

Media release

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Basic information

Name: Signe Væth Email: vaeth@dadlnet.dk Phone: 24222041

Department of: Clinical Medicine

Main supervisor: Professor Uffe Birk Jensen

Title of dissertation: "Charcot-Marie-Tooth disease- genetic and epidemiological aspects"

Date for defence: 17/3 at (time of day): 13:00 Place: Auditoriet, Science Center Skejby, Brendstrupgårdsvej 21 A, 8200 Aarhus N

Media release (Danish)

Genetiske og epidemiologiske aspekter af Charcot-Marie-Tooths sygdom

Charcot-Marie-Tooths sygdom i Danmark: En undersøgelse af forekomst, dødelighed og ny genetisk analyseteknik. Et nyt ph.d.-projekt fra Aarhus Universitet, Health. Projektet er gennemført af Læge Signe Væth, der forsvare sin afhandling d. 17/3.

Charcot-Marie-Tooth sygdom (CMT) er den hyppigste arvelige sygdom i det perifere nervesystem, men den epidemiologiske viden om sygdommen er sparsom. Projektet er baseret på data fra Landspatientregistret og præsenterer beregninger af forekomst af og dødelighed ved af CMT i Danmark. Projektet er det første nationale registerbaserede studie af CMT, og det første der sammenligner dødeligheden blandt personer diagnosticeret med CMT med dødeligheden i baggrundsbefolkningen. Indtil for få år siden har det kun været muligt at tilbyde analyse af ganske få gener når en patient skulle udredes for CMT, dette selvom mere end 80 gener er forbundet med sygdommen. En ny teknik har nu muliggjort analyse af mange gener samtidigt (Next Generation Sequencing). Dette projekt opgør resultaterne af den genetiske diagnostik for CMT i Danmark frem til 2012, og undersøger effekten af den nye analyseteknik ved at udføre samlet analyse af 63 CMT-relaterede gener på 195 patienter der tidligere har været undersøgt for CMT uden fund af sygdomsgivende mutation.

Forsvaret af ph.d.-projektet er offentligt og finder sted den 17/3 kl. 13 i Auditoriet, Science Center Skejby, Brendstrupgårdsvej 21 A, 8200 Aarhus N. Titlen på projektet er " Charcot-Marie-Tooths sygdom - genetiske og epidemiologiske aspekter. Yderligere oplysninger: Ph.d.-studerende Signe Væth, e-mail: vaeth@dadlnet.dk, tlf. 24222041.

Media release (English)

Genetic and epidemiological aspects of Charcot-Marie-Tooth disease

Charcot-Marie-Tooth disease in Denmark: A study of prevalence, mortality and new genetic analysis technique. The project was carried out by MD Signe Væth, who is defending her dissertation on 17/3.

Charcot-Marie-Tooth disease (CMT) is the most common inherited disorder of the peripheral nervous system, yet the epidemiological knowledge of CMT is scarce. This project is based on data from the Danish National Patient Registry, and presents estimates of prevalence and mortality of CMT in Denmark. The project is the first national register based study of CMT, and the first to compare the mortality in CMT diagnosed patients with that of the general population. Until recently, only few genes were analyzed in a patient suspected of having CMT, although more than 80 genes are related to the disorder. A new technique has now made it possible to perform simultaneous analysis of many genes (Next Generation Sequencing, NGS). This project reports the results of genetic CMT analysis in

Denmark until 2012, and investigates the effect of implementing NGS by performing analysis of 63 CMT related genes on 195 patients previously analyzed for CMT without a genetically confirmed diagnosis.

The defence is public and takes place on 17/3 Science Center Skejby, Brendstrupgårdsvej 21 A, 8200 Aarhus N. The title of the project is "Charcot-Marie-Tooth disease- genetic and epidemiological aspects". For more information, please contact PhD student Signe Væth, e-mail: vaeth@dadlnet.dk, Phone +45 24222041.

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