Academic productivity from rare neuromuscular disease registries: A systematic review

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BACKGROUND

The goal of the TREAT-NMD network is to accelerate pre-clinical and clinical research and clinical innovation in the field of neuromuscular disease with the aim of improving patient care and treatment options. One of TREATNMD's primary focus areas is supporting a network of NM disease registries. The purpose of this study was to determine the academic output from these registries and further understand the registry factors that contribute to successful academic productivity.

OBJECTIVES

- The **main** objectives of this systematic review were to assess:

- 1) Asses the current academic output from the TREAT-NMD neuromuscular disease registries
- 2) Assess the types of studies published using data from this network

METHODS

- A systematic search was conducted in **May 2019:**

→ **Identify publications that use the registry data and focuses on a neuromuscular disease or a series of neuromuscular diseases**

→ **Results were then sub-grouped into four different categories: Profile or methodologic, clinical research/guidelines, epidemiologic and basic science**

→ **Results were sub-grouped into type of academic product: Abstract or manuscript**

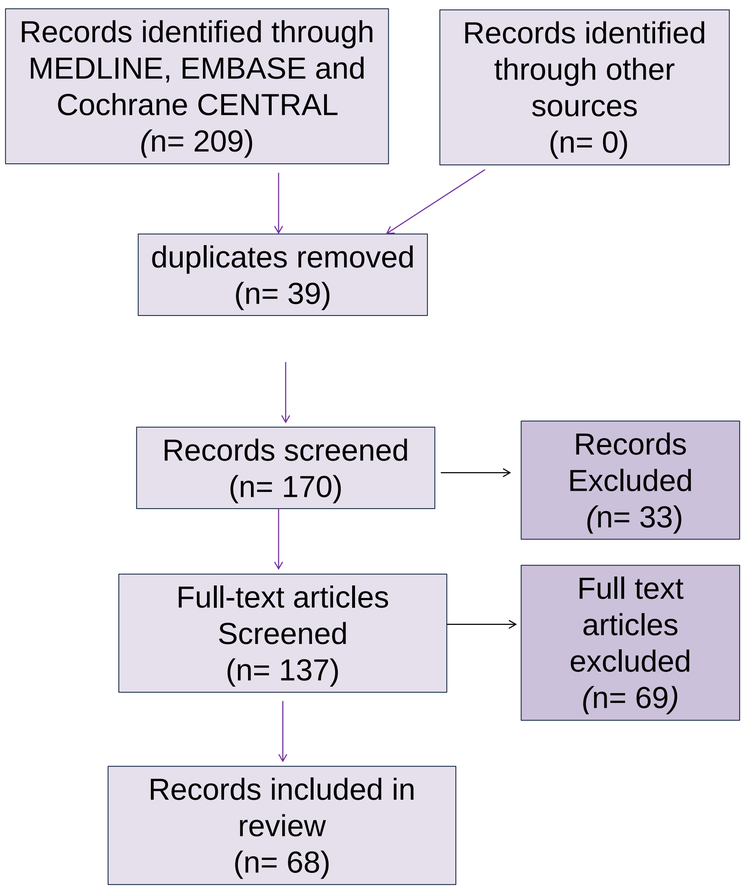
METHODS

Table 1: Electronic Databases used in Systematic Review

Databases
MEDLINE
EMBASE
Cochrane CENTRAL

- 3-stage process
- Two authors independently screened records against the inclusion criteria

RESULTS



The search yielded a total of 68 included studies:
basic science = 0
epidemiology = 5
clinical research/guidelines = 10
profile/methodologic papers = 53

The type of academic product included:
Abstracts: 28
Manuscripts: 40

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Table 2. Included Records (n = 68)

Type of study (N)	Abstracts	Manuscripts
Basic science (n= 0)		
Clinical Research/ Guidelines (n =10)	1) First drug registry in Duchenne muscular dystrophy (DMD) to assess Translarna (Ataluren) use, safety, and effectiveness in routine clinical practice 2) How reference networks develop, implement, and monitor guidelines 3) Clinical research of becker muscular research using nationwide patient registry 4) Care provision for adults with Duchenne muscular dystrophy in the UK: Compliance with international consensus care guidelines 5) Health related quality of life in European adults with DMD: Results from the Care-NMD-project 6) Molecular profile of 307 Portuguese patients with dystrophinopathy, including 39 new variants	1) Corticosteroids in Duchenne muscular dystrophy: Major variations in practice 2) Compliance to care guidelines for duchenne muscular dystrophy 3) Psychometric properties of the Zarit Caregiver Burden Interview administered to caregivers to patients with Duchenne muscular dystrophy: a Rasch analysis 4) Mapping the differences in care for 5,000 Spinal Muscular Atrophy patients, a survey of 24 national registries in North America, Australasia and Europe
Epidemiology (n= 5)	1) The burden of Duchenne muscular dystrophy: An international, cross-sectional study 2) Estimates of spinal muscular atrophy (SMA): Results from the TREAT NMD research program 3) European Cross-Sectional Survey of Current Care Practices for Duchenne Muscular Dystrophy Reveals Regional and Age-Dependent Differences	1) Clinical Outcomes in Duchenne Muscular Dystrophy: A Study of 5345 Patients from the TREAT-NMD DMD Global Database 2) A multi-source approach to determine SMA incidence and research ready population
Profile/ Methodologic (n = 53)	1) TREAT-NMD (translational research in Europe, assessment and treatment for neuromuscular disorders) 2) The national Dutch dystrophinopathy patient registry 3) Advancing diagnosis, care and treatment for people with neuromuscular diseases around the world: A network of excellence to catalyse research infrastructure globally 4) Preliminary data of national romanian registry of DMD patients 5) The UK Myotonic Dystrophy Patient Registry: A key tool in the facilitation of clinical research 6) EURO-NMD, a reference network for neuromuscular diseases 7) Australasian neuromuscular disease registry 8) Promoting rare disease policies across Europe 9) DMD HUB: Expanding clinical trial capacity for Duchenne muscular dystrophy in the UK 10) GNE myopathy patient registry 11) Current status of dystrophinopathy national registry in Japan 12) Current status of national neuromuscular patient registries in japan: Remudy 13) Infrastructure for new drug development to treat muscular dystrophy - Current status of patient registration in Japan: REMUDY 14) DMD/BMD patient registry in Japan: Remudy 15) Treat-NMD: Advancing diagnosis, treatment and care in neuromuscular rare diseases 16) The UK myotonic dystrophy patient registry: A key tool in the facilitation of clinical research 17) New patient registries for Myotonic dystrophy and Facioscapulohumeral muscular dystrophy in the United Kingdom 18) The New Zealand neuromuscular disease registry-A review of diagnoses confirmed by molecular test 19) The impact of 25 years ENMC workshops	1) National registry system for Duchenne Muscular Dystrophy and Spinal Muscular Atrophy 2) Spinal muscular atrophy national registry of Turkey 3) The UK myotonic dystrophy patient registry 4) The myotonic dystrophy registry of Japan: Current status and analysis for clinical research 5) UK Facioscapulohumeral Muscular Dystrophy (FSHD) patient registry 6) UK patient registry for facioscapulohumeral muscular dystrophy (FSHD) 7) A comprehensive database of duchenne and becker muscular dystrophy patients in children's hospital of fudan university 8) Meeting report of the "Regulatory Exchange Matters" session at the 5th International TREAT-NMD Conference:Lessons in communication: How an early dialogue between patients, regulators and academics can further therapy development for neuromuscular disorders 9) The Italian neuromuscular registry: A coordinated platform where patient organizations and clinicians collaborate for data collection and multiple usage 10) TREAT-NMD A European network for neuromuscular diseases 11) TREAT-NMD A European network for neuromuscular diseases 12) The TREAT-NMD duchenne muscular dystrophy registries: Conception, design, and utilization by industry and academia 13) The TREAT-NMD DMD global database: Analysis of more than 7,000 duchenne muscular dystrophy mutations 14) Collaborating to bring new therapies to the patient--the TREAT-NMD model 15) The creation of a network after an international conference 16) Remudy, Japanese national registry for neuromuscular diseases 17) The infrastructure for the clinical research of muscular dystrophies: Remudy and MDCTN 18) Remudy 19) Translational Research in Europe for the Assessment and Treatment for Neuromuscular Disorders (TREAT-NMD) 20) The TREAT-NMD patient registries for spinal muscular atrophy and Duchenne muscular dystrophy 21) Registry of muscular dystrophy (Remudy). Construction of the patient self-report registry and collaboration with overseas network 22) Infrastructure for new drug development to treat muscular dystrophy: current status of patient registration (remudy) 23) Characteristics of Japanese Duchenne and Becker muscular dystrophy patients in a novel Japanese national registry of muscular dystrophy (Remudy) 24) REMUDY-DMD/BMD patient registry in Japan 25) New horizons in the Duchenne Connect registry 26) The prospective German Charcot-Mary-Tooth patient registry 27) CARE-NMD: The role of patient registries in an international study of care in Duchenne muscular dystrophy 28) The TREAT-NMD care and trial site registry: An online registry to facilitate clinical research for neuromuscular diseases 29) THE New Zealand neuromuscular disease registry-A review of diagnoses confirmed by molecular test 30) The New Zealand Neuromuscular Disease Registry: Rate of diagnoses confirmed by molecular testing 31) National registry of Japanese dystrophinopathy patients: Remudy 32) Natural history, trial readiness and gene discovery: Advances in patient registries for neuromuscular disease 33) Patient Registries and Trial Readiness in Myotonic Dystrophy - TREAT-NMD/Marigold International Workshop Report 34) Eight years after an international workshop on myotonic dystrophy patient registries: Case study of a global collaboration for a rare disease.

CONCLUSIONS

- Results suggest the publications from the TREAT-NMD registry network are mainly descriptive or methodologic
- Papers may not have been discovered if the manuscripts did not mention TREATNMD or TGDOC in the searchable fields
- Two additional systematic reviews with a broader scope will be conducted to help identify the type of academic output from these neuromuscular disease registries