Assessing the potential of a European Reference Network for Neuromuscular Diseases: outcomes of an ENMC workshop

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Introduction

Centres of Expertise (CE) and European Reference Networks (ERN) for rare diseases are priorities of the EU Health policy. The Cross-Border Healthcare Directive (EC, 2011) is intended to bridge the gap, providing the framework for Reference Centres identified by Member States to function as CE (EUCERD, 2011) in ERNs, delivering equal access to the most up-to-date care and advice to all individuals, regardless of where they live.

As with other rare diseases, the rarity and diversity of Neuromuscular Diseases (NMD) pose specific challenges for healthcare provision and research as well as for the development and marketing of therapies that could be fruitfully addressed through the existence of an ERN.

Background

NMD are complex, progressive, multi-system disorders, mostly of genetic origin that can present from childhood to adulthood. Around 500,000–600,000 patients suffer from rare Neurodegenerative diseases /Neuromuscular diseases in Europe [NEUROMICS (Integrated European Project on Omics Research of rare Neuromuscular and Neurodegenerative Diseases) (http://rd-neuromics.eu)]. NMD most often require multidisciplinary care, frequently provided in a scattered and uncoordinated way.

The general concept and method of implementing ERNs has been developed by the European Commission. They will be built within the scope of the Cross-Border Healthcare Directive, as a result of Member States’ cooperation, and regulated by the Delegated and Implementing Acts. The vision entails European Networks of healthcare providers sharing highly specialised expertise and covering a great number of low-prevalence and complex diseases. Member States’ role will be to establish the connections between healthcare providers and to foster their participation in ERNs. It is also contemplated that healthcare providers/centres of reference/centres of expertise may voluntarily apply for participation in an ERN if they meet the requirement and conditions established in paragraph 4 of article 12 of the Cross-Border Healthcare Directive (converted to measures through the Delegated and Implementing Acts).

Aims of the workshop

1. exchange of knowledge and expertise in processes for the delivery of neuromuscular care
2. assessment of existing resources both at national and international level
3. identification of gaps which need to be addressed
4. decide on a guideline document for the implementation of ERN/ERNs in the neuromuscular field

Discussion

Current status of specialized neuromuscular centres in Europe

- A few countries (ex: Belgium and France) have officially appointed CE. In several other countries, patients’ organizations as well as academic institutions have had a crucial role in recognizing the CE.
- Most CE are involved in the care of all groups of NMD adding to that a super-specialized area of expertise/research.
- Transition from paediatrics to adult clinics, although contemplated in NP, is not universally implemented.
- Most countries have registries/networks in place for NMD and there is a strong association between national networks and the TREAT-NMD Alliance

Experience in the neuromuscular field on networking activities and Biobanks:

- European Neuromuscular Centre (ENMC): It is a non-profitable organization, mainly founded trough patients organizations. It’s main aim is to encourage and facilitate collaborative research in Europe in the area of NMD through small workshops that function as a starting point for new consortia, a breeding ground and a facilitator in topics such as diagnostic criteria, characterization of disease genes, clinical trials protocols and practical care standards.

- TREAT-NMD Alliance: It is a network for the neuromuscular field designed to provide an infrastructure that ensures the most promising new therapies are reaching patients as quickly as possible. For example, the “Care and Trial Site Registry” (CTSR) (http://www.treat-nmd.eu/industry/trial-sites-and-patients/ctsr/) is one of the keys dimensions of TREAT-NMD, enjoying multi-stakeholder involvement and functioning as a platform for trial readiness.

- Biobanks: National/Local biobanks are in most cases part of the Telethon Network of Genetic Biobanks (TNGB) and/or the EuroBioBank (EBB)

- RD-Connect: aims at the development of an integrated platform connecting databases, registries, biobanks and clinical bioinformatics for rare disease research.

The role of the learned societies in an ERN:

- Scientific societies like ENS, EFNS, EPNS or PNS could play a major role in an ERN through teaching and training. Their current resources, such as e-learning programs, teaching courses and guidelines should be integrated into a future ERN.

- The scientific societies can also contribute to the establishment of a European Neuromuscular curriculum and to the structure of the European Board Examination.

- e-health: E-learning programmes are already in place through the scientific societies and can be adjusted to different needs.

- Other resources in the area of e-health are being assembled through projects like the cross border EU project SIGN which is establishing a telegenetics system in order to perform genetic counselling and clinical genetics consultations via the web.

Conclusions

- There is a high level of networking experience in the NMD field, which would be a strong starting position for applying for ERN status.

- The added value of implementing CEs, registries and networking can be demonstrated in different countries.

- The existing structures in the NM field are already fulfilling most of the criteria established and are in a position to apply for an ERN.

- The scientific societies have a relevant role in the areas of training and education as well as in the development of standards of care and best practice guidelines.

- The current involvement of patients and patient representatives is well established in the different projects either at European or National levels.

- An ERN in the neuromuscular field should encompass all the different groups of NMD, to avoid fragmentation of resources and the exclusion of patients with ultra-rare diseases.

- A future ERN could include an extended number of healthcare providers across Europe.

- Funding, harmonization of concepts and identification of healthcare providers in different countries were the main problems identified.

List of participants

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