Mitochondrial diabetes- don't ignore clinical clues!

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Background

Mitochondrial diabetes has an incidence of 1%, is maternally inherited and leads to gradual beta cell failure. A high index of clinical suspicion and a strong family history may help establish the diagnosis.

Case presentation

An 18 year old, obese (BMI 30) female with recent onset of diabetes mellitus, was referred because of an atypical presentation and a strong family history of diabetes (mother, grandmother and great-grandmother). She was asymptomatic at presentation (no osmotic symptoms, tiredness or weight loss). However, fasting plasma glucose was 7.2 mol/l, and an oral glucose tolerance test confirmed diabetes. She was ketone free. Essential hypertension (on perindopril) and “fibromyalgia” (intense muscle pains and tenderness) had been diagnosed previously. Genetic screening established that she was negative for HNF and glucokinase mutations but she had the m.3243A>G mutation confirming the clinical suspicion of mitochondrial diabetes. Her HbA1c fell from 63 to 48 mmol/mol on metformin (MF) and diet alone. However, MF was withdrawn because of the risk of lactic acidosis, but her HbA1c deteriorated on sitagliptin, and MF was restarted as she wished to avoid sulphonylureas and insulin because of the risk of weight gain.

Discussion

(1) Diabetes mellitus is part of the clinical picture in subjects presenting with mitochondrial mutations. (2) Some have deafness, myopathy, lactic acidosis and heart disease associated with diabetes. (3) An atypical presentation (young and asymptomatic in our subject) coupled with a strong maternal family history should alert clinicians to the diagnosis. (4) Our patient had the commonest mutation identified with this syndrome. (5) Genetic counseling and family case detection should be undertaken and involvement of other systems should be excluded. (6) Beta cell dysfunction is progressive in this condition and many progress to require insulin at some stage.