

From Patient to Practitioner: A Future Doctor's Perspective on Rare Disorder Genomics

Advances in genomics are rapidly transforming the diagnosis and management of rare disorders. As a future doctor, I anticipate that genomic medicine will become an increasingly important part of clinical practice, particularly for people living with rare conditions who often face prolonged diagnostic journeys and uncertainty. My perspective on this issue is both professional and personal. Living with Ehlers-Danlos syndrome, a rare connective tissue disorder, has given me first hand insight into the challenges patients experience while navigating healthcare systems that are not always equipped to manage rare conditions.

I believe that one of the most significant opportunities genomics presents is the potential to shorten the “diagnostic odyssey” experienced by many individuals with rare disorders. Genetic sequencing technologies can help identify underlying causes that may otherwise remain undiagnosed for years. For patients and their families, receiving a diagnosis can be profoundly meaningful. It can validate lived experiences, guide clinical management, and connect individuals with appropriate support networks. As a future clinician, I hope to recognise when a rare disorder may be present and advocate for appropriate genomic investigation or specialist referral when needed.

While the potential benefits may be significant, genomic medicine also raises important ethical considerations. Genetic information often extends beyond the individual patient and can have implications for wider whānau. Approaching genomics in clinical practice requires careful communication, informed consent, and sensitivity to patient autonomy. Clinicians must ensure that patients understand the potential outcomes of genomic testing, including uncertain findings or incidental results. My own experiences as a patient undergoing genetic testing have reinforced the importance of clear, compassionate communication when navigating complex medical information.

Equitable access to genomic medicine is also essential. In Aotearoa, health inequities continue to affect Māori and Pacific populations, as well as other underserved communities. If genomic technologies are implemented without addressing these disparities, they risk further widening gaps in healthcare access. As a future health professional, I believe it is important to advocate for systems that ensure genomic services are accessible, culturally safe, and responsive to the needs of diverse communities. This includes engaging respectfully with patients and their whānau, recognising the importance of trust, and ensuring that innovations in healthcare benefit all patients.

Patient-centred care should remain central to the integration of genomics into clinical practice. Rare disorders can affect many aspects of a person's life. Genomic results can carry emotional and practical implications. My experiences living with a rare disorder have deepened my appreciation for clinicians who listen, validate patient experiences, and take concerns seriously. As a future doctor, I hope to bring this same empathy and understanding to my own practice.

Ultimately, genomics offers powerful opportunities to improve the lives of people living with rare disorders. By integrating these technologies thoughtfully, health professionals can help ensure that genomic medicine empowers patients, rather than simply advancing scientific knowledge.