

Living in the shadow of hereditary disease

23/9/2013

By Alan Baker

Temple Street Hospital is Dublin's famous northside children's hospital. Opened in 1879, it's a dilapidated shack of a place, with Victorian-era buildings that are unsuitable for the dispensing of modern medicine.

However, a value cannot ever be put on the work it has done for Irish children in its time. It's also a pretty incongruous place for a forty-something man to be sitting in a waiting room, with Scooby Doo characters on its walls, and a sticking plaster featuring ladybirds and polar bears on his arm...

Most people familiar with the hospital are probably unaware that it houses an arm of the National Centre for Medical Genetics, a clinical service for people who are at risk of genetic disorders.

Things like cystic fibrosis, or Huntingdon's. My possible opponent is a little less well known than that....

Cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy. It's quite a mouthful. It sounds like something serious – and it is. Stick the opening letters of each word together and you get CADASIL.

Unless you know a family that's affected, the chances are you've never heard of it. It's a rare disease which, in medical terms, is a form of hereditary stroke disorder. In human terms, it is a blight that can haunt families from generation to generation.

It started - for us - with my mother. About two years ago, she started having problems with balance and other issues that led her to her GP and then to hospital with what doctors suspected was a tumour on the base of her brain.

I can remember at the time how scared we all felt and how pessimistic the outcome looked, if confirmed. But then she was diagnosed with an AVM, a malformation of arteries and veins; a problem, but one that could be treated. Oh, how we celebrated.

A few days later, she had her first stroke.

Over the next few weeks, she had a series of over a dozen more, including at least one that affected her spinal cord. For a while, I was convinced she wouldn't leave the hospital. There were days when it seemed the next bad turn would be the last.

It's hard to put into words how we all grew to hate that hospital, not because of the care she received, which was wonderful, but the months of never knowing what the next day would bring. And the noises. And the smells. And the dodgy cafe. That damn place...day after day, month after month...

She's a fighter though, my mum; she rallied and after the best part of a year we got her home a week before Christmas 2011.

She was back to herself mentally and sharp as a button, but, sadly, rendered an invalid. She'll need constant care for the rest of her life and that's rough, both for her and for my dad.

Her spirit is fantastic, though, and she's a typical Irish mammy, she hides the dark thoughts from us and never wants us to worry. We love her and we'll always be there for her.

It was in the early days of her hospitalisation when I first heard the term CADASIL. It turns out there was a family history; relatives who'd been affected many years before. I'd known nothing about it (that Irish mammy thing again). But I quickly became an expert.

CADASIL affects small arteries in the brain, which can close, causing strokes and other issues. Prevalence is about 1 in 50,000, though it is suspected to be more common, as it is sometimes misdiagnosed as cardiovascular disease, or even MS.

Onset is usually in the 40s; sufferers can have recurrent strokes over a period of years, damaging the brain and leading to mental and/or physical deterioration. Migraines with auras are a common occurrence. Other possibilities: depression; incontinence; dementia; worse...

Tests proved our mum's diagnosis. For my siblings and I, the chance of each of us having inherited the condition is 50:50. And if any of us are on the wrong side of that 50:50, there's a 100% certainty that it will strike one day.

It's as stark as that. There's no way of knowing when the day might come - just that the day will come. A month away, a year...maybe 20 years. And I've no way of knowing how severe it will be. It's difficult to come to terms with something like that.

Oh, and there is no treatment.

Now we face an uncertain journey. It wasn't that long ago when a CADASIL diagnosis would usually only be made after autopsy. Nowadays there's a simple blood test that spells out your future for you. There's a few steps involved beforehand - two visits to a genetic counsellor and one to a psychiatrist before actual blood testing takes place.

Of course, there's no pressure to do it, no requirements, but little peace of mind. I can choose not to know - many in my position do just that. Life happens and nobody can say what the next day will bring, so why add to the uncertainty, why risk gambling a comfortable life for knowledge when there's little to be done either way?

What would you do? It's something I've wrestled with for months now. There's a stubborn part of me that says forewarned is forearmed, that I can handle the truth. I've always been an optimistic person - and that 50% chance of NOT having it is worth pursuing. I'd never have to be concerned about it again, wouldn't have to worry each time I felt a bit of a headache coming on...

The other half of me is too bloody scared to contemplate the wrong result.

That brings me back to Temple Street. It's here I have my appointments with the genetic counsellor and give my blood samples (they test two in the end). In our first session, we discussed the disease itself, whether I've had any problems that may be symptomatic (I haven't) and what I can expect of the disease.

Part of this conversation is about how people react to learning they have it; how depression often follows, or lethargy, or impulsive behaviour. A bullet is coming that you can't dodge - time to buy a Harley maybe...I'd probably be scraped off the road after two days.

Round two sees me asserting myself - I want the damn test, take my blood (after which I exit bearing that lovely plaster). Then there's the psychiatrist - everything stops if he forms a medical opinion about the threat to your mental health.

One of the toughest hours of my life and feeling very emotional when I left, but the journey continues. The whole process has taken six months - but in eight to ten weeks, I will be back to the counsellor who will have an envelope for me.

Even at that point, plenty of people have bottled it. The counsellor has a drawer full of envelopes for people who changed their minds on the day. She's torn up plenty more. As a family, there are four of us facing that envelope.

I can't say four of us will open it though. I'm determined right now, but who knows what I'll do on that day and how my life will change after it.

A mutation on a gene on an out-of-the-way chromosome. A tiny thing really. And devastating.

You can locate this article at <http://www.irishhealth.com/article.html?id=22635>