

## Media release

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### Basic information

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Department of: Clinical Medicine

Main supervisor: Prof. Claus Gravholt

Title of dissertation: Marfan syndrome - Diagnostics, epidemiology and aortic events

Date for defence: 14/3-2017 at (time of day): 14 Place: Konferencerum for afdelingen for Hjertesygdomme, Palle Juul-Jensens Boulevard 99, 8200 Aarhus N

Media release (Danish)

#### Opgørelse af sjældnen men farlig sygdom

Marfans syndrom er en sjælden arvelig sygdom der svækker bindevævet i kroppen. Patienter med Marfan syndrom kan, pga. bindevævspåvirkningen, få en svækkelse i hovedpulsårens væg der udvides og i yderste konsekvens kan bryde og medføre pludselig død.

Diagnosen Marfan syndrom er kompleks og stilles ud fra en kombination af kliniske, genetiske og billeddiagnostiske undersøgelser.

I et nyt ph.d.-projekt fra Aarhus Universitet, Health, afklares hyppigheden af Marfan syndrom i Danmark herunder i hvilken alder diagnosen stilles. I afhandlingen undersøges også hvor mange patienter med Marfan syndrom der har alvorlig sygdom i hovedpulsåren ved en opgørelse af antallet af patienter der har behov for en operation af hovedpulsåren eller hvor hovedpulsåren er bristet.

I afhandlingen undersøges desuden kvaliteten af de genetiske databaser der anvendes til at evaluere om patienter har genetiske forandringer der fører til Marfan syndrom.

Ph.d.-projektet er gennemført af læge Kristian Ketill Ambjørn Groth, der forsvare sin afhandling den d. 14/3-2017

Forsvaret af ph.d.-projektet er offentligt og finder sted den 14/3 kl. 14 i konferencerummet for Hjertesygdomme, Aarhus Universitets Hospital i Skejby, Palle Juul-Jensens Boulevard 99, Aarhus N. Titlen på projektet er "Marfan syndrome - Diagnostics, epidemiology and aortic events". Yderligere oplysninger: Ph.d.-studerende Kristian Ketill Ambjørn Groth, e-mail: [kristian.groth@clin.au.dk](mailto:kristian.groth@clin.au.dk), tlf. 20719356.

Media release (English)

#### Evaluation of rare and lethal disease

Marfan syndrome is a rare heritable disease affecting the connective tissue. As bloodvessels contains connective tissue patients with Marfan syndrome have weak arterial vessels and some patients experience dilatation of the aortic vessel and ultimately fatal rupture.

Diagnosing Marfan syndrome is complex and involves clinical, genetic and radiologic examination.

A new PhD project at Aarhus University, Health, examined the frequency of Marfan syndrome in Denmark and what age the patients are diagnosed. The thesis also examines the number of patients

with severe aortic disease by determining the number needing surgery of the aorta vessel or experienced a ruptured aorta.

The thesis also examined the quality of the genetic databases used to evaluate genetic changes causing Marfan syndrome.

The project was carried out by MD Kristian Ketill Ambjørn Groth, who is defending his dissertation on 14/3-2017.

The defence is public and takes place on 14/3-2017 at 14 o'clock in the Conference room at department of Cardiology, Aarhus University Hospital, Palle Juul-Jensens Boulevard 99, 8200 Aarhus N. The title of the project is "Marfan syndrome - Diagnostics, epidemiology and aortic events". For more information, please contact PhD student Kristian Ketill Ambjørn Groth, email: kristian.groth@clin.au.dkX, Phone +45 20719356.

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