AARHUS UNIVERSITY

Press release

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Basic information

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Department of: Biomedicine

Main supervisor: Professor Ulf Simonsen

Title of dissertation: Pulmonary alveolar microlithiasis - clinical and genetic aspects

Date for defence: Thursday February 6, 2020 at (time of day): 14:00 - 16:00 Place: Søauditorierne - Eduard Biermann Auditoriet (Building 1252, room 204), Aarhus University, Bartholins Allé 3, 8000 Aarhus C

Press release (Danish)

Kliniske og genetiske aspekter af lungesygdommen pulmonal alveolær mikrolithiasis

Pulmonal alveolær mikrolithiasis (PAM) er en sjælden lungesygdom, der skyldes en genfejl i et transportmolekyle for stoffet fosfat. På nuværende tidspunkt er den eneste effektive behandling lungetransplantation. I et nyt ph.d.-projekt fra Aarhus Universitet, Health er flere nye genfejl blevet påvist og sammenhængen mellem typen af genfejl og sygdommens sværhedsgrad er blevet nærmere belyst. Projektet er gennemført af Åsa Lina Alle Madsen (née Jönsson), der forsvarer det d. 6/2-2020.

PAM er en sjælden lungesygdom, hvor der ophobes små kalksten i lungerne. Færre end 1100 mennesker menes at lide af sygdommen. Sygdommen har en varierende sværhedsgrad, men oftest bliver patienterne langsomt mere syge, får behov for ilt og dør ultimativt af lungesvigt. Den eneste effektive behandling er lungetransplantation. Man ved at sygdommen skyldes genfejl i et transportmolekyle for fosfat, men på nuværende tidspunkt er der kun ganske få patienter, der er blevet genetisk undersøgt. Formålet med dette ph.d.-projekt var at påvise nye og undersøge forskellige typer af genfejl og se på sammenhængen mellem disse og sygdommens sværhedsgrad. Der blev inkluderet 14 patienter fra USA og flere Europæiske lande. Gennem dette forskningsnetværk blev der indsamlet blodprøver eller spyt og kliniske oplysninger om patienterne. Herudover blev der lavet cellestudier for at undersøge effekten af forskellige typer af genfejl på celleniveau. Hos patienterne fandt vi flere nye genfejl i fosfattransport molekylet, som ikke tidligere er beskrevet. Vores undersøgelser tyder også på, at der kan være en sammenhæng mellem sygdommens sværhedsgrad og typen af genfejl. Projektet bidrager med væsentlig ny viden, der på sigt forhåbentlig kan lede til udvikling af en effektiv medicinsk eller genetisk behandling af sygdommen.

Forsvaret af ph.d.-projektet er offentligt og finder sted den 6/2 2020 kl. 14:00 i Søauditorierne - Eduard Biermann Auditoriet (Bygning 1252, lok. 204), Aarhus Universitet, Bartholins Allé 3, 8000 Aarhus C. Titlen på projektet er ”Pulmonary alveolar microlithiasis - clinical and genetic aspects”. Yderligere oplysninger: Ph.d.-studerende Åsa Lina Alle Madsen (née Jönsson), e-mail: jonsson@biomed.au.dk, tlf. +45 40513516.

Bedømmelsesudvalg:
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Jens Michael Hertz, Professor, Klinisk Institut, forskningsenheden Human Genetik, Klinisk Genetisk Afdeling, Odense Universitetshospital, Danmark

Press release (English)

Pulmonary alveolar microlithiasis - clinical and genetic aspects

Pulmonary alveolar microlithiasis (PAM) is a rare lung disease caused by genetic mutations. No effective treatment exists except for lung transplantation. In a new PhD project at Aarhus University, Health, several new mutations in patients with PAM have been discovered. In addition, a possible association between the severity of the disease and the genetic defects have been investigated. The project was carried out by Åsa Lina Alle Madsen (née Jönsson), who is defending her dissertation on February 6, 2020.

PAM is a rare lung disease with less than 1100 patients worldwide. It is characterised by accumulation of calcium-phosphate stones in the lungs. The clinical course is variable, most often slowly progressive leading to pulmonary failure and death. The only effective treatment is lung transplantation. The disease is caused by genetic mutations in a sodium-phosphate transporter. Up until now, only a few patients have been genetic tested. The aim of the current PhD project was to detect and investigate different mutations and to explore whether there may exist any association with the disease severity. In this research collaboration, patients were included from USA and several European countries and we collected blood samples or saliva and clinical data from 14 patients. Furthermore, the impact of different genetic mutations on the function of the transporter was investigated in cellular studies. We identified several novel mutations in the patients. In addition, data from our study suggest that there may exist an association between the disease severity and the type of mutation. The project contributes with substantial new knowledge about PAM that hopefully will lead to development of new treatment options.

The defence is public and takes place on February 6, 2020 at 14:00 in Søauditorierne - Eduard Biermann Auditoriet (Building 1252, room 204), Aarhus University, Bartholins Allé 3, 8000 Aarhus C. The title of the project is ”Pulmonary alveolar microlithiasis - clinical and genetic aspects”. For more information, please contact PhD student Åsa Lina Alle Madsen (née Jönsson), e-mail: jonsson@biomed.au.dk, phone: +45 40513516.

Assessment committee:
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