Belgium

EUROPLAN NATIONAL CONFERENCE

in the framework of the EU Joint Action RD-ACTION

Brussels, 8 June 2018

"WHAT ASPECTS ARE UNIQUE TO RARE DISEASES?

HOW ARE THEY BEING TACKLED BY THE RARE DISEASES PLAN,

AND WITH WHAT RESULTS?"

FINAL REPORT

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FOREWORD

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

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

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

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

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

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

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

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

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

GENERAL INFORMATION

Country	Belgium
National Alliance (Organiser)	RaDiOrg – Rare diseases Belgium
Date & place of the national workshop/conference	8 june 2018 – Brussels
Website	www.radiorg.be
Members of the Steering Committee	Eva Schoeters, RaDiOrg Peter Raeymaekers, LyRaGen bvba Jonathan Ventura, consultant
List of Themes addressed	• What aspects are unique to rare diseases? • Which action points in the rare diseases plan have been most effective in improving patients' lot? • What supplementary actions are needed in the rare diseases plan?
Annexes :	I. Programme in English II. List of Participants (by stakeholders' categories)

FINAL REPORT

I. Introduction/ Plenary session

The aim of this second EUROPLAN round-table organised by RaDiOrg, the Belgian Rare Diseases Organisation, is to have a clear analysis of the current situation around rare diseases in Belgium. To achieve this, the round-table brings together representatives of all parties involved: patient associations, scientific associations and professional medical bodies, administrators and political figures in the field, and the pharmaceutical industry.

The conclusions of this round-table will also allow RaDiOrg to set its priorities for the years ahead, and will allow the patient associations involved to co-ordinate their activities better.

RaDiOrg also wishes to clarify that this round-table is not intended to evaluate the Rare Diseases Plan.

II. Themes

THEME 1: WHAT ASPECTS ARE UNIQUE TO RARE DISEASES?

The issue of what aspects are unique to rare diseases has been the subject of numerous studies and publications over the past two decades, in a context in which specific policies in this area have evolved.[i] To capitalise on this information already at hand, participants were given a list, in preparation for the round-table, enumerating some of the aspects specific to rare diseases.[ii]

The aim of the first session of the round-table was to confirm that these characteristics are still up-to-date and, where necessary, to augment the proposed list (see below):

DIAGNOSTICS
The right diagnosis is often not made for months or years.
First-line medical support is insufficiently aware of rare diseases, which is contributing to the length of delay before proper diagnosis.
Due to the delay in diagnosis, there is a major risk of wrong treatment of the disease or of preventable and irreversible progression of the disease.
Owing to wrong diagnoses, and thus equally owing to a lack of genetic counselling, illnesses are often passed on unawares.
The delay or lack of diagnosis, and the slight incidence of these diseases, are preventing the accretion of properly detailed registers, which is hampering both recognition of these diseases and research into care options and treatments (the link to clinical trials).
80% of rare diseases have a genetic cause (whether heritable or due to fresh mutations), and 50% of them manifest themselves in childhood.
FOLLOW-UP AND TREATMENT
There are few or no therapies specifically tailored to a given rare disease.
There is no treatment (at least, none that will cure) for most rare diseases; the only possibility is to treat the symptoms (to a limited extent).
It is hard for patients to access the most suitable available follow-up and treatment, particularly orphan medicines.
The complex and rare nature of these illnesses makes it particularly challenging to organise integrated and best-quality follow-up and treatment. In fact, following up on these illnesses requires the involvement of many different medical and paramedic specialities.
There are few or no guidelines available on managing the condition.
There are major differences in care models for a given illness between one hospital, region or country and another.
The scarcity of expertise on most rare diseases calls for that expertise to be concentrated at national expertise centres and for transnational networks to be set up.
MANIFESTATION OF THE DISEASE
The manifestation of most rare diseases is very varied (with a great spread of symptoms and of degree of severity of symptoms).
Rare diseases are serious, chronic, and often degenerative and/or fatal.
Rare diseases tend to be complex.
KNOWLEDGE OF THE DISEASE
There is a lack of information on the disease at patient level.
There is a lack of scientific knowledge of these diseases, due largely to the lack of data available (from registers).
Few studies are carried out on rare diseases, because the cost required outweighs the potential yield.
Given the scarcity of scientific expertise, it is essential to foster international co-operation on scientific research.
There is a lack of interest and awareness of rare diseases among the public, even though 6 to 8 per cent of the population have to deal with one.

PSYCHO-SOCIAL ASPECTS
Due to the lack of diagnoses or poor diagnoses, there is often a lack of support or understanding among patients' relatives.
Patients often feel entirely alone; others fail to grasp their situation or problems.
Those affected by rare diseases are often forced to become experts on their own illness. For many patients, this is the most reliable way of obtaining the best possible care, but it does mean pressure and an additional burden of responsibility on the patient or those close to him/her. Moreover, it entails that those less favoured by circumstance are likely to be given lower-quality care.
It is of vital importance that patients enjoy contact with a well-informed patient organisation, both for the sake of gleaning information and for the sake of communicating with fellow patients.
The impact of these illnesses upon patients transcends the purely medical aspect and affects all aspects of life: access to education and the job market, access to recreational activities, shunning, isolation, exclusion from social groups, and types of discrimination in access to insurance or credit.
The impact of a rare disease affects all the patient's nearest and dearest in various ways: psychologically, socially, culturally and financially. There is a vital need to develop support structures for close relatives (such as respite care homes).

This preliminary consultation enabled us to confirm that the list of characteristics of rare diseases does indeed remain applicable, with the caveat that we needed to highlight three aspects needing adjustment:

(1) The need to involve first-line care in the care of rare diseases

"Personally, I think we ought to try to optimise the connections between generalists and genetic research centres and centres of expertise. The proportion of prescriptions written by generalists is poor; less than 5%."

"Reinforce first-line care: general medical training and training of health care practitioners in the widest sense on rare diseases …"

"We need better information / awareness-raising of first-line carers on rare diseases, so that 'red flags' can be identified more quickly and the patient can be transferred sooner to the appropriate centre of expertise."

(2) The need to raise public awareness of the issue of rare diseases

"One major characteristic that sets us apart from other kinds of diseases is the persistence of lack of awareness and thus dissatisfaction. Besides, mainstream media don't recognise this issue … it's really a necessity to draw more attention to rare disease patients."

"A suggestion would be to specify that we have to do with a lack of interest and awareness concerning precisely the rarity of rare diseases, because, even though some particular rare diseases are receiving significant public attention, the public is not necessarily putting these in the bracket of rare diseases."

(3) Access to the best care and treatment options. On this point, the participants in the preliminary consultation emphasised the following specific issues:

- Too few specific treatments for rare diseases
- Too few healing treatments
- Excessive cost of orphan medicines
- Difficulty of accessing treatments abroad, particularly if the treatment is not (yet) one reimbursed by insurers in Belgium
- Length of time taken to decide to reimburse treatments (three months could make a life-or-death difference)

As a final gain, the consultation allowed us to highlight some of the advances made as compared with the situation fifteen years ago: there is more knowledge and better concentration of expertise; more multi-disciplinary work; more familiarity with and interest in these diseases; more research; better diagnostics; more co-operation; better advice being given; etc.

Following group discussion of these various aspects, the key conclusions of this first session of the round-table were as follows:

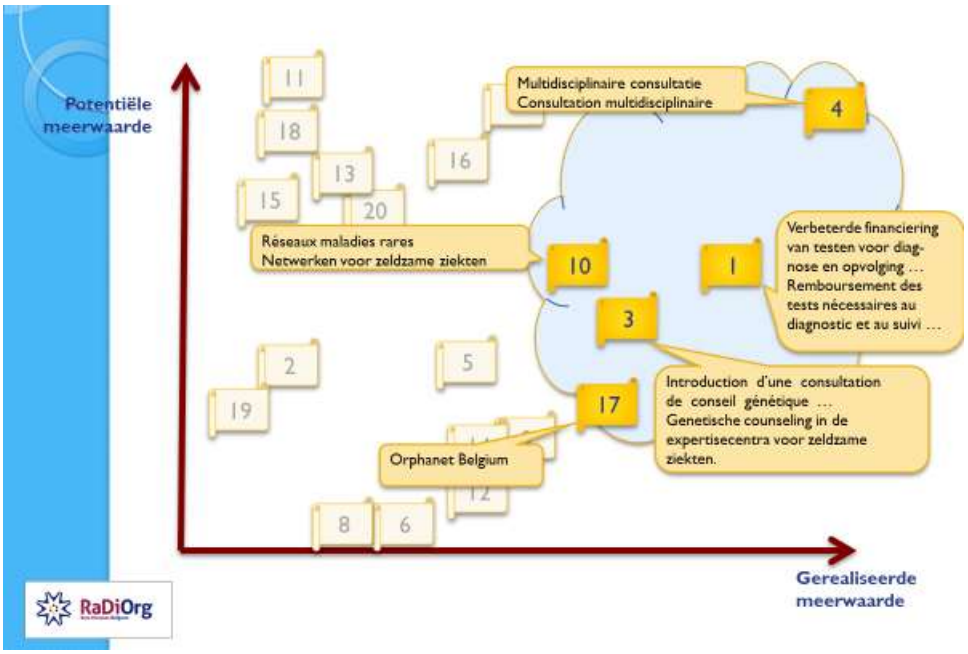
- It is important to make mention of the launch of the European Reference Networks and the Flemish Network for Rare Diseases, and the start of efforts to set up a similar network in Wallonia, as major advances made in Belgium for the care of these diseases.
- While networks will facilitate co-operation and communication, they will not necessarily help concentrate expertise. These networks will, for example, facilitate communication between first-, second- and third-line care, which is essential for looking after rare disease patients properly.
- Although patient organisations do acknowledge that improvements have come about at the overall level (policy and administrative decisions), on the ground, these improvements are still not tangible to patients in terms of care and follow-up.
- In order for rare disease patient needs to be seen to more quickly by public authorities, there must be a more pronounced engagement and a truly proactive stance on the part of the authorities, particularly in financing the measures provided for in the Rare Diseases Plan, comparably to what is done for cancers, for instance.
- For patients, the connection with first-line care is really very important.
- There needs to be wide scale communication on rare diseases to demystify them, and again this is comparable to what has been done in the past for cancers. To this end, it would be useful to determine the total number of patients who fall under the generic term “rare disease”. This is the main way by which rare diseases can be expected to become a priority in funded public health. More specifically, this awareness-raising among the general public might help nudge public choices, fostering a willingness to pay for treatments and care for rare diseases.
- We need to come up with innovative reimbursement mechanisms for orphan medicines; the currently-applicable contractual negotiation mechanism (Article 81) is unsustainable. Moreover, besides orphan medicines that require major funding, there are promising genetic therapies which in future will exert more pressure on the scope for reimbursement.

THEME 2 : WHICH ACTION POINTS IN THE RARE DISEASES PLAN HAVE BEEN MOST EFFECTIVE IN IMPROVING PATIENTS' SITUATION?

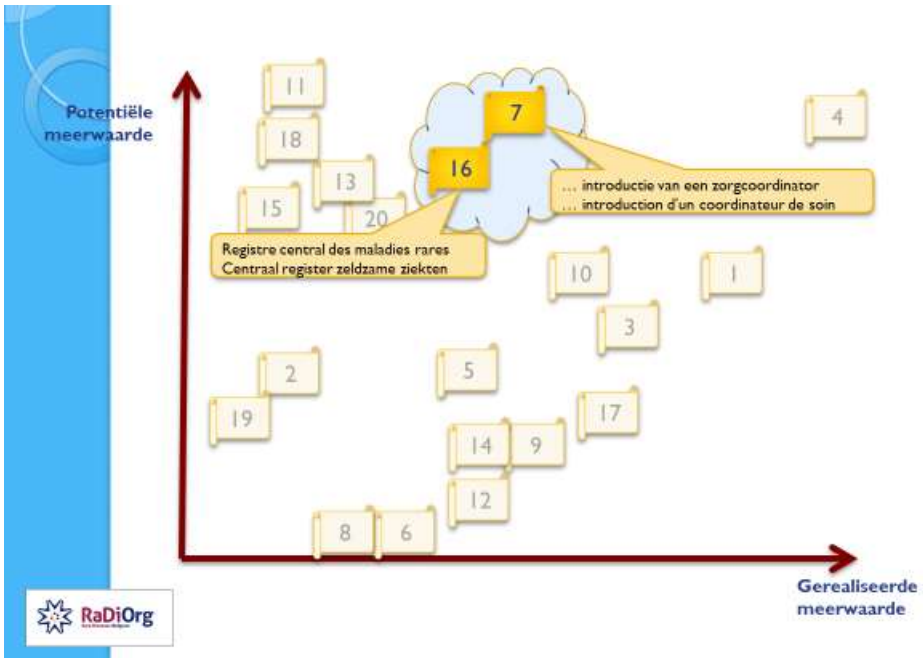
The Rare Diseases Plan[iii] is made up of 20 action points. The aim of this session is to map the actions (whether achieved or not) in the Rare Diseases Plan and to identify their expected or actual impact for patients.

Once again, this exercise was the subject of a prior consultation of participants. The findings of that consultation were presented for discussion in this session.[iv]

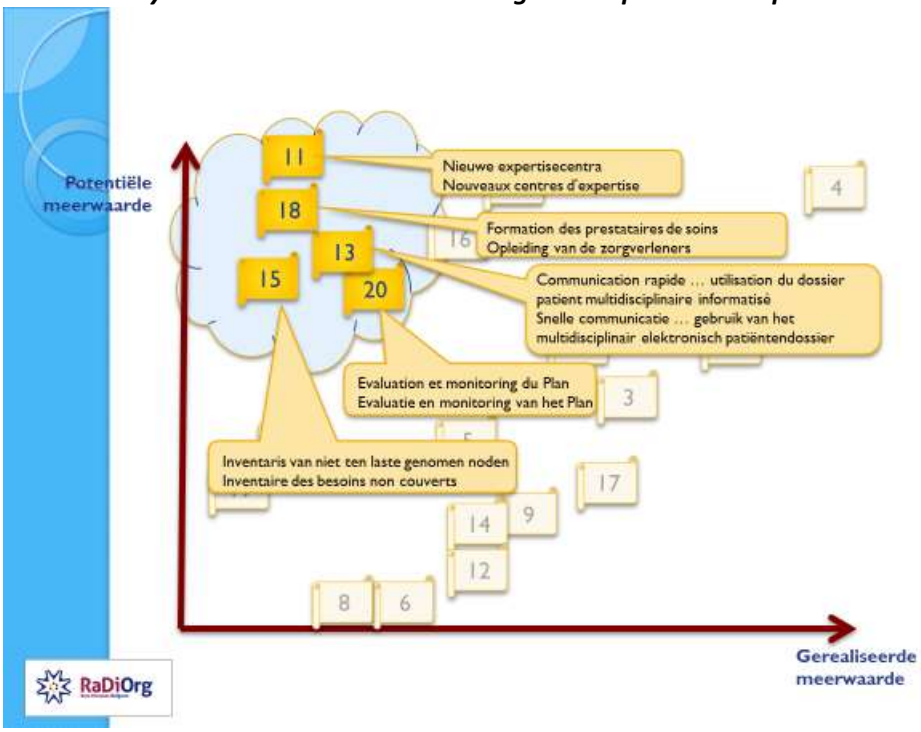
(1) The most effective actions already accomplished up until now



(2) The actions presently being taken that have the greatest potential impact



(3) The actions yet to be taken that have the greatest potential impact



Following group discussion of these various elements, the key conclusions of this second session of the round-table were as follows:

- As regards Orphanet, it seems useful for professionals but not yet sufficiently so for patients; for this cause, it is vital that information be expressed understandably for patients (adapt the language used to make it comprehensible to all). It is currently a stagnant tool but it has great potential for improvement.
- There is an importance, felt by all parties, that data and records should be harvested. However, it seems that a whole series of obstacles would have to be cleared for the central register of rare diseases to be better filled out. Some of these obstacles are already known (e.g. the lack of a professional obligation to enter patients' data into it; the lack of availability of technical tools at hospital level; the poor quality of the data reported), but we need to identify all of the obstacles that have to be addressed.
- Developing centres of expertise is of prime importance, but we also have to set up collaborative working systems and communications between those centres and non-expert centres if patients' lives are to be made easier. Defining guidelines and care pathways could help establish these connections between the different levels of care.
- We need to define what we mean by "expertise": are we talking about diagnostic expertise, care expertise or other expertise? Is expertise determined by the number of patients seen, the number of specific medical interventions performed, or indeed the size of participant base in research projects or networks, or by something else?
- Grouping rare diseases (as is done for the European Reference Networks) could help concentrate expertise more realistically, since it is impossible to set up a centre of expertise for every rare disease there is.
- We absolutely have to reach a balance between concentrated centres of expertise and health carers close at hand. This will address the problem of how to move patients towards the appropriate expertise. To achieve this action point, it will be essential to invest in carer training, including the training of immediate carers.

- The role of care co-ordinator is a fundamental one for patients. This concept needs to be understood as one encompassing more than just medical and paramedic care, so as to cover all aspects of the care of patients affected by rare diseases.
- Non-covered needs, including “unmet medical needs” (measure 14 of the Belgian plan for rare diseases), must continue to be a priority in the objective of improving quality of life for patients with rare diseases.
- All parties involved must work together to exert pressure against price hikes for orphan medicines.

THEME 3: WHAT SUPPLEMENTARY ACTIONS ARE NEEDED IN THE RARE DISEASES PLAN?

Finally, in the third session in this round-table, participants were invited to name new action points that should be included in any update that might be made to the Rare Diseases Plan or if a second Rare Diseases Plan should be adopted. The following were the main themes mentioned:

- Support provided by public authorities to RaDiOrg — the national association for rare diseases — ought to be bolstered, so as to afford it the same degree of legitimacy as other platforms, such as LUSS or VPP.
- State priorities for particular pathologies.
- More transparency on how orphan medicine prices are determined, and how innovative mechanisms are identified, so as to ensure they are developed and ultimately that the costs are reimbursed.
- There is an equally important need to state objectively all the needs of patients besides those seen in a purely medical perspective. Meeting these other needs is just as vital to ensuring good quality of life for patients affected by rare diseases.
- Reinforce the linkage between first- and second-line health care.
- Boost and improve the 20 measures already being taken.
- Ensure that patients have access to the right information; signpost patients towards patient associations (at the same time strengthening RaDiOrg’s role as the national alliance for rare diseases).
- Aim to achieve better cover in specialized beds, down to a more local level.
- Integrate rare diseases into the Tackling Chronic Diseases Plan (*Suivi des maladies chroniques*) more specifically: it is simply impossible to gather “rare disease” expertise within each cluster of “chronic diseases”, since “rare diseases” often are also “chronic diseases”.
- Better training for immediate caregivers, and ensure they have access to effective, usable guides.
- Formalise patient-doctor relationships and foster more exchange placements between them.
- Work up and implement a communications plan for the general public, particularly raising awareness of just what percentage of the whole population is affected by “rare diseases” as a general concept.
- Integrate rare diseases into care courses (e.g. nursing and general medicine courses).
- Allow orphan medicines to be administered by routes other than hospital pharmacies.
- Arrange follow-up for general health objectives stated by the government, such as the health gap (an issue included in general objectives for public health), which is a problem probably even more pronounced with rare diseases.

III. Conclusions

Conclusions are gathered at the end of each theme in the text above (bullet points format).

IV. Appendix

(1) Agenda of the meeting

13.00 Welcome
13.05 Introduction and aims of this round-table
13.15 What aspects are unique to rare diseases?
13.45 Which action points in the Rare Diseases Plan have been most effective in improving patients' lot?
14.40 What supplementary actions are needed in the Rare Diseases Plan?
15.00 End of round-table

(2) List of participants

Name	Institution
Albert Counet	Ligue Huntington (Huntington's Disease Patient Association)
André Loir	Federal Agency for Medicines and Health Products
Bénédicte Gombault	King Baudouin Foundation
Benoit Mores	Minister's Office at Federal Service of Public Health
Chantal De Boevere	Conference of Belgian University Hospitals
Chantal Mathy	RIZIV/INAMI National Institute for Health & Disability Insurance
Chris Van Haecht	Mutualité Chrétienne (health insurer)
Chris Van Hul	Collège intermutualiste (Health Insurers' Association)
Christine Verellen	Institute of Pathology and Genetics
Danny Reviers	ALS (Motor Neuron Disease)
Eva Schoeters	be-TSC (Tuberous Sclerosis Complex)
Fanny Duysens	Doctoral student, Social & Political Science (University of Liège)
Freddy Raymackers	UZ Brussels (University Hospital)
Geert Mortier	UZ Antwerp (University Hospital)
Geert Peuskens	Flemish Network for Rare Diseases
Hilde Ketels	BOKS (Metabolic Diseases Patient Association)
Jonathan Ventura	RaDiOrg (Rare Diseases Organisation Belgium)
Ken de Marie	MUCO (Cystic Fibrosis)
Luc Matthysen	HTAP (Pulmonary Arterial Hypertension)
Marc Dooms	Hospital pharmacist
Maria Barea	VASCAPA (Vascular Anomaly Patient Association)
Mimi Minsiemi	Sickle Cell Diseases
Muriel Mignolet	Sanofi/Genzyme (Lysosomal Storage Diseases)
Peter Raeymaekers	*Moderator*
Romain Alderweireldt	Marfan/f101g (Marfan Syndrome / 101 Genomes Foundation)
Saskia Van den Bogaert	Federal Service of Public Health
Valentina Bottarelli	Eurordis (Rare Diseases Europe)

[i] https://eur-lex.europa.eu/legal-content/EN/TXT/HTML/?uri=CELEX:32009H0703(02)&from=FR

[ii] https://www.eurordis.org/IMG/pdf/Princeps_document-FR.pdf

[iii] https://www.health.belgium.be/sites/default/files/uploads/fields/fpshealth_theme_file/plan_belge_maladies_rares.pdf

[iv] This was a straightforward consultation aimed at preparing for the round-table discussions, with no scientific input. At most, 25 people from the various participating bodies took part in this consultation.