

## Press release

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### Basic information

Name: Cecilie Ejerskov Pedersen

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Department of: Clinical Medicine

Main supervisor: Annette Haagerup, MD, DMSc, Head of Dept. and Associate Professor,, NIDO | danmark, Regional Hospital West and Institute of Clinical Medicine Aarhus University

Title of dissertation: Gastrointestinal symptoms and NF1 mutational spectrum in neurofibromatosis type 1

Date for defence: March 2nd at (time of day): 13h30 Place: Auditorium B entrance G2

Press release (Danish)

Forsvar af ph.d.-projekt om neurofibromatose type 1

Sammenhængen mellem den forholdsvis sjældne sygdom neurofibromatose 1 og symptomer i mavetarmkanalen er for første gang undersøgt i et nyt ph.d.-projekt fra Aarhus Universitet, Health. Projektet er gennemført af læge og ph.d.-studerende Cecilie Ejerskov Pedersen, der forsvarer det d. 2. marts 2018.

Neurofibromatose er en arvelig og uhelbredelig sygdom, der blandt andet er karakteriseret ved lysebrune pigmentforandringer i huden (café-au-lait-pletter) og tumorer i nervesystemet. Sygdommen kan give mange forskellige symptomer afhængig af, hvor tumorerne udvikler sig.

Projektet, der undersøger 277 danske patienter ved hjælp af blandt andet kliniske og genetiske test, er et vigtigt skridt på vej mod at tegne et mere komplet billede af neurofibromatose 1 – både hvad angår de genetiske mekanismer, der ligger til grund for sygdommen, samt sygdommens symptomer og behandling af disse.

Forsvaret af ph.d.-projektet er offentligt og finder sted den 2. marts 2018 kl. 13.30 i B auditorium, Aarhus Universitetshospital, Palle Juul-Jensens Boulevard 99, 8200 Aarhus N. Titlen på projektet er "Gastrointestinal symptoms and NF1 mutational spectrum in neurofibromatosis type 1". Yderligere oplysninger: Ph.d.-studerende Cecilie Ejerskov Pedersen, e-mail: [ceej@clin.au.dk](mailto:ceej@clin.au.dk), tlf. 25673729.

Bedømmelsesudvalg:

Christian Lodberg Hvas, Associate Professor, MD, PhD - chairman of the committee and moderator of the defence

Department of Hepatology and Gastroenterology, Aarhus University Hospital, Aarhus, Denmark

Ludwine Messiaen, Professor, PhD, FACMG, Director

Medical Genomics Laboratory, Department of Genetics, The University of Alabama at Birmingham, Birmingham, Alabama, USA

Sue Huson, Honorary consultant, MD, FRCP

Manchester Centre for Genomic Medicine, St Mary's Hospital, Central Manchester University Hospitals NHS Foundation Trust, Manchester Academic Health Sciences Centre, Manchester, United Kingdom

Press release (English)

PhD defence on neurofibromatosis 1

A new PhD project from Aarhus University investigates the link between neurofibromatosis type 1 and gastrointestinal complaints, a hitherto unexplored research area. The project was carried out by PhD student Cecilie Ejerskov Pedersen, who is defending her dissertation on March 2nd, 2018.

Neurofibromatosis type 1 is a rare, hereditary, and incurable condition characterized by changes in skin pigmentation and the growth of tumors along nerves. The symptoms of this condition vary widely among affected people, dependent on the specific location of the tumors.

The project examines 277 Danish patients using clinical and genetic tests among others and uncovers important new insights about neurofibromatosis 1. The study holds promising perspectives for future research in the genetics and pathophysiology of neurofibromatosis 1, as well as for the treatment of this complex disease.

The defence is public and takes place on March 2nd 2018 at 13.30 i B auditorium, Aarhus Universitetshospital, Palle Juul-Jensens Boulevard 99, 8200 Aarhus N. The title of the PhD project is Gastrointestinal symptoms and NF1 mutational spectrum in neurofibromatosis type 1. For more information, please contact PhD student Cecilie Ejerskov Pedersen, email: ceej@clin.au.dk, Phone +45 25673729.

Assessment committee:

Christian Lodberg Hvas, Associate Professor, MD, PhD - chairman of the committee and moderator of the defence

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Ludwine Messiaen, Professor, PhD, FACMG, Director

Medical Genomics Laboratory, Department of Genetics, The University of Alabama at Birmingham, Birmingham, Alabama, USA

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