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# Ectodermal dysplasias in Denmark: identification and characterization of a nationwide cohort

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## Introduction & Objectives:

Ectodermal dysplasias (ED) constitute a group of rare genetic disorders of the skin and skin appendages. Common features of EDs include hypodontia, hypotrichosis, and hypohidrosis. Population-based studies into the epidemiology of ED are lacking. We aimed to utilize Danish health registries to identify and characterize a nationwide cohort of patients with ED allowing population-based investigations of the disease epidemiology.

## Materials & Methods:

We searched the Danish National Patient Registry for hospitalizations and outpatient contacts with International Classification of Diseases 10th Revision (ICD-10) diagnosis codes indicative of ED from Jan 1st, 1995 to Aug 25th, 2021. We applied three search algorithms identifying patients with ICD-10 codes for (1) EDs specifically, (2) $^{3}$2 cardinal features, or (3) 1 cardinal feature with $^{3}$2 minor features (only at Aarhus University Hospital). Algorithms 2 and 3 also included records of hypodontia in the Danish Central Dentistry Registry (SCOR). We supplemented the search with patients registered in the Danish RareDis Database (2007–2021), the Danish Database of Genodermatoses (2018-2021), and local genetic databases. One author then reviewed the patient's medical records for validation and detailed patient characterization. We estimated the minimum birth prevalence for birth cohorts 1995–2011 to ensure a minimum attained age of 10 years for the population at the end of the study period.

## Results:

Our database search identified 845 patients suspected of ED, for which 792 (93.7%) medical records were available. Our validation confirmed an ED disorder in 320 cases with an additional 77 possible cases, yielding a combined positive predictive value (PPV) of 50.1% [95% confidence interval (CI): 46.6–53.6]. The PPVs of the three search algorithms were (1) 67.0% (95% confidence interval (CI): 62.7–71.0%), (2) 8.2% (95% CI 4.6–14.3%), and (3) 1.8% (95% CI 0–7.2%), respectively. The estimated minimum birth prevalence for all EDs combined (n=161) was 15 cases per 100,000 live births (95% CI: 12–17 per 100,000 live births). Of 397 validated cases, 245 (61.7%) were females. A molecular genetic diagnosis was available for 242 (61%) patients, including *EDA* (n=100), *IKBKG* (n=55), *WNT10A* (n=21), *TRPS1* (n=18), *EDAR* (n=10), *P63* (n=9), *GJB6* (n=9), *PORCN* (n=7), and other rare genes.

## Conclusion:

We used the Danish health registries to identify and characterize a validated nationwide cohort of ED patients. With detailed clinical and molecular data, the cohort provides a unique resource for future ED research. The low PPVs of the search algorithms also emphasize the importance of diagnosis validation. The ED prevalence estimated in our study is lower than previously reported, a possible result of this stringent validation and updated disease definition.