The Cost of Fear: how embracing AI could transform the diagnosis of rare disorders

"We are on the verge of a vast expansion of human potential, made possible by technology. Our tools amplify our capabilities, our reach, and our understanding."
– Carl Sagan (1980)

Change is unsettling - the notion of AI-mediated healthcare is uncomfortable for many, both within healthcare professions and beyond. But the populations who stand to gain the most from these technological leaps and bounds are also among our most vulnerable. I believe that learning to engage with and advocate for the development of AI will facilitate early and accurate diagnoses for people with rare disorders, even if this proves intellectually and logistically challenging in practice.

The arduous process of receiving a diagnosis creates a massive healthcare barrier for persons living with rare disorders (PLWRD) - nearly 20% of PLWRD reported a diagnostic journey of over ten years (HealthNZ, 2024). This is not good enough. As a medical student, I believe that advocating for AI research and development *in consultation* with our PLWRD communities is essential to improving these diagnostic outcomes. AI holds the promise of remarkable pattern recognition using vast data sets - something that is simply not achievable by an independent clinician, no matter how extensive their knowledge. Research is already showing the power of AI to recognise facial features to augment the diagnosis of rare and ultra-rare disorders (Hsieh et al., 2022). My goal is to safely enlist future patients in clinical trials that can accelerate this research, and make noise among my peers about the advantages of engaging with AI in our practice.

However, it would be remiss to discuss AI in healthcare without acknowledging some well-founded ethical and logistical concerns. AI is still new and relatively unregulated technology, thus warranting a healthy dose of skepticism. In the next 25 years I can too easily picture a landscape where we avoid it altogether to alleviate those feelings of mistrust. But this cannot happen. Systems must be built, policies written, and evidence compiled - dare I say, egos must be quashed. What an act of hubris to go on (inadequately) diagnosing, treating, and monitoring populations with diseases that we do not fully understand, while ignoring powerful tools that could ease their suffering. I make a stand now that I will not allow my pride or time constraints to stop me from

pursuing the best possible technologies available for my patients. I make the commitment to overcome the discomfort of upturning the status quo, to make life better for those who are consistently at the margins.

Nobody chooses to be sick, much less to live with a rare disorder. It is no longer acceptable in this global age of information and data processing to shrug our shoulders and say, “I don’t know, sorry, let’s see how you get on.” AI is here to fill the gaps in our knowledge, but of course nothing can ever replace the human connection of a therapeutic relationship. So let’s pick up and adapt to AI the way we did with Google, tele-consults, e-prescriptions, MRI, and genome studies. Health professionals aren’t going anywhere, but long, drawn-out diagnoses need to become a thing of the past.

References

HealthNZ. (2024). *Impact of Living with a Rare Disorder in Aotearoa New Zealand*. Rare Disorders NZ. Retrieved March, 2025, from https://raredisorders.org.nz/media/pages/file/91/5295-rdnz-white-paper-layout-web.pdf

Hsieh, T., Bar-Haim, A., & Moosa, S. (2022). GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. *Nature Genetics*, *54*(1), 349-357. https://doi.org/10.1038/s41588-021-01010-x