

GENETIC MUTATIONS PREDICTIVE OF TYPE 2 DIABETES

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Technology description

BACKGROUND:

In recent years, there has been a dramatic increase in the incidence of diabetes worldwide. According to the International Diabetes Foundation, about 246 million people worldwide have diabetes and this number is expected to increase to 380 million by 2025. Type 2 diabetes accounts for about 90% of all cases. The current gold standard for diagnosing diabetes is a test for elevated blood sugar level following an overnight fast. Another test frequently used is the oral glucose tolerance test. However, these tests based on blood glucose can overlook approximately 25% of people who will develop type 2 diabetes, yet have normal blood glucose levels several years before diagnosis. Just in the US alone, 57 million people have pre-diabetes with blood glucose levels that are higher than normal but not high enough to be classified as diabetes. Recent research has shown that some long-term damage to the body, especially the heart and circulatory system, may already be occurring during pre-diabetes. Therefore, there is a need for assays to detect risk for type 2 diabetes a) prior to the onset of pre-diabetes as well as b) in individuals who have normal blood glucose levels.

DESCRIPTION:

Researchers at UCSF in collaboration with researchers at the University of Catanzaro have discovered mutations in a nuclear regulatory gene that are predictive of type 2 diabetes. The gene encodes a transcription factor necessary for cells to make insulin receptor, and low levels of insulin receptor lead to insulin resistance and type 2 diabetes. Researchers sequenced genomic samples from 3000 type 2 diabetes patients and 2000 sex and age matched non-diabetic controls. These mutations were observed in 10% of type 2 diabetes patients, while mutations were observed in less than half a percent of control patients. Since these mutations were 20-fold lower in controls, a robust diagnostic with high positive predictive value can be developed. Such a gene-based approach to assessing susceptibility to type 2 diabetes provides a more definitive diagnosis than today's diagnostics, allowing physicians to change patient care. Furthermore, genetic tests are accurate as early as infancy, when diet and exercise habits are being formed.

Application area

Genetic screening for type 2 diabetes

Design of treatment strategies for the subset of patients with nuclear gene mutations

Discovery and development of therapies for type 2 diabetes

Predictor of complications

Advantages

No other gene is predictive of as many type 2 diabetes cases

Gene-based approach predictive at early ages allows for early diagnosis and treatment

High positive predictive value diagnostic leads to change in care

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