Jill Prawer

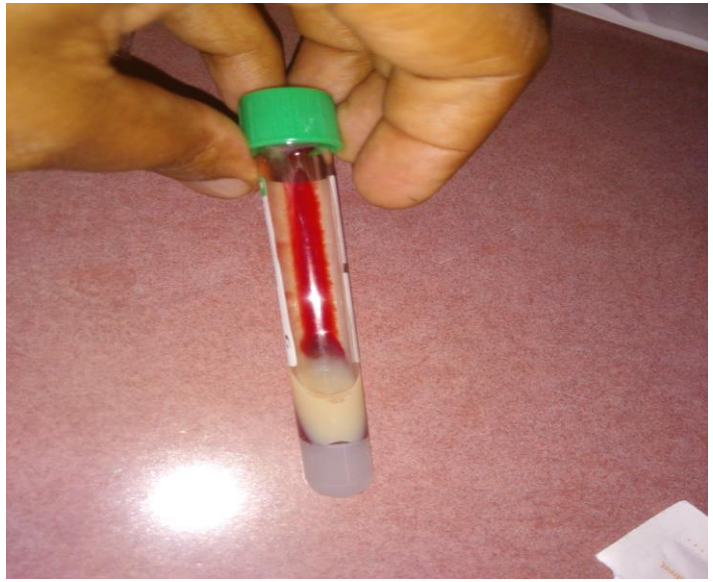
EUPATI FELLOW

Hello!

Diagnosed with Lipoprotein Lipase Deficiency in 1964 (aged 2)

What is LPLD/FCS (Familial Chylomicronemia Syndrome)

- It is an ultra-rare recessive genetic disorder (1 to 2 : 1,000,000).
- People with the condition don’t produce any or enough of the enzyme lipoprotein lipase, responsible for metabolising fats.
- If fat is eaten it travels around the bloodstream, giving it its classic creamy-white appearance - and causes symptoms



The blood of a young child with LPLD and very high trigycerides

Symptoms

High triglycerides lead to (among other things):

- **abdominal pain, often severe**
- **fatigue (called post-prandial fatigue)**
- **depression (isolation?)**
- **poor memory**
- **poor concentration**
- **joint pain**
- **yellow fat-filled spots (xanthoma)**
- **pancreatitis, which can be life-threatening**
- **chronic pancreatitis**

Treatment

- No useful medication currently available
- Patients need to limit their diet to between 10g and 20g of (any) fat per day (extraordinarily restrictive) and drink no alcohol.
- Glybera – first gene therapy authorised in Europe in 2012
  - But – under special circumstances and for a tiny minority of patients
  - And – dubbed ‘the world’s most expensive drug’
  - Is currently awaiting a decision from NICE about whether it will be assessed

Complications

- **Pregnancy can be dangerous unless a very strict diet is adhered to, especially if the mother develops gestational diabetes.**

- **People with LPLD are at a high risk of developing type 2 diabetes in their middle age or earlier if they have experienced many episodes of pancreatitis**

- **Diabetes and LPLD are not a good mix as high blood sugar levels are turned to fat exacerbating the LPLD and as high levels of carbohydrates are generally eaten to replace the energy of fat this leads to the necessity of managing the diabetes often with insulin.**

Patient Advocate

Since childhood – dream of breaking isolation and sharing experiences

late 1990s tentative meetings with patients - difficult

2008: AMT, EMA, HEART UK

2012 –LPLD Community on RareConnect

2013 EURORDIS Summer School on medicines regulation

Why EUPATI?

- **Felt isolated as a patient advocate – wanted a peer group**
- **Needed help navigating the relationship between patient advocates and pharmaceutical companies**
- **Help with my lack of self-belief (from having grown up with an ultra-rare condition)**
- **A strong desire to learn all I could to get medicines to people with LPLD who desperately need it.**

EXPERIENCE OF THE EUPATI COURSE

- **It's a lot of information that seemed quite remote and not immediately applicable - I found the format dry and hard to focus on**

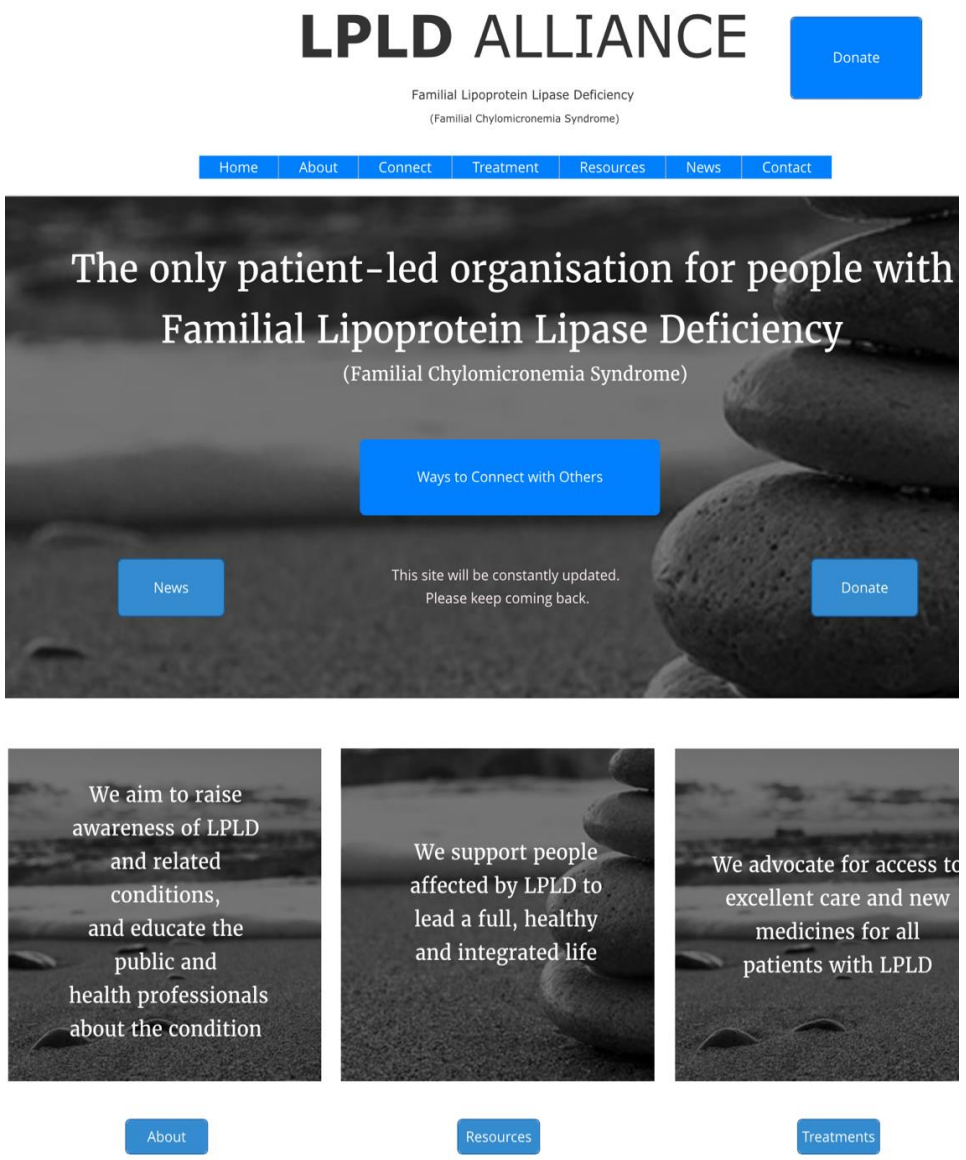
- **The first face-to-face was difficult as it was a lot more information delivered in a similar style and I felt 'unanchored'**

- **Personally I have difficulty being away from home with people who don't know me as my dietary restrictions are difficult to manage**

- **Second face-to-face was much better, we knew each other a bit and the format was much more engaging.**

- **The toolbox is really useful to return to!**

Post Course



Our Aims

- to raise awareness of LPLD and related conditions
- educate the public and health professionals about the condition
- support people affected by LPLD to lead a full, healthy and integrated life
- advocate for access to excellent care and new medicines for all patients with LPLD

What has the EUPATI course done for me?

- **Establishment of a supportive Peer Group of other Fellows**
- **Confidence to create the charity**
- **An understanding:**
  - **of the language used in medicines development**
  - **of how pharma works**
  - **of how to manage the relationship between us**
  - **of the pressures on pharma representatives and where we as patients might be being manipulated!**
  - **of many of the joint aims between pharma and patients**

How has EUPATI impacted on my advocacy work/involvement in medicines development?

- Worked with pharma and hosted a Patient Advisory Board
- Consulted on a burden of disease questionnaire which will truly capture the experience of living with LPLD
- Through contact with patients the company have plan to run a study with the patient-centred endpoint of relaxing the diet
    - With assurance that patients will be involved in the design from the outset (?)

How has EUPATI impacted on my advocacy work/involvement in medicines development… Contd

- **I'm a lay reviewer for NIHR and have commented on a couple of prospective clinical trials**

- **LPLD Alliance represented patients at NICE at the scoping phase for Glybera – improving the odds in favour of getting the therapy to patients**

Future initiatives

- **Hope to keep abreast of developments in the LPLD drug development world and be involved at an earlier stage of clinical trials**

Also - Brain Tumours

I have three children now 18, 15 and 13

- **2005 David aged 4 – metastatic medulloblastoma (he's now 15)**
- **Treatment saved his life but devastated it. He is now living a life that will always involve carers**
- **We have many late effects of treatment to look forward to (can be 20 or 30 years time – eg, heart problems, another cancer, fertility issues, problems with teeth etc).**

Brain Tumours

- **2012 I set up the telephone counselling service at Brainstrust (www.brainstrust.org.uk) and offered counselling for people affected by brain tumours (adults)**

- **2015 became a member of the steering group of SUCCESS (www.successcharity.org) a revitalised charity supporting long-term survivors of childhood brain tumours**

- **2016 was invited to join PORT through a chance meeting**

What is PORT?

- **Paediatric Oncology Reference Team (PORT) is a team of parents in the UK who have direct experience of children's cancer.**

- **An independent body which has developed autonomously although we enjoy very close links with CRCTU, the Cancer Research UK Manchester Institute, GOSH, Royal Marsden, and other paediatric oncology primary treatment centres.**

- **Members of PORT sit on the NCRI Children's Cancer and Leukaemia Clinical Study Group, the NCRI Leukaemia subgroup and CCLG Publications Committee among others.**

PAEDIATRIC ONCOLOGY REFERENCE TEAM (PORT)

Enhancing patient outcomes through research and education

Paediatric Oncology Reference Team (PORT) is a team of parents in the UK who have direct experience of children's cancer. Some of our children are post treatment and are well, and some of us are bereaved. We all have experience of working with various cancer related charities and bring a wealth of professional experience to PORT.

PORT is an independent body and has developed autonomously although we enjoy very close links with CRCTU, the Cancer Research UK Manchester Institute, GOSH, Royal Marsden, and other paediatric oncology primary treatment centres. Members of PORT sit on the NCRI Children's Cancer and Leukaemia Clinical Study Group, the NCRI Leukaemia subgroup and CCLG Publications Committee.

PORT's aims

•Assist medical professionals in the writing and reviewing of information for parents and children (of all ages) – particularly patient information sheets
•Advise clinicians on the practicality and value of clinical trials and studies
•Provide generic representation for families of children living with cancer
•Provide advice to clinicians and others on the reality and practicalities of caring for a chronically ill child

What does PORT do?

PORT's activities in the world of paediatric oncology is varied but the majority of what we do is review paediatric oncology clinical trial documentation for parents and patients. Click here for more information.

What doesn't PORT do?

PORT's remit does not include helping families resolve issues regarding their treatment, doctor or hospital.

Clinical trial documentation reviewed by PORT

Since 2013 PORT has reviewed over 20 paediatric oncology trials. Click here for a complete list.

How to get clinical trial documentation reviewed by PORT?

Click here to find out how to get PORT to review the clinical trial documentation for a paediatric oncology trial.

How to get assistance from PORT?

In addition to reviewing documentation for clinical trials we also give advice on ethical and consent issue and review websites, leaflets, posters. If you are a doctor or researcher and would like assistance or advice from PORT please email contactus@port.uk.com.

PORT needs new members

We are looking for parents whose children are no longer on treatment to help review documentation. If you are a parent and you are interested in volunteering your time for PORT please email contactus@port.uk.com. We would love to hear from you.
We are a virtual group and work from home when we have the time. We try and meet up once a year.

How to get in touch with PORT?

Email us at: contactus@port.uk.com.

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LPLD Alliance

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PORT

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