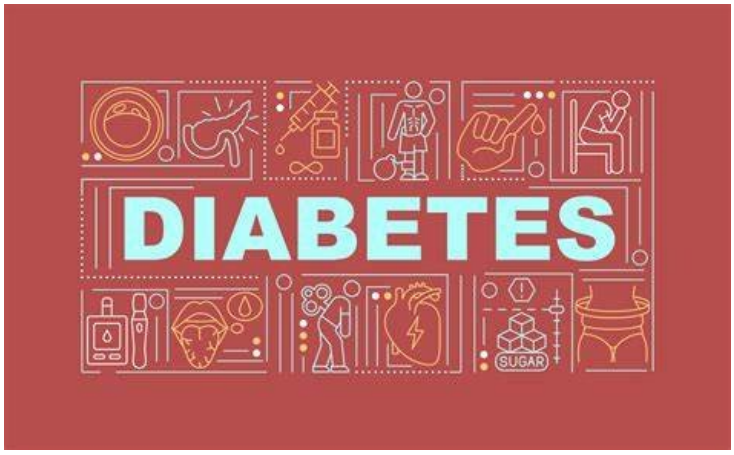
BioSpectrum
the business of Bio & Health Sciences

MDRF & US scientists discover new diabetes subtype in global breakthrough study

09 May 2025 | News

Newly identified genetic mechanism could revolutionise diagnosis and treatment of Maturity-Onset Diabetes of the Young (MODY)



In a major breakthrough, researchers from the Madras Diabetes Research Foundation (MDRF) in Chennai, and Washington University School of Medicine in St. Louis, Missouri, USA have jointly announced the discovery of a new subtype of MODY diabetes, potentially transforming how certain forms of the disease are diagnosed and treated worldwide.

The discovery centers around a rare, inherited form of diabetes called Maturity-Onset Diabetes of the Young (MODY), a genetic form of diabetes typically diagnosed in childhood and adolescence and has been published online ahead of print in the prestigious journal '*Diabetes'*, published by the American Diabetes Association.

The study, based on a collaborative investigation involving detailed genetic and functional analyses of Indian patients clinically diagnosed with MODY, reveals a groundbreaking mechanism behind a subtype of MODY affecting the ABCC8 gene which plays a crucial role in pancreatic ? cell function.

Prof Colin G Nichols, the lead researcher of this work from Washington University School of Medicine, St Louis, Missouri, USA states: "Through our collaborative work with MDRF, using various experiments in the laboratory, we were able to show some novel mutations in the Indian patients with MODY which occur as Loss Of Function (LOF). LOF mutations, abolish or reduce the activity of protein and they normally lead to Congenital Hyperinsulinism (CHI) which presents as persistent low blood glucose levels (hypoglycemia) in childhood."

Dr Radha Venkatesan, Executive Scientific Officer and Head of Molecular Genetics, who is the lead researcher of this work from MDRF, emphasizes the importance of the work and states, "Through our work in the lab and follow-up of our patients, we propose that diabetes driven by K_{ATP}-Gain of Function and K_{ATP}-Loss Of Function mutations should be officially be recognised as distinct disease subtypes, with different molecular basis and different clinical and therapeutic implications".

Dr V. Mohan, Chairman of MDRF adds, "Patients with this new (Loss of Function) MODY subtype do not respond to

Sulphonylureas unlike other forms of MODY like MODY 3 & MODY 1 & MODY 12. Further studies are needed to assess the best antidiabetic medicines to treat this novel type of diabetes. This study also opens up new avenues for discovery of novel drug targets in diabetes treatment".

MODY is a rare, inherited form of diabetes caused by mutations in a single gene, typically presenting in adolescents and young adults. While 13 MODY subtypes have been recognised so far, this newly identified variant upends long-standing assumptions about how the disease develops.

The discovery not only expands the scientific understanding of MODY but also underscores the urgent need for wider access to genetic screening — particularly in countries like India, where such testing is not yet part of routine diabetes care. This breakthrough could mark a turning point in advancing personalized diagnosis, treatment, and long-term management for thousands of individuals living with undetected or misclassified forms of diabetes.