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New study maps a group of rare genetic diseases for the first time

A new study from Aarhus University reveals fewer cases of ectodermal dysplasia in Denmark than previously believed. This first mapping of the diseases holds crucial importance for diagnosis, treatment, and future research.

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Behind the term ectodermal dysplasia lies a wide range of genetic disorders that can have a major impact on patients' lives and quality of life. Some cannot sweat, others are born with missing or cone-shaped teeth, while others have very little hair, skeletal deformities, or abnormal nails. A total of 49 diagnoses fall under the term, but until now we have not had an overview of how many Danes actually suffer from an ectodermal dysplasia disorder.

IMAGE:

A NEW STUDY FROM AARHUS UNIVERSITY BY PHD-STUDENT AND MD, LAURA KROGH HERLIN, MAPS FOR THE FIRST TIME A GROUP OF RARE GENETIC DISEASES THAT FALLS UNDER THE TERM ECTODERMAL DYSPLASIA. THE STUDY DEMONSTRATES, AMONG OTHER THINGS, THAT ONLY 67% OF DANISH PATIENTS DIAGNOSED ACTUALLY HAVE A CONGENITAL ECTODERMAL DYSPLASIA.

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A new study from Aarhus University has for the first time provided an overview of the prevalence of the condition in Denmark, and the study shows that there are fewer patients with ectodermal dysplasia than was previously thought to be the case.

In fact, the study shows that only 67% of Danes registered with an ectodermal dysplasia diagnosis actually have congenital ectodermal dysplasia. And this is one of the reasons why it is important to study these rather rare diseases, says doctor and PhD student Laura Krogh Herlin from the Department of Clinical Medicine at Aarhus University and Department of Dermatology, Aarhus University Hospital, who is one of the researchers behind the study:

"It's important that we don't just study the most common diseases, but also study and learn more about rare diseases. Overall, it is believed that as many as 36 million people in the EU are affected by a rare disease. Even though individuals with ectodermal dysplasia only make up a small part of that number, it's crucial for patients affected by a rare disorder that we gain new knowledge, so we can both diagnose and treat better in the future," she says.

Laura Krogh Herlin's study shows that approximately 1 in 7000 Danes are born with ectodermal dysplasia, but the study has also mapped the key characteristics most often seen in connection with an ectodermal dysplasia diagnosis. This is important knowledge because the diagnoses often involve many disciplines and require collaboration across the healthcare system, she says:

"The study shows that 81% of patients have abnormal teeth, while 59% of patients have skin issues and 27% have nail issues. Approx. 1/3 of patients sweat less than normal, leading to a risk of overheating, e.g., if they have a fever. This means that patients are first seen by a wide

range of medical specialists, ranging from dermatologists, paediatricians, dentists and clinical geneticists, who need to be able to work together to help the patients. This is why it's crucial that we map the symptoms they should specifically look for in patients."

With the new study, researchers can now delve even deeper into the conditions and their prognosis, and Laura Krogh Herlin hopes that this will mean both faster diagnoses and better treatment in the future for patients born with ectodermal dysplasia.

"Patients with rare diagnoses often feel left behind due to their disease, and it can take longer to get the correct diagnosis and treatment. Hopefully, population-based knowledge about ectodermal dysplasias will help to ensure that this patient group can also receive fast and good treatment in their encounter with the healthcare system," she says.

FACTS

- The study identified a total of 396 patients in Denmark with ectodermal dysplasia. The most common form is X-linked hypohidrotic ectodermal dysplasia, which was found in 100 individuals.
- The incidence (birth prevalence) of ectodermal dysplasia in Denmark is estimated at 14.5 per 100,000 births, corresponding to around 1 in 7000. This figure is lower than previously estimated.
- The study describes key characteristics of ectodermal dysplasia, including phenotypic features involving teeth (81%), hair (59%), skin (59%), sweat glands (34%) and nails (27%) as the most common findings.

The research results - more information

- **Type of study:** Nationwide population-based cohort study with diagnosis validation
- **Partners:** Aarhus University Hospital: Mette Sommerlund, Sigrún A.J. Schmidt, Kirsten Rønholt, Jenny Blechingberg, Lise Graversen, Trine H. Mogensen, Hans Gjørup; Danish Genodermatosis Group: Anette Bygum, Annette Schuster, Ulrikke Lei, Mette Mogensen, Gabrielle R. Vinding; Rigshospitalet: Xenia B. Hermann, Malene Djursby, Hanne Hove; London School of Hygiene and Tropical Medicine: Sinéad M. Langan.
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  Read more in the scientific article: https://jamanetwork.com/journals/jamadermatology/article-abstract/2815804

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COI STATEMENT

None