

Resident Spotlight

Dr. Adrienne Elbert

Medical Genetics & Genomics



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Medicine for everyone

So often, the way we're exposed or introduced to a new disease or condition — whether in the media or in our education — is by seeing the most extreme instances of that disease. We are shown how a disease manifests itself in the worst case scenario and the poor outcomes for the patient and their family.

But for many conditions the most common manifestation is a much milder version. And this often results in the condition going undetected, undiagnosed, or dismissed by authority figures and even by physicians.

Adrienne Elbert, PGY3 resident in Medical Genetics & Genomics, wanted to bridge this gap and provide diagnoses and treatments to those who experience a disease



across the spectrum of its phenotypic expression.

"There is definitely a bias in identifying patients that are more severe," Adrienne stated. "We see this all the time. One example was



THE UNIVERSITY OF BRITISH COLUMBIA
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with a condition called Nonketotic Hyperglycinemia.”

She continued, “the severe manifestation was the *‘image’* people had of this disorder. But with wider testing, we’re identifying individuals that have a milder form.”

These patients have the same genes, and their struggles are just as valid, but different. And this difference in manifestation caused the disease to be overlooked during childhood.

“We weren’t even looking at them,” Adrienne exclaimed. “They may have a learning disorder in school but were just told to *‘try harder’* while they had an actual, treatable, metabolic condition.”

Recognizing and understanding how diseases manifest at all levels helps to put families at ease and provides important information, and even sometimes provides a community, to concerned parents and patients.

“If we can say *‘this is the cause for you’* it really ends the diagnostic odyssey for that family,” Adrienne stated. “This is very empowering.”

She continued, “this also makes it more likely to identify other individuals who may have the same or related disorder, and can help provide prognosis or likely outcomes to a concerned family.”

Often, parents put blame on themselves or on their pregnancy when there is a *“difference”* with their child that has no explanation. Having a proper diagnosis usually puts their mind at ease and can often be a way to find a community of similar experiences and a network of support.

Working with undiagnosed patients and rare diseases brings a sense of fulfillment and pride to Adrienne, and it was part of her reason for choosing UBC to do residency.

“We’re at the forefront of medicine and seeing new things each day. In the past, there was a concern that there wasn’t a lot of treatable conditions and therapies,” Adrienne stated.

“I always had an idea of what a doctor should be like, but being in Genetics & Genomics at UBC has definitely changed my perception.” She continued, “I had a very paternalistic image of what medicine was. But upon shadowing

the genetic councillor, I really began enjoying working with the undiagnosed. It felt like I was on an episode of *House MD*,” Adrienne joked.

“My vision of what a physician was changed, and I saw that I could be in a teaching role and close to families in a guiding role — helping them through diagnoses at an accessible level.”

Adrienne attributes her curiosity and ability to troubleshoot to completing her PhD in Biochemistry at the University of Western Ontario. She states that doing a PhD gets you used to failure. Or at the very least, it gets you used to not getting it right the first time and having to rework your hypothesis, or tackle a problem from a different point of view.

“Not every question that you ask in a PhD is the right question, and when you design an experiment, it doesn’t always work and you have to tweak your methods,” Adrienne stated.

“In genetics, it’s intimidating if you have a patient with a condition you’ve never seen before. Doing a PhD has made me more prepared

for the unknown. And being fearless like that and undertaking something new is really supported through a PhD.”

While Adrienne’s fondness of kayaking in False Creek may be on hold with the recent cold snap and squalls. She plans on spending many cozy evenings playing *The Legend of Zelda* with her 15 year old dog Mitsu by her side — who came to Vancouver with her many years ago and has been on every adventure thus far.

