



Patient story

Jill, now aged 47 years, explains what it was like to grow up with lipoprotein lipase deficiency (LPLD) and how she copes with this rare disease.

“Being in the hospital for three months at the age of two had a big psychological impact on me. Today, I manage my diet very well but all the restrictions exclude me from a lot. I miss spontaneity. Eating out, especially with my children, is almost impossible. I find traveling anywhere difficult, particularly abroad.

As a baby I would stop feeding for days and arch my back to stretch out my stomach area. Doctors didn't know what was wrong. When I was two my mum gave me lots of chocolate buttons to stop my incessant crying, and I eventually came out in yellow spots. That was when I was taken into hospital, where I stayed for three months. This had a big and lasting impact on me, especially as in those days parents were not allowed to stay with their children. After I was diagnosed, my mum had frequent contact with dieticians and was given an eating plan for me. However, in the sixties, before the days of food labeling and nutritional analyses, it was incredibly difficult to know which food was low-fat. Skimmed milk had to be ordered specially from the milkman.

There are no visible signs of this disease (apart from the spots) but it made me feel different, isolated and misunderstood. I was asked to do the same things as anybody else, but I had this huge burden of trying to keep myself healthy. When you are young that is incredibly difficult. When I was at university, everybody was eating fast food and cooking for each other.

You need to be a strong character to deny and explain yourself all the time. I didn't keep to the diet as I should have, and was ill all the time but I was never hospitalized when I had severe stomach aches. I was so aware that it was because I had eaten the wrong food that I felt too guilty to go to the doctor. Instead, I self-medicated and stayed in my room. I wouldn't eat for five days and took painkillers.



It's incredibly disruptive to your relationships and life generally when you have to suddenly disappear. No support was offered to help me to deal with the issues that made managing the diet difficult. A period of extreme depression when I was 27 was picked up by my GP, who started me on therapy, which after several years has gotten me where I am today.

I work hard to stay healthy but I have to plan almost everything and have to exclude myself from many things.

Since I got married and settled down my health has improved dramatically. The most difficult time in this period has been having children – I have three. I couldn't breastfeed, as my milk doesn't have the correct nutrients – maybe it's low-fat! I found this upsetting as I'd always imagined myself breastfeeding. In my second pregnancy I became gestationally diabetic. This is a risky situation, as in pregnancy fat levels are always raised, but when you have high sugar levels due to diabetes the excess sugar is also turned to fat. This creates the potential for pancreatitis, which puts both the mother and baby at risk of dying. Because of the LPLD, I now have diabetes permanently.

Today, I manage the diet properly and have found ways to be able to participate more fully in "normal" life. I can now eat out in a Japanese restaurant, where I can eat about five of the sushi toppings out of the whole menu on offer.

If I go out for supper with friends, I send them a list of foods I can and can't eat and ask them to check out their menu plan beforehand. I always have to ask people to make an extra effort for me and I can often feel I have to reward this by being extra entertaining. I have been at many meals where people haven't made this effort. This does not make me feel loved and included!

There are so many situations in my life where food becomes an issue. Each individually isn't severe, but cumulatively, considering that eating is unavoidable, I'm left feeling separated from the normal flow of life and that the extra work I have to put in just to be healthy goes unnoticed and unacknowledged."