

**Short status report regarding NDA Research Grant 2021 for the NDA board
November 13th 2022**

Recipient: Laura Krogh Herlin

Project: Ectodermal dysplasia (ED) in Denmark

Aim:

To identify and characterize a large nationwide cohort of patients with ED allowing population-based epidemiologic studies.

Current status:

We have now obtained the full dataset on putative ED patients identified through a search of the Danish National Patient Registry (DNPR) from 1995-2021 (data provided from the Danish Health Data Authority). The search was based on their diagnosis history (ICD-10 diagnosis codes). Moreover, the search has been complemented by searches of RareDis (nationwide registry of rare diseases) and the Danish Genodermatosis Database to ensure completeness. In total, we have identified 1091 putative ED patients. At this point, diagnosis validation has been completed for a total of 136 patients of which 80 patients had ED. This preliminary status shows the utility of DNPR data to identify a large cohort of patients with a rare disease, but also the need for diagnosis validation to identify the true ED patients eligible for subsequent analyses.

The generous research grant (30,000 SEK) from NDA has been used to cover expenses for obtaining individual-level DNPR data from the Danish Health Data authority.

Dissemination:

The project and our preliminary results from the DNPR search were presented at the NCDV annual meeting April 2022 by the grant recipient.

The first manuscript describing the cohort identification and search validity is expected around Fall 2023. Hereafter, derived studies of long-term outcomes in ED patients utilizing our validated cohort will follow.