

**SWEDEN**

**EUROPLAN NATIONAL CONFERENCE**

**FINAL REPORT**

**26 November 2012, Stockholm**

# FOREWORD

**The EUROPLAN National conferences are aimed at fostering the development of a comprehensive National Plan or Strategy for Rare Diseases addressing the unmet needs of patients living with a rare disease in Europe.**

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

The EUROPLAN National conferences are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS – the European Organisation for Rare Diseases. For this purpose, EURORDIS nominated 10 EURORDIS-EUROPLAN Advisors - all being from a National Alliance - specifically in charge of advising two to three National Alliances.

**EUROPLAN National conferences share the same philosophy, objectives, format and content guidelines.** They involve all stakeholders relevant for developing a plan/strategy for rare diseases. According to the national situation of each country and its most pressing needs, the content can be adjusted.

During the period 2008-2011, a first set of 15 EUROPLAN National Conferences were organised within the European project EUROPLAN. Following the success of these conferences, a second round of up to 24 EUROPLAN National Conferences is taking place in the broader context of the Joint Action of the European Committee of Experts on Rare Diseases (EUCERD) over the period March 2012 until August 2015.

The EUROPLAN National Conferences present the European rare disease policies as well as the EUCERD Recommendations adopted between 2010 and 2013. They are organised around common themes based on the Recommendation of the Council of the European Union on an action in the field of rare diseases:

1. Methodology and Governance of a National Plan;
2. Definition, codification and inventorying of RD; Information and Training;
3. Research on RD;
4. Care - Centres of Expertise / European Reference Networks/Cross Border Health Care;
5. Orphan Drugs;
6. Social Services for RD.

The themes “Patient Empowerment”, “Gathering expertise at the European level” and “Sustainability” are transversal along the conference.

## I. GENERAL INFORMATION

<b>Country</b>	<b>Sweden</b>
<b>Date &amp; Place of the National conference</b>	26th of November 2012 Stockholm, Sweden
<b>Organiser</b>	Rare Diseases Sweden - Riksförbundet Sällsynta diagnoser
<b>Website</b>	<a href="http://www.sallsyntadiagnoser.se/">www.sallsyntadiagnoser.se/</a>
<b>Members of the Steering Committee</b>	<ul style="list-style-type: none"> <li>• Elisabeth Wallenius, President, Rare diseases Sweden;</li> <li>• Maria Gardsäter, Rare diseases Sweden/EUROPLAN advisor;</li> <li>• Susanne Bergman, National Board of Health and Welfare;</li> <li>• Hans Winberg, Leading Health Care</li> </ul>
<b>Eurordis – Europlan Advisor</b>	Maria Gardsäter, Rare diseases Sweden
<b>List of Workshops, Chairs and Rapporteurs</b>	
<b>Workshop 1</b>	<p><b>Definition, description, information and education</b></p> <p><b>Chair:</b> Anders Fasth, Information Centre for Rare Diseases</p> <p><b>Rapporteur:</b> Caroline van Mourik, Ehlers-Danlos Association</p>
<b>Workshop 2</b>	<p><b>Care Recommendations/ programmes for medical and non-medical needs</b></p> <p><b>Chair:</b> Kerstin Westermark, European Medicines Agency Scientific Committee for Orphan Medicinal Products, EMA/COMP</p> <p><b>Rapporteur:</b> Raoul Dammert, Rare diseases Sweden</p>
<b>Workshop 3</b>	<p><b>Research and registries of quality</b></p> <p><b>Chair:</b> Paul Uvebrandt, Centre for Rare Diagnoses at the Queen Silvia Children's Hospital</p> <p><b>Rapporteur:</b> Britta Berglund, Rare diseases Sweden/Ehlers-Danlos association</p>

<p><b>Workshop 4</b></p>	<p><b>National and international networks, regional centre and cross-border healthcare</b>  <b>Chair:</b> Robert Hejdenberg, CEO, Ågrenska  <b>Rapporteur:</b> Ulf Larsson, Rare diseases Sweden/AMC association</p>
<p><b>Workshop 5</b></p>	<p><b>Pharmaceutical / e-health</b>  <b>Chair:</b> Hans Winberg, Leading Health Care  <b>Rapporteur:</b> Veronica Hübinette, Rare diseases Sweden/MPS association</p>
<p><b>Workshop 6</b></p>	<p><b>Patient involvement, user involvement and empowerment</b>  <b>Chair:</b> Nicole Silverstolpe, Director of the processes at Regional Centre Stockholm  <b>Rapporteur:</b> Malin Holmberg, Rare diseases Sweden</p>
<p><b>Horizontal theme</b></p>	<p><b>Sustainability and user participation</b></p>
<p><b>Annexes</b></p>	<ul style="list-style-type: none"> <li>- Programme</li> <li>- List of participants in the workshops</li> </ul>

## II. INTRODUCTION

The programme was developed together with all stakeholders through the whole process. A reference group of the national strategy and function for rare diseases in Sweden had meetings several times prior to the conference, when also the EUROPLAN project was discussed.

Elisabeth Wallenius, Rare diseases Sweden; Maria Gardsäter, Rare diseases Sweden/EUROPLAN advisor; Susanne Bergman, National Board of Health and Welfare; Hans Winberg, Leading Health Care were in close contact prior to the conference.

A stakeholder meeting was held with following participants:

- Susanne Bergman, National Board of Health and Welfare
- Cecilia Gunnarsson, head of Regional Centre of Rare Diseases, Linköping university
- Nicole Silverstolpe, head of Patient Processes at the Regional Centre of Cancer in Stockholm
- Veronica Wingstedt de Flon, director of the National Function of Rare Diseases
- Kristina Gustafsson Bonnier, Regional Centre for Rare Diseases, Karolinska University hospital, Stockholm
- Per Gunnar Holmgren, journalist and moderator at the conference
- Hans Winberg, secretary General of the Academic think tank Leading Health Care
- Malin Holmberg, Rare diseases Sweden
- Britta Berglund, Rare diseases Sweden
- Paul Uvebrant, Regional Centre of Excellence at Sahlgrenska University hospital, Gothenburg
- Eli Fogelman, responsible for the web-streaming

Also invited to the meeting:

- Daniel Zetterberg, Ministry of Health and Social Affairs
- Fredrik Andersson, Ministry of Health and Social Affairs
- Billie Pettersson, Merck Sharp & Dohme (Sweden) AB
- Ulrika Eriksson, Swedish Association of Local Authorities and Regions (SALAR)
- Niklas Hedberg, The Dental and Pharmaceutical Benefits Agency (TLV)

### **EUROPLAN in Sweden is an intense success story**

The EUROPLAN Conference took place on Monday, 26 November 2012.

Participants, lecturers and organizers have been very satisfied. We had a fantastic day, to summarize the impressions of Maria Gardsäter, project manager at Rare Diseases Sweden, the National Swedish Alliance for Rare Diseases.

Approximately 150 people attended. Additional listeners followed the conference via webcast. Virtually all stakeholders were represented:

- Members of the Parliament (MPs),
- decision-makers at regional level,
- representatives from the competence centre for rare diseases
- officials from all relevant organizations, agencies and networks.
- 35 representatives from patient associations for rare diseases.

### **Starting position: Heavy criticism of a thin strategy proposals**

The national Board of Health and Welfare had published its proposal for a national strategy for rare diseases, and submitted it to the Ministry of Health and Social Affairs, only a few weeks before the EUROPLAN conference. However, the strategy had already been criticized before the conference by major stakeholders, including parliamentarians and professors at centres of excellence. The criticism was that the strategy was considered thin.

On 22 November 2012, a highly critical article on the strategy was published by Rare diseases Sweden's President Elisabeth Wallenius on the website Dagens Medicin ("Today's Medicine"), the important Swedish journal of health:

<http://www.dagensmedicin.se/debatt/strategiforslag-for-sallsynta-sjukdomar-ett-antiklimax/>

Rare diseases Sweden had, as a result of this "thin" draft strategy, made a shadow strategy of its own with additional suggestions. The shadow strategy was based on the priority areas identified in the draft strategy by the National Board of Health and Welfare. However, the shadow strategy gave many examples of what could be included in an action plan.

### **Preparation: Workshop for member organisations**

During the annual meeting of the chair persons of the Rare disease Sweden member organisations, the weekend before the EUROPLAN conference, Rare disease Sweden organized a workshop on the national strategy. About 50 representatives of member associations participated. This preparation was very appreciated. It also provided great pay off at the conference. Several members expressed that, after the workshop, they could use their personal experiences and stories on "another level", and increasingly contribute to the discussions.

Indeed, the members were also able to share their views and opinions during the EUROPLAN conference.

### **Procedure: Presentations, panel discussions and think-tanks**

Initial presentations during the plenary session were conducted in a brisk pace and the presentations were very interesting and rewarding. Firstly, was "the Swedish model", with the presentation of the Strategy proposed by the investigator Susanne Bergman; then the presentation of the Swedish National Function of coordination and information (NFSD) by

the director Veronica Wingstedt de Flon. This was followed by a comparative analysis between Swedish, French and Danish systems with regard to rare diseases, presented by Kerstin Westermark (EMA/COMP). Then, the sessions were followed by panel discussions with a number of respected and influential people who commented on the draft strategy.

The goal of the discussions in the morning was that they would be inspiring for think-tanks in the afternoon. Think-tanks/group discussions were also much appreciated as subjects and proposed measures for the strategy's priority areas. The outcomes were reported to all conference participants. A summary of the working groups' proposed measures are included in this conference report.

### **Results: Cooperation, centre steering, “scooter and motorcycle”**

Some keywords that constantly recurred during the conference were: cooperation, regional centre and central control. But how could the draft national strategy for rare diseases be implemented, so that the strategy is not just an empty gesture? This question was raised and analysed during the conference, with proposals made.

President Elisabeth Wallenius, of Rare Diseases Sweden, underlined that the Alliance would soon be 15 years old, which according to Swedish tradition means that you are allowed to drive a scooter/moped. “We are a messy 15-year-old, who is dissatisfied with social structures that do not function for those who have rare disorders”. She continued: “Instead of a scooter, we have a strategy that is currently parked in the Ministry's garage. From there, it must roll on for something to happen. But so far, it is making only 30 km per hour”. Elizabeth mentioned that scooters admittedly easily can be tuned up. Actually it is a powerful motorcycle that is needed, not a leisurely bike, and this is why the EUROPLAN conference was organised. If we tune up the scooter enough, maybe it will be a motorcycle in the end. That was the point of Elizabeth's message.

The metaphor of the “moped” (scooter) was a common thread throughout the conference. It was pointed out that “the scooter must have an engine” in the form of regional centres for rare diseases, and fuel in the form of special incentives, necessary if the tuning up is to be successful.

Many professionals, both within and outside the health care sector, work with rare diseases. A dilemma that was identified was that nobody has the mandate to coordinate all the different occupational groups that, each on its own, help people who have rare diagnoses. Some areas of activity were called “isolated islands”. Employees in charge of the coordination of care are needed.

Another parallel was that even when a bathroom is renovated, special funds are set aside for someone with the responsibility to supervise and coordinate the artisans.

But that does not happen when it comes to health care for those who have a rare diagnosis. As a matter of fact, a lot of efforts are put in the care and treatment of a patient with rare

diseases. However, considering that a patient who has a rare disease needs multidisciplinary care and access to specialised social services, a coordinator would be needed.

An invitation by a panel discussion was to “tune right”, adding that “an agile scooter is better than a dangerous gadget”. One of the speakers made a comparison with the situation in Denmark and France. The conclusion from the experiences of these two countries was that a Steering Committee is needed, with both muscles and a mandate to take actions.

Some of the content of Rare Diseases Sweden’s shadow strategy was presented and mentioned, appreciated and welcomed by members of the National Alliance as well as other participating stakeholders. For the very first time, MPs and regional politicians suggested to allocate a special budget for the implementation of the Strategy.

Elisabeth Wallenius recalled that this conference was the third one on the way towards a national strategy. The first one took place a few years ago and gathered some 30 participants. The second one was in fact the first EUROPLAN conference held in 2010, with 80 participants. On November 2012, the second EUROPLAN conference brought together more than 140 participants. This was a clear sign that rare diseases have gained more and more attention in recent years.

#### **Continued: Conference Conclusions presented to the Ministry of Health and Social Affairs and other stakeholders**

The result of the EUROPLAN conference will be reported to our European partner organization EURORDIS, conducting the EUROPLAN project. We will of course also present our conclusions from the conference, including the think-tank action proposals, to the Ministry of Health and Social Affairs and ask the following question: What will happen with the Board's policy proposals from October 2012? Other influential stakeholders, such as the Parliamentary Social Affairs Committee and the Swedish Association of Local Authorities and Regions (SALAR), will be made aware of the outcome of the National Association Rare diseases EUROPLAN conference 2012.



### III. PLENARY AND WORKSHOPS

#### Plenary Session

##### Summary of the plenary presentations

**1) National strategy for rare diseases**, Susanne Bergman, investigator at the National Board of Health and Welfare

Rare diseases Sweden initially thanked Susanne Bergman for her dedicated work and the tremendous knowledge she has obtained through working on the report. Susanne introduced the background of the national strategy, the conditions for further development and its implementation.

The Swedish government mandated the National Board of Health and Welfare to lay down the foundation for a national strategy for rare diseases, following the Recommendation of the Council of the European Union “on action in the field of rare diseases” (9 June 2009). The Council recommended that Member States “elaborate and adopt a plan or strategy as soon as possible, preferably by the end of 2013 at the latest”.

Summary of the background to the strategy:

- The Council of the European Union adopted a Recommendation for rare diseases in 2009.
- The European co-funded project, EUROPLAN (2008-2011) has developed a set of Recommendations. These are a ‘guidance’ document for the Member States for the development of National Plans and Strategies on Rare Diseases. It comes as an additional document to help implement the contents of the main European documents on rare diseases, and in particular the Council Recommendation on Rare Diseases.
- In October 2011, the National Board of Health and Welfare received the mission by the Government to develop a proposal for a national strategy for rare diseases. This came in addition to the task (or mission) of establishing a national function for RD, resulting in NFSD (see next section).
- In October 2012, the National Board did submit a proposal to the government on a national strategy for rare diseases.

The recommendations provided in the EUROPLAN project were the starting points for the proposal of a national strategy, and these recommendations were adapted to the Swedish national situation.

Outcomes: The objectives of the national strategy are as follows:

- People with a rare disease should have access to adapted health care and care of high quality.
- People with a rare disease should be treated according to the unique situation of the disease.

- Health care, social care and other community agencies should be coordinated, based on individual needs.
- People with a rare disease should have an established, individual care plan.

### Priority areas

In the national strategy proposal, seven priority areas have been identified. Work in these areas will help to achieve the objectives.

#### **1<sup>st</sup> area: Common definition.**

There is a reason to create a common concept. Swedish definition of a rare diagnosis is “1 in 10 000 inhabitants”, while the European definition is “5 per 10 000 inhabitants”.

#### **2<sup>nd</sup> area: Care recommendations and care programmes.**

These recommendations and programmes are needed to improve care for people with rare diseases.

#### **3<sup>rd</sup> area: Research.**

Research is a way to increase awareness of rare diagnoses.

#### **4<sup>th</sup> area: Drugs.**

An important part of the area is orphan drugs; prescribed medications for a rare disease. An ongoing government investigation into the pricing of drugs will also consider the funding of orphan drugs.

#### **5<sup>th</sup> area: E-health.**

E-health are tools that provide opportunities to create combined operations that require high skills that are accessible regardless of geographic location.

#### **6<sup>th</sup> area: National networks and regional centres.**

This area concerns the development of centres of regional excellence, which gathers expert knowledge, fosters research and professional development, serves as a unifying hub, providing advice and support, and is part of a network of expertise. In addition, regional competence centres also facilitate the formation of the national network.

#### **7<sup>th</sup> area: Patient Participation.**

Many patients with a rare disease have needs that require coordinated action from various public bodies. Many players have to interact and perform different activities to meet individual needs.

Susanne Bergman concluded by stating that the strategy documents now lie with the government for consideration by the Cabinet Office. The future will tell us what their decisions will be and what it will mean for the patients.

**2) National function for rare diseases (NFSD), Veronica Wingstedt de Flon, director, NFSD**

Veronica Wingstedt de Flon explained that NFSD will work in order to improve the life of people with a rare disease in the community and increase awareness of various social initiatives. The NFSD's mission includes the following tasks:

- Contribute to the increased coordination of health care resources for people with rare diseases, including social insurance, employment service, social services, voluntary organizations and others.
- Contribute to the dissemination of knowledge and information to all parts of the health service, other relevant public bodies and people with a rare disease and their relatives.
- Contribute to the exchange of knowledge, information and experience between the actors operating in the area.
- Make an inventory of available resources for people with rare diseases.
- Identify and create opportunities for the exchange of knowledge, experiences and information with other countries and international organizations.

2012 was the first year of operation for NFSD. During the first fiscal year, the following activities were prioritized:

- Participate in strategic planning.
- Mapping of patient organisations and social actors.
- Mapping actors involved in information management.
- Identification of knowledge materials that describe the continuum of care.
- Identification of information needs.
- Identify areas of particular concern.
- Initiate efforts to create regional and national networks.
- Establish networks for knowledge exchange.

The survey among patient organisations confirmed what already has been stated repeatedly:

- that social institutions that patients meet lack knowledge of the rare disease.
- the patient is forced to become an expert in the diagnosis.
- the need for the patients to be their own coordinator, to bridge lack of coordination between the bodies involved, mainly health insurance, employment service and social services.

Another finding was that 80 percent of the survey respondents lacked information about which rights you have.

The corresponding survey among social actors had resulted in the following conclusions:

- In health care, there are in Sweden about 15,000 different entities that are organized in many different ways.
- Health care providers are seeking collaboration to create care plans/guidelines and quality.
- Primary care should have clear referral pathways.
- There are good examples of effective activities to coordinate/collaborate on issues related to rare diseases.
- Many social actors are lacking internal guidelines and information about the specific needs that patients with rare diseases may have.
- The results depend to a large extent on the ability of the civil servants that are responsible of the case.
- More collaboration/cooperation is needed.
- More, easily accessible, comprehensive and customized information is required.

### **3) The project EUROPLAN, the Swedish strategy for rare diseases in comparison with other strategies**

Kerstin Westermark, Committee for Orphan Medicinal Products (COMP) of the European Medicines Agency (EMA)

Kerstin Westermark described the main characteristics of a rare disease:

- Late diagnosis – sometimes too late ...
- Lack of experts and centres for the treatment.
- No – or inadequate – treatment.
- Premature death and/or reduced quality of life.
- Impact on families.
- High healthcare costs – very expensive drugs.

Then Kerstin explained what we can learn from France and Denmark. France had been chosen because it is a pioneer in the field. Denmark was included in the comparison because it is a Nordic country which has many similarities with Sweden.

Her conclusions of what should be done in Sweden, stemming from comparisons with those two countries, were:

- Use the EU-wide definition of “rare disease”: No more than 5 in 10 000 individuals in the EU – provided for collaboration on rare diseases/disorders/diagnoses.
- Urge the national authorities to designate national centres of excellence based on the EUCERD Recommendations on “quality criteria for centres of expertise for rare diseases in

Member States” that interact with European Reference Networks. EUCERD is the “European Union Committee of Experts on Rare Diseases “ of the European Commission.

- Establish a steering committee with the mandate to decide and take actions – a lesson from both France and Denmark.

Kerstin continued by listing the factors of success:

- What is best done at the EU level should be done at EU level, for instance approval of orphan drugs, European reference networks, registries and treatment guidelines.
- What is best done at the national level should be done at national level, for instance pricing, benefit assessment, designation of national centres of expertise, provision of care, information.
- For a successful outcome: Coordinate the national level with the EU level.

Kerstin summed up what needs to be done, both nationally and internationally, to achieve success in the work of improving living conditions for those with rare diseases:

Cooperation, more collaboration and cooperation even more!

Four panel discussions were carried out, after the introductory presentations. The discussions are documented here:

[http://sallsyntadiagnoser.se/virtupload/sallsynta/content/32/Europlankonferens\\_Sallsynta\\_diagnoser\\_2012.pdf](http://sallsyntadiagnoser.se/virtupload/sallsynta/content/32/Europlankonferens_Sallsynta_diagnoser_2012.pdf)

## **EUROPLAN, workshop 1: Definitions, coding, education on classification and coding**

Moderator: Anders Fasth

Proposed measures:

a) Sweden should introduce the EU definition of rare disease as a disease affecting not more than 5 per 10 000.

Justification:

It is better to use the same definition as most of the other EU Member States.

- To achieve consistency, Sweden should therefore use the same concept definition as elsewhere.
- The connection to the orphan drugs: we should not have different boundaries, one for the group of patients concerned (1 in 10,000) and another (5 at 10,000) that control which drugs are intended.

b) Include both the incidence and prevalence, as this would include also the number of people who are having a particular diagnosis at a given point in time.

Justification:

- In order to make as accurate a forecast as possible over number of diagnostic support, one must reckon both the incidence and prevalence of a disease.
  - The incidence is needed in addition to improving knowledge on diagnosis and treatment.
- c) Consistently use the word “rare” instead of “unusual”.

Justification:

- “Rare” is perceived to have a more positive connotation than “unusual”.

d) Educate health care professionals on rare diseases.

Justification:

- Training of health workers is necessary to increase health care awareness of rare diseases.

Excerpts from group discussion:

- Should we continue to use our Nordic definition? Or is it time to start applying the much more extensive EU definition of what constitutes a “rare” diagnosis?
- The EU has a much broader approach that also includes rare forms of cancer.
- The conference discussed whether we should change ourselves in the Nordic countries, even though we were the first to recognize and define the term “rare” diagnosis. That is the reason why Sweden, and other Nordic countries, have a different definition than the rest of Europe: the Nordic pioneering work on rare disorders which began in the 1980's. But as the EU does not control health care of the member countries, Sweden has been able to maintain its narrower definition even after the adoption of the EU Regulation on Orphan Drugs (2001).
- When EU began to take an interest in the field of rare diseases, the definition for a rare disease was influenced by commercial purposes. The EU definition “no more than 5 per 10,000” is a good incentive to develop orphan drugs.
- We concluded that it is impractical to have two different conceptual definitions, one in Europe and another one in Sweden.
- An additional conclusion was that the term “diagnosis” is preferred by those who have the condition and their related parties. The word “disease” can be perceived as a stigmatizing label. However, when it comes to care, it turns out that people have a disease. A physician treats a disease, not a diagnosis. So it is not surprising that health care professionals also use the term “illness”, it was said too.
- Consensus was that “rare” is preferable, compared to “unusual”, when considering words denomination. The patients have experienced that “rare” has a more positive connotation. It is not easy to work with the words “disease” and “handicap”, which sound negative. Then it is better to be “rare”.

- It is important to educate health care professionals and raise awareness of rare diseases.

## **EUROPLAN, Workshop 2: Care Recommendations/programs medical and non-medical needs**

Moderator: Kerstin Westermark

### Proposed measures:

a) Determine: What is meant by a “care program”.

Justification:

- Definitions vary: Is a care program only a medical chart indicating the specialists, or does it refer to a chain of different social activities, within and outside the health care sector?
- Sometimes a strictly medical care programs suffices, but not when a patient belongs to several clinics and also needs non-medical interventions.

b) Determine: Who will pay for the care programs?

Justification:

- The money is available regionally. However, there is a need for a body/authority to settle cases when not everyone agrees.
- Funding is needed to develop an overall care program template model, and for those who do diagnosis-specific care programs based on the template model.
- Examine: To what extent are there already care programs for rare diseases? Alternative: What do current care programs for rare diseases contain?
- Starting point: Develop a template/checklist of a good model for a care program. Start with a minimum level; do not attempt to “do it all” at once.
- Note: If possible, start from the rare disease database of the National Board of Health and Welfare and extend it with information about treatment recommendations/programs.
- Investigate: What can/should care programs do?

Action Points:

- Rare diseases Sweden is responsible for developing a general model to be produced. There should be a “program of care” template model for all rare diagnoses, with both medical and non-medical needs considered. The general template is an incentive to do diagnosis-specific care programs.
- When the diagnosis-specific care programs are to be developed, we will also need to turn to the EU level to find specialists who could be involved in the specific programs.
- Provide specialist associations (associations of medical specialists in various fields) to be commissioned to write care programs.
- Other professions, such as physiotherapists, should also be able to initiate care programs.

- Use Orphanet as a model, translate and pick info from there.
- Relevant patient organizations must also be able to initiate care programs. Thus, the patient organizations must be given the resources necessary to educate their representatives in this field.
- Health programs linked to regional centres. The programs should be linked to all the areas inside and outside health care.
- It is assumed that health programs are linked to the quality of monitoring and evaluation.

### **EUROPLAN, Workshop 3: Research and registers of quality**

Moderator: Paul Uvebrant

#### Proposed measures

- a) Research on the rare diagnoses requires special resources.
- b) Research funding is needed for basic infrastructure research, especially adopted for rare diseases.

Comments:

- The National Associations of Rare Disorders (Rare diseases Sweden or Swedish patients' organisations in general) should lobby for the new research bill, so that rare disorders are addressed in the research bill and research funds will be earmarked for rare diagnostic groups.
- Other organizations, such as those connected to common diseases, are lobbying against the new research bill and already have considerable influence.

- c) Collaboration between centres is important from a research standpoint.

Justification:

- Some form of national connectivity is needed, which involves collaboration between different centres.

- d) Build patient records as a basis for research.

Justification:

- Information management in health care need to be clarified.
- Patient records describe the diagnosis, which measures are taken by health care professionals and other actors, and what are the results of their efforts.
- Significantly, not only medical research and care efforts, even social security should be included in patient records.
- The causes of symptoms need to be clarified, facilitated by patient registries.
- Medical records should be linked to the register, in order to avoid double entries of data.



e) National structures to support the registry. This refers to organization of the records, who handles and finance them, how records are, how to get the data from the registers and who has access to registry data.

Justification:

- The Central Statistical Office collects data on population and the Government checks the result. This is important to rare diseases in this context, so that the Government gets indications concerning public health in the rare diagnosis groups.
- Co-operation is necessary when the registers are built up. This is undoubtedly a task for professionals, but we must find forms of patient participation.
- Transnational research is important. There patient registries also play a role. But it is unclear what applies to provide data to another country.

#### **EUROPLAN, Worskhop 4: National and international networks; regional centres and cross border health care**

Moderator: Robert Hejdenberg

Action Points: Each patient must be supported, even those who did not receive a diagnosis. Collaboration with patient organizations is very important.

- A national cooperation must be achieved.

#### Proposed measures

- a) Make an inventory of the fields of expertise at each of the seven university hospitals
- b) Establish regional centres for rare diseases.

Justification:

- Someone has to control the establishment of the centres, with some form of authorization.
- Patients should have a say in the establishment of the centre.
- Special incentives must be provided to allow interested parties to be involved.
- Teamwork is needed to guarantee professional's competence, both within and outside the healthcare sector.

c) Cross-border care is very important. It should be easy to access health care in other EU countries when the best care can be found outside the native country.

Justification:

- Regional centres are the solution to many problems.
- There must be a sharing of knowledge between centres, at national and EU level and even at international level. (EU level is not always enough to find specific expertise.)

#### Action Points, e-health

##### Discussion, summary

- E-health has great potential. It quickly reaches out with knowledge about rare diseases, both concerning diagnosis and among the professionals.
- If the necessary knowledge is not available in a country, the base for a question, e.g. X-rays, can be easily transferred to a skilled centre abroad for an assessment.
- However, it is difficult to evaluate the information spread through e-health instruments, and assess its quality.

##### Proposed measures, other:

- National medical records, the patient may recognize that even other hospitals may share their medical records.
- The information provided using e-health instruments should be quality marked.
- It would be good if the National Board of Health and Welfare had more information relevant to e-health.

### **EUROPLAN, group 5: Pharmaceuticals / e-Health**

Moderator: Hans Winberg

#### Actions points:

- Let specialized doctors at a medical centre determine whether treatment should be initiated for the current diagnoses (those who may need orphan drugs).

#### Proposed measures

a) There should be a national fund for orphan drugs, financed either by the State or jointly by the county councils.

#### Justification:

- The current model, with decentralized payment of expensive orphan drugs means that access to treatment of orphan becomes arbitrary. It may depend on the individual physician's knowledge and point of view.
- The decentralized payment system may also mean that even if a treatment were approved, the payment of the treatment may be delayed to the following year, which is unreasonable in degenerative disease.
- A national funding prevents the risk of delay for economic reasons, providing durability.

## **EUROPLAN, group 6: Patient Participation, user influence and empowerment**

Moderator: Nicole Silverstolpe

Questions:

- How to design a financial system for rare diagnosis groups' basic organizational activities.
- How can patients' representatives be part of the development of care program for rare diseases?
- Experience has shown that patients always (often) are consulted at a late stage in the decision process, when planned activities are already in place.
- When regional and, later, national centre, are going to be established, patients need to be included from the outset.

Patients must participate in the entire process of establishing centres.

- We, as patient organizations, are expected to participate in the organisation of care. In order for patient organizations to be able to do just that, they need to have resources available for their participation. It must be possible to deploy the most appropriate representative of the group that each patient organization chooses.
- Make sure that patients are involved when the centre is established and care programs are in place, by creating the conditions for an efficient user participation, including available resources for compensation of their representatives.
- The rules for which associations are eligible must be reviewed in general. Contribution rules are based on an outdated approach to volunteer involvement. Advocacy is not the same as it used to be, the voluntary sector does not attract young people.

However, we do not have time to wait for a review of the benefit system. For rare diagnosis groups, we must find a special arrangement, for example to fund our member meetings.

Action Points:

We should not designate the financial support for the associations of rare diseases. However, we will clarify the justification for such a support

The following can be cited:

- A compelling argument is that patient organizations play an important role in knowledge generation. Many patients' representatives are expected to set up non-profit organisations, to help health care professionals as well as other patients in providing information about the diagnosis and being a place to turn to for help.
- Would it be possible to find out how much voluntary work is carried out by patient organizations for the health care system? It is important to present the results to decision makers as a justification for financial support to the patient organizations.

- Can such a survey serve as a basis for compensation, where patient organizations have a mandate to spread knowledge on the diagnosis to stakeholders (patients and the profession) and hence are given financial compensation for this task?
- All associations involved in rare disorders should be able to lift government subsidies (in the form of core funding) on specific basis.
- There should also be a grant for patient representatives' participation in studies/investigations relevant to rare diagnosis groups.

#### Proposal: funding for international cooperation

- Sharing and disseminating knowledge within the EU is important, particularly concerning the rarest diagnostic groups (“ultra rare”).
- Today the associations of the patient groups struggle economically to send representatives to participate at the European level committees/conferences.

#### Proposals: care programs

- When care programs are developed for specific diagnoses, representatives from the relevant rare disease association should be included already at an early stage, thus not only representatives from the National Alliance.
- Give the task to develop a generic form (template) for a care program for rare diseases to the National Alliance, Rare diseases Sweden.
- We must include “the ordinary into the extraordinary”, otherwise the task to make care program will be too overwhelming.
- There are few doctors who are ready to cooperate, ignore their own prestige and take on “best practices”.
- We are so scarce in some diagnostic groups, that in some cases we may have to work at the European level to provide a care program.
- Give to NFSD the task of coordinating the work of care program for rare diseases.
- Patient representatives need training to be able to act patient representatives and be representative.
- Rare diseases Sweden could administer such programs. Patients need to learn to “read between the lines”, when the profession presents its arguments.

#### Sustainability

- Right now, the doctors and health administratives/civil servants' specific interests control focus on the different centres.
- We need to establish criteria for these centres, which guarantee a better life for people who have rare diseases.
- We might need a common criteria for all of Europe.

- We must also have procedures for evaluating and monitoring regional activities that are emerging (national monitoring responsibility).
- The sustainability of patient organizations is an important thing that require funding.
- The strategy provides no information about who is responsible for evaluation and monitoring. Could this responsibility be given to NFSD?

#### Patients' involvement

- It is important that Rare diseases Sweden acts as a consultative body to government for investigations.

#### General user participation

- In medical school knowledge about rare diseases ought to increase, which in turn can generate interest for research diagnoses. The doctors cannot learn all diagnoses, but they must be aware of the concept.
- One of the big problems is that carers must understand and accept that parents or patients know their own diagnosis or situation.
- Administrative structures on the national level for patient programs (e.g. Ågrenska), instead of training grants to be sought from their home county.
- We must work together with EURORDIS to create European Reference Networks as some diseases are so rare, that we need to pool the expertise at the EU level.

## Annexe I - Final Programme of the National Conference

09:00 **Registration and breakfast**

09:30 **Rare Disorders since 1998 in five minutes**, Elisabeth Wallenius, Rare diseases Sweden

09:40 **National strategy for rare diseases**, Susanne Bergman, National board of health and Welfare

09:55 **National function for rare diseases**, Veronica Wingstedt de Flon, Nationella Funktionen Sällsynta Diagnoser (NFSD)

10:10 **EUROPLAN Project, the Swedish strategy for rare diseases in comparison with other countries' strategies**, Kerstin Westermark, European Medicines Agency/COMP

10:30 Debate group: **What effect will come out of the proposed National Strategy for RD?**  
Moderator: Per Gunnar Holmgren. Participants: Susanne Bergman, National board of health and Welfare; Hans Winberg, Leading Health Care; Veronica Wingstedt de Flon, NFSD; Andor Wagner, EUCERD, Elisabeth Wallenius, Rare diseases Sweden

10:50 **Break**

11:15 Debate group: **Public opinion, politics, profession, or user needs – who control priorities in caring for the rare?** Moderator: Per Gunnar Holmgren. Participants: Lena Hallengren, Member of Parliament/MP (Social Democratic Labour Party); Finn Bengtsson, MP (Moderate Party); Birgitta Rydberg, The medical mission of Swedish Association of Local Authorities and Regions/SALAR (Moderate Party); Marie Wedin, Medical Association; Ulf Larsson, Rare diseases Sweden

11:40 Debate group: **Care according to Swedish law and equal access to orphan drugs, how is it possible?** Moderator: Per Gunnar Holmgren. Participants: Anders Blanck, LIF/association for the research-based pharmaceutical industry; Niklas Hedberg, The Dental and Pharmaceutical Benefits Agency (TLV), Anders Hallberg, NLT-group (Nya läkemedelsterapier/"New pharmacotherapies") at SALAR; Fredrik Andersson, pharmaceutical and pharmacy investigation; Penilla Gunther, MP (Christian Democrats)

12:00 **Patient-centred care, how is it rare for patients with complex health care needs?**  
Moderator: Per Gunnar Holmgren. Participants: John Assarsson, Patient Power Inquiry; Jon Rognes Founder of Leading Health Care; Barbro Westerholm, MP (Liberal Party); Nanda Holm, who has personal experience of living with a rare diagnosis; Robert Hejdenberg, Ågrenska

12:30 Lunch, 13:30 The program will continue in the group rooms.

13:30 **Think-tanks focusing on the implementation of the strategy, how do we translate the wise words into action?**

### **1. Definition, designation, information and education.**

Moderator: Anders Fasth, Information Centre for Rare Diseases

**2. Care recommendations/programmes for medical and non-medical needs.**

Moderator: Kerstin Westermark, European Medicines Agency scientific committee for Orphan Medicinal Products/COMP

**3. Research and registries of quality.**

Moderator: Paul Uvebrandt, Centre for rare diseases at the Queen Silvia Children's Hospital

**4. National and international networks, regional centres and border healthcare.**

Moderator: Robert Hejdenberg, Ågrenska

**5. Orphan Drugs/e-Health**

Moderator: Hans Winberg, Leading Health Care

**6. Patient Participation, user involvement and empowerment.**

Moderator: Nicole Silverstolpe, director of the processes at the regional cancer centre in Stockholm

15:15 Afternoon snack

15:30 **The chairs of the think-tanks presents the outcome of the discussions**

16:15 **What do we do now?** Elisabeth Wallenius, Rare diseases Sweden and Anders Olason, President and founder of Ågrenska, also representing European Patients' Forum

16:30 **Mingle in the conference foyer**

## Annexe II – Participants List

<b>Name</b>	<b>Organization</b>	<b>Stakeholder</b>
Ann-Christin Andersson	Avdelningen för kunskapsstyrning och FoU Region Skåne	Academic/Researcher
Anna Brommée	Lunds universitet	Academic/Researcher
Siri Hennessey	Lunds universitet	Academic/Researcher
Karin Fernler	Leading healthcare	Academic/Researcher
Hans Kollberg	Uppsala Universitet	Academic/Researcher
Jon Rognes	Leading healthcare	Academic/Researcher
Jan Wahlström	Sahlgrenska	Academic/Researcher
Hans Winberg	Leading healthcare	Academic/Researcher
Johanna Wiss	patentmaktsutredningen	Academic/Researcher
Gunnar Ahlsten	Akademiska barnsjukhuset	Clinician/GP
Birgitta Bergendal	Odontologiska Institutionen	Clinician/GP
Anders Fasth	Drottning Silvias barnsjukhus	Clinician/GP
Cecilia Gunnarsson	Universitetssjukhuset i Linköping	Clinician/GP
Ulf Kristoffersson	Labmedicin Skåne	Clinician/GP
Helena Magnusson	SUS	Clinician/GP
Else Månsson	Barn- och Ungdomshabiliteringen	Clinician/GP
Ricard Nergårdh	Karolinska metabolteam ALB	Clinician/GP
Britt Nilsson	Habiliteringen NSÖ	Clinician/GP
Magnus Nordenskjöld	Karolinska institutet	Clinician/GP
Johanna Norderyd	Kompetenscentre	Clinician/GP
Ann Nordgren	Karolinska universitetssjukhuset	Clinician/GP
Hans Olsson	Överläkare Länssjukhuset Ryhov	Clinician/GP
Johan Richter	Överläkare Hematologi	Clinician/GP
Lotta Sjögren	Mun-h-centre	Clinician/GP
Pia Sjölund	Neuro- och hjärnskade-Team Norr	Clinician/GP
Eva-Lena Stattin	Akademiska	Clinician/GP
Jimmy Sundblom	Uppsala Universitet	Clinician/GP
Johan Svensson	Överläkare SUS	Clinician/GP
Alexandra Topa	Sahlgrenska	Clinician/GP
Paul Uvebrant	Drottning Silvias Barnsjukhus	Clinician/GP
Ulrika von Döbeln	Karolinska	Clinician/GP
Marie Wedin	Sveriges Läkarförbund	Clinician/GP
Rolf Zetterström	Centrum för Medfödda Metabola Sjukdomar Karolinska	Clinician/GP
Britt Marie Anderlid	Klinisk genetik, Karolinska	Clinician/GP
Anna-Lena Johansson	Arbetsterapeut, vuxenhabiliteringen	Clinician/GP
Christina Lundqvist	Förbundet Sveriges Arbetsterapeuter	Clinician/GP
Daniel Nowinski	Akademiska sjukhuset	Clinician/GP
Christina Clementsson Kockum	Klinikchef Barnkliniken Lund	Clinician/GP
Karin Samuelsson	Förbundet Sveriges Arbetsterapeuter	Clinician/GP
Ann Nachemson	Sahlgrenska sjukhuset	Clinician/GP



Desirée Gavhed	Orphanet	Healthcare Professional (other than clinician or GP)
Christina Greek Winald	Ovanliga diagnoser	Healthcare Professional (other than clinician or GP)
Kristina Gustavsson-Bonnier	Habilitering & hälsa	Healthcare Professional (other than clinician or GP)
Rune Johansson	Föreningen Sveriges Habiliteringschefer	Healthcare Professional (other than clinician or GP)
Staffan Larsson	Vuxenhabiliteringen Örebro	Healthcare Professional (other than clinician or GP)
Stina Löf	Vuxenhabiliteringen Örebro	Healthcare Professional (other than clinician or GP)
Nicole Silverstolpe	SLL	Healthcare Professional (other than clinician or GP)
Rula Zain Luqman	Regionalt kompetenscentrum för sällsynta diagnoser, KS	Healthcare Professional (other than clinician or GP)
Elizabeth Hernberg Ståhl	Late Phase Solutions Europe AB	Industry
Karolina Antonov	LIF	Industry
Anders Blanck	LIF	Industry
Eva Bremberg	Pfizer	Industry
Marianne Eriksson	Bayer	Industry
Kerstin Falck	Pfizer	Industry
Tomas Gloveus	Genzyme	Industry
Björn Hellström	Celgene	Industry
Lena Kristoffersson	Genzyme	Industry
Bernd Kuntscher	Orphan Europé	Industry
Jan Lagerström	Genzyme	Industry
Dag Larsson	Bayer	Industry
Åsa Magnusson	Actelion	Industry
Anita Nyström	Biomarin	Industry
Billie Pettersson	MSD Sweden	Industry
Peter Rybäck	Alexion	Industry
Jonas Sandgren	Celgene AB	Industry
Astrid Torstensson	Astra	Industry
Pål Faxén	Grifolds	Industry
Mattias Bankel	Shire	Industry, insurer
Finn Bengtsson	Socialförsäkringsutskottet	Medical /Learned society
Robert Hejdenberg	Ågrenska	Other
Per Gunnar Holmgren	journalist/Leading healthcare	Other
Hanna Isaksson	Ex-centre	Other
Marie Wikström	Ex-centre	other
Inger Ahlbin	Alfa-1 Sverige	Patient representative
Britta Berglund	Sällsynta styrelse	Patient representative
Kerstin Bjernevik	Riksföreningen mot Porfyrisjukdomar	Patient representative
Olle Bäckebo	Fabryföreningen	Patient representative
Ulf Carlsson	PKU-föreningen	Patient representative
Ulf Carlsson	Alfa-1	Patient representative

Helene Cederroth	Willefonden	Patient representative
Mikk Cederroth	Willefonden	Patient representative
Raoul Dammert	Riksförbundet Sällsynta diagnoser	Patient representative
Daniel Degerman	Sällsynta diagnosers styrelse	Patient representative
David Eriksson	Föreningen för Tuberös skleros	Patient representative
Mari Eriksson	Föreningen för Tuberös skleros	Patient representative
Eva Falk Carlsson	PKU-föreningen	Patient representative
Maine Forsberg	Sällsynta styrelse	Patient representative
Rangela Fredsjö	Möbius syndromföreningen	Patient representative
Maria Gardsäter	Riksförbundet Sällsynta diagnoser	Patient representative
Annika Hallberg Juhlin	TSC föreningen Sverige	Patient representative
Monica Hedman	Ehlers-Danlos syndrom	Patient representative
Malin Holmberg	Riksförbundet Sällsynta diagnoser	Patient representative
Veronica Hübinette	MPS diseases	Patient representative
Agneta Hult	LAM Academy	Patient representative
Elisabeth Ivarsson	Svenska Noonanföreningen	Patient representative
Ulf Larsson	AMC	Patient representative
Anneli Larsson	PIO	Patient representative
Lise Lidbäck	NHR	Patient representative
Beth Lindecrantz	Sturge Weber syndrom	Patient representative
Barbro Medén	Fabryföreningen	Patient representative
Neven Milivojevic	Aniridi Sverige	Patient representative
Monica Nanclèr	NF-förbundet	Patient representative
Joachim Nilsson	Fabry disease	Patient representative
Alexandra Puccini	NF-förbundet	Patient representative
Catrine Reuterborg	Alfa-1 Sverige	Patient representative
Björn Rosenkrantz	Kraniofaciala föreningen	Patient representative
Erik Rosenkrantz	Kraniofaciala föreningen	Patient representative
Caroline Sandberg Kvick	Riksföreningen mot Porfyrisjukdomar	Patient representative
Ingela Sjöberg	NF-förbundet	Patient representative
Mats Sjöberg	Aniridi	Patient representative
Darko Tilldal	Svenska EB föreningen	Patient representative
Caroline van Mourik	Ehlers Danlos syndrom Riksförbund	Patient representative
Anette von Kock	LAM Academy	Patient representative
Ingrid Wadenheim	Noonan syndrom	Patient representative
Elisabeth Wallenius	Sällsynta diagnoser	Patient representative
Janne Wallgren	HSO Uppsala	Patient representative
Annika Wettlén	RMT	Patient representative
Dan Wikström	Möbius syndrom	Patient representative
Anna Winberg	Beckwith-Widerman syndrom BWS	Patient representative
Lise Lidbäck	NHR	Patient representative
Håkan Sjunnesson	Neurologiskt Handikappades Riksförbund	Patient representative
Margaretha Nordin	Hypofysis	Patient representative
Birgitta Rydberg	SKL Sjukvårdsdelegationen	Politician
Penilla Gunther	Näringsutskottet	Politician
Anders Hallberg	SKL NLT	Politician

Lena Hallengren	Socialutskottet (s)	Politician
Lars-Joakim Lundquist	SKL Sjukvårdsdelegationen	Politician
Barbro Westerholm	Socialutskottet (fp)	Politician
Fredrik Andersson	Apoteks och Läkemedelsutredningen	Public administration (local, regional or national)
Susanne Bergman	Socialstyrelsen	Public administration (local, regional or national)
Ulrika Eriksson	SKL	Public administration (local, regional or national)
Gunilla Gunnarsson	SKL	Public administration (local, regional or national)
Anders Olauson	Ågrenska	Public administration (local, regional or national)
Ulla Reiderstedt	Spsm	Public administration (local, regional or national)
Anders Tollmar	Landstinget Gävleborg	Public administration (local, regional or national)
Monica Ulrikson	Samordnare Handikappfrågor	Public administration (local, regional or national)
Andor Wagner	Socialstyrelsen	Public administration (local, regional or national)
Kerstin Westermark	EMA	Public administration (local, regional or national)
Lennart Magnusson	Nationellt kompetenscentrum Anhöriga	Public administration (local, regional or national)
Niklas Hedberg	TLV	Public administration (local, regional or national)
Johan Assarsson	Patentmaktsutredningen	Public administration (local, regional or national)
Susanne Fägersten Sabel	Ovanliga diagnoser, Socialstyrelsen	Public administration (local, regional or national)
Erik Bissessar	Alexion	Social worker