The Nuances of Genomics in Rare Disorders

Although I have no personal experience, my short time in the clinical world has shifted my perspective of rare disorder genomics from feeling like an abstract theory to a responsibility I will carry in my daily practice. I have seen how uncertainty characterises patients’ journeys, from the dissatisfaction of not being believed, to the missed diagnoses and delayed answers. For those living with rare disorders, this uncertainty is prolonged.

However, genomics alters that journey by shortening the diagnostic process, guiding management and providing opportunities for research and targeted therapies. Although this seems like routine care in the health system, the rise of genomics provides substance that has been historically missing for this community: visibility and connection. This is crucial as it validates patients' experiences while helping them connect with others of similar backgrounds. It also enables me to become a better health professional by working in a wider team of clinical geneticists, counsellors, and researchers to provide care tailored to the patient's needs.

Genomics will contribute to recognising unusual patient presentations or having a korero regarding life-changing results. However, it is not just about ordering tests, but interpreting and communicating results with empathy and cultural safety. This will be a learning experience that I will need to navigate through in this field, particularly since it is rapidly evolving. Subsequently, it fosters professional growth as I need to be adaptable and reflect on my own practices. Additionally, I am also aware of the limitations rooted in this side of healthcare. There will be times when I will not have the answers for my patients, further pushing me to be a healthcare professional committed to lifelong learning.

In the future, I will face difficulty regarding the ethical aspects embedded in genomics. Although informed consent and privacy are important, it must not be forgotten that genomic information extends beyond the individual to current and future relatives. Personally, the most difficult aspect as a future healthcare professional will be dealing with uncertainty. For example, how do I go about expressing incidental findings and ensuring they are physically, mentally, and socially equipped in that moment?

However, access to genomic services is not equal across Aotearoa New Zealand, and any advances may increase existing disparities. This particularly affects Māori, Pacific and rural populations, who face a myriad of factors when trying to access specialist care. Consequently, I see my role as an advocate for marginalised populations, so that inequities do not widen and our practice of care is culturally safe.

Overall, a patient-centred approach is essential in this aspect of care. Although genomics can provide specific diagnosis, rare disorders are very complex. They comprise physical, social, mental and financial aspects, making it a multi-faceted challenge. Therefore, genomics should aid the holistic care we strive to provide. Eventually, as a health professional, my responsibility will be to harness these opportunities equitably, so that I can deliver more personalised care and contribute to a future where rare does not mean unseen.