#RWD131

A nationwide feasibility study and study concept to assess the burden of Duchenne Muscular Dystrophy (DMD) in Finland

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BACKGROUND

- Duchenne muscular dystrophy (DMD) is a rare, severe, progressive muscle disease.[1]
- DMD is associated with a substantial burden to patients, their families, and societies that increases dramatically with disease progression and severity.[2]
- Systematic characterization of the disease epidemiology and understanding of the total burden of DMD in the Finnish real-world (RW) setting are lacking.
- Finland offers equal access to tax-funded healthcare to its 5.5 million inhabitants. Detailed individual-level data recorded in social and healthcare registers provide a unique opportunity for nationwide burden of disease (BoD) studies.
- This study had two objectives:
  1. To verify whether the ICD-10 code G71.06 can be used to reliably identify the patients with DMD; and to subsequently assess the epidemiology of DMD in Finland (a feasibility study).
  2. To design a RW study concept for a comprehensive description of the epidemiology, patient characteristics, treatment practices, and the burden of DMD in Finland.

METHODS

Feasibility study:

- The incidence, prevalence, treatment management, and healthcare resource use (HCRU) of DMD patients in Finland were evaluated in a nationwide feasibility study.
- The number of unique patients and all visits with the ICD-10 code G71.06 (DMD) between 2011-2020 were extracted in aggregate from the health records of the National Institute for Health and Welfare.

Real-world study concept:

- A study concept for the comprehensive characterization of the epidemiology and burden of DMD in Finland was designed based on feasibility results.
- The individual-level data will be retrospectively collected from the Finnish national social and healthcare registers and linked using personal identification numbers. In addition, unstructured data from the electronic health records (EHRs) will be collected manually at Helsinki University Hospital (HUS) in order to collect information on the mutations in the Dystrophin gene, and examine disease progression and functional ability of the patients.

RESULTS

Feasibility study:

- The feasibility study showed that there were approximately 120 patients with DMD in Finland per year (Figure 1A). On average, 5.3 (2–7) new cases were diagnosed annually (Figure 1B).
- The mean age of patients at the time of DMD diagnosis was 5.5 (range: 0–31) years.
- Patients with DMD had an annual average of 1.1 (0.5–1.4) secondary care inpatient periods, 7.0 (3.8–10.7) secondary care visits, and 0.5 (0.2–1.0) primary care contacts (including visits, inpatient periods, and other contacts).

Prevalence and incidence of DMD in Finland

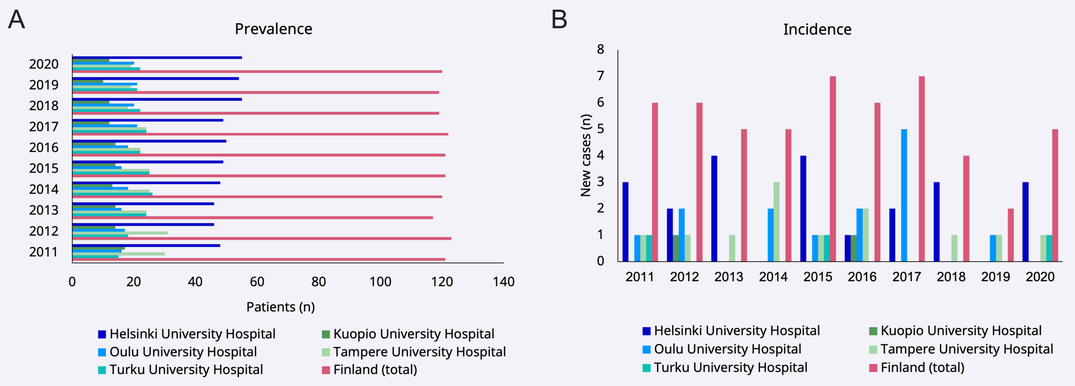

Figure 1. (A) Prevalence and (B) incidence of DMD in Finland. Prevalence is defined by the number of unique patients with a G71.06 diagnosis recorded at healthcare visits during the year. Incidence is defined by the number of new unique G71.06 cases per year. The catchment areas in the study are indicated by the names of the respective university hospitals.

Patient age at the time of DMD diagnosis (G71.06)

	Mean age of patients (min, max)									
Region	**2011**	**2012**	**2013**	**2014**	**2015**	**2016**	**2017**	**2018**	**2019**	**2020**
Helsinki University Hospital	6.0 (4.0, 9.0)	4.0 (3.0, 5.0)	3.8 (1.0, 6.0)	.	3.8 (2.0, 5.0)	4.0 (4.0, 4.0)	12.5 (8.0, 17.0)	5.0 (4.0, 6.0)	.	3.3 (2.0, 5.0)
Kuopio University Hospital	.	30.0 (30.0, 30.0)	.	.	.	4.0 (4.0, 4.0)	.	.	.	.
Oulu University Hospital	4.0 (4.0, 4.0)	2.0 (0.0, 4.0)	.	11.3 (6.0, 14.0)	6.0 (6.0, 6.0)	2.5 (2.0, 3.0)	8.6 (1.0, 31.0)	.	4.0 (4.0, 4.0)	.
Tampere University Hospital	2.0 (2.0, 2.0)	1.0 (1.0, 1.0)	5.0 (5.0, 5.0)	7.0 (7.0, 7.0)	4.0 (4.0, 4.0)	5.5 (4.0, 7.0)	.	1.0 (1.0, 1.0)	5.0 (5.0, 5.0)	9.0 (9.0, 9.0)
Turku University Hospital	4.0 (4.0, 4.0)	.	.	.	0.0 (0, 0)	.	.	.	.	1.0 (1.0, 1.0)
Finland (in total)	4.7 (2.0, 9.0)	7.2 (0.0, 30.0)	4.0 (1.0, 6.0)	9.2 (6.0, 14.0)	3.6 (0.0, 6.0)	4.0 (2.0, 7.0)	9.7 (1.0, 31.0)	4.0 (1.0, 6.0)	4.5 (4.0, 5.0)	4.0 (1.0, 9.0)

The catchment areas in the study are indicated by the names of the respective university hospitals.

Healthcare resource utilization among patients with DMD in Finland

	Year (total number of patients)										
Resource	**2011 (121)**	**2012 (123)**	**2013 (117)**	**2014 (120)**	**2015 (121)**	**2016 (121)**	**2017 (122)**	**2018 (119)**	**2019 (119)**	**2020 (120)**	**Average**
Per year in total, n											
Primary care contacts, n	37	40	58	51	35	20	63	121	77	56	55.8
Primary care visits, n	29	37	53	41	35	18	59	105	61	37	47.5
Primary care contacts other than visits, n	8	3	5	10	0	2	4	16	16	19	8.3
Secondary care contacts, n	679	626	772	865	958	1012	1076	1121	1222	1346	967.7
Secondary care visits, n	504	473	640	722	822	871	939	1000	1134	1282	838.7
Secondary care inpatient periods, n	175	153	132	143	136	141	137	121	88	64	129.0
Per year per patient, n											
Primary care contacts, n	0.3	0.3	0.5	0.4	0.3	0.2	0.5	1.0	0.6	0.5	0.5
Primary care visits, n	0.2	0.3	0.5	0.3	0.3	0.1	0.5	0.9	0.5	0.3	0.4
Primary care contacts other than visits, n	0.1	0.0	0.0	0.1	0.0	0.0	0.0	0.1	0.1	0.2	0.1
Secondary care contacts, n	5.6	5.1	6.6	7.2	7.9	8.4	8.8	9.4	10.3	11.2	8.1
Secondary care visits, n	4.2	3.8	5.5	6.0	6.8	7.2	7.7	8.4	9.5	10.7	7.0
Secondary care inpatient periods, n	1.4	1.2	1.1	1.2	1.1	1.2	1.1	1.0	0.7	0.5	1.1

Real-world study concept:

- The RW study will be based on individual-level data for patients and their caregivers in the Finnish social and healthcare registers from 1996 to 2022 (Figure 2).
- The study protocol and statistical analysis plan (SAP) have been prepared.
- The aims of the RW study will be to examine epidemiology, clinical characteristics, treatments and treatment outcomes, healthcare and social care resource utilization, and associated direct and indirect costs of DMD patients over the course of the disease.
- The Finnish Social and Health Data Permit Authority Findata has granted a permit for data collection and linkage in September 2023.
- All analyses will be conducted on individual-level pseudonymized data, and in a secure operating environment provided by Findata. Only aggregate level results will be extracted from the analysis environment, ensuring the anonymity of the patients.

Registries for individual-level data collection

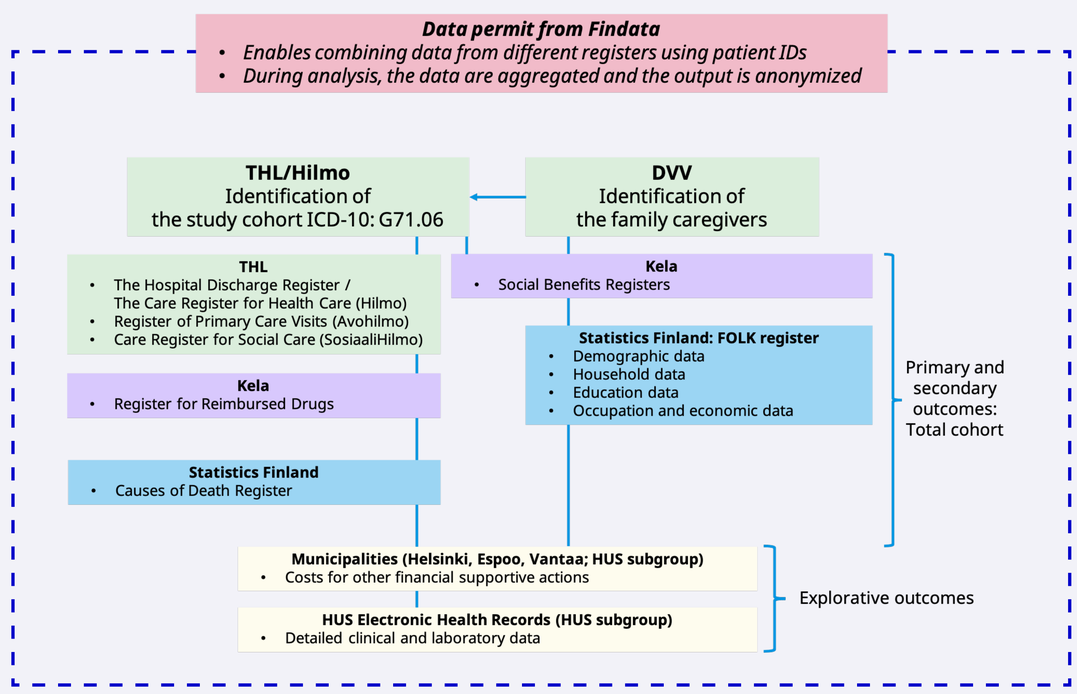

Figure 2. Overview of study registries by outcomes. Avohilmo, Register of Primary Care Visits; DVV, Digital and Population Data Services Agency; Hilmo, The Care Register for Health Care; HUS, Helsinki University Hospital; ICD-10, International Classification of Diseases, 10th revision; Kela, The Social Insurance Institution of Finland; SosiaaliHilmo, Care Register for Social Care; THL, The Finnish Institute for Health and Welfare.

CONCLUSIONS

- Based on the review of the feasibility results by expert clinicians, the ICD-10 code can be used to reliably identify DMD patients in the health records.
- A RW study is a necessity in order to address clinical characteristics, disease progression, and management of DMD patients and total BoD.
- Based on the RW study concept developed here, it is possible to obtain an unparalleled, comprehensive nationwide dataset on DMD management and BoD and thus, obtain information that cannot be gained from the feasibility or aggregated data.

REFERENCES

1. Mercuri E. & Muntoni F. Muscular dystrophies. *Lancet* 2013;381(9869):845–860.
2. Ryder S., *et al*. The burden, epidemiology, costs and treatment for Duchenne muscular dystrophy: an evidence review. *Orphanet Journal of Rare Diseases* 2017;12(1):79.

FINANCIAL DISCLOSURE

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