Management challenges in familial partial lipodystrophy (FPLD): a case report

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We report a 48 year old Omani lady with a history of diabetes diagnosed at the age of 30. When she was seen in our Diabetes Clinic she had unsatisfactory glycaemic control (HbA1c 70mmol/mol) on metformin and glimepiride. Of note were previous history of hypertriglyceridaemia, hepatitis C, an episode of pancreatitis, and a strong family history of ischaemic heart disease.

On examination she had a body mass index of 18.2kg/m^2, marked reduction in subcutaneous fat (weight 42.6kg) and phlebectasia; features strongly suggestive of familial partial lipodystrophy (FPLD). Investigations revealed deranged lipid profile and hepatic steatosis.

She was commenced on basal insulin with resulting improvement in glycaemic control (HbA1c <53mmol/mol), but was noted to need high doses of over 70 units/day. Furthermore, finding suitable injection sites were difficult due to lack of subcutaneous fat. Glycaemic control was unpredictable. She transitioned to insulin pump treatment with new issues in locating suitable sites for glucose sensor insertions and inaccuracies in glucose readings.

Genetic analysis showed heterozygunits/dayosity for a missense mutation in *LMNA* pR482W; the commonest pathogenic mutation implicated in FPLD.

FPLD is an autosomal dominant condition characterised by gradual loss of adipose tissue beginning at puberty. FPLD has been reported primarily in European populations with cases described among those of African or Asian ethnicity. Diagnosis should be considered in patients who require disproportionately high insulin doses. Genetic diagnosis allows targeted screening for associated cardiac, renal and other complications, individualised treatment focusing on improving insulin resistance, and identification and early management of affected relatives.