

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: Ida Charlotte Bay Lund

Email: idalund@biomed.au.dk Phone: 2298 2281

Department of: Biomedicine

Main supervisor: Professor, Dr.Med, Ida Vogel

Title of dissertation: Analyzing Placental DNA -methods, clinical use, prevalence and complexity of placental mosaicism.

Date for defence: January 15, 2021 at (time of day): 2 PM Place: Zoom

Press release (Danish)

Fosterdiagnostik og moderkagens DNA

Moderkagens DNA har været hovedfokus i et nyt ph.d.-projekt fra Aarhus Universitet, Health.

Projektet er gennemført af læge Ida Charlotte Bay Lund, der forsvare det d. 15/1-2021.

Tidligt i graviditeten kan en moderkageprøve eller en blodprøve (Noninvasiv prænatal test, NIPT) fra den gravide anvendes til at diagnosticere sygdomme hos fosteret. Projektets opgørelse af nationale data viste, at de fleste danske gravide (80%), som blev tilbudt fosterdiagnostik i perioden 2013-2017, fik foretaget moderkageprøve frem for NIPT.

Resultater fra moderkageprøver er generelt meget pålidelige og mere præcise end resultater fra NIPT. I sjældne tilfælde, <4% viser projektet, indeholder moderkageprøven både en abnorm og en normal cellelinje – det kaldes mosaicisme. Mosaicisme er som oftest afgrænset til moderkagen, men kan være til stede i fostret og dermed give anledning til sygdom eller misdannelser. Projektet demonstrerer, at mosaicisme i moderkagen for mindre delmængder af kromosomer indebærer samme risiko for sygdom hos fostret som hele kromosomer. Undersøgelser på fødte moderkager med mosaicisme viste dog også, at der kan være mange forskellige samtidige forandringer i moderkagen, uden at disse er til stede i fosteret. Mosaicisme i moderkageprøver er derfor komplekst og skal udforskes nærmere.

Forsvaret af Ph.d.-projektet er offentligt og finder sted online den 15/1 kl. 14.00 via Zoom. Se venligst nedenstående kontaktoplysninger, hvis link til forsvar ønskes. Titlen på projektet er "Analyzing Placental DNA -methods, clinical use, prevalence and complexity of placental mosaicism". Yderligere oplysninger: Ph.d.-studerende Ida Charlotte Bay Lund, e-mail: idalund@biomed.au.dk, tlf. 2298 2281.

Bedømmelsesudvalg:

Erik Iwarsson, MD, PhD, Associate professor and consultant in clinical genetics Department of Molecular Medicine and Surgery and Center for Molecular Medicine, Karolinska Institutet, Karolinska University Hospital, Stockholm, Sweden.

Karin Sundberg, MD, DMSc, Consultant and Head of Center of Fetal Medicine and Pregnancy, Department of Obstetrics, Copenhagen University Hospital Rigshospitalet, Denmark.

Ole Mogensen, MD, DMSc, Clinical professor (chairman and moderator of the defence) Department of Obstetrics and Gynecology, Aarhus University Hospital, Aarhus, Denmark.

Press release (English)

Fetal diagnostics and placental DNA

Placental DNA is the center of a new project from Aarhus University, Health. The project was carried out by Ida Charlotte Bay Lund, who is defending her dissertation on January 15, 2021.

Early in pregnancy, placental DNA can be obtained through chorionic villus sampling but also through maternal blood sampling (Non-invasive prenatal test). National data from the project showed that the

majority of Danish pregnant women (80%), who were offered prenatal diagnostics in 2013-2017, had chorionic villus sampling carried out instead of NIPT.

Generally, analyses on chorionic villus samples provide reliable results and these results are more accurate than results from NIPT. However, in rare cases (<4%, data from the project), results from chorionic villus samples show two or more distinct cell lines; also known as mosaicism. Mosaicism is frequently confined to the placenta but may involve the fetus and cause disease or malformations. The project demonstrated that the risk of fetal mosaicism was equally high in cases of mosaicism for small chromosomal parts and whole chromosomes. Analyses on chorionic villus samples from postpartum placentas revealed that many different variations, without fetal involvement, can be found in the same placenta. Therefore, the complexity of mosaicism in chorionic villus samples is evident and needs to be explored further.

The defence is public and takes place online on January 15, 2021 at 2 PM on Zoom. The title of the project is "Analyzing Placental DNA -methods, clinical use, prevalence and complexity of placental mosaicism". For more information including online access to the defence, please contact PhD student Ida Charlotte Bay Lund, email: idalund@biomed.au.dk, Phone +45 2298 2281.

Assessment committee:

Erik Iwarsson, MD, PhD, Associate professor and consultant in clinical genetics Department of Molecular Medicine and Surgery and Center for Molecular Medicine, Karolinska Institutet, Karolinska University Hospital, Stockholm, Sweden.

Karin Sundberg, MD, DMSc, Consultant and Head of Center of Fetal Medicine and Pregnancy, Department of Obstetrics, Copenhagen University Hospital Rigshospitalet, Denmark.

Ole Mogensen, MD, DMSc, Clinical professor (chairman and moderator of the defence) Department of Obstetrics and Gynecology, Aarhus University Hospital, Aarhus, Denmark.

Permission

By sending in this form:

- I hereby grant permission to publish the above Danish and English press releases.
- I confirm that I have been informed that any applicable inventions shall be treated confidentially and shall under no circumstances whatsoever be published, presented or mentioned prior to submission of a patent application, and that I have an obligation to inform my head of department and the university's Patents Committee if I believe I have made an invention in connection with my work. I also confirm that I am not aware that publication violates any other possible holders of a copyright.