Abstract for 'The evaluation of the National Strategy for rare diseases'

The following evaluation presents a status of the implementation of both the recommendations from the national strategy from 2014 and the status evaluation from 2018, as well as providing a new overview on the matter. Moreover, this abstract provides an overview of each of the eight themes, which have been elaborated in Part 2 of the analysis.

This field of research is notably complex which manifests in a number of ways. In part, the prevalence of people in Denmark with rare diseases is so low that it is difficult to perform a national comparison. Furthermore, the recommendations are noticeably influenced by a high degree of specialised expertise, as well as individualised approaches to a variety of different treatment plans. This is in part due to the patients having such rare diseases that there are in fact so few in the world with the same diagnosis, hereby causing the practical experience to be exceptionally limited.

The evaluation has focused on assessing the implementation, effect and relevance of all 28 recommendations within the appointed eight themes. These eight themes were established based on a preliminary analysis (Cf. Appendix 3) of all recommendations presented by the national strategy from 2014 and the status evaluation from 2018.

The preliminary analysis found that the status evaluation from 2018 contains, to some degree, all approx. one hundred recommendations issued in 2014. Yet, the level of depiction varies, which is why the preliminary analysis elucidates and expands upon these themes, hereby enabling the entire period, from the release of the national strategy in 2014 to today, to be covered by the evaluation.

Theme 1: Rare patients in the hospital and municipal systems

Attending to people with rare diseases affects many different stakeholders within the healthcare system. At Rigshospitalet in the Capital Region and at Aarhus University Hospital in Central Jutland Region, centres for rare diseases have been established. In practice, these centres have close collaborations with several specialties, in particular the Clinical Genetics specialty and Paediatrics. Yet, as diagnosis and treatment often require a wide range of expertise, there is also collaboration with other specialties (Cf. Specialised hospital services – Principles of national planning in Denmark) as well departments which perform treatment at the basic hospital service level. Additionally, the centres collaborate with general practitioners as well as the municipality who fulfil needs within the following areas: children, family, health, employment, disability and care.

With regards to the treatment in the hospitals, the intention is that the centres act as an 'assembly mechanism' for the patients who do not naturally belong to other specialties (Cf. Specialised hospital services – Principles of national planning in

Denmark). However, this has proven to be difficult in practice, as it requires the knowledge of each individual doctor to identify and treat symptoms. This is in turn complicated by the fact that patients typically are not treated locally and challenged further by the fact that visitation guidelines have not been produced as per recommended, hereby exemplifying an unfulfilled potential.

A central focus in the strategy from 2018 is to ensure better transitions for people with rare diseases from child to adult. It is in this phase that a transition occurs from a collaboration between the centres for rare diseases and paediatrics, to a potential collaboration between the centres and a number of organ-specific 'adult specialties', where a patient often belongs to several. There have been initiatives at both centres to address this problem, yet it still appears to be an exceedingly difficult task.

Furthermore, extensive development has been made within clinical genetics. Moving forward, it could be beneficial to focus on employing the potential of the improved methods, which includes working systematically to evaluate patients without a definitive diagnosis. In addition, this development offers other opportunities for capturing and ensuring the establishment of good patient care.

Furthermore, it may be beneficial to ensure that municipalities are provided a sufficient basis for implementing holistically oriented and coherent patient care. It should also be noted that the terminology used in relation to the municipalities' function is less obligating in the evaluation from 2018 than the strategy from 2014.

Interventions for people with rare diseases is often complex and draws upon several different municipal initiatives. This means that people with rare diseases, and their families, often meet several different municipal authorities and practitioners, despite intentions to the contrary. There especially appears to be issues associated to the municipalities' interventions being granted based on a functionality assessment, rather than applying knowledge of the disease itself. This means that for this group of patients, there are several experiences of being offered interventions that are not targeted their needs. Thus, there is potential in directing the interventions by developing and maintaining frameworks for how to secure that the municipalities have the relevant knowledge; and how and when they should seek it out.

Theme 2: Sectorial transitions, collaboration and coordination

Much like the right intervention is crucial, it is also essential that there is collaboration and coordination across sectors. Since the status evaluation from 2018, work has begun on describing a generic patient care model. However, due to Covid-19, this has yet to be completed. This includes a number of relevant elements that relate to ensuring process coordination for this patient group, which often requires input from several different specialties.

Although there is collaboration about i.e., the exchange of patient-specific information in practice, there is still work to be done to integrate the work with rare diseases more strategically, e.g., through the health agreements. The evaluation indicates that this has only happened to a very limited extent. It is also essential to

continuously monitor the nature and processes of the collaboration, which has not yet occurred as intended.

Theme 3: Patient education, coping mechanisms and empowerment

Due to the rarity, the task of ensuring patient education with a focus on coping mechanisms and empowerment is particularly difficult for several reasons. Firstly, the rarity means that patients and relatives must have comprehensive knowledge about their disease, as they will often be met by both professionals and others in their network who have no knowledge of it. In addition, the rarity means that the human need to be among other like-minded individuals is often difficult, as there will rarely be someone with the same disease 'just around the corner'. Finally, it is also significant that the rarity often entails more complexity in the treatment, hereby placing greater demands on patients and relatives to be able to navigate a complex hospital system. Finding that the need for good patient support through patient education is still highly relevant.

The evaluation finds that patient associations have largely taken on the task of patient education and information and have come a long way with this. Yet challenges remain. The patient associations under Rare Diagnoses are very different and there are, due to the prevalence of the individual disease, considerable differences in the number of members and therefore resources. This creates a risk of bias with regards to the offers that are available. It is also clear that the patient associations are dependent on financial subsidies for their work, which is why the level of security surrounding grants for e.g., Helpline and further development of existing models is crucial for ensuring continued work in the area. Prioritization, range, and stability of offers are therefore still central.

Theme 4: International collaboration

The international collaboration has changed since 2014. The first recommendations contained a number of different elements, but since 2018 the focus has mainly been on establishing and participating in the European Reference Network (ERN). ERN is comprised of 24 networks which, among other things, works with different groups of rare diseases with the intention of exchanging knowledge and experiences across the EU's health services and health personnel. Denmark is now represented as a full member in 22 out of 24 networks and as an Affiliated Partners in two ERNs (ERN Cranio and Ern EpiCARE), which has been achieved based on extensive efforts. There is a recognition of the opportunities and potential of international cooperation among the regions, and the importance of employing and benefiting from international competences when initiatives for people with rare diseases are discussed and organised.

The evaluation finds that a focus on expanding and maintaining Danish participation in international networks could strengthen care for people with rare diseases in Denmark, including organising systematic knowledge sharing internally and across regions.

Theme 5: Education and competencies

The evaluation finds that the efforts to ensure training and competencies within the area has a very low degree of implementation overall. A possible explanation for this may be the difficult conditions for skills development during the covid-19 epidemic. Knowledge of rare diseases is therefore not currently included in the revision of the specialty education, no description has been made of what an expert education might look like, just as additional training of 'other health personnel' has taken place without any shared structure for this.

The low degree of implementation contrasts with the importance and relevance of having specialist staff who are qualified for the task; this includes specialist doctors, dietitians, therapists and the municipality's social and healthcare staff. Therefore, it is deemed relevant that moving forward, a clear plan is made for upskilling within this area, based both on practical learning and more traditional education.

Nevertheless, the applicability of the Lægehåndbogen's articles on rare diseases has been good. However, no further funding has yet been allocated towards this.

Theme 6: Registering, documenting and knowledge

The effort to establish a systematic and universal registration is underway, however it is also an area where there simultaneously has occurred extensive development. Therefore, a number of decisions about the direction and need for data are pending. These decisions act as a basis for agreement and from which a universal registration practice can be formed, constituting the possibilities offered by the international collaborations and coding systems as well as the electronic patient records. This is however also an area where special attention should be paid to the connection between the expected outcome and the resources required.

Furthermore, the analysis finds that, within the area of research, there has been a significant reduction in the focus of the recommendations from 2014 to 2018. The analysis also indicates that there are limited opportunities to prioritize resources towards it. Without a strategic focus on strengthening this research moving forward, it may prove difficult to realise the potential within this area.

Theme 7: Availability of treatment

The analysis has found that although the status evaluation from 2018 does not explicitly include the three themes from the strategy in 2014 on the development of medicine, participation in experimental treatment and access to research treatment abroad, there has still been extensive development within the area.

With the placement of assessments of new medicines in the Medicine Council, a system has been established and expertise gathered, which also includes the special conditions for rare diseases. The assessment process is shaped by a number of dilemmas, for example in relation to the uncertainty in assessments based on a precarious dataset, fused with high costs for new medicines for small patient groups.

The evaluation finds that the opportunity for patients to participate in experimental treatment is in practice limited. Yet, the relevance of the recommendation to ensure the possibility of treatment abroad has changed, as systems for this have been established, as well as with the establishment of European Reference Networks, efforts are being made towards bringing the expertise closer to the patient via knowledge sharing.

Theme 8: Implementation

The last theme considers how the implementation process for the strategy maintains focus during the period. It is therefore a significant observation, that a mechanism for follow-up is essential. The annual status meetings and status notes have proven useful, especially as rare diseases is an area under constant development.

In the following report, the status of and experiences with the work with the national strategy for rare diseases are reviewed.