



# DENMARK

## EUROPLAN NATIONAL CONFERENCE

### FINAL REPORT

23 January 2015, Copenhagen area

# 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>	Denmark
<b>Date &amp; place of the National Conference</b>	January 23., Copenhagen Area (IBOS, Hellerup) 2015
<b>Website</b>	Venue: <a href="http://www.ibos.dk">www.ibos.dk</a> Website for Conference: <a href="http://sjaeldnediagnoser.dk/europlan-konference-om-den-nationale-strategi-for-sjaeldne-sygdomme/">http://sjaeldnediagnoser.dk/europlan-konference-om-den-nationale-strategi-for-sjaeldne-sygdomme/</a>
<b>Organisers</b>	Rare Diseases Denmark (RDD)
<b>Members of the Steering Committee</b>	<p>Rare Diseases Denmark:</p> <ul style="list-style-type: none"> <li>- Birthe Holm, President</li> <li>- Sven Fandrup, Member of RDD Executive Committee</li> </ul> <p>The Danish Health and Medicines Authority (DHMA)</p> <ul style="list-style-type: none"> <li>- Marianne Jespersen, Senior Medical Officer</li> </ul> <p>The National Board of Social Services (NBSS)</p> <ul style="list-style-type: none"> <li>- Vibeke Lubanski, Academic Consultant</li> </ul> <p>Centres of Expertise for Rare Diseases:</p> <ul style="list-style-type: none"> <li>- Allan Meldgaard Lund, Consultant Paediatrician, MD, DMSc</li> <li>- Hanne Hove, MD, DMSc</li> <li>- John Østergaard, Professor, M.D., DMSci</li> <li>- Stense Farholt, Consultant Paediatrician, MD, Ph.d.</li> </ul>
<b>Names and list of Workshops</b>	<p><b>Workshop 1: The way forward for Danish rare policy / national strategy</b></p> <p><b>Workshop 2: Information and education on rare diseases</b></p> <p><b>Workshop 3: Research in rare diseases and handicaps</b></p> <p><b>Workshop 4: Diagnostics, treatment and more - Centres of Expertise</b></p> <p><b>Workshop 5: Medicine and other treatment of rare diseases</b></p>

	<b>Workshop 6: Social services for people living with rare Diseases</b>
<b>Workshop Chairs (and Rapporteurs, where applicable)</b>	<p><b>WS1: Chair:</b> Acting Head of Office Kirsten Brøndum, The NBSS. <b>Rapporteur:</b> Mette Grentoft, Rare Diseases Denmark/Danish Society for Williams syndrome</p> <p><b>WS2: Chair and rapporteur:</b> Managing Director Lene Jensen, Rare Diseases Denmark</p> <p><b>WS3: Chair:</b> Professor Karen Brøndum Nielsen. <b>Rapporteur:</b> Søren Lildal, Danish Apert Patient Society</p> <p><b>WS4: Chair:</b> MD, DMSc Henning Bundgaard. <b>Rapporteur:</b> Liselotte Wesley Andersen, Rare Diseases Denmark/ Danish Patient Society of Tuberous Sclerosis</p> <p><b>WS5: Chair:</b> President Birthe Byskov Holm, Rare Diseases Denmark. <b>Rapporteur:</b> Professor Karen Brøndum</p> <p><b>WS6: Chair:</b> Managing Director Lene Jensen, Rare Diseases Denmark. <b>Rapporteur:</b> Søren Lildal, Danish Apert Society</p>
<b>Annexes</b>	<ol style="list-style-type: none"> <li><b>1. Programme</b></li> <li><b>2. List of Participants</b></li> </ol>

## II. MAIN REPORT

### GLOSSARY

CE RD	Centres of Expertise for Rare Diseases. In Denmark, two centres for multiple rare diseases exists, CE RD-RH at Rigshospitalet (Copenhagen University Hospital) and CE RD-AUH at Aarhus University Hospital
DHMA	The Danish Health and Medicines Agency
KRIS	The Coordination Committee of Managed Introduction of Hospital Medicine
LGD	Local Government Denmark – organisation of all municipalities in Denmark
”Lægedage”	A specific GP seminar for continuing professional development etc.
NBSS	The National Board of Social Services
NCS	The National Coordination Structure, hosted at the NBSS
NGS	Next Generation Sequencing
OMP	Orphan Medicinal Product
PLWRD	People Living With Rare Diseases – patients and relatives
Raredis	The Nordic database for rare diseases
Specialty plan	DHMA plan of treatment of specific diseases by appointing national and regional functions to hospitals (not only rare diagnosis, but all diagnosis requiring highly specialised treatment)
VISO	The National Organisation for Knowledge and Specialist Consultancy, hosted at the NBSS

#### Plenary Report – Opening Session

Moderator: President Birthe Byskov Holm, Rare Diseases Denmark

#### Opening speech

The conference was opened by **Bent Hansen**, Chairman of Danish Regions. Bent Hansen's point of departure was that despite being very different, rare diseases share common characteristics. Their complexity makes specialised services across sectors necessary. At the same time, there is only little knowledge about rare diseases - more knowledge and better information needs to be produced. Communication, cooperation and coordination across sectors are very important in the effort for rare patients as well as relatives – People Living With Rare Diseases, PLWRD.

Bent Hansen stressed that the **Regional Health Agreements** between the municipalities and the regions are an important tool, in order to ensure correlation and quality in the courses of PLWRD.

In future, Danish Regions will put spotlight on "The Health Service of the Citizens" - a proposal will be presented in April 2015. The goal is to bring the requests, needs and experiences of the citizens into play. This is also important for PLWRD, where it is about creating a balance between the highly specialised services and services to be handled at other levels. The limited knowledge needs to be unified and developed. The Centers of Expertise for Rare Diseases (RD CE) are located at the level of highly specialised functions, which they should be - as should many of the rare patients. At the same time there has to be an inter-disciplinary focus on maximizing synergy and joint results.

Bent Hansen finished his welcome speech by emphasising that international cooperation is important in regards to rare diseases.

### European policy and guidelines

EURORDIS-EUROPLAN-Advisor **Lene Jensen**, Rare Diseases Denmark, presented the European framework for rare diseases. The point of departure is that because there are few patients and even fewer specialists, rare diseases and handicaps are an evident area of cooperation in Europe. Lene Jensen briefly presented a number of key documents:

- Regulation (EC) no 141/2000 of the European Parliament and of the Council of 16 December 1999 on Orphan Medicinal Products
- Commission Communication on rare diseases: Europe's challenge of 11 November, 2008
- Council Recommendation on an action in the field of rare diseases of 8 June 2009
- Setting-up of EUROPLAN I and II, 2008-2011, 2012-15
- Setting-up of EU Committee of Experts on Rare Diseases 2010-2013, and Commission Expert Group on Rare Diseases 2014 -
  - Recommendations
  - State-of-the-art publications on rare diseases
- Development of core indicators meant for monitoring and assessing the implementation of national plans for rare diseases in the EU
- Directive of the European Parliament and of the Council of 9 March 2011 on patients' rights in cross-border healthcare
- Joint Actions - a new one is on its way!

In the presentation, all documents were provided with links to the original sources. Lene Jensen underlined a mutual European effort to make things better for PLWRD - most important is the Recommendation to make national plans or strategies in all member countries. EUROPLAN's 22 core indicators were briefly presented and finally a slide of the map of Europe was shown to illustrate in how many countries there are now strategies or plans for rare diseases.

**Laurs Nørlund** (LN), the representative of the European Commission in Denmark, subsequently addressed the conference and underlined the importance of European collaboration on rare diseases and handicaps. It is about collecting experience and expertise as well as spreading knowledge and to have broad cooperation across borders. This makes even better sense, when patients as well as the expertise are rare. This was the main reason why the Commission and the Council of Ministers in 2008/2009 found it natural to create a European framework for this work. Two thirds of the member countries have presented and passed plans or strategies.

LN pointed out that rare diseases have received a special place in the Directive on cross-border healthcare, because especially during the treatment of rare patients it is of great usefulness that patients and expertise can meet across borders. He expressed a hope for an efficient implementation of the Directive, also in Denmark.

### Opening speech - II

**Else Smith** (ES), Director General/CEO of The Danish Health and Medicines Authority (DHMA), talked about the effort for PLWRD. ES stated that Denmark traditionally has asserted itself strongly in the field of rare diseases. The DHMA is proud to have produced a National Strategy, where all interested parties of the rare disease field have contributed. The strategy is a good basis for future work and cooperation. A special effort has to be made, if PLWRD are to meet the same quality, services and efforts as other patients.

ES promised that The DHMA will make a point of attending to its role and its tasks in the field in the next few years, i.e. concerning the Danish Specialty Plan for healthcare and more. The National Strategy is not a plan of action, but it contains points of orientation, focus areas and recommendations, which can be transformed into specific initiatives.

**Knud Aarup** (KAA), CEO of The National Board of Social Services (NBSS), also addressed the situation of PLWRD. They need rehabilitating efforts cut across several sectors. The big challenge in the social area is the lack of knowledge about the actual effect of interventions. This can be learned from the memorandum recently issued by The NBSS on rare handicaps. The NBSS is working on creating better knowledge in the social area, also inspired by the area of health care.

PLWRD are often in contact with a great number of specialists. It is a highly prioritised focus area of The NBSS to strengthen the holistic and the collaborative approach, also across sectors. Focal points regarding the social effort are knowledge, coordination, strain on the entire family and the families' needs to meet others in the same situation. Two new mechanisms in The NBSS are about the needs of PLWRD:

- New social supervision with focus on the quality of social services
- The National Coordination Structure (NCS) for specialised social services that ensures the existence of the right services and knowledge environments for small target groups

with complex needs.

KAA also accentuated the importance of international cooperation.

The NBSS and the municipalities are going to cooperate with the patient organisations to try out some relevant empowerment initiatives. The patient organisations must still attend to their tasks and the volunteers are of an immeasurable value. However, new methods have to be found. Finally KAA expressed his great expectations for future cooperation with all interested parties.

### Country Status - Denmark

Senior Medical Officer Marianne Jespersen from The DHMA and Randi Lykou Head of Office at The NBSS presented the contents of the Danish National strategy:

By way of introduction, **Marianne Jespersen** (MJ) explained the Danish definition of rare diseases, which is maintained in the national strategy: Rare diseases are rare, most often congenital, hereditary complex and serious diseases and conditions that demand special knowledge and expertise. Prevalence is approx. 1-2 out of 10.000 or less, which is to say approx. max. 500-1.000 persons in Denmark. There has to be a need for a particularly well-planned effort consisting of highly specialised diagnostics, healthcare, follow-up and check-ups, which profitably can be united in 1 -2 places in the healthcare system. The criteria for the positioning of the highly specialised functions in the Danish Specialty Plan are rare occurrence, complexity and resource consumption. There is no need for an absolute definition of "rare diseases" in Denmark, as there are no specific rights or similar attached to them.

Subsequently, MJ went through the recommendations of the national strategy in the health care area. There are a number of challenges for the healthcare system, but there are also certain new diagnostic and treatment possibilities:

- Referral, diagnostics and coordination: In the strategy, there is a focus on strengthening earlier and timely diagnostics, i.e. by ensuring the possibility for further referral directly from a specialty to other specialties in specifically planned coordinated diagnosing procedures, also including further referral of patients to more highly specialised functions and to other relevant areas of specialisation. There is focus on strengthening coordination and cooperation between highly specialised functions in the diagnostic process. And CE RD have a particular obligation to give counselling, guidance and to coordinate.
- Multidisciplinary teamwork: There is a need for well-organised multidisciplinary diagnosis and treatment based on existing highly specialised functions. Based on the CE RD, work is done to develop models and agreements on multidisciplinary teamwork for children as well as adults. There is a specific need to strengthen the effort for grown-up patients with rare and complex diseases with multi-organ involvement.
- Children and adults together: As complex rare diseases do not comply with age, organ,

or expertise limits; it is recommended that the grown-up patients are also treated at the CE RD or at the same hospital units as the child patients. If so the patient is ensured continuity and correlation over time and the practitioners will get more experience, knowledge and routine. A desirable transition from childhood to adulthood is guaranteed by taking into account at an early stage which specialty should attend to responsibility of the procedure and this specialty is involved in the treatment of the patient in good time before transition.

- Coordination and treatment responsibility: A team should be established. Coordination responsibility lies with a doctor from the CE RD or from the specialty, which is in charge of diagnosing and treating the individual's dominating problem. Professionals with other specific specialties are also a part of the multidisciplinary teamwork of the special functions e.g. nurses, dieticians, occupational therapists, physiotherapists, psychologists, dentists and social workers.
- Not mentioned, but not forgotten: Patients with a rare or suspected rare disease, congenital or genetically determined, who are not placed elsewhere in The Danish Specialty Plan, can be referred to one of the two CE RD. The Centers can ensure that patients are assessed through multidisciplinary teamwork at the Centre on hospital grounds or by preadmission assessment proceedings to one of the specialty specific highly specialised functions at the hospital.
- Treatment: Tentative experimental treatment should continue to be possible in relevant cases. Attention should be paid to the possibility of referral to research based treatment across borders, when there is a relevant possibility for this. When assessing the effect of treatment modalities, a breadth of scientific methods and approaches should be included.
- Genetics: Genetic examinations should generally require a relevant clinical report/ diagnosing of the patient. Guidelines should be contemplated for, in which situations it is relevant to offer exhaustive genetic examinations to a patient. It has to be evaluated, if there is a need for pooling and task allocation of genetic expertise functions, including pooling of examination and guidance for selected rare diseases at fewer genetic departments. It is further recommended that patients are given the possibility to be genetically reassessed after a suitable time span, when relevant.
- Medicine: (Continued) access to necessary Orphan Medicinal Products (OMP) in Denmark needs to be ensured.

There is a need for knowledge accumulation, also in the health care sector:

- Knowledge accumulation and counselling: As far as possible, recommendations on patient procedures, possibly based on international guidelines, best practices, should be used. General Practice and the main and regional function levels of the hospitals need to have an easy access to valid and updated information regarding rare diseases. Qualified counselling and competent feedback from the CE RD and the multidisciplinary teams should also be available.

- The Learned Societies are encouraged to be more attentive of the specific challenges that the rare diseases entail for specialist training, further training, quality development of treatment and more, research etc. The Societies are further recommended to increase focus on timely diagnosis and treatment, rehabilitation, research and development on rare diseases within the field of the individual specialty. Furthermore, the Societies are recommended to relate to the specific challenges that rare complex diseases, not belonging to any specialty, entail.

With regard to organising the rehabilitating effort, there are a number of challenges across the healthcare system and social services:

- There should be focus on coherent continuity of diagnosis, treatment and care and social services, rehabilitation and more. Often, it will be a case of lengthy, possibly lifelong parallel procedures.
- Persons of authority, who need to elucidate and make a decision regarding services for PLWRD, should, as far as possible, be obliged to find relevant knowledge and use this in the case management.
- Knowledge on successful interdisciplinary and coordinated efforts between relevant stakeholders should be collected, including the possibility to establish new divisions of tasks and interdisciplinary cooperation etc. (e.g. the Spiel Meyer-Vogt teamwork).
- Regional Health Agreements are an instrument for ensuring cooperation between regions and municipalities. The Regional Health Agreement should cover the problems that PLWRD meet in order to receive coherent continuity of treatment, care and rehabilitation and more.

Regarding documentation, registration, research etc. the recommendations of the strategy are as follows:

- Documentation: The CE RD are obliged to register rare disease patients in the joint Raredis database and thereby ensure improvement and maintenance of knowledge. The database needs a consolidation. In general, a basis for registrations of relevant rare groups of patients in registers and clinical databases should be ensured. Furthermore, an overview of the databases and registers etc. available should be established, with a view to improve knowledge on occurrence as well as quality parameters.
- Registration and disease classification: Systematic registration should be given high priority, in preparation for quality assessment, research and development. An as accurate and uniform diagnosis classification as possible should be used. The so-called McKusick/OMIM numbers should still be used. Concurrently, the development of Orpha codes should be complied with.
- Greater focus on research: National and international research in the health care environment should be given higher priority, e.g. via more PhDs. The Learned Societies, relevant research committees and foundations are informed about the national

strategy and the perspectives in the international interest in research on rare diseases. Research should entail a holistic approach to the patient - from basic molecular disease mechanisms to rehabilitation and social efforts.

- Education: There should be more focus on rare diseases in the training at medical school and other health education programmes. There should be continuing education for General Practitioners regarding rare diseases e.g. in relation to “Lægedage” - a specific GP seminar for continuing professional training development etc. Development of interdisciplinary and possibly inter-sectorial training and education should be contemplated.

Furthermore, there is a focus on international cooperation as well as implementation and evaluation in the strategy:

- Denmark should, like so far, be attentive, participate and seek influence on the work under EU auspices.
- Implementation and evaluation: The strategy is to be implemented before 2018 and evaluation will be undertaken after 3-5 years (starting year: 2014).

Finally, MJ pointed out that there are a total of **97 recommendations in the strategy**, addressed to: health authorities, regions, municipalities, research foundations, Learned Societies, professional organisations, general practitioners, other professionals, hospital administrators, social authorities, the education sector and researchers, GP’s and others as well as to the patient organisations.

Subsequently, **Randi Lykou** (RL) went through the recommendations in the field of social services. The point of departure is that PLWRD are very different individuals. The municipalities cannot know something about all diagnoses. Therefore it is particularly important that the municipalities have access to information and draw on specialised expertise. The rehabilitating effort must be inter-disciplinary, holistic and with a perspective of lifelong support when necessary. The recommendations of the strategy include:

- A focus on the access to the social service initiatives and good cooperation with authorities/ municipalities and professionals.
- That continuity and stability in the cooperation between families with rare diseases and the social services of the municipality is ensured. Preferably, with one coordinating social worker per family, who can be the facilitator in relation to the different administrations and departments in the municipality as well as potentially the contact with the healthcare-system?

There is a need for user-friendly information about the many rare diagnoses for PLWRD and relevant professionals. It is of great importance in order to be able to prepare for the progression and prognosis of the disease, and for the planning of efforts. Among the recommendations of the strategy are:

- A breeding ground for inter-disciplinary knowledge environment should be created in order to ensure relevant knowledge about the rare diagnoses and on the reduction in functional capacity, which is typical for the individual diseases and handicaps. It has to be possible for the social workers and the municipalities to draw on professional social service counselling and knowledge, when they are planning rehabilitating efforts for PLWRD.
- The Rare Diagnosis Information Database is to be quality assured, passed on to a highly qualified, professional environment and improved in the next few years. It is recommended that the Database is transferred from The NBSS to a platform in a strong, sustainable and robust health professional environment with the possibility for input on social services.
- The NBSS acts as a source of knowledge for the municipalities, given that:
  - VISO - The National Organisation for Knowledge and Specialist Consultancy - gives guidance to citizens and professionals and can assist the municipalities.
  - The task of ensuring access to up-to-date social service knowledge belongs to the Office of communication on handicaps, rare handicaps and special needs education.
  - The National Coordination Structure (NCS) has to follow the development in target groups, services and efforts. NCS also has to ensure an amount of services necessary for target groups with the most specialised needs, develop and communicate knowledge as well as strengthen cooperation across regions and municipalities. This is done by monitoring, central announcements and instructive professional procedure recommendations. NCS is in constant dialogue with municipalities, regions, patient organisations and other interest groups.
- Cross-border cooperation is prioritised - including Nordic and European cooperation, amongst others regarding Rarelink, Orphanet and EURORDIS.

RL emphasised the need for an empowerment focus, patient education and patient organisations: Meeting others in a similar situation and/or with similar experience can strengthen management of one's own disease. Sometimes management of own disease as well as the acknowledgement of it is paramount for the effect of the rehabilitating efforts. PLWRD are often the experts on their diagnosis. Among the recommendations of the strategy are:

- A particular focus on empowerment;
- Patient education should be an element in rehabilitation, also for patients with rare diseases. Efforts should be made so that there is possibility for more specific patient education for PLWRD;
- PLWRD should have the possibility to join relevant networks and to participate in these activities. The Rare Network for PLWRD should continue to be a service for those, who are not able to participate in other relevant networks/associations due to the rarity of

the disease;

- Patient organisations are to be involved in experience aggregation, participation etc.;
- Inter-disciplinary specific guidance and counselling should be developed for PLWRD.

Concerning clinical databases and research there is a need for more knowledge about research in relation to social efforts; and how the municipalities can support the best possible rehabilitation for children, youths and adults with rare handicaps. Recommendations of the strategy includes a focus on:

- Which methods promote that more PLWRD benefit from education and support in childhood and youth years.
- What kinds of habilitation, support and help have the best results as to increase self-determination and quality of life for PLWRD, and increase the possibilities to live life autonomously.
- The role of relatives, networks and other environments in relation to helping and supporting children, youths and adults with rare diagnoses.

Everybody, who works in the field of social services, should have a general knowledge of how a rehabilitating effort can be organised in the best possible way, when it comes to a citizen with complex needs. Once again, it points in the direction of the need for easily accessible and understandable knowledge. Among the recommendations of the strategy are:

- That particular attention is paid to further develop information on specialised social service for PLWRD and reductions of functional capacity.
- A focus on upgrading qualifications of employees who work with PLWRD in the fields of social service, education and employment.
- That professional knowledge environments are developed that can ensure this upgrading of qualifications.

### **Workshops - playground of solutions**

As a build-up to the next step of the conference, **Birthe Byskov Holm** presented the purpose and aim of six workshops. The purpose of these workshops is to develop concrete ideas and suggestions in order to make a reality of the strategy (turn the strategy into reality) and better support the effort for PLWRD. Workshop Chairs have to bring two-three suggestions back to the afternoon plenary, as all discussions and suggestions will be included in the written report of the Conference.

## Report of Workshops

### Theme 1 - Methodology, Governance and Monitoring of the National Plan

#### Sub-Themes:

- 1.1 Mapping policies and resources
- 1.2 Development of a National Plan /Strategy
- 1.3 Structure of a National Plan /Strategy
- 1.4 Governance of a National Plan
- 1.5 Dissemination and communication on the National Plan
- 1.6 Monitoring and evaluation of the National Plan
- 1.7 Sustainability of the National Plan
- 2.1 Definition of RD

#### Workshop 1: The way forward for Danish rare policies / the national strategy.

Relevant chapters in the national strategy:

- Chapter 3 - definition and occurrence
- Chapter 5 - recommendations on salience in the health care specialty plan and awareness in the Learned Societies
- Chapter 15 - implementation and evaluation

First, the **Chair Kirsten Brøndum** (KB) from The NBSS presented the recommendations of the Danish strategy in relation to implementation and evaluation:

- The recommendations are to be implemented and integrated in the day-to-day work with rare diseases;
- The recommendations of the strategy should be implemented before 2018;
- Evaluation three-five years after the drawing up ;
- The evaluation should result in a short progress report.

Furthermore, the Chair presented the Danish understanding of rare disease, as it was explained in the plenary, and pointed out that the definition is narrower than the European one (approx. 1-2 out of 10.000 in Denmark compared to “no more than 5 per 10 000 persons” in the EU definition) . However, the definition was not up for great discussion, as there is a general support of it in Denmark.

CEO **Leif Vestergaard** (LV), The Danish Cancer Society, gave a presentation on concrete experiences of translating strategies into action:

Initially it is important to acknowledge that the drawn up recommendations need more stakeholders. Few people alone cannot translate the recommendations into action. All stakeholders must work together.

LV pointed out that The DHMA (Danish Health and Medicine Authority) and The NBSS each have their professional traditions and way of thinking. In both government agencies there are committed professionals, who would like to make progress and each of them has to maintain their professional competence, but they also need to reach out to each other. The Health and Medicines Authority's way of working is very much based on evidence and knowledge, whereas The NBSS traditionally work is more experience-based. It is difficult to coordinate efforts between social and health systems, but when it succeeds, it will be of benefit to many other groups of patients, including those with rare cancer diagnoses.

It is a good thing to search for knowledge, but it is worth remembering that it is difficult to procure evidence in this field. The best evidence is at the CE RD. Also, many municipal employees hold important knowledge and experience. LV pointed out that a knowledge network already exists, but it has to be more systematic, as it can quickly become complicated due to the many stakeholders, who are in play; the municipality, the region, large hospitals, small hospitals, the doctor, the pedagogue etc.

LV thought that some of the recommendations of the national strategy are non-specific - they need to be unfolded and specified. Here, the stakeholders need to cooperate and develop a plan for implementation. A task group should be formed to assess the process and report every second year, with the participation of The NBSS, The Health and Medicines Authority, Local Government Denmark, Danish Regions and Rare Diseases Denmark.

Furthermore, Rare Diseases Denmark, the CE RD and VISO should jointly cooperate on solving the knowledge challenges.

LV saw a big challenge in putting PLWRD on the political agenda in the municipalities and the regions. An inquiry should be initiated in order to discover if the concrete services work well for the service users. Based on this, the five regional health coordination committees have to consider what changes should be made. Each municipality should contemplate how to manage in areas of weak management that administer the municipal services, without weakening professional competence. It is important to realise that there are systems within the social service and healthcare system, which are lagging behind and need to be challenged.

LV also brought the vulnerable patients into focus in relation to languid parties (parties, who are not prepared to follow national/regional recommendations) proposing that the CE RD should be able to make a complaint when failure occurs in the municipal and regional social services and healthcare system. It is not appropriate, that the citizen him-or herself has the responsibility to take action. LV suggested that The NBSS should have a specific responsibility to go through the complaints to The Council of Appeal in the field of PLWRD, in order to learn more. Likewise, The DHMA should be obliged to examine complaints in the field of PLWRD for The National Agency for Patients' Rights and Complaints, so everybody knows where the

problems of the systems are.

Afterwards, Senior Medical Officer **Marianne Jespersen** (MJ), The DHMA, presented the contents of the strategy for rare diseases regarding implementation, monitoring and evaluation with specific focus on the field of healthcare. The national strategy for rare diseases is to be implemented before 2018 and there is to be made an evaluation after 3-5 years from when it is issued - this is to be adjusted to the cadence of The Danish Specialty Plan and The NBSS' work with The National Coordination Structure (NCS). Many of the recommendations cannot be finalised, as it is more a case of continuous work. The Strategy belongs to every one of us and therefore, we should all take on the recommendations and take them to heart.

Within The Danish Specialty Plan, medical highly specialised functions have been established in connection with the revision of The Specialty Plan.

MJ presented the tools for the work that has been done with regards to The Specialty Plan: Monitoring, progress reports, various routine statistics, statements from clinical databases and patient satisfaction surveys, where some of it may be specifically targeted to some of the rare patients. In this context, Rare Diseases Denmark has been instrumental at collecting data and delivering material that has made The DHMA more knowledgeable in that field.

Furthermore, MJ accounted for the exciting challenge The DHMA stands before, with the establishment of European Reference Network (ERN), where CE RD cooperate internationally. How the European work is going to be organised is still under discussion.

Finally, MJ pointed out that in order to put recommendations of the strategy into action, the strong stakeholders need to contribute: The Danish Cancer Society, Rare Diseases Denmark, Learned Societies, research committees and the research foundations etc. Small committees within this framework will have to take a specific look at PLWRD, and then it may be the spearhead for something that can benefit many. The DHMA will do its part of the job, within the framework of the government agency and based on what is promised.

Subsequently, Head of Office **Randi Lykou** (RL) from The NBSS presented the contents of the strategy regarding implementation, monitoring and evaluation with specific focus on the field of social services. Implementation in 2018 is a short deadline, but The NBSS is ready to work hard and has already made an effort via NCS.

RL told about the NCS, which has to:

- Follow developments in target groups, services and efforts.
- Ensure a sufficient choice of services for target groups with the most specialised needs.
- Organise and communicate knowledge.
- Strengthen cooperation across municipalities and regions.

How does NCS do this?

- Monitoring - construction of database with target groups.
- Central communications: report target groups/efforts.
- Instructive professional procedure recommendations.
- Continuous dialogue with municipalities, regions, patient organisations and other service user- and interest groups.

The NBSS/NCS cannot make professional procedure recommendations for all 800 rare diagnoses. Procedure recommendations for disease groups of a certain capacity with special needs for highly specialised services will be made where VISO has difficulty managing specific issues. RL told that The NBSS gives the municipalities' social service knowledge on how to handle PLWRD via a new knowledge memorandum, via VISO and via empowerment programmes.

RL also told that **The Rare Diagnosis Information Database** has the goal to give access to easily comprehensible information to PLWRD and professionals about many rare diagnoses. It is of great importance, both in relation to inform PLWRD of the expected progression/prognosis and for those planning the service. Therefore, there has to be a quality control of The Rare Diagnosis Information Database, and the Database has to be continued and improved in future years and preferably in cross-border cooperation.

In closing, RL pointed out that The NBSS seeks a dialogue with Local Government Denmark with a view to implement the recommendations from the national strategy. The Council of Appeal has at present begun an initiative of investigations into the practice, in form of spot checks, of case management of the municipalities, seen in relation to cases of complaint in the field of social services.

The last introductory speaker was the President of Rare Diseases Denmark, **Birthe Byskov Holm** (BBH), who related what is done in other countries. BBH stated that Denmark is one of the last countries in the EU to launch a national strategy for rare diseases and therefore is one of the last to have a EUROPLAN conference. The idea of EUROPLAN is to learn from each other. We should draw on best practice experiences from other countries, but work with EUROPLAN has not come that far yet.

BBH pointed out that the Danish strategy is a professional strategy. In some countries, it is a political strategy. This can be of significance in relation to economic resource prioritisation. These are examples of how other countries have set about the work with national strategy or plan:

- In Ireland the strategy was presented by the Minister for Health.
- In Finland, a task group has been formed in the wake of the national strategy, with the task of making specific action plans for the individual areas to ensure progress.

- In France, concrete economic priorities have been made.
- In Belgium, the strategy has received an autonomous budget.
- In Germany, “seed money” has been earmarked for the different objectives.

Finally, BBH made a concrete proposal, which originates in the concern of Rare Diseases Denmark of how the national strategy is to be put into action. A lot has to be coordinated and many knots have to be tied in order to link reality to the paperwork. Therefore a national forum for rare diseases and handicaps should be formed/a mechanism to gather the threads. Such a forum would also be able to contribute to the yearly work of The DHMA of collecting information for the "State of the art of rare disease activities in Europe", EUROPLAN indicators for the strategy and its results etc.

Afterwards, the **Chair Kirsten Brøndum** opened the floor and several views and ideas were introduced:

- In many places, very few passionate people run the health professional effort. It works surprisingly well, but there is a lack of management organisation and management involvement in order to make it a success.
- A medical professional pointed out the need to have the right diagnosis codes in order to register the patients in the right way. In other countries a sum of money comes with the patients for this, which is not always the case in Denmark.
- From a patient point of view there is a huge distance between what was said in the morning opening address and the real world. Patient organisations put a lot of voluntary effort into the work for patients with rare diseases. The patients have to be involved in the implementation of the national strategy in order to qualify it.
- When you have a rare disease it is often the service user himself/herself, who has to bring his/her own case around the whole system. The experience of this is that it is a concrete problem to find out where to hand in the case.
- In Sweden, there is a competence center for rare diseases and handicaps, Agrenska, where courses are offered alongside counselling and information. The center also accommodates The Swedish National Function, which is responsible for cooperating, coordinating and disseminating information about rare diseases and handicaps. In Denmark, there is no equivalent, but there is a need for it. The Swedish model should be exported to Denmark. In Sweden they also have good experience with participation of rare disease patients at public meetings, where patients meet politicians and make them aware of the issues.
- There was support from several participants and different stakeholders to establish a national forum. On the patients' part there was a concrete suggestion to transform the recommendations from the national strategy into a checklist and use it in the work of implementation. On the part of the municipalities, there was also support in favour of the idea of a national forum and it was pointed out that all professionals should be involved. Finally, the private association Orphan Council was suggested as a platform.

- On the part of the patients more knowledge collection and communication was looked for, especially in connection with the municipalities.
- Also on the part of the patients, it was suggested that economic resources are injected into the field of rare diseases and that a “rare package” with diagnostics and well-coordinated procedures across the social service and healthcare systems, inspired by the cancer area, is established.
- A participant representing a region pointed out that recommendations have different status in the social service and healthcare systems, respectively. In the health sector, recommendations are understood as a downright work tool, whereas in the social sector they do not have the same definite status.

**LV** underlined the importance of converting the recommendations of the national strategy into concrete actions. LV suggested the initiation of a national rare forum. Some national initiatives have to be put in place. If things are not working out, money is wasted.

**BBH** summed up that several general examples had arisen in the workshop as well as some suggestions for the continued work with the national strategy for rare diseases. Rare Diseases Denmark has many ideas inspired by its Board of representatives and other work. Many things have already been achieved in the rare field, but a lot more needs to be done. Everybody has to take on responsibility to move forward; and it will not happen by itself. All stakeholders have to work together.

## Theme 2 - Definition, codification and inventorying of RD

### Sub-Themes:

- 2.4 Information on available care for RDs in general, for different audiences
- 2.7. Training healthcare professionals

### Workshop 2: Information and education about rare diseases

Relevant chapters in the national strategy:

- Chapter 10 – information.
- Chapter 13 - education and competency development.
- Section 4.6 - general practice and rare diseases.

As introduction, the **Chair Lene Jensen** from Rare Diseases Denmark presented the sections in EUROPLAN guidelines and the EU Recommendation regarding the topics of the workshop. The Danish Strategy's recommendations in the field were also presented:

Information and knowledge:

- The Rare Diagnosis Information Database: should be quality assured, passed on from the NBSS to a sustainable, highly competent body and improved in future years -

rooted in a strong, sustainable and robust health environment with possibility for social professional input.

- General practice and hospitals: easy access to valid and updated information and possibility for counselling from the CE RD and other relevant environments.

#### Education:

- Particular attention on rare diseases in health education.
- Development of interdisciplinary and potential intersectorial training and education should be contemplated.
- Courses for general practice, e.g. at “Lægedage”.
- Increased attention towards the specific rare challenges, e.g. in relation to the specialist medical education, further training, quality development etc.

**Senior Research Fellow at KORA** Leif Olsen (LO) gave the first presentation. The presentation concerned the different knowledge arenas of rare diseases and handicaps. The point of departure is that people have to find a way in which to get the most out of life with rare and often complex diseases, diagnoses, disabilities and handicaps. There is a demand for structural and procedural answers to the challenges in relation to e.g. knowledge and coordination in practice across professions and sectors.

The present way of thinking is that a coordination effort needs to occur on behalf of the family affected by rare diseases, as a great number of professionals are interacting with the family. This effort is insufficient at present and the patients often stand alone. LO pointed out that it would be appropriate to change the way of thinking as to see the family as a party in the coordination effort, as it is the patient/the relatives, who have to put their problems into words. Their resources, knowledge and competencies have to be brought into play in relation to the solution. At the same time, unnecessary strain on the family needs to be actively prevented. Concurrently, clear and common goals need to be established.

It is hardly realistic with just one coordination model for everybody as the needs of the individual family may change over time. There are different ways to better communication and sharing of knowledge. When preparing further efforts in the field, it is important to meet the needs in the short run and to develop efforts that can influence both structures and processes in the long run.

Afterwards, **Holger Bang-Møller** (HBM), father of a child with Albrights Hereditary Osteodystrophy with Pseudohypoparathyroidism, presented the family's daily life with focus on being in pursuit of knowledge. The diagnosis is characterised by a tremendous plurality of serious problems and the family has been in contact with a very great number of authorities and professionals. In one year, they have been in contact with 38 authorities - and some of these authorities cover a larger number of persons. The family acts as knowledge source for

quite a few of the authorities and the family obtains knowledge through the internet and a few medical contacts. A lack of knowledge has among other things led to malpractice, life threatening situations and tremendous discomfort for the patient.

HBM pointed out the need for a unified database, where social workers, doctors as well as parents can find information about the diagnosis and living with it. The diversity of each diagnosis should be reflected. The database should be a tool with which to improve the quality of the effort of the medical as well as the social services. Concurrently, patients and relatives need to be able to find information themselves, to make it easier to handle everyday life.

**Lars G. Johansen** (LGJ), Chairman of The Danish College of General Practitioners, then came with his suggestion for what has to be done in order to make General Practitioners more aware of rare diseases. The point of departure is that the GP is a professional for the same patient over a long period of time and that the GP sees the rare disease patients for other problems than those related to the rare diagnosis. They are experts in the individual patient, not in the individual rare disease.

LGJ pointed out that the General Practitioners' role has very little significance in the strategy and that this is to be criticised. He pointed out a number of necessary initiatives:

- Further training.
- Accurate and updated information - e.g. via an inclusion of The Rare Diagnosis Information Database on the Danish e-Health Portal sundhed.dk. This is where the GPs already look for information.
- One place, with one phone number, where they can ask medical questions and get information about how to act regarding social service matters.

In addition to this, there should systematically be allocated more money for research and knowledge as well as experience collection.

Subsequently, focus was levelled at the question of whom in the social service and healthcare sector, should be trained in handling the tasks regarding services for rare disease patients. Head of Department, **Lykke Jensen** (LyJ) from The Metropolitan University College stated that nobody should be trained in 800 rare diseases, but everybody should be trained in handling the needs that 50.000 Danish PLWRD have. The important thing is not the diagnosis, but the person. They have normal needs that demand a special effort.

It necessitates training and further training. A battering ram is needed to neutralize the differences between social services and healthcare. Interdisciplinary needs to be taught as well as what it means that the service user is central. The evidence line of thinking needs to be challenged: All parties need to have access to specialised knowledge - but value creation only takes place, when knowledge is transformed into action via cooperation and the courage to

introduce a radical inclusion. Finally, focus must be put on the interaction between disease management of the professions and PLWRD management of professional knowledge.

The last introductory speaker was Consultant Paediatrician, MD, PhD **Stense Farholt** (SF), CE RD-AUH. The point of departure is that the highly specialized functions in the health care system meet many rare disease patients - the general level does not. Therefore, interaction is necessary.

CE RD must be updated professionally in the niche area that one has and should inform, cooperate, educate, communicate and collect new knowledge - and see the patients. The knowledge of CE RD comes from the patients, from research, from international contacts and literature, knowledge databases, registers etc. CE RD are not experts in 800 different rare diseases and cannot educate others to become so either. However, one can be trained in the manner in which to think and work, in order to have a holistic approach to PLWRD. Rarity demands knowledge sharing, cooperation and professional network.

SF pointed out a number of areas, where action should be taken with an intensified effort:

- Increase awareness via training and further training (the medicine studies, the specialist educations, other courses).
- The Rare Diagnosis Information Database and guidelines in Danish, preferably entrenched on The Danish e-Health Portal sundhed.dk - there must be easy access to knowledge, which is updated and there must be a unified entry and access to it.
- Research and registration.

Afterwards, the Chair opened the floor to the participants of the workshop to put forward their points of view and to come up with suggestions for the future effort:

- Several speakers from different stakeholder groups backed the idea of attaching greater priority to The Rare Diagnosis Information Database and an entrenchment at [www.sundhed.dk](http://www.sundhed.dk). It was informed that The NBSS at present is in a process of public procurement regarding the phasing out of The Rare Diagnosis Information Database for entrenchment elsewhere.
- On the part of the patients as well as the health professionals it was furthermore pointed out that quite a lot of knowledge, evidence and research actually exists, which makes it even more important to get on with the information effort; but we do not know everything - and there is also a need for new knowledge.
- From the medical side it was pointed out that geneticists have the necessary competences to be the case managers and that they are willing to act. It was also pointed out that the interdisciplinary approach must not prevent specialisation. Furthermore, it is important to remember that some PLWRD also have cognitive difficulties.

- From the patient side concrete steps were called for regarding how the social services and the healthcare system can become better at working together. It was underlined that the municipal social worker often does not trust the knowledge they are presented with.

**LyJ** said that at The Metropolitan University College silos have been broken down, for example by offering further training in classes, consisting of professionals from the healthcare sector as well as from social services. There is currently also an ongoing process of digitalising the education and making knowledge sharing simpler.

**LO** agreed that there is enough knowledge in order to act now. In the field of cancer there has been created a unified access to knowledge resources.

- From the patients point of view there was backing to the idea that it is very important to put effort into training professionals. The experts must learn to collaborate and work together interdisciplinary, with respect for each other's knowledge and domains.
- The patient organisations ought to be recognised as an important knowledge resource in relation to living with a rare disease. Rare Diseases Denmark can be "umbrella for PLWRD" - an access point to knowledge. The **Chair** pointed out the need for an editorial or steering group with patient representation regarding The Rare Diagnosis Information Database.
- It was stated that the Rehabilitation Center for Muscular Dystrophy conducts a research on how the patients' stories can be told in order for them to feel recognized. There is a great need for this in order to make professionals understand the realities of having to handle rare diseases.
- Furthermore, information was given on how The NBSS is working on a 4-year project about empowerment for families with a handicapped child. It is among other things the ambition to test if empowerment courses etc. can be made in cooperation with patient organisations and municipalities.

**LyJ** was an advocate for the fact that the training of professionals is not good enough - among other things, there is a lack of focus on empowerment and rehabilitation.

**HBM** thought that patient organisations are important – and we should not forget all those patients, whose disease is so rare that they do not have an organisation at their disposal. They also need to meet others and to take part in a course; this should be facilitated by The Rare Network for PLWRD.

### Theme 3 - Research on RD

#### Sub-Themes:

- 2.2 Codification of RD and traceability in national health system
- 2.6 Training healthcare professionals to recognise and code RD
- 2.3 Registries and databases
- 3.1 Mapping of existing research resources, infrastructures and programmes for RDs
- 3.2 Dedicated RD research programmes and governance of RD research funds
- 3.3 Sustainability of research programmes on RD
- 3.4 Needs and priorities for research in the field of RDs
- 3.5 Fostering interest and participation of national laboratories and researchers, patients and patient organisations in RD research projects
- 3.6 RD research infrastructures and registries
- 3.7 EU and international collaboration on research on RD

#### Workshop 3: Research in rare diseases and handicaps

Relevant chapters in the national strategy:

- Chapter 12 - databases, registers, codification, research
- Section 3.2.1 - occurrence in proportion to the planning of the healthcare systems effort

As an introduction the **Chair Professor Karen Brøndum Nielsen** presented the sections in the EUROPLAN Guidelines and the EU Recommendation that relate to the topic of the workshop. The Danish strategy's recommendations in the field were also presented:

Focus on research:

- More focus on research in rare diseases, nationally and internationally, i.e. via more PhDs.
- Make the Learned Societies and relevant research committees aware of the national strategy and the intentions, i.e. for the purpose of encouraging Danish and international research in the field.
- Research with a holistic approach to the patient, natural history, health service research and biology is needed.

Registers, databases:

- Upgrading and consolidation of the Raredis database.
- Ensure basis for registrations of rare patient groups in registers and clinical databases, also for the purpose of quality supervision.
- Better overview of existing databases, bio banks and registers.

Classification, diagnosis codes:

- Use of accurate and uniform diagnosis classification, e.g. OMIM numbers.

**Professor Ebba Nexø** (EN) made the first presentation, talking about challenges for research in the rare disease field. EN had chosen to extend the topic to include *possibilities*. The national strategy creates very good framework conditions. It is important that the strategy is put into action.

Rare disease research is a small part of the total healthcare research, which is framed by three parties: The patient, the researcher and society as a whole. There are several challenges to set about: How to get researchers interested in rare diseases and related problems? How to get significant results with few patients and how to get the pharmaceutical industry interested in making medicinal products? However, the possibilities of the field are many, e.g. solid patient organisations, where patients as well as networks have the will and the ability to take part in research projects, also as partners.

In the rare disease field, there are only few research environments and few ways, in which young researchers can see themselves in a top position. Even so, there are also many possibilities: You can conduct research and fully understand correlations and conduct research in a variety of topics from the entirely basic to the entirely practice-oriented. There is also a unique possibility to gain solid patient involvement in the research and there are many international perspectives. If this potential is to be realised, talented researchers must be recruited and retained - passionate people, who can keep focus. A possibility is to recruit an international first class researchers who can lift a field and attract the young, who have to do the future research. A few PhDs is not enough - young researchers have to be recruited and retained in lengthy processes.

EN pointed out that society as a whole is more aware on the importance of research - with regard to the national economy as well as the pharmaceutical industry, but also because there is a wish for a healthcare system based on research. Society wants good framework conditions to make it possible to initiate research. It can be seen in the organisation of the hospitals - e.g. in the Danish Specialty Plan, where it is an aim to collect the specialised treatments.

Infrastructures like databases and bio banks are important. In the rare disease field, the bio banks are of enormous importance. With them, research related hypotheses would quickly be possible to test, without having to wait for decades of patients' increase. Nevertheless, we cannot carry out research in all rare diseases in Denmark - focus is necessary and there must be mechanisms for attracting and implementing international knowledge.

The field of rare diseases can often be used as a model on how to handle other diseases. Small groups make it "easier" to try out models of connections between research and treatment.

EN pointed out that this cannot be done without resources. People working in the field must be aware that they have to find funds on equal terms with everybody else. We have to compete on quality and we have to prove that research does not only solve a concrete problem for a small group, but that it also addresses more general problems. EN underlined once again that there should be an open mind towards the international possibilities.

**Professor Troels Staehelin Jensen (TSJ)**, who presented the practice of The Danish Council of Independent Research, gave the next presentation. He pointed out that he represents the council of "Health and Disease", but the problems of PLWRD can easily belong in several of the other research councils.

TSJ gave a factual exposition of The Danish Council for Independent Research: its division into different fields of research and the proportions of the means that have been applied for and granted, respectively. Approx. 20 % of the applications are met and the criteria for a grant is strictly quality. The topics range from the basic to the clinical and to the field of social services. No particular considerations are shown for any of the disease groups. There are many different types of means, which are offered. Most interesting for the rare field is probably "Project 1" and "Project 2" (limit up to 1.8 m DKK and 4.5 m DKK, respectively). Additionally, there is a program for shared positions, where you can research "part time", concurrent with taking care of your clinical work.

Finally, TSJ mentioned that the research committees are aware of the field of rare diseases. It can however, not be prioritised as it does not lie within the mandate of the research committees.

The Chair asked, if The Danish Council for Strategic Research cannot support chosen fields? TSJ answered, that this is no longer the case as this duty is now placed in The Danish National Innovation Foundation. The Chair pointed out that there is "Era-net" in the EU, which i.e. has a focus area called "Rare Disease Research" (E-Rare). The member countries have to contribute in order for the researchers of the country to be able to apply for means, but Denmark does not do this.

Consultant pediatrician, MD, DMSc **Allan Meldgaard Lund (AML)**, CE RD-RH, who presented Danish participation in international research, gave the next presentation. By way of introduction, AML pointed out that the many barriers related to few patients, lacking diagnoses, lack of homogeneity in the patient group etc. can be overcome by working internationally. This can be in the form of EU financed Consortia or cooperation between European universities - with or without the pharmaceutical industry or it can be European

networks, where knowledge databases are established. There are some good examples of Denmark's participation in all this. According to EUCERD's "State of the Art" publication, there were 30 EU research projects with Danish participation in 2013. The starting point is good: good patient registers, The Danish Civil Registration System, bio banks, good collaboration with patient organisations, industry etc.

AML pointed out that more has to be done. Firstly, we should map out research at national level and where we especially excel. Afterwards, we should make a strategy, because we cannot embrace everything. The good databases and registers have to be improved and made visible internationally. The cooperation between the university environments and the pharmaceutical industry must be strengthened. Denmark is a good country for clinical trials, but we lack more and better trials facilities. There is especially an insufficiency in the field of paediatrics.

Furthermore, AML pointed out that it is important to develop partnerships with patient organisations nationally as well as internationally - in order to recruit participants in clinical trials, but also in order to get input for research projects, including selection of end-points for e.g. medicine testing.

In relation to the EU, European cooperation in the field of research is mentioned in the national strategy, but the effort should be strengthened concretely. AML called for ways in which to handle the comprehensive administrative effort that is especially related to EU applications. A lot of the European research that will be conducted in future years will originate from the European Reference Networks (ERN) that are under establishment, to which Centers can apply to become a part of. Therefore, the Danish CE RDs must have resources to be able to enter into the ERNs. There is also a need for more earmarked public means for rare disease research.

Afterwards, the word was passed on to Consultant paediatrician, MD, **Hanne Hove** (HH), CE RD-RH, who talked about identifying the patients who can take part in research. It should be easy, but when one is supervised in the system, you are registered under the ICD10 codes. The rare diseases do very often not have any specific ICD10 code. Later, when the patients need to be identified, many will inevitably "be lost". Another challenge is that with new knowledge, new diagnoses will arise all the time.

We therefore need a diagnosis system, which is updated all the time. It exists, but is not in use. When we are approached by international researchers, we very quickly have to be able to find rare disease patients based on combination of specific diagnosis, age, gender etc. Therefore, we need databases, where registration is very specific. We also need databases in order to make follow-up on quality and best practice, which again has to lead to the best possible treatment.

HH pointed out that the rare disease field is characterised by the fact that many passionate professionals have made registers for their patients in certain fields of disease. However, these are "island systems", which live independent lives and where the data quality perhaps is not always optimal. An overview should be created over the databases in the rare disease field. Perhaps, they can be put under Raredis, the Nordic database for rare diseases, and in the process data can be quality assured. The goal must be that they are collected in national databases for rare disease patients, where we can register the procedures.

There should be an obligation for all CE RD to strengthen research, to carry out continuous registration and ensure quality development of the treatments for PLWRD.

Vice President of Rare Diseases Denmark **Liselotte Wesley Andersen** (LWA) gave a presentation on the research political agenda of PLWRD:

The patients want an increase in focus on research in rare diseases in order to get the treatments of tomorrow developed. An increase in research makes it possible to take part in more clinical trials, which again leads to the fact that the patients can get medical treatment earlier than usual. It will create more knowledge and will also contribute to the understanding of more frequently occurring diseases. Tuberos Sclerosis (TS) is a good example. It is cancer related and the two genes which cause TS have been identified. Suddenly this has brought about a lot of attention from cancer research. It tells that there are good examples of knowledge passing from the rare to the more widespread disease areas.

LWA suggested allocation of research funds in order to subsidy research specifically in rare diseases. This can perhaps be through The Danish National Innovation Foundation. Databases and registers have to be strengthened because they are a prerequisite for Danish participation in international research projects, which are very necessary due to the rarity.

Patients also want knowledge transfer and continuity to be ensured - not just via PhD positions, but also by international researchers. We are very dependent on few passionate people. Patients also want courses to be established for students concerning rare diseases - e.g. in medicine, psychology, nursing - at relevant times in the course of their training.

The patient organisations should be involved in research at an early point in time, because the patients are experts in their own disease and in what actually is possible, when clinical trials are set up. Therefore, it is important to strengthen cooperation between the patient organisations and the research institutions.

Finally, LWA pointed out that the patients also want a holistic approach to research, where the patient is put in the center of things and where interdisciplinary and intersectorial topics and staff are involved.

Afterwards, the **Chair** opened the floor to the participants of the workshop. Comments were made and ideas stated:

- Astonishment was voiced that genetics were not more highly represented in the presentations. Almost all rare diseases are genetically determined. If we had a bigger effort within genetics, we would be able to identify the patients earlier and thereby organise a much better preventive effort or course of treatment. For a relatively small expense, all Danes could be sequenced, but it would require an ethical discussion. The **Chair** replied that genetics are highly prioritised in Denmark. Propositions have been made to allocate 500 m DKK to genome sequencing of chosen patient groups - it is in hearing presently.
- From medical side it was pointed out that sequencing has limited value, if it is not compared to what is actually wrong with the patients. It is only when these data are compared with sequence data that new knowledge arises.
- From medical side a discussion was also raised regarding the terms on which to make databases. A database for Genodermatoses for example is under development, but there are no resources for diagnosing and sequencing all relevant patients. It would be clever to have a national pool, from where means could quickly be released for molecular diagnosing. It was pointed out that some registers are degenerating and if laboratory findings cannot be correlated with reality, much will be lost. Conversely, there are also a lot of Danish databases and bio banks etc. that are very valuable. They can e.g. be used to identify the people, who are bearers of a genetic disease, but not necessarily have it in outbreak. In the US it is a very large problem that they do NOT have these registers.
- Finally, it was pointed out from a medical point of view that there is an enormous international network in the EU and other places, but Danish researchers can often not participate, because we do not contribute as a nation. There should be ways in which to find the means e.g. in The Danish National Innovation Foundation.
- A representative from the pharmaceutical industry thought that we are able to do so much in Denmark that we need a common focus. Where do we excel nationally and where do we want to present ourselves internationally and brand ourselves?
- Different stakeholders pointed out that there is a lack in career directions for those, who enter the field of rare diseases. The national strategy is all right, but it is lacking the resources.

**TSJ** pointed out that there is a need for the regions to allocate means for research at the CE RD and other highly specialised departments.

The **Chair** told that there is an international consortium for "International Rare Diseases Research" (IRDiRC). The goal of the consortium is to develop 200 new medicinal products for rare diseases in few years. In order to become a member you have to allocate 10 MEUR over a 5-year period. If you are a member, there are a number of possibilities. Should we try to

promote the project, so Denmark can become a member and get access to the means?

The **Chair** proceeded by accounting for the task group that is currently formed by the circle of Deans at the universities about systematic whole genome studies. The proposal requires 500 m DKK. The question is, which patient groups ought to be whole genome sequenced? It can result in a great amount of knowledge about the diseases, if we systematically complete whole genome sequencing. It must be remembered, however, that it demands a great deal of knowledge to interpret these data. "Human knockouts" exist: The genes say that one ought to be ill, but one is not. Therefore, it can be difficult to interpret a given gene deviation. However, Denmark has an obvious possibility in connecting our unique databases to genome data. This has been done successfully in Finland. Here, they have sequenced 3-4.000 exomes, and have compared them to similar European data. As a result i.e. certain genetic variants in the Finns have been unfiltered, which protect against cardiovascular diseases.

Finally, the **Chair** summed up the discussion by pointing out that several presentations focused on involving the patients in research - regarding evaluation as well as relevant end points. Various presentations underlined that registers and bio banks are important. An overview has to be created in Denmark and small registers must be unified, e.g. in Raredis. The important role of genetics was also touched upon. It was also pointed out that genome knowledge has to be interconnected with health information in order to make sense. The necessity of international cooperation was a general mantra, and there was a big request for a stronger national will to contribute to European and international forums.

#### **Theme 4 – Care for RDs - Centres of Expertise and European Reference Networks for Rare Diseases**

##### **Sub-Themes:**

- 4.1 Designation and evaluation of CE
- 4.2 Scope and functioning of CEs
- 4.3 Multidisciplinary, healthcare pathways & continuity of care
- 4.4 Access to information
- 4.5 Research in CEs – How to integrate research on RDs and provision of care
- 4.6 Good practice guidelines
- 4.7 Diagnostic and genetic testing
- 4.8 Screening policies
- 4.9 European and international collaboration – Cross-border healthcare and ERNs (European Reference Networks)
- 4.10 Sustainability of CEs

#### Workshop 4: Diagnostics, treatment and a lot more - Centres of Expertise

Chair and rapporteur: MD, DMSc Henning Bundgaard - Liselotte Wesley Andersen, Rare Diseases Denmark.

Relevant chapters in the national strategy:

- Chapter 4 - organisation of the health professional effort
- Chapter 5 - challenges for the health service
- Section 14.1.2 + 14.1.3 - Centers of Expertise, European Reference Networks

By way of introduction, the **Chair Henning Bundgaard** specified the terms of Centers of Expertise (CE) and European Reference Networks (ERN):

CE:

- National expert centers for one or more rare disease(s)
- A number of quality criteria have been outlined by EUCERD for CE, among them
  - Perform diagnostics, treatment and research.
  - High clinical and research quality.
  - Multidisciplinary and coordinated approach.
  - Sufficient capacity.
  - Committing to collaborate and sharing information.
  - Holistic approach in relation to the individual ("integrating medical and social aspects").
  - Participation in international research and collaboration.
  - Information and communication for healthcare personnel.
  - Education activities for healthcare personnel.
  - Collaboration with patient organisations.

ERN:

- Network between national CEs and other healthcare providers, creating a flexible framework for healthcare pathways for the patients.

Afterwards, the Chair presented the recommendations from the national strategy in this field:

- About referral, diagnosing and coordination:
  - Strengthen earlier and timely diagnostics.
  - Ensure possibility for further referral directly from a specialty in concrete planned coordinated diagnosing procedures.
  - Both vertical and horizontal referral to other relevant areas of specialisation.
- About multidisciplinary treatment, follow-up:
  - A well organised multidisciplinary diagnostic process and treatment.
  - Specific attention on and strengthening of the effort for adult patients and focus

on the transitions in the systems, e.g. childhood -> adulthood.

- Based on CE RD, work should be continued to develop models and agreements on multidisciplinary teamwork for adults as well as children - and that children and adults, when relevant, can be treated in the same place, e.g. at CE RD.
- Connection to a well-defined medical team responsible for the course and coordination of diagnosing and treatment course.
- If not mentioned elsewhere in the area of the Danish Specialty Plan: reference to CE RD can take place.

The first speaker was Regional Chief Operations Officer **Svend G. Hartling** (SGH) from The Capital Region of Denmark. The presentation was about the rare diseases in the health care system and challenges related to this.

SGH pointed out the problems with getting a timely diagnosis and subsequent treatment. Among several, one cause for this is that the doctors often do not contemplate the possibility of rarity, but take as their point of departure that it is a standard disease with unusual symptoms. There is a need for a change in culture, where the doctors refer to other specialists and defy the medical pride they might have.

Next, SGH touched upon the problem with the increasing number of patients at the CE RD and the lack of resources that can be the result of such an increase. Another challenge for the Centres is to become prepared to be able to monitor more adult patients. Here, especially adults with multi organ problems are mentioned, while the specialties in the field of adults are divided into specific organs. According to SGH, a solution could be to focus on coordination persons/case managers, who can function across age groups and organ specialties. The Centres are aware of the problem and try to find solutions. The collaboration between CE RD-RH and CE RD-AUH is fruitful, but there is still a lack of an interconnection from the Centers to the municipalities and the social area.

Afterwards, Senior Medical Officer **Marianne Jespersen** (MJ) from The DHMA presented an overview of the Danish model for the specialised health care planning. In 1993, it was the first time that The DHMA enforced a Specialty Plan of treatment of specific diseases, by appointing national and regional functions to hospitals. The Specialty Plan was last revised in 2008-2010 and in 2016 the ongoing revision has to be finished. In the present Specialty Plan approx. 100 rare diseases are specifically mentioned. When a rare diagnosis is not mentioned specifically it does not mean that it has been forgotten.

MJ stated that The Specialty Plan is thought of as a generic model for all the different diseases. The Specialty Plan is very important, but it is impossible to secure all knowledge and competences via The Specialty Plan. For rare complex diseases not belonging to any specialty, it is necessary to build a teamwork between the organ-based specialties. It can perhaps be

done by changing the structure of the specialties itself or the way people collaborate, but the experiences of the past show that it is not done by establishing specific hospitals or isolated units for the individual rare diagnoses.

Finally, MJ pointed out the importance of planning the continuity of care for the patient with a long-term perspective.

Afterwards, Consultant Paediatrician, MD, PhD **Stense Farholt** (SF) CE RD-AUH presented the challenges in getting CE RD to function according to the criteria. Rare diseases do not respect age or organ-based specialties, and this call for a holistic approach disregarding age and with a specific eye for coordination and correlation. Rare patients take up relatively little space by way of number and are easily squashed between larger groups of patients, e.g. the cancer and the heart disease areas.

SF talked about the CE RD-AUH model. Here, the aim is to get a combined specific and "brick less" centre, where there is collaboration with specialists across fields. Such collaboration is very necessary in order to secure the effort for the adult rare disease patients. Collaboration between social service experts and psychologists is also being attempted. The continuity of care for the patient is closely linked with the specialty, where the patient has the biggest needs for treatment - this can be at the Centre or in a specialty function. It is important that the departments speak the same language. This can perhaps be ensured by employing doctors part time at the Centre and part time in the relevant specialty function. In this way, a higher degree of knowledge sharing and correlation is ensured.

SF pointed at challenges of inter-disciplinarity internally at the Center and "out of the house" and correlation between the social service and the healthcare sector. In order to make the collaboration a success, there needs to be a paradigm shift in i.e. medical training, by creating a focus on training in knowledge of cooperation across specialties and subjects, and awareness of rare diseases.

SF closed the presentation by pointing out the necessity of prioritising the rare diseases higher with more resources and to put forward a specific proposition of developing a "rare package".

The last introductory speaker was President of EURORDIS and The Danish Hemophilia Society **Terkel Andersen** (TA), who spoke about the necessity of a holistic approach. TA commenced by telling that in the past 30 years a positive development regarding diagnostics and treatment in the field of rare diseases has taken place, although there is still room for improvement in relation to the organisation regarding the patients and their families. The healthcare, social service and education sectors have to become better at cooperating and incorporating the effort in relation to the PLWRD.

A patient/family may meet 50 different professionals from different sectors in one year and that places great demands on inter-sectorial cooperation and coordination. TA gave an example of how such cooperation can proceed, based on hemophilia, which has had a CE for more than 30 years. There is a demand for resources and at present the effort is not as good as it could be.

TA pointed out that future interaction/cooperation between the social services and the healthcare sector could profit from including patient organisations. In addition, one could be inspired by how continuity of care is handled elsewhere around Europe.

The **Chair** illustrated a very long course of diagnosis with a case: An 8-year-old boy is diagnosed with cystic fibrosis; his lung function is 40 %. Why has it taken so long to establish a diagnosis? Who is keeping an eye on the general and the overall course? The Chair called for specific recommendations on how to secure faster diagnosis and subsequently opened up the floor for the participants of the workshop to put forward their points of view and to come up with suggestions for future efforts.

- From a patient point of view it was expressed that medical pride might be a reason for trouble concerning diagnostics and collaboration between specialists. Inter-disciplinary teams have to be established and new technology and screening methods must be used. There should also be a diagnosing guarantee for the rare disease patients.
- A doctor suggested that one should learn from the experience of the establishment of The American National Institute of Health (NIH). From a medical point of view the GPs were pointed out as important players in relation to having a suspicion that a rare condition might be at play. A suggestion was made, to make a board consisting of different experts/specialists in order to establish a diagnosis, so the patient does not have to go from doctor to doctor.

**The Chair** pointed out that there should be a far more liberal referral policy: If unexpected and unknown symptoms occur, the patients should be referred to other professionals.

**SF** pointed out the need for a cultural change in the education of medicinal, social services and psychology and more. Professionals have to learn to think "rare disease" and cooperate across sectors. It should be contemplated if referral to other specialists could be relevant, if necessary to a center function, and to have a more liberal and smooth referral pathway. It should be avoided to call patients to an examination, in order to send them home again without taking care of their problems. There has to be an increase in collaboration between the hospitals.

**SGH** agreed and stated that the doctors are getting better and better at cooperating across specialties and fighting the Tarzan syndrome. If a diagnosis board is established as suggested, it will also require a food chain of professionals.

**The Chair** asked SGH if he, as decision-maker at the regional level, could imagine announcing the need for a more liberal system of referral and facilitating this way of thinking and make the paths more resilient. SGH answered that this had already been the case, however not with a rare disease.

- From a patient point of view, it was pointed out that some rare diseases are so difficult to diagnose that it demands that you end up with a doctor, who has seen a patient with the same diagnosis earlier on. It is important to gather the expertise. Another patient representative told that there is good coordination at CE RD, but there are only few patients in her diagnosis group, who is assigned to the CE RD. The rest of the group is treated in local hospitals, where there is poor knowledge and a lack of coordination.

**The Chair** told about the new possibilities for diagnosing with neonatal screening programs. A lot is written about genetics in the national strategy, as 80 % of the rare diseases are genetic. The Chair asked MJ if she can picture that we in 15 years make much larger programs for neonatal screening with The DHMA in the lead as a road ahead for early diagnosis. **MJ** answered that with new tools like genetic testing and neonatal screening programs this can become relevant in the longer run, but MJ does not believe that the problems will be solved with technological tools.

**The Chair** pointed out that in relation to treatment we do not have to start from scratch. Many things are well functioning at the highly specialised departments and there must be some knowledge that we can learn from - but it has to be converted. Can we make a generic protocol, as it was done for the 2001 report? Could the model from CE RD-AUH perhaps be used? Should we use the Centers? Who is responsible for this and who will take action?

**The Chair** also pointed out that on the other hand we should also learn from the rare diseases and not place them under a cheese bell. There are also other more frequent diseases, which have multi organ problems and with these, we could reciprocally exchange experiences. We have to learn from each other.

- From a patient point of view, it was underlined that the problems are not new - the patients have been there all the time; but the treatment of them stalls, and therefore, there is very much a need for a case manager function to coordinate the treatments for the patients. Particularly a case manager, who can incorporate the family and who is willing to bring in the experts.
- From a medical point of view, it was stated that the training of doctors is changing and here it could perhaps be a possibility to emphasise rare diseases. Each specialty should identify some rare disease patients. More awareness is needed in order for the rare disease patients to be treated at the same level as other patients.

**The Chair** asked MJ, if the revision of the Specialty Plan will bring more attention to the rare

diagnosis groups? MJ stated that she, at the present time being in the revision process of The Specialty Plan, could not say anything specific. However, the highly specialised functions, which are described in The Specialty Plan, are also very much about the rare diseases, regardless of whether they are specifically mentioned or not. MJ ascertained that one should also remember that The Specialty Plan can do quite a few things, but it cannot solve all problems.

**The Chair** called for a direct specification from The DHMA to have every specialty develop a structure for diagnosing and handling of the patient with e.g. the more difficult or more serious, not well-described diverging course of a disease. One could imagine that each specialty selects one or more departments, which specifically take on a "rare function". **SGH** pointed out that not everything can be solved with The Specialty Plan. It is more a question of a need for cultural change; each individual doctor has the responsibility to refer the patients to the right professionals.

- From a patient point of view it was pointed out that The Specialty Plan perhaps can be formulated in a way that clarifies that all rare diseases is included in the Plan. This must be underlined, so that it is unmistakable.
- From a medical point of view, it was underlined that it is very important that the national strategy for rare diseases and The Specialty Plan are supported by the regions and at the hospitals.

At the end of the debate a consultant, who works with Rett Syndrome, told about their interdisciplinary teams, who go out locally teaching about the syndrome and give special counseling at schools and at institutions.

## Theme 5 – Orphan Medicinal Products

### Sub-Themes:

- 5.1 Support to Orphan Drug (OD) development
- 5.2. Access to treatments
- 5.3. Compassionate use programmes
- 5.4. Off label use of medicinal products
- 5.5. Pharmacovigilance

### Workshop 5: Medicine and other treatment of rare diseases

Relevant chapters in the national strategy:

- Chapter 14.2 - EU initiatives for development of medicine.

By way of introduction, the Chair **Birthe Byskov Holm** (BBH), President of Rare Diseases Denmark, presented the sections in EUROPLAN Guidelines and the EU recommendation that relate to the topics of the workshop. The Danish strategy's recommendations in the field were

also presented:

- Regarding medicines, (ongoing) access to the necessary Orphan Medicinal Products (OMP) in Denmark should be ensured.
- Additionally, there are a number of recommendations about treatment etc. as well as the possibilities for research, development and initiatives, but not explicitly in relation to the development and the managed introduction of OMP.

The Chair took the fact that the topic is not sufficiently treated in the national strategy as a starting point. Therefore, the workshop was mainly of an informative character, and was only characterised by discussions and specific suggestions to a minor degree.

First, Managing Director of The National Danish Innovation Foundation **Peter Høngaard Andersen** (PHA) gave a presentation about clinical trials in the field of rare diseases - what are the challenges? He went through the process of development of new medicinal products. It is an extremely protracted process from initial idea to marketing, which can take up to 10-20 years and cost billions of DKK. He advocated for a much more widespread use of genetics consisting of mapping of the genome of the individual and connection of this information with health registers. There are extended possibilities for Denmark in this area. He predicted that frequent diseases also will be divided into many subgroups in the future, which then individually become "rare". The healthcare system needs an extensive technological upgrading.

Finally, PHA accounted for the fact that it is difficult to make controlled clinical trials for rare diseases. There is a need for new ways of thinking the format of clinical trials, and patient involvement is truly necessary. New models for managed introduction of new medicinal products are also necessary.

Chief Medical Officer **Jens Ersbøl** (JE) from The DHMA spoke about the principles for authorisation and managed introduction of new medicinal products for rare diseases. The principles for authorisation are the same for medicines for rare as well as common diseases, where benefit is weighed up in proportion to risk. Applications for authorisation are submitted to the European Medicines Agency (EMA), whereupon the Committee for Medicinal Products for Human Use (CHMP) examines quality, safety and effect. There can be a process of questions to the applicant before a potential marketing authorisation, which is given by the European Commission. However, there can be exceptions, where a medicine is authorised without prior controlled tests (in the rare disease field) due to the very small number of patients.

As of January 2015, there are 112 medicinal products, which have gained EU marketing authorisation with Orphan status. EU marketing authorisation is binding for Denmark, but managed introduction and questions of payment are a national matter of each individual

country. In Denmark, there is no institute dealing with prioritisation, as e.g. the National Institute for Health and Care Excellence (NICE) in the UK.

The DHMA assess if the medicine should be reimbursed, when it is a prescription medicine, which is to be reimbursed by the regions (The Reimbursement Committee). The DHMA has little influence on the managed introduction and payment of medicines that are reserved for the hospitals, which include all medicines with an Orphan designation. The DHMA gives dispensing authorisations for medicines without marketing authorisation. JE stressed that Denmark is one of the countries where the process up until managed introduction is fast.

**Steen Werner Hansen** (SWH) Chairman of The Coordination Committee of Managed Introduction of Hospital Medicine (KRIS) presented the procedures of KRIS: As a rule, there is a simple system for managed introduction of medicine in Denmark, which results in quick managed introduction. KRIS commits to establish whether or not new treatment should be implemented nationally as standard treatment for all relevant patients, but does not commit itself to the treatment of the individual patient. KRIS purely makes a professional assessment of the effect, side effects etc. of the medicine, because KRIS base their opinion on EMAs assessment. Economy is not a part of the basis of evaluation.

Most of the applications that KRIS receives are for cancer medicine, but there have also been a few for rare diseases, like e.g. cystic fibrosis. SWH gave some examples of criteria for KRIS' handling of applications on standard treatment:

- Is the application on standard treatment consistent with the indication(s) that the medicine is authorised for by EMA?
- Is the application on standard treatment consistent with the patient target groups of the existing studies?
- Is there an actual effect of the medicine?
- What is the relation between effect and serious side effects?
- What other available treatment option exists for the same disease?
- Do the data based on EMAs opinion reflect the treatment regime in Denmark?

KRIS is not a institute dealing with prioritisation like e.g. NICE in the UK. KRIS has authorised products for standard treatment in Denmark, which NICE has not authorised for use in England based on an economic evaluation. In a few cases, KRIS has arrived at the conclusion to reject an application of managed introduction as standard treatment. It does not prevent a department etc. in using the product anyway.

SWH did not think that KRIS is the answer to quickly put medicine for rare diseases into use. He also indicated that KRIS is not suitable for assessing medicines for very rare diseases, with only very few patients, who are typically treated in the same place. Therefore, medicines (only) for a few rare patients cannot be characterised as standard treatment. There are good treatment

environments for rare diseases in Denmark, who are better than KRIS at deciding which patients would benefit from the new medicine.

**Allan Melgaard Lund (AML)** Consultant paediatrician, MD, DMSc from CE RD-RH, presented a medical multicenter for European medicine testing based at Rigshospitalet. He stressed that Denmark is a good country for clinical trials. AML mentioned that only 7 % of medicinal products with orphan status reach market authorisation after trials. There are several disadvantages, of which two important ones are: the few patients make it easy to overlook an effect. Furthermore, it is tough on the families to take part in clinical trials that do not lead to a medicinal product.

He also pointed out that patients in Denmark get economic coverage for most of the authorised medicines for rare diseases, when the medicine has been found to be clinically meaningful for the patient by professional assessment. Compassionate use is possible in Denmark and off-label is also used; off-label use as a result of drug-repurposing can perhaps become more common.

It can be difficult to assess the effect of the medicines and the long-term follow-up is essential. There is only little data on safety and the side effect profile, and collecting evidence just like traditional medicinal products is often not possible. Therefore, registers for long-term follow-ups are important.

It was ALMs opinion that there should perhaps be found a European solution for documentation and use of Orphan Medicinal Products, also including follow-up and effects in the long run. A certain patient involvement in the long-term evaluation would be desirable.

**Jannie Schymann (JS)**, Alpha-1 patient, concluded the series of presentations with a personal account of participation in clinical trials. The motivation for taking part in the first trial was a sense of duty and enthusiasm about being able to get medical treatment for Danish patients on par with the treatment that is i.e. given in Germany. But the results of the trial was not approved, i.e. the measuring methods were disputed. To take part in the next trial was a way to get medicine herself, even with a good effect. However, this second trial did not lead to patients getting the medicine either. She has declined to be part of a third clinical trial with the same kind of treatment out of fear for the side effects of a great number of CT scans - and for lack of trust in "the system".

**JS** suggested that more clear rules for arranging trials should be developed, i.e. including a clarification on measuring methods, side effects and evidence requirements. In order for the patients to be able to trust the management of a clinical trial, there has to be involvement of the patients and be given thorough information throughout. There has to be transparency and a clear division of responsibilities in relation to managed introduction of medicines – and there

has to be a concrete obligation to initially take an initiative to bring effective medicines in use.

At this point in time, the Chair opened the floor:

- From a patient point of view, it was asked what the patients should do when the medicine they receive is suddenly not produced anymore. Nobody had an answer to this. **JE** pointed out that it is the responsibility of the manufacturer to deliver. However, there are no national possibilities for sanctions, if they do not.
- From a patient point of view it was also asked if the medicine for Alpha-1 patients has not been tested in other countries, since the treatment is used in several countries. Why are those data not used in order to determine the Danish treatment regimen, when there are so few patients? It underlines the need for registers. **JE** thought that the procedures regarding the Alpha-1 patients have not been very pretty. Personally, he believed that patients, who voluntarily take part in clinical trials and who have a positive effect from it, should also have access to the medicine after the end of the trial.

**The Chair** addressed the question whether or not KRIS is the solution in relation to securing that medicinal products aimed at rare conditions reach the patients. The Chair summed up that KRIS does not think so, but there are examples of KRIS processing orphan medicinal products e.g. for cystic fibrosis. This indicates that there is a need for assessment of these products, even though it may primarily be for diagnosis groups of 200-300 patients, when it makes sense to talk about standard treatment. There is no need for a Danish NICE, but perhaps for a mechanism, which can help patients through the system. Perhaps the way forward is to centralise the systems/collect expertise further, so that it will be the clinical environments that have to put the medicine into use?

**SWH** answered that if it is a question of several hundred patients and products based on randomised trials, then it could be a possibility to use KRIS. But when it is a question of very few patients, then KRIS is not suitable. Regarding treatment of the rare metabolic diseases, it is best linked with the clinical environment, where it is now. This also applies to other rare diseases, and SWH is personally a great advocate of relating to existing data at a clinical level, when it is a question of very few patients and coordinating treatment nationally. When there is a critical clinical assessment of effect and costs KRIS will not be needed for this.

## Theme 6 –Social Services for Rare Diseases

### Sub-Themes

- 2.5 Help Lines
- 6.1. Social resources for people with disabilities
- 6.2. Specialised social services for rare diseases
- 6.3. Policies to integrate people living with rare diseases into daily life
- 6.4. International–supranational dimension

### Workshop 6: Social service for PLWRD

The relevant chapters in the national strategy:

- Chapter 7 - organisation of services in the municipalities
- Chapter 8 - new structure in the field of social services
- Chapter 9 - coordination and coherence
- Chapter 11 - empowerment

By way of introduction, the Chair **Lene Jensen** from Rare Diseases Denmark presented the sections in EUROPLAN Guidelines and the EU Recommendation that relate to the topics of the workshop. The Danish strategy's recommendations in the field were also presented:

- Organisation in the municipalities:
  - Focus on access to the services of municipalities.
  - Focus on transition between childhood and adulthood.
  - Access to updated, valid and interdisciplinary knowledge.
  - Continuity and stability in cooperation between municipality and family.
- Coordination and coherence:
  - Coherence in patient/citizen procedures.
  - Preferably one coordinating case manager as coordinator.
- The National Coordination Structure (NCS):
  - To secure knowledge and competences in relation to the most complicated rare groups.
- Empowerment:
  - Specific focus on the rare field.
  - Access to patient education and relevant networks / activities.
  - Development of counseling and services for PLWRD across health care and social services.

**Kirsten Denning** (KD), Manager of Social Services and Handicaps at Gentofte Municipality, gave a presentation on rehabilitation of PLWRD. First, KD made it clear that the municipalities play an important role in many of the lives of PLWRD, because there is a need for social service and support. In the municipality of Gentofte, with 75.000 citizens, social workers report that they

on average meet a citizen affected by a rare disease approx. once a year.

The point of departure for KD's presentation was re/habilitation in relation to the citizens' needs and specific problems. Work is based on the following understanding of the terms: "Rehabilitation is a goal-oriented collaboration process, limited for a certain period of time, between a citizen, relatives and professionals. The aim is, for the citizen, who has or is at risk of getting significant limitations in his/her physical, psychological and/or social functional capacity, to have an independent and meaningful life. Rehabilitation is based on the whole life situation and decisions of the citizen and consists of a coordinated, interconnected and knowledge-based effort."

The method of accounting for the functional capacity of adults was briefly covered – the method results in a description of the functional capacity of the citizen and its importance for participating in all sections of society. It is strived for to have a focus on resources - not just to focus on what is not possible! But the system has to understand the severity of the situation of each individual and the citizen must know why the caseworker focuses on resources. KD accounted for the central elements in the re/habilitation work:

- Systematic social service reports with a view to be able to evaluate the entire functional capacity of the citizen.
- Focus on the resources of the citizen and on where skills can be improved. A focus on the need for an effort in order to maintain a functional capacity.
- Focus on the network of the citizens and on the requests and goals of the citizen.
- To determine the right effort, possibly by consulting persons with knowledge in the rare field.
- Specific goals have to be established, on which follow-ups can be made. It is central that the goal of the citizen is in focus.

Regarding coordination of the effort for PLWRD, KD pointed out that there are often a lot of contacts to keep track of. Coordinating meetings around the individual rare patient are really useful - it is known from the field of brain damage and can also be used regarding PLWRD. As it is now, municipalities are presumably drawing too much on the relatives. They always have to be involved, but in many cases their responsibility to coordinate is far too big.

KD believed that in general, a lot of knowledge about disabilities exists, but this is not the case when it comes to *rare handicaps*:

- General knowledge about diagnoses and consequences for the functional level of the citizen/ *PLWRD: smaller amount of fingertip knowledge. Need for professionals to look for knowledge.*
- Many citizens have coordination needs e.g. in the field of brain damage/ *PLWRD: generally a big need for coordination (health, social support, education, employment etc.).*

- Need for more knowledge about which methods and efforts have an actual effect/*this is also the case in the rare disease field.*
- Challenges of transition e.g. when turning 18/*this is also the case for the rare disease field.*
- The family needs empowerment/*this is also the case in the rare disease field.*
- Often possibilities to meet others in the same situation/*PLWRD: difficult to meet others in the same situation.*
- Services for the target group exist, often within a reasonable distance/*PLWRD: the need for some kind of service can be prioritised prior to target groups, age etc., resulting in less focused services.*

Finally, KD pointed out that when it comes to rare diseases and handicaps, it is important that the municipality can draw on knowledge, because it is impossible to have specialised knowledge about everything. The municipalities depend on being able to find knowledge at The NBSS or in the organisations for PLWRD.

Afterwards, the word was passed on to **Jes Rahbæk** (JR) Managing Director and Consultant from The National Rehabilitation Center for Neuromuscular Disorders, who talked about rehabilitation with focus on the citizen. JR told that he has been involved in the rare field back in the 1990s. At that time, many conferences were held and there was a thought provoking similarity between the problems that were put forward then - and those that have already been put forward at this conference. According to JR, the strategy is good, but there is nothing new about it.

JR defined "rehabilitation" as a term in the same way that KD did in her presentation and underlined that the citizen has to be involved. The citizen is in charge. JR put this into perspective by illustrating how many professionals and groups a typical citizen with muscular dystrophy is in contact with during the rehabilitation process. It is an extremely complex network with the citizen in the eye of the hurricane.

Furthermore, JR mentioned that significant progress has been made in the past 10-20 years in the healthcare sector, where expertise has been centralized. From having had 20 centers, the muscular dystrophy citizens now have three Centers and the treatment available is on par with that of other countries. In the municipalities, on the other hand, there are a number of challenges. The citizens often experience services of the municipality to be "divided into silos" and uncoordinated. An effort is needed to improve this - a heavy task lays ahead for the service users in telling the municipalities which efforts have effect.

Afterwards, the word was given to **Kirsten Brøndum** (KB) acting Head of Office at The NBSS, who talked about the experiences of VISO (the National Organisation for Knowledge and Specialist Consultancy) regarding PLWRD. KB told that VISO renders specialised counseling to

citizens and professionals regarding specialised, professional areas of effort.

The point of departure is that many challenges for the individual person are not connected specifically to a diagnosis, but to the disability/ies, which can be addressed generally. VISO has contracts with more than 110 specialists, who can become involved. In 2013, there were about 1.500 cases that had been initiated at VISO of which 69 were related to rare diseases.

VISO has a certain focus on the rare field. In 2013, a professional network of VISO specialists with specific competences in the rare field was established. The experience is that it makes sense and that it shall continue.

Subsequently, the word was passed to **Else Lund Frydensberg (ELF)**, Head of Office for The National Coordination Structure (NCS) under The NBSS. ELS told that NCS came into existence after a period of decrease in demand from the municipalities for specialised counseling, service and information. It gave rise to a fear of undesirable de-specialisation. Therefore, NCS has been politically recommended, and became operational in July 2014.

NCS is meant to bring services and knowledge about small, complex target groups, including PLWRD, into focus, to:

- Ensure the necessary range of services - also for small target groups.
- Strengthen quality and collaboration.
- Ensure that the services are developed dynamically along with the needs.
- Ensure specialised knowledge, nationally.
- Create an overview of the most specialised areas and monitor where knowledge can be found etc.
- Make instructive procedure recommendations.

Furthermore, NCS can make announcements if there is a concern for undesirable de-specialisation. Everybody is encouraged to report and create awareness, if and when this happens.

The last speaker was Managing Director **Lene Jensen (LJ)** from Rare Diseases Denmark, who spoke about empowerment and PLWRD as a resource. LJ's approach was that a prerequisite for an effective effort is that the citizen and/or the family can handle his/her own situation. The same is true if PLWRD should to be a knowledge resource.

LJ pointed out that contact with others in the same situation is what PLWRD need the most. This has been established in several studies and this is difficult in the rare field for obvious reasons. Rare Diseases Denmark has 49 membership associations, which cover approx. 200 diagnoses, as well as The Rare Network for PLWRD and their relatives representing approx. 170 other diagnoses. Volunteers run most of the associations. 800 different diagnosis are

estimated to be present in Denmark

There are good objectives and goals in the National Strategy in this field, especially in the recommendations in chapter 11, but there is no clear plan for how they are to be carried out. LJ suggested that an action plan should be made for a NGO-managed “rare package”, which relates to:

- Creating better possibilities for meeting and establishing networks for patients and relatives, render mutual service, regardless if there is an association at one's disposal or not.
- To create access to specific patient education meeting the needs of PLWRD.
- Establishment of a Help Line for PLWRD, where patients as well as relatives - and perhaps also professionals - can call and get counseling on how to handle life with a rare diagnosis and how to get on in "the systems".

The need for the last-mentioned is apparent every day in the small secretariat of Rare Diseases Denmark, where many enquiry's are made without enough resources to be handled well enough. The need is acknowledged in the strategy, but with no clear responsibility. LJ said that Rare Diseases Denmark would be glad to take on the responsibility, if the necessary resources were allocated.

Then the floor was open for suggestions and comments from the other participants of the workshop. A number of topics were brought into play:

- From a patient point of view, it was stated that there is a lack of knowledge about the interaction between daily life and the services offered. The municipalities have to find the necessary knowledge, also at Rare Diseases Denmark - and the municipality has to accept the knowledge and information that is put at their disposal.
- From a patient point of view, a request was made to grant Rare Diseases Denmark a social worker. The individual rare citizen has a need for competent professional guidance in order to get the right social services from the municipalities, where there is a lack of knowledge about managing life with a rare handicap.
- Ågrenska in Sweden was mentioned, where a number of activities take place with regard to create empowerment in the rare families. The whole family takes part in a course in order to become more knowledgeable on life with a rare disease and subsequently they become a lot more socioeconomically advantaged.

**KD** told that the municipality looks for knowledge at VISO and at The NBSS. But there is a need for better access to more knowledge - how can we make better use of each other? The patient organisations are an obvious resource.

**KB** thought that rehabilitation has to be seen in a broad perspective - there is not only a need for an effort in the social service area, but also in employment, training etc.

**JR** told that family courses are a regular service for people affected by muscular dystrophy. Everybody is offered to go to a family course, typically for a weekend, approx. every second year. Furthermore, JR pointed out that many diseases are progressive. In that context, rehabilitation can be difficult to understand. VISO refers back to the municipalities and VISO are often rejected, because they indicate suggestions that involve expenses. The Muscular Dystrophy Foundation goes to the homes of the citizens / the municipalities in order to facilitate the processes.

- From a medical point of view, it was pointed out that there is also a need to focus on those who do not have an evident diagnosis or have to fight for a long period of time to get it. They constitute a specific challenge.
- A patient representative pointed out that it is not enough for NCS to prevent de-specialisation, there is also a great need for upgrading of specialisation. The patient representative also thought that social work has to relate to the complexity of problems as a whole. In some municipalities a specific expertise in relation to a given diagnosis exists and therefore a free choice of social workers across the municipality boundaries should be established; it would provide a higher professional standard and better service and perhaps even administrative cost reductions.

**JR** pointed out that there is nothing as economical as qualified counseling. It is important to have specialists, but coordination is extremely important, when one has to commit to rehabilitation.

**ELF** agreed that specialised counseling can contribute with systematising and mapping the knowledge at one's disposal, with focus on the individual disabilities.

**KB** pointed out that in many areas, there is no documented knowledge to be found. Here, we need the professionals, who work with the citizens to submit documentation of their experiences.

**Additional Workshops (optional)** – no additional workshops

### **Report of the Closing Session – Conclusions**

Moderator: Managing Director Lene Jensen, Rare Diseases Denmark

#### **Brave new rare world - what does tomorrow bring?**

**Michael Bjørn Petersen** Clinical Director, Professor, MD, PhD (MBP) from Aalborg University Hospital gave a presentation on the development of diagnostic methods based on genetics.

MBP went through specific cases and concluded that with Next Generation Sequencing (NGS) we have new technology, which can be used in order to scan the whole genome for genes

carrying diseases. This ensures better diagnostics of rare diseases, where the cause cannot be identified in other ways. High-tech platforms are used as well as very advanced bioinformatics. Prices are falling, which makes the technology a realistic diagnostic tool in practice.

When the genome is sequenced, you will get a number of incidental findings. It is a big and important discussion. The Danish Association for Medical Genetics (DSMG) is drawing up a new proposition for informed consent, which also includes genome sequencing.

MBP referred to the American College of Medical Genetics, who are suggesting that when the genome is being sequenced in its entirety, 55 high-risk genes should actively be searched for, which then have to be reported on to the commissioning doctor - even though they were not the actual reason for the examination. According to the American recommendations, this should be done regardless of the age of the patient, when it is a question of diseases with high penetration and possibility for prevention/treatment. We have not reached this far in Denmark - it is a completely new set of problematics.

MBP concluded that the new technologies definitely will make diagnostics of rare diseases better. Concurrently, new ethical dilemmas are arising, which need to be addressed.

### **Feedback to Plenary**

The feedback session was attended to by the **Chair** of each workshop and had to consist of (some of) the workshop's specific suggestions for implementation of the strategy and a better effort for rare patients and their relatives.

### **Workshop 1: The road ahead for the Danish rare policy / the national strategy**

During the workshop focus had been on the specific recommendations in relation to i.e. the implementation and monitoring of the strategy and coherence of the effort as well as knowledge development. There were especially two specific suggestions, which were repeated by the participants of the workshop:

- Establishment of networks: Ensuring a continuous focus on the implementation of the national strategy via a forum or a network with participation of the central stakeholders (state, municipal, patient's organisations and professionals) across sectors. Based on the strategy, it needs to be discussed what can be done.
- Connection of people who possess knowledge: Several of the participants were focusing on the fact of rarity of both patients and specialists, which makes it necessary to connect them. This connection can be made between sectors, but also within a sector. Suggestions were made to use the Regional Health Agreements in order to ensure this. It was also pointed out that it would be an advantage if one became better at sharing knowledge and information at the local municipal level.

### **Workshop 2: Information and training in rare diseases**

The overall focus of the workshop was about information and training. Additionally, the main presentation of the workshop focused on thinking about coordination in another way, so the families of PLWRD are better prepared to take part in the coordination task. A number of specific suggestions were made during the workshop:

- About information: there is a need for valid and quality assured information in Danish, preferably with one access point to all information. There was massive support of the recommendations of the strategy for The Rare Diagnosis Information Database. It was pointed out that it could be suitable to set up a steering group or the like regarding this Information Database. There was also a focus on the need to create, collect and communicate new knowledge. The GPs made a request for a hotline or a place to contact (one phone number) to get medical knowledge as well as knowledge on whom to contact regarding social services and support.
- About training: it is not sufficient to look at further training, even though it is important, i.e. in relation to the General Practitioners. Instead, in the basic training (social service and healthcare), it should be taught how to think when you meet a patient with a rare diagnosis - what the problems are and what is distinctive for rare diseases.

In plenary, it was pointed out that if you have to collect all this knowledge in one place, Sundhed.dk or CE RD's homepages could be a possibility. It was also stated that there is an enormous need for more quality assured information in Danish.

### **Workshop 3: Research in rare diseases and handicaps**

The workshop was based on the patients' uniqueness and interest in contributing to research, and this is a resource which should be cherished. The patients should perhaps be included more in the planning as well as the evaluation of the research projects. There were especially two specific suggestions/requests that recurred in the debate:

- Better possibilities for clinical research at CE RD, because it would improve the quality of the treatment. Here, the regions have a responsibility to ensure that there are enough resources - but have the regions forgotten to give money for research?
- Danish researchers, who conduct research in the rare field, need the possibility to participate in international cooperation and EU programs on rare diseases. Researchers can apply for these programs, if their country is a participant, but Denmark is not. The question is who is going to support this participation (The National Innovation Foundation, The Research Committees)?

In plenary, it was pointed out that the regions have given some money for research.

### **Workshop 4: Diagnostics, treatment and a lot more - Centres of Expertise**

During the workshop there had partly been focus on delayed diagnosis - and partly on treatment and procedures. Regarding diagnostics it was agreed that earlier and more timely diagnostics demand a cultural change - less 'Tarzan behavior' (i.e. doctors' reluctantly in referring patients from one specialist to another) is called for as well as a more liberal referral

policy, matched by a more liberal receiver policy. A preamble should be made about this. Additionally, concrete suggestions were made:

- Making “rare packages<sup>1</sup>” attached to a schedule in relation to referral and placement of the referral procedures. It should also be contemplated that in connection with problems with establishing a diagnosis it should be possible to involve a board consisting of “expert experts”, who should function on an ad hoc basis and with the involvement of different specialties.

Regarding treatment and procedure, it was underlined, that there are many procedures, which work really well - at CE RD as well as in other places.

- When better procedures are made, they should be based on the well-functioning procedures that actually exist. Better procedures can also be inspired by other fields, where patients also need coordination and a coherent procedure, e.g. diabetes patients. The rare disease field has to interact with other specialties, to avoid isolation.

It was pointed out in plenary that if something effective is to be done, it has to be done nationally. If “rare packages” are formulated, there is a demand for acceptance in the same manner as for example “cancer packages”. It should entail diagnosing and interdisciplinary collaboration as well as research - the rare field needs to be organised to ensure interdisciplinarity. There ought to be specific focus on adult patients, also in the transition from childhood to adulthood.

#### **Workshop 5: Medicine and other treatment of rare diseases**

The National Strategy does not deal much with the themes of the workshop. Therefore, the workshop was organised differently with more presentations of an informative nature. There was only little time for discussion, but the workshop covered many topics.

The difficulties of developing medicinal products for rare diseases were discussed, including the authorisation procedures at EU level and nationally. Despite not having a structured system for managed introduction of medicine (such as NICE, the National Institute for Health and Care Excellence in the UK) in Denmark, it is going quite well and there is no need for a “Danish NICE”.

- Medicine gets to the patients quite fast, but there is room for improvement - there are several examples of this. An initial obligation has to be placed on managed introduction of medicines and transparency is important.

The positive narrative about development, testing and treatment with medicines for metabolism disorders were highlighted. In Denmark, there are good clinical places, with knowledge and capacity to handle and evaluate new treatments.

- This is also needed for other areas and it should be developed along the lines, which are pointed out in the strategy: Gathering of expertise, establishment of knowledge

<sup>1</sup> In the Danish Healthcare system exists several disease specific “package procedures” is defined as a set of patient rights and deadlines regarding diagnosis, treatment and more is defined. There are no “packages” dedicated to rare diseases.

centers across specialties etc., in order for the clinical environments to be responsible for bringing the medicine into use.

#### Workshop 6: Social service for PLWRD

The focus of the workshop had partly been social services for PLWRD, including coordination of the effort and partly empowerment of PLWRD. Coordination was also a topic of workshop 2.

- In workshop 2 as well as 6 there was focus on the fact that coordination is not a static mechanism, where the same mechanism can be used over the years for all PLWRD. It has to be thought in a more dynamic way and as an interaction between all stakeholders; there were suggestions for developing new models for this.
- About knowledge, systematised knowledge was called for and it was suggested that The NCS has focus on avoiding de-specialisation as well as upgrading of specialisation, where the services are not good enough. Furthermore, there was a discussion about access to social worker function.
- Additionally, a social NGO-driven “rare package” was suggested consisting of more systematic patient education, better conditions for the establishment of networks and establishing a Help Line.

In plenary, it was stated that the NCS does not only have to focus on de-specialisation, but also on specialised knowledge being developed and implemented. It was also pointed out that The National Rehabilitation Center for Neuromuscular Disorders has a model, which could be used as an example for imitation regarding citizens affected by complex conditions.

#### The closing of the EUROPLAN conference

After the feedback in plenary, the word was given to **Birthe Byskov Holm**, President of Rare Diseases Denmark, for closing remarks.

Birthe Byskov Holm thanked the participants for the many discussions, ideas and suggestions. The conference is reported in Danish and English in the context of the EUROPLAN project. Moreover, regarding the further procedures, Birthe Byskov Holm pointed out that it is all about taking responsibility in the different areas. If it is the responsibility of everybody, no one is really responsible. Rare Diseases Denmark will try to live up to their responsibility in relation to what the patient organisations can do. However, their role will also be to attend to the interests of all PLWRD by continually insisting on all other responsible stakeholders also attending to their part.

### III. ANNEXES

#### ANNEXE 1: PROGRAMME

#### **Programme of the Danish EUROPLAN-conference**

**23 January 2015**

**IBOS, Rymarksvej 1, DK-2900 Hellerup**

#### **Welcome coffee (09.30am)**

#### **Opening session (10.00 – 11.30am)**

Moderator: Birthe Byskov Holm, President of Rare Diseases Denmark

10.00-10.15: Opening speech, Chairman Bent Hansen, Danish Regions

10.15-10.25: The European context, EUROPLAN Advisor Lene Jensen, Rare Diseases Denmark and Representative Laurs Nørnlund, European Commission in Denmark

10.25-10.40: Efforts for people living with rare diseases, Director General/CEO Else Schmidt, The Health and Medicines Authority and CEO Knud Aarup, National Board of Social Services

10.40-11.15: A Progress Report on Denmark: The National Strategy, Senior Medical Officer Marianne Jespersen, The Health and Medicines Authority and Head of Office Randi Lykou, National Board of Social Services

11.15-11.20: Workshops - solutions' playground, Birthe Byskov Holm

#### **Workshop 1- 3 (11.30 – 13.00)**

#### **Lunch (13.00 – 13.45)**

#### **Workshop 4 - 6 (13.45 – 15.15)**

#### **Coffee break (15.15 – 15.30)**

#### **Plenary session (15.30 – 16.25)**

Moderator: Lene Jensen, Managing Director Rare Diseases Denmark

15.30-15.45: Brave new rare-world - what will tomorrow bring? Clinical Director, Professor, MD, PhD Michael Bjørn Petersen, Aalborg University Hospital

15.45-16.25: Two excellent ideas from each workshop - and what to do next?

#### **Closing of the EUROPLAN-konferencen (Kl. 16.25 – 16.30)**

16.25-16.30: Thank you - Birthe Byskov Holm, President of Rare Diseases Denmark

#### ***Workshop 1: The way forward for Danish rare-policy / national strategy***

Chair and rapporteur: Acting Head of Office Kirsten Brøndum, National Board of Social Services – Mette Grentoft, Rare Diseases Denmark/Danish Society for Williams syndrome

- From strategy to action, CEO Leif Vestergaard, The Danish Cancer Society
- Other contributions:
  - o The strategy's content about implementation, monitoring and evaluation, with particular focus on the area of health, Senior Medical Officer Marianne Jespersen, The Health and Medicines Authority

- The strategy's content about implementation, monitoring and evaluation, with particular focus on the social field, Head of Office Randi Lykou, National Board of Social Services
- How do they manage in other countries?, President Birthe Byskov Holm, Rare Diseases Denmark

**Workshop 2: Information and education on rare diseases**

Chair and rapporteur: Managing Director Lene Jensen, Rare Diseases Denmark

- How does the arena of knowledge look in the rare field - a researcher's encounter with the rare knowledge challenge - Leif Olsen, Senior Research Fellow at KORA, the Danish Institute for Local Government Analysis and Research
- Other contributions:
  - Where is the entrance to knowledge? Holger Bang-Møller, the father of a girl with a rare disease
  - What knowledge / information must be available to doctors, so they recognize rare diseases? Chairman Lars G. Johansen, The Danish College of General Practitioners
  - Who in the social- and health sector must be trained to perform the task on the rare diseased? Head of Department Lykke Jensen, Metropolitan University College
  - The highly specialised knowledge suppliers - what does it take in daily life and education? Consultant Paediatrician, MD, Ph.d. Stense Farholt, CE RD-AUH

**Workshop 3: Research in rare diseases and handicaps**

Chair and rapporteur: Professor Karen Brøndum Nielsen - Søren Lildal, Danish Apert Patient Society

- Challenges for research in the rare field, Professor Ebba Nexø
- Other contributions:
  - Research Council's practice, Professor Troels Staehelin Jensen, Representative for The Danish Council for Independent Research
  - Danish participation in international research - what happens? Consultant paediatrician, MD, DMSc Allan Meldgaard Lund, CE RD-RH
  - Finding the patients, Consultant paediatrician, MD, DMSc Hanne Hove, CE RD-RH
  - Rare citizens political research Agenda, Vice Chairman Liselotte Wesley Andersen, Rare Diseases Denmark / Danish Patient Society of Tuberous Sclerosis

**Workshop 4: Diagnostics, treatment and more - Centres of Expertise**

Chair and rapporteur: MD, DMSc Henning Bundgaard – Liselotte Wesley Andersen, Rare Diseases Denmark/ Danish Patient Society of Tuberous Sclerosis

- The rare diseases in the hospital service - a challenge? Regional Chief Operations Officer Svend G. Hartling, The Capitol Region of Denmark
- Other contributions:
  - The Danish model for highly specialised healthcare, Senior Medical Officer Marianne Jespersen, The Health and Medicines Authority
  - Wauv... to be a Center of Expertise... , Consultant Paediatrician, MD, Ph.d. Stense Farholt, CE RD-AUH
  - An holistic approach to the patient, President Terkel Andersen, EURORDIS / The Danish Haemophilia Society

### **Workshop 5: *Medicine and other treatment of rare diseases***

Chair and rapporteur: President Birthe Byskov Holm, Rare Diseases Denmark – Professor Karen Brøndum

- Clinical trials in the rare area: What are the challenges? Managing Director of The National Danish Innovation Foundation Peter Høngaard Andersen
- Approval and entry into service of new medicine - black box or crystal shrine? Chief Medical Officer Jens Ersbøl, The Health and Medicines Authority
- Other contributions:
  - o Access to new treatment – is KRIS the answer? Chairman Steen Werner Hansen, The Coordination Committee of Managed Introduction of Hospital Medicine, Danish Regions
  - o Entry into service of medication - examples from the metabolic area. Consultant paediatrician, MD, DMSc Allan Meldgaard Lund, CE RD-RH
  - o To participate in the testing of new medicines - duty or opportunity? Jannie Schymann, Alfa-1-patient

### **Workshop 6: *Social services to people living with rare diseases***

Chair and rapporteur: Managing Director Lene Jensen, Rare Diseases Denmark - Søren Lildal, Danish Apert Society

- Rehabilitation of people with rare disabilities - what does it take and how to ensure coordination? Manager Kirsten Dennig, Social Services and Handicaps at Gentofte Municipality
- Other contributions:
  - o With the citizen in center, Managing Director and Consultant Jes Rahbek, The national rehabilitation center for neuromuscular disorders
  - o Specialist consultancy, experiences from VISO about the rare field, Acting Head of Office Kirsten Brøndum, National Board of Social Services
  - o National coordination for rare diseases, Head of Office Else Lund Frydensberg, National Board of Social Services
  - o Empowerment – PLWRD as a resource, Managing Director Lene Jensen, Rare Diseases Denmark

## **ANNEXE 2: LIST OF PARTICIPANTS**

### **Stakeholder Groups:**

- Academic/Researcher
- Clinician/GP
- Healthcare Professional (other than clinician or GP)
- Industry
- Insurer
- Medical /Learned society
- Patient representative
- Politician
- Public administration (local, regional or national)
- Social worker
- Other

### **Roles:**

- C:** Chair
- R:** Rapporteur
- KS:** Speaker in plenary or key note speaker in workshop
- S:** Speaker
- M:** Moderator
- P:** Participant

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Name	Institution	Role	Group
Aasa Magnusson	Alexion	P	Industry
Allan Bayat	KC/PKU	P	Clinician/GP
Allan Meldgaard Lund	CSS/CMS Rigshospitalet	S	Clinician/GP
Amalia Egle Gentile	EUROPLAN Project	P	Other
Anders Olausson	Aagrenska	P	Other
Ann Hjort	Novartis	P	Industry
Anne Andersen	VISO, Socialstyrelsen	P	Public administration
Anne Elming	Rigshospitalet	P	Social worker
Anne Grethe Lauridsen	Gaucher-foreningen i Danmark	P	Patient representative
Anne Hamann	Synscenter, Refnæs	P	Social worker
Anne-Lis Mark	Rigshospitalet	P	Social worker
Anne-Marie Bisgaard	Rigshospitalet	P	Clinician/GP
Anne-Marie Gerdes	Rigshospitalet	P	Clinician/GP
Annette Vind	Sundhed.dk	P	Public administration
Antonio Atalaia	EUROPLAN Project	P	Other
Bent Hansen	Danske Regioner	KS	Politician
Bente Bjerring Nielsen	Mitokondrie-foreningen i Danmark	P	Patient representative
Bente Møller	Sundhedsstyrelsen	P	Public administration
Birgit Meister	MSA-foreningen	P	Patient representative
Birgitte Ravn Sølvhøj	Sundhed.dk	P	Public administration
Joan Andersen	Turnerforeningen	P	Patient representative
Birthe Byskov Holm	Sjældne Diagnoser, OI-foreningen	C,KS,M,S	Patient representative
Birthe Damkjær	Frederikssund Kommune	P	Public administration
Bo Libergren	Region Syd	P	Politician
Bodil Højgaard	PKU-foreningen	P	Patient representative
Bodil Kallesen	VISO, Socialstyrelsen	P	Public administration
Britta Christensen	DUKH	P	Social worker
Britta Ortiz Echeverria	Region Sjælland	P	Public administration
Charan Nelander	Komiteén for Sundhedsoplysningen	P	Public administration
Charlotte Agger	Demenscentret Pilehuset	P	Healthcare Professional
Charlotte Munk	Smith-Magenis Syndrom Foreningen	P	Patient representative
Christine i Dali	Rigshospitalet	P	Clinician/GP
Diana Holm	Foreningen for ATAKSI/HSP	P	Patient representative
Dorte Haubek	Dr. odont, ph. d	P	Clinician/GP
Dorthe Gaarsdal	MLD-Foreningen Danmark	P	Patient representative
Dorthe Lykke	Foreningen for ATAKSI/HSP	P	Patient representative
Dorthe Lysgaard	TrygFonden	P	Other
Ebba Nexø	Professor	KS	Academic/ Researcher
Elisabeth Nørgård Andreasen	VISO, Socialstyrelsen	P	Public administration
Ellen H. Thomsen	Region Sjælland	P	Politician
Else Danø	RCfM	P	Social worker
Else Lund Frydensberg	Socialstyrelsen	S	Public administration
Else Schmidt	Sundhedsstyrelsen	KS	Public administration
Erik Wendel	Landsforeningen til bekæmpelse af Cystisk Fibrose	P	Patient representative

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Flemming Møller Mortensen	Sundhedsordfører (S)	P	Politician
Gertrud Rex Baungaard	Sundhedsministeriets departement	P	Public administration
Gitte Ludvig	SOBI	P	Industry
Gunhil Nørhave	Alfa-1 Danmark	P	Patient representative
Gurli Erikke Wolf	Børneamb, Hosp.enheden Vest	P	Healthcare Professional
Hanne Hove	Rigshospitalet	S	Clinician/GP
Hanne Jepsen	CFD	P	Healthcare Professional
Hanne Marie Damgaard Kristensen	Socialstyrelsen	P	Public administration
Hanne Merete Sørensen	Spurvetofte	P	Social worker
Hanne Stig Andersen	Socialministeriets departement	P	Public administration
Hans Christian Laugaard-Jacobsen	Aalborg UH	P	Clinician/GP
Hans Gjørup	Odontologisk videntcenter AUH	P	Clinician/GP
Helle Harder	Herning Kommune	P	Public administration
Henning Bundgaard	Rigshospitalet	C	Clinician/GP
Henrik Ib Jørgensen	Muskelsvindfonden	P	Other
Holger Bang-Møller	Forældre til barn	S	Patient representative
Ida Vogel	Ledende overlæge	P	Clinician/GP
Inge Kristensen	Ballerup Kommune	P	Public administration
Ingrid Reimer	Crouzonforeningen	P	Patient representative
Irene Kibæk Nielsen	Uddannelsesansvarlig overlæge	P	Clinician/GP
J. Michael Hasenkam	Dr. med. Professor i eksperimentel hjertekirurgi	P	Clinician/GP
Jacob Brønnum	Socialstyrelsen	P	Public administration
Jannie Schymann	Alfa 1-Danmark	S	Patient representative
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Jens Michael Hertz	Odense Universitets Hospital	P	Clinician/GP
Jes Rahbek	RCfM	S	Clinician/GP
Jette Daugaard-Jensen	Odontologisk Videnscenter	P	Clinician/GP
Johnny Krogh Hansen	Synageva BioPharma Corp		Industry
Julie Bergtorp	Socialministeriets departement	P	Public administration
Julie Trolle	Baxter	P	Industry
Jørgen Jeppesen	RCfM	P	Other
Karen Binger Holm	Danmarks Bløderforening	P	Patient representative
Karen Brøndum	Professor	C,R	Academic/ Researcher
Karina Kragerup	Kennedy Centret	P	Social worker
Karsten Jensen	Dansk Forening for Osteogenesis Imperfecta	P	Patient representative
Kasper Steenberg	Socialstyrelsen	P	Public administration
Kirsten Brøndum	Socialstyrelsen	C,S	Public administration
Kirsten Dennig	Gentofte Kommune	KS	Public administration
Kirsten Washuus	CFD	P	Healthcare Professional
Kirsten Rasmussen	Vejle sygehus	P	Clinician/GP
Kirstine Stochholm	CSS Aarhus Universitets Hospital	P	Clinician/GP
Kis Laursen	Det Unikke Barn	P	Other
Knud Aarup	Socialstyrelsen	KS	Public administration
Lars Dreier	MLD-Foreningen Danmark	P	Patient representative

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Lars Ege	Socialstyrelsen	P	Public administration
Lars G. Johansen	DSAM	S	Medical/ Learned society
Laurs Nørlund	Europa Kommissionen	KS	Other
Leif Olsen	KORA	KS	Academic/ Researcher
Leif Vestergaard	Kræftens Bekæmpelse	KS	Patient representative
Lene Jensen	Sjældne Diagnoser	C,KS,M,S	Patient representative
Lene Lind	Dansk Forening for Neurofibromatosis Recklinghausen	P	Patient representative
Liat Damsbo Lund	Rigshospitalet	P	Social worker
Lilian Bomme Ousager	Odense Universitets Hospital	P	Clinician/GP
Linda Bødker	Næstved Kommune	P	Public administration
Lis Callesen	Genzyme ApS		Industry
Lisbet Ottesen	WilsonPatientforeningen	P	Patient representative
Lisbeth Tranebjærg	Bispebjerg Hospital	P	Clinician/GP
Liselott Blixt	Sundhedsordfører (DF)	P	Politician
Liselotte Wesley Andersen	Sjældne Diagnoser, TS-foreningen	S,R	Patient representative
Lone Bech Christensen	RCfM	P	Healthcare Professional
Lone Sunde	MD, PhD	P	Clinician/GP
Louise Boserup	Dansk Forening for Neurofibromatosis Recklinghausen	P	Patient representative
Lykke Jensen	Metropol	S	Public administration
Marianne Jespersen	Sundhedsstyrelsen	KS, S	Clinician/GP
Marianne Verdel	Center for Hjerneskade	P	Healthcare Professional
Marie Louise Mølgaard Binderup	Panum Institutet	P	Clinician/GP
Marie Schack	Dansk forening for Albinisme	P	Patient representative
Marlene Borst Hansen	MF, Sundhedsordfører (RV)	P	Politician
Merete Daugaard Gabay	Rigshospitalet	P	Social worker
Mette Grentoft	Sjældne Diagnoser, Williams-foreningen	R	Patient representative
Mette Sommerlund	overlæge, Ph.d., klinisk lektor	P	Clinician/GP
Michael Petersen	Aalborg Universitets Hospital	KS	Clinician/GP
Niels Illum	Odense Universitets Hospital	P	Clinician/GP
Nina Tuxen	WilsonPatientforeningen	P	Patient representative
Patricia Egge	Børneklíniken	P	Healthcare Professional
Peter Høngaard	Innovationsfonden	KS	Other
Pia Illum	Region Hovedstaden	P	Politician
Preben Sindt	Sjældne Diagnoser, Rett-foreningen	P	Patient representative
Randi Lykou	Socialstyrelsen	KS,S	Public administration
Rasmus Prehn	Uddannelses- og forskningsudvalget	P	Politician
Signe K. Mortensen	Sjældne Diagnoser	P	Patient representative
Sonja Raabjerg Bang-Møller	Forældre til barn	P	Patient representative
Steen Werner Hansen	KRIS	S	Other
Stense Farholt	CSS Aarhus Universitets Hospital	S	Clinician/GP
Stephanie Jøker Nielsen	Sjældne Diagnoser	P	Patient representative
Stig Yndgaard	AUH + Region Midt	P	Public administration
Stine F. Mønster	Sjældne Diagnoser	P	Patient representative
Susan Fugger	Synscenter Refsnæs	P	Healthcare Professional

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Susanne Marquardsen	Porfyriforeningen i Danmark	P	Patient representative
Susanne E. Boonen	Uddannelsesansvarlig overlæge	P	Clinician/GP
Susanne Romlund	Sjældne Diagnoser	P	Patient representative
Svend G. Hartling	Region Hovedstaden	KS	Public administration
Sven Kreiborg	Professor, dr. odont., Ph.D.	P	Clinician/GP
Søren Brostrøm	Sundhedsstyrelsen	P	Public administration
Søren Lildal	Sjældne Diagnoser, Apert-foreningen	R	Patient representative
Søren Schytt	Synscenter Refsnæs	P	Healthcare Professional
Søs Marie Luise Bisgaard	Københavns Universitet	P	Clinician/GP
Tamara Steiner	Sundhedsstyrelsen	P	Public administration
Terkel Andersen	EURORDIS	S	Patient representative
Thomas Pihl	Region Hovedstaden	P	Public administration
Tina Buhl	Celgene	P	Industry
Toke Nahuel Pepe Grøn	KORA	P	Academic/ Researcher
Torben Kajberg	Danske Handicaporganisationer	P	Patient representative
Torben Schroeder	Sundhed.dk	P	Healthcare Professional
Trine Juul	Sjældne Diagnoser	P	Patient representative
Troels Staehelin Jensen	Det Sundhedsfagl. Forskningsråd	S	Academic/ Researcher
Uffe Koppelhus	Aalborg Universitet	P	Clinician/GP
Ulla Werlauff	RCfM	S	Healthcare Professional
Valentina Bottarelli	EURORDIS	P	Other