Affiliation to the Treat-NMD Consortium Using an Agreed Minimum Dataset Allows Small Registries and Large Registries to Collaborate Together

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Introduction

Treat-NMD is an international collaboration aimed at bringing treatments to people with neuromuscular diseases. The TREAT-NMD Myotonic Dystrophy Working Group was established in 2017 specifically to develop collaborations amongst TREAT-NMD DM registries.

Aim

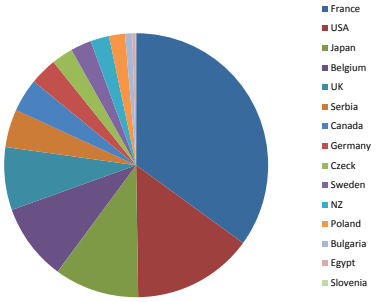
To further develop the TREAT-NMD Global Registry Network by highlighting the registries that are affiliated and full members of TREAT-NMD and to explore the strengths and weaknesses of the collaboration.

Methods

A postal survey was undertaken of the 24 Myotonic Dystrophy registries who have expressed interest in the TREAT-NMD network. The survey included questions about total numbers and demographics of participants. We were also interested in what data were collected, particularly with respect to an agreed minimum dataset and how children with myotonic dystrophy were classified, whether data about patients with DM2 as well as DM1 were included and whether 'at risk' participants were enrolled. The registries' formal relationship in terms of whether the registry has a formal Confidential Disclosure Agreement was gauged.

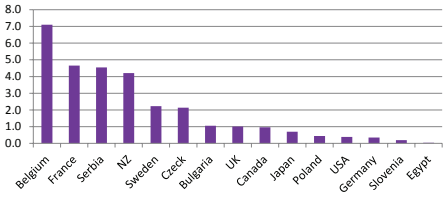
Results

Total patients per registry. Grand Total = 8603



Responses were received from 14 of the 25 registries. Total Enrolments ranged from just 4 patients to over 3000 in the French registry. The majority of registries only enrol those who are affected by the disease. The large majority of registries update their patient details annually.

Registry Enrolments / 100 000 national inhabitants



The majority of registries claim to enrol patients from their whole nation. Considering rates of enrolment per head of population, the Belgian registry has the highest rate and may be getting close to full population registration.

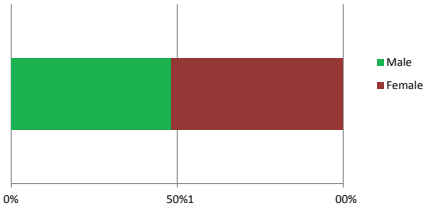
Rates may be influenced by ethnic differences with international studies varying in their estimates from 0.5 / 100 000 to 18.1 / 100 000[1].

Some low enrolment rates represent newer registries or registries aimed at a paediatric or other specific population.

Registry Demographics and Categorisation of early onset disease

Only two registries formally subdivided early onset myotonic dystrophy into Congenital, Childhood and Juvenile onset and were able to provide precise definitions of these subgroups.

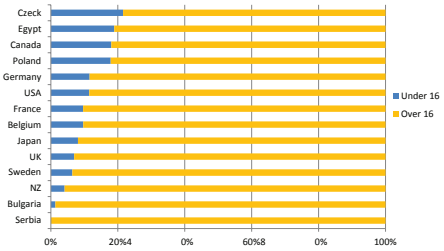
Sex Distribution



Overall there was a slight predominance of female enrolments in the registries.

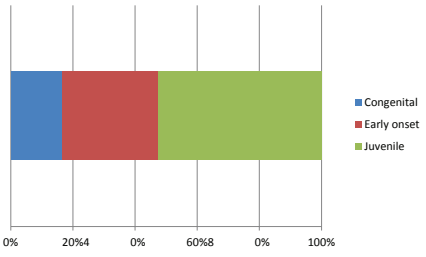
Given that myotonic dystrophy may be more difficult to recognise in women as the classical features are less prominent[2] this may represent a tendency for women to be happier to volunteer for research[3].

Proportion of DM1 Registry under the age of 16



The proportion of DM1 patients under 16 years of age is likely to reflect enrolment criteria (e.g Slovenia is a paediatric registry), or the underlying population structure.

Subdivisions of patients < 16 years

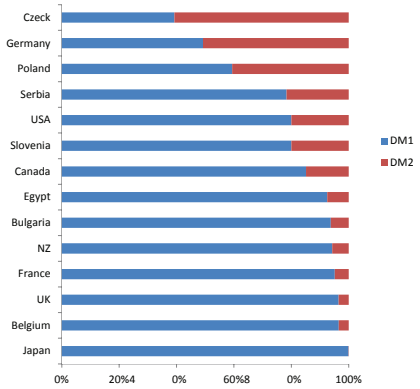


Data presented is from the French registry.

For some years now different syndromes with different prognoses have been identified between congenital, childhood onset and early adult onset myotonic dystrophy[5]. However, only 2 registries formally subdivided early onset myotonic dystrophy into Congenital, Childhood and juvenile onset and were able to provide precise definitions of these subgroups. Other groups appeared to group all patients with early onset together or identified just a specific Congenital group with age of onset within the first month of life.

DM2

Ratio of DM1 patients to DM2 patients in each registry



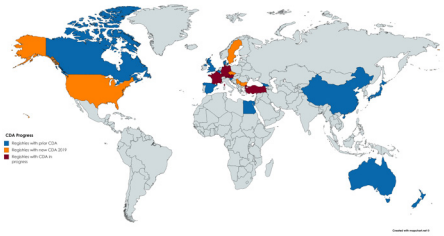
Fourteen of the 15 registries have enrolled patients with DM2 as well as DM1. A total of 1478 patients with DM2 have been enrolled.

As expected[4], registries from central Europe had higher proportions of DM2 patients in their registries, which is likely to represent underlying disease frequency differences.

This disease appears to be much less frequent in eastern Asia.

Just nine out of the 1478 patients with DM2 were aged < 16.

Relationship with TREAT-NMD



This Confidentiality Agreement (CDA) allows Treat-NMD to circulate to individual registries the details of Pharma and other research enquiries.

Signing up for the CDA is required for full membership but Treat-NMD does still support other Registries without these CDAs.

For small countries it is unlikely that they will be given the opportunity to take part in trials without joining forces with an organisation which can then act as a beacon attracting research enquiries.

In 2018 and 2019 the Myotonic Dystrophy working group has held meetings at three international meetings emphasising the importance of the CDA. With the help of the TREAT-NMD secretariat seven new registries have signed CDAs in 2019

All but two registries collect an agreed Core Dataset allowing sharing of data and joint publications[6].

Conclusion

- The TREAT-NMD Collaboration works because of an agreed minimum dataset and allows registries from all around the world, even from small countries, to combine their data.
- Consideration should be given to introducing specific criteria for early onset disease such as those used by the French registry as a further minimum dataset requirement.
- Registries benefit most from the TREAT-NMD collaboration when they have a CDA in place as this allows registries to benefit from a centralised enquiry system. Repeated face-to-face meetings has markedly increased understanding of this and sign up to CDAs in 2019.

References

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