

Living with ASMD Niemann–Pick Disease Type B

My name is James. I have ASMD Niemann-Pick type B (NP-B). I have been asked to give an account on what it has been like to live with NP-B. This is quite a difficult task as I spend most of my life with my head buried in the sand and only think about it when things go wrong or I am forced to by events.

I thought it was quite normal for all children to spend a great deal of time at the doctors and hospital. To be examined by a stream of medical students, to have my liver and spleen palpated until my stomach was sore and always having blood tests.

I suppose school made me realise that I was different, I was made to have an afternoon nap until my mum found out and that soon stopped, both my parents were very keen that I should have as normal life as possible even if sometimes that was hard. I was not allowed however to do contact sports due to my enlarged liver and spleen.

My doctor put my name forward to go on Dreamflight an organisation that takes terminally ill children to Disney World, I had a lovely time, however from that moment on the word terminal kept cropping up

My mum and dad were always truthful with me and always told me that what ever I wanted to do I should try for, whatever the obstacles.

Practically my main problems were keeping my trousers up because of my large stomach and movement especially bending and getting up from laying down also seeing my feet was also a neat trick when standing up, however, all that changed when I had my partial splenectomy, suddenly I had a relatively flat stomach and I grew. I spent a couple of years having hormone injections to help me grow but noticed little improvement in my height.

The growth thing did bother me and has been a problem, (although I am now nearly 6ft), kids gave me a hard time at school as I was so small and looked so young, before I had a driving licence as proof of age I was always being refused in shops and pubs ect, this part was very frustrating and annoying, although living in a small town where most people know me has helped, having a brother who at 17 was 6ft 5 didn't, no-one ever questioned him!

I have spent my whole life trying to prove everyone wrong, much to the despair at times of my parents. If I was told this was something I would be unable to do, then that was exactly the thing I wanted to do.

I have now been through main school education, achieved my GCSES, went to college to do Lens based Media and gained a distinction. I have applied to the Ambulance service and Fire Service, I own my own shop, now in partnership with my brother, selling Menswear, who now sees to the day to day running of the shop, while I am off learning to drive Double Decker Buses and that's when I am not out on my motorbike.

How do I cope with living with NPD? I live my life to the max, of course it scares me a bit but life is a lottery and I am lucky that I have been able to achieve most of the things that I have wanted and that I have great people around me who support and encourage me.