**Genetic Testing for Hypertriglyceridaemia in the UK**

Georgios Kounidas

Lipoprotein lipase deficiency (LPLD) is a rare disorder of lipid metabolism characterised by severe elevation of plasma triglycerides/chylomicrons. Between 2013 and 2019, NHS Grampian hosted the only genetic testing service for suspected cases of the condition in the UK. We aimed to report the "real world" genetics data by retrospectively reviewing laboratory records for cases from the UK, referred to the national centre for LPLD genetic testing (LPL, APOC2, GPIHBP1, APOA5 and LMF1). A total of 292 referrals requesting LPLD analysis were identified between November 2013 and June 2019; 181 were from cases in England, 99 patients from Scotland. Primary referral reason [N=186] was confirmed hypertriglyceridemia/hyperlipidaemia on biochemical testing, followed by pancreatitis [N=66]. The mean age at referral was 42 and most patients were male (62%). The majority, in whom LPLD was confirmed, had homozygous (35.5%) or heterozygous (64.5%) variants in LPL (62/108), and homozygous (10.7%) or heterozygous (89.3%) variants in APOA5 (28/108). This is the first comprehensive review of LPLD mutations from the UK. In contrast to previously reported series, mutations in genes other than LPL were found to be common, reflecting the outbred nature of the UK population, and highlighting the utility of genetic diagnosis in LPLD.

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