I pepper these personal stories with a few general discussions of genetic testing, screening, and gene therapy. I use the word "mutation," because it is easier for a general audience to understand than the term "variant" (pathogenic or benign) that has come to replace it.

Both the American Cancer Society (ACS) and the American Heart Association (AHA) recommend genetic testing for those with a family history of disease.[1] The AHA advises testing even without family history if a patient has a condition commonly caused by a genetic mutation. Knowing the disease mutation lets other family members check if they have it and get preventive care. Unfortunately, despite widespread agreement among experts on the value of such testing, the clinic often fails to implement it.[2]

My family story has driven me to promote genetic screening in addition to genetic testing. I posit that proactive screens for mutations that cause treatable medical conditions regardless of family history would save lives and improve health outcomes. Such tests are now quite cost-effective. However, ClinGen is concerned that we neither have enough physicians who are sufficiently trained nor the follow-up clinical resources required.[3] Also, the American College of Medical Genetics and Genomics asserts more research is needed to understand how the risks of certain disease-causing mutations vary in different populations.[4] Another concern is that knowing you have a higher risk of illness may cause emotional distress even if treatment is available. Also, if revealed, genetic results could lead to social and employment discrimination and the denial of life insurance.

I hope my family's story will inspire more action toward tackling these serious issues and will raise awareness of the value of genetic testing and screening.