

Fat, Cholesterol and Genetics 2019

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Genetic lipoprotein disorders are uncommon clinical entities. Chylomicronemia is very rare but can lead to acute pancreatitis. The combination of chylomicronemia with severe hypertriglyceridemia in the context of diabetes, obesity and poor lifestyle is a major risk factor for ASCVD and a therapeutic challenge. The Canadian and US guidelines recommend lifestyle and statin therapy. Lipoprotein Lp(a) is considered a major cardiovascular risk factor for aortic stenosis and ASCVD. Current treatment is statin therapy and novel therapies, (anti-sense RNA therapies) are being developed for elevated Lp(a). Familial hypercholesterolemia is the most frequent genetic disorder seen in clinical practice. The prevalence is 1/250 in the general population and approx. 1/125 in the acute cardiac population. Tools for the diagnosis of FH are now available. Treatment consists of statins +/- ezetimibe. New guidelines recommend a target LDL-C level of < 2.0 mmol/L in ASCVD patients. The use of PCSK9 inhibitors is recommended in patients who do not reach their target levels.