



LOWE SYNDROME PATIENT INFORMATION

I am writing to highlight a problem raised by Joel Lunardi in France which may affect Lowe children. The problem is associated with a defect in Lowe platelets which can be problematic in bleeding especially during certain types of surgery. My son, Oscar, was tested prior to surgery and he was confirmed to have a defect in platelet aggregation and has been recommended to take a drug prior to and after surgery to counteract the platelet problem.

To determine whether your son might be at risk, a PFA100 platelet screening test should be organised at your local hospital. It is quick and easy although about 20mls of blood is taken which might be a little bit of a problem with babies or younger children.

We are interested to find out whether this platelet problem exists with all Lowe children/adults or just some and so your results would be very interesting.

Whilst writing, if you did not complete the LSA survey sent out last year, I would be grateful if you could complete the attached questionnaire. The Trust is also eager to find out whether your son was diagnosed by DNA, Skin Biopsy or clinical diagnosis. If the diagnosis was by DNA, we would be grateful if you could forward a copy of the DNA sequence which can be obtained from your Doctor or Hospital.

Please contact me at any time to discuss in more detail. My email is lowetrust@gmail.com and telephone is 0207 794 8858.

Best wishes

Yours sincerely

Lorraine Thomas
Trustee
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