Several doctors provide various evaluations which lead sometimes to a lack of consistency. Two committees are involved: the Medical Committee and the Guidance Committee.
The discussion on a job, tailored to the specific needs of the person, occurs too late, only when the reclassification file is finalised. If internal redeployment is not possible, then the person becomes unemployed.

Civil servants do not benefit from the same system as private sector employees; they do not have the possibility of reclassification. They are entitled to a compensatory allowance.

ADEM (the Agency for Employment) accompanies people with its educators and psychologists. It has the possibility to allocate 6 additional days leave to disabled people, at their request, when they fall ill. The Agency reimburses the employer for the costs that this entails. ADEM also provides training to the persons involved.

6.4. Healthcare reimbursement

Long term care insurance ("assurance dependence") treats all cases equally regardless of the pathology and according to the definition of the word "dependence" as defined in the Social Security Code (article 348, 1): "the state of a person who thereafter a physical, mental or psychological illness or a deficiency of the same nature, has an important and regular need of assistance from a third person for his basic living needs". According to this definition, dependence must result from a disease, an advanced age is not sufficient in itself.
However, it is the need of assistance for basic living needs which defines dependence and not the pathology. Thus, certain persons affected by a rare disease do not meet the necessary conditions to qualify for reimbursement under long-term insurance.

Physiotherapy costs are reimbursed by the national health insurance fund (CNS).

Occupational therapy is not reimbursed by CNS. However, the beneficiaries of long-term care insurance who are recognised as dependant in their basic living needs may receive acts of supporting nature by occupational therapists under certain conditions and be reimbursed by the long-term care insurance.

6.5. Proposals for improving the care of persons (children or adults) affected by a rare disease:

Recognition of the uniqueness of rare diseases:

- Define a specific status 'rare diseases' at the level of the national health insurance fund (CNS);
- Reflect on the creation of a Ministry of Disability or of a Rare Diseases Unit attached to the Ministry and recruit disabled employees which could contribute to the implementation of the strategy on rare diseases.

Improvement of information on rights and existing means:

- Organise a Central Office which would provide information on all procedures, rights, means, and support available (medical, social, professional);
- Update the guide on rare diseases which had been structured in 2009 by MARA (Working Group on Rare Diseases) and the guide Info-Handicap;
- Create a new website section "MARA" on Guichet.lu, with all explanations and forms.

Simplification, optimisation of care :

- Simplify procedures (to be recognised as a worker with reduced capacity/ to benefit from an adjustment of the working conditions, lay out);
- Simplify procedures for doctors who have to write several certificates in order for the patient to apply for various services (invalidity pension, parking card...);
- Set up a coordination cell for all social and professional formalities. Adopt a similar scheme as the one used in Germany, by disabled persons in all their dealings (one application is sufficient to get all cards which the disabled person needs to benefit from the various existing services);
- Create a "master file" for the patient, a single electronic medical file, accessible to the patient and which could be used by all the different services (cf. shared care file, in the framework of the ongoing project for e-health);

- Create a single commission (medical and guidance);
- Appoint a doctor who would be in charge of the entire patient's file (referral).

Transition in the social system between child-adulthood:

- Set up a co-ordination service providing the link between all organisations and associations
- Review the role of "case managers" and define who should coordinate the various services in different areas (during all stages of life);
- Create a service to provide support to the patient from a very young age until his death (see "transition programme" in France). Support the family (spouse and children throughout life);
- Provide coaching services to accompany and guide particularly disadvantaged persons;
- Consider the option of implementing a hotline for psychological support knowing that today psychologists' fees are not reimbursed by the national insurance fund;
- Give a budget to persons who do not benefit from long-term care insurance so that they can afford the costs of services provided at home (ex: Stëftung Hëllef Doheem);
- Improve access to local medical care.

Information and training:

- Link services, and organise workshops and training days for the professionals working in different services and concerned by social care for rare diseases.

7. CONCLUSIONS

The Europlan Conference has been a major event in the recognition of rare diseases in Luxembourg. It raised high expectations for the future. It has been evaluated positively by the participants who have regretted not have had more time to further discuss the various themes which were debated in the workshops.
The conference gave the opportunity to review all the existing actions and measures already implemented under the supervision of different Ministries or on the initiative of patients associations, to improve the situations of people with rare diseases. It also highlighted the fact that the participants who contributed with enthusiasm to the workshops were willing to contribute to the elaboration and implementation of a national plan.

Important issues and well identified problems were underlined. Main areas for improvement have been listed:

- a lack of knowledge and skills on rare diseases by the stakeholders (patients being often the most knowledgeable of all concerning their condition),
- no comprehensive and up-to-date data on rare diseases in Luxembourg,
- a general lack of information on existing measures to help patients and their families,
- a dispersion of numerous initiatives,
- a lack of coordination between healthcare professionals, laboratories, institutions, ministries, patient associations,
- a complex legal framework and corresponding procedures,
- gaps due to the fact that rare diseases do not have a specific status in the healthcare policy,
- ethical issues related to biotechnology, genetic testing, use of personal data,

- and, finally, a chronic lack of resources to adopt a structured and efficient approach in order to improve the situation of patients.

The discussions allowed all stakeholders to have a say and some solutions have been proposed to address these problems.

At this stage it is considered that the national plan should focus on the following priorities:

- The adoption of the **new codification system** and the **creation of a patient registry** which will allow to have a clear view on the existing situation and the corresponding needs. Even though the adoption of the new codification system may be delayed due to being under discussion, the necessary implementation conditions could be prepared for when it becomes official.
- The **setup of a rare diseases platform** would contribute to solve many of the problems mentioned above concerning the need for information, coordination, simplification, definition of healthcare paths, cross border cooperation, training, quality control, ethical issues... It will help to professionalize medical and also psycho social care. As an umbrella organisation, it will give the logic and coherence which is lacking today.
- The **creation of an alliance** will guarantee that the patients have their say in all the decisions affecting them. This could also contribute to create greater coherence between the associations and optimise their resources.

The commitment of the new government, which was published two weeks after the conference, is a very positive step and the best basis for the development of a national plan on rare diseases in Luxembourg.

ANNEXE 1: PROGRAMME

EUROPLAN NATIONAL CONFERENCE

PLENARY SESSION

19th November 2013

Salle des fêtes "Mamerschlass", Mamer

18h30	Welcome	Gilles Roth, Mayor of Mamer
18h40	Introduction of the conference	Shirley Feider-Rohen, President ALAN
18h50	Testimonies	Christiane Romitelli and Chris Denis Garzaro
18h50	National strategy on rare diseases in Luxembourg	Mars di Bartolemeo, Minister of Health and Social Security – represented by Dr. Yolande Wagener
19h25	European Union and Rare Diseases	Michael Hübel - Head of the Unit "Programme Management and diseases"
19h35	National strategy within European perspectives	Yann Le Cam, CEO Eurordis
20h00	Cocktail –Networking	

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PLENARY SESSION – WORKSHOPS - CONCLUSIONS
20th November 2013
Maison d'Accueil des Soeurs Franciscaines, Luxembourg

8h00	*Registration*	
8h30	Welcome	Xavier Bettel, Mayor of Luxembourg
8h45	Introductory speech: Historical background on the recognition of rare diseases issues in Luxembourg.	Dr Yolande Wagener, Ministry of Health
9h00	The conference within the Europlan context: Issues and challenges of today and for the future.	Antoni Montserrat, DG Sanco
9h20	Reminder of the Europlan objectives - Presentation of the workshops and the topics for discussion.	Lily Cannon, Eurordis Advisor
9h35	**Workshops** :	**Chairs**:
	Workshop 1. Health care system supporting people with RD - cross border healthcare	Prof. Karin Dahan and Dr Yolande Wagnene
	Workshop 2. Creation of a platform for rare diseases - cross border healthcare	Dr Michel Hoffmann and Dr Fernand Pauly
	Workshop 3. Creation of a registry - codification - research - orphan drugs	Dr Anna Chioti and Dr Jos Even
	Workshop 4. Social services	Gwenn Crohin and Dr Marguerite Leches
10h30	*Break*	
10h45	**Workshops: continuation**	
12h30	*Lunch*	
14h00	Presentation of the workshops' conclusions - Q&A-Debate	Chairs with moderator Cécile Hemmen
15h15	Closing speech - What next?	Dr Fernand Pauly, Groupe de travail Maladies Rares
15h30	Thanks - End of the conference	Shirley Feider-Rohen, President ALAN

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ANNEXE 2: LIST OF PARTICIPANTS

WORKSHOPS PARTICIPANTS	
WORKSHOP_A **Healthcare system supporting people with RD** (social security benefits related to genetic testing, diagnosis, treatments/orphan medicinal products)	**WORKSHOP_B** **Creation of a platform for rare diseases**
Chairs: Prof. Karin Dahan - Dr Yolande Wagener	**Chairs :** Dr Michel Hoffmann - Dr Fernand Pauly
Participants Ministère de la Santé : Dr Martine Debacker, Betty Heimermann Contrôle médical : Dr Gérard Holbach, Dr Raoul Hartert CHL, cardiologie: Dr Jean Beissel Agence eSanté : Cindia Bessa Assurance dépendance : Andrée Kerger Laboratoire National de la Santé : Dr Arantzazu De Perdigo CHL, cardiologie pédiatrique : Dr Kerstin Wagner Orphan drugs : Prof. Henri Metz CHL, laboratoire : Georges Gilson Laboratoires Réunis : Prof. Bernard Weber Gynécologie : Dr Didier Van Wymersch University of Newcastle : Dr Antonio Atalaia	Participants Ministère de la Famille : Pierre Biver Ministère de la Santé : Dr Françoise Berthet, Olivier Lepanto Contrôle médical : Dr Edith Miller-Schintgen, Yves Bruch, phamacien Collège médical : Dr Pit Büchler Rehazenter : Dr José Pereira Rééducation fonctionnelle CHL : Dr Emmanuelle Pizon CHL, pédiatrie : Dr Armand Biver Médecin généraliste : Dr Patrick Tabouring ALAN : Gaby Turmes Fondation kriibskrank Kanner : Anne Goeres ALLM (mucoviscidose) : Christiane Hoffmann, Viviane Zimmer EURORDIS : Ariane Weinmann
WORKSHOP_C **Inventorying of RD - Creation of a registry - Research**	**WORKSHOP_D** **Social services**
Chairs: Dr Anna Chiotti - Dr Jos Even	**Chairs:** Gwenaëlle Crohin - Dr Marguerite Leches
Participants Ministère de la Santé : Guy Weber Contrôle médical : Dr Isabelle Rolland-Portal eSanté : Frederic Haas Comité National d'Ethique de Recherche (CNER) : Dr Georges Michel CHL, endocrinologie pédiatrique : Dr Carine de Beaufort Integrated Biobank : Dr Catherine Larue Laboratoire National de Santé : Dr Patricia Borde Een Häerz fir kriibskrank Kanner : Diane Lux Istituto Superiore di Sanità : Dr Domenica Taruscio	Participants Ministère de la Famille : Joëlle Floener Ministère de la Santé (action socio -thérapeutique) : Dr Juliana D'Alimonte Agence pour le développement de l'emploi (ADEM) : Jeff Hurt Assurance Dépendance : Dr Jacques Lück Hellef Doheem : Anke Trabut CHL : Severine Chef Tricentenaire : Nadine Medinger Info-Handicap : Vera Bintener Croix Rouge : Jean-Philippe Schmitt ALAN : Gaby Damjanovic, Lis Goergen ALAEC (Association Luxembourgeoise d'Aide pour les Enfants Cardiaques) : Michel Colin Syndrome de Marfan : Denis Garzaro Fondation kriibskrank Kanner : Marie-Thérèse Probst EURORDIS : Lily Cannon