AARHUS UNIVERSITY

Press release

Please fill in this form and return it to graduateschoolhealth@au.dk in Word format no later than three weeks prior to your defence.

Basic information

Name: Didde Haslund Email: didde.haslund@biomed.au.dk Phone: 31515190

Department of: Biomedicine

Main supervisor: Jacob Giehm Mikkelsen

Title of dissertation: "Dominant negative intracellular retention of C1 inhibitor in hereditary angioedema"

Date for defence: 10/5 at (time of day): 1.00 PM Place: Auditorium 1253-211 (Merete Barker)

Press release (Danish)

Ny forståelse af den molekylære sygdomsmekanisme bag den sjældne sygdom arveligt angioødem

Et nyt ph.d.-projekt fra Aarhus Universitet, Health giver for første gang et bud på, hvad der forårsager arveligt angioødem, hvilket er afgørende for det videre arbejde med genterapi som et fremtidigt behandlingstilbud for patienter med arveligt angioødem. Projektet er gennemført af Didde Haslund, der forsvarer sin afhandling d. 10. maj 2019.

En spontan hævelse i armen, en opsvulmet mave, samt frygten for at næste hævelse kan være livstruende vanskeliggør hverdagen for patienter med arveligt angioødem. Hævelserne skyldes, at patienterne med arveligt angioødem mangler C1-inhibitor proteinet i blodet, hvilket gør blodkarrene utætte. Siden den første beskrivelse af sygdommen for godt 50 år siden har det været et mysterium, hvorfor disse patienter producerer så lidt af C1-inhibitor proteinet. Didde Haslund har i forbindelse med sit ph.d.-projekt kortlagt, hvad der sker i de leverceller som producerer C1-inhibitor proteinet og hvorfor patienterne mangler C1-inhibitor. Det viser sig nemlig, at de arvelige genmutationer, som patienterne har arvet fra den ene af deres forældre, resulterer i produktionen af et ændret C1-inhibitor protein. Og det er netop denne ændring, der forhindrer at almindeligt fungerende C1-inhibitor protein udskilles normalt. Således har Didde Haslund vist, at det normale gen påvirkes negativt af det muterede gen. Den nye cellulære forståelse af det manglende C1-inhibitor protein i blodet hos patienter med arveligt angioødem bringer patienterne det først skridt i retningen af genterapi som et fremtidigt behandlingstilbud.

Forsvaret af ph.d.-projektet er offentligt og finder sted den 10/5 kl. 13.00 i Auditorium 1253-211 (Merete Barker Auditoriet), Aarhus Universitet, Bartholins Allé 3, 8000 Aarhus C. Titlen på projektet er "Dominant negative intracellular retention of C1 inhibitor in hereditary angioedema". Yderligere oplysninger: Ph.d.-studerende Didde Haslund, e-mail: didde.haslund@biomed.au.dk, tlf. +45 3151 5190.

Bedømmelsesudvalg:

Coen Mass, Associate Professor, PhD
University Medical Center Utrecht, Clinical Chemistry and Hematology
Utrecht, The Netherlands

Lars Ellgaard, Professor, PhD
Section for Biomolecular Sciences, Department of Biology, Copenhagen Biocenter, University of Copenhagen,
Copenhagen, Denmark

Mette Nyegaard, Associate Professor, PhD (Chairman)
Department of Biomedicine, Aarhus University
Aarhus, Denmark

Press release (English)

New understanding of the molecular disease mechanism in the rare disease hereditary angioedema

A new PhD.-project from Aarhus University, HEALTH, provides for the first time a detailed description of the cellular defects that cause hereditary angioedema, which is essential for further work on gene therapy as a future treatment option for patients with hereditary angioedema. The project was carried out by Didde Haslund, who is defending her dissertation on May 10, 2019.

An arm that spontaneously swells to twice its normal size, a swollen stomach, and the constant fear that the next edema attack can be life-threatening make everyday life difficult for patients with hereditary angioedema. The swellings occur in patients with hereditary angioedema due to the lack of C1-inhibitor protein in their blood, which causes the blood vessels to leak fluid. Since the first description of the disease just over 50 years ago, it has been a mystery why these patients produce so little C1-inhibitor protein. During her PhD project Didde Haslund has identified events that take place in liver cells producing C1-inhibitor protein and described why angioedema patients lack the C1-inhibitor protein. It turns out that the genetic variations, which the patients have inherited from one of their parents, result in the production of a functionally altered C1-inhibitor protein. Precisely this change prevents normal C1-inhibitor protein from being secreted normally. Thus, Didde Haslund has shown that the normal gene is negatively affected by the mutated gene. The new cellular understanding of the lack of C1-inhibitor protein in the blood of patients with hereditary angioedema brings patients the first step in the direction of gene therapy as a future treatment option.

The defence is public and takes place on 05/10 at 1.00 PM in Auditorium 1253-211 (Merete Barker), Aarhus University, Bartholins Allé 3, 8000 Aarhus C. The title of the project is "Dominant negative intracellular retention of C1 inhibitor in hereditary angioedema". For more information, please contact PhD student Didde Haslund, email: didde.haslund@biomed.au.dk, Phone +45 3151 5190.

Assessment committee:

Coen Mass, Associate Professor, PhD
University Medical Center Utrecht, Clinical Chemistry and Hematology
Utrecht, The Netherlands

Lars Ellgaard, Professor, PhD
Section for Biomolecular Sciences, Department of Biology, Copenhagen Biocenter, University of Copenhagen,
Copenhagen, Denmark

Mette Nyegaard, Associate Professor, PhD (Chairman)
Department of Biomedicine, Aarhus University
Aarhus, Denmark

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