Göteborg, 2025

Genodermatoses: epidemiology and application of advanced laser microscopy techniques

Akademisk avhandling

Som för avläggande av medicine doktorsexamen vid Sahlgrenska akademin, Göteborgs universitet kommer att offentligen försvaras i Arvid Carlsson, Academicum, Medicinaregatan 3, Göteborg, den 25 april 2025, klockan 13.00

av **Rahime Inci**

Fakultetsopponent:

Enikö Sonkoly, Professor

Uppsala universitet, Sverige

Avhandlingen baseras på följande delarbeten

I. *Zagoras T*, Inci R*, Kantere D, Holmström P, Broström J, Gillstedt M, Polesie S, Peltonen S.* Incidence and Prevalence of 73 Different Genodermatoses: A Nationwide Study in Sweden. Acta Derm Venereol. 2023 Aug 24;103: adv12404. *Equal contribution

II. *Inci R, Zagoras T, Kantere D, Holmström P, Gillstedt M, Polesie S, Peltonen S.* Porokeratosis is one of the most common genodermatoses and is associated with an increased risk of keratinocyte cancer and melanoma. J Eur Acad Dermatol Venereol. 2023 Feb;37(2):420-427.

III. *Inci R, Gillstedt M, Kallionpää RA, Peltonen S, Polesie S.* Patients with Darier disease have an increased risk of keratinocyte carcinoma: A Swedish registry-based nationwide cohort study. Orphanet J Rare Dis. 2024;19(1):463.

IV. *Inci R, Neittaanmäki N, James J, Ericson M.B, Peltonen S, Kantere D.* Applicability of novel laser scanning microscopy techniques in demonstrating characteristic features of porokeratosis. (Manuskript)

SAHLGRENSKA AKADEMIN
INSTITUTIONEN FÖR KLINISKA VETENSKAPER

GÖTEBORGS UNIVERSITET

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Genodermatoses: epidemiology and application of advanced laser microscopy techniques

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Abstract

Genodermatoses encompass a wide range of hereditary skin disorders that significantly affect the quality of life of patients and pose substantial challenges for research. Most genodermatoses are rare, and their incidence and prevalence often are only roughly estimated. This thesis comprises three registry-based retrospective cohort studies and an experimental prospective study.

The primary aim was to ascertain the incidence and prevalence of genodermatoses in Sweden, along with their healthcare needs, and to provide a comprehensive overview of these conditions. The secondary aim was to investigate the cancer risk associated with specific genodermatoses, including, porokeratosis and Darier disease. The experimental section assessed the applicability of reflectance confocal microscopy (RCM) and multiphoton microscopy (MPM) in diagnosing porokeratosis compared to traditional histopathology.

We described a local patient cohort by searching the local patient registry at Sahlgrenska University Hospital in Gothenburg, Sweden. This included patients who visited the Department of Dermatology and Venereology during the period from 2016 to 2020. To gather nationwide data, we used the National Patient Registry, where we identified both inpatients and outpatients using ICD-10-SE codes between January 1, 2001, and December 31, 2020. The same patient cohorts were used for **Papers I, II,** and **III.**

A total of 73 genodermatoses were registered in Sweden, and the incidence and prevalence of these diseases were reported in **Paper I**. The results showed that porokeratosis and Darier disease were among the most common genodermatoses.

We observed an association between keratinocyte cancer and melanoma among patients with porokeratosis, as discussed in **Paper II**, and Darier disease, as discussed in **Paper III**. A prospective descriptive study presented in **Paper IV** evaluated the potential of the laser microscopy techniques RCM and MPM as alternatives to histopathology, with the goal of identifying a rapid, non-invasive diagnostic tool.

Keywords: Genodermatoses, porokeratosis, Darier disease, epidemiology, laser-scanning microscopy, reflectance confocal microscopy, multiphoton microscopy.

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