Helping patients identify if familial chylomicronemia syndrome (FCS) is the reason for their symptoms

Some people with extremely high triglycerides (a type of fat) have an inherited condition called familial chylomicronemia syndrome (FCS). FCS is caused by genetic changes (also known as *disease-causing variants* or *mutations*) in one of six genes.

Most commonly, FCS is due to genetic changes in the *LPL* gene. The *LPL* gene makes a protein, called *lipoprotein lipase*, that helps break down triglycerides. In people with FCS, this gene does not work as it should, and fat builds up in the blood leading to certain health problems. Previously, FCS was known as *lipoprotein lipase deficiency* or *LPL deficiency*.

Variants in five other genes may also lead to FCS: *APOA5, APOC2, GPD1, GPIHPB1,* and *LMF1*[1].

Genetic testing looks for *variants* in the genes that may cause FCS.

The purpose of genetic testing for FCS is to:

1. Confirm an FCS diagnosis
2. Guide treatment and management

continue on reverse >

1 Hegele RA, Ginsberg HN, Chapman MJ, et al. The polygenic nature of hypertriglyceridaemia: implications for definition, diagnosis, and management. Lancet Diabetes Endocrinol. 2014;2(8):655-666.

For more information about Genetic Testing visit **FCSFocus.com/genetic-testing**

Since not all genetic tests are the same, speak with your doctor or a genetic counselor to understand what test is right for you.

FCS Genetic Testing

FCS focus

Get your no-cost genetic test now

The FCS Genetic Testing program is confidential and available at no-cost to certain patients. Talk with your healthcare provider to see if you qualify for the program. You qualify if you have:

- Extremely high (severe) triglyceride levels (more than 750 mg/dL (or 8.4 mmol/L)), on two fasting blood tests in a row.
- No other known causes of high triglyceride levels.

Receive genetic counseling before and after testing

The FCS Genetic Testing program has partnered with GeneMatters to provide information on familial chylomicronemia syndrome, as well as support surrounding the genetic testing process.

The FCS Genetic Testing program offers no-cost genetic counseling through GeneMatters, an independent genetic counseling service.

How to get genetic testing for FCS

- ❑ Make an appointment with a healthcare provider to discuss genetic testing.
- ❑ Share the **FCSFocus.com/genetic-testing** web pages with your healthcare provider. Your healthcare provider will check if you qualify for the no-cost, confidential FCS Genetic Testing program.
- ❑ If you qualify, your healthcare provider can order a genetic test at **Akcea.PreventionGenetics.com.** Testing involves either a saliva sample, an oral swab or a blood draw.
- ❑ All tests can be done at your doctor's office, or a saliva kit or oral swab kit can be mailed directly to your home.
- ❑ If a saliva kit or oral swab kit is sent to your home, the kit will arrive a few days after ordering. Follow instructions for collection and how to mail the kit to the lab.
- ❑ Testing takes about 3-4 weeks. When results are available, talk with your healthcare provider. You may be scheduled to talk with a genetic counselor about your results and what they mean.
- ❑ To learn more about FCS genetic testing, visit **FCSFocus.com/genetic-testing** or call the GeneMatters hotline number **+1 888.478.1494**.