

EIGHTH WORKSHOP ON PARTNERING FOR RARE DISEASE THERAPY DEVELOPMENT

The Reality of Orphan Medicines

**Copenhagen, 18-19 October 2007
Danish Parliament, Denmark**

**With the participation of Her Royal Highness
Crown Princess Mary of Denmark**



- The report and its aim
 - 1. How to estimate the value of an orphan drug
 - 2. How many orphan drugs – for how many patients?
- 3. How to communicate about orphan drugs in a real-life setting?



The Co-Chairs with the Director and the Chairman of EPPOSI
S. Aymé, E. Tambuyzer, G. Asta, B. Holm, A. Kent

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Contributors: Integral, Zymenex

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Cover picture : HRH Crown Princess Mary of Denmark, Kristine, Mari, Alastair Kent

The report and its aim

This publication is a report of the Eighth EPPOSI Workshop on Rare Disease Therapy Development and Partnering, held on 18 and 19 October 2007 in the Danish Parliament, in Copenhagen.

The starting point for the workshop was simple: patients with rare diseases have no choice over their disease but are as entitled to a treatment as patients with more common conditions. The aim was to build a platform for consensus on what can be done, rather than to establish an artificial one in an area where there are many viewpoints and too few data. This short report, accordingly, is not intended to be a consensus view. Rather, it seeks to highlight the main points that emerged during two intense days of discussion and debate. The rapporteur, Peter Wrobel, takes full responsibility for this work.

What is a rare disease?

A rare disease is a life-threatening or chronically debilitating condition affecting not more than five in 10 thousand (or 1 in 2 thousand) persons in the Community.

Source: Reg EC 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products.

Executive summary

What is the reality of orphan medicines in Europe? The truth is that no one really knows the full picture. It is hard enough to find out which medicines are actually on sale in which European countries, and equally difficult to say how many patients could benefit from orphan medicines. When it comes to saying how many orphan medicines may be on the market in five or ten years' time, or how large the potential market is for these medicines, there is too much guesswork operating.

In this uncertain climate, three approaches are flourishing – hope, fear and frustration.

First, hope: for patients and their families who for the first time in human history can start to see treatments or even cures for intractable, serious and often-life-threatening diseases.

Second, fear: for the payers who look at the prices of orphan medicines now and wonder if the current crop of orphan medicines is just the tip of the iceberg – that they face an avalanche of new medicines and ballooning claims on their already constrained budgets.

And lastly, frustration: for patients who see new medicines but cannot gain early and equitable access to them, and for the researchers and companies who have created these medicines only to see access to them delayed or denied to many, many patients.

Against this background, EPPOSI brought together around 120 stakeholders from patient groups, industry, research, hospitals and authorities from both sides of the Atlantic, and the discipline of health economics to a workshop in Copenhagen in October 2007. The aim: to see where the consensus lies, what facts we know, how we estimate the value of innovative medicines now (and in the future), what further research is needed, and how we might go about finding data and hardening up forecasts.

Despite all the uncertainties, a number of definite conclusions emerged as the workshop progressed through three sessions: *How to estimate the value of an orphan drug; How many orphan drugs for how many patients; and How to communicate about orphan drugs in the real-life setting.* These conclusions, amplified in the report below, should make worrying reading for policymakers but – unexpectedly, perhaps – reassuring reading for the payers.



A. Kent, C. Lavery, W. Goettsch, E. Jessop, E. Tambuyzer, T. Grønnebæk, F. Borlum Kristensen, F. Meyer

What is an orphan drug or an orphan medicinal product?

"Orphan drugs", or in the EU "orphan medicinal products", are medicines intended to treat diseases so rare that sponsors (companies) are reluctant to develop them under usual marketing conditions.

The process from the discovery of a new molecule to its marketing is long (10 years on average), expensive and, since much work is done in formerly uncharted territory, very risky (of many molecules tested, none or only one may have a therapeutic effect).

Under normal market conditions a company developing a drug intended to treat a rare disease may not recover the capital invested for its development.

Industry and health authorities have jointly argued, first in the US and then at national and European levels (the European Commission) for the incentives required to stimulate the development of orphan drugs; these efforts led in 2000 to the European Parliament unanimously approving the EU Regulation on Orphan Medicinal Products (EC 141/2000).

The review of all marketing applications for orphan medicines is centralised through EMEA – the European Medicines Evaluation Agency) with the goal of making rapidly available, for rare diseases, medicines with a level of quality, efficacy and safety equivalent to that required for any other medicine.



The auditorium

Put very briefly, the key messages are these:

- 1 Despite uncertainty about exact numbers, there is no likelihood of an avalanche of expensive new orphan medical products within the next five or even ten years – not such good news for patients, but payers need not be alarmed about setting precedents that would lead to an imminent exponential rise in costs.
- 2 The real cost of orphan medicines may be much lower than thought, once account is taken of reduced costs in other areas, plus the contribution that some patients may be able to make to society through employment.
- 3 Equitable and timely access to orphan medicines varies not just between countries but also within them. That is a problem not just for patients but for Europe as a whole.
- 4 Health technology assessment is a useful – indeed vital – tool for decision makers if adapted to the scarcity of available data, but it can only be one element of the decision-making process. But patients need to be both won over to the concept and included in the process as partners; and politicians should recognise that the final decision is theirs, a political rather than solely an economic one.
- 5 More research can and should be done to establish the real value of orphan medicines; the number of patients in Europe with orphan conditions; the likely number of those conditions for which treatments (and perhaps cures) will be found; and the timescale over which those will be developed.
- 6 Good databases and patient registries – preferably international – are essential to establish how orphan medicines actually work in real life.
- 7 In all these areas, there are examples of good practice that can be followed and developed.
- 8 Dialogue, partnership and transparency – nationally and internationally – will hold the keys to progress.

1. How to estimate the value of an orphan drug

The scene was set by a powerful account of inequalities in access to medicines. "One never imagines parents could be put in a situation where they had to beg for their child's treatment," said a patient representative describing precisely that situation as it applied to two children in Scotland with exactly the same life-threatening condition, an enzyme defect. One of those children was enrolled in a clinical trial, and therefore continued to receive treatment from the company organising the trial at their cost after the (successful) trial ended. The other – the only other child in Scotland meeting the criteria for treatment with the new drug – was diagnosed fractionally too late to be on the trial, and was denied funding for the treatment when it became available.

The absurdity here is that governments all over Europe support companies to develop innovative medicines with economic incentives and sometimes research money, but keep their hands firmly in their pockets when it comes to paying to use the medicines developed. For example, NICE, the UK National Institute for Clinical Excellence, regularly uses economic arguments to control patients' access to innovative drugs. There is a significant discontinuity between this and the policies adopted by other governments towards the funding of biomedical R&D – seen as a strategically important element of the UK's future prosperity.



F. Meyer, F. Borlum Kristensen, A. Rappagliosi, A. Kent, E. Jessop, W. Goettsch, P. Wrobel



Andrea Rappagliosi

There are better ways of deciding these matters. For example, Christine Lavery, from the MPS Society and Eurordis, presented a model that is used in England to regulate access to orphan medicines for lysosomal storage disorders including strict diagnosis criteria, centres

of excellence, registries, treatment guidelines and post-marketing follow up, and which may be adaptable to other regions or disease areas. And in fact a similar model exists in France.

Earlier, introducing the workshop, EPPOSI Chair Alastair Kent had laid down a challenge to health technology assessment: "The rhetoric is unarguable. Of course we need to know what works, for whom, what the cost is, and so on. But the practice is imperfect."

What is Health Technology Assessment (HTA)?

Health Technology Assessment (HTA) is a multidisciplinary process that summarises information about the medical, social, economic and ethical issues related to the use of a health technology in a systematic, transparent, unbiased, robust manner. Its aim is to inform the formulation of safe, effective, health policies that are patient focused and seek to achieve best value.

Source: EUnetHTA, www.eunethta.net

Despite its policy goals, HTA must always be firmly rooted in research and research methods.

That challenge was met, at least on the part of EunetHTA, the European project on health technology assessment, with a detailed explanation of what health technology assessment is, and what it isn't. It can and should take account of patients' views. It is rooted in research and research methods. But its output is information on the basis of which politicians and payers make decisions, not those decisions themselves.

Is cost really an issue? With some drugs costing more than €100,000 per patient per year, that might seem like an odd question. But it's not many patients, and not many drugs. And in any case, when you have such expensive drugs, and patients who will die without them, the purely economic criteria of health technology assessment cannot apply. So society must make moral judgements.

Even so, we are in general woefully ignorant of the true value of a successful orphan medicine. The workshop heard of a promising way forward, represented by a long-term survey of



Cees Smit

haemophilia patients in The Netherlands. For 30 years, the patient association has been tracking a range of medical and social parameters, and can now show that treated patients were staying at work an average of 17 years longer in 2001 than they were in 1972. The study also revealed the cost of hospitalisation where treatment is denied: up to €100,000 a year.

In a later discussion about communication, results from the latest survey from Eurordis, the European Organisation for Rare Diseases, showed that variations in the actual ex-factory price of orphan medicines in different European countries were in a fairly narrow band – between 94 per cent and 110 per cent of the mean. Given such small variation, could there really be a valid argument against the concept of a centralised European ex-factory price?

Conclusions

Health technology assessment for orphan medicines

- Given that the questions being asked of Health Technology Agencies are broadly the same throughout Europe, it would make sense to develop, in consensus, a model at EU level for looking at health technology assessment in the orphan medicines area.

This would simplify the task for national agencies, make efficient use of scarce skills and knowledge, and speed the time between development and access. A period of piloting work would be needed.

- One model or not, there needs to be better coordination internationally to reduce duplication of assessments.
- The link between health technology assessment and healthcare policymakers needs to be strengthened.
- Countries with limited experience of health technology assessment should be supported.
- Health technology assessment should stay transparent, comprehensive, and be firmly rooted in established research methods.
- Patient representatives should be included in the conduct of health technology assessment, in the same way as they are now included in many other aspects of the assessment of new medicines.
- Work needs to continue on alternative statistical models where randomised controlled trials are not practicable ways of demonstrating the quality, safety and efficacy of novel therapies (for example, by virtue of low patient numbers).
- Health technology assessment is a research methodology that informs political decisions taken by our elected representatives, and we all need to hold them to account for the way they use the data supplied.



Jens Gruenger

- We face European healthcare systems with capped budgets that require hard decisions to be taken. We must ensure the decisions are taken on the best possible ground and the hardest evidence.

QALYs

- There needs to be more discussion whether QALYs (quality-adjusted life years) and other tools for the estimation of cost and clinical effectiveness can play a role, and if so, how, in the assessment of the value of orphan medicines, especially for very rare diseases.
- Surveys of public opinion consistently show that people are ready to pay more than the accepted cost-per-QALY ceiling for treating patients with serious, life-limiting conditions.

Value and price

- It is possible to do far more detailed work on the values created by orphan medicines, in particular in relation to the reduction of hospital care, the cost of disabilities for patients and their families, the ability to enter the labour market and contribute to wealth creation, and the contribution made accordingly via taxation...
- ...but there seems to be unwillingness to undertake the research. This failure needs to be rectified.
- Industry needs to be transparent about how it reaches its prices, and, as importantly, be seen to be transparent.
- It is not currently possible to forecast the price of an innovative medicine several years before its launch, when its development is not complete.
- A centralised European price might be easier to achieve than has been thought, given how small the actual variations between different countries are.

2. How many orphan drugs – for how many patients?

It's often the simplest questions that are hardest to answer. And so it turned out when the workshop moved on to discussing some basic issues that everyone – not least the payers – would like to know. How many rare diseases are there? How many patients? And of these, how many are diagnosed? How many and which conditions are treatable now? How many might be treatable in the foreseeable future? How many will need treatment?

In short, no one really knows the details yet. But regulatory agencies on both sides of the Atlantic indicate it is exceptionally unlikely that payers will face a flood of new orphan medicines in the next ten years, or longer. That's perhaps not such good news for patients, but forecasts like this should moderate what many see as rising anxiety among payers: whatever the future holds, orphan medicines are not going to bankrupt them, at least for a decade or more.

Yet the uncertainties are startling. A broad review of the published literature conducted for the workshop revealed that definitions of rare diseases vary, that many papers muddle prevalence and incidence (and confuse different types of prevalence). Many patients remain undiagnosed. The stark conclusion is that the overall prevalence of rare diseases in Europe is unknown.

What is prevalence?

Prevalence is the total number of cases of a disease or of a specific characteristic in a defined population at a given moment.

What is incidence?

Incidence is the number of new cases in a defined population during a defined period of time. Therefore, the incidence measures the occurrence of diseases.

Source: Glossary of Public Health Technical Terms, European Commission, 2006

The one ray of light here is that although the exact number is not known, we do at least have an idea. From the literature, a prevalence

of rare diseases, all taken together, of around 3.5 per cent can be inferred; from an Italian registry (in the Veneto), around 1 per cent; from one study of hospital discharges, around 1 per cent.

Of course, many of the rare diseases are almost vanishingly rare. Overall, one estimate is that if there were treatments for all orphan diseases in the categories of rare diseases where there are already orphan medicinal product designations, about 1.6 per cent of the population could be defined as treatable patients. Knowing that only 1 in 10 medicinal product in development successfully reach the market, the figure becomes 0.16 percent of the population. But given that medicines will not exist for most theoretically treatable orphan diseases for a long time, and that many treatable patients are not diagnosed, the number of treated patients will be even smaller.

As to the number of rare diseases, it seems to be in the region of 5,000 to 6,000 (depending on how you define a disease), with around 5 or 6 new diseases being described per month.

But the data are weak, and much more research on numbers is needed.

The workshop also heard three forecasts, two from Europe and one from the US FDA, about how many new orphan drugs can be expected to enter the market in the foreseeable future.

One European forecast, from Eurordis, based on a detailed statistical model, concluded that the number of new orphan medicines entering the market over the next 10 years is most likely to be in the region of 85 to 105, depending on the rate of drug discovery, time taken to develop the drugs, and success rates in gaining marketing authorisation. The American forecast was slightly, but not greatly higher.

The second European forecast, from the EMEA, focused not so much on actual orphan medicines but on applications for marketing authorisation. It saw no dramatic change in the current rate of increase. Indeed, number of applications could even plateau at 15 a year, roughly the level they reached in 2006; 46 per cent of applications so far are in the area of cancer.

From industry the workshop heard the same warnings about the difficulties of establishing prevalence. Significantly, though, it also heard about the contribution that a programme of screening of newborn children could achieve. A pilot programme has been conducted in Taiwan: 135,000 children have been screened for Pompe disease, establishing a prevalence of around 1 in 34,000, with 4 patients identified. The proviso: in other countries, the genetics may vary – we just don't know yet.



T. Nguyen, P. Tomasi, K. Westermark, F. Bignami, S. Aymé

Conclusions

Registries

- Patient registries are exceptionally valuable repositories of data about specific diseases, but there are too few of them.
- Registry design is a crucial area: think of the data you would want to capture, even if only for a subset of users (such as biomarkers, for regulatory approval), and think about future needs.
- Reimbursement authorities should be involved in registry design, so that the data can answer not only questions relating to marketing authorisation but also those that arise at the time reimbursement decisions are made.
- Registries are resource-intensive to maintain. It's not enough to set them up, they need resources to operate and be maintained; otherwise their data will be useless.

- It is not sensible to set up registries at the country level for very rare diseases – for these, registries should be set up on the European level.
- We should consider prioritising the creation of registries for the more severe diseases that are amenable to treatment.

Access

- We need a much wider public discussion about how the claimed willingness to provide the resources for access can be delivered meaningfully and sustainably.
- There is a lot of rhetoric about equity, solidarity and access, but families have to fight tooth and nail to get even a part of what the law says they should have as of right.

Forecasts

- We need more research to validate the forecasting models being used and developed.
- Forecasts should look at future costs as well as numbers of treatments.



Ségolène Aymé

3. How to communicate about orphan drugs in a real-life setting?

Communication challenges crop up everywhere. This is true not least in finding out just how many patients with rare diseases are actually benefiting from orphan medicines in different parts of Europe.

You would think it would be a simple matter: ask the Member States what medicines are authorised and which are reimbursed...ask the companies how many treatments they are providing, and at what price...and voilà, you have the answer.

Sadly, that's not how communication in real-life Europe works today, as the fourth survey on access by Eurordis showed. While many countries and companies are cooperative, some are not – either through a culture of secrecy, or perhaps because they don't know, or maybe because they just can't be bothered to answer a simple questionnaire.

The results – though qualified by the difficulties in data collection – indicate that it is still taking too long from European-level authorisation to patients actually gaining access to new orphan products. These products are supposed to be accessible in each European Union country at the latest one year after authorisation, yet in many countries they are not.

One hopeful finding was that most countries do eventually provide access, though this may take many years. But generally speaking, the smaller the country, the worse your chances of access – which doesn't say much for European solidarity. And while patients wait, many die, as well.

Why all the secrecy about basic information? There are no clear answers to this. The European Union's centralised procedure lays down that companies must provide a breakdown of sales by country if asked by EU authorities – but the authorities are not asking. Instead, it is a voluntary organisation, Eurordis, that is trying to find out. Why should patient organisations have to act like detectives to winkle out the facts?

Communication with patients, policymakers and politicians has its own challenges. Everyone needs to recognise that the climate has changed. Before the orphan drug legislation in Europe, the talk was of the need for incentives. Then came the regulation, implemented in an atmosphere of optimism about the opportunities for constructive partnering across all the stakeholder groups.

By 2005, however, a surge in approvals led to payers becoming concerned over costs, while patients were faced with huge diversity in access. Scepticism about progress is rising. Policy makers and politicians are starting to mutter that the proponents of orphan medicines were making too much fuss and demanding too many privileges.

And industry itself faces big challenges. It is perceived negatively by big swathes of society – it is, some say too profit-oriented; only positive trial results are communicated; and patents are over-protected. But there is another view – that industry innovates for unmet needs, and is a major source of healthcare products; that models of success generate interest in the field; and that market exclusivity does not equate with monopoly.

In some members of the European Union, there is little communication between regulators, patients and industry; even the basic structures of dialogue do not exist. All stakeholders must work at creating those structures.

The answer is clear: the challenges of the new climate must be faced head on, openly and transparently, using the facts. And if the facts aren't there yet, then find them out.

Conclusions

Transparency

- Countries and companies should be more willing to share data and be transparent and open about the access they are providing to orphan medicines.
- The European Union should use its powers, where necessary, to extract this information and publish it.

Data

- Good data are not just the responsibility of industry. Physicians must improve their diagnostic skills as well in order to improve knowledge of the true prevalence of rare diseases and of the unmet health needs that they represent.
- Standardised databases are vital, but standardisation has to be built-in at the start of a project using criteria agreed across all stages of the process of developing a novel therapy and making it available to patients.
- Some companies say either they don't have the time to provide data, or that the data are confidential. They need a discussion to make them aware that unnecessary concealment of data is counter-productive, and that greater openness brings benefits to all.



B. Leufkens, E. Tambuyzer, T. Andersen, Y. Le Cam

Communication

- We need to be better at communicating the fact that innovation is a learning process – one that can be aided by better registries and innovative data sources.
- We are not good enough at communicating the value of orphan medicines. Eastern European countries in particular depend upon clearly presented communication to establish access.
- Industry, which provides the breakthrough medicines, is often perceived unfavourably. In rectifying this industry needs to engage on a constructive dialogue with other interested parties. It cannot shy away from controversial issues.
- Personal engagement is always the most powerful.



Birthe B. Holm,
Chair of the Workshop

“We need to talk to the public, not just each other, and engage personally. We need a recipe for success in communication: we need to share knowledge, but we also need a well-informed public, transparent grounds for decisions and an open dialogue between stakeholders... and not least, brave politicians to make the right decisions.”

– Birthe B. Holm, workshop chair, closing the meeting.

Programme

Thursday 18 October 2007

- Opening** – Alastair Kent, GIG / EGAN, Chair of EPPOSI
 – Birthe B. Holm, Sjaeldne Diagnoser / EURORDIS, Vice-Chair COMP, Chair of the Workshop
 – Torben Grønnebæk, Sjaeldne Diagnoser / EURORDIS

SESSION 1: How To Estimate The Value Of An Orphan Drug?

Chairs: Alastair Kent, GIG / EGAN and Andrea Rappagliosi, Merck Serono

Introduction – Andrea Rappagliosi, Merck Serono

The patient's view on health technology assessment for Orphan Drugs: are there models?

– Christine Lavery, MPS Society / EURORDIS

Is HTA an appropriate tool to promote access to Orphan Drugs?

– Finn Borlum Kristensen, Danish Center for Evaluation and HTA (DACEHTA), EUnetHTA

Access in a real-life setting: Member States experiences go live

– France: François Meyer, HAS

– The Netherlands: Wim Goettsch, CVZ

– UK: Edmund Jessop, Department of Health

Questions & Answers

A case study: industry experience – Jens Grueger, Novartis

The patients' view – Cees Smit, Dutch Genetic Alliance - VSOP

Interactive session with audience participation

Concluding remarks – Alastair Kent, GIG / EGAN

Dinner Debate – **The Communication on the Consultation regarding the European Action in the Field of Rare Diseases**

Friday 19 October 2007

SESSION 2: How Many Orphan Drugs – For How Many Patients?

Assessing Treatable Rare Diseases And The Proportion Of Patients Eligible For Treatment

Chairs: Torben Grønnebæk, Danmarks Bløderforening / EURORDIS and

Kerstin Westermark, COMP & Medical Products Agency, Sweden

Forecast of Orphan Drugs from epidemiological data – Ségolène Aymé, INSERM / Orphanet

Forecast of Orphan Drugs: perspective from modelisation – Fabrizia Bignami, EURORDIS

Forecast of Orphan Drugs: the FDA perspective – Tan Nguyen, FDA

Forecast of Orphan Drugs: the EMEA perspective – Paolo Tomasi, EMEA

Forecast of Orphan Drugs: an Industry perspective – Geoff McDonough, Genzyme

Interactive session with audience participation

Concluding remarks – Ségolène Aymé, INSERM / Orphanet

SESSION 3: How To Communicate About Orphan Drugs In The Real Life Setting?

Chairs: Terkel Andersen, Danmarks Bløderforening / EURORDIS and

Bert Leufkens, Dutch Steering Committee on Orphan Drugs

Patients' Access to Orphan Drugs in the EU: an EURORDIS survey – Yann Le Cam, EURORDIS

Orphan Drugs today: what are the communication challenges? – Bert Leufkens, Utrecht Institute for Pharmaceutical Sciences

Industry's communication about orphan drugs – Erik Tambuyzer, Chair EBE/EuropaBio Orphan Drugs Task Force, Genzyme Europe

Interactive session with audience participation

Conclusion – Birthe B. Holm, Sjaeldne Diagnoser / EURORDIS, Vice-Chair COMP, Chair of the Workshop

Press Release

Medicines for the few –

how can society better understand the reality of medicines for rare diseases?

Patients' Organisations, scientists, Biotechnology and Pharmaceutical industries highlight the need to better understand the reality of treatments for rare diseases in Europe and make concrete proposals to better communicate on the issue.

COPENHAGEN, Denmark, 19th October 2007 – One in 33 babies is born with a rare and serious genetic disease – most of which have no treatment. Yet the burden of rare diseases is immense and affects millions of individuals across Europe. This week more than 120 stakeholders from patients' organisations, academia and the biotechnology and pharmaceutical industries came together for the 8th EPPOSI Workshop on Partnering for Rare Disease Therapy & Development in the Danish Parliament, in the presence of HRH Crown Princess Mary of Denmark, to assess how European society can better understand the reality of rare diseases in Europe.

During the workshop, participants identified nine key areas that need to be addressed to tackle the continuing issue of rare disease treatments and their availability to patients.

1. Several factors need to be solved simultaneously in order to ensure rare disease patients have access to treatments. That's why cooperative dialogue such as this is key.
2. Access to treatments remains a political policy decision. Health technology assessments can be used to inform these decisions, but they remain policy decisions to be based on societal values and preferences.
3. It is reasonable to ensure that we only pay for treatments that work, but mechanisms are needed to ensure that this does not delay or prevent access while the evaluation is happening.
4. Suitable models do exist in several member states to make sure that patients do get access. These could serve as examples in other countries.
5. Access to rare disease treatments needs public acceptance, support and solidarity. We all need to have faith in our partners. Transparency and trust is vital – another reason that we all need to work together.
6. The EU's orphan Regulation is a success. It is anticipated that, within the next 5 years, Europe will have authorised between 85 to 105 new treatments, thanks to the Regulation. A third of them are currently developed by Small & Medium-sized Enterprises (SMEs).
7. Predictions of an avalanche of treatments eating into healthcare budgets are largely unfounded. The evi-

dence shows that there is a large gap between theoretical numbers of patients and those actually treated. Prevalence is largely over-estimated at the time of designation, not all patients are eligible for treatment, not all eligible patients have access to treatment at country level, and not all patients are diagnosed in a timely enough manner to allow them to be treated. While it is clear we do not have this data, information on the real situation should be gathered to allow us to identify potential solutions.

8. Registries should continue to be established to gather real-life evidence of the effect of available treatments through a coordinated efforts at European or even global level. The data should be made available to all researchers and external audiences.
9. The European Clinical Trials Directive has a negative effect on clinical research and development of treatments in the field of rare diseases. It raises costs and complexity – therefore, many companies choose to conduct their trials outside Europe and academic researchers do not organise trials here, meaning that rare disease patients do not benefit from such research.

Torben Grønnebæk, chairman of Rare Disorders Denmark and member of the European Organisation EURORDIS, knows the difficulties of suffering from a rare disease. He suffers from Wilson's syndrome, which threatened his physical mobility until he was finally diagnosed and received the appropriate treatment; commenting on the recommendations, he stated:

"Rare diseases are often overlooked, and because of this the first obstacle is get the proper diagnosis. Then it becomes a matter of treatment – does a treatment form actually exist? It is not very lucrative to develop and produce medicine for treating rare diseases, which is why we need to create a better platform for research and the development of this type of medicine. We cannot allow ourselves to ignore the sufferings of people afflicted by rare diseases simply because there a few of them."

EPPOSI aims to build dialogue and foster understanding between all parties with an interest in human healthcare. The joint recommendations developed in Copenhagen will be shared with European experts and decision-makers at all levels.

LIST OF PARTICIPANTS

* Co-Chair ** Member Organising Committee

Terkel ANDERSEN, Danmarks Bløderforening/
EURORDIS, Denmark

Muriel ASSOULINE, Alexion Europe, France

Giovanni ASTA**, EPPOSI, Belgium

Sécolène AYMÉ*, INSERM/Orphanet, Board
Member EPPOSI, France

Igor BEITIA ORTIZ DE ZARATE, French Institute
for Rare Diseases, France

Fabrizia BIGNAMI, EURORDIS, France

David BOOTHE, BioMarin Europe, United Kingdom

Finn BORLUM KRISTENSEN, Danish Center for
Evaluation and Health Technology Assessment/
EUnetHTA, Denmark

Karen BRONDUM-NIELSEN, Kennedy Center,
Denmark

Nikolai BRUN, Genzyme, Denmark

Bente BUNDGAARD, Journal of the Danish
Medical Association, Denmark

Mathilde CABANETTES, EPPOSI, Belgium

Francesca CAPRARI, Celgene, Italy

Jean-Jacques, CASSIMAN, University of Leuven,
ESHG, Board Member EPPOSI, Belgium

Emmanuel CHANTELOT**, European
Biopharmaceutical Enterprises, Belgium

Kjeld CHRISTENSEN, Dansk Tourette Forening,
Denmark

Catherine COURTIN, UCB Pharma, Belgium

Helen DAHL-HANSEN, Nordic Hypopara-
thyroidism Organisation, Norway

Loredana D'AMATO SIZONENKO, Geneva
University Hospitals, Switzerland

Jaap DE BOER, Genzyme, The Netherlands

Jérôme DEL PICCHIA**, Vienna Medical
Academy, Austria

Renate DOERNER, Genzyme, Germany

Annick DUBOSQ, INSERM, France

Androulla ELEFThERIOU, Thalassaemia
International Federation, Cyprus

Panos ENGLEZOS, Thalassaemia International
Federation, Cyprus

Safiye ER, Danish Medicines Agency, Denmark

Christina FASSER, Retina International,
Switzerland

Christian FRIIS, Zymenex, Denmark

Martina GARAU, Office of Health Economics,
United Kingdom

Ester GARNE**, Kolding Hospital, Denmark

Rüdiger GATERMANN, CSL Behring, Germany

Wim GOETTSCH, Health Care Insurance Board
(CVZ), The Netherlands

Michael, GRIFFITH, Fighting Blindness, Board
Member EPPOSI, Ireland

Torben GRØNNEBÆK, Sjaeldne Diagnoser/
EURORDIS, Denmark

Jens GRUEGER, Novartis Pharma, Switzerland

Angus GUNN, UCB Pharma, United Kingdom

Erica HACKENITZ, ZonMw/E-Rare, The
Netherlands

David HAERRY, European AIDS Treatment Group,
Switzerland

Harald HEEMSTRA, Utrecht Institute for
Pharmaceutical Sciences, The Netherlands

Jiri HERMANEK, Genzyme, Czech Republic

Lisse-Lotte HERMANSSON**, Genzyme,
Denmark

Birthe HOLM*, Sjaeldne Diagnoser/EURORDIS,
Denmark

Iben HOLTEN, Danish Cancer Society, Denmark

Wills HUGHES-WILSON, Genzyme, Belgium

Jolanda HUIZER, Steering Committee on Orphan
Drugs, The Netherlands

Michel HUYBRECHTS, Belgian Knowledge Health
Care Centre, Belgium

Erik HVIDING, Norwegian Medicines Agency,
Norway

Ulrike JAEGLE, Novartis International, Switzerland

Karsten JENSEN, Dansk forening for
Osteogenesis Imperfecta, Denmark

Lene JENSEN**, Rare Disorders Denmark/
EURORDIS, Denmark

Jørgen JEPPESEN, Rehabiliterings Center for
Muskelsvind, Denmark

Edmund JESSOP, National Commissioning
Group, NHS, United Kingdom

Kristina, JOHANSSON, Integral, Denmark

Sarah JONES, Genzyme, The Netherlands

Poul Eik JORGENSEN, Danish Haemophilia
Society, Denmark

Jacob KALUSKI, BioMarin Europe, United
Kingdom

Dennis KAYSEN, Integral, Denmark

Alastair KENT**, EGAN, GIG, Chair EPPOSI,
United Kingdom

David KING, Novo Nordisk, Denmark

Sophie KOUTOUZOV, French Institute for Rare
Diseases, France

Inge KRISTENSEN, National Centre for Rare
Diseases, Denmark

Rasmus KRISTENSEN, Integral, Denmark

Anne-Grethe LAURIDSEN, Gaucher, Denmark

Christine LAVERY, Society For Mucopoly-
saccharide Diseases, United Kingdom

Michael LEADER**, The European Association for
Bioindustries - EuropaBio, Belgium

Yann LE CAM**, EURORDIS, Board Member
EPPOSI, France

Lugdivine LE DEZ, Alexion Europe, France

Susanna LETO DI PRIOLO, Novartis Farma, Italy

Bert LEUFKENS, Dutch Steering Committee on
Orphan Drugs, The Netherlands

Catherine LEVINSON**, Merck Serono
International, Switzerland

Michele LIPUCCI DI PAOLA, EURORDIS, Italy

Asa LOMMELE, Alexion Europe, France

Kevin LOTH, Pharmion, United Kingdom

Gitte LUDWIG, Swedish Orphan International,
Denmark

Ellenor MATHISEN, Free-Lance Journalist,
Norway

Geoff McDONOUGH, Genzyme, United States

Tom MEULEMAN, BioMarin, Belgium

François MEYER, Haute Autorité de Santé,
France

Pawel MISKIEWICZ, Genzyme, Poland

Rod MITCHELL, European Federation of Crohn's
and Ulcerative Colitis Associations, Board
Member EPPOSI, United Kingdom

Kevin MOER, Journalist, Denmark

Eibhlin MULROE, IPPOSI, Ireland

Kirsi MYLLYNIEMI, In Medias Res, Finland

Gerard NGUYEN, Rett Syndrome Europe, France

Tan NGUYEN, FDA - Office of Orphan Products
Development, United States

Christel NOURISSIER, EURORDIS, France

Cor OOSTERWIJK, VSOP/EGAN, The
Netherlands

Anita OSBORNE, Rheoscience, Denmark

Irlin PLAMBECH, Alfa-1, Denmark

Ysbrand POORTMAN, International Genetic
Alliance, Board Member EPPOSI, The
Netherlands

Jes RAHBK, Rehabiliterings Center for
Muskelsvind, Denmark

Andrea RAPPAGLIOSI**, Merck Serono
International, Board Member EPPOSI,
Switzerland

Lillian REJKJAER, Copenhagen University,
Denmark

Pia RINGHOLM, Zymenex, Denmark

Clare ROBINSON, DEBRA Europe, United
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Maria RODRIGUEZ SANCHEZ, Baxter World
Trade, Belgium

Saskia ROMBACH, University of Amsterdam, The
Netherlands

Kathrin ROMMEL, INSERM, France

Fernando ROYO, Genzyme, Spain

Jacek RUSZKOWSKI, Centre for Public Health -
Leon Kozminski Academy of Entrepreneurship
and Management, Poland

Kjartan SAELENSMINDE, Norwegian Directorate
for Health and Social Affairs, Norway

Rosa SANCHEZ DE VEGA, Federacion Espanola
de Enfermedades Raras, Spain

Arrigo SCHIEPPATI, Clinical Research Center for
Rare Diseases, Italy

Francois SCHILTZ, Schiltz Health Care
Consulting, Switzerland

Ramona SCHMID, HRA-Pharma, France

Ad SCHUURMAN, Steering Committee on Orphan
Drugs/CVZ, The Netherlands

Tsveta SCHYNS, European Network for Research
on Alternating Hemiplegia, Austria

Cees SMIT, VSOP/EGAN, Honorary Member
EPPOSI, The Netherlands

Christos SOTIRELIS, UK Thalassaemia Society,
United Kingdom

Rumen STEFANOV, Information Centre for Rare
Diseases and Orphan Drugs, Bulgaria

Florence STEINHAUSLIN, Alexion Europe, France

Erik TAMBUYZER*, Genzyme, Board Member
EPPOSI, Belgium

Louise TAYLOR, INSERM, France

Geraint THOMAS, GlaxoSmithKline, United
Kingdom

Paolo TOMASI, European Medicines Agency,
United Kingdom

Sonja VAN WEELY**, Dutch Steering Committee
on Orphan Drugs, The Netherlands

Lisbeth VESTERGAARD, Swedish Orphan,
Denmark

Gudrun Ruth VIDARSDOTTIR, Hypopara-
thyroidism Europe, Iceland

Laurence VINDEVOGHEL, Neurochem,
Switzerland

Jean-Marie VLASSEMBROUCK, Baxter World
Trade, Board Member EPPOSI, Belgium

Philipp VON ROSENSTIEL, UCB, United States

François VUILLET, HRA-Pharma, France

Kerstin WESTERMARK, Medical Products
Agency, Sweden

Peter WROBEL, Clarity in Science
Communication, United Kingdom

Barbara WUEBBELS, BioMarin Pharmaceutical,
United States



A patient-led EU partnership of patients, academic science and industry, working together to advance healthcare policies for the prevention and treatment of serious diseases

EPPOSI was founded in 1994 on the initiative of patients' organisations, for the exchange of information and the discussion of human healthcare policies in the EU.

EPPOSI puts patients first in this dialogue, providing a forum for patients, academia, authorities and industry to discuss innovation and policies for healthcare, health technology, and the health outcomes for patients, especially those affected by chronic, life-threatening diseases – including rare diseases.

EPPOSI's ambition is to develop strategies that benefit present and future generations.

Objectives

- To encourage timely and regular exchange of information between stakeholders on the latest developments in human healthcare related to (bio-)medical research, policy and regulations; on the ethical, social, legal and political aspects of this type of research, and on biotechnology, notably for its application to human healthcare
- To promote a mutual understanding between patients' organisations, science, industry, and EU institutions
- To contribute to equal access for all to human healthcare products and services in the EU
- To support patients' organisations in presenting timely and effective contributions to the European political debate on all matters that concern them
- To raise public awareness in Europe on the opinion of patients and their organisations
- To help sustain a dialogue within society on progress in medical science through new technologies
- To advocate the development of therapies for unmet medical needs and to facilitate partnerships within society
- To function as an information coordination centre that encourages discussion, opinion-forming, and public debate in the area of human healthcare

Achievements

EPPOSI focuses on building dialogue, consensus positions and policy recommendations for the benefit of EU patients and consumers.

These consensus positions have provided building blocks for:

- the establishment of the European Orphan Medicinal Products Regulation
- the advancement of biomedical research and the value of innovation
- the timely access to innovative medicines
- several rare-disease therapy developments and partnerships
- East-West European collaboration among patient groups
- bio-banking

For more information, reports and publications, please visit our website or contact the EPPOSI office:

Rue de l'industrie 4, B-1000 Brussels, Belgium

Tel.: + 32 2 503 13 07

Fax: + 32 2 503 31 08

info@epposi.org - www.epposi.org