Genetic empowerment: Extreme athlete probes own genetics to streamline diagnosis

In 2002, extreme athlete Kim Goodsell was diagnosed with Charcot–Marie–Tooth disease, “a genetic disorder that affects the peripheral neurons carrying signals between the spinal cord and the extremities.” When Kim reviewed her doctor’s notes, she found it odd that he made no mention of her other rare genetic condition, previously diagnosed: arrhythmogenic right ventricular cardiomyopathy (ARVC). Was she really so unlucky to have been struck by genetic lightning twice, or was there an unseen connection? So she started doing her own research, teaching herself genetics in the process. Ed Yong writes:

> Kim looked at every gene linked to Charcot–Marie–Tooth – there are more than 40 overall, each one imparting a slightly different character to the disease. One leapt out: LMNA, which codes for a group of rope-like proteins that mesh into a tangled network at the centre of our cells. This ‘nuclear lamina’ provides cells with structural support, and interacts with a bunch of other proteins to influence everything from the packaging and activation of genes to the suicide of damaged cells. Given this central role, it makes sense that mutations in LMNA are responsible for at least 15 different diseases, more than any other human gene. These laminopathies comprise a bafflingly diverse group – nerve disorders (like Charcot–Marie–Tooth), wasting diseases of fat and muscle, and even premature ageing.
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> As Kim read about these conditions and their symptoms, she saw her entire medical history reflected back at her – the contracted muscles in her neck and back, her slightly misaligned hips and the abnormal curve in her spine. She saw her Charcot–Marie–Tooth disease.
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> She also saw a heart disorder linked to the LMNA gene that wasn’t ARVC but which doctors sometimes mistake for it. “Everything was encapsulated,” she says. “It was like an umbrella over all of my phenotypes. I thought: This has to be the unifying principle.”
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> “People have been talking about empowering consumers since there was an internet,” says Eric Topol, a geneticist at the Scripps Clinic. “But finally, we’ve reached a point where someone can delve into their condition beyond what the top physicians at the Mayo Clinic could. They couldn’t connect the dots. She did.”

Read the full, original article: DIY diagnosis: How an extreme athlete uncovered her genetic flaw

Additional Resources:

- Patient cracks her own mysterious dual diagnosis, Medscape
- Woman with aggressive cancer delays treatment in the name of science, Yahoo! News
- One of a Kind: What do you do if your child has a condition that is new to science?, New Yorker