EUROPEAN COMMISSION
DIRECTORATE GENERAL HEALTH AND CONSUMER PROTECTION

Pan-European Patient Network for Information on Rare Diseases and Orphan Drugs - PARD 3 -

A project conducted by

EURORDIS
European Organization for Rare Disorders

and co-funded by

European Commission

AFM TÉLÉTHON

Under the

Programme of Community Action on Rare Diseases

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Final Activity Report
June 2004

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I - Introduction

This document describes the work carried out under the project entitled "Pan-European Patient Network for Information on Rare Diseases and Orphan Drugs", internally referred to as "PARD 3 project". This project was conducted from December 30th, 2002 to March 31st, 2004.

This is the third part of a long-term project initiated by Eurordis in the framework of the Programme of Community Action on Rare Diseases - 2000/2003, the objective of which is to create and animate a European trans-national network on rare disorders, geared at patient organisations and patients.

The report begins with an executive summary of the project. It is followed by a description of the project management, the activities achieved and the methodology used. Results are then assessed and conclusions drawn.

II - Executive Summary

The objectives of the "PARD 3 project" were:
- to gather information needed to contribute to building a public policy on rare diseases,
- to improve the access to quality information on rare diseases and orphan drugs.

The methodology was based on **a survey**, including both a qualitative and a quantitative phase, **workshops** both at European and national level, **an awareness action** through the first European Rare Disease Awareness Week and **the writing of guidance and pedagogical documents.**

This project involved over 500 organisations from 19 European countries and largely mobilised and interested the rare disease community in Europe around the key topic of information.
The work accomplished is therefore very much the result of sharing experiences. The collaborative work accomplished reflects opinions from all around Europe. Tools are adapted to the European situation whilst taking into account the disparity of administrative and legal contexts between the countries. The project approach was to keep the patient at the heart of the process.

Deliverables planned have been achieved.
- The manual in English, divided into three booklets and guidelines available in 10 languages published are visible and practical tools to achieve improved quality and greater access to information on rare diseases and orphan drugs.
- Pilot training sessions in new European Union member states have enhanced the educational dimension of the project.
- The comprehensive report as a result of the quantitative survey contains valuable evidence from which to distil specific reports and guide future policy for rare diseases.
- Face-to-face contacts with individuals and groups interacting at European level have improved communication and joined-up thinking for policy shaping for rare diseases.
- Each activity attached to the outcomes of this project was achieved with the patient at the heart of the process.

The project has created a solid basis from which information on rare diseases can be further improved. It has initiated a dialogue on the importance of information quality and availability between the various organisations concerned with a common pan-European reference.

III - Progress Report

1. Team

Project Leader
- Lesley Greene, Regional and Development Manager, *C*limb National Information and Advice Centre for Metabolic Diseases (UK) and Eurordis President until May 24th, 2003

Project Coordinator
- Claire Marichal, Eurordis Project and Administrative Officer

Project Manager (from May 2003)
- François Houÿez, Eurordis Project Manager, Access to Care

Information Manager/Webmaster (from August 2003)
- Julia Fitzgerald, Eurordis Information Manager/Webmaster

Additional Steering Committee members
- Elisabeth Kampmann-Hansen, Director, CSH (Danish Centre on Rare Disorders, Denmark)
- Michele Lipucci di Paola, Associazione Veneta per la Lotta alla Talassemia (Italy) and Eurordis Vice-President
- Anne Schaetzel, General Delegate, Maladies Rares Info Service (France)
- Yann Le Cam, Eurordis Chief Executive Officer, Vice-Chairperson of the Committee for Orphan Medicinal Products (France).

Between the submission of the project and its approval by the European Commission, Ms. Youngs, the proposed Project leader, changed jobs and was appointed Director of Public Affairs at the UK Dyslexia Association. As dyslexia is not a rare disease and as her new job was very demanding, it was decided to appoint a new project leader having the experience and skills to take over the project without any additional delays.
As long as the web portal based on the PARD 2 project was not fully operational, it was felt that a more effective way of implementing the staff resources available for this project would be to postpone the recruitment of an Information manager / Webmaster to Summer 2003, and meanwhile to recruit a project manager with expertise in managing European public health actions to assist the project coordinator and project leader, which was done in May 2003.
At the same time, the project coordinator required sick-leave for three weeks, which necessitated the involvement of the Eurordis Chief Executive Officer for that period.
Additionally, some changes were made to the originally proposed Steering Committee in order to involve national helplines as much as possible.

2. Project monitoring

Throughout the project, the Steering Committee was in charge of defining the strategy of the actions and of supervising the implementation of the project. Meetings took place once a month, mostly by phone. They were preceded by agreed agendas and followed up with action points to monitor the project implementation.

In addition, four face-to-face project team meetings took place to discuss in detail the implementation of the strategy defined by the Steering Committee.

3. Project schedule

The PARD 3 project was conducted from December 30th, 2002 to March 31st, 2004. The actions took place as shown in the final schedule below.

Action	**Jan. 03**	**Feb. 03**	**Mar. 03**	**Apr. 03**	**May 03**	**Jun. 03**	**Jul. 03**	**Aug. 03**	**Sep. 03**	**Oct. 03**	**Nov. 03**	**Dec. 03**	**Jan. 04**	**Feb. 04**	**Mar. 04**
European Workshop in Paris															
Qualitative survey: face-to-face interviews in 9 countries															
European Workshop in Namur															
European Awareness Week on Rare Diseases															
Development of guidelines															
Development of manual															
Quantitative survey: questionnaire preparation and fieldwork															
Quantitative survey: result analysis and report															
Pilot test of charter and manual and training sessions															
European Workshop in Paris															
National workshops															
Dissemination															

4. Activities achieved

1. Paris Workshop (17-18 January 2003)

This first workshop had three objectives: design the quantitative questionnaire, validate the methodology of the quantitative and qualitative phases and plan the first European Awareness Week on Rare Diseases.
We indeed preferred to ask our partners for their inputs at design stage rather than at validating stage.

Date : 17-18 January 2003

Location : Paris, France

Chaired by : Lesley Greene (Project Leader)

18 participants : 1-2 participants per partner (Denmark, Italy, France, Spain, Sweden, UK, Belgium, The Netherlands, Estonia), CEO and staff. (5 participants were sponsored by Eurordis).

The project coordinator first gave an overview of the project and the team, emphasising the objectives, the means and the role of the partners and briefly presenting the survey.
Participants were split into five groups to study the structure and wording of the quantitative questionnaire as well as the organisation of the fieldwork and the methodology of the qualitative phase.
After the four groups had reported back the results of their discussions, the European Awareness Week was discussed.

2. Qualitative survey (April-July 2003)

A survey of information practices was the key action to obtain an overview of the situation. It served as a starting point to identify good practices.
As we started contacting market research institutes to discuss the quantitative part of the survey with them, we progressively realised that it would be helpful to conduct the qualitative phase first, instead of starting with the quantitative phase, as planned in the initial project. The results of the qualitative survey indeed helped us to refine the quantitative questionnaire.

We therefore conducted 31 in-depth interviews in nine countries in April and May 2003, using the same methodology in each case.
(See Annex 1a for the list of interviews and Annex 1b for the template of the qualitative interview reports.)

A first synthesis was provided to the participants in the Namur workshop of May 2003.
The transcription work continued until July when interviewers and respondents were asked to validate the interview reports.
Additional interviews planned in Sweden and Portugal could not take place for reasons beyond our control.

A synthesis of the situation by country was finally not made as the data was not appropriate for this exercise.

3. Namur Workshop (23-25 May 2003)

During the Paris workshop of January 2003, the participants decided to organise the next meeting around the Eurordis General Assembly to be held in Namur, Belgium on May 24th, 2003, as it was a unique opportunity to gather so many organisations for two days.

Date : 23-25 May 2003

Location : Namur, Belgium

Chaired by : Lesley Greene (Project Leader)

25 participants : 1-4 participants per partner (Denmark, Italy, France, Spain, Sweden, UK, Belgium, Hungary, Germany, Greece), CEO and PARD 3 Steering Committee.

As this meeting took place just after the series of in-depth interviews in 9 countries, the aim of this workshop was to report on the qualitative survey, analyse its results and validate the work process from interviews to the development of the guidelines.
It was essential for the participants to define what was considered as good practices and what practices should be avoided, as well as what obstacles paved the way to good information and how to overcome them. The final step consisted in drafting a tentative structure for the manual.

4. European Awareness Week on Rare Diseases (24-31 May 2003)

The preparation of the first European Awareness Week on Rare Diseases started in June 2002 at the National Workshop of National Alliances held in Barcelona when the National Alliances were consulted about the type of event we should organise.
It was then further discussed at the Workshop held in Paris in January 2003 and at PARD 3 steering committees early 2003.
Finally, an announcement was sent to Eurordis members on May 6th, 2003 and an announcement was posted on the Eurordis website to facilitate its coordination and implementation.
The European Awareness Week was launched in Namur on May 24th, 2003 on the occasion of the Namur Workshop, and lasted until May 31st, covering events both at Eurordis and country level.
A debrief of this event was included in the European workshop held in Paris in October 2003.

(See Annex 2 for personal profiles of rare diseases)

5. Development of guidelines and a manual (August 2003 - February 2004)

Based on the qualitative interview reports and the discussions of the Namur workshop of May 2003 leading to the identification and analysis of good practices, the project team started working on guidelines and a draft manual (initially called "support and training package").

- The guidelines list the basic rules recommended when handling information on rare diseases. *(See annex 5)*
- The manual lists good practices and practical advice based on the principles listed in the guidelines. It provides advice on how to start and manage an information service, whatever the level of development and the resources. *(See annex 6)*

Both documents were submitted to the participants in the Paris workshop of October 2003.
After groups had worked on both documents, a new version of the guidelines to be submitted to the PARD 3 Steering Committee was validated. Consensus was that this document would be called "Guidelines" rather than "Charter" and that it would comprise "principles" covering the good practices identified.
With regard to the manual, comments gathered during the Paris workshop and the national workshops as well as new content submitted by participants helped create a new version.

As the various comments emphasised the need for both documents to be easy to read and to understand, quotation requests were sent to 6 companies or free-lance designers. Baptiste Ferrier was selected. This graphic designer was commissioned to work on the layout of the documents. The manual was printed as a set of three booklets whereas the guidelines were printed both as a leaflet and as a poster.

6. Pilot sessions (October 2003 - December 2003)

As it was planned to test the guidelines and manual in new member states of the European Union, we decided to organise pilot training sessions in Hungary and Estonia. The principle was to invite rare disease patient organisations representative of a country to a two-day meeting hosted in said country and moderated by 2 trainers from Eurordis.

Objectives of a pilot session were to:

- help by working together, to set foundations for a national alliance of rare disease patient groups;
- help to create a project of access to and delivery of information on rare diseases with minimum quality standards;
- assist participants at face-to-face level to recognise and deliver good information practice.

Hungary: 1st session
The training session took place in Budapest, Hungary, on October 2nd and 3rd, 2003.
10 diseases were represented during the meeting: Williams Syndrome, mucopolysaccharidosis, achondroplasia, cystic fibrosis, phenylketonuria, coeliac disease, Rett Syndrome, Usher Syndrome (deaf-blindness), autism, Down's Syndrome.

Each group made a presentation about their organisation. This exercise helped the participants to learn from each other and to recognise shared problems at an early stage.
After the Eurordis trainers presented the guidelines and the manual, still in a building process, they divided the group into small sub-groups whose task was to order the principles of the guidelines into a triangle to evaluate their ranking. The need to choose rapporteurs helped identify leaders and thinkers.
The participants were also eager to learn about existing national alliances and European institutions. To answer this need in the following training sessions, we prepared specific presentations to tackle both subjects.
An action plan was then completed, based on the guidance provided by the workshop. The action plan focussed on getting to know each other better and on creating a shared newsletter on rare diseases.

Estonia: 2nd session
The second training session took place in Tartu, Estonia, on December 11th and 12th, 2003.
7 rare diseases were represented by patient representatives in the meeting over the two days: Cri du Chat Syndrome, haemophilia, autism, genetic skin diseases, cystic fibrosis, phenylketonuria, cleft palate. One representative of the Estonian Chamber of Disabled People and four health professionals were also present.
Based on the experience gained in Hungary, the programme included case studies and presentations on rare diseases national alliances and European institutions in addition to the presentations made by the participants and that of the trainers on the guidelines and manual.
The action plan they prepared at the end of the meeting was detailed and promising. The aim of the action plan is to increase the provision of information on rare diseases in Estonia. The various steps include identifying human resources, translating the guidelines, establishing contacts with national alliances and centres on rare diseases in Denmark and Norway, starting to build a database, organising an information seminar on rare disorders during 2004 and issuing articles in newspapers.

These meetings confirmed the ability for different groups to work in synergy on tasks with a common goal with mutual benefit. Involvement of the participants was very good and the evaluation forms filled after the training session showed that they had been very pleased with the training.

Portugal: additional session
The national workshop organised in Portugal on February 27th-28th, 2004 included a training session to strengthen the national alliance there. Main features were the presence of 2 Eurordis trainers and the ORPHANET representative in Portugal, a meeting over two days instead of one, presentations by the participants, introduction of the guidelines and manual by the trainers and the development of an action plan.

(See Annex 7 for training package used for pilot session in Estonia)

7. Quantitative Survey (September 2003 - February 2004)

A draft questionnaire in English was first discussed with the partners during the Paris workshop and refined later, taking into account the results of the qualitative survey.

To select a provider for the quantitative survey, we contacted 3 companies with the same requests. Research International was selected. Their role was to finalise the questionnaire, plot the data and analyse the results.
(See Annex 3 for final questionnaire and accompanying letter, Annex 4 for the highlights of the qualitative and quantitative survey.)

Because of the holidays, Eurordis and its partners conducted the fieldwork from the end of September to the beginning of December 2003 in order to optimise chances of a high response rate. Translation was done by the project partners into 7 languages: Dutch, French, German, Hungarian, Italian, Portuguese and Spanish. 1402 questionnaires were sent out, covering 19 European countries. The project partners also advertised the survey and redirected their visitors to the Eurordis website where the questionnaire had been posted.
405 questionnaires were received (response rate=28.9%), of which 372 (i.e. 26.5% of the questionnaires sent out) could be processed. This is a relatively high response rate.
Eurordis fine-tuned the report on the basis of the analysis provided by Research International, highlighting what was behind the figures and the main differences between the regions.

The aim of this fact-finding survey was to provide a more quantitative idea of the state of information services and practices and to highlight obstacles to a good access to information in the various countries.

8. Paris Workshop (17-18 October 2003)

The 7th European Workshop of National Alliances took place in Paris just after the 2nd European Conference on Rare Diseases and Disabilities held in Evry, France.

Date: October 17-18 2003.

Location: Paris, France

Chaired by: Lesley Greene

Participants: 41 participants from 13 countries.

The meeting was opened by an informal dinner during which participants had the opportunity to get to know each other better and exchange experiences of lessons learned during the course of the project. The following day, participants were updated on the project, the charter and manual and the pilot training session that had taken place a few days before in Hungary.

Small groups were built to discuss the charter and the manual, checking their organisation and layout and identifying what was missing, what should be changed and who among the participants felt they could send useful additional contributions for the manual.
After the reports from the various groups, a new version of the charter to be submitted to the PARD 3 Steering Committee was validated. Consensus was that this document would be called "Guidelines" rather than "Charter" and that it would comprise "principles" covering the good practices identified.

The preparation of the national workshops included a detailed presentation of logistics, objectives and timetable as well as exercises to get accustomed to the SWOT methodology and to building an action plan, using the SMART concept. The

improvement of the European Awareness week was taken as an example, which allowed a debrief on that event in a friendly way.

ORPHANET and NEPHIRD representatives were present. We had also invited EMEA, EFPIA, EuropaBio and European Commission representatives but they were unable to attend the meeting at that date.

9. National Workshops (November 2003 - February 2004)

National workshops took place in 9 countries (Belgium, Denmark, France, Germany, Italy, the Netherlands, Spain, Sweden and the United Kingdom) in November and December 2003.

Their main goal was to develop improvement plans at national level.
Each one-day meeting therefore studied the situation at country level, suggested an improvement plan at country level and identified needs for future European integrated projects.
These meetings were also an opportunity for the participants to discuss the implementation potential of the manual and guidelines in their country. All comments were taken into account and implemented according to the context.

Partners were asked to report results, using a template to ensure that results would be comparable. Analysis followed until March 2004.

Key messages are :

- The manual is of generic value for all of Europe but it must be implemented in conjunction with reference to national laws.
- Overall the opportunity to have a reason to come together at a national workshop is of positive value, and discussing access to and quality of information is an attractive reason.
- The subject matter was felt worthwhile because the participants could see the value of improving information delivery.
- The SWOT analysis threw up information into relief about the national alliances that these hadn't identified before.

(see Annex 8 for national workshop synthesis)

In addition, the Portuguese national workshop was held in February 2004 for two days and included a training session on good practices. *(see "6. Pilot sessions")*

10. Dissemination (January-March 2004)

- From the end of August 2003 onwards, i.e. as soon as the Eurordis Webmaster/Information Manager joined the team, we posted on the Eurordis website as much information as possible, particularly for the quantitative survey. Dissemination through the Internet increased the outreach of the project.

- The last phase of the project, at the end of March 2004, was dedicated to the dissemination of:
  - "Guidelines for organisations providing information on rare diseases"
  - "Information services for rare diseases: a manual to guide their creation and development".

In total, 309 manuals and 911 sets of guidelines (posters and leaflets) were disseminated.
We sent printed versions of both documents in English to the partners of the project, the PARD 3 Steering Committee, all Eurordis members as well as the organisations who participated in the face-to-face interviews.
Participants in the quantitative survey as well as contact people in the European administration, the pharmaceutical industry, the media and other interested parties such as the Task Force on Rare Diseases set up on the initiative of the European Commission also received the guidelines.

- In addition to a distribution by mail, the following documents were also posted on the Eurordis website (www.eurordis.org):
  - the English version of the documents,
  - the translation by the partners of the guidelines into 9 languages: Dutch, Estonian, French, German, Icelandic, Italian, Portuguese, Spanish and Swedish,
  - a report on the findings of the survey using the interviews conducted during the qualitative phase and the analysis generated by Research International from the quantitative survey.

- A discussion forum on www.eurordis.org was launched to enable the exchange of comments on the guidelines.

IV - Description of the methodology used

The methodology was based on:

- **a survey**, including both a qualitative and a quantitative phase, to gather data;

- **workshops** both at European and national level, either to gather input or to validate the work done, involving:
  - working groups to benchmark and debate results, thus developing experiences of working together,
  - short presentation exercise, SWOT analysis and development of national action plans using the SMART concept to strengthen existing national alliances,
  - pilot training sessions to test the deliverables and transfer know-how;

- **an awareness action** through the first European Rare Disease Awareness Week;

- **the writing of guidance and pedagogical documents:**
  - training package
  - guidelines
  - manual.

Overall the methodology was inclusive, involving rare disease patient groups and information services throughout Europe. The project actions were open to interested parties and coordinated with major rare disease information providers such as Orphanet and national centres or help lines. Outreach in the rare disease community was ensured by a broad dissemination.

V - Description of the results achieved

1. Deliverables

Expected deliverables	Delivered	Actual deliverables
• Results of the fact-finding survey (quantitative questionnaire + 31 face-to-face interviews in 9 countries)	YES	• Detailed report of quantitative phase • Highlights of both phases • Brief article
• Guidelines on producing and assessing quality of information on rare diseases and orphan drugs	YES	• "Guidelines for organisations providing information on rare diseases" from the qualitative survey
• Identification of best-practices for improved information and access to information for patients	YES	• "Information services for rare diseases: a manual to guide their creation and development", divided into three booklets
• Know-how transfer through support and training package particularly for new national alliances and accession countries	YES	• Training session package created, including an evaluation form
• Strengthening of co-operation between existing national structures	YES	• 3 European workshops of National alliances: January, May and October 2003
• Development of at least 2 new national alliances of patients' organisations	YES	• Creation of a national alliance in Greece, first steps in Hungary and Estonia
• Further development of Web-prototype into an interactive platform structuring the network	YES	• The web portal prototype was further developed during the project and launched online.
• First European Awareness Week on Rare Diseases	YES	• First European Awareness Week on Rare Diseases (24-31 May 2003)

Additional deliverables identified:

- National workshops held in Belgium, Denmark, France, Germany, Italy, Portugal, Spain, Sweden, the Netherlands, the UK: synthesis of recommendations from each national workshop (national and European recommendations)
- Training sessions held in Hungary, Estonia and Portugal.

2. Assessment of the project specific aims

1. Gather information needed to build a public health policy

The idea was to obtain an overview of the situation of information on rare diseases in Europe, to guide those in charge of building a public health policy, particularly with regard to rare disease information provision.
Through a quantitative questionnaire answered by 372 organisations representing 18 countries and in-depth interviews with 31 rare disease patient groups in 9 European countries, data was gathered on the organisations concerned, their needs, sources, tools, services and expectations.

As an insight into results of the **quantitative questionnaire**: 62% of respondents were organisations dealing with a single rare disease, and 5% were dealing with more than 50 diseases. Sources of funding were members (87%) and private donors (69%). National governments only funded 26% of organisations, and the European Commission only 2%. There was a link between source of funding and source of information, demonstrating that funding is never neutral.

When analysing sources of information, it was clear that pharmaceutical industry was a minor source of information (11%) as compared to the rare disease network (52%), or specialist medicine doctors (60%). Patients' organisations were considered as the most reliable source.

Information needs varied by region, reflecting European cultural diversity. Whether regional, national or European-wide, information services were all facing a common difficulty, as language was mentioned as the most frequent obstacle to access information.

Whereas patients themselves often enquire, the mother was much more often quoted than the father (40% versus 2% respectively). Questions most often asked relate to specific information on the disease, current research, and expert doctors.

From the **qualitative interviews**, important recommendations were suggested, such as the transfer of knowledge from patients to patient group/information service. This is essential to ensure the group is the voice of the patients and is developing actions that answer their needs rather than a group driven solely by transfer of professional knowledge to patients which may not meet their needs.

2. Identify and benchmark best practices to improve patients' access to information on rare diseases and the availability of orphan drugs

After carrying out 31 qualitative interviews in 9 countries according to the same questionnaire template, interview reports were collected and a synthesis developed. These were in turn submitted to the project partners gathered at the Namur workshop in May 2003.

- 26 common practices were identified ranging from the structure of the organisation, initial establishment history, use of communication tools, and missions.

- 99 different practices were noted from Northern and Southern Europe.
- 81 observations identifying problems and offering advice were noted

Discussion took place about what constitutes acceptable, unacceptable and desirable practice before splitting into working groups. These working groups were asked to discuss the findings of the survey and from these identify good practices with justification of choices and practices to be avoided.

This intermediate work prepared the actual writing of the guidelines.

3. Develop consensual guidelines around information

Once identified, the good practices were split into three categories: organisational, ethical and procedural principles. A preamble, an introduction and facts about rare diseases were added to build the "Guidelines for organisations providing information on rare diseases".

These guidelines were then validated during the European workshop of October 2003 in Paris and translated into 9 languages (the Danes felt the English version would be understood in their country). 911 sets of guidelines in English were sent by mail. Electronic files were also posted on the Eurordis website in all languages for a broader dissemination.

To disseminate the guidelines in their countries, national alliances suggested to send them to all their member associations who would forward them to their local representatives and to the persons in charge of a helpline.
Some countries planned to send the English version to the big patient organisations in their country and the translated version to the smaller patient organisations.

The comments received from the project partners after the national workshops encouraged Eurordis to start a discussion forum on the guidelines and manual to gather feedback on suggested changes and potential implementation difficulties.

4. Develop guidelines on how to create and manage national help lines

The work planned at first at the level of national help lines was extended to all help lines. By help line we mean any kind of information delivery, whether it is run from a home or from an office, by phone, e-mail or regular mail.

Guidelines relating more specifically to the creation or management of information services were included in the "Guidelines for organisations providing information on rare diseases" under "organisational principles".
Implementation details can be found in the manual: "Information services for rare diseases: a manual to guide their creation and development".

5. Develop a Support and Training Methodology

Objectives of a pilot session were to:
- help by working together, to set foundations for a national alliance;
- help to create a project of access to and delivery of information on rare diseases with minimum quality standards;

- assist participants at face-to-face level to recognise and deliver good practice.

The training package we developed therefore contains:
- examples of the genesis of other national alliances to gain from others' experiences as well as a national action plan template to summarise their discussion on what they want to achieve together
- the guidelines and manual and a presentation of their content and objectives
- case studies to implement the approach explained in the guidelines and manual

but also
- a presentation of the European institutions for accession countries to get acquainted with the roles of the various institutions
- an evaluation form to measure the satisfaction of the participants.

6. Assist each Partner based in the Member States in defining an Improvement Action Plan

During the national workshops, the participants were asked to work on an improvement action plan, following instructions sent by Eurordis on how to use the template provided in the manual.
To prepare this exercise, they were asked to do a SWOT analysis (strengths, weaknesses, opportunities, threats) beforehand. The aim was to take into account the current situation of their organisation at national level so as to be realistic as to the objectives and actions envisaged. In Spain, a comparison was drawn with the SWOT analysis conducted in 2001 and an improvement was stated.

Actions decided at national level cover the following areas:

Information/Communication

- All targets
- Have a "Communicator" in the staff, specialist on how to provide information, an Information and System department, etc.
- Build an efficient communication and image plan with appearance in mass media.
- Work on web page.
- Work on newsletter.
- Organise a conference on orphan drugs.
- Help the Steering Committee on Orphan Drugs set up a database (disease, patient organisation, treatment availability, treatment and/or information expert centres and at a later stage also possibly clinical trials).

- To patients and patient organisations
- Develop a network for very rare diseases.
- Implementation of the guidelines and manual including:
  - IT training sessions for patients with rare diseases or rare disabilities
  - complementing the manual with country-specific advice.
- Publication of an information guide on questions of interest for patients with rare diseases or rare disabilities.

- To health professionals
- Develop a collaboration with health professionals in hospitals, general practitioners.

- Visit hospital coordinator, director, urgencies, social workers, requiring good practice, and for example, a psychologist when diagnosis.
- Promote conferences on rare diseases in hospitals and universities.
- Provide information on rare diseases to make disease and the national alliance known and ask to hand it out to patients with rare diseases.
- Make pharmaceutical industry aware of rare disease patients' needs and of the advantages the have to develop new products for rare diseases.

- To the general public
- Communication campaign directed to the public (in order to improve awareness, obtain external and broad public visibility).
- Organise a press conference.

Lobbying/contacting administration
- For improved recognition at institutional level, particularly at regional level for decentralised administration systems.
- Recommendations to governmental institutions and proposal of legislation changes.
- Focus not only on the heath side of the disease but also on the social side. Advocate also in this issue since most of these disease are chronic, life long, degenerative and disabling.
- Address the subjects in a political way, make a political use of it and lobby the policy makers.

Strengthen the organisation
- Formalise the alliance.
- Expand the base by trying to reach more organisations.
- Improve fund-raising activities.
- Exert the best efforts to build a stronger base of operation for further improvement and expansion of activities of the Alliance.
- Strengthen links between rare disease support groups, get to know them and motivate them for group work.
- Organise a national meeting.
- Keep data base up to date and give the possibility to share data, to contact people with the same pathology.
- Elaborate an Action Plan with the services the national alliance offers its members and internal rules to coordinate the activities of its branches.

Other
- Estimate the number of people affected by rare diseases

7. Identify needs/gaps for future European integrated projects

Participants in the national workshops suggested the following areas of development:

Act at European Union level
- Advocate for the creation of incentives and laws able to bring interests to the rare disease field.
- Advocate for the consideration in the European policy of the social aspects of rare diseases.
- Follow ongoing projects, such as the implementation of the Regulation of Orphan Medicinal Products.

Promote
- More collaboration and exchange among EU countries.

- Incentives to develop new orphan medicinal products and put them in the market.
- Good practices and training: gather further examples of good practice in Europe, disseminate material regarding success stories of patients' organisations in other countries of Europe, organise a pan-European meeting for the presentation of the examples, carry out comparative studies about good practice in health and social assistance (facilities and technical aids) in the various European countries.

Disseminate information
- Comparison of various national and regional legislations.
- How to transform information into knowledge.
- Increase awareness on rare diseases and create a conscience for personal commitment in the "third sector".

Investigate the daily problems of patients affected by rare diseases in the European Union
- Difficulties of access to orphan medicinal products.
- Current inequality in health assistance in the EU countries.
- Reduce delays for diagnosis.

Create and develop reference centres and improve their access for rare disease patients
- Gather all the information on national reference centres, mostly from patient organisations.
- Try to be clearer on the needs (quantitative and qualitative approach). How to regroup different disorders?
- Work on the criteria that will be necessary for the definition of a reference centre.
- Focus on financial problems (travelling from one European country to another where there is a reference centre can be expensive).
- Disseminating this information to the medical sector.
- Find inspiration in other countries, whether European or not.
- Remember that reference centres must deal also with social aspects of rare diseases.
- List the best practices in the field of social care of rare diseases.

Encourage exchanges among National Alliances
- How to guide a new alliance
- Mutual exchange of ideas and knowledge
- Organise convention, meetings and joint special interest workgroups

Increase Eurordis communication
- Increase the information about the activities it carries out, not only in English
- Further develop the Eurordis web portal

Support research proposed by members

8. Use and implement the Website prototype for networking, sharing of information and promoting project aims

During the PARD 3 project, the web prototype was further developed and new content was built but it was put on line only at the end of the project. It could therefore not be used to share information on the project.

However we used the existing Eurordis website to increase the outreach of the project by disseminating information about the Rare Disease Awareness Week and the quantitative survey and by making questionnaires and deliverables available in various languages. The project intermediate report was also posted.
As soon as the new web portal was on line, a discussion forum on the guidelines and manual was launched.

9. Co-ordination and organisation of the 1st European Awareness Week for Rare Diseases

The theme chosen for the first European Rare Disease Awareness Week held from May 24th to May 31st, 2003 was "Patients' Voices".

1. In preparation for this milestone event Eurordis invited National Alliances to contact their membership for offers to be interviewed by local media as a patient with a rare disease.
Attracting media attention in this area remains a constant challenge. Sweden, Belgium, the Netherlands, the UK and Spain sent out press releases. Spanish efforts led to significant activity with interviews with radio, TV and newspapers over two weeks. Two articles appeared in UK papers. There was also coverage in national press about a rare metabolic disorder (MCADD) linked to a court case, and an interview in a popular women's magazine. Belgian TV covered the launch in Namur. Associazione Italiana Sclerosi Laterale Amiotofica participated in a successful TV programme entitled "La Sclerosi Laterale Amiotrofica e le Malattie Rare" on a local channel, STV1.

2. Alternatively, members were invited to send profiles directly to Eurordis in order to create an exhibition at the launch of the week at Namur, Belgium and for oral presentation on the day.
A total of 30 patient profiles were received from 6 EU countries. Eurordis intends to continue to collect profiles focused on living with a rare disease and to create an ongoing electronic Eurordis Registry for enduring record and reference.
The launch itself in Namur was attended by delegates from Denmark, France, Germany, Greece, Hungary, Italy, Spain, Sweden, Switzerland, and the UK. Following the Eurordis General Assembly, participants gathered to send off 180 blue, yellow and white balloons. Each balloon carried a label dedicated to a rare disease, and a request that the finder return it to Eurordis HQ in Paris.

3. Partners were also asked to communicate on the event through their websites, which led to dedicated information being posted on seven websites throughout the period, in addition to Eurordis.

Impact is directly correlated to the resources available for planning and preparing an event and its media coverage.
In view of the response from patients, families and patient organisations, despite a belated request and a clear lack of means, the concept of a European Rare Disease Awareness Week, based on a pan-European grass-roots approach is welcomed.

This awareness week was complemented throughout 2003 by other events. The Netherlands organised special activities for rare diseases during the awareness week for the chronically sick (7-14 November 2003). The European Conference on Rare Disorders and Disabilities (Paris, 15-17 October 2003) also played a major role in driving people's attention to rare diseases. UNIAMO organised the first

awareness day for rare diseases in Italy on August31st-September 1st, 2003 in Venice.

10. Publish and disseminate throughout Europe (a guide) a manual

The "guide" initially planned has been renamed "manual" in order to distinguish it from the "guidelines" that contain principles to be followed when delivering information on rare diseases.

This manual is intended for the creation or the development of information services in the field of rare diseases. It is agreed that no information service can exist or be delivered outside an organisational structure, usually a patient group. It is for that reason that attention is given to establishing a robust organisation that can deliver the service.
The decision was made to write a manual without listing the various differences from country to country but to remind readers that they need to refer to legal requirements specific to their own country.

Objectives
The objectives are to guide the creation or the development of such delivery services, emphasising the main principles, mentioning possible obstacles and proposing tested solutions, i.e. good practices. They focus on specific requirements and issues related to information on rare diseases, and provide some insights on general recommendations for the management of the service. Technicality is not addressed in great detail.
Information in the field of rare diseases is primarily intended for people who are affected by a rare disease or a rare disability, their families and relatives, and also for a broader audience among health professionals and the general public.

Targeted audience
This manual targets people who would like to create information services about a rare disease with little or no support: isolated patients or parents willing to initiate a local or a national programme, patients' groups that are already structured and active but that do not provide information yet. Institutions, medical departments, social services or governmental organisations may be considering creating such information services, and resources exist to support such initiatives.
Patients' groups that are already providing information services will benefit from this manual as they can compare their practices with what counterpart organisations have validated elsewhere.

Sources
Principles and examples of practices described in this manual result from a qualitative survey conducted in most European Union countries from March to May 2003. 31 organisations were interviewed about the information programmes they provide, and the results were discussed during the European Workshop of National Alliances, Namur, May 2003.

Format
To facilitate its use, the manual is divided into three booklets. Its design has been particularly privileged to make it accessible to all. It is also available as electronic files on the Eurordis website.

3. Assessment of the project general aims

The project primarily aimed at

- Improving patients' access to information on rare diseases and the availability of Orphan Medicinal Products, by identifying and benchmarking national and EU best-practices for information on rare diseases and orphan medicinal products
- Developing practical means and recommendations for the improvement of information and its access for professionals and patients

Through these overall objectives, the project also aimed at

- Strengthening trans-national collaboration and network between rare diseases patient organisations
- Strengthening recently created national alliances of patient organisations and developing contacts to create new ones in current and future Member States
- Transferring know-how and support to patient organisations in accession Countries
- Further building consensus and common understanding on key issues addressed by the EU policy on rare diseases and orphan drugs

1. Improving patients' access to information on rare diseases and the availability of Orphan Medicinal Products, by identifying and benchmarking national and EU best-practices for information on rare diseases and orphan medicinal products

Patients' access to information can be difficult in several cases. This project contributed to the improvement of patients' access to information as illustrated in the following scenarios.

- **People don't know they have a rare disease**

Before people obtain a diagnosis, awareness must have been raised for help lines to be identified. For that purpose the manual contains advice for organisations to raise awareness and to face the media and for help lines to be well informed on diagnosis centres.
This project has also identified the need for governments to help to disseminate information through the health network, care centres, social coverage services and schools and for doctors to attend special courses on rare diseases for them to learn to more rapidly refer to a specialist when they cannot provide a diagnosis.

- **A rare disease has been diagnosed but people don't know where to find information and support**

Among the principles listed in the guidelines written during the project, "sign-posting" consists for a helpline in knowing who to refer enquiries to when the answer is not known.
(See above for the need for organisations to raise awareness and for governments to circulate information.)

- **There is no or very little information because the disease is extremely rare**

One of the principles stated in the guidelines refers to isolated people, encouraging help lines to establish structured channels of information for very rare diseases.

Some national alliances or national help lines already offer a service to find other affected people when the disease is so rare that no organisation exists. In the framework of the national workshops organised during the project, additional countries such as The Netherlands for instance, decided to work specifically on that issue. The Eurordis web portal will keep track of the resources available and continue to guide people with a very rare disease in their search for information.

- **People know where to find information but they can't access it:** they don't have a computer or don't have access to the Internet; some web sites are not adapted to some patients' disabilities; differences in languages and cultures are additional obstacles.

Some national alliances already organise some training sessions to the Internet. During the national workshops, additional countries (such as Belgium) identified training sessions as one of the actions of their improvement plan.
Accessibility rules are explained in the manual so that organisations providing information through the Internet make sure their information is accessible to disabled patients.
The project enabled to further develop the new EURORDIS web portal (www.eurordis.org) and to make the multi-lingual ORPHANET website (www.orpha.net) better known in the various countries.

- **People have found information but they cant' understand it or they don't know how to sort the information out**

The guidelines and manual explain how to deliver information, adapting it to life events and age of enquirer.

- **The information they have accessed is not validated**

Among the principles listed in the guidelines written during the project, "Validation" consists for a helpline in revisiting information regularly and checking its validity systematically. The manual details how to do it.

2. Developing practical means and recommendations for the improvement of information and its access for professionals and patients

Beyond the principles listed in the guidelines and the practical recommendations gathered in the manual, we need to emphasise that good practices alone will not dramatically improve the access to information on rare diseases for patients and professionals.
Based on the in-depth interviews conducted in 9 countries, the feedback received from 10 national workshops and the results of the fact-finding survey, we describe below an overview of the situation for information on rare diseases today in Europe.

2.1. Progress made

In most countries, there are many organisations representing the interests of patients. National alliances are representative actors advocating the interests of people with rare diseases, whatever the disease. Due to their experience and their contacts with a high number of affected people, they are well aware of patients' problems. Some of them are well-known by political counterparts. In some cases they have a professional secretariat.

Databases on rare diseases are now available in several languages. New information technologies provide increased communication possibilities. Rare diseases are on the political agenda in some countries. In addition, there is an

opportunity for the rare disease cause to gain a higher profile with the public by integrating the European perspective into national activities.

2.2. Remaining obstacles

- The main obstacle for disease-specific patient organisations, national alliances or national help lines is the lack of finance to support the organisation. This can be due to patients not always realising the value of being members of support groups and contributing to its funding activities, or due to national governments lacking interest in voluntary organisations and the pharmaceutical industry feeling rare diseases provide insufficient market to be worth sponsoring.
- Another major obstacle is the lack of human resources, whether of qualified personnel or people actively involved in the management of the organisation. Even when suitable individuals are recruited, there are problems, both in retaining their skills long-term (because of work-overload or poor salaries) and in identifying successors to continue their roles when they move on.
- It continues to be a challenge to get support for rare diseases at European level.
- The difficulty in accessing the media (this is rare so it will not be of interest to the general population who are our audience) and therefore the lack of awareness by the public are also true obstacles to the improvement of access to information.
- Examples of cooperation between health professionals and patient organisations are still scarce.
- Lack of political power and of strategy are felt as additional weaknesses.
- Some national alliances feel that their activities should be more accessible in regions.
- In some countries an additional obstacle consists in the absence of a national policy on rare diseases because of decentralisation and local autonomies. This can affect funding of organisations and the reimbursement of treatment (post-code prescribing). The fragmentation of jurisdiction both in the medical and political arena makes it difficult to move smoothly at a national level. Moreover politics are often deeply ingrained in the inner mechanisms of institutions thus requiring an extra effort in diplomacy and independence.

For enquirers

- The major problem is the difficulty to access proper diagnosis; it can take years depending on the area.
- Families are scattered, which makes it hard for them to have access to information or to find people with the same disease and hard for the organisations to identify patients.
- Doctors undervalue or do not recognise the importance of information for patients.
- Although a lot of information is available on the Internet, it is hard for people to identify which information has been validated. Texts are sometimes hard to understand.
- Access is not always free
- Some people (older people or people without a computer) can't access the information on the Internet.
- Differences in languages and cultures are additional obstacles.
- Information is still missing on some rare diseases, particularly very rare ones
- Information is lacking on social topics, where to be treated, where to get psychosocial counselling on how to organise life and financial issues.
- Epidemiology data is poor.

2.3. Recommendations

- The European Union should look at national reimbursement policy.
- The European Union should provide funding in a more accessible way to patient organisations.
- National Governments should set up reference centres, walk-in centres and specialised training courses, develop activities in genetic research and provide funding to patient organisations and encourage national public funding.
- Governments should help to disseminate information through the health network, care centres, social coverage services and schools.
- Young researchers should be encouraged to work on rare diseases
- Doctors should attend special courses on rare diseases for them to learn to more rapidly refer to a specialist when they cannot provide a diagnosis
- Nurses and physiotherapists should be more involved, mainly to guide people.
- Pharmaceutical industry should invest part of their profit in less profitable drugs.
- Healthcare insurances should pay more attention to care and treatment of patients with rare disorders
- More cooperation should be encouraged between patient representatives and other help lines or databases as well as with health professionals.
- National alliances should keep raising awareness among the general public, involving more patients to get their stories out to the public and acting as go-betweens with the media as they represent more patients.

- Eurordis will continue with its actions at European level, advocating for better research and treatment and equable access to treatment and for an increased cooperation between states, not only on orphan medicinal products but on all aspects of living with a rare disease.

3. Strengthening trans-national collaboration and network between rare diseases patient organisations

The three European workshops gave the representatives of the rare disease patient organisations present the possibility to talk together about subjects of common interest and exchange experiences of lessons learned during the course of the project.

The publication of the manual giving examples of good practices experienced by patient organisations in other countries of Europe is a very helpful tool in giving motivation to people willing to commit themselves, showing them what can be done, how and where. The documents created by this project will empower these people and give them the right answers to the thousands of questions which will trouble them on the road to excellence in the service of the people with rare diseases in need of their help and guidance.

4. Strengthening recently created national alliances of patient organisations and developing contacts to create new ones in current and future member states

Both in **Hungary** and in **Estonia**, the pilot training session was the first opportunity for rare disease groups to meet together and to learn about each other. The presentation by each participating group helped to recognise shared

problems at an early stage and therefore start building the feeling of a community.
Contacts were developed and an action plan developed to transform this first meeting into the first step of the creation of a common initiative in those future member states.

In **Portugal**, the group started in 2001 had not met since and a management team needed to be set up. The national workshop organised during the PARD 3 project gave patient organisations the opportunity to gather and plan very concrete actions to reorganise the group and to register officially the alliance.

In the **other countries** where national workshops took place, these meetings were also an occasion for members to meet, discuss priorities at national alliance level and obtain consensus about actions to be developed.

The impact of the actions successively conducted since the PARD 1 project was measured by the Spanish alliance who stated a distinct improvement. Some weaknesses and threats have disappeared and some strengths and opportunities have increased. In these two years, Feder´s members have increased considerably, it has now four branches with qualified staff and technical equipment and it has improved resources and awareness, even if insufficiently.

To strengthen the rare disease national alliance in **Belgium**, Eurordis organised the second European workshop and the launch of the first European Awareness week in Namur. It gave the national alliance an occasion to raise awareness among potential funding sources.

In **Greece**, the Greek alliance for rare diseases was officially created towards the end of 2003. Although Greece was not an official partner of the project we invited delegates to the European workshops to develop contacts with other national alliances at an early stage.

5. Transferring know-how and support to patient organisations in accession countries

The manual and guidelines are tools designed to transfer know-how to help lines, in particular those who are just starting. They contain good practices covering what has been experimented in other countries.
We tested these documents in two accession countries: Hungary and Estonia.

In addition to presenting the content of the manual and guidelines, the trainers used case studies to encourage interaction and joint working, and to practice the advice for the various stages of the development of a help line given in those documents.

The need to choose rapporteurs helped identify leaders and thinkers.

The SWOT analysis and the SMART concept for building an action plan were introduced to develop rational thinking.

in Hungary, for instance, developing an action plan as described in the manual and applying this to the creation of a shared newsletter transferred skills to the development of a tool for information delivery on rare diseases.

6. Further building consensus and common understanding on key issues addressed by the EU policy on rare diseases and orphan drugs

Eurordis took the opportunity of the pilot training sessions to include in the training package a presentation on the roles of the various European institutions in the area of rare diseases.
This presentation raised a high interest in the countries where we trialled it, which suggests a specific requirement for further training.
However this topic deserves to be handled as a subject in itself, which was not possible at this stage as information was the main topic of the project.

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4. Additional result achieved: cooperation with other actors in the area of rare diseases

The cooperation initiated with ORPHANET during the PARD 2 project continued. and an amendment to the initial agreement signed in 2002 was signed in March 2004.

Eurordis also signed cooperation agreements with three further projects funded by the European Commission in the framework of the Programme of Community Action on Rare Diseases:

- NEPHIRD (Network of Public Health Institutions on Rare Diseases),
- EUROCAT (European surveillance of congenital anomalies)
- ENERCA (European Network for Rare Congenital Anaemias).

During the course of the project, we involved our partners at several stages:

- During the qualitative phase of the survey an interview was conducted at the National Centre on Rare Disorders in Italy, who co-ordinates the NEPHIRD project.
- During the quantitative phase of the survey, ORPHANET provided a link from its own website to the quantitative questionnaire posted on the Eurordis website.
- The paper version of the questionnaire was also sent to the patient organisations listed in the ORPHANET directory.
- Representatives of ORPHANET and NEPHIRD attended the October workshop held in Paris.
- The representative of ORPHANET in Portugal also attended the national workshop held in Lisbon on February 27th-28th, 2004.
- Meetings were organised with ENERCA during the 2nd European Conference on Rare Diseases and Disabilities held in Evry, France and on January 20th, 2004 in Luxembourg.

(see annexes 9a to 9d for cooperation agreements signed with ENERCA, NEPHIRD, EUROCAT and ORPHANET)

VI - Final conclusions

1. This project was successfully completed

This project involved over 500 organisations from 19 European countries and largely mobilised and interested the rare disease community in Europe around the key topic of information.
The work accomplished is therefore very much the result of sharing experiences. The collaborative work accomplished reflects opinions from all around Europe. Tools are adapted to the European situation whilst taking into account the disparity of administrative and legal contexts between the countries. The project approach was to keep the patient at the heart of the process.

Deliverables planned have been achieved.

- The manual in English, divided into three booklets and guidelines available in 10 languages published are visible and practical tools to achieve improved quality and greater access to information on rare diseases and orphan drugs.
- Pilot training sessions in new European Union member states have enhanced the educational dimension of the project.
- The comprehensive report as a result of the quantitative survey contains valuable evidence from which to distil specific reports and guide future policy for rare diseases.
- Face-to-face contacts with individuals and groups interacting at European level have improved communication and joined-up thinking for policy shaping for rare diseases.
- Each activity attached to the outcomes of this project was achieved with the patient at the heart of the process.

The project has created a solid basis from which information on rare diseases can be further improved. It has initiated a dialogue on the importance of information quality and availability between the various organisations concerned with a common pan-European reference.

2. This project enshrines guidance and tools for improvements

A patient organisation that is setting itself up or launching an information service is not forced to reinvent or rethink everything. It will find in the manual and the guidelines a help to start its activity, taking into account what others have already experienced and learnt. This both saves time and reduces risks of discouragement and failure.

- It is recommended to people to join forces rather than to create additional duplicate organisations, thus increasing the fragmentation of the community.

- By being warned of possible obstacles before they appear, patient organisations can prepare themselves and set up some solutions, even at a small scale.

- By being encouraged to develop the service progressively and to choose paths that are compatible with their resources, they also save money and energy and increase their survival chances. This is the only way to a sustainable and reasonable development.

- The emphasis is laid on the quality of information and on the way it is delivered. Information must be selected and validated. People delivering information must always put themselves in the enquirer's shoes so that they can adapt their answer to what the enquirer can hear and understand. It is indeed not sufficient to deliver some information for it to be understood.

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3. This project opens new perspectives

This project is not the end of an effort but builds firm foundations for future work and carries hopes of new perspectives:

- the hope that as soon as possible there no longer exist obstacles preventing people from accessing the information they need and they can understand;

- the hope that the tools developed guide people through the extreme diversity of the new medias, thus reducing confusion linked to contradictory or false information while still enabling them to exploit the greater fund of resources made possible through these diverse medias (printed newsletters, websites, scientific magazines…)

- the hope of further permanent and stable networking between national information centres or help lines. This should lead to the pan-European observation of rare disease information needs, common best information practices and to more links between people affected by very rare diseases;

- the hope that some professions or people who are reluctant to work together in providing information on rare diseases overcome this reluctance. Even if difficulties in sharing knowledge exist, applying the principles listed in the guidelines should contribute to reducing them;

- the hope that strengthening the role, content and functions of the Eurordis web portal through quality information will contribute to building the rare disease community, in cooperation with other partners.

IX - List of annexes

N°	Type of document
1a	Qualitative Interviews: list of interviews
1b	Qualitative Interviews: template of qualitative interview reports
2	European Awareness Week (May 24th-31st, 2003): personal profiles of Rare Diseases
3	Quantitative Survey: accompanying letter + final questionnaire (English)
4	Quantitative and qualitative survey: highlights
5	Guidelines
6	Manual
7	Training package for pilot session in Estonia
8	National Workshop Synthesis
9a	Cooperation agreement with ENERCA
9b	Cooperation agreement with NEPHIRD
9c	Cooperation agreement with EUROCAT
9d	Amendment to cooperation agreement with ORPHANET