

Prevalence of Genodermatoses in Finland from 1995 to 2022: A Nationwide Registry Study

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Genodermatoses are a large heterogenous group of skin diseases of monogenic disorders originating either from inherited or *de novo* pathogenic variants. Clinical manifestations of genodermatoses vary in severity from mild to life-threatening (1) and can have a substantial impact on patients' quality of life (2, 3). Some genodermatoses, such as neurofibromatosis type 1 (NF1), may also involve extracutaneous organs (4).

Few previous population-based studies (5–10) have examined the epidemiology of genodermatoses in the Nordic Countries. We therefore designed the present study to analyse the periodic prevalence of genodermatoses in Finland and describe the age at diagnosis, over a 2-decade period, through the use of national registry data.

MATERIALS AND METHODS

All study data were obtained from the database of the Finnish Care Register for Health Care (CRHC), which is a statutory nationwide registry maintained by the Finnish Institute for Health and Welfare. It includes information on all healthcare visits to Finnish hospitals and includes inpatient data from 1969 onwards and specialized healthcare outpatient data from 1998 onwards. Each patient's record includes a unique identifying number, along with data from each visit, including primary and subsidiary diagnoses, and type of visit and provider (11). Data from the years 1995 to 2024 inclusive were retrieved for all individuals with a diagnosis of any genodermatosis, as identified by the relevant ICD-10 codes (**Table I**). The study population included only individuals whose record featured at least 2 entries of at least 1 genodermatosis code. A sensitivity analysis was performed with a single entry of a code. Individuals who had a code indicating an "other" or "unspecified" condition in the same category as a more specific diagnosis were counted only in the specific category. For example, if a record contained codes for both ichthyosis vulgaris and "other congenital ichthyosis", the individual was counted only as having the former. If a record contained entries of 2 or more different genodermatosis diagnoses, the individual was included based on only the latest of them. To avoid bias, age at diagnosis was calculated only for subjects born in 1995 or later.

Data are presented as proportions and medians with interquartile ranges. The periodic prevalence of each genodermatosis at study period was calculated by dividing the number of cases by the actual average population of Finland over the study period, as reported by Statistics Finland. All statistical analyses were performed using the R software package (version 4.2.2; R Core Team, Vienna, Austria) within RStudio (version 2023.03.1 Build 446; RStudio, PBC, Boston, MA, USA).

RESULTS

A total of 5,356 individuals with a diagnosis of genodermatosis and fulfilling the inclusion criteria were found in the National Finnish Registry during years 1995–2024. The 5 most common disorders were NF1, tuberous sclerosis, inherited palmoplantar keratosis, benign familial pemphigus (Hailey-Hailey disease) and unspecified neurofibromatosis. In total, these diagnoses accounted for just over 62.6% of all cases, with periodic prevalences varying from 4.7 to 37.3/100,000 (Table I). In a sensitivity analysis with only single entry of a code, frequencies were higher, and especially the numbers of unspecific diagnoses heightened (Table SI).

Neurofibromatoses were the largest group, comprising 2,392 (44.7%) of all cases. Most of these were NF1. The median (Q1–Q3) age at diagnosis for NF1 was 3.0 (1.0–6.0) years, whereas type 2 tended to be diagnosed later, with a median (Q1–Q3) age at diagnosis of 14.0 (8.2–17.0) years.

Overall, median age at the time of diagnosis varied across diagnostic groups. Incontinentia pigmenti had the lowest median age at diagnosis and inherited dyskeratosis the highest.

DISCUSSION

This study reports the prevalence of genodermatoses in Finland and the median ages at the time of diagnosis for a period spanning almost 30 years. The only previous national estimate in Finland of a genodermatosis was for the incidence of neurofibromatoses, which found 1,471 cases of NF1 between 1987 and 2015 (12). Our cohort included 1,997 individuals with NF1 between 1995 and 2024, suggesting an increase in the incidence compared

Table 1. Frequencies of genodermatoses in Finland between 1995 and 2024

ICD-10	Name of diagnosis	N total	N ^a	Median age at first recorded diagnosis [Q1–Q3] ^a	Prevalence per 100,000 (95% CI)
Q80.0	Ichthyosis vulgaris	237	65	6.0 [3.0–11.0]	4.4 (3.9–5.0)
Q80.1	X-linked ichthyosis	193	94	5.5 [1.0–11.0]	3.6 (3.1–4.2)
Q80.2	Lamellar ichthyosis	90	40	2.5 [0.0–7.5]	1.7 (1.4–2.1)
Q80.3	Congenital bullous ichthyosiform erythroderma	36	12	5.0 [0.8–9.2]	0.7 (0.5–0.9)
Q80.4	Harlequin fetus	6	<3	15.5 [14.8–16.2]	0.1 (0.0–0.2)
Q80.8	Other congenital ichthyosis	126	65	2.0 [0.0–7.0]	2.4 (2.0–2.8)
Q80.9	Congenital ichthyosis unspecified	145	58	2.0 [0.0–6.8]	2.7 (2.3–3.2)
Q81.0	Epidermolysis bullosa simplex	133	62	2.0 [0.0–15.0]	2.5 (2.1–2.9)
Q81.1	Epidermolysis bullosa letalis (Herlitz)	16	8	6.0 [3.8–9.2]	0.3 (0.2–0.5)
Q81.2	Epidermolysis bullosa dystrophica	73	26	2.0 [0.0–4.8]	1.4 (1.1–1.7)
Q81.8	Other epidermolysis bullosa	36	13	2.0 [0.0–5.0]	0.7 (0.5–0.9)
Q81.9	Epidermolysis bullosa unspecified	47	16	1.5 [1.0–5.0]	0.9 (0.6–1.2)
Q82.1	Xeroderma pigmentosum	93	34	1.5 [1.0–4.8]	1.7 (1.4–2.1)
Q82.3	Incontinentia pigmenti	117	70	0.0 [0.0–0.0]	2.2 (1.8–2.6)
Q82.4	Ectodermal dysplasia	154	72	4.0 [1.0–6.0]	2.9 (2.4–3.4)
Q82.80	Benign familial pemphigus (Hailey–Hailey)	301	11	2.0 [1.0–9.5]	5.6 (5.0–6.3)
Q82.81	Follicular keratosis (Darier disease)	205	14	14.5 [10.8–15.8]	3.8 (3.3–4.4)
Q82.82	Inherited palmar and plantar keratosis	379	114	9.5 [5.0–17.0]	7.1 (6.4–7.8)
Q82.83	Dyskeratosis congenita	35	10	16.0 [12.0–21.0]	0.7 (0.5–0.9)
Q82.84	Pseudoxanthoma elasticum	28	4	12.5 [9.0–15.8]	0.5 (0.3–0.8)
Q82.85	Acrokeratosis verruciformis	11	<3	12.5 [9.8–15.2]	0.2 (0.1–0.4)
Q82.86	Gorlin's syndrome	74	34	6.5 [1.2–12.0]	1.4 (1.1–1.7)
Q85.00	Neurofibromatosis type 1	1,997	825	3.0 [1.0–6.0]	37.3 (35.7–39.0)
Q85.01	Neurofibromatosis type 2	143	26	14.0 [8.2–17.0]	2.7 (2.3–3.1)
Q85.09	Neurofibromatosis unspecified	252	22	8.0 [3.0–12.0]	4.7 (4.1–5.3)
Q85.1	Tuberous sclerosis	424	180	1.0 [0.0–4.0]	7.9 (7.2–8.7)

^aAnalysed in those born in 1995 or later.

CI: confidence interval; ICD-10: International Classification of Diseases, 10th version.

with the earlier study. In a similar comparison, it appears that the number of patients with NF2 has also increased (12). However, it is of note that the diagnoses in the previous study were based on chart review and re-verified based on diagnostic criteria, while ours were based on registry data. Thus, it is possible that this finding may also originate from other reasons, such as diagnostic developments including genetic testing of milder presentations.

We found 5,356 individuals diagnosed with a genodermatosis for the study period. In the Swedish study, they found over 13,000 patients with a genodermatosis diagnosis in the national patient registry over a 19 year period (5). Our study required at least 2 recorded diagnosis entries in the patient system to be included in the dataset, whereas it is not reported in the Swedish national study how many recorded entries were required (5). This makes direct comparison to the Swedish study difficult and thus it is most likely that our study has higher specificity due to stricter criteria. In addition, in our dataset patients were only allowed to be included under one genodermatosis diagnosis, whereas in the Swedish study patients could have more than one diagnosis.

Neurofibromatosis, specifically NF1 was the most common genodermatosis in our study, followed by tuberous sclerosis. There are differences in diagnosis coding between Finnish and Swedish versions of ICD-10, which hampers the comparison with previous studies: For example, the conditions with highest incidence in the Swedish data, porokeratosis of Mibelli and keratosis follicularis spinulosa decalvans, cannot be separately identified in the Finnish national ICD-10 coding system due to lack of subcodes. Correspondingly, the Swedish study did not report the incidence or prevalence of neurofibromatosis at national level, due to unspecificity of the Swedish ICD-10 codes but they found it as second most common genodermatosis (n=32/297, 11%) in their hospital cohort (5).

In our study, the prevalence of EB varied from 2.5 (EB simplex) to 0.3/100,000 persons in more severe forms of EB being only partly comparable with the prevalence in Swedish data with prevalences of 3.8/100,000 for EB simplex and 1.0/100,000 for dystrophic EB (5). These slight differences between the incidences in Finnish and Swedish populations may partly reflect migration-related differences in genetic background of the population (13). However, in both countries, prevalence varied markedly between EB subtypes, as described previously in other countries as well (14, 15).

Previous Danish studies have reported birth prevalence estimates for both ectodermal dysplasia (10) and incontinentia pigmenti (9) based on nationwide data collected during time periods broadly comparable to ours. For ectodermal dysplasia, the Danish birth prevalence estimate (14.5 per 100,000 live births) was markedly higher than the prevalence observed in our study (2.9 per 100,000). Since diagnoses in the Danish study were retrospectively verified through medical record review, this difference is unlikely to be explained by diagnostic misclassification alone. In contrast, our estimate for incontinentia pigmenti was highly consistent with the Danish birth prevalence estimate (2.2 vs 2.37 per 100,000), despite the use of retrospective diagnostic validation in the Danish study as well. However, comparing birth prevalence and periodic prevalence may include bias especially for diagnoses which affect survival. In addition, other aspects like emigration and failure to capture mild cases may affect the differences between birth and periodic prevalences.

The main strengths of this study are the national coverage, long follow-up period and ability to identify hospital-based cases from tax-funded healthcare system. The registry-based nature of the design left us unable to confirm the diagnoses. We admit that the validity of the diagnoses is generalized from other studies (16, 17) and diagnostic validation was not possible because medical

records were not reviewed. However, all diagnoses in CRHC are set in specialized healthcare which increases the accuracy of coding. Most diagnoses (>75%) were recorded in the specialty of dermatology, second most commonly in the specialty of paediatric neurology, reflecting the high number of neurofibromatosis diagnoses found in our study. As no primary care or private practitioners data were included, we admit that there may be some underreporting as registry-data may only capture most severe cases, and variation may occur between diagnoses as several genodermatoses may present with relatively mild phenotypes, which may not be referred to hospital clinics. However, it is of note that hospital dermatology services are publicly available to everyone living in Finland by doctors' referral, and thus, we consider that most individuals with genodermatosis, who have sought medical care are included in the study, as genodermatoses diagnoses are typically verified in hospital clinics.

We conclude that genodermatoses are rare in Finland and many of them, especially NF type 2, are diagnosed in late childhood or adolescence. As well as skin symptoms, these diseases may also have extracutaneous manifestations (1). Thus, it is important to be aware of these conditions and to employ modern genetic diagnostic techniques in a timely fashion to offer the most accurate diagnosis as early as possible. As hospital-based registry data have its limitations, these diseases may occur more commonly, and thus specific care registries for genodermatoses or preferably prospective studies are needed. Accurate knowledge of the prevalence of the genodermatoses would advocate the needs of these individuals in the healthcare system.

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Data availability statement: Our data are from the Finnish Care Register for Health Care (CRHC), which is maintained by the Finnish Institute of Health and Welfare. According to Finnish laws and regulations, the data in the social welfare and healthcare registers and documents are confidential. As a responsible authority, FinData can, on a case-by-case basis, grant permission to use the registers and documents for the purposes of scientific research. More information on research authorization applications can be found on www.findata.fi/en.

Ethics committee: The study was exempted from institutional review board ethics review because it was retrospective and based only on data from medical records.

The authors have no conflicts of interest to declare.

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