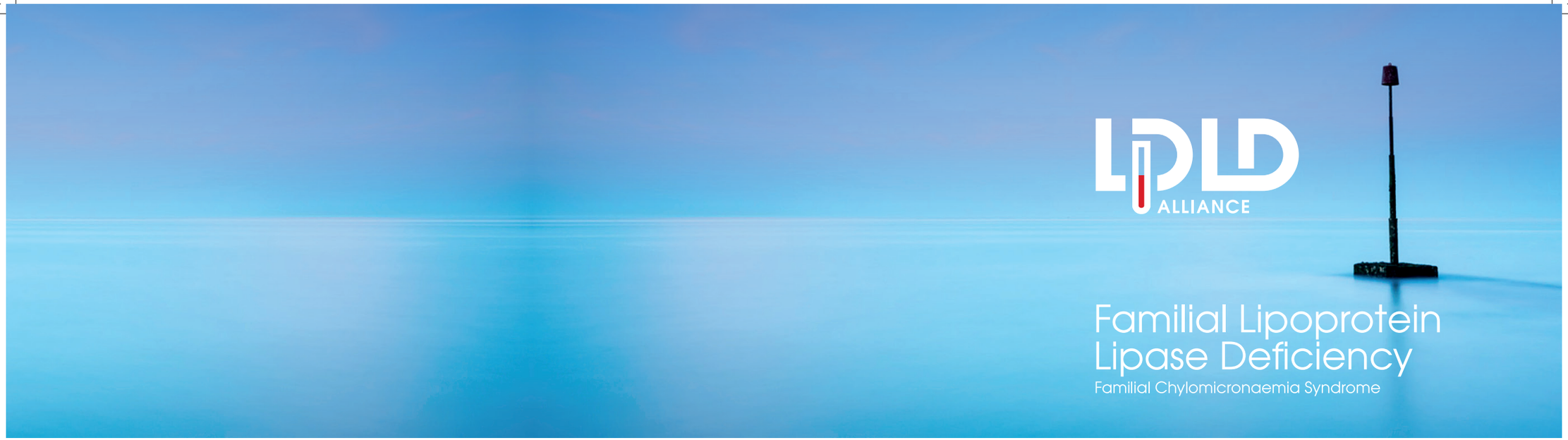

Familial Lipoprotein Lipase Deficiency

Familial Chylomicronaemia Syndrome

For healthcare professionals

LPLD/FCS is ultra-rare and under-diagnosed. Once diagnosed people can struggle to manage the restrictions it imposes and are often isolated.

We provide information on symptoms and on the day-to-day experience of living with LPLD/FCS to increase understanding of the burden placed on these individuals and those around them. We have a growing list of professionals with an interest and expertise in the condition, and information on medicine developments and treatment options.

We would be grateful if you could help us to ensure that every person with LPLD/FCS is aware of us. Please help them to access their community by passing on a copy of this leaflet.

www.lpldalliance.org

Want to find out more?

Visit our website at:
www.lpldalliance.org

Email us at
jill@lpldalliance.org

Ring us on:
07517 752168

Visit our Facebook page:
@LPLD Alliance

Follow us on Twitter:
@LpldAlliance

Join the LPLD Community on Rareconnect
www.rareconnect.org

RareConnect.org
A EURORDIS INITIATIVE

LPLD Alliance is a charity registered in England and Wales
Charity number 1165873

If you have Lipoprotein Lipase Deficiency (LPLD) or its related condition Familial Chylomicronaemia Syndrome (FCS) LPLD Alliance is here to help.

We raise awareness of this little-known condition among health professionals and the wider public.

We are building a supportive community of people with or with an interest in the condition.

We advocate for excellent care and access to new medicines.

www.lpldalliance.org

What is LPLD?

Familial lipoprotein Lipase Deficiency is an ultra-rare genetic condition that affects 1 to 2 people in a million. It affects men and women equally.

People with LPLD have difficulty properly digesting certain fats because they lack, or have limited amounts of, the enzyme lipoprotein lipase.

If too much fat is eaten it does not get cleared from the body and instead travels in the bloodstream giving it the typical creamy white appearance, the most obvious symptom seen by a doctor.

What is FCS?

Familial Chylomicronaemia Syndrome is the name given to all the genetic conditions which result in limited ability or an inability to metabolise fat. People with LPLD account for about 80% of those with FCS.

How do I know I have LPLD/FCS?

LPLD/FCS is most commonly identified in early childhood when the baby fails to thrive and has abdominal pain. A simple blood test shows the creamy white appearance of the blood. LPLD/FCS can be diagnosed later in life.

Women can sometimes be diagnosed during pregnancy when their blood is being monitored and high levels of triglycerides are noticed or if they suddenly have pancreatitis for seemingly no reason.

How much fat can I eat?

The amount of fat that someone can eat and remain symptom-free seems to vary from person to person. The recommended range is anywhere between 10g to 25g of any type of fat daily.

What happens if I eat too much fat?

Symptoms of LPLD/FCS range from mild to severe. Pancreatitis is the most severe - and potentially life-threatening. People with LPLD/FCS can also experience severe abdominal pain, an enlarged liver, yellow spots (xanthoma, fatty deposits), and report high levels of fatigue, lack of concentration, poor memory and depression.

Diabetes

People with LPLD/FCS are at a higher risk of developing Type 2 Diabetes, especially after repeated attacks of pancreatitis. High blood glucose levels are turned to fat in the body increasing the effect of the LPLD/FCS. Having diabetes restricts the amount of simple sugars and carbohydrates that can be eaten.

Pregnancy

Pregnancy can be difficult for women with LPLD/FCS as triglyceride levels rise naturally in the third trimester (6-9 months). Close monitoring and a very strictly controlled fat intake is recommended. Women need to be monitored for gestational diabetes which can exacerbate triglyceride levels.

Treatment

Currently the only effective treatment for LPLD/FCS is restricting the intake of all types of fat.

A number of new medicines for these conditions are at various stages in the medicines development process. We are working with all the relevant stakeholders to ensure patients have access to these medicines as soon as is possible.

Support

Through our website, our private Facebook group (LPLD Discussion Community), and the online community we host on Rare Connect (the LPLD Community), we encourage people to connect with others living with the same restrictions, providing the opportunity to share experiences and to give and receive support.

Meeting others

We provide opportunities for people affected by LPLD/FCS to meet each other and to learn more about this condition and its management.

Further details about our meetings may be found on our website.

Website

Our website has up-to-date information on LPLD/FCS, on managing the necessary dietary restrictions and on current treatment option. The website invites people affected by the condition to share tips and recipes and to add their own story.

www.lpldalliance.org