EUROPLAN National conference

FINAL REPORT

I. General information

Country	**Sweden**
Date & place of the National Conference	**Nov. 11, 2010, Stockholm**
Website	**www.sallsyntadiagnoser.se**
Organisers	**Elisabeth Wallenius, President of Rare diseases Sweden** **Britta Berglund, Advisor EURORDIS**
Members of the Steering Committee	**Elisabeth Wallenius, President of Rare diseases Sweden** **Britta Berglund, Rare diseases Sweden, Advisor** **Anders Olauson, Ågrenska** **Annika Larsson and Désirée Gavhed, Rare Diseases Portal, Rare Diseases Platform (Orphanet), EUROPLAN, Secretariat ICORD, Karolinska Institutet** **Marie Lawrence and Jonas Karnström, National Board of Health and Welfare**
Names and list of Workshops	**Theme 1 - Methodology and Governance of a National Plan/Strategy, Definition, codification and inventory of RD** **Theme 2 - Research on RD** **Theme 3 - Standards of care for RDs - Centres of Expertise (CoE)/ European Reference Networks (ERN) and Orphan Drugs** **Theme 4 - Patient Empowerment and Specialised Services**
Chairs and Rapporteurs of Workshops	**WG 1. Chair Anders Fasth, professor, Gothenburg University** **rapporteur Maine Forsberg, Rare diseases**

	Sweden **WG 2. Chair Maria Westfält, Ministry of Education and Research** **rapporteur Britta Berglund, Rare diseases Sweden** **WG 3. Chair Birgitta Bergendal, Odont.dr, National Oral Disability Centre, Jönköping rapporteur Ulf Larsson, Rare Diseases** **WG 4. Chair Robert Hejdenberg, Ågrenska, rapporteur Raoul Dammert, Rare diseases Sweden**
Attachments (programme, list of participants, etc.)	**Programme** **Invitation** **List of participants**

ll. Main Report

Discussions about the programme started in 2009 and four meetings were held in the steering group in 2010 with the latest meeting on Oct. 15, 2010. Mr Anders Olauson was moderator during the day. People from all parts of the society, 85 stakeholders, participated. Mr Anders Olausson meant that all decision makers that matter were present, the list of participants was a dynamic network! He continued with a short description of how the awareness of rare diseases started to grow in Sweden. During the 90s some funding was available to apply for from the National Board of Health and Welfare in order to stimulate development in this area. Many projects started, but were later closed down since new funding was not found. One project that has been kept ongoing is the data base of the National Board of Health and Welfare. It was first named 'Small and less known diagnoses' but is now named 'The Database for Rare Diseases'. EURORDIS was established in 1997 and the Swedish Rare Diseases Association was formed in 1998. The EU Council Recommendation in 2008 is now the pillar stone for the conferences held in 15 countries during 2010, which is fantastic. After the introduction by Mr. Olauson, Ms Elisabeth Wallenius, who is the President of Rare Diseases Sweden, greeted all participants welcome to the conference.

Ms Ulrica Sundholm, political adviser at the Ministry of Health, was the first speaker and presented the strategies in the EU recommendation. She emphasised that the focus is that those who are in need of care and treatment should have that. She mentioned that the National Board

of Health and Welfare are to investigate how the newly established coordinating function will work in Sweden. The focus is from getting the correct diagnosis to how life works for those who are experts on their disease. Work has started to clarify how this will be organised.

The report from the National Board of Health and Welfare earlier this year shows a complex picture and concludes that the patients' position should be strengthened. At the moment the Swedish Association of Local Authorities and Regions (SKL in Swedish) and the Social Ministry are negotiating on some areas cocerning drugs. The prescription of orphan drugs is different in different parts of the country, there is a need to map and identify those differences. Another issue is how solidaric financing is defined and can be instituted. She mentioned the dental care where changes have already been made. There is a proposition that 500 millon SEK will be set aside up until 2012 to cover the costs of general reimbursement in order to protect for high costs for those who need extra dental care.

Ms Désirée Gavhed presented the EUROPLAN project, which will have its final meeting in Rome 2011. She stated the importance of having this conference as it focuses on what is needed for the future. Expected results are that discussions will be stimulated, the development of national plans will be supported and pushed forward and tools will be supported. Several reports (Work Packages-WP) are created with recommendations for the national plans. Today`s conference is planned to contain four workshops. Désirée guided the audience through the seven areas in the recommendation.

In WG 1 plans and strategies for rare diagnoses, definitions, information and education.

In WG 2 research, programmes, clinical projects, guidelines for cooperation, network

In WG 3 Centres of Excellence, network, national centers, telemedicine, cooperation.

In WG 4 patient empowerment, support for organisations, integration in school and work, sustainability

Ms Britta Berglund then presented some experiences of national plans for rare diagnoses in different countries. The Nordic countries Finland, Norway and Iceland have no national alliances.

Next speaker was Ms Elisabeth Wallenius, President. She mentioned that the council recommendation supports the work the Rare Diseases Alliance has been active in during the 12 years of its existence. However, a raised general awareness is needed to get support in politics for the rare diseases. These groups are today not prioritised in care and society. Everybody wants to have secure, correct and good care. Who I am, how old I am, what care I need, where I live in the country should not be important. A national plan gives opportunities for care and development of care and knowledge, since evidence-based decisions for care and treatment are difficult to make. Coordination of resources is cost effective, it saves the doctor´s time. Another benefit is equality in the country, today there is a lack of coordination in many counties. A national plan gives possibilities to guide, coordinate and cooperate and it is also important to be aware of the caregivers who do a great job.

Mr Jan-Inge Henter, pediatrician at the Karolinska University Hospital, talked about the perspectives of health care. As a physician he aims to find the correct diagnosis, to be able to offer good treatment and measure its effect, to have reliable information about the disease, and to understand the biology. As an example of the importance to get the correct diagnosis he used histiocytosis. When guidelines were instituted in 1991, international cooperation was possible. Such cooperation results in more knowledge which in the end gives better care. With more data from many patients we know more than if data are from a few patients, and cooperation is important also to get knowledge about complications, not only about life and death. Development of knowledge is important; not the least in clinical research. For identification of new drugs, cooperation with industry is important. Information is needed for clinically active staff and patients. Through clinical research, syndromes can be identified and drugs and treatments developed. As an example he mentioned the increased survival with less complications for children diagnosed with cancer. He talked about the ‘added value of research’, meaning that not only patients and families with rare diseases will benefit from research on rare diseases, but also patients with other related and non related diseases. One example was cytotoxic therapy of influenza (Henter et al. 2010, in press). He meant that it is important to support clinical research of already marketed drugs for other diagnoses, as it is a cost-effective way to find new treatments for rare diseases. He also mentioned the ICORD that started in Stockholm 2005.

Before lunch, Mr Anders Olausson introduced the chairs for each workshop. The discussions started immediately between the participants, it was obvious that there was a great need to discuss and next paragraphs follow the discussions in the worskhops.

Workshop 1 : Definitions, codification and inventoring

Chair : Mr Anders Fasth, professor pediatric immunology, Gothenburg University

Rapporteur : Ms Maine Forsberg, Rare diseases Sweden

Discussions in the workshop :

During workshop 1 it was discussed if Sweden should change its definition of a rare disease to the European definition. In Sweden a diagnosis is considered rare if it affects less than 1 in 10 000. Although many agreed that it would be more practical to have the same definition as the majority of other European countries, concerns were raised regarding the 'ultra rare' diseases. Participants feared that if Sweden would adopt the European definition, the voice of the persons with ultra rare diseases would become comparably smaller in the rare disease community, as many more people with more common diagnoses would be included. It was also mentioned that it would be beneficial to have a definition based on incidence, as many newborns are affected by RD with a genetic origin and it would be easier to get information on for example survival.

The ICD-10 classification is currently used in Sweden and it was considered ineffective when it comes to rare diseases. It was requested that Sweden should adopt ICD-11 as soon as possible after its release. Participants in the workshop asked for a more flexible classification system where new diagnoses could be added also after ICD-11 has been launched.

As sources of information about RD the databases of the National Board of Health and Welfare, Unique, Genetest and Orphanet are used. The information for professionals is quite good, but for patients more information is needed. The information on diagnoses in the database of the National Board of Health and Welfare was considered very good and informative. However, only about 280 diagnoses are presented in this database. More diagnoses need to be included. Many patients ask for help-lines and more sources of information in Swedish, in particular information on social issues, e.g. how you cope in every day life. Care giving institutions need to improve knowledge about the rare diagnoses through data bases, information material and

project reports. It is important to learn from and listen to the patient associations when producing information as the patients possesses much knowledge and experience.

The Swedish Information Centre for RDs also has a help-line but it is not so well known. Ågrenska has a help-line for grown-ups, which is much appreciated since many patients feel that they don't get enough information once they leave the paediatrics.

More patient registers are needed. However, more financing is needed to establish new registers. Their sustainability is important. One has to consider integrity issues if only a few persons are included in the register due to an ultra rare disease.

The participants of the workshop stated that a Swedish national plan must ensure that patients have the right to the same health care quality, independent of where they live and their diagnosis. Also, it was stressed that the plan should not only be about health care but also about good quality of life, hence many players in the society should be included, such as the Swedish Social Insurance Office (Försäkringskassan) and the Swedish Public Employment Service.

In a national plan the medical centres must have a very visible role. The care plan decided must be carried out in spite of increasing costs for the county councils. The society must be part of this in order to secure its quality and function, as well as ensuring correct support and care to get life functioning. A national function to coordinate this is therfore needed.

Conclusion :

- EUs definition is not accepted and incidence figures are needed, not only prevalence
- Adopt ICD-11 as soon as possible after its release
- The data base of the National Board of Health and Welfare needs to be updated with more diagnoses
- It is important to add information in the plan on how it is to live with a diagnosis
- Several new patient registers are needed for the rare diagnoses

Workshop 2 : Research on rare diagnoses

Chair : Ms Maria Wästfelt, Ministry of Education and Research

Rapporteur : Ms Britta Berglund, Rare diseases Sweden

Discussions in the workshop

A lively discussion started about what research is desireble and how can it be done. In Sweden, research based on themes is not as common as in the rest of Europe. Currently there is no funding dedicated purely to rare diaseses, rather all research projects are put together and supported by a bottom-up procedure. Funding is given to the best application. Big finansiers as FAS and VINNOVA do not focus on rare diagnoses today. Today it is difficult to find information about ongoing research projects in the RD field. However, information on the projects are being collected (title of funded project, type of project and name of researcher) can be found on Orphanets webpage (www.orpha.net). It is difficult to get funding for research on rare diagnoses and also as a researcher to be connected to a research group. The medical profession and other groups can apply for funding from ALF-funds that are linked to county councils and universities. In some county councils projects about rare diagnoses are prioritised from several professions. However, the resources are unevenly distributed in the country.

The item ICF was briefly discussed, it is used mostly in child rehabilitation but rarely in other parts of society. A problem is that codes for rare diagnoses are lacking.

Funding is available to apply for, especially for research in breast and child cancer. One strong statement was that funding for rare diagnoses need to be supported by the government. For this to happen, political pressure is needed. National funding is important if research is to be sustainable and if many people are interested in the results it is easier to find funding. Funding via EU has been possible in the seventh framework programme where Sweden had 7 of 18 projects, mostly clinical trials. The EU-commission supports groups that cooperate (ERA-net). Sweden is currently not a partner of the EU-funded projects E-Rare 1 and 2. The prerequisites of taking part of E-Rare is that the project is a transnational research collaboration on rare diseases and that the project is jointly sponsored by the sponsored by the participating country. The Delegation for clinical research may consider financing of rare disease research, but they would like to evaluate pilot projects on rare diagnoses. Interesting topics are: how is it to live with a disease, what is needed, daycare, school, work, the situation of next-of-kins, loss of employment, quality of life and so on.

International competent experts are an important factor when selecting research projects and a visible structure is needed. A change in attitudes is important for research on rare diagnoses. The medical companies AstraZeneca and Phizer are interested in these diagnoses if a medical need can be shown.

Changes are needed concerning patient registers. They must be sustainable and financed. One example is the prescription registers, where all patients with some need of medication are found. The national centres should apply for funding for registers and work with these, decrease consumption of care and agree about the location of centres. A central EU-register owned by patient organisations is one possible idea.

It´s a long process to reach out to all physicians with information about rare diagnoses. Many patients have problems with multiple functions. It is difficult to get help if you don't have national centres. A good infrastructure in health care where patients meet gives possibilities for research. Swedish centres should be conected to other centres in Europe and specialised physicians should be connected to the centres. Every center must have an independent board that cooperates with patient organisations. Parents are good resources as well. Education of health care personnel and school staff is important as well as showing them where to turn for information.

Questions were raised about the centres. Should they be called competence centres? Who will be the owner of the registers? How can funding be connected for the national coordination? How rare is a diagnosis to be interesting enough for creating drugs?

What centres are active in Sweden today ? Some examples :

- Cystic fibrosis has centres in several hospitals
- Retinoblastoma is taken care of at St Eriks hospital in Stockholm
- Tuberousis sclerosis: 25 patients were cared for by more than 100 physicians
- Retts Centre in Östersund, has been active since 2003 with governmental support, works with research
- Centre for Porphyria, with research and information for patients. It started based on a patient register, funded by patient organisations
- Astrid Lindgrens Hospital, in Stockholm, wants to start a centre for spina bifida
- Ågrenska centre of competence, in Gothenburg has education for families and adults and so on
- Several registers about different issues (but not on RDs) can be found on the SBU website (www.sbu.se).

Conclusions

- Thematic research is needed with funding for rare diagnoses
- Quality registers for rare diagnoses are needed : structure needs to be clarified, as well as long term funding
- There is a need of a fund for research on treatments with concrete suggestions for research
- National medical centres are important for researchers and patients. The centres should have an independent board that cooperates with patient organisations.

Workshop 3 : Centres of Expertise, network, national centres, telemedicine, cooperation

Chair : Ms Birgitta Bergendal, odont.dr, National Oral Disability Centre, Jönköping

Rapporteur : Mr Ulf Larsson, Rare diseases Sweden

Discussions in the workshop

CoE or informal networks in the university hospitals are necessary. Genetic guidance, a responsible physician helping the family, and habilitation are important since the needs of rare disease patients are life long.In dental care, centres of competence have been active in Gothenburg (Mun-H-Center) and Jönköping (National Oral Disability Centre).

The rare diagnoses are different. National coordination is important. We are talking not only about known diseases, but also about diseases that are less visible and those that make their debut in adulthood. The definition of a rare diagnosis must be clarified. Transmission of knowledge to patients must be acknowledged and the knowledge of the patients as well. Financial management is a problem, funding is lacking. The county councils are not very familiar with these diagnoses and have the notion that one specialist is enough, which is inaccurate. National health care is important but nothing happens for the rare diagnoses.

Regional centres are suggested. The least acceptable base for a university hospital is 4 million inhabitants. Habilitation, where many problems are handled, was emphasized. Development of knowledge is important, as well as to establish local contacts, education of local staff is

important. Many diseases need only specialist knowledge. Surgical care is easier to centralise than the type of care needed for rare diseases.

One key-word is registers and we must find out the prevalence and incidence of a rare diagnosis. The national concept is important, the centre of competence can search for the undeveloped and unknown diagnoses. A centre must be organised so it will function even if the person in charge is leaving. Existing centres need to be registered in a catalogue.

Orphan Drugs. Patients particpating in studies wit orphan drugs may improve their health status. But what happens when the study ends? Why should OD be treated differently than other drugs? There are big differences in willingness to pay depending on where in the country you live. Before permission is granted a drug is not allowed to use, even if that is possible in other countries. Sweden should follow the recommendations from EU about off-label use. Different financing of drugs is discussed by The Medical Products Agency and by the Swedish Association of Local Authorities and Regions (SKL). CoE would facilitate for clinical trials in Sweden. Today clinical trials are problematic as the health care is spread in so many places of the country.

Conclusions

- To clarify the diagnoses of rare diseases is important
- Coordination of CeO is needed
- National centres in the university hospitals should take care of registers and follow ups
- Patients need to have a right to go to a CoE
- Training and education of young professionals in order to keep up the work is important
- Funding of OD is necessary

Workshop 4. Patient Empowerment
Chair : Mr Robert Hejdenberg, Ågrenska

Rapporteur : Mr Raoul Dammert, Rare diseases Sweden

Discussions in the workshop

At first, basic prerequisities of rare diagnoses and empowerment were discussed. The definition of empowerment was described. Knowledge is needed to strengthen the individual´s possibilities to be independent, to be able to formulate one´s own goals and to take power over one´s own life. In one family up to 40 different contacts with health care, social security, school, social network and other authorities are needed to get every day life to function. Mr Hejdenberg gave some examples from the work at Ågrenska during its 21 years. Ågrenska arranges family weeks, camp activities and courses devoted for adults with rare diseases.

It is fundamental that everybody is able to master the daily situation. In order to do this, relevant knowledge and exchange of experiences is important for the family, parents, brothers and sisters and the person who has a rare diagnosis. This must be offered by the society with higher priority than today. Education for those who have a diagnosis gives parents decreasing reponsibility when a child becomes an adult.

The national function for coordination was discussed as well and the patient organisations must be involved. Patient organisations are higly valued for creating meeting places for exchange of experiences for both the person with a diagnosis and relatives. Another positive factor is that the organisation is available in different phases in life. Empowerment programs are important to be professionally managed.

Today´s reality is that patient organisations have very limited resources to work with the different tasks. Support is needed from a national centre to ensure sustainability. Socioeconomic studies are needed to describe the saving of resourses if different efforts are made. It is also a great need to get infrastructural support to help organisations to make and update a homepage.

The WG agreed that organisations should be invited to reference groups, to be included in discussions on changes or development (such as research) that concern the organisations target groups. To ínclude organisations for referrals is good, but it is often too late to influence decisions when all basic preparations are already made. The patient organisations should work for investigations to start, get finished and follow up.

The organisations for rare diagnoses have difficulties to get economic resources because they do not fit in the existing systems of funding. An organisation for a rare diagnosis is not the same as a local football club and the society does not always understand their huge responsibilities, for example when giving information to their members, something that in fact the society should do. If persons with rare diagnoses are guided from the start about where to turn for health care and support, they do not have to be lost in the health care system.

Funding is organised differently in different countries. For example in Denmark charity events are arranged, which have provided money to some of the patient organisations in the rare disease community. This is not a common way to support RDs in Sweden.

The Swedish disability movement is divided into many small organisations instead of working together. Nobody will listen to an association with 40 members, it is better to join in a big organisation. One third of members in The Swedish Association of Persons with Neurological Disabilities (NHR) have a rare diagnosis, some of them are also members in the National Alliance of Rare diseases. Many rare diagnoses do not belong to a specific category, i.e. neurology and many of them are therefore members of bigger organisations such as The Swedish National Association for Persons with Intellectual Disability (FUB) and others.

The patient organisations of rare diseases should work and be seen together.It is important to emphasise what is common instead of differences. The best would have been if all rare groups joined the National Alliance of Rare diseases. A suggestion came up that all organisations that have rare groups should be invited to the Alliance.

When getting a rare diagnosis, life is changed and you need somewhere to turn to for aid. It is important that there is someone to call when you have questions about the diagnosis and the consequences of it. The Information Centre for Rare diseases is a kind of helpline, but you can not talk as patient to patient, but is referred to documentation and networks by the centre. Ågrenska has helpline on the homepage and receives many calls from patients and relatives. Many questions concern different perspectives in life and society support, that Ågrenska has knowledge and experiences about. Such information is always included in the programs with development of competence that Ågrenska is working with.

Availability of Orphan Drugs is a problem since the individual counties decide over their own budget. A common solution is asked for so the drugs can be available for those who need them. This can be coordinated or, even better, if the state takes this reponsibility. What drugs should be reimbursed, that is a question. The society can save a lot of money if correct care is given directly.

The national coordinating function

We have enormous expectations on the national coordinating function. It must start from the needs of patients and their relatives with a whole life perspective. The question is whether tasks and resources could be delegated the National Alliance of Rare diseases? The national function must become an operative unit contributing to the development of existing resources in society, but it will not be able to solve everything. Good communication with the county councils is

essential. The patient organisations have a great reponsibility, but how do they get influence? Their representatives should be part of the management of the function. The professionals contribute as a part of their work, while the patients are doing it on unpaid time. It should be possible for patients to contribute with salary.

The organisations often have a good knowledge of where the good physicians and other professionals are located. In Norway, Competence Center for learning has created structures for patient education. During the training sessions the patients or relatives are part of the education as experts with a salary. The Nordic region should be an uptake area for most of the rare diagnoses and patient organisation should be a part of the national function.

Conclusions

- Development of competence is needed during all different phases in life.
- There is a need to create meeting places for exchange of experiences with other groups.
- Patient organisations need support for infra structure (home page and so on).
- Patient organisations should be representated in many different contexts.
- Patient organisations should be included in reference groups and be a referral body.
- Solidarity funding is needed for Orphan Dugs.
- Different helplines are needed. Education is important for those working with the helplines.
- Invitation to other groups for mapping needs and strengthen all.
- National centres are needed for those who have a rare diagnosis and their families.

Concluding remarks

Mr Finn Bengtsson, member of the Swedish Parliament and of the Committee on Health and Welfare, Consultant/professor, Linköping marked the end of the conference. He has submitted several motions on rare diseases to the Parliament. (www.riksdagen.se/webbnav/index).

He said that all kinds of activities are good as it will havean influence on politics. It is important that all political parties work for the rare disease community and that there is communication across party lines. He meant that it is ingenius to collect data bases on how health care is developing. As other countries we get an older population. It is important to find new solutions, not to decrease care. The debate must continue.

Then Ms Wallenius thanked all participants and closed the conference.

The work to achieve a national plan continues and a meeting in the reference group is planned to Jan. 21, 2011. The Rare Diseases in Sweden has as well made an application for funding to make the intentions of the recommendations possible in Sweden.