

# Familial Lipoprotein Lipase Deficiency (LPLD/Chylomicronaemia)

LPLD is a rare condition that affects about one in a million of the population. People who have the condition are unable to produce enough lipoprotein lipase, the enzyme which breaks down fats that enter the bloodstream after a meal. To stay well, people with this condition need to eat a diet with a very restricted fat content. The amount of fat a person with LPLD can tolerate varies from individual to individual, but ranges from 5g-20g a day. For people without LPLD, a healthy level of fat intake in the UK is considered to be no more than 70g a day for women and 95g for men.

## How are people affected?

LPLD affects each person differently and some people are more badly affected than others. Because it is so rare, most consultants only ever see a few cases, so it is important that each person with LPLD works with their consultant to find the best diet and treatments for them.

## Why does dietary fat cause a problem

Because fats do not normally mix with water they are carried around the blood stream in particles called lipoproteins (mixtures of proteins and fats).

Chylomicrons are the largest of these lipoprotein particles. They appear in the circulation shortly after eating fatty foods. Normally they are cleared from blood over a few hours. In LPLD, clearance of chylomicrons from the blood (or plasma) is impaired.

Triglycerides are the major fatty component of chylomicrons. The failure to break down chylomicrons results in high levels of triglyceride accumulating in the blood. This gives the plasma a milky appearance.

## Diagnosis

LPLD usually starts showing effects in childhood, with periods of stomach pain, repeated attacks of pancreatitis, yellow spots (eruptive xanthomata) and periods where the liver and/or spleen are enlarged, causing pain. Males and females are equally affected.

Approximately a quarter of affected children show signs before the age of one year, and the majority develop symptoms before 10, but some women only show symptoms during pregnancy. Because this condition is so rare, it is possible that people with LPLD may not be formally diagnosed until adulthood.

The biggest concern is that high levels of triglycerides (usually over 2000mg/dl or 22.6mmol/l) can cause acute pancreatitis, although how they do this is not completely understood. Raised triglycerides are also a precursor for developing diabetes in later life.

## How did I get it?

LPLD is an autosomal recessive condition. This means that to have LPLD you need to have inherited two faulty LPL genes (one from your mother and one from your father). A faulty gene is often referred to as an altered gene, or gene mutation. Most parents of a child or adult with LPLD will not have the condition themselves, but both parents are carriers. Having one altered LPL gene does not result in LPLD – this is the meaning of the word 'recessive'.

## Can I drink alcohol?

The medical advice is that alcohol should be avoided because the process of metabolising alcohol in the liver produces fat which then can't be dealt with by patients with LPLD. However, it is possible to drink moderately, with no ill effects, as long as the rest of the diet is good. Beware that alcohol can cloud your judgement about what is acceptable to eat, and that hangovers may produce cravings for forbidden foods. You will need to find the right food/alcohol balance for you.

### What are the chances of passing it on?

If both parents are carriers:

Each child born to parents who are both carriers of the altered gene will have

- A 25% chance (1 in 4) of developing the condition (ie the pair of genes they inherited would have been the disease-causing gene from both parents)
- A 50% chance (1 in 2) of being carriers of the disease (the disease-causing gene plus the healthy gene, one from each parent)
- A 25% chance (1 in 4) of being completely clear of the disease (inheriting the healthy gene from each parent)

### If one parent has LPLD will they pass it on?

Individuals with the condition will have two disease-carrying genes

- Individuals with the condition will have two faulty LPL genes
- If they have a child with a partner who is a carrier (difficult to assess the number of carriers, as there are no outward signs, but not common) each child will have a 50% chance (1 in 2) of having the condition, and a 50% chance (1 in 2) of being a carrier
- More likely, the individual with LPLD will have a child with an individual who is disease-free, meaning the child will be a carrier, but will run no risk of developing the condition themselves

### If I am a carrier does it affect my health?

People who are carriers for this condition will not present any problems but may have mildly raised blood fat levels (mild hypertriglyceridaemia) and may be at risk for premature atherosclerosis (hardening of the artery wall) and heart disease. If a member of your family has the condition there are tests available to determine whether you are a carrier. You will need to ask your GP.

### What would happen if I relaxed my diet for a couple of days?

This depends on the individual. Some people with LPLD are very sensitive to any extra fat in their diet, whereas others seem to be able to tolerate quite high levels of fat without any noticeable consequence. Symptoms also differ. Some will experience periods of abdominal pain (which isn't pancreatitis) others report being sick, while others feel nothing until they have an attack of pancreatitis.

There are some reports that high fat levels result in concentration difficulties and breathlessness, whilst others notice nothing. The condition isn't understood well enough to answer this question fully, but it is safe to say that relaxing the diet for a few days, on the whole, won't cause any immediate difficulties other than feeling unwell. This is what makes living with the condition very hard, especially for children. It can be difficult for them to make the link between increasing dietary fat and feeling bad a few days later. The 'just a little bit won't hurt' type approach, on the whole is not advisable.