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Original article

Epidemiology of childhood neuromuscular diseases in two hospital districts in Finland

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1. Intro

Neuromuscular diseases are a group of rare, often inherited diseases with varying effect on muscular functioning and daily activities. They can be roughly grouped into four categories based on which part of the motor unit is affected: the anterior horn cells of the spinal cord, peripheral nerve, nerve-muscle junction or muscle fiber.

There are few existing epidemiologic studies that include all neuromuscular diseases in children. In previous studies, the overall prevalence of childhood neuromuscular diseases has been estimated to be 21.4–63.1 $\times 10^{-5}$ [1–5]. There is variation between studies as to which diseases are included, and many studies focus on only one specific neuromuscular disorder. Further, especially for the X-linked disorder of Duchenne muscular dystrophy (DMD), comparison between studies is difficult, because some authors use the whole population as the study population while others only include males or males of a certain age [6, 7]. Few studies have been conducted using all publicly available patient records of a certain geographical region. There is currently no published data concerning the epidemiology of childhood neuromuscular diseases in Finland.

There is a need for more accurate data on the epidemiology of pediatric neuromuscular disorders to help plan healthcare services. With advanced genetic diagnostic opportunities and emerging new treatments, the topic is very relevant. The purpose of this study was to estimate overall and specific prevalence and cumulative incidence for pediatric neuromuscular diseases in two hospital districts in Finland.

2. Materials and METHODS

2.1. Study population

A retrospective registry-based study in the Pirkanmaa and Päijät-Häme Hospital Districts (Fig. 1) was carried out. The area covers 22 491,24 km^2, which is 6.6 % of the total area of Finland. On the prevalence day (December 31, 2018), the entire population of the area was 724 727, which is 13.1 % of the population in Finland. The population of individuals under 17 years old was 128 358 (65 592 males), accounting for 12.8 % of the total population in this age group. 195 982 (100 284 males) children were born in the study area between 1992 and 2018. Both hospitals are the only hospitals with public pediatric health care services in their geographical area.

2.2. Procedures

The study aimed to identify all under 17-year-old individuals with a neuromuscular disorder living in the study areas on December 31, 2018 and all under 17-year-old individuals who received a neuromuscular diagnosis between 2008 and 2018. Thus, the patients included in the study were born between 1992 and 2018. We searched the patient records of both Tampere University Hospital (Pirkanmaa Region) and Päijät-Häme Central Hospital for the following ICD-10 codes: G12, G60-G64 and G70-G73.

2.3. Terms and measures

Neuromuscular diseases were defined as diseases affecting the

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anterior horn cells of the spinal cord, peripheral nerve, nerve-muscle junction and muscle fiber. We excluded patients, who had had one episode of rhabdomyolysis but no suspicion of an underlying neuromuscular disease. Prevalence was defined as the number of patients affected who were alive and living in the area on the prevalence day. This was compared to the total population under 17 years of age on the prevalence day. The male population under 17 years of age was selected for X-linked conditions. Cumulative incidence was defined as the number of patients born between 1992 and 2018 and who developed the disease before the age of 17 years while living in the study area in relation to the total number of live births during the same period.

2.4. Data collection

The patients with a neuromuscular diagnosis were included in the study population. The inclusion criterion was a neuromuscular diagnosis

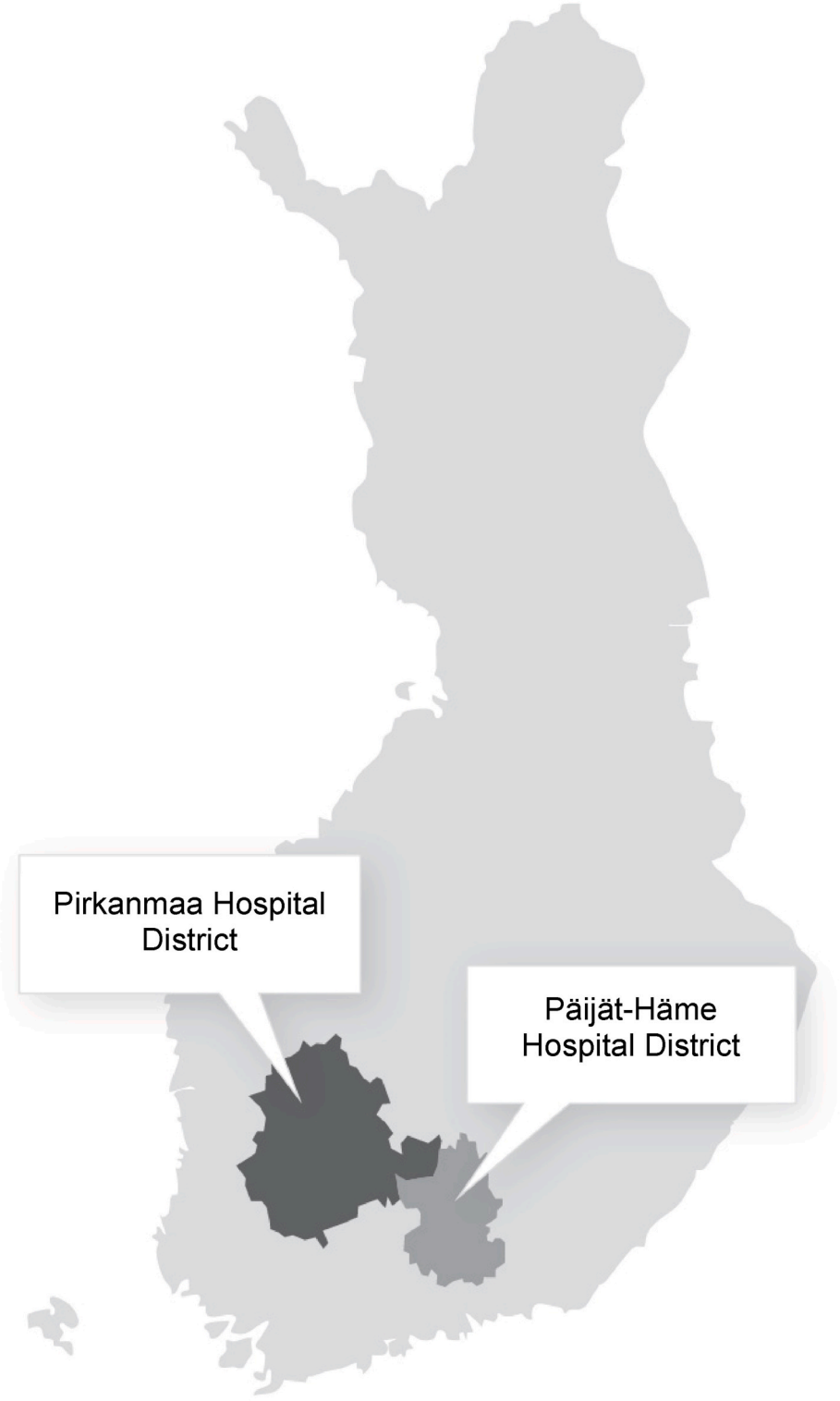

Fig. 1. Pirkanmaa and Päijät-Häme hospital districts in Finland.

established by the treating pediatric neurologist. In case of an unclear diagnosis, the medical records were reviewed and if there was not enough evidence of a neuromuscular disease, the patient was excluded. All included patients had to have a clinical picture of a neuromuscular disease and/or abnormal findings on electromyography (EMG), nerve conduction studies, muscle biopsy or genetic testing.

2.5. Statistical analysis

Wilson's 95 % confidence intervals (95 % CI) were calculated for cumulative incidences and point prevalences.

3. Results

We identified 120 patients from the medical registries of which 30 were excluded. The flowchart of included and excluded patients are shown in Fig. 2. A total of 36 prevalent and 90 incident cases were ascertained, giving a prevalence of 28.0 $\times$ 10^{-5} and a cumulative incidence of 45.9 $\times$ 10^{-5}. The prevalences and incidences for separate diseases are given in Tables 1 and 2.

Of the incident hereditary diseases, 56 (73.7 %) had a molecular genetic confirmation of their diagnosis. However, in some cases the electronic data did not contain all diagnostic information. Patients lacking a genetic diagnosis, but whose clinical data and course of the disease were compatible with the given diagnosis were included in the study. These included three DMD and one Becker muscular dystrophy (BMD) patient whose genetic confirmation was lacking but based on their symptoms, muscle biopsy findings, and lifespan fitting to DMD or BMD they were included.

The most prevalent myopathic disease was DMD (16.8 $\times$ 10-5) and the most prevalent neurogenic disease was Charcot-Marie-Tooth-disease (6.23 $\times$ 10^{-5}). We identified 30 patients with a hereditary neuropathy, 25 with CMT, four with hereditary neuropathy with liability to pressure palsy (HNPP), and one with congenital insensitivity to pain and anhidrosis (CIPA). Six patients with CMT1, five patients with CMT2 and one patient with HNPP remained without a genetic confirmation, but were included based on clinical symptoms, positive family history and/or electrophysiological findings suggestive of CMT or HNPP.

There was no mention of a genetic diagnosis in the electronic medical record for two patients from Päijät-Häme with diagnoses of unspecified myopathy and fascioscapulohumeral dystrophy (FSHD) because they had been diagnosed at Helsinki University Hospital, but they were included in the study. The patient with unspecified myopathy had been using an electric wheelchair since the early school years and the patient with FSHD had weakness in the abdominal and shoulder muscles and a myopathic EMG. One patient from Pirkanmaa had a diagnosis of distal SMA without genetic confirmation, but was included because his EMG findings, nerve conduction studies and biopsy were compatible with the disease.

We identified 14 patients with an acquired neuromuscular disease. Seven patients had Guillan-Barre syndrome (GBS), one had chronic inflammatory demyelinating polyneuropathy (CIDP), two diabetic polyneuropathy, and four myasthenia gravis. Six of the GBS patients were diagnosed based on EMG and nerve conduction study findings and one on MRI findings. The patient with CIDP had demyelinating findings on nerve conduction studies and on a nerve biopsy. The patients with diabetic polyneuropathy had findings compatible with polyneuropathy on nerve conduction studies. The patients with myasthenia gravis had either positive acetylcholine receptor antibodies or a positive tensilon test.

4. Discussion

This is the first reliable epidemiological study on pediatric neuromuscular diseases in Finland. Earlier reports on epidemiological studies from different countries have shown variable results, and there is variation as to which disease groups have been included and how diagnoses have been verified. None of these studies have been done exactly the same way as ours, which complicates comparison but our results for overall prevalences are either close to [1,2] or lower than what has previously been reported [3–5].

The circumstances for conducting an epidemiological study in Finland are good because the examination and follow up of practically all children with motor difficulties are done within our public health

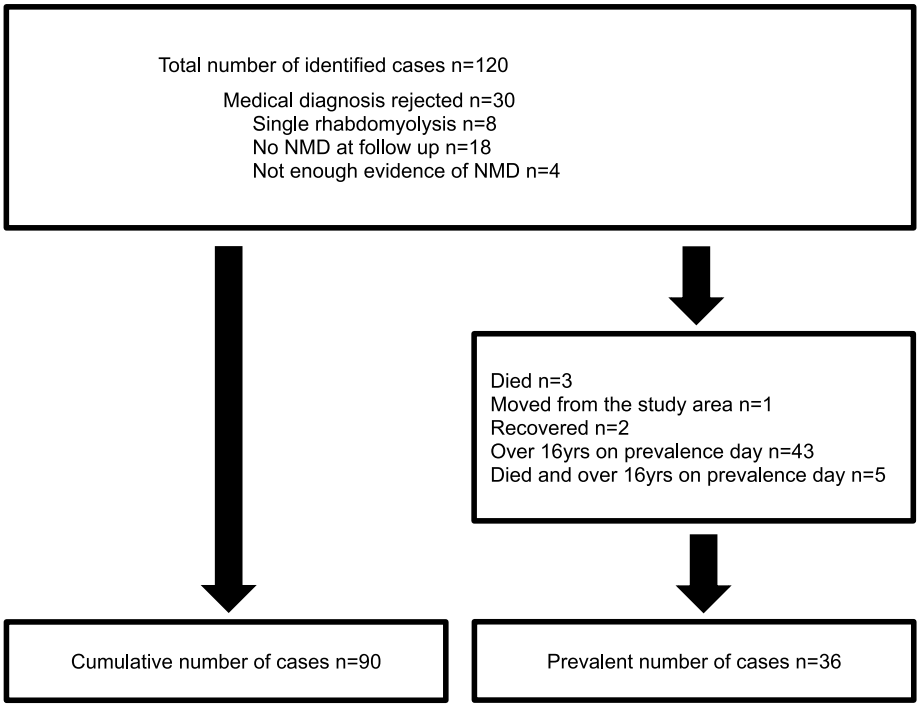

Fig. 2. Flowchart of included and excluded patients.

Table 1
Prevalence of different neuromuscular disorders in Pirkanmaa and Päijät-Häme Hospital Districts.

Disease	Prevalent cases n	Point prevalence /100 000	95 % Confidence Interval/ 100 000	Genetically confirmed cases n (%)
All diseases	36	28.0	20.3–38.8	
Hereditary diseases	33	25.7	18.3–36.1	27 (81.8)
Muscular dystrophies				
Duchenne	11	16.8	9.4–30.0	11 (100)
Becker	0	0	0–5.9	
Fascioscapulohumeral muscular dystrophy	0	0	0–3.0	
Limb-girdle muscular dystrophy 2F	2	1.56	0.4–5.7	2 (100)
Myotonic dystrophy	0	0	0–3.0	
LMNA-related muscular dystrophy	1	0.78	0.1–4.4	1 (100)
Merosin negative congenital muscular dystrophy	1	0.78	0.1–4.4	1 (100)
Bethlem myopathy	1	0.78	0.1–4.4	0
Myopathy NAS				
Mitochondrial myopathie	0	0	0–3.0	
Mitochondrial myopathy				
Non-dystrophic myotonias	2	1.56	0.4–5.7	2 (100)
Myotonia congenita				
Anterior horn cell disorders				
SMA I-III	2	1.56	0.4–5.7	2 (100)
SMALED	0	0	0–3.0	
Distal SMA	1	0.78	0.1–4.4	0
Myasthenic syndromes				
Myasthenia gravis	2	1.56	0.4–5.7	
Congenital myasthenic syndrome	2	1.56	0.4–5.7	2 (100)
Hereditary neuropathies				
CMT	8	6.23	3.2–12.3	4 (50)
CMT1	4	3.12	1.2–8.0	2 (50)
CMT2	3	2.34	0.8–6.9	1 (33.3)
CMT4G	1	0.78	0.1–4.4	1 (100)
HNPP	0	0	0–3.0	
CIPA	1	0.78	0.1–4.4	1 (100)
Autoimmune neuropathies				
GBS	0	0	0–3.0	
CIDP	0	0	0–3.0	
Other polyneuropathies				
Diabetic polyneuropathy	1	0.78	0.1–4.4	

Abbreviations: SMA, spinal muscular atrophy; CMT, Charcot-Marie-Tooth disease, HNPP, hereditary liability to pressure palsies; CIPA, congenital insensitivity to pain and anhidrosis; GBS, Guillain-Barre syndrome; CIDP, chronic inflammatory demyelinating polyneuropathy.

Table 2
Incidence of different neuromuscular disorders in Pirkanmaa and Päijät-Häme Hospital Districts.

Disease	Incident cases n	Cumulative incidence /100 000	95 % Confidence Interval/ 100 000	Genetically confirmed cases n (%)
All diseases	90	45.9	37.4–56.4	
Hereditary diseases	76	38.8	31.0–48.5	56 (73.7)
Muscular dystrophies				
Duchenne	17	17.0	10.6–27.2	14 (82.4)
Becker	4	4.0	1.6–10.3	3 (75)
Fascioscapulohumeral muscular dystrophy	2	1.0	0–3.7	1 (50)
Limb-girdle muscular dystrophy 2F	2	1.0	0–3.7	2 (100)
Myotonic dystrophy	1	0.51	0–2.9	1 (100)
LMNA-related muscular dystrophy	1	0.51	0–2.9	1 (100)
Merosin negative congenital muscular dystrophy	1	0.51	0–2.9	1 (100)
Bethlem myopathy	1	0.51	0–2.9	1 (100)
Myopathy NAS	1	0.51	0–2.9	0
Mitochondrial myopathies	4	2.04	0–5.3	4 (100)
Mitochondrial myopathy				
Anterior horn cell disorders				
SMA I-III	7	3.57	1.7–7.4	6 (85.7)
SMALED	1	0.51	0–2.9	1 (100)
Distal SMA	1	0.51	0–2.9	0
Myasthenic syndromes				
Myasthenia gravis	4	2.04	0–5.3	
Congenital myasthenic syndrome	2	1.0	0–3.7	2 (100)
Hereditary neuropathies				
CMT	25	12.76	8.6–18.8	14 (56)
CMT1	14	7.13	4.3–12.0	8 (57.1)
CMT2	10	5.10	2.8–9.4	5 (50)
CMT4G	1	0.51	0–2.9	1 (100)
HNPP	4	2.04	0–5.3	3 (75)
CIPA	1	0.51	0–2.9	1 (100)
Autoimmune neuropathies				
GBS	7	3.57	1.7–7.4	
CIDP	1	0.51	0–2.9	
Other polyneuropathies				
Diabetic polyneuropathy	2	1.0	0–3.7	

Abbreviations: SMA, spinal muscular atrophy; CMT, Charcot-Marie-Tooth disease, HNPP, hereditary liability to pressure palsies; CIPA, congenital insensitivity to pain and anhidrosis; GBS, Guillain-Barre syndrome; CIDP, chronic inflammatory demyelinating polyneuropathy.

care system. The health care system is similar in all Nordic countries and therefore our results should be comparable to those of Sweden and Norway. However, our overall prevalence of 28.0×10^{-5} is lower than the 63.1×10^{-5} and 57.8×10^{-5} reported from similar studies in western Sweden [1] and northern Norway [4]. Finland has a history of bottlenecks and isolation in the population, and this has caused some autosomal recessive diseases to be more common in the Finnish population than elsewhere [8,9]. Because of our unique genetic heritage, some disease-causing genetic variants are less common in Finland than in other Nordic countries, which might at least partly explain the differences in the prevalence of some neuromuscular diseases.

DMD was the most prevalent single diagnosis in our study. Its point prevalence is identical to 16.8×10^{-5} reported from western Sweden [5]. Previous studies estimating prevalence in the young male population have reported prevalences varying from 9.8×10^{-5} to 16.2×10^{-5} [1,2,10,11] A recent review [7] reports a pooled global DMD prevalence of 7.1 per 100 000 males but this includes data from studies which comprised males of all ages.

Some studies have estimated the prevalence of DMD in the whole population, but since it is a disease occurring only in males and leading to death in the early adult years, we think prevalence should be determined according to the young adult male population.

Our cumulative incidence for DMD (17.0 per 100 000 live male births) is similar to the cumulative incidence of 19.7 per 100 000 live male births reported from Australia [12] and the pooled global birth prevalence of 19.8 per 100 000 live male births reported by a recent meta-analysis [7]. Most of the studies included in the meta-analysis reported period birth prevalences which were calculated in the same way as the cumulative incidence of our study.

There is marked variability in the prevalences of 5q-related SMA combined. Our prevalence of 1.56×10^{-5} is in the range of 1–2 x 10^{-5} regarding all age groups reported in a review from 2017 [13]. The

prevalence in children has been previously reported to be slightly higher [3,5,14]. The cumulative incidence of 3.57×10^{-5} for SMA in our study is low compared to 14.5×10^{-5} reported by a recent study from Germany [15]. Finland does not have a newborn screening program for SMA and therefore it is possible that we could have missed patients that did not come for diagnostic evaluation until later. However, Verhaart et al. [13] report an estimated birth incidence of 8 per 100 000 lives births, despite having included studies from before the era of newborn screening in their review. Interestingly, a newborn screening study from New York [16] with a similarly sized population as ours, also reports an unexpectedly low incidence of 3.55×10^{-5}. The variation may be explained by the low number of cases in the source populations, but regional differences might also exist.

Our prevalence of 6.23×10^{-5} for CMT is lower than the 9.1×10^{-5} and 13.6×10^{-5} reported from New Zealand [17] and Sweden [5] for the pediatric population and lower than the estimated pooled prevalence of 17.69×10^{-5} for the whole age population [18]. Marttila et al. [19] have reported a prevalence of 34.6×10^{-5} in the adult population of Northern Ostrobothnia, Finland. The differences in the results might be explained by differences in the genetic background of the populations.

The prevalence of 1.56×10^{-5} for myotonia congenita is slightly higher than the previously reported childhood prevalences of 0.52×10^{-5} and 0.83×10^{-5} from China [1] and western Sweden [5]. The occurrence of myotonia congenita has been reported to be exceptionally high in northern Finland with a prevalence of 7.3×10^{-5} in all age groups [20], but there is no previously published data on the prevalence of myotonia congenita in the Pirkanmaa and Päijät-Häme regions. Comparison between the areas is difficult due to the differences in the ages of the study populations.

We did not identify any Finnish patients with limb girdle muscular dystrophy (LGMD) in our study; the two siblings with a diagnosis of LGMD were of different ethnicity. The prevalence of LGMD is higher in adult populations [4,21] than in children [1,2,5] and it seems that the prevalence of childhood-onset LGMD is exceptionally low in our study areas. In addition, there was only one patient with myotonic dystrophy type 1 (DM1) and no prevalent cases in either of the areas. The prevalence of DM1 in childhood has previously been reported to be $3.3 \times 10^{-5} - 5.0 \times 10^{-5}$ [1,2,5] and the pooled prevalence of all age groups has been estimated to be 9.99×10^{-5} [22]. Since DM1 might present with predominant cognitive and behavioral symptoms in childhood [23,24], it is possible that there are some unrecognized DM1 cases in our study areas but at least partly the lower prevalence may be explained by a different genetic background of the populations. It is also noteworthy, that we did not identify any patients with a metabolic myopathy. A previous screening study [25] suggests that Pompe disease is very rare in the Finnish population further emphasizing our unique genetic heritage.

Neuromuscular diseases are rare; therefore, the prevalence and incidence of a certain disease can be influenced considerably if a few cases are missed or diagnosed incorrectly, especially when studying small populations. We reviewed the accuracy of the diagnoses and only included patients whose diagnosis of a neuromuscular disease we considered valid. It is possible that patients with mild symptoms have not sought healthcare services or have not been diagnosed correctly and therefore would be missed by our search with ICD-10 codes. The results are therefore more likely to be an underestimate than overestimate.

Altogether, the overall prevalence of pediatric neuromuscular diseases in this study was in the lower range of what has previously been reported from other countries. There is a need for accurate epidemiological data on childhood neuromuscular diseases to aid in the planning of health care services. The results from our study add knowledge about this disease group.

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Conflict of interest

The authors declare they have no competing financial interests or personal relationships that could have appeared to influence the work reported in this paper. The author J. Palmio is a member of the European Reference Network for Rare Neuromuscular Diseases (ERN EURO-NMD).

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