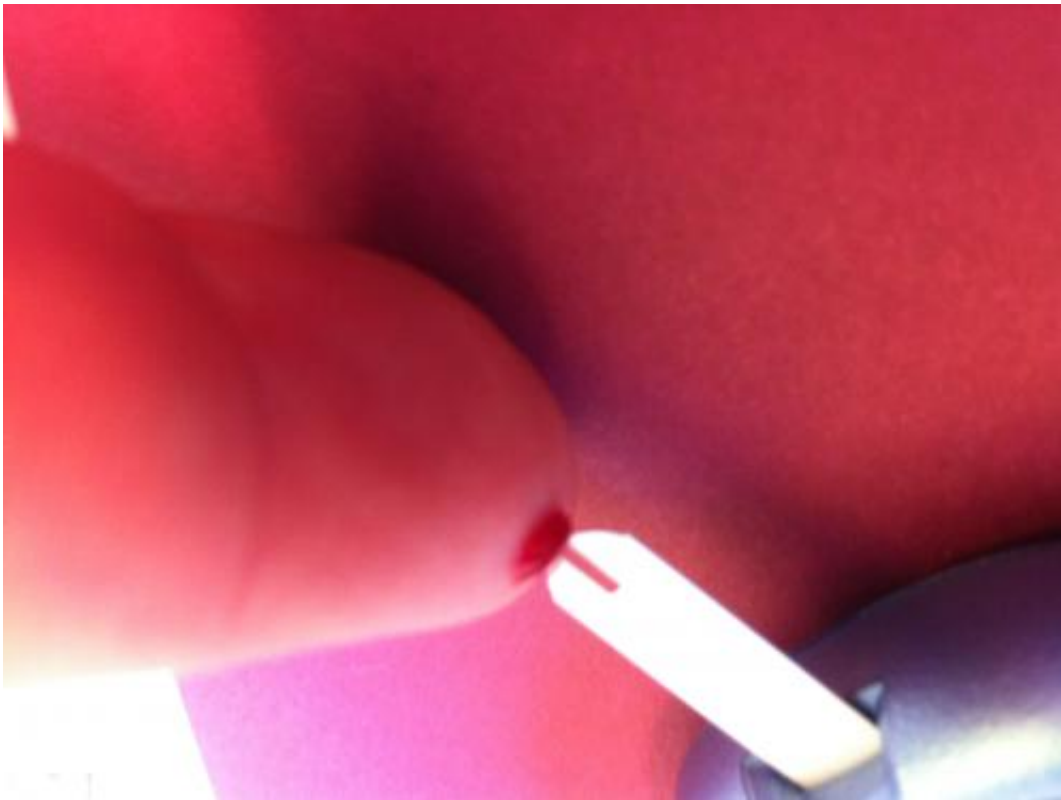


Discovery of human genetic mutation could lead to new treatments for type 1 diabetes

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This image shows a measurement of blood glucose by a patient with type 1 diabetes following a pricking of the finger with a lancet. Self-monitoring of blood glucose is recommended for people with type 1 diabetes before each injection of insulin and when hypoglycemia (low blood glucose) is suspected. Credit: *Cell Metabolism*, Bionis-Lauber et al.

In type 1 diabetes, the immune system destroys insulin-producing cells in

the pancreas, but the precise cause has not been clear. A study published by Cell Press on March 5th in *Cell Metabolism* reveals that a single mutation in the "longevity gene" SIRT1 can cause type 1 diabetes in humans. The findings unearth the role this gene plays in human autoimmunity and disease and also offer new avenues for treating a range of autoimmune disorders.

"We describe one of the first single gene defects leading to [type 1 diabetes](#), as well as the first human mutation in the SIRT1 gene," says senior study author Marc Donath of University Hospital Basel. "Our findings reveal a potential mechanistic basis for the development of a treatment for type 1 diabetes and other [autoimmune diseases](#)."

Type 1 diabetes is a lifelong, potentially fatal disease, typically diagnosed in young individuals, in which [beta cells](#) in the pancreas do not produce enough of the [hormone insulin](#), resulting in high levels of blood sugar. Animal studies have shown that a gene called SIRT1, which is well known for its role in promoting longevity and protecting against age-related diseases, also regulates [insulin secretion](#) and the development of autoimmune conditions, and activation of this gene can protect against type 1 diabetes. But until now, the role of SIRT1 in human autoimmunity and disease was not known.



This image shows a patient with type 1 diabetes injecting herself with insulin. Patients with the disease have to inject insulin before each meal. Credit: *Cell Metabolism*, Biason-Lauber et al.

In the new study, Donath and his team described a family carrying a mutation in the SIRT1 gene. All five affected members developed an autoimmune disorder, and four developed type 1 diabetes. Using a combination of gene-sequencing techniques, the researchers identified a previously undocumented mutation that caused an amino acid substitution in the SIRT1 protein. Moreover, inactivation of the SIRT1 gene in mice led to the destruction of the insulin-producing region of the pancreas, resulting in high levels of blood sugar.

"The identification of a gene leading to type 1 diabetes should allow us to understand the mechanism responsible for the disease and may open up new treatment options," Donath says. "To follow up on this study, we are creating a mouse that carries the mutation, with the hope of developing an animal model for human type 1 diabetes, and we are exploring the possibility of conducting a clinical study with SIRT1 activators."

More information: *Cell Metabolism*, Biason-Lauber et al.: "Mutation of SIRT1 in a Family with Type 1 Diabetes."
[dx.doi.org/10.1016/j.cmet.2013.02.001](https://doi.org/10.1016/j.cmet.2013.02.001)

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