

Introduction

Although international consensus guidelines exist for optimal multi-disciplinary care for Duchenne muscular dystrophy, their level of implementation and impact on health outcomes remains unclear.

The EU-funded CARE-NMD project analysed the current care situation and health impact of patients with DMD in 7 European countries (Bulgaria (BG), the Czech Republic (CZ), Denmark (DK), Germany (DE), Hungary (HU), Poland (PL) and the United Kingdom (UK)).

Patient registries offer a valuable approach to engaging with the patient community, both to disseminate information and to survey their experiences

Registries in clinical trials

Registries permit the identification of a patient population with a precise genetic diagnosis, and are thus essential to the development of novel, mutation-specific therapeutic approaches such as exon-skipping. A core driving factor in their development has often therefore been clinical trial readiness: e.g. determining trial viability for a specific genetic mutation

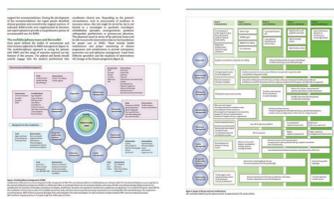


However, as registries also enable contact with a patient population, they offer research opportunities outside the clinical trial domain. These include surveying availability of high-quality care and quality of life issues. Furthermore, registries permit the distribution of care information aimed at that particular audience. CARE-NMD has utilised patient registries in both of these contexts

Registries to promote best-practice care

CARE-NMD has promoted knowledge of best-practice care via the translation and dissemination of the Family Guide to the DMD Care Standards.

Multidisciplinary, international consensus standards for best-practice DMD care were published in *The Lancet Neurology* in early 2010.



TREAT-NMD, MDA, PPMD, and UPPMD worked together to produce a Family Guide to the Care Standards. It is written in language that is more accessible to those without a medical background than the original scientific paper.

It has been translated by local patient organisations and experts into Bulgarian, Chinese, Czech, Danish, Dutch, English, Farsi, Finnish, French, German, Greek, Hebrew, Hungarian, Japanese, Lithuanian, Norwegian, Polish, Romanian, Russian, Serbian, Slovenian, Spanish, and Turkish.

National patient registries are a very important way of distributing information such as the Family Guides.



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Surveying care and quality of life in people living with DMD

CARE-NMD has conducted the largest ever survey of care and quality of life in patients and families living with DMD.

1,677 questionnaires were distributed via the National Patient Registries in each country. For accessibility, these were in the local language, in both paper and online versions.

Questions were asked about the care received by each family via a custom questionnaire, and about their perceptions of quality of life using standardised instruments such as PedsQL, WHO-QOL, and SF-36.

Data collection began in September 2011, and was completed in April 2012.

As different age groups of respondents required different versions of the questionnaires, distribution had to be stratified by age. However, respondent anonymity also needed to be preserved. Using the National Patient Registries to distribute the questionnaire allowed both conditions to be satisfied.

Each registry provided the CARE-NMD project leaders with the number of patients in each age group, and each were supplied with a sufficient number of unique IDs for their patient cohort.

The National Registries distributed the questionnaires, but when these were returned to the CARE-NMD project leaders, only the unique IDs were included - no personally identifiable data was provided from the Registries to the project.

1,180 questionnaires were received, with 109 of these not being included in the evaluation.

Reasons for non-evaluation include:

- Duplicate questionnaires
- Empty questionnaires
- Respondents identified by the national registry as BMD
- Clinical data clearly indicating a non-DMD phenotype

This left 1,071 questionnaires to be evaluated after data cleansing, i.e. 63.9% of those sent out.

This response rate is higher than anticipated given the length of the questionnaire. We believe this is in part due to a genuine engagement by those registered in National Registries, and their desire to help improve care for DMD.

The use of patient registries for this purpose has been extended beyond the project: the Japanese National Patient Registry has also distributed the CARE-NMD questionnaire in the same manner, building on the experience of this project.

Partners



CARE-NMD is led by Dr Janbernd Kirschner at the University Clinic Freiburg.

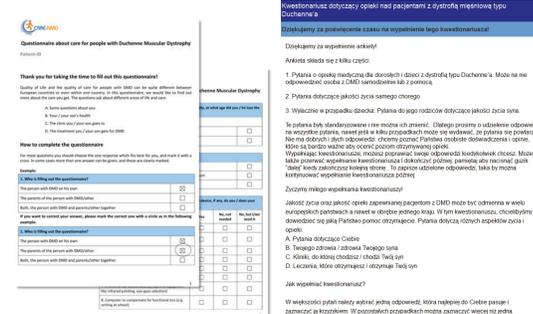


The UK academic partners are Prof Kate Bushby and Prof Hanns Lochmüller at Newcastle University.



Both Action Duchenne and the Muscular Dystrophy Campaign are UK partners, and have been actively involved in the project.

Other funded partners include the Bulgarian Neuromuscular Disorders Society (BGNMDS), Brno University in the Czech Republic, the Danish Rehabilitation Centre for Muscle Diseases, the Hungarian National Institute for Environmental Health, and Warsaw Medical University in Poland.



National Patient Registries		CARE-NMD Anonymised Data	
UK National Registry	Unique ID Name UK1001 Joe Bloggs UK1002 ...	Unique ID Questionnaire Question 1 Questionnaire Question 2 UK1001 A ... UK1002 B ...	
Danish National Registry	Unique ID Name DA2001 Hr. Jensen DA3002 ...	DA2001 A ... DA3002 B ... DE10001 B ... DE4001 B ...	
German National Registry	Unique ID Name DE1001 M Mustermann DE4001	

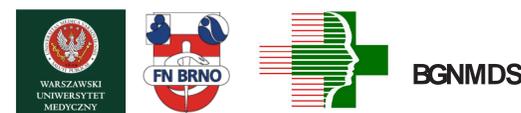
Diagram 1: Personally identifiable information, such as the names and contact details of patients, is retained by the National Patient Registries (left). Only anonymised data collected by the questionnaires is passed to the CARE-NMD project leaders (right). The only link between the two sets of data is the Unique ID.

Country	Sent	Evaluated	% Evaluated
Bulgaria	73	40	54.8%
Czech Republic	191	90	47.1%
Denmark	131	90	68.7%
Germany	545	424	77.8%
Hungary	70	57	81.4%
Poland	246	142	57.7%
United Kingdom	421	228	54.2%
Overall	1677	1071	63.9%

Table 1: sent and evaluated questionnaires by country

The use of Patient Registries to identify patient cohorts for clinical trials is well established.

However, our experience in the CARE-NMD project demonstrates that Patient Registries are also very valuable tools for the dissemination of disease-specific information to rare disease patients, and to discover other factors influencing care and quality of life in rare diseases.



Unfunded partners include clinicians, universities, and patient organisations across more than 20 countries. For a complete list of partners, see the CARE-NMD website.