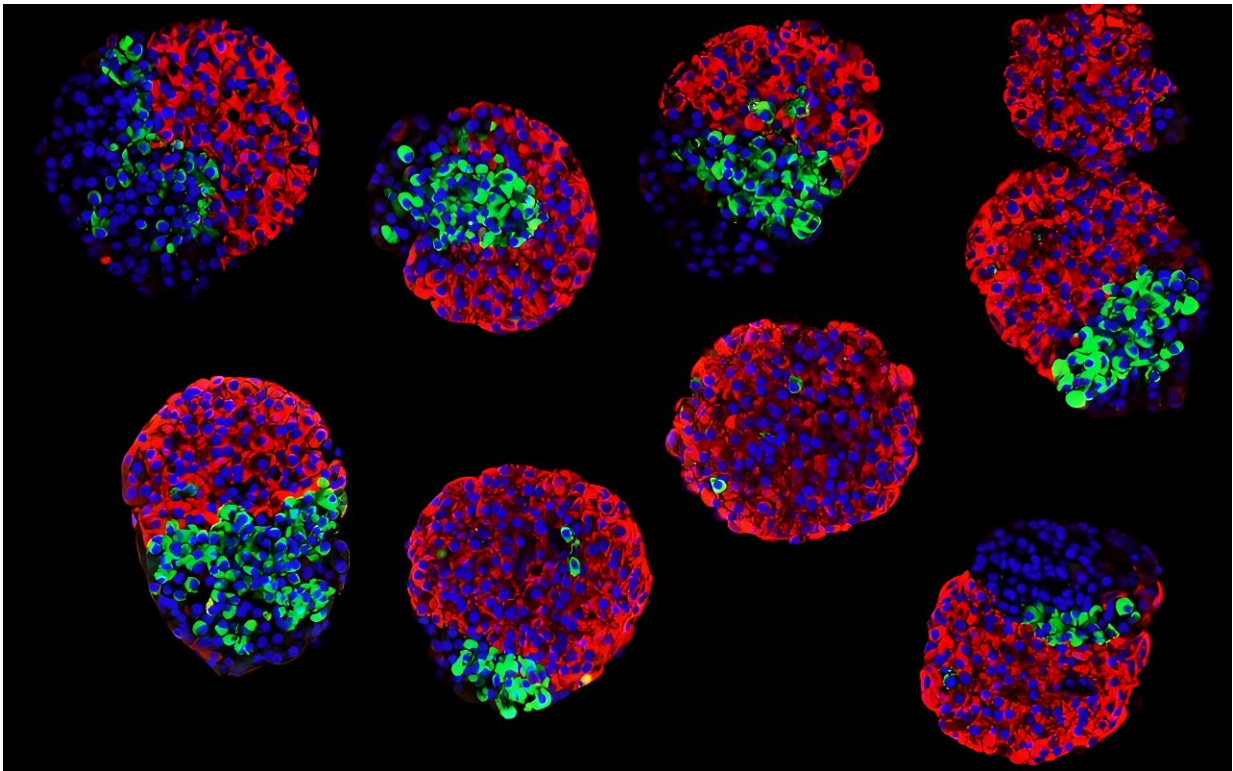


Study uncovers the link between RFX6 gene mutation and diabetes

July 3 2024



The study was conducted using stem-cell-derived islets. Credit: Hazem Ibrahim

A recent study by the University of Helsinki has uncovered that a mutation in the RFX6 gene significantly impairs insulin secretion, thereby elevating the risk of type 2 and gestational diabetes among carriers. Notably, this gene mutation is more prevalent in Finland than in

other parts of the world.

The regulatory factor RFX6 is crucial for the development of pancreatic beta cells, which are responsible for insulin production. Timo Otonkoski's research group employed genetically engineered stem cell models to explore, for the first time, how the RFX6 gene mutation influences the development and function of these essential beta cells.

Their findings revealed that while the gene mutation does not alter the number of beta cells, it does impair [insulin secretion](#), predisposing carriers to diabetes.

"Carriers of the RFX6 gene mutation face about double the risk of developing type 2 and [gestational diabetes](#)," explains Hazem Ibrahim, the lead researcher of the study. This discovery is particularly significant, as the [gene mutation](#) is enriched in the Finnish population, occurring in approximately one in every 250 individuals.

"This discovery enhances our understanding of the genetic mechanisms underlying diabetes and highlights the critical role of genetic factors in the disease's onset. Understanding these connections could pave the way for improved [prevention](#) and treatment strategies for diabetes in the future," Ibrahim adds.

The findings are [published](#) in the journal *Diabetologia*.

More information: Hazem Ibrahim et al, RFX6 haploinsufficiency predisposes to diabetes through impaired beta cell function, *Diabetologia* (2024). [DOI: 10.1007/s00125-024-06163-y](https://doi.org/10.1007/s00125-024-06163-y)

Provided by University of Helsinki

Citation: Study uncovers the link between RFX6 gene mutation and diabetes (2024, July 3)
retrieved 23 July 2024 from <https://medicalxpress.com/news/2024-07-uncovers-link-rfx6-gene-mutation.html>

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.