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Monogenic Diabetes

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Abstract

Most children with diabetes have type 1 or 2. In these patients no single gene has been implicated and the genetic factors are limited to some single oligonucleotide variants, protective or predisposing HLA genotypes.

Over the last 2 decades, advances in molecular genetics led to identifying a small group of patients with diabetes due to single gene defect, the so called monogenic diabetes (MGD). MGD is rare; but could be under-diagnosed as patients often misdiagnosed as either type 1 or type 2diabetes. To date, mutations in > 40 genes have been associated with different forms of MGD. These mutations can either cause β-cell dysfunction leading to insulin deficiency or interfere with insulin receptors resulting in insulin resistance. The commonest forms of MGD are maturity onset diabetes of the young (MODY) and neonatal diabetes (NDM). In Arabs, NDM is more frequent than other populations and has different spectrum, while MODY appears to be rare or under-diagnosed.

Making the correct diagnosis of MGD has important implication as it can guide the best treatment for these patients. For example, individuals with GCK MODY don't require pharmacological therapies, while patients with HNF1A and KATP mutations can achieve better glycaemic control on sulphonylurea than insulin. This concept of personalized therapy (pharmacogenomics) is also shown in few patients with SLC19A2 diabetes in whom early genetic diagnosis led to starting thiamine and discontinuation of insulin. The presentation will highlight the genotype and phenotype of MGD with clinical examples of from the ASPED countries, where the concept of pharmacogenomics was vividly illustrated.