

PATIENTS PUT SKIN IN THE DYSTONIA RESEARCH GAME

By Paul A*

I need your help, and in ways you might not realise, you need your help too.

I've just become the 25th person to have undergone a skin biopsy, one of life's simplest medical procedures, at the hands of Neurologist Assoc Prof Kishore Kumar, in his quest to ultimately find 92 people willing to volunteer a tiny piece of themselves to help solve the mysteries of dystonia.

Dr Kumar's research at Sydney's Garvan Institute of Medical Research is gaining momentum after winning international attention for discovering genes that cause dystonia and for his peer-reviewed scientific papers disclosing his findings. Knowing that, so far, only about one in five sufferers have had a defective gene associated with their condition he has pondered what else might be found to be contributing to distressing dystonic behaviour.

Now, he is finding some answers to that question by not just looking at the DNA in a gene, but also the RNA.

While the DNA might be organised to deliver someone a happily normal life, RNA and the messaging protein it produces is calling the shots and telling the DNA what to do, creating all sorts of havoc which then leads to the manifestation of dystonia. Which is how and why I found myself to be Dr Kumar's 25th biopsy volunteer, all in the name of science, research, discovery and ultimately the greater good.

So, here I was, lying on a bed in a procedure room in the neurology department on the 5th floor of Concord Hospital in Sydney in May, and the mood was cheerful. With a relaxed Dr Kumar was neurologist Dr Laura Rudaks, a PhD candidate and fellow researcher on his team, and another brilliant young mind in PhD graduate and research scientist Dr Bianca Grosz who was at hand to take delivery of my skin sample so she could grow fibroblasts from it for the purpose of harvesting my RNA.

What transpired was one of the easiest and simplest medical procedures I'd ever had - better by a country mile over a visit to the dentist, a prostate check or removing a splinter! Dr Kumar expertly and gently delivered a little local anaesthetic to his target zone - my upper forearm - and before I knew it, he had painlessly used a special little device to instantly take a tiny skin sample and deliver it to Dr Grosz. No worries!

I'd had a biopsy done before, on my head to diagnose a skin cancer, and in both cases, they were a walk in the park, which is really something coming from me as a person who hates the thought of any medical procedure.

Despite the simplicity and ease of the procedure, Dr Kumar says that many people find the move from providing a blood sample to a tiny skin sample more of a leap than a simple step and that pioneering research is being slowed or prevented from achieving important goals. Perhaps remarkably, those associated with dystonia sufferers have been more readily forthcoming in offering a skin sample as part of a control group than sufferers themselves.

Already the biopsies have yielded other important findings and some patients have been delivered simple therapies that have led to vastly improved symptoms. Dr Kumar recently discovered one volunteer's symptoms were vastly improved by being dosed with therapeutic levels of common caffeine. Another was found to be responsive to being dosed with dopamine.

In research, minimum sample sizes are required to validate, prove or disprove a theory. Dr Kumar needs 32 skin samples for his current research objective, but in an expansion of his work through substantial additional research funding to support him and his team for another four years, he needs an additional 60 volunteers to achieve definitive research outcomes.

Not everyone with dystonia is eligible despite their goodwill. Dr Kumar especially needs skin samples from people with generalised or segmental forms of dystonia (two or more parts of the body affected), dystonia with other neurological symptoms (combined, complex dystonia), have early onset dystonia (acquired before the age of 29) or a family history of it.

And going back to my original point, I need your help because my wife and I have found fulfilment in financially contributing to Dr Kumar's work for 9 years and while he could be given all the money in the world, he will not achieve anything if people like you and me can't give him the skin and blood samples he needs to validate his early findings. We as sufferers of dystonia hold the keys to the discovery of its causes and can contribute to the body of science which we hope can one day lead to cures.

Neurologists have powers of observation, now they need to be empowered by the dystonia community's willingness to find answers to the mysteries of the condition, not necessarily for themselves but for others whose lives are impacted by it. Let's not just talk about it, let's do something about it. Both me, and you.

To help, please talk or write to your neurologist to check if you are eligible and ask your doctor to contact Dr Kumar at Garvan to make suitable arrangements. You'll be glad you did.

Links to Dr Kumar's work are here:

<https://www.garvan.org.au/people/researchers/kishore-kumar#publications>

<https://dystonia.org.au/media/videos-and-webinars/>

Dr Kumar works with neurologists in Sydney (Westmead, St Vincent's, NeuRA, and around Australia).

If you are interested in having a genetic assessment of dystonia, your neurologist could refer you to A/Prof Kishore Kumar (Mail: Neuromuscular Clinic, Concord Repatriation General Hospital, Concord, NSW, 2139; Phone: 02 9767 6416 or 02 9767 5623 Fax: 02 9767 7807; Email: kishoreraj.kumar@health.nsw.gov.au), or print out a copy of this article to discuss with your neurologist.

*Paul developed cervical dystonia almost 40 years ago at age 28 and has been an advocate for patient support and research since 1988. He lives in Sydney.