Is your patient a potential candidate for FCS genetic testing?

Assign your patient a score for each familial chylomicronaemia syndrome (FCS) parameter, calculate the total score and then refer to the table on the right.

FCS PARAMETER	Score		Your patient's score
	Yes	No	
TRIGLYCERIDES			
Has the patient had fasting TGs of >10 mmol/L for three consecutive blood analyses?	+5	0	
Has the patient had fasting TGs of >20 mmol/L at least once?	+1	0	
Has the patient had fasting TGs of <2 mmol/L at least once?	-5	0	
MEDICAL HISTORY			
Does the patient have a history of pancreatitis?	+1	0	
Does the patient have unexplained recurrent abdominal pain?	+1	0	
Does the patient have a family history of familial combined hyperlipidaemia?	0	+1	
DIFFERENTIAL DIAGNOSIS			
Have you excluded secondary factors (except pregnancy and ethinylestradiol)?*	+2	0	
Has the patient failed to respond to hypolipidaemic treatment (TG decrease <20%)?	+1	0	
How old was the patient when their symptoms first appeared?			
≥40 years	0	0	
<40 years	+1	0	
<20 years	+2	0	
<10 years	+3	0	
		TOTAL	

* Secondary factors for hypertriglyceridaemia include alcohol consumption, diabetes, metabolic syndrome, hypothyroidism, corticotherapy and additional drugs. If diagnosis is made during pregnancy, a second assessment is necessary to confirm diagnosis post-partum.

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The likelihood of a patient having FCS can be determined on the basis of clinical presentation. FCS is diagnosed by presence of homozygous or compound heterozygous mutations in the following genes: *LPL, APOC2, APOA5, LMF1* or *GPIHBP1*.[1] FCS is characterised phenotypically by the presence of key signs and symptoms.[1,2]

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Score	
≥10	FCS very likely
=9	FCS unlikely
≤8	FCS very unlikely

References:
1. Moulin P, *et al. Atherosclerosis.* 2018;275:265–272.
2. Hegele R, *et al. Curr Opin Lipidol.* 2015;26(2):103–113.

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