AARHUS UNIVERSITY

Press release

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Basic information

Name: Agnethe Berglund Email: agnetheberglund@dadlnet.dk Phone: + 45 23 84 38 48

Department of: Clinical Medicine

Main supervisor: Professor, Claus Højbjerg Gravholt

Title of dissertation: The epidemiology of females with 46,XY disorders of sex development and males with 46,XX disorders of sex development: a nationwide study

Date for defence: December 14 2017 at (time of day): 14.00 Place: The Eduard Biermann Lecture Hall, Lakeside Lecture Halls, Aarhus University, Aarhus C

Press release (Danish)

Nyt dansk ph.d. studie kortlægger medfødte tilstande hvor genetiske piger fødes som drenge og genetiske drenge fødes som piger

Den typiske kønskromosomsammensætning for en mand er 46,XY medens den for en kvinde er 46,XX. Medfødte tilstande, hvor kvinder har den mandlige kønskromosomsammensætning 46,XY og mænd har den kvindelige kønskromosomsammensætning 46,XX forekommer, omend sjældent. Hidtil har viden omkring forekomsten af disse sjældne tilstande været begrænset, men et nyt nationalt ph.d.-projekt fra Aarhus Universitet har kortlagt forekomsten og fastslår at omtrent 6 per 100,000 nyfødte piger fødes som genetiske drenge mens omtrent 4-5 per 100,000 nyfødte drenge fødes som genetiske piger. I studiet har man også set på hvornår disse patienter typisk diagnosticeres, og medens størstedelen af pigerne/kvinderne blev diagnosticeret i den tidlige barnealder eller i slutningen af teenageårene blev størstedelen af drengene/mændene først diagnosticeret i voksenalderen i forbindelse med udredning for infertilitet. En del blev dog først diagnosticeret sent i livet. Udfra registerdata har man i studiet også undersøgt sygelighed, dødelighed samt socioøkonomiske forhold blandt disse patienter og sammenlignet med den danske baggrundsbefolkning. Ser man bort fra de følgetilstande, som helt forventligt er en del af det at være født med disse tilstande, var den generelle sygelighed og dødelighed ikke øget blandt patienterne, hvorimod både kvinderne og mændene var udfordret på forskellige socioøkonomiske parametre. Studiet er unikt, da det for første gang nogensinde beskriver en national kohorte af kvinder med kønskromosomsammensætningen 46,XY og mænd med kønskromosomsammensætningen 46,XX, hvorfor det også udgør et væsentligt bidrag til vores grundlæggende forståelse af disse sjældne tilstande. Projektet er gennemført af læge, Agnethe Berglund, der forsvarer det d. 14/12 2017

Forsvaret af ph.d.-projektet er offentligt og finder sted den 14/12 kl. 14.00 i Eduard Biermann Auditoriet (bygning 1252, lokale 204), Søauditorierne, Aarhus Universitet, Bartholins Allé 3, 8000 Aarhus C. Titlen på projektet er ”Epidemiologien for kvinder med 46,XY disorders of sex development og mænd med 46,XX disorders of sex development”. Yderligere oplysninger: Ph.d.-studerende Agnethe Berglund, e-mail: agnetheberglund@dadlnet.dk, tlf. 23 84 38 48.

Bedømmelsesudvalg:
Lars Rejnmark, professor, overlæge, Medicinsk Endokrinologisk Afdeling, Aarhus Universitetshospital, 8000 Aarhus C

Dorte Hansen, overlæge, klinisk lektor, H.C. Andersens Børnehospital, Odense Universitetshospital, 5000 Odense C

Syed Faisal Ahmed, professor, University of Glasgow, Academic Child Health, UK

Press release (English)

A new Danish ph.d. study benchmarkes congenital conditions in which genetic girls are born as boys and genetic boys are born as girls

The normal male sex chromosome constitution is 46,XY whereas it for females are 46,XX. Congenital conditions, where females have the male sex chromosome constitution 46,XY and males have the female sex chromosome constitution 46,XX occur, although rarely. Previously, knowledge concerning the frequency of these conditions has been sparse, but a new national ph.d. project from Aarhus University has estimated the frequency and concludes that around 6 per 100.000 newborn girls are born as genetic males, while around 4-5 per 100.000 newborn boys are born as genetic girls. The age at diagnosis of these patients has been studied, and where the main part of the girls/women were diagnosed during early childhood or in the late teens the main part of the boys/men were diagnosed in adulthood in relation to infertility treatment. A considerable part was, however, not diagnosed until late in life. Using registry data, morbidity, mortality and socioeconomics were studied in a comparison to the general population. Disregarding diseases, which are expected as a natural consequence of being born with these conditions, morbidity and mortality were not increased. However, both the men and the women were challenged when observing different socioeconomic parameters. The study is unique, as it is the first time that national cohorts of females with the sex chromosome constitution 46,XY and males with the sex chromosome constitution 46,XX are described, and thus it makes a considerable contribution to our general understanding of these rare conditions. The project was carried out by Agnethe Berglund, who is defending her dissertation on 14/12 2017.

The defence is public and takes place on 14/12 2017 at 14.00 in the Eduard Biermann Lecture Hall (building 1252, room 204), Aarhus University, Lakeside Lecture Halls, Bartholins Allé 3, 8000 Aarhus C. The title of the project is "The epidemiology of females with 46,XY disorders of sex development and males with 46,XX disorders of sex development. For more information, please contact PhD student Agnethe Berglund, email: agnetheberglund@dadlnet.dk, Phone +45 23 84 38 48.

Assessment committee: Consultant and clinical associate professor, Dorte Hansen, H.C. Andersen's Children's Hospital, Odense University Hospital, Denmark

Professor, Syed Faisal Ahmed, University of Glasgow, Academic Child Health, UK

Consultant and Professor, Lars Rejnmark, Department of Endocrinology and Internal Medicine, Aarhus University Hospital, Denmark

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