AARHUS UNIVERSITY

Press release

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Basic information

Name: Thomas Wisbech Skov Email: tws@biomed.au.dk Phone: 28148123

Department of: Biomedicine

Main supervisor: Jacob Giehm Mikkelsen

Title of dissertation: CRISPR/Cas9-based Gene Editing in Human Hematopoietic Stem Cells for Treatment of GATA2 Deficiency

Date for defence: 12/11-2025 at (time of day): 13:00 Place: 1262-101 Samfundsmedicinsk auditorium

Press release (Danish)

Ph.d.-forsvar om forskning i genetisk reparation af immundefekten GATA2-defekt

Ny forskning undersøger, hvordan genredigeringsteknologien CRISPR/Cas9 kan bruges til at korrigere den sjældne arvelige immundefekt GATA2-defekt. Afhandlingen udforsker mulighederne for en personlig og mere sikker behandling ved brug af patientens egne stamceller.

GATA2-defekt er en sjælden, arvelig immundefekt, der skyldes mutationer i genet GATA2. Den eneste nuværende behandling er baseret på allogen hæmatopoietisk stamcelletransplantation, hvor patienten transplanteres med stamceller fra en donor. Dette er en risikabel behandling med høj dødelighed, primært fordi donorcellerne kan genkende patientens raske væv som fremmed og angribe det. Denne afhandling undersøger muligheden for en behandling baseret på transplantation af patientens egne hæmatopoietiske stamceller, hvori den sygdomsfremkaldende mutation er blevet repareret. CRISPR/Cas9-teknologien er blevet anvendt til at undersøge forskellige måder at udføre denne genetiske reparation på. Afhandlingen beskriver optimering af sådanne genetiske korrektionsstrategier samt undersøger effekten af genetisk korrektion i hæmatopoietiske stamceller fra en patient med GATA2-defekt. Derudover belyses også potentielle uønskede bivirkninger ved genkorrektionen.
Projektet er gennemført af Thomas Wisbech Skov der forsvarer d. 12/11-2025.

Projektet er gennemført af Thomas Wisbech Skov der forsvarer d. 12/11-2025.
Forsvaret af PhD projektet er offenligt og finder sted d. 12/11-2025 klokken 13:00 I Samfundsmedicinsk Auditorium (1262-101), Bartholins Allé 2, 8000 Aarhus C. Titlen på projektet er "CRISPR/Cas9-based Gene Editing in Human Hematopoietic Stem Cells for Treatment of GATA2 Deficiency"
For yderligere oplysninger kontakt: Ph.d.-studerende Thomas Wisbech Skov, e-mail: tws@biomed.au.dk, tlf. 28148123.

Bedømmelsesudvalg:
Professor Matthew Porteus, Stanford University.
Associate Professor Eric Paul Bennett, Novo Nordisk.
Professor Christian Kanstrup Holm, Aarhus University.

Press release (English)

PhD defence on research in genetic repair of the immunodeficiency GATA2 deficiency

New research investigates how the gene editing technology CRISPR/Cas9 can be used to correct the rare hereditary immunodeficiency GATA2 deficiency. The dissertation explores the possibilities for a personalized and safer treatment using the patient's own stem cells.

GATA2 deficiency is a rare hereditary immunodeficiency caused by mutations in the GATA2 gene. The only current treatment is based on allogeneic hematopoietic stem cell transplantation, in which the patient is transplanted with stem cells from a donor. This is a risky treatment with high mortality, primarily because the donor cells can recognize the patient's healthy tissue as foreign and attack it. This dissertation investigates the possibility of a treatment based on transplantation of the patient's own hematopoietic stem cells in which the disease-causing mutation has been repaired. The CRISPR/Cas9 technology has been used to study different ways of performing this genetic repair. The dissertation describes optimization of such genetic correction strategies and investigates the effect of genetic correction in hematopoietic stem cells from a patient with GATA2 deficiency. In addition, potential unwanted side effects of the gene correction are also investigated.

The project has been carried out by Thomas Wisbech Skov, who will defend on 12/11-2025.

The defence is public and takes place on 12/11-2025 at 13:00 in Samfundsmedicisk Auditorium, Aarhus University, Bartholins Allé 2, 8000 Aarhus C. The title of the project is "CRISPR/Cas9-based Gene Editing in Human Hematopoietic Stem Cells for Treatment of GATA2 Deficiency". For more information, please contact PhD student Thomas Wisbech Skov, email: tws@biomed.au.dk, Phone +45 28148123.

Assessment committee:
Professor Matthew Porteus, Stanford University.
Associate Professor Eric Paul Bennett, Novo Nordisk.
Professor Christian Kanstrup Holm, Aarhus University.

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