

# 100,000 GENOMES PROJECT IN SCOTLAND



On Rare Disease Day (28 February) the Scottish Government announced that people in Scotland will now be able to join the Scottish arm of the 100,000 Genomes Project. SWAN UK member Sharon tell us what this could mean for her family.

I'm Sharon and I live in Fife with my undiagnosed son, 12-year-old Luuk, his brother James, five, and my husband Remy. Luuk is our walking, talking miracle. We were told that he would not survive birth and for the first five years of his life it was touch and go.

Developmentally Luuk is about eight or nine years old. His short-term memory doesn't work well and he has difficulties with abstract concepts. He is socially very vulnerable, and because of this in combination with the risks posed by his liver, Luuk requires constant adult supervision. He remains completely tube-fed, needs to use a wheelchair for distance and has to do daily physio especially now puberty has struck. He experienced his first joint dislocation a few weeks ago and due to growth spurts, lax joints and hormones is at risk of more dislocations. His speech can be difficult to understand due to his palate being very short and he is currently awaiting major surgery to hopefully help with his speech. However, this surgery could have far-reaching negative effects for Luuk too; it's a very fine balance weighing up risks versus benefits of any surgery for him.

With Luuk being so anatomically unique and with so many of his bodily systems being involved, we don't know what the future holds for him. I know that applies to everyone, but having an undiagnosed genetic condition means that as his parent and advocate my brain never gets a break.

I worry constantly that something awful will happen to him that we'd be able to prevent if we had a diagnosis and were looking out for it. Every little twinge causes us to panic.

Will he make it to adulthood? Will his organs just give up because of their position? Will he get cancer? Will he get one more infection that will kill him? Will he develop seizures or other neurological problems? The list is endless. We try and hide this from Luuk but he knows he's different. Regardless of how awful knowing might be, not knowing, for

me, is far worse. Everything gets magnified, it grows, and I find myself thinking of the most awful things, when he simply has a bug!

Potentially, Luuk being involved in 100,000 Genomes Project could go one of three ways:

- He gets a diagnosis and we get a better understanding of the longer term effects his condition might have on him.
- He gets a diagnosis but is the only one with it and there is no information.
- We get no diagnosis.

If the second or third option happens, we have to accept that we won't know any more or how his condition is likely to affect him as he ages. This means that the worry and fears for his health won't end, but if he gets a diagnosis the constant search for an answer might end and will free up brain time for me meaning it might have a rest now and again.

Luuk was one of the first children recruited to the DDD Study and unfortunately they weren't able to find a diagnosis for him. We remain hopeful that we will get answers for him through the 100,000 Genomes Project.

For us, Luuk's undiagnosed genetic condition is a huge big black cloud that hovers over us each and every day, indoors as well as out. And each day we hope it doesn't rain on us. We look up at it and try to determine if it will rain or stay dry for a while; maybe the sun might even shine through. Sometimes we guess right and have an umbrella at the ready or we don't bother with a coat and stay dry. Sometimes we get it wrong and we're completely soaked and wet and struggle to get dry again. For us, that's what having a child with an undiagnosed condition means – constantly thinking and trying to guess what the cloud will do next.

