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MBD. Monogenic Beta-cell Diabetes

NIHR BioResource – Rare Diseases study project

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Summary

Patients with monogenic beta-cell diabetes get diabetes because of a change in a single gene which results in them producing a reduced amount of insulin. The condition can be broadly subdivided into two groups according to the age at onset; neonatal diabetes is diagnosed within the first 6 months of life whilst Maturity-Onset Diabetes of the Young (MODY) tends to be diagnosed in late childhood/early adulthood.

Prof Andrew Hattersley, MBD project Lead

At least 40 different genetic causes of monogenic diabetes have been described. These different genetic causes result in different types of diabetes and in some cases may also have additional clinical features. The rarity of these types of diabetes means we still have a lot to learn about their diagnosis, clinical features and treatment.

The NIHR BioResource will benefit patients by assisting research into how best to diagnose and treat the different sorts of monogenic beta-cell diabetes. It will also help identify suitable patients for clinical trials, aimed at assessing old and new therapies.

Recruitment Criteria

Inclusion

Individuals who have a confirmed genetic diagnosis of monogenic beta-cell diabetes (typically maturity-onset diabetes of the young (MODY) or neonatal diabetes): this would include all subtypes with the commonest being *GCK, HNF1A, HNF1B, HNF4A, KCNJ11, ABCC9, INS,* and *3243tRNA$_{leu}$*.