- Actions with authorities and laboratories for NS medication to be recognised and costs reimbursed (e.g. growth hormone effective against impaired growth in children due to corticosteroids)
- Vigilance against the abandonment of low profitability products like levamisole.
- An annual information meeting between families and doctors, researchers

10 TRAINING AND INFORMATION

10.1 Best practice guidelines for care and management

help-lines and written information

When parents and patients are going through the experience of a rare disease, there is important information they need immediately:

- on the accuracy of the diagnosis,
- whether any research is being done,
- where specialists or centres of excellence / centres of reference are to be found,
- what treatment options there are,
- what the future may hold.

SPEAKER

“Lesley Greene,
Climb National Information and Advice Centre for Metabolic Diseases

→ WHO NEEDS TO KNOW?

Patients and parents differ by the rare and ultra rare conditions, different cultures and languages, geographic localisation, etc.
In order to survey the needs for information, Eurodis launched a project in 2003, the PARD3 project, with the support the Rare Disease Programme of Directorate C “Public Health and Risk Assessment” & AFM (Association Française Contre les Myopathies –French Association for Muscular Dystrophy).

→ THE PARD SURVEY

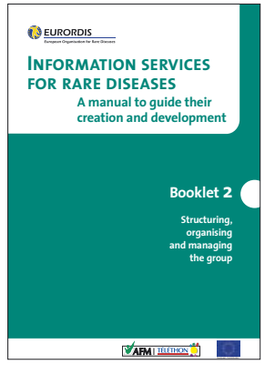
Programme and Actions for Rare Diseases (PARD 3) consisted in:

- A Qualitative Survey interviewing 31 associations
- A Quantitative Survey: analysis of 372 questionnaires returned from 18 countries, an overview of needs, information sources, tools, services and expectations

Two solutions were brought forward:

→ SOLUTION ONE : A MANUAL AND GUIDELINES (see figure 36)

One of the main objectives was to identify the knowledge base for help-lines. There were two main sources: a specialised doctor, and medical advisors. The rare disease network came next (see figure 37). Web sites were only fourth. “Own physician” and “Governmental organisations” were poor information providers for rare diseases: this demonstrates to what extent the rare disease community is lacking in information.

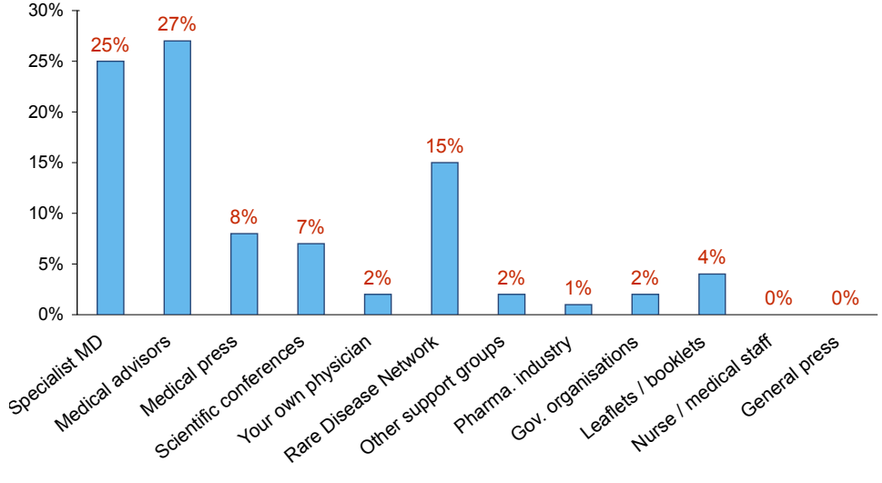


(figure 36)

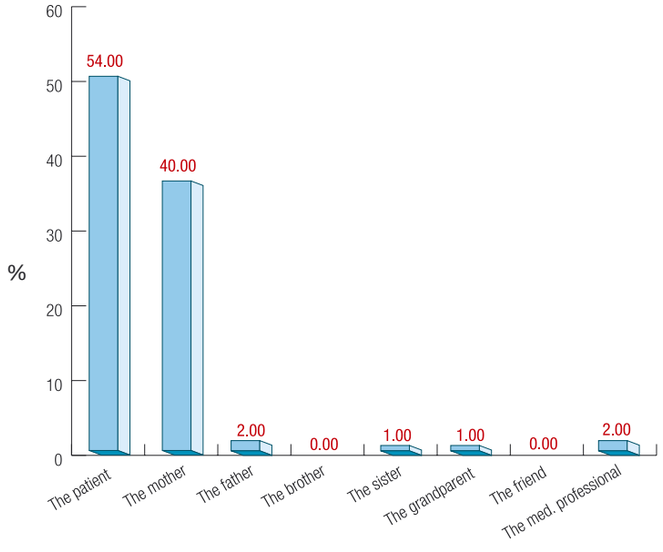
1. Sources of funding

The survey also explored funding sources for services: organisations mainly relied on member and private donors, followed by fundraising events. Private and public administrations represented secondary sources for funding. Countries with regional governments (Spain, Italy) expected the region to be the funding source, and where support for research by groups was greater (Western Europe), industry might have funded more. Governmental organisations and European institutions were a long way down the list. Fragile sources of funding means that any service is going to have problems with consistency in being able to continue to work effectively.

MAIN SOURCES OF INFORMATION (figure 37)



PERSONS WHO ASK FOR INFORMATION (figure 38)

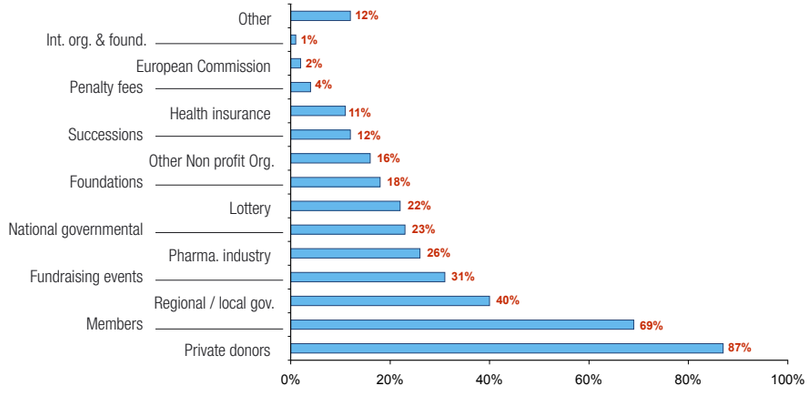


2. Quality of helplines
Another important aspect was to assess how quality of help-lines was achieved and monitored.
63% of help lines used volunteers, and for 81% of help-lines the service was in a confidential area. 86% kept track of the enquirer so they do develop a relationship with the enquirers.

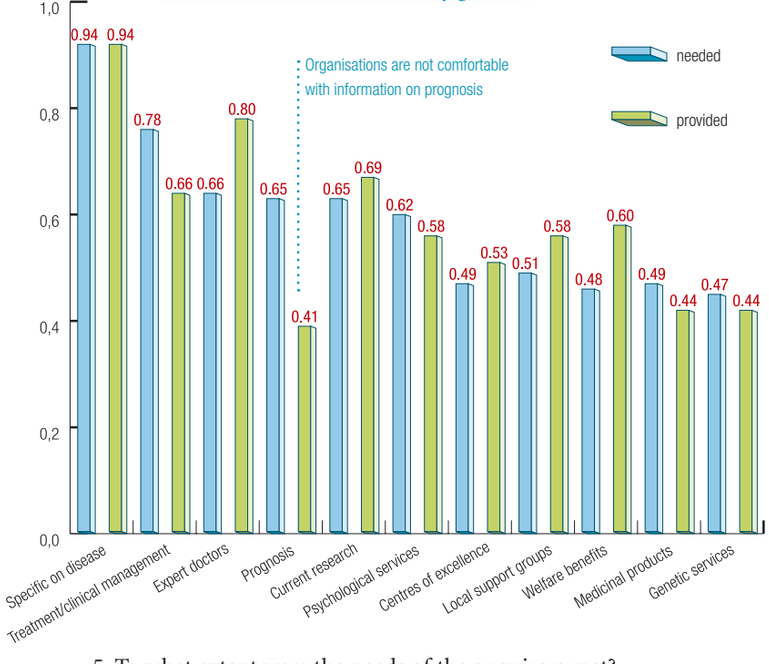
3. Who are the persons who ask for information?
Patients themselves are actively searching for information: they represented 54% of enquiries, even though rare diseases are often disabling. There was a clear difference between mother and father: roles are distributed between the mother as the carer and the father as a source of income for the family. Fathers may sometimes refuse to acknowledge the disease.

4. To what extent were the help lines accessible?
Help lines run at home by unpaid people are available 24 hours per day seven days a week. Once you get paid staff in, probably because the help lines develop the e-force (web site, etc.), then help lines operate at office time.

SOURCES OF FUNDING: percentage of organisations quoting source (figure 39)



INFORMATION NEEDED COMPARED TO INFORMATION PROVIDED (figure 40)



5. To what extent were the needs of the enquirers met?
Overall the needs of the enquirers were met, and the help lines were able to give the information needed. The organisations showed reluctance to discuss or reveal information on prognosis.

6. What is the impact of lack of information?
The very negative impacts of the lack of information are listed below:

Type of impact	Mentioned by % of respondents
Isolation/inappropriate care	63%
Wrong decision making	59%
Frustration	58%
Powerlessness	49%
Fear	48%
Insufficient financial support	36%
Anger	32%

7. Conclusions
Help lines and written information are the main services provided by patient organisations with patient and mother as primary users.
To ensure quality of help lines, the following requirements were agreed on:

- Volunteers and paid staff need to be trained and the delivery monitored and evaluated
- Help lines need to develop a common tool
- Mature services can mentor developing ones
- The funding base needs to be supported by governments and the EC for stability and continuity

To ensure access to help lines:

- Patient groups, as the most reliable source of information, need to increase networking
- Interested professionals need a central source to which they can refer with confidence
- Leaflets need to be uniform, high quality, in a language appropriate to users in each region (also appropriate to the level of education)
- Websites need to be user-friendly and linked to each other for maximum delivery

→ SOLUTION TWO : A NEW EURODIS INITIATIVE, THE RAPSODY PROJECT
Rhapsody: Rare Disease Patient Solidarity (project submitted for the 2005 call for proposals, EC public health programme).
This project aims at improving access and quality of essential services at EU level. Within this project, a concerted action for rare disease help lines in Europe (CARHE) is planned.

- It will establish an EU network of rare disease help lines (paid and unpaid)
- It will develop standardised tools for collection of profiles
- It will give pilot training for helpline advisors
- It will build a European Observatory to collect process and record data
- And it will develop a system of identifying and networking very isolated patients

10.2 Internet resources for the rare disease community

SPEAKER

« Prof. Jörg Schmidtke,
Coordinator Orphanet Germany, Institute of Human Genetics, Hannover Medical School

Internet is a powerful tool, both to disseminate information To all stakeholders, with potent tools to adapt it, so that global outreach can become very large.
It also creates virtual networks

- To end isolation
- To create communities
- To promote collaboration

→ INTERNET NETWORKS
From 1997 to 2004, DG Health and Consumer Protection Directorate-General has supported networks that largely use internet as a communication tool. To mention a few ones that could not function without Internet:

- Rare pulmonary diseases: set up of diagnostic criteria and reference / training centre (Prof. Popper, Austria)
- Information network for immune-deficiencies (Prof. Vihinen, Finland)
- Euromusclenet: muscle diseases as a prototype of rare and disabling disorders: creation of a European information network (Prof. Spuler, Germany)
- Severe Chronic Neutropenia: European network on the epidemiology, patho-physiology and treatment (Dr Schwinzer, Germany)
- Paediatric rheumatic diseases: a European information network (Prof.. Martini, Italy)
- Transfer of expertise on rare metabolic diseases in adults (Prof. de Valk, NLD)
- Rare congenital anaemia: European information network (Prof. Vives Corrons, Spain)
- Charge association and Usher syndrome in Europe (Mr. Hawkes, UK)
- Rare forms of dementia (Alzheimer Europe)

All projects run by health professionals included more or less the same type of approach:

- Production of information for patients and health care professionals
- Directory of services (clinics, laboratories, support groups)

- Web-based system for collecting epidemiological data
- Discussion forum between professionals

→ LIMITS OF INTERNET AS A SOURCE OF INFORMATION

Drawbacks are often mentioned when referring to internet as a potent source for information

- not too little but too much information
- no validation rules
- can take time to find the information sought for
- can be a frustrating experience

Therefore an important question is…

Selected further Internet-based information and educational tools

- ***www.eshg.org (ESHG)***
- ***www.genetests.org (Genetests)***
- ***www.kumc.edu/gec/prof/genecour (University of Kansas)***
- ***www.humgen.umontreal.ca/int/ (Prof. Bartha M. Knoppers)***
- ***www.vh.org (Virtual Hospital)***
- ***www.ncbi.nlm.nih.gov/entrez/query.fcgi?db=OMIM (OMIM)***

→ HOW TO SURF (IVE) IN THE AREA OF INFORMATION ON RARE DISEASES ?

One solution is proposed by Orphanet:

- single-entry point for all documented rare diseases
- peer-reviewed information
- revised annually
- updated permanently

The service is not just providing information. An ambitious goal is to identify the gaps that exist in fighting rare diseases and to structure the information in a way that facilitates collaborations and contacts e.g.:

Issues addressed	Tools provided
Lack of information	Encyclopaedia of rare diseases • review articles and abstracts • expert-authored • peer-reviewed • over 1300 diseases
Scarce expertise	Experts' directory
Too few collaborations and partnership	Directory of research projects (OrphanXchange, see infra)
Difficulties in enrolling volunteers in clinical trials	Directory of clinical trials On-line recruitment service

ISSUES ADDRESSED	TOOLS PROVIDED
Lack of information	Encyclopaedia of rare diseases • review articles and abstracts • expert-authored • peer-reviewed • over 1300 diseases
Scarce expertise	Experts' directory
Too few collaborations and partnership	Directory of research projects (OrphanXchange, see infra)
Difficulties in enrolling volunteers in clinical trials	Directory of clinical trials On-line recruitment service

Orphanet is also a directory of services, in 20 countries:

- Clinics
- Tests
- Research projects
- Support groups
- Networks
- Registries
- Clinical trials

The participating countries joined the project in a staggered manner, due to financial constrains :

1997 ..... France
2001 ..... Belgium, Italy, Switzerland, Germany
2002 ..... Spain, Austria
2003 ..... Portugal
2004 ..... Ireland, United Kingdom, Finland, Denmark, Estonia, Latvia, Romania, Greece, Turkey, Bulgaria, the Netherlands, Hungary

→ ORPHANET USERS'TYPOLOGY

Among 12 000 daily users in March 2005					
Patients and families	**33.3%**	**Health professionals**	**51.6%**	**Others**	**15.1%**
Patients	14.1%	Medical doctors	30.4%	Other professions	7.3%
Parents	8.3%	Non MDs	16.0%	Others	7.8%
Family	10.9%	Medical students	5.2%		

→ CURRENT CONTENT OF THE DATABASE

- 3 713 diseases and synonyms
- 2 463 abstracts (translation ongoing)
- 624 review articles
- 751 diagnostic labs for 943 diseases
- 1 952 research projects on 1 154 diseases
- 858 patients organisations linked to 1 431 diseases
- 1 312 specialised clinics
- 4 832 health professionals
- 4 379 other web pages

→ FUTURE DEVELOPMENTS

- to move towards comprehensiveness of data
- to establish a lay-people oriented encyclopaedia
- to obtain additional national funding
- to edit and distribute country specific print versions
- to establish itself as a partner of stakeholders in rare diseases at national levels

10.3 Training families and carers in Norway

→ A WORD ABOUT FRAMBU

- Frambu is a national centre for rare disorders
- It covers approximately 60 disorders
- It offers services to the users, families and to the local/regional professionals
- It also offers supplementary services to health and social services
- It is financed by the Ministry of Health and Care Services
- Frambu is one of 17 centres for rare disorders in Norway

SPEAKER

Britta Nilsson,
Frambu centre for rare disorders, Norway

Norway is benefiting from a long experience in National plans for rare diseases, the initial one was launched in 1990 and lasted until 1993; the most recent was launched between 1994 and 1997.

→ CLUSTERS OF DISEASES COVERED BY FRAMBU

- Genetic syndroms and disorders with developmental delay
- Sex chromosome disorders
- Overgrowth syndroms
- Muscular disorders with appearence in childhood
- Progressive encephalopathies
- Neurocutaneous syndroms

→ A PLACE FOR DIALOGUE

Frambu is a place of dialogue for the users, the families, the siblings and the professionals. Users/families can acquire knowledge, consult with professionals, exchange experiences, discuss different topics, enjoy physical activities, focus upon coping, relations, self-esteem, and empowerment and make lasting friendships.....

→ IN-HOUSE ACTIVITIES

Frambu is offering a wide range of services to its users:

- information courses for users and families
- summer camp
- school and kindergarten
- seasonal gatherings for representatives of the user organisations
- workshops, seminars and conferences

→ HOW MANY PEOPLE PARTICIPATE TO FRAMBU ACTIVITIES?

Information courses for families and users: altogether 1 160 persons stayed at Frambu in 2004; 453 users (patients), 499 parents and 208 siblings.

Four different summer camps were organised in 2004, for 101 children aged 10 – 16 years and for 74 others aged 17 – 30 years. These camps lasted for two weeks and the children attend the camps on their own, no parents and no siblings are present.

→ TRAINING

Courses for users and families

The main training is related to the diagnosis but not only that: there are social rights and prognosis, genetics and daily life are also covered. It consists in a two week introductory training course followed by a one week course on schooling, social rights, friendship, moving to your own house, technical aid, leisure time…

When parents are listening to lectures, participating in discussion groups or consulting with experts, children are in kindergarten or at school. Both kindergarten and school welcome children with a disease along with their siblings. Special educators have been trained for more than twenty years.

« At Frambu participants can stay awake all night to discuss, to cry, to love, to encourage, to support and to make lifelong friendships. »

→ OUTREACH ACTIVITIES / TRAINING

Information dissemination, collaboration and counselling in the local community where the user lives:

- Workshops and seminars in different regions, communities
- Collaboration with clinical and research institutions

A total of 150 users and their professionals were able to be visited from Frambu in 2004. Usually two professionals, for instance a medical doctor and a special educator, travel to the user's home-community for a one- or two-days information dissemination and collaboration with the local and regional professionals.

- Genetic syndroms and disorders with developmental delay
- Sex chromosome disorders
- Overgrowth syndroms
- Muscular disorders with appearence in childhood
- Progressive encephalopathies
- Neurocutaneous syndroms

→ RESEARCH

- data collection on every day experiences
- research projects

→ DOCUMENTATION AND INFORMATION

- Internet and intranet
- Publications - booklets, brochures, books
- Videos
- Videoconferences
- Online information (telephone, internet)

→ LEARN MORE ON FRAMBU

www.frambu.no Most of the site is in Norwegian.

10.4 The Ågrenska Foundation: a family programme

SPEAKER

“Anders Olauson,
Ågrenska Chairman,
Göteborg

A child's disability affects all members of a family; therefore the Family Programme at Ågrenska is directed towards the entire family. The Family Programme offers a unique opportunity for families to meet and exchange experiences concerning the same rare disease.
During the stay, the parents are offered a programme containing the most recent medical and psychosocial information, information on the consequences of the disorder and on the support offered by society.

Professionals from the child's home environment are invited to attend the parental programme for two days. The siblings and the children with the disease are offered a programme that suits their needs.

To better integrate children with a rare disease in our society, educational tools themselves must be considered as part of the treatment.
The objectives of the Family Programme developed by the Ågrenska Foundation in Sweden are to obtain information on educational consequences and spread it to teachers, pre-school teachers and others who meet the children in daily activities

“In order to understand how it is to be a parent to a disabled child, you have to be a parent of a disabled child yourself!”
Åke Martinsson

→ METHODS

- Using a controlled and validated observations material which is approved by the University of Göteborg.
- Systematic observations of the children with the same diagnosis are performed in Ågrenska school activities, during the family programme.
- Lectures of the syndrome to parents and the children's accompanying professionals.
- Report back and connection with the children's local teachers at home.

→ RESULTS

- From 1990 to 2004, 140 different rare diagnosis's were observed at Ågrenska
- From all parts of Sweden
- More than 2 200 visiting families
- More than 3 300 visiting parents
- More than 2 400 visiting children/ adolescents with a rare diagnosis
- Nearly 3 000 visiting siblings

→ RESULTS FROM THE FAMILIES' PERSPECTIVES

- Parents feel "normal"
- Families feel "In power", by meeting others in the same situation
- Parents obtain new knowledge, in order to be in control of their own lives
- The diagnosed children meet others who have the same diagnosis
- Siblings meet other siblings

→ OVERVIEW OF THE FAMILY PROGRAMME, AUTUMN 2005

Week 35 ..... Achondroplasia
Week 37 ..... Usher's syndrome, type 1
Week 38 ..... Osteogenesis Imperfecta (OI)
Week 40 ..... 22q11 - deletion syndrome
Week 41 ..... Neurofibromatosis, type 1
Week 43 ..... Spastic paraparesis
Week 45 ..... Dysmelia – Limb deficiency
Week 46 ..... Langerhans cellular histiocytosis
Week 48 ..... Hydrocephalus, (without Myelomeningocele, MMC)
Week 49 ..... Anal atresia

→ NEWSLETTER

A journalist at Agrenska summarises and compiles lectures and information from the parents programme during a Family Programme for a newsletter on the disease.
Before the information is made available to the public, the lecturers read and register their opinions on the summaries. The medical information is updated continuously, in cooperation with the lecturers. A single case description is included in the newsletter on every disease, describing the every day challenges that the family meet.

→ OTHER PROJECTS

Agrenska launched an initiative with a designer school to help find ergonomic solutions for all sorts of disabilities that people with rare disease may encounter in life.
As an illustration, figure x shows some of the ideas suggested by the participating students. Not all will become reality, but at least this demonstrates that efforts to improve the daily life of disabled persons are possible: eating, washing up, expressing yourself, finding your way, having fun.

10.5 Training on genetic medicine, new technologies

❝ Poster 60,
Giovanni Romeo
– European Genetics Foundation – Bologna

European Courses in Genetic Medicine and Genetic Interest Groups

During the last 50 years the scientific achievements in all areas of life sciences have led to a common basis of unified knowledge and also to common methodological approaches including the specialised areas of medicine. The central role of genetics/genomics in medicine is now widely acknowledged both for biomedical research and for the advanced training of the new generations of scientists. Thanks to the results of the Human Genome Project, the genes responsible for an increasing number of rare diseases can now be identified. The term "genetic medicine" implies that the use of genetics as a tool in biomedical research and in advanced training is becoming one of the main features of modern medicine.
The European School of Genetic Medicine (ESGM) is at the leading edge of advanced training in the field of Genetic Medicine and its courses have been attended by more than 5000 students during the last 18 years (see www.eurogene.org). During the last three years the ESGM training has been marked by the experimentation of new technological and methodological approaches. Using web-casting technology the ESGM is now offering its courses to virtual participants unable to travel to the Main Training Centre located in Bertinoro (Italy). Following this model the virtual version of the courses will be web-cast to Satellite Training Centres all over the Europe. This "hybrid courses" format is intended to attract new participants at the ESGM courses without requiring them to invest time and resources for travel.
In the near future the ESGM, in collaboration with professional associations and patient organizations will offer a series of courses aiming at increasing awareness and understanding of genetic disorders and intends to collaborate with Genetic Interest Groups in the application for European grants.

Participants at the conference

11 PATIENTS' RIGHT :
mobility, care in a foreign country. Decisions of the European Court of Justice.

11.1 Trans-border access to care: a view from the European Court of Justice

→ SUMMARY

The organisation of healthcare and social security is a matter for which the Member States have not transferred powers to the European Union. In the organisation of their national healthcare systems, Member States must however take into account basic principles of European law, such as the right of patients to free movement. In a series of judgments, starting with the Kohll and Decker cases, the Court of Justice made clear that any national rule which makes reimbursement of medical treatment provided abroad dependent on prior authorisation, must be justified by objectives of general interest such as the financial balance of the social security system and the need to maintain a balanced medical and hospital service open to all.

SPEAKER

Dr. Piet van Nuffel,
Legal Secretary at the Court of Justice of the European Communities, Luxembourgof the European Communities, Luxembourg

Whereas prior authorisation may thus be justified for hospital treatment abroad, this is normally not the case for ambulatory care abroad. In the latter case, the requirement of prior authorisation will be an unjustified restriction of the freedom to receive services, irrespective of whether the home State applies a system of reimbursement or benefits-in-kind.

Even though European law does not preclude a system of prior authorisation for hospital care abroad, it requires any such system to be based on objective and non-discriminatory criteria. Under this condition, Member States are free to determine which treatments will be paid for by their social security system. Where prior authorisation is dependent on the necessity of the treatment abroad, authorisation may be refused only if treatment which is the same or equally effective for the patient can be obtained without undue delay in the home Member State. Prior authorisation cannot be refused solely because there are waiting lists on the national territory, that is to say undercapacity. The existence of waiting lists is central to the pending Watts case, in which the Court of Justice has been asked whether the need to allocate resources according to medical priorities might justify refusing certain patients to receive treatment abroad at the expenses of the national health service.

Rights to medical care abroad under EC regulation :

1. If insured person abroad needs treatment in State of stay (E111 form)

2. If insured person is authorised to go to other State to receive there treatment (E112 form)
- Entitlement to benefits under terms of the host State
- Authorisation not to be refused by home State if:
  - benefits covered in home State
  - treatment not available there within time-limit medically justifiable given patient's state of health

→ RIGHTS TO MEDICAL CARE ABROAD OUTSIDE EC REGULATION :

1. Kohll and Decker case: patient did not seek for authorisation prior to care in state different from state of residence

Articles 30 and 36 of the EC Treaty preclude national rules under which a social security institution of a Member State refuses to reimburse to an insured person on a flat-rate basis the cost of a pair of spectacles with corrective lenses purchased from an optician established in another Member State, on the ground that prior authorisation is required for the purchase of any medical product abroad.

Requirement of authorisation under scrutiny :
- As it can be considered as a barrier to free movement of goods or services
- Can it be justified? In general, member states object that in the absence of such an authorisation,
  - Financial balance of social security system could be impaired,
  - Thus endangering the objective of maintaining a medical and hospital service open to all.

Art. 22 Reg. 1408/ 71 is intended to allow an insured person, authorised by the competent institution to go to another Member State to receive there treatment appropriate to his/her condition. It is not intended to regulate and hence does not in any way prevent the reimbursement by Member States, at the tariffs in force in the competent State, of the cost of medical products purchased in another Member State, even without prior authorisation.

The obligation to obtain prior permission must be categorised as a barrier to the free movement of goods, since they encourage insured persons to purchase those products in the national territory rather than in other Member States, and are thus liable to curb their import. They are not justified by the risk of seriously undermining the financial balance of the social security system, since reimbursement at a flat rate of the cost of spectacles and corrective lenses purchased in other Member States has no significant effect on the financing or balance of the social

security system, nor are they justified on grounds of public health in order to ensure the quality of medical products supplied to insured persons in other Member States, since the conditions for taking up and pursuing regulated professions have been the subject of Community directives.

2. Requirement of authorisation justified for hospital care (Smits/Peerbooms and Van Riet cases)
This requirement for hospital care is considered as justified, as the impact of foreign visitors consulting or seeking care can be significant:

- necessity of planning the number of hospitals, their geographical distribution, their mode of organisation, their equipment and the nature of the medical services
- aim of controlling costs and preventing wastage of financial, technical and human resources
- ensuring sufficient and permanent access to high-quality hospital treatment

→ **CONDITIONS FOR AUTHORISATION**

- Hospital care must be insured and reimbursed in state of origin (Smits/Peerbooms)
- Requirement of « necessity » for treatment abroad (Smits/Peerbooms)
  - No necessity if same or equally effective treatment available without undue delay
  - Are waiting lists relevant? (Müller-Fauré/Van Riet; Watts?)

→ **WHEN IS AN INSURED PERSON COVERED FOR TREATMENT ABROAD?**

- Treatment while staying abroad
  - no authorisation needed (E111)
  - covered under host State terms
- Ambulatory care abroad
  - if authorised with E112: covered under host State terms
  - without prior authorisation: reimbursed under home State terms
- Hospital care abroad: prior authorisation needed
  - if authorised with E112: covered under host State terms
  - reimbursed under home State terms if authorised otherwise

→ **PATIENTS' MOBILITY RIGHTS: CHALLENGE FOR HEALTHCARE?**

- Administrative complication
- Stimulus for structural change?
- Will the Court eventually get guidance from our political representatives?

→ **CONCLUSION**

The Court's case law has prompted the Commission to include provisions on patient mobility in its proposal for a Directive on services in the internal market. In the current political context, it is all but sure that discussion of this "Bolkestein proposal" will result in any codification of the Court's case law. Still, legislative intervention in this field would certainly enhance transparency and legal certainty for all stakeholders.

12 STRATEGIES FOR PREVENTION

12.1 Strategies based on the assessment of epidemiological evidence

« Prof. Helen Dolk, EuroCat, University of Ulster

Prevention of rare diseases is possible, to some extent.
1. Primary Prevention consists in:

- pre-disease
  - Attacking basic cause(s) of disease
  - Altering environment or resistance/susceptibility

2. Secondary Prevention consists in:

- Disease has started but symptoms have not appeared
  - Detecting and treat early to prevent disease development
  - e.g. newborn screening for phenylketonuria

3. Tertiary Prevention consists in:

- Disease has become symptomatic
  - Curing, controlling or preventing complications

Epidemiology in relation to primary prevention strategies:

- What is the incidence of the disease in the population?
- What distinguishes who does and does not get the disease in the population?
  - Age, sex, time, place, social status
- What causes the disease?
  - Environment, genes, and their interaction
  - Causal pathways and networks
- How much and why do populations differ in incidence?
- How much disease could we prevent with different prevention strategies?
- How would different prevention strategies affect inequalities in disease incidence?
- How successful has the implementation of a prevention strategy been in relation to its potential in reduction of disease and reduction in inequalities in disease?

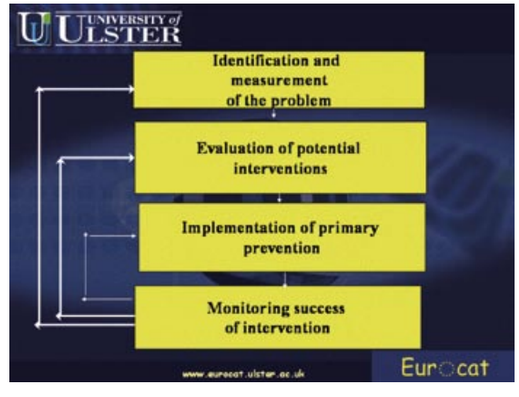


figure 41: an action plan for prevention, from problem identification to the monitoring of the success.

→ **EUROCAT : EUROPEAN SURVEILLANCE OF CONGENITAL ANOMALIES.**

- European network of population-based registries for the epidemiologic surveillance of congenital anomalies.
- Started in 1979, now funded by EU Public Health Programme
- More than 1.2 million births surveyed per year in Europe
  - 40 registries in 19 countries
  - 30% of European birth population covered
  - Quality at the expense of completeness of geographical coverage

→ **POSSIBLE PRIMARY PREVENTION STRATEGIES**

- Periconceptional folic acid supplementation
- Vaccination e.g. congenital rubella
- Preconceptional and pregnancy care for high risk women e.g. diabetes, epilepsy
- Genetic counselling for high risk families
- Reduction of abuse of recreational drugs/alcohol
- Pre-marketing drug testing, pharmacovigilance and health technology surveillance
- Reduction of exposure to environmental pollutants (precautionary where necessary) and enviro-vigilance

→ **IN CONCLUSION**

- Epidemiology underpins planning and evaluation of all levels of prevention
- Primary prevention of rare diseases is as much an equality issue as secondary and tertiary prevention
  - Termination of pregnancy following prenatal diagnosis should not be an alternative to primary prevention
  - Whole population measures may sometimes be needed to prevent rare diseases
- Population-based registries, networked at a European level, provide the means to carry out epidemiologic research and surveillance for prevention

12.2 Prevention of genetic diseases

→ **TO PREVENT THE CLINICAL OUTCOME OF A LATE-ONSET GENETIC DISEASE**

Gene	Disease	Treatment
BRCA1-2	Breast cancer	Mammectomy
RET	MEN	Thyroidectomy
MHC1-L	Hemochromatosis	Iron chelators
Myosin	Cardiomyopathy	Follow-up
n ….	Deafness / blindness	Special schooling

“Prof. Stanislas Lyonnet, Hôpital Necker, France.

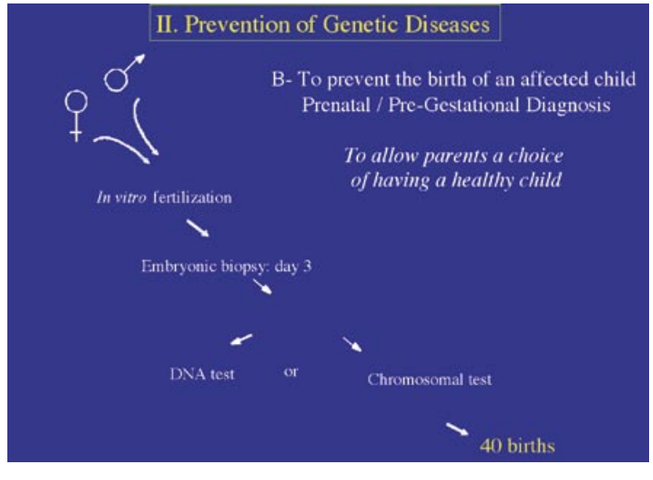
Detection of a genetic risk is best indicated in families where the risk exists, i.e. where cases are already diagnosed. Individualised follow-up can then be implemented. For people who are carrying detrimental genes like those favouring blindness or deafness, even though no treatment exist, early detection of the genes helps deciding how to organise the life of the person years before the disability/disease becomes symptomatic. Schooling for example can be adapted to the needs of the person.

→ **TO PREDICT OR TO SLANDER?**

- Who wants to know?
- When duly informed about the utility of testing, not all patients or persons at risk decide to undergo genetic testing. For Huntington Chorea for example, only 18% of persons at risk and who are consulting for genetic testing actually do the test.
- What do we know? Modifier genes exist, they can be detected, they do not provoke a disease but may increase or decrease the expression of detrimental genes. Predictions are then difficult.
- What is the social impact of prediction? Insurance companies, employers could divert genetic testing from its medical purpose.

Prenatal and pre-implantation genetic testing

Figure 42 : Pre-implantation testing has nothing to do with cloning. It consists in in vitro fertilisation, for parents who are at high risk to give birth to a sick child. One cell is captured (embryonic biopsy) and analysed (DNA test, chromosomal test), and if the genes are healthy, then the corresponding embryo is



13 CLOSING OF THE CONFERENCE

13.1 Moving forward in Europe

My first message is simple: "Europe, Europe and Europe", and I am very sorry to be from a member state which missed the point a few weeks ago by rejecting the Constitutional Treaty. It is obvious that for rare diseases this is the only level where we can achieve something meaningful. Even if some citizens have not understood the message, we want to work at the European level, and even better, at the global level.

SPEAKER

« Dr. Ségolène Aymé,
Orphanet,
Inserm SC11, France.

Then, as health care professionals, we have several messages to the Commission:

- To DG health and consumer protection: we are very satisfied for the support and initiatives in the field of information and surveillance, and this should be continued. However solutions to ensure sustainable and longer term funding are expected, as most of the initiatives really useful for the rare diseases community are developed to collect data and disseminate information and results and these are long-lasting efforts. If funding can only be granted for 2 to 3 years, it may not be worth starting the project at all.

- To DG research: in the recent years, few research projects were supported on rare diseases. In FP7, new research projects on rare diseases are expected to be funded. We need to advocate in order ensuring that no budget cuts will affect this good will. If the research budget has to be decreased, we hope this will not affect projects on rare diseases.

- Currently, rare diseases appear in the genomics strand of the research programme. Genomics is an important part of research but it does not constitute all what we need. Research in epidemiology, health care provision and services are equally important.

- Expectations are very high in the rare diseases community, and sometimes the researchers' agenda does not totally coincide with patients and families' agendas. Immediate needs of the community should be listened to.

- Efforts to provide information exist and are welcome; they may not always be adapted to all publics. Paramedical professions, patients and their representatives for example need adapted information. Collaboration and partnership between information sources are certainly a way to address this issue.

- The need for more imagination: even complex problems always have a solution. Even when the damage is present at birth, there are therapeutic solutions that can help and improve the quality of life. More solutions can be found.

- Messages from the Task Force on Rare Diseases: the DG Health and Consumer Protection created this task force and we all welcome this initiative. Funding has been granted and a newsletter thus created to convey information to all stakeholders. It is a link for all of you, so please register if you haven not already done so. The URL where to register is: www.rdtf.org. You are invited to send information to the newsletter team if you wish to disseminate it through the community.

- Other actions of the Task Force for the coming years are ambitious as well: coding and classification of rare diseases. We want a code for each of the rare diseases; all deserve to be visible in the health care systems. We will be working on health indicators, in order to compare outcomes in different member states, and to bench mark best practices in Europe. Of course we will act as advisors to the European Commission, and your participation is key. The force will come from you.

13.2 A society where rarity does not affect opportunity

SPEAKER

« Christel Nourissier,
Eurordis

After one year of preparation and a two day conference, we now have a clear vision for rare diseases: ten years from now, people living with rare diseases across Europe will have the same opportunities as their fellow citizens in European society.

→ **HOW CAN WE REACH THIS GOAL?**

By enforcing a number of very practical changes in the health care systems across Europe:

- Well trained doctors or paediatricians able to detect a rare condition right away,
- Diagnostic laboratories exchanging blood samples, tissues, DNA and results across the EU,
- Radiologists sending medical images from any care centre to a specialised centre,

- Accurate diagnosis being made as early as possible with the support of telemedicine,
- If necessary, patient and family travelling to the centre of reference, regardless of borders. Or better still, professionals from the centre of reference training local doctors and their families.
- Simpler paperwork to obtain financial compensation, care, etc.
- Financial aspects being handled with the help of social workers,
- Families getting the support of a local patient group and meeting with other families.

→ WE WANT A CLEAR PATH FOR PATIENTS AND THEIR FAMILIES:

- Necessary support, immediately after the diagnosis, during critical situations, and for the duration of the patient's lifetime
- Local health professionals with access to treatment and care guidelines for medical and paramedical care
- Emergency units using updated information from medical web sites, telephone lines,
- Adequate information and training at school, at the work place, at home and in residential homes.

→ TO MAKE OUR DREAM BECOME A REALITY, EUROPE CAN CERTAINLY HELP:

- With the co-ordination of public health and research programmes, ensuring continuity of actions
- With the collection of epidemiological information (Morbidity and Mortality Working Party and the Task Force for Rare Diseases)
- With a permanent support of a network of European reference centres connected with national / regional centres of reference
- With the definition of best practice guidelines for care,
- for the integration of children at school, for the integration of adults at the work place
- With the cartography of existing resources: hospitals, respite care, summer camps, and the identification of urgent needs together with patient associations
- By providing a strong European environment for innovative therapeutic interventions and innovative medicines

→ MANY ACTIONS ARE TO BE IMPLEMENTED AT A NATIONAL LEVEL IN EACH MEMBER STATE :

- Encourage national plans for rare diseases,
- Support information centres with more public funding,
- Train and educate health care professionals, and also volunteers and staff for patient groups,
- Create and support national or regional centres of reference for rare diseases,

- Facilitate access to medical and paramedical care, devices and equipment,
- Improve access to already marketed orphan drugs; continuously push for the development of other orphan and paediatric drugs,
- Better compensate disabilities: human resources and technical aids
- Empower rare diseases patient groups, inform and educate patients.

→ ALL THESE EFFORTS WILL NOT ONLY BENEFIT RARE DISEASES, BUT EFFORTS FOR RARE DISEASES CAN PLAY THE ROLE OF A CATALYST FOR OTHER DOMAINS :

- European networks of biobanks, patient registries and centres for clinical research
- Rare diseases as models for more common diseases
- Pluri-disciplinary research aiming at a better life for patients
- Innovative methods for clinical trials, innovative drugs
- Involvement of strong, active and well supported patient groups

→ HOW CAN PATIENTS AND THEIR FAMILIES GET THERE?

- At national level: by strengthening groups and national alliances,
- At European level: with Eurordis representing the rare disease community,

→ OUR STRATEGY : A GLOBAL APPROACH OF THE PATIENTS

- Integrate patient networks, research, diagnosis, and treatment structures.
- Make the best use of our current knowledge, of medical and paramedical care, of education and rehabilitation schemes
- Publish and publicise best practice guidelines, when and wherever they exist
- Involve patients at all stages: clinical trials and research protocols, dissemination of outcomes.

Our rare diseases community will meet again to discuss achievements and plans for the future at the next European conference for rare diseases, in Lisbon, Portugal, October 2007.

13.3 The word of the European Commission

The Commission would like to thank the organising committee of this conference, Eurordis, all partners involved, and also the Luxembourg government and in particular the health minister Mars di Bartolomeo, for what I think has been a very successful EU level event.
I wanted specially to complement the cultural performance which was an excellent show, very professional and inspiring.
I also wanted to thank the other sponsors, who helped us to build up this conference together with the EU health programme.
For 2 days we have been taking through what is really going on in many areas in the field of rare diseases. Those participating have been able to get a valuable of where the problems lie.
We learned the results of the Eurordis study on the delays in diagnosis, identifying sometimes considerable time lags before rare diseases are identified and treated, where appropriate and when possible.
There was also a discussion on bench marking initiatives to improve care, comparison of national plans, practices on trans-border access to care, leading to a discussion on the need for and the role of reference centres in the context of rare diseases.

Targeting research to improve quality of life, increasing the coherency of research by avoiding fragmentation, establishing larger networks, and the contribution of our EU research programme where other issues which were discussed. I can confirm that rare diseases per se will be included s an eligible disease category within the next framework programme of the Commission proposal for the period 2007 to 2013.
The Commission will also take forward the conclusions of the research workshop which took place on April 13th.

We also heard about the problems encountered in data collection and management. How and why you need to improve coding and classification, a challenge not only for the European Union but other countries around the world and our international partners such as WHO.
The specific aspects of registries were examined; and national and trans-national options were looked at. We looked at clinical trials, treatments using orphan drugs, and access issues.
Finally I must mention the important statement given to us just now by Dr. Ségolène Aymé who is the chairman of our Rare Diseases Task Force within DG Sanco which plays a very important role for our work and we hope to include its recommendations in our forthcoming work plan.

SPEAKER

“John F. Ryan,
Head of Health Information Unit, Public Health and Risk Assessment Directorate, DG Health and Consumer Protection, EC

On behalf of the Commission’s public health services, I have been most interested together with my colleagues to hear the very high level presentations during these last two days. This conference has given us a better direction, defining priorities in the projects that can be made, and also in the context of our new programme for 2007-2013.
I haven been encouraged by some of the presentations to see where we could play a bigger role at EU level in supporting the work of health professionals in improving training, sharing information resources, and providing concrete resources and supports for patients and carers.

You may have seen outside on our information stand some of the completed projects’ reports that the Commission has financed and we are very happy that this is given some recognition and publicity to those efforts. We also hope that the future EU health portal which we hope to launch this year will be another step in this direction.
It is our hope that the recommendations made during this conference will be implemented in the coming years, we hope that the future Rare Diseases White Book that you are hoping to develop according to the results of this conference, will bring together these ideas and strengthen European cooperation. We also commit ourselves to taking forward the work within the Task Force that we have set up last year.
The Commission is finally convinced that this is not a single or a one-off event, we are very happy to hear about the announcement of the Lisbon event in 2007 and we would look forward to seeing everybody there and giving our support again.

Thank you ladies and gentlemen, thank you minister.

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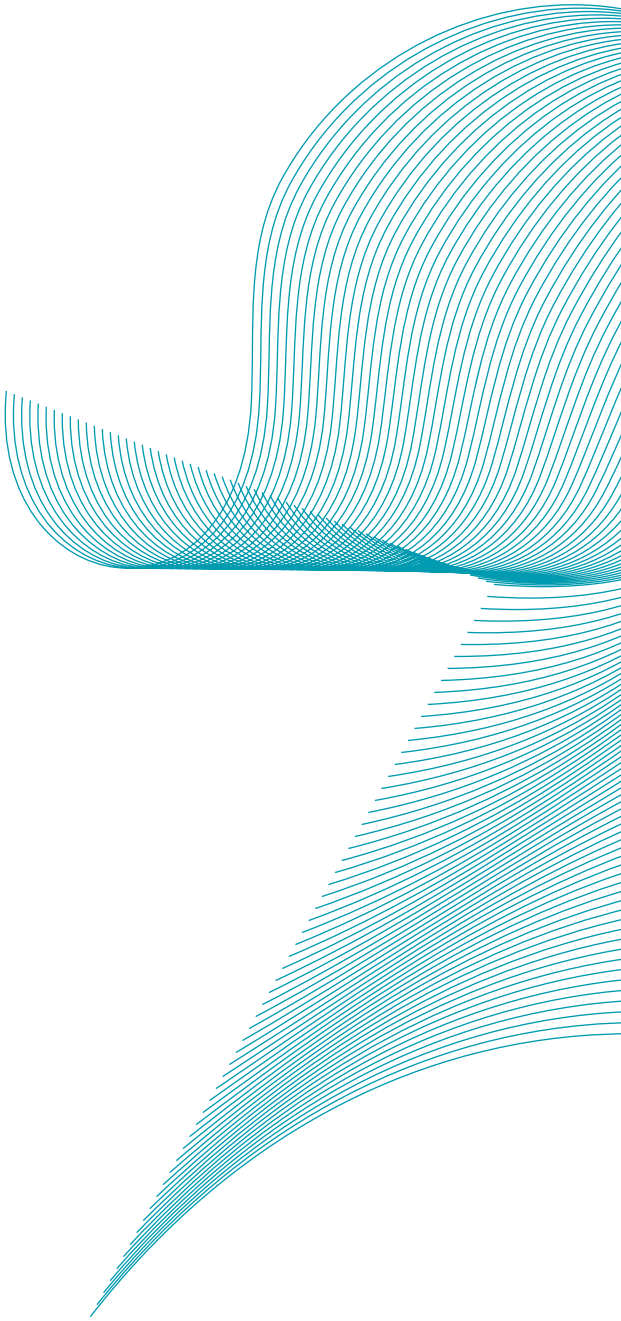

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