

Rare new form of diabetes discovered in newborns worldwide.

Scientists have discovered a rare form of neonatal diabetes caused by a mutation in the TMEM167A gene, opening up new avenues of research on insulin and diabetes.

A little-known gene turns out to play a crucial role in insulin production in the early stages of life.

Thanks to recent advances in DNA sequencing technology, combined with modern stem cell research methods, an international team of scientists has identified a previously unknown form of diabetes that appears in infancy.

Researchers at the University of Exeter School of Medicine, in collaboration with the Free University of Brussels (ULB) in Belgium and other organizations, have determined that mutations in the TMEM167A gene are the cause of this rare form of neonatal diabetes.

Diabetes can appear in some children within the first six months of life, and in over 85% of cases, the cause stems from inherited gene mutations. In a study led by the University of Exeter, scientists analyzed six children with neonatal diabetes who also had neurological problems such as epilepsy and microcephaly. The results showed that all of them shared changes in a single gene: TMEM167A.



The role of the TMEM167A gene in stem cell research and gene editing.

To clarify the function of this gene, Professor Miriam Cnop's research team at ULB used stem cells and directed them to develop into pancreatic beta cells – the type of cells responsible for insulin production. The team then applied CRISPR gene editing technology to manipulate TMEM167A.

The results showed that when the TMEM167A gene is disrupted, insulin-producing cells no longer function normally. This causes the cells to enter a state of prolonged stress and ultimately leads to cell death, severely impairing their ability to secrete insulin.

Dr. Elisa de Franco, a researcher at the University of Exeter, said that identifying the DNA changes that cause diabetes in infants offers a unique opportunity to find genes that play a key role in insulin production and secretion. According to her, discovering specific DNA alterations in six patients helped the team elucidate the function of a little-known gene, TMEM167A, and highlight its central role in regulating insulin secretion.

Professor Cnop also emphasized that the ability to create insulin-producing cells from stem cells has allowed scientists to study in detail the malfunctions in beta cells of patients with rare forms of diabetes as well as other types of diabetes. This is considered an extremely valuable research model for understanding disease mechanisms and testing new treatments.

Broader implications for diabetes research

This finding suggests that the TMEM167A gene is not only essential for the normal functioning of insulin-producing beta cells, but also plays a crucial role in nerve cells. Conversely, this gene appears to be less essential for many other cell types in the body.

These findings contribute to a better understanding of the key steps in insulin production and open up new avenues of research for other forms of diabetes, a disease currently affecting nearly 589 million people worldwide.

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