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FCS: Enhancing the Diagnosis

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Dr. Gaudet:
This is a CME on PACE-CME and ReachMD, and I'm Dr. Daniel Gaudet from University of Montreal. Let's dive right into the key characteristics of familial chylomicronemia syndrome.

Recently, the National Lipid Association and the American Society of Preventive Cardiology agreed on a joint expert consensus on persistent chylomicronemia. The most severe form of persistent chylomicronemia is familial chylomicronemia syndrome, FCS, which is an ultrarare autosomal recessive disease where TG levels are sustainably above 880 mg/dL, or 10 mmol/dL, as it's associated with increased acute pancreatitis risk and other morbidities.

In general, it's non-responsive to most traditionally available TG-lowering treatments, including fibrates and omega-3. It's an unmet therapeutic need. However, recently, olezarsen, an APOC3 antisense oligonucleotide, has been approved in the US, whereas plozasiran, an APOC3 small interfering RNA received a new drug application acceptance in the US.

Clinical signs and symptoms of FCS include recurrent abdominal pain, acute and potentially lethal pancreatitis, eruptive xanthomas, lipemia retinalis, hepatosplenomegaly, hepatic steatosis, vigilance issues, and other complications. It hugely affects the quality of life of the patients at work, with the family, the diet is extremely restrictive, traveling is an issue, insurance is an issue. And if you think of having an acute pancreatitis, just think of the risk of having recurrent acute pancreatitis, which is one of the most severe pain that an individual can suffer. It's kind of a Damocles sword over the head of the affected patients.

Genetic testing and FCS clinical diagnosis scores are available and may help to distinguish FCS from other causes of severe hypertriglyceridemia and persistent chylomicronemia. There has been a recent North American clinical diagnosis scoring system published.

So as a conclusion, the key takeaway message here is that familial chylomicronemia syndrome is a severe form of persistent chylomicronemia associated with a huge risk of acute pancreatitis. When you meet a patient with TG levels above 880 mg/dL, sustainably, think of the possibility of familial chylomicronemia syndrome.

That's all the time we have today. I hope this has been valuable to you, and thank you very much for listening in.

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