More than a Diagnosis

Written in the POV as a future GP.

Another morning in clinic, twenty years into my career, and Jenny sat across from me describing months of unsteadiness and frequent falls. She sat with dystonic posturing, her movements filled with sudden jerks. As I finalised my clinical notes, a notification appeared:

Possible rare disorder detected:
Myoclonus dystonia. Click here for more information.

The national rare disorders registry had identified patterns in my documentation using AI, linking clinical features to differentials for rare disorders. With Jenny's consent, I opened the suggested pathway referring to genomic testing and patient resources. What once required years of referrals could now begin within a single consultation.

Moments like this return me to my fourth year of medical school in 2025. I met Gerald, whose symptoms mirrored Jenny's, but whose journey unfolded differently. For five years, he moved between cities and clinics searching for an answer. When the diagnosis of myoclonus dystonia had finally been made, relief filled the room, alongside a quiet awareness of time lost. Gerald was referred to genomic testing and had to wait months for his results to return from Finland. It confirmed a pathogenic SGCE variant. I realised then that a diagnosis is not merely a label; it is permission for patients to understand their own stories.

Many years later, whole-genome sequencing is easily accessible through primary care, supported by a national rare disorders registry co-designed with Māori and Pacific communities, embedding principles of data sovereignty and equitable governance. The registry collects information from patients with rare disorders across New Zealand and consolidates it into a central database that provides clinicians with resources, guidelines, and pathways. Genomic testing that once had to be sent overseas could now be done locally. Diagnostic odysseys have shortened from years to weeks.

However, this progress has expanded, rather than simplified, my responsibilities. Earlier in my training, I believed the purpose was to reach answers quickly. Genomic results may clarify a diagnosis, but they can also exacerbate uncertainty, reveal incidental findings, or raise implications for whānau who didn't choose testing themselves. It begins with listening, and consent is not a single signature but an ongoing dialogue. Some patients choose to know, others choose uncertainty. Both decisions deserve equal respect.

Weeks later, Jenny's results confirmed SGCE myoclonus dystonia. Together, we explored the next steps. Although treatment options remain limited, the impact of having a diagnosis was profound. With her consent, her anonymised data contributed back to the national registry, improving recognition for patients yet to come.

Rare disorder genomics has transformed medicine since my student days. As healthcare professionals, our role is to steward these advances with care, ensuring genomic innovation is guided not by what technology permits, but by what patients need most. For people living with rare disorders, genomics doesn't simply accelerate diagnosis: it restores them time. It offers more than diagnosis: it offers the visibility of being seen within a health system that has too often overlooked them.