key points

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Maternally inherited diabetes and deafness (MIDD) is a rare form of diabetes, only affecting an estimated 0.1-0.2% of patients with diabetes in the UK. It is caused by an A to G (adenine to guanine) substitution at position 3243 of the mitochondrial DNA encoding the gene for tRNA.

Affected women pass the condition on to both daughters and sons. All their daughters and sons will inherit the mutation but due to phenotypic variance some will have both diabetes and deafness, some will only have deafness, some will only have diabetes and others may have no problems at all. However, affected men cannot pass the condition on to any of their offspring. It has high penetrance, with more than 85% of carriers developing diabetes during their lifetime.

Diagnosis is often challenging with some patients, as in this case, initially assumed to have type 1 diabetes; approximately 8% of patients present with diabetic ketoacidosis. Absent or only weakly positive GAD antibodies and/or a normal C-peptide level may suggest MIDD rather than type 1 diabetes. Cases may also be misdiagnosed as type 2 diabetes mellitus depending on age and presentation. The average age at diagnosis of MIDD is approximately 37 years but can range from 11 to 68 years.

Patients presenting with a type 2 diabetes picture can initially be treated with lifestyle modification with or without medication. However, metformin is contraindicated because of the risk of lactic acidosis. The majority of patients with MIDD will be insulin dependent within two years of diagnosis.

Bilateral sensorineural hearing loss occurs in most cases. The hearing loss typically develops in early adulthood and often precedes the onset of diabetes. Other complications include: rapidly progressive heart failure or arrhythmias; nephropathy due to focal segmental glomerular sclerosis; proximal myopathy; macular dystrophy; stroke; and constipation.