Identification of an adrenaline receptor mutation in a family with atypical lipodystrophy

16 June 2016

Lipodystrophy syndromes are characterized by an abnormal distribution of adipose tissue and metabolic dysfunction. These disorders are rare and the underlying genetic abnormalities that lead to altered fat distribution are not fully known.

In this issue of *JCI Insight*, Abhimanyu Garg and colleagues at UT Southwestern Medical Center identify a genetic mutation that caused atypical lipodystrophy in a single family. Affected individuals had a marked loss of fat from the limbs but excessive accumulation of adipose tissue in the face and neck.

Additionally, family members with this lipodystrophy went on to develop metabolic complications. Genome sequencing revealed the presence of a heterogeneous mutation in the gene encoding adrenoceptor ? 2A (ADRA2A), which reduces the breakdown of lipids in adipocytes when activated by adrenaline. The mutation resulted in loss of ADRA2A function, which could explain excessive fat loss in some adipose deposits in these individuals.

The results of this study add to the list of known lipodystrophy-causing mutations.

More information: Abhimanyu Garg et al, Whole-exome sequencing identifies ADRA2A mutation in atypical familial partial lipodystrophy, *JCI Insight* (2016). DOI: 10.1172/jci.insight.86870