

Press release

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Basic information

Name: Maria Rasmussen Email: marirasm@rm.dk Phone: 45 55 06 09

Department of: Clinical Medicine

Main supervisor: Lone Sunde

Title of dissertation: "FETAL KIDNEY ANOMALIES: GENETICS, PREVALENCE, AND OUTCOME"

Date for defence: 31st of March at (time of day): 13:00 Place: Science Center Skejby,
Brendstrupgårdsvej 21A, 8200 Aarhus N

Press release (Danish)

Føtale anomalier af nyrerne

Anomalier af nyrerne kan ses ved 2. trimesterskanningen. På baggrund af skanningsfundene alene kan det være svært at forudse fosterets prognose.

Et nyt PhD-projekt fra Health, Aarhus Universitet har undersøgt genetiske årsager til føtale anomalier af nyrerne, prævalensen af fostre med anomalier af nyrerne samt betydningen af sådanne fund. Projektet er gennemført af Maria Rasmussen, som er læge og forsvarende hendes afhandling d. 31. marts 2017.

Som led i projektet har vi påvist en mulig genetisk årsag hos ét ud af fem fostre med svære anomalier af nyrerne. Vi har herunder identificeret nye gener knyttet til manglende udvikling af nyrerne.

Med udgangspunkt i de unikke danske sundhedsregistre har det været muligt at etablere en national kohorte af fostre med anomalier af nyrerne blandt alle fostre ultralydsskannet fra 2007 til 2012. En tredjedel af graviditeterne blev afbrudt. Blandt de børn, som var levendefødte, var der en overhyppighed af misdannelser i mave-tarm systemet. Vi har også vist, at en væsentlig andel af børnene, som var diagnosticeret med anomalier af nyrerne under graviditeten, havde en eller flere urinvejsinfektioner inden de fyldte to år.

Vores forskningsresultater styrker grundlaget for rådgivningen af forældre, som venter et barn med anomalier af nyrerne.

Forsvaret af PhD-projektet er offentligt og finder sted den 31. marts 2017 kl. 13:00, Science Center Skejby, Brendstrupgårdsvej 12A, 8200 Aarhus N. Titlen på projektet er "FETAL KIDNEY ANOMALIES: GENETICS, PREVALENCE, AND OUTCOME". Yderligere oplysninger: PhD-studerende Maria Rasmussen, e-mail: marirasm@rm.dk, tlf. 78 45 55 06 09.

Press release (English)

FETAL KIDNEY ANOMALIES

Fetal kidney anomalies may be diagnosed at the second trimester ultrasound scan. Based on the prenatal ultrasound findings only, it can be difficult to predict the prognosis.

A new PhD project from Health, Aarhus University, studied genetic causes, prevalence, and outcome of prenatally diagnosed kidney anomalies. The project was carried out by Maria Rasmussen, a medical doctor, who is defending her dissertation the 31st of March 2017.

In this recently completed PhD project, a possible genetic underlying cause was identified in one of five fetuses with severe kidney anomalies. Novel genes associated to failure of kidney development were proposed.

Using the unique Danish health registers, it was possible to identify a nationwide cohort of fetuses with kidney anomalies among all ultrasound-scanned fetuses from 2007 to 2012. About one third of these pregnancies were terminated. Additional malformations in the gastrointestinal system were found to be particularly prevalent among the live born infants. Furthermore, it was found that a substantial proportion of infants prenatally diagnosed with kidney anomalies experienced one or more urinary tract infections before the age of two years.

The findings in this project will strengthen the basis for parental genetic counseling, when kidney anomalies are diagnosed in a fetus.

The defence is public and takes place on Science Center Skejby at Brendstrupgårdsvej 12A, 8200 Aarhus N. The title of the project is "FETAL KIDNEY ANOMALIES: GENETICS, PREVALENCE, AND OUTCOME".

For more information, please contact PhD student Maria Rasmussen, email: marirasm@rm.dk, Phone +45 78 45 55 06 09.

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