

Genetic Consultation

At present, surgically, we stand at awaiting facet joint injections before surgery. I am also currently awaiting the date for surgery to be processed, my consultant is happy to go for April, I am vying for the end of May time. Pre-op will roughly be six weeks to whatever date is finalised for surgery. Surgery will be some form at an Anterior Cervical Discectomy and Fusion (ACDF), however my consultant think he will have to do half of it from the front, turn me over and go in through the posterior too, so there will be two incisions - I won't know until after surgery of course if he has.

Parallel to the surgical side, I have finally taken myself to genetics. As per protocol there was a genetic counsellor present during the consultation. She started drawing out my family tree with me starting from grandparents, I am fortunate to know the names and DOBs of most of my family so it was fairly straightforward. The genetic consultant joined us shortly after, and continued with the family tree.

They wanted to know if there was any medical family history, there was none that I know of so that didn't take much time either. Then came my reasoning for genetic testing, this was slightly irksome as I don't like talking about the subject at the best of times. I don't have an entire concrete version of why I want genetic testing, but I know it is something I have to do.

She told me then that even if I had acquired KFS as a sporadic mutation, I will essentially have the gene and therefore there will be a possibility it could be inherited. This is something I have to discuss further with the genetic counsellor apparently. Next up came the physical examination, nobody had ever mentioned that this would be part of the consultation so that came as a shock. In short, she wanted me to strip down to my underwear so she could take pictures. It may sound humourous, but I was by no means amused. She just about got the pictures she needed but I did remain semi-clothed with a blanket.

After I was dressed again, she went through what tests she wanted to do and explained why - most were blood tests, she wanted x-rays too. I am aware the genetic consultants do seem to request a whole heap of stuff but I have the advantage of already having a whole load of them done already, so she gained my consent to request all my medical records instead. I also gave consent for my medical images to be used anonymously - this isn't at all necessary but I don't see the harm in it.

She told me she would write up our consultation and send it to me with further details and her opinion of the blood tests results. It has been advised that I speak to the genetic consultant when I receive these letters and I will see her in about 6 months to discuss the medical stuff in more detail face-to-face when she has finished processing everything she has to. Blood was taken before I left so that my DNA can be analysed in several different ways for a whole range of stuff in addition to some more specific conditions that could be relevant. Being that this was done on 23rd December, I am still waiting for the letters.