EUROPLAN National Conferences

FINAL CONFERENCE REPORT

I. General information

Country	**Denmark**
Date & place of the National Conference	**19 November 2010, Rigshospitalet (University Hospital of the Capital Region of Denmark)**
Website	**http://sjaeldnediagnoser.dk/00787/01151/**
Organisers	**Rare Disorders Denmark (RDD)**
Members of the Steering Committee	**Birthe Holm, President, RDD** **Liselotte W.Andersen, Vice President, RDD** **Lene Jensen, CEO, RDD** **Marianne Jespersen, Chief Physician, National Board of Health** **Professor John Østergaard, DMSc** **Hanne Hove, DMSc, Chief Physician** **Henriette Hutters, Head of Centre** **Britta Berglund, Eurordis Advisor**
Names and list of workshops	**All themes were dealt with in the plenary session. The speaker prepared the presentation of the various themes in close co-operation with the Steering Committee and RDD.**
Chairs and rapporteurs of workshops	**Conference rapporteur: Ms Lene Jensen, CEO, RDD**
Attachments (programme, list of participants, etc.)	**Programme, list of participants, minutes of the conference (in Danish)**

II. Main Report

In August and September, invitations to the conference were sent to RDDs member associations, other patient groups, politicians, officials, researchers, social- and health-care professionals and the pharmaceutical industry.

The conference was originally planned for 75 participants, which would ensure real opportunities for dialogue in plenary sessions, as no workshops were planned. Almost 90 applications were received, and all applicants were invited to attend. Attendance was free, except for representatives of the pharmaceutical industry, whose fees helped to fund the conference.

Prior to the conference, individual speakers prepared their presentations in dialogue with the steering committee and RDD, who also co-ordinated the content of the presentations. Participants received a dossier containing most of the presentations, as well as background material, e.g. pertaining to EUROPLAN and the Health Ministers' recommendation.

The conference was to be opened by the Health Minister. Unfortunately, he had to cancel, as a number of health proposals were being discussed in parliament at the time. Instead, Dr Marianne Jespersen of the National Health Board bade everyone welcome and offered a number of observations. The chair of RDD, Birthe Holm, then presented the Health Ministers' recommendation and the EUROPLAN project.

Marianne Jespersen outlined the Danish rare-disorders landscape – from the report in 2001 to the actual professional settings for the work of the Danish healthcare system, including the medical specialisation planning outlined in 2010. Against this background, a number of professional presentations were given on treatment and diagnosis, records and databases, research, information and social conditions, as well as empowerment and the role of patients and their associations.

The Conference concluded with a debate between the presenters and the attendees1. Both the presentations and the debate yielded many valuable contributions that will be included in future work on a national strategy and action plan for rare disorders and disabilities. In her closing address to the conference, Birthe Holm pointed out the following:

- A national plan for rare disorders and disabilities will serve as the basis for intensified efforts on behalf of citizens suffering from rare disorders. Sufferers from rare disorders should have the same rights and access to treatment and social support as everyone else.

[1] In the following report, the suggestions and views presented during the debate is integrated under each theme

- Work on a national plan should transcend sectors. It is not only health conditions that are of major importance – social factors and circumstances relating to the labour market and education also have to be taken into consideration.

- Drawing up a national plan should be an inclusive process. All stakeholders must commit and be given the chance to provide input if the process and the implementation of the national plan are to succeed.

Main Themes

Theme 1 – Methodology and Governance of a National Plan/Strategy (NP)

Denmark does not at present have a national strategy or action plan for rare disorders and disabilities as defined in the EUROPLAN project. However, the National Board of Health points out that a medical specialisation plan exists that grants the Board the power to appoint regional functions of medical specialisation, and to determine the requirements for and approve the placement of medical specialisation functions in public- and private-sector hospitals. The Board has issued 2,500 approvals of medical highly specialised functions in Denmark, and estimates that at least 100 of these are related to various diagnoses or groups of diagnoses that can be classified as rare. However, there is a need to further develop and update the strategy so that patients with rare disorders become an integral part of the health service's planning.

RDD points out that there is a need to draw up an actual national strategy or action plan for rare disorders and disabilities, as defined in the EUROPLAN project. Tangible provisions should be made for diagnosis, treatment, control and support measures. Research into rare disorders should also be promoted. The approach must be interdisciplinary and inter-sectoral – it must incorporate health, social affairs, the labour market, education, etc. It also has to focus on the role and influence of patient associations.

During the conference, it was widely agreed among most participants, that a national plan for rare disorders and disabilities is necessary. Also that the drawing up, implementation and operation of the plan should be an inclusive process that involves all stakeholders in the field of rare disorders.

Theme 2 – Definition, codification and inventorying of RD

Birthe Holm explained that in Denmark there is no clear, accepted definition of rare disorders.

The National Board of Health usually operates on the principle that a disorder or disability is rare if no more than 500 people in Denmark suffer from it (approximately one in 10,000).However, the Centre for Rare Diseases and Disabilities (CSH) and RDD states, that up to 1,000 people in Denmark may be affected by a rare disorder (about two in 10,000). The Danish definition is therefore significantly more restrictive than the European one. In addition, the Danish definition does not build solely at the number of patients, but also at the degree of complexity – the general rule is that it must be a rather severe, genetic disorder. Therefore, rare cancers are not automatically considered to be rare disorders, nor are infectious diseases considered rare in Denmark.

The conference did **not** support introduction of the European definition in Denmark.

Hanne Hove from the Clinic for Rare Disabilities explained that there is no central register for rare diseases and disabilities in Denmark. However, there are several different research and bio-bank registers, e.g. the Danish Cytogenetic Central Register, which records chromosomal abnormalities at national level.

There is also RAREDIS, a joint Nordic database funded by the Nordic Council of Ministers, in which the Danish authorities record rare hereditary diseases that are treated at the Clinic for Rare Disabilities, Copenhagen University Hospital, and the Centre for Rare Diseases, Aarhus University Hospital.

There is no joint registration tool for the rare disorders area, and some rare diseases do not even have a clear ICD-10 code. (ICD, which stands for International Classification of Diseases, is a system developed by the WHO.) This is a problem. Without a unique code, it is not clear cut to identify the exact diagnosis, which makes it difficult to work together at international level.

During the debate it was also pointed out that:

- there is a need for more systematic registration
- ongoing registration of rare diseases must be organised in such a way that the workflow, as far as possible, is integrated into existing administrative systems
- increased registration requires more resources.

Theme 3 – Research into RD

In Denmark, no research resources are specifically earmarked for rare diseases and disabilities. Karen Brøndum of the Kennedy Centre outlined the Danish system as follows:

Universities receive basic funding for independent research. The genetic institutes do perform some research into rare hereditary diseases. The university hospitals also conduct research, but this often competes with the need to devote time to patients – who, for good reasons, are accorded first priority.

The research-council system allocates funding to both independent and strategic research. The number of grants for rare disorders is not large and they have never been a focus theme.

Then there are the sector-research institutions, e.g. the Kennedy Centre, home to the national medical specialisation function for PKU and for Rett Syndrome, but which also conducts general research into disorders that impair mental faculties and a wide range of hereditary eye diseases.

Finally, there is the Danish National Research Foundation, which broadly supports “centres of excellence” in science and biology. There is no centre dedicated to rare disorders.

However, Karen Brøndum argued that, even though there is no official research platform, Danish researchers have a relatively high profile in the field of rare disorders. In the last two years, at least 98 Danish articles dealing with rare disorders were published. Rare cancers accounted for many of them, but neurological and psychiatric conditions were also well represented. Seventeen of them were about rare inherited genetic disorders and congenital deformities.

Denmark has positions of strength, as well as barriers, when it comes to generating more research in this area. It is a barrier that rare disorders are not a defined as a priority and that relatively little research is conducted into them. It is a strength that records, databases and bio-banks are generally good, and that patients have shown great interest in participating in research projects.

Several proposals to promote research into rare disorders and disabilities were put forward, including:

- promoting high-tech co-operation between industry and academia in order to develop new drugs for rare disorders within the existing strategic research-funding frameworks
- defining rare disorders and disabilities as an actual theme in both independent and strategic research
- creating an independent research programme for rare disorders and disabilities
- prioritising Danish participation in E-Rare
- establishing a Danish platform on “Next Generation Sequencing” in order to identify genetic changes and mutations that were previously unidentifiable.

It was also pointed out that a wide-ranging approach to research into rare disorders must be multi-sectoral. In addition to the medical research, there is a need for research into quality of life, living conditions, etc.

Theme 4 – Standards of care for RDs: Centres of Expertise (CoE)/European Reference Networks (ERN)

The structure of the Danish rare-disorders area stems from a 2001 report by the National Board of Health. Among others, RDD participated in drafting the report, which highlighted the need to, e.g. improve and increase hospital efforts to deal with rare disorders and disabilities.

Part of the solution was to establish two centres for rare disorders and disabilities. Their remit included the co-ordination of patient-care programmes, treatment protocols and databases, and taking care of medical highly specialised tasks in agreed partnerships (both internally and transcending the general hospital service). The centres have responsibility for a limited number of diagnoses, while others are rooted elsewhere in the hospital system, particularly in highly specialised medical functions.

The centres, which were established in the eastern (“Clinic” at Copenhagen University Hospital) and the western (“Centre” at Århus University Hospital Skejby) part of Denmark, were charged with a range of

responsibilities: diagnostics, treatment, records, research, international partnerships, counselling and advice, co-ordination, collation and dissemination of knowledge, teaching, updating of guidelines, etc.

Two years after the establishment of the centres, RDD conducted a survey that revealed that 75% of patients felt they had received better and more coherent treatment.

John Østergaard from the Centre for Rare Diseases reviewed the Danish structure and identified a number of challenges:

- There are too few resources available in relation both to the many tasks the centres perform and the rapidly growing number of patients. As the centres cannot compromise on diagnosis and treatment, there is less time not only for advice and guidance for the individual patient, but for collecting and disseminating knowledge, research, development and international cooperation.
- The centres are historically located in children's wards to which many adult patients do not have access. This causes problems both for the patients whose disease does not make its appearance until adulthood and for those young people who must change wards as they grow from childhood into adulthood.
- Most rare diagnoses are not covered by the centres, but belong elsewhere in the hospital system, where the approach is largely organ-specific. This means that many rare-disorder patients are dealt with by multiple specialists and in practice have to act as their own co-ordinators.
- There is insufficient focus on training and education for health workers.
- There is a lack of connection between the social and medical spheres.

In Østergaard's view, the two centres meet eight of the ten criteria that must be met to qualify as a Centre of Expertise. The other approx. 100 highly specialised medical functions in the hospital service that, among other things, deal with rare-disorder patients fulfil far fewer of the criteria. However, there is no comprehensive overview of this.

As far as Orphan Drugs are concerned, the situation is that out of the 64 drugs approved for marketing in the EU, 54 are approved in Denmark. The approval process usually takes one-and-a-half to two months.

On the other hand, the picture is a little mixed in relation to what is known as "compassionate use" of medicine for rare-disorder patients, i.e. circumstances in which a doctor can use a drug for a patient even though it has not been approved for his or her condition.

Several suggestions emerged from the debate:

- It is a priority to resolve the problem of transition between childhood and adulthood, as well as access to the centres for patients with a disease that does not make its appearance until adulthood.
- Experiences from the work in the two centres should be much more widely utilised in future endeavours – both at the centres and throughout the hospital system. One prerequisite for this will be a greater emphasis on education and training
- The two centres should be decoupled from the children's wards and other wards that take care of rare-disorder patients, and need to be upgraded – not just medically, but in an interdisciplinary manner that better incorporates social, pedagogical and psychological aspects.
- A pricing and settlement system that takes into account the specific conditions faced by rare-disorder patients should be introduced into the Danish hospital system in order to ensure a better balance between actual tasks and resources.

Theme 5 – Patient Empowerment and Specialised Services

Torben Grønnebæk of the Danish Wilson Association spoke about the need for empowerment. His main point was the importance of retaining control over your own life, even in the event of a rare disorder. In this context, the patient associations play a crucial role, as they provide knowledge about living with rare disorders and disabilities. RDD conducted a survey in 2005 that showed that 80% of the families thought it was important to meet others in the same situation.

It is therefore crucial that society provides resources for patient associations, even though they are voluntary in nature. A series of grants are allocated both to the associations and to the families participating in their activities, but at present, these are subject to cut backs. This bodes ill for the associations, for families and for society. It will cost the public purse more if families do not attend courses via the associations and develop the skills to cope with life while suffering from a rare condition.

It is precisely because the associations possess knowledge about rare disorders and disabilities that it is imperative that they are involved in policy-making and structural development. Not least, this also applies to the forthcoming national action plan for rare disorders and disabilities.

Torben Grønnebæk also pointed out another challenge for the associations – i.e. that it is mainly resource-rich families who have the energy and vision to be active. It is also a challenge that the vast majority of rare-disorder patients have no association to join, precisely because their disorder affects such a small number of patients.

Henriette Hutters from the Centre for Rare Diseases and Disabilities (CSH) outlined the social aspects. The patients' diagnoses are getting ever-greater importance in order to get the necessary support from the social sector, which is a challenge for many rare patients. Also to the transition from childhood to

adulthood poses a challenge. When adulthood is reached, patients no longer have a single caseworker in the social system, but three or four, each of whom deals with a different aspect of life as an adult with a rare disorder. Education and training also pose a number of challenges.

Problems also exist in relation to the referral of rare-disorder patients to the social, specialised services, because GPs and others in the system simply do not know about these services and/or do not know who is eligible. For example, CSH operates a phone and e-mail helpline for patients and for professionals involved in their care. CSH also has a website with descriptions of rare diseases, written in a way that professionals in local authorities and other institutions can build upon and use in their work with rare-disorder patients. The starting point is interdisciplinary, i.e. based on both the nature of the diagnosis and the social needs.

As of 1 January 2011, CSH will no longer be a separate centre but part of a new overall Knowledge Centre for Disability and Social Psychiatry. In other words, there will be no independent centre for rare diseases and disabilities. Several conference participants strongly regretted this, and pointed out the need for the new structure to ensure that provisions are made for patients and professionals dealing with rare disorders. It was also pointed out that there is a need to do something for the many rare-disorder patients who do not have relevant patient associations and who have hitherto made use of CSH.

The following proposals were among those that emerged from the discussion about empowerment and social support:

- The patient associations deserve far greater support than they currently receive.
- The patient associations should be involved directly in the determination of regional medical specialisation planning, since it is in the process, decisions are made about where in the hospital system the different diagnoses should be anchored.
- Local authorities should provide far greater financial support to families, so that they can participate in the associations' courses and other relevant activities.
- There is a strong need to adopt a holistic approach to each individual citizen. The transitions in the social system, especially between child- and adulthood, must be much more appropriate than at present.
- Social profiles, which will act as a dialogue tool between rare-disorder patients and social caseworkers and other professionals, should be drawn up for all rare diagnoses.

Conclusion of the Final Report

After the conference, an evaluation form was distributed to the participants. The following were the most important aspects of the feedback:

- 95% were satisfied with the advance information about the conference. However, several pointed out that they knew of others (especially health care professionals) who would have liked to participate but did not receive an invitation.
- There was widespread satisfaction with the various presentations, with 80–90% describing them as good or very good.
- There was some criticism that there was too little time for the final debate, during which it was possible to make position statements. It was pointed out, that apart from the two centres; other medical highly specialised groups who take care of rare-disorder patients ought to have had time to speak.
- A number of suggestions were made for other topics that should be included in future work, including:
    - patient training for the families of rare-disorder patients
    - distinguishing between assessment and diagnosis
    - the Danish regions' role, so that bodies other than the National Board of Health are involved
    - telemedicine – both in the contact between specialists and in the contact between specialist and patient
    - bridge-building between scientific research, medical care, social work, psychological, educational and therapeutic practice.

Overall, the conference was evaluated extremely positively by the participants and generated high expectations for the future development of the process.

The EUROPLAN recommendations played an important role at the conference in relation to illustrating and defining what a national plan of action actually is.

Both the EUROPLAN recommendations and indicators are assed to be most useful when RDD is lobbying politicians and civil servants to promote the process of developing a national action plan.

Whether the indicators can be used in relation to evaluate the process and its progress depends on how the process unfolds. At present, this is still uncertain.

Some of the recommendations may be applicable in a Danish context, but others are not relevant. In addition, some of the recommendations – most notably in relation to a European definition of rare disorders based on the current European definition – did not attract support.

III. Document history

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