Press release

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Basic information

Name: Sofie Eg Jørgensen Email: segj@clin.au.dk Phone: 26746639

Department of: Clinical Medicine

Main supervisor: Trine Hyrup Mogensen

Title of dissertation: Studies on Primary Immunodeficiencies - Genetic susceptibility to severe influenza and NEMO branch-point mutation as a novel genetic mechanism of EDA-ID

Date for defence: 31.05.18 at (time of day): 13.00 Place: Læsesalen i Bartholin, bygning 1242 rum 211, Aarhus Universitet, Wilhelm Meyers Allé 4, Aarhus.

Press release (Danish)

Karakterisering af genetiske varianter hos patienter med svær influenza og identifikation af en ny genetisk mekanisme bag EDA-ID

Primære immundefekter forårsages af genetiske varianter, som nedsætter immunsystemets funktion og dermed øger risikoen for infektioner. Influenza virus infektion resulterer i de fleste tilfælde i mild sygdom, men kan hos nogle patienter udvikle sig til en alvorlig livstruende infektion. I første del af et nyt ph.d.-projekt fra Aarhus Universitet, Health, er tilstedeværelsen af genetiske varianter i det medfødte immunsystem blevet undersøgt hos patienter med svær influenza. Studiets resultater viser at genetiske varianter i immunreceptoren RIG-I dysregulerer immunresponset og fører til øget produktion af pro-inflammatoriske molekyler, dvs. molekyler, der inducerer en betændelsestilstand i kroppen. Denne betændelsestilstand kan være medvirkende til at influenza infektionen bliver mere alvorlig.

I anden del af ph.d.-projektet er en ny genetisk mekanisme bag immundefekten EDA-ID blevet identificeret. Studiet af en dansk patient med EDA-ID viste tilstedeværelsen af en branch-point mutation i patientens NEMO gen. Mutationen påvirker evnen til at producere korrekt NEMO protein, hvilket er essentielt for aktivering af et inflammatorisk respons under infektioner. Dette studie er det første, der beskriver branch-point mutationer som en mekanisme for udvikling af EDA-ID. Projektet er gennemført af Sofie Eg Jørgensen, der forsvarer det d. 31/5.

Forsvaret af ph.d.-projektet er offentligt og finder sted den 31/5 kl. 13 i Læsesalen i Bartholin, bygning 1242 rum 211, Aarhus Universitet, Wilhelm Meyers Allé 4, Aarhus. Titlen på projektet er ”Studies on Primary Immunodeficiencies - Genetic susceptibility to severe influenza and NEMO branch-point mutation as a novel genetic mechanism of EDA-ID”. Yderligere oplysninger: Ph.d.-studerende Sofie Eg Jørgensen, e-mail: segj@clin.au.dk, tlf. 26746639.

Bedømmelsesudvalg:
Professor Uffe Birk Jensen (Formand), Klinisk genetisk afdeling, Aarhus Universitetshospital, Aarhus, Danmark.
Professor Anne Puel, Genetique humaine des maladies infectioueses, Imagine Institute, Universite Paris Descartes-Sorbonne, Paris, Frankrig.
Lektor Christopher Duncan, Institute of Cellular Medicine, Newcastle University, Newcastle, England.

Press release (English)

Characterisation of genetic variants in patients with severe influenza and identification of a new genetic mechanism of EDA-ID

Primary immunodeficiencies are caused by genetic variants, which impair the immune system and thus increase the risk of infection. Influenza virus infection usually causes mild disease but can in some cases develop into a severe life-threatening infection. Genetic variants in the innate immune system in patients with severe influenza have been investigated in the first part of a new PhD project from Aarhus University, Health. The results indicate that genetic variants in the innate immune sensor RIG-I dysregulate the immune response leading to increased production of pro-inflammatory molecules. Increased inflammation can contribute to a more severe influenza disease course.

A new genetic mechanism of the primary immunodeficiency EDA-ID has been characterised in the second part of the PhD project. The study of a patient with EDA-ID identified a branch-point mutation in the NEMO gene. NEMO is essential for induction of inflammatory immune responses, and the mutation was demonstrated to impair production of the protein. The study is the first to describe a branch-point mutation as a mechanism of EDA-ID. The project was carried out by Sofie Eg Jørgensen, who is defending her dissertation on 31/5.

The defence is public and takes place on 31/5 at 13 o'clock at Læsesalen in Bartholin, building 1242 room 211, Aarhus University, Wilhelm Meyers Allé 4, Aarhus. The title of the project is "Studies on Primary Immunodeficiencies - Genetic susceptibility to severe influenza and NEMO branch-point mutation as a novel genetic mechanism of EDA-ID". For more information, please contact PhD student Sofie Eg Jørgensen, email: segj@clin.au.dk, Phone +45 26746639.

Assessment committee:
Professor Uffe Birk Jensen (Chairman), Department of Clinical Genetics, Aarhus University Hospital, Aarhus, Denmark.
Professor Anne Puel, Genetique humaine des maladies infectioueses, Imagine Institute, Universite Paris Descartes-Sorbonne, Paris, France.
Associate professor Christopher Duncan, Institute of Cellular Medicine, Newcastle University, Newcastle, England.

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