Rare Disease Day 2009

Rare Disease Day

28 February 2009

INFOPACK

"Patient care, a public affair"

Merel, Epidermolysis Bullosa

Winner of EURORDIS Photo Contest 2008

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EURORDIS
Rare Diseases Europe

Contents

1. Introduction
2. Theme for 2008-2011
3. Focus 2009
4. Organisation principles
5. Country by Country
6. What are the objectives?
7. What will happen at European level?
8. Tools
    a. Website
    b. Logos, poster & bookmark
    c. Fact sheets:
        - What is a rare disease?
        - What is an orphan drug?
        - Rare disease patient groups in the EU
        - Paediatric drugs and rare diseases
    d. Accessories
    e. Press Release

9. Annexes

a) Agreement between EURORDIS and participating organisations
b) Agreement between National Alliances and participating organisations
c) Review of events and media coverage
d) Declaration of Common Principles on Centres of Expertise and European Reference Networks
e) Executive Summary of Eurordis Care Survey book

1. Introduction

- **Read the whole pack**

It will give you background information on Rare Disease Day 2009. It will help you understand how we can achieve our main goal of awareness raising

- **Spend time thinking about the theme that was chosen for 2009**

If necessary, according to the rare disease environment in your region or country, think about a sub-theme or subtitle that would translate the general theme to your own conditions.

- **Slogan**

Translate the slogan in your own language

- **Specific targets**

Decide on specific targets for Rare Disease Day 2009 for your country or region

- **Suggestions for local events & activities**

Think about which ones you could organise in your region or country

- **Complete, personalise and translate the press release**

The press release is to be sent to the media in your region or country

- **Download logos and posters**

Download from www.rarediseaseday.org

- **Use the whole pack or only the parts relevant to your region**

You may need to translate it in your own language to share it with your members

For any questions, please contact Anja Helm anja.helm@eurordis.org

2. Theme for 2008 - 2011

"Rare Disease: a Public Health Priority"

The theme was chosen in 2008 for its broadness. Some of our main targets, the general public in particular, are not yet aware of what rare diseases are, what impacts they have on patients' lives, and what needs to be done.

This concept needs to be looked at in the current context of development of rare disease legislation, both at European and national levels: **European Commission Communication on Rare Diseases**, development of **national plans for rare diseases** and development of **European reference networks of Centres of Expertise.**

This year's theme encompasses these issues, which must be adapted to local and national environments. For example, some countries have a national plan in place, others are in the process of creating one, and others don't have anything yet.

- The rare disease patient is the **orphan of health systems**, often without diagnosis, without treatment or research
- **Rare diseases are life-threatening or chronically debilitating diseases** with a low prevalence and a high level of complexity. 6000 - 8000 rare diseases have been identified, affecting 30 million European citizens

  Patients with very rare diseases and their families are particularly isolated and vulnerable. Life expectancy of rare disease patients is significantly reduced; many have disabilities that become a source of discrimination and reduce or destroy educational, professional or social opportunities
- **Research on rare diseases is scarce**
- **Lack of specific health policies and the scarcity of expertise** translate into delayed diagnosis and difficulty of access to care. The national healthcare services for diagnosis, treatment and care of rare disease patients differ significantly in terms of their availability and quality. European citizens have unequal access to healthcare services and to orphan drugs
- Since 1999, the European Union has taken measures to fight against rare diseases and their impact on patients' lives, and has made **rare diseases a priority of its public health programmes**: EU Regulation on Orphan Medicinal Products (1999), EU Regulation on Pediatric Drugs (2006), EU Regulation on Advanced Therapies (2007), Community Action Programme in the Field of Public Health (1999-2003), Community Action Programme in the Field of Public Health (2007-2013), EU 7th Framework Program for Research (2007-2013)
- Over the last few years, a certain number of Member States have developed **National Plans for Rare Diseases**

3. Focus 2009

"*Patient-Centred Care*"

EURORDIS advocates for the best interest of patients at all times, they are at the centre of all our activity. Patient satisfaction is our goal. Patient-centred care is the right care!

Through the publication of the EurordisCare Survey Book, "The Voice of 12,000 Patients", the patient's perspective can go beyond patients' anecdotes and be additionally represented by the analysis of data collected through the EurordisCare2 and EurordisCare3 surveys.

These surveys directly investigated patients' experiences and expectations regarding access to diagnosis and to health services, for a variety of significantly relevant rare diseases across Europe.

EURORDIS has and will continue to organise discussions empowering patient representatives in seeking multidisciplinary and comprehensive care, as well as promoting adequate public policies in 2009, through:

- Adoption of the "Declaration of Common Principles on Centres of Expertise and European Reference Networks" by EURORDIS at its European Membership Meeting in May 2008 in Copenhagen (a list of key functions, proposed and agreed on by patients, to be included in the functions of all Centres of Expertise).
- Promotion of a policy on Centres of Expertise and European Reference Network both in the *Directive on Cross Border Health Care and Patient Mobility*, as well as in the *European Commission's Communication on a European Action in the field of rare disease.*
- Participation in the work of the *EU High Level Group on Health Services and Medical Care* as well as in the *DG SANCO Rare Disease Task Force*
- Participation in EUROPLAN, (European Project for Rare Disease National Plans Development), a DG SANCO funded project aimed at developing recommendations defining a strategic plan for rare disease at national level. This will result in a patient-centred contribution to the development of a key tool in the implementation of one significant recommendation in the European Commission's Communication on a European action in the field of rare diseases
- Close work with its 16 national rare disease alliances on national policies on Centres of Expertise as well as with its 25 rare disease specific federations on European Reference Networks
- Collaboration with the pilot European Reference Networks supported by the European Commission (often involving EURORDIS as a partner or advisor)

Through these intense efforts EURORDIS speaks out on behalf of all rare disease patients in Europe by promoting a sound European Union policy for European Reference Networks of Centres of Expertise.

4. Organisation principles

Rare Disease Day is an annual, awareness-raising event co-ordinated by EURORDIS at the international level and National Alliances at the national level. Who can take part? Anyone who wants to: National Alliances, Patient Organisations, Centres of Expertise, NGOs, Ministries of Health - the more, the better!

Role of EURORDIS:

- Decision on dates, themes and content
- European coordination of Rare Disease Day
- Management of agreements with international organisations or organisations in countries without a National Alliance for Rare Diseases
- Management of graphic identity (logo, poster, visuals)
- Rare Disease Day website
- Development of tools
- European-level event (in Brussels)
- Evaluation of results and analysis at European level

International organisations or partners in countries without a national alliance should sign the "Letter of Agreement between Eurordis and its partners for participating in the Rare Disease Day" **(Annexe A)** with **EURORDIS** and send it to anja.helm@eurordis.org with the list events they would like to see featured on the Rare Disease Day website

Role of National Alliances:

- Coordination at national level
- Management of agreements with organisations at national level
- Finding a patron and obtaining video or written message of support
- Contacting MEPs and candidate MEPs to invite them to visit Centres of Expertise in their country
- Updating of information and events on www.rarediseaseday.org
- Adaptation of common tools and development of own tools
- Funding for local actions
- Data gathering (results) & evaluation
- Press / media contacts

All other **participating organisations in countries** where a National Alliance for rare diseases already exists, should contact their National Alliance to coordinate plans, in order to maximize the success of the event. Participating organisations shall sign the "Letter of Agreement between National Alliance for participating in Rare Disease Day 2009" with their **national alliance (Annexe B)** and send it to this alliance with the list of events they would like to see featured on the Rare Disease Day website.

4. Organisation principles

National Alliances participating in the Rare Disease Day 2009:

Country	Alliance	Telehone	Contact e-mail
Argentina	GEISER	54 261 424 00 76	virginialejandrallera@yahoo.com
Belgium	RaDiOrg	32 498 70 15 03	lut@boks.be
Bulgaria	NAPRD	359 888 323 748	tomov@gaucherbg.org
Canada	CORD	1 416 969-7464	durhane@sympatico.ca
Croatia	Rijetke Bolesti	38 1 481-28-46	rijetke.bolesti@gmail.com
Denmark	Rare Disorders Denmark	45 33 14 00 10	mail@raredisorders.dk
France	Alliance Maladies Rares	33 1 56 53 53 40	lecerf.chantal@wanadoo.fr
Germany	ACHSE	49 30 33 00 7080	mirjam.mann@achse-online.de
Greece	PESPA	30 210 76 60 989	tsahellas@ath.forthnet.gr
Hungary	HUFERDIS	36 1 326 74 92	pogany@williams.ngo.hu
Ireland	GRDO	353 709 30 50	avril.daly@fightingblindness.ie
Italy	UNIAMO	39 0412410886	bellagambi.estero@uniamo.org
Luxembourg	ALAN	352 266 112 1	bvogel@pt.lu
Netherlands	VSOP	31 35 603 40 43	c.oosterwijk@vsop.nl
Romania	RONARD	40 260 616 585	doricad@yahoo.com
Spain	FEDER	34 91 534 43 42	direccion@enfermedades-raras.org
Sweden	Sällsynta diagnoser	46 8 764 49 99	dammert@sallsyntadiagnoser.nu
UK	Rare Diseases UK	442 077 043 141	Melissa@raredisease.org.uk
USA	NORD	1 203 744 01 00	mdunkle@rarediseases.org

4. Organisation principles

Rare Disease Day 2009 is flexible at national/local level:

Depending on individual and national environment, National Alliances can choose to organise Rare Disease Day around one single day, one weekend, several days, or a week. It is preferable for the day to happen every year in a given country. However, a National Alliance could decide differently. Some National Alliances may prefer not to make the day a fundraising event. EURORDIS suggests a variety of tools for National Alliances, which they are free to adapt and translate if necessary. Target audiences may vary from country to country.

Common features:

VIP/ Patron/First Lady

National Alliances are encouraged to secure the support of a patron: a first lady, royalty, VIPs or other well know persons in their country. The role of the patron is to provide a message of support, written or video, to the national alliance, to be displayed on www.rarediseaseday.org and given to the media.

At a national level, patrons may host conferences, receptions, or give out awards, depending on what is agreed upon between the national alliance and the patron.

Member of the European Parliament

National Alliances are encouraged to contact their MEP or candidate MEP (especially those following health issues) to visit a Centre of Expertise. This could be an opportunity to invite your health minister, other politicians and media.

Who is Rare Disease Day 2009 for?

Primary targets:

- European Institutions, authorities & policy makers
- National institutions & policy makers
- Media
- Centres of Expertise
- Pharmaceutical industry
- General public

5. Country by country

Activities taking place around Rare Disease Day in each country are the decision of the National Alliance. EURORDIS suggests the following possible activities. These may vary depending on your resources, time, strategy and available funds.

Coordinate letter writing or email campaign to local or national policy and decision makers. Urge them to act for people living with rare diseases. The topics covered in the letter need to be adapted to the particular situation of rare disease patients in your region/country

Send a press release to the media

Organise a visit of an MEP/ health minister to a Centre of Expertise. This could be done with patient representatives, and the media should be invited.

Organise interviews with the media to talk about the situation of rare disease patients (magazines and newspapers, television, radio)

Organise events around rare diseases and the topics at heart of patients (conference, workshops, walks, demonstrations, sports events etc)

Approach a special/famous person to be the patron of the day or use the existing patron of your organisation

Give awards to people who are recognised for having acted effectively or outstandingly for the cause of rare disease patients

Hold a competition centred on rare diseases: photo competition, art contest, essays etc

Set up a help line to respond to enquiries or promote your existing help line

Display posters, images or other awareness raising displays

Distribute stickers and flyers to people on the street, in schools, hospitals, or universities

Make appointments to meet with your local and national authorities

Organise fundraising events or a special fundraising campaign: Collect donations from people on the street/Collect donations from your website/Organise a fundraising dinner with a special guest/Special mailings/Sell specific items/Approach corporate organisations for donations or long-term partnerships

6. What are the objectives?

The main objective of Rare Disease Day 2009 is to raise awareness with policy makers and the public about rare diseases and their impact on patients' lives. Thus the aim of the day is to reinforce their importance as:

(i) A public health priority

We have succeeded in making rare diseases a public health priority in the EU. However, for the period 2008-2013 a new **Public Health Programme** should replace the existing one (if approved by the Council & Parliament). Therefore we need to make sure that rare diseases remain a priority of the new Programme.

For more information please download the EURORDIS *Princeps* document:

http://www.eurordis.org/IMG/pdf/princeps_document-EN.pdf (ENGLISH)

http://www.eurordis.org/IMG/pdf/princeps_document-FR.pdf (FRENCH)

http://www.eurordis.org/IMG/pdf/princeps_document-GM.pdf (GERMAN)

http://www.eurordis.org/IMG/pdf/princeps_document-IT.pdf (ITALIAN)

http://www.eurordis.org/IMG/pdf/princeps_document-SN.pdf (SPANISH)

(ii) The need for Centres of Expertise

For more information please read Annexe D or download the Declaration on Centres of Expertise, which is available on the Rare Disease Day website.

http://www.rarediseaseday.org/article/index/id/1

Other objectives of Rare Disease Day are to:

- Raise awareness on rare diseases
- Strengthen one voice of patients
- Give hope and information to patients
- Bring stakeholders closer together
- Coordinate policy actions in different countries
- Inspire continued growth of the awareness of rare diseases
- Get equity in access to care and treatment

7. What will happen at European level?

Events in Brussels:

EURORDIS is organising events in the European Commission in Brussels on the week of the 28th of February 2009.

Its objectives are to stress the importance of the role of the European Commission and of the EU in fighting against rare diseases in Europe. A press release will be issued by EURORDIS.

Press conference in the European Commission. The press conference will include two presentations:

1. EURORDIS' *Declaration of Common principles on Centres of Expertise and European Reference Networks for Rare Diseases*.

2. Publication of Eurordis Care Survey book: "The Voice of 12,000 Patients - Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe"" (see executive summary in background information on this Info Pack)

It is anticipated that European Commissioner for Health, Androulla Vassiliou, will act as patron for the European event. The event will directly target European policy makers and the media. Presentations will be followed by a cocktail reception.

Copies of the EURORDIS Care Survey Book will be offered to attendees.

Dinner Debate in the European Parliament

The event will be hosted by MEP and Rapporteur on the Commission Communication Professor Trakatellis (PPE, Greece). The debate will focus on the European Commission Communication on Rare Diseases and the Council Recommendations on Rare Diseases. The dinner will be attended by MEPs, representatives of DG SANCO, DG Research, DG Enterprise, members of RDTF and COMP, patient organisations as well as pharmaceutical industry representatives.

Video Contest

Eurordis is launching it's first ever Rare Disease video competition.
The Rare Disease community is invited to upload original video entries no longer than 10 minutes. Videos can be uploaded here. www.rarediseasday.org
Anyone can submit a video about living with a rare disease. This could include rare disease patients in every day life, in the medical environment or in school/work.

8. Tools

a. **Website**: **www.rarediseaseday.org**

b. **Poster**: can be downloaded from:
http://download.rarediseaseday.org/RARE_DISEASE_DAY_POSTER_2009_&_FONTS.zip

Logos: can be downloaded from www.rarediseaseday.org or send request to anja.hel@eurordis.org

c. **Fact sheets**:

- What is a rare disease? (www.eurordis.org/IMG/pdf/Fact_Sheet_RD.pdf)
- What is an orphan drug? (www.eurordis.org/IMG/pdf/Fact_Sheet_OD.pdf)
- Rare disease patient groups in the EU (www.eurordis.org/IMG/pdf/Fact_Sheet_PO.pdf)
- Paediatric drugs & rare diseases (www.eurordis.org/IMG/pdf/Fact_Sheet_Paediatrics.pdf)

d. **Accessories**

You can order Rare Disease Day bracelets/badges from the following **OR** do it yourself in your country:

Basile Boels
TransEuroGift
VDM International s.p.r.l, 362 Chaussée de Malines, 1950 Kraainem, Belgium
Tel: +32 2 771 73 91
Fax: +32 2 770 58 66
Mobile: +32 485 76 46 48
E-mail: basile@transeurogift.com / www.transeurogift.com

Please quote "EURORDIS" as a reference. Price varies on quantity. See:
http://www.transeurogift.com/price_list_EN/wristbands_EN.pdf

e. **EurordisCare Book** "The Voice of 12,000 Patients". You will receive a copy if you sign the Letter of Agreement and it will also be available to download on the website.

8. Tools

Press Release

Rare Disease Day

28 February 2009

[*In CAPS = to be filled in by your alliance*]

PLACE, DATE

[***YOUR NATIONAL ALLIANCE***] announces Rare Disease Day 2009 in [***REGION/COUNTRY***] on 28 February 2009.

The main objective of Rare Disease Day 2009 is to raise awareness of rare diseases and of their impact on patients' lives, and reinforce their importance as a public health priority.

Organised by National Rare Disease Alliance members of EURORDIS and partners, it is hoped that the day will help raise awareness of life threatening or chronically debilitating rare diseases and the needs of the patients suffering from them.

"As a direct result of the attention we expect Rare Disease Day 2009 will raise, we hope national healthcare systems improve the availability and quality of diagnosis, treatment and care for rare disease patients throughout Europe", said Yann Le Cam, CEO of EURORDIS.

Rare diseases are chronic, progressive, degenerative, and often life-threatening with high levels of pain. There is no cure today for the 6000 - 8,000 rare diseases, 75% of which affect children.

In [***YOUR REGION/COUNTRY***], there are [***X***] people living with rare diseases. [*Elaborate on situation of rare disease patients in your region/ country, what their needs are, and what is urgent to do*].

[*Contact person details*]

-ENDS-

9. Annexes

ANNEXE A

Rare Disease Day

Letter of Agreement between EURORDIS and its partners for participating in Rare Disease Day 2009

EURORDIS, in collaboration with National Alliances member of the Council of National Alliances, has established the annual 'Rare Disease Day', held on the last day of February every year.

EURORDIS has registered the trademark "Rare Disease Day" with the logo .

EURORDIS is dedicated to promoting the Rare Disease Day at the European and international level. Eurordis has entitled the National Alliances, which are members of Eurordis, to use this Logo and Trademark subject to the conditions stipulated by Eurordis. Furthermore, the National Alliances are entitled to grant use of these to organisations participating in Rare Disease Day 2009 (hereafter the "participating organisations") in their country.

Participating organisations are entitled to use the following materials:

- Rare Disease Day website: www.rarediseaseday.org, featuring all relevant information, events, photos, videos etc.
- Rare Disease Day name and logo
- Rare Disease Day poster
- Rare Disease Day Info Pack + media pack + press release
- EurordisCare Survey Book, "The Voice of 12,000 Patients"

These materials can be downloaded on www.rarediseaseday.org

Participating organisations agree to the following:

1. Use the Rare Disease Day logo as provided by EURORDIS, with no changes
2. Use the theme and focus as defined by EURORDIS and the Council of National Alliances: Theme: "Rare Diseases – a public health priority"/ Focus 2009: Patient -centred care
3. Use the Rare Disease Day poster, including logo, adapting it to their local language but no other major changes
4. Create a link to the Rare Disease Day website (www.rarediseaseday.org) on their website
5. Place sponsor's logos underneath the organiser's logo on the poster, using the following sentence: "with the support of…"
6. Fill out the "Review RDD 2009" and send it to Eurordis for a consolidated report of events and media coverage.
7. Use the logo and trademark only in the context of Rare Disease Day 2009. The status of "participating organisation" has to be renewed each year for Rare Disease Day.
8. Comply with any additional conditions of their National Alliance, e.g. an obligation to submit the actual use of the logo for approval.

All **participating organisations, corporations, public bodies or other entities** coordinate the planning of their activities with the National Alliance, in order to maximize the success of the event. European organisations or organisations in countries without a National Alliance coordinate their activities directly with Eurordis.

Only National Alliances will have access to update their National page on the Rare Disease Day website. Other participating organisations will send their information to anja.helm@eurordis.org for approval and publication on www.rarediseaseday.org.

Name of participating organisation:	Eurordis:
Place:	Place:
Date:	Date:

9. Annexes

ANNEXE B

Rare Disease Day

Letter of Agreement between the National Alliance XY on Rare Diseases recognised by EURORDIS and its partners for participating in Rare Disease Day 2009

EURORDIS, in collaboration with National Alliances member of the Council of National Alliances, has established the annual 'Rare Disease Day', held on the last day of February every year.

EURORDIS has deposited the trademark "Rare Disease Day" with the logo .

EURORDIS is dedicated to promote the Rare Disease Day at the European and international level. Eurordis has entitled the National Alliances, which are a member of Eurordis, to use this Logo and Trademark subject to the conditions stipulated by Eurordis. Furthermore, the National Alliances are entitled to grant the use of the same to organisations participating with the National Alliance (hereafter the "participating organisations") within their specific country for the Rare Disease Day 2009.

Participating organisations are entitled to use the following elements:

- Rare Disease Day website: www.rarediseaseday.org, featuring all relevant information, events, photos, videos etc.
- Rare Disease Day name and logo
- Rare Disease Day poster
- Rare Disease Day Info Pack + media pack + press release
- EurordisCare Survey Book "The Voice of 12,000 Patients"

These elements can be downloaded on www.rarediseaseday.org

Participating organisations agree to the following:

9. Use the Rare Disease Day logo without any changes
10. Use the theme and focus as defined by EURORDIS and the Council of National Alliances: Theme: "Rare Diseases – a public health priority"/ Focus 2009: Patient centred care
11. Use the Rare Disease Day poster, including logo, adapting it to their local language but without any other major changes
12. Link Rare Disease Day website (www.rarediseaseday.org) on their website
13. To fill out the "Review RDD 2009" and send it to Eurordis for a consolidated report of events and Media coverage.
14. To use Logo and Trademark only in the context of the Rare Disease Day 2009. The Status of being a "participating organisation" has to be renewed each year for the Rare Disease Day.
15. To comply with any additional conditions entered into with the National Alliance, e.g. an obligation to submit the actual use of the Logo for approval.

All **participating organisations** coordinate the planning of their activities with the National Alliance, in order to maximize the success of the event.

Only National Alliances will have access to update their National page on the Rare Disease Website. Participating organisations will send their information to their contact partner at the National Alliance for approval.

Name of participating organisation:	Name of National Alliance:
Place:	Place:
Date:	Date:

ANNEXE C

In order to measure and assess the success of RDD 2009, it is important to conduct a review of the events / activities organised, as well as the media / press coverage around that day.

Please fill out this form and send any relevant video and press clippings to paloma.tejada@eurordis.org

Review Rare Disease Day 2009

Your organisation:

Number participating organisations in your country:

Events

Please list events that took place in your country, including date, place & attendance of VIPs, if any.

1.

2.

3.

4.

5.

Political Awareness

- Did you succeed in securing a visit of an MEP/ Health Minister/policy maker to a Centre of Expertise? If so, please give details:

- Did you have involvement from any of your Members of Parliament? Please give details.

Public Awareness

Did you manage to secure involvement of a patron? Please give details.

Increase of phone calls to your alliances/or help line?

Increase in visits to your website?

Increase in membership?

Greatest achievements/highlights of Rare Disease Day 2009:

1.

2.

3.

Any other comments or suggestions?

Media Analysis

Number of articles published in national/regional/local print media: ___
Number of TV interviews or appearances: ___
Number of radio interviews or reports: ___

Date	Media Source Name	Article Title	Print / audiovisual / radio	Highlight / quote

N.B Please send any media articles to EURORDIS: paloma.tejada@eurordis.org

ANNEXE D

Declaration of Common Principles on
Centres of Expertise and European Reference Networks for Rare Diseases

In 2008, EURORDIS adopted this Declaration of "Common Principles on Centres of Expertise and European Reference Networks for Rare Diseases", in order to improve patient care throughout Europe.
Rare disease patients call upon National Health Authorities to endorse, publicise and implement the following Declaration to contribute to the identification of Centres of Expertise and to support them financially.

Rare diseases are often complex diseases

1. Centres of Expertise shall aim at providing a multi-disciplinary approach[1] [2]

2. Centres of Expertise shall aim at providing patient centred-care. Multidisciplinarity shall be managed in a coordinated manner[3], and shall not result in disconnected medical services

3. Centres of Expertise shall represent a reliable source of accurate diagnosis, and shall include genetic testing and genetic counselling

4. Centres of Expertise shall share their competences at both national and European levels[4] and shall endeavour to constantly increase and update their level of expertise

5. Centres of Expertise should join in European Reference Networks for Rare Diseases.

Rare disease patients are too often excluded from health systems and socially marginalised, in spite of their tenacious personal commitment[5] [6]

6. Centres of Expertise shall be places where patients feel welcome and safe [7] and where patients are received by knowledgeable and understanding professionals

7. Centres of Expertise shall facilitate and improve the autonomy of the patient

8. Centres of Expertise shall provide access[8] to social assistance[9], which respond to the special needs of the disease[10]

Centres of Expertise shall not only be "care giving structures", but shall also engage in the following activities:

9. Centres of Expertise and European Reference Networks shall actively involve patients and their representatives in the establishment and performance, management and evaluation of the centre (11). These evaluations should be made publicly available

10. Centres of Expertise shall exchange information with local professionals(12), including general practitioners (13)

11. Centres of Expertise and European Reference Networks shall disseminate information on the diseases to social and other relevant stakeholders involved (14)

12. Centres of Expertise shall provide training (15) to all stakeholders involved, including health care professionals, patients and their representatives

13. Centres of Expertise and European Reference Networks shall provide guidelines on the most appropriate care management for patients, closely integrating both medical and social aspects. They should involve patients and give them an active role as recognised partners at all stages.

14. Centres of Expertise and European Reference Networks shall facilitate the coordination of both basic and clinical research activities and infrastructures, including clinical trials, registries, biobanks, exploration of innovative techniques, etc. They should also be required to publish and disseminate research results, irrespective of whether the results are positive or negative

15. Access to Centres of Expertise must be ensured to all patients, regardless of their country or region of origin

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The following figures are based on the **EurordisCare3 Survey** on access to health services, for which a total of 5995 responses were received from 22 countries for 16 diseases, thanks to the active involvement of 130 patient organisations.

(1) Each patient went through an average of four different types of medical consultation, three kinds of examination and 2.4 types of treatment over the last two years, in relation to his/her disease.

(2) During the same period, almost half of these (47%) spent time in hospital for an average of three times for 20 days in total.

(3) 94% of patients consider that “coordinating the sharing of medical information on the patient between all professionals who care for him/her in the specialised centre” is essential (70%) or useful (24%).

(4) 95% of patients consider that “communicating with other specialised centres and professional networks to harmonise treatments and research at the national and European level” is essential (67%) or useful (28%).

(5) An average of 59% of the respondents (up to 64% for the low income group) had to reduce or stop their professional activity because of their disease or to take care of a relative affected by a rare disease.

(6) On average, 16% of patients (up to 24% for the low income group) were forced to move house because of their disease.

(7) One out of 5 patients (18%) experienced rejection linked to their disease from healthcare professionals. The patient perceived reason of rejection is linked to the disease (80% of cases due to reluctance because of the

complexity of the disease), and/or to the physical conditions of the patient: 10% for disease-related behaviour, 11%, for communication difficulties and 15% for physical aspect.

(8) Every year, 28% of the patients needed the assistance of a social worker. For about one-quarter of these, access to this assistance was difficult: difficult access (18%), very difficult (9%) or even impossible (4%).

(9) 92% of patients consider that “informing patients about their rights and guiding them toward social services, schools, leisure activities or vocational guidance” is essential (55%) or useful (37%).

(10) Globally, social assistance services respond inadequately to the expectations and needs of rare diseases patients (only 37% of patients are satisfied), especially when the demands are specific to the disease: 27% for assistance to obtain exceptional financial support, such as the purchase of a wheel chair, 32% for assistance with social integration, school, leisure or professional integration. This inadequacy of the social assistance is more severe for the low income patients (only 26% of satisfied).

(11) 96% of patients agree that “a specialised centre should involve patient organisations in order to benefit from their knowledge of daily life and needs of patients”.

(12) 90% of patients consider that “creating material for teachers, employers, social services, insurance companies and the general public to inform them about patients’ needs and improve social integration of patients” is useful or essential

(13) 44% of patients disagree that “the role of general practitioners consists mainly in looking after health problems not related to the rare disease”.

(14) 95% of patients consider that “coordinating the sharing of medical information between health professionals of the specialised centre and local health professionals” is useful or essential.

(15) 93% of patients consider that “training local professionals to respond to the specific needs of patients” is useful or essential.

9. Annexes

ANNEXE E

Executive Summary of Eurordis Care Survey book:

"The Voice of 12,000 Patients - Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe"

Rare diseases are often chronic, progressive, degenerative, life-threatening and disabling diseases. Many rare disease patients are denied their right to the highest attainable standard of health and continue to advocate their need to overcome common obstacles. Through the publication of this book, "The Voice of 12,000 Patients", the patient's perspective can go beyond patients' anecdotes and be additionally represented by the analysis of data collected through the EurordisCare2 and EurordisCare3 surveys. These surveys investigated patients' experiences and expectations regarding access to diagnosis and to health services, for a variety of significantly relevant rare diseases across Europe.

Key findings of the EurordisCare2 Survey on delays in, and conditions surrounding the announcement of diagnosis include:

- **25% of patients** reported waiting between 5 and 30 years from the time of first symptoms to a confirmatory diagnosis of their disease
- **40% of patients** were initially misdiagnosed leading to severe consequences such as inappropriate medical interventions, including surgery and psychological treatment
- **25% of patients** had to travel to a different region to obtain a diagnosis and **2%** had to travel to a different country
- In **33% of cases**, the diagnosis was announced in unsatisfactory terms or conditions. In **12.5% of cases**, it was announced in unacceptable ones
- The genetic nature of the disease was not communicated to the patient or family **in 25% of cases**. This is paradoxical, given the genetic origin of most rare diseases
- Genetic counselling was only provided in **50% of cases**

Key findings of the EurordisCare3 Survey on the experience and expectations of rare disease patients on access to care include:

- The average patient required more than **nine different medical services, over the two-year period preceding the survey**. This number ranged from approximately four different medical services to nearly 12 different medical services depending on the disease
- **26% of patients reported difficult, very difficult or impossible access to services.** Lack of referral was the greatest barrier in accessing essential medical services
- **10% of patients** reported that the essential services they sought poorly met their expectations or did not meet them at all
- **29%** of respondents required the assistance of a social worker in the 12 months preceding the survey. While the majority of patients reported easy or very easy access, more than one-third met one with difficulty or could not meet one at all

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Rare Diseases Europe

- **0%** of respondents seeking social services reported that their expectations were only met “somewhat” or even “not at all”

- **16%** of respondents had to move house, usually to relocate to one better adapted to their health needs
- **29%** of respondents reported a patient in their family had to reduce or stop professional activity as a result of their disease and an additional **30%** of respondents reported one member in the family had to reduce or stop professional activities to take care of a relative with a rare disease
- **18%** of respondents experienced rejection by a health care professional. The majority of patients reported a reluctance of professionals to treat them due to the complexity of their disease

Survey participants considered the following functions provided by a Centre of Expertise as the most essential:

- Coordinating the sharing of medical information on the patient between all professionals who care for him/her in the Centre of Expertise
- Communicating with other Centres of Expertise and professional networks to harmonise treatments and research at the national and European levels
- Coordinating the sharing of medical information between professionals of the Centres of Expertise and local professionals, to facilitate the continuity of the patients’ follow-up
- Collaborating with research teams working on the rare disease (in particular for clinical studies)

Survey participants most strongly agreed with the following statements regarding the potential implementation of Centres of Expertise:

- A Centre of Expertise should involve patient organisations to benefit from their knowledge of daily life and needs of patients
- Rare diseases are not well known by the majority of health professionals; it is therefore preferable to travel to a Centre of Expertise for consultations and most specialised care
- To maintain the skills and experience of its professionals, a Centres of Expertise must follow a sufficient number of patients affected by a specific disease
- Rather than concentrating all the expertise and competences in a single national Centre of Expertise, sharing them between several Centres of Expertises would be preferable and more accessible to patients

To address the common challenges concerning timely and accurate diagnoses and accessible and quality medical and social services, stakeholder discussions have revolved around the encouragement of rare disease national plans in each European country in which the organisation of rare diseases care is structured around Centres of Expertise. The creation of European Reference Networks, physical or virtual networking of knowledge and expertise, would provide the potential for a higher European added-value, while respecting the responsibility of Member States for the organisation and management of their health care systems.

EURORDIS has and will continue to organise discussions empowering patient representatives in seeking multidisciplinary and comprehensive care and promoting adequate public policies through:

- a series of national and European Workshops held in 2007 involving nearly 300 participants from 12 countries;

- the adoption of the "Declaration of Common Principles on Centres of Expertise and European Reference Networks" by EURORDIS at its

- European Membership Meeting in May 2008 in Copenhagen; a list of key functions, proposed and agreed on by patients, to included in the functions of all Centres of Expertise;
- promotion of a policy on Centres of Expertise and European Reference Network both in the Directive on Cross Border Health Care and Patient Mobility as well as in the European Commission's Communication on a European Action in the Field of Rare Diseases;
- participation in the work of the EU High Level Group on Health Services and Medical Care as well as in the DG SANCO Rare Disease Task Force;
- participation in EUROPLAN, the European Project for Rare Disease National Plans Development, a Directorate General for Health and Consumers funded project that aims at developing recommendations on how to define a strategic plan for rare disease at the national level will allow a patient-centred contribution to the development of a key tool in the implementation of one significant recommendation in the European Commission's Communication on a European Action in the Field of Rare Diseases.
- close work with its 16 national rare disease alliances on national policies on Centres of Expertise as well as with its 25 rare disease specific federations on European Reference Networks;
- collaboration with the pilot European Reference Networks supported by the European Commission (often involving EURORDIS as a partner or an advisor).

Through these intense efforts EURORDIS speaks out on behalf of all rare disease patients in Europe by promoting good health outcomes and contributing to a sound European Union policy for European Reference Networks of Centres of Expertise.