

Lipoatrophy in a patient with type 1 diabetes: genetic or immunological?

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We report the case of a 28-year-old Emirati lady with type 1 diabetes (weight 51 kg, BMI 20.82 kg/m²) diagnosed at 3 years of age who presented to our centre with problems at her insulin injection sites. She had poor glycaemic control and her diabetes was complicated by retinopathy; other past medical history included vitamin D deficiency. All her seven siblings had diabetes and were on insulin; both parents had type 2 diabetes.

In 2017, she was switched from insulin glargine to insulin degludec and 7 months later she developed pronounced lipoatrophy (both upper frontal aspects of thigh) at the injection sites. She also developed hyperpigmented and hypertrophic reticular lesions on both axillae and neck which were histologically confirmed to be acanthosis nigricans/ reticular papillomatosis. She was treated with Minocycline and this resulted in some improvement.

Further investigations included serum leptin and C3 nephritic factor which were within normal range. Genetic testing showed a heterozygous variant in the Lipase E (LIPE) gene [c.746C>T p.(Thr249Met)] which was reportedly of uncertain significance. Attempted adipose tissue transplant was unsuccessful as transplanted tissue also atrophied within 2 weeks.

The lipoatrophy in this patient was severe and continued in spite of avoidance of injections at the lesions. It is likely that immunological factors are important in its aetiology. The heterozygous variant in LIPE gene is very rare and has not been previously described in the Emirati population. The strong family history suggests genetic cause for this patient's diabetes which may also be relevant in the aetiology of her lipodystrophy. In the reported case here, there were also major psychological and cosmetic sequelae which contributed to her poor glycaemic control which have proved challenging to treat.