

Part 2: Carrier Testing

The daughter decided to go through with the genetic testing to see if she is a carrier of a *BRCA1* mutation. Your job is to look at a sequence obtained from her normal white blood cells and compare it to the human *BRCA1* reference sequence. If a mutation is found, there is a 55% risk of having breast cancer.

Identify the mutation present and if one is found, then what type of mutation is it (i.e. insertion, deletion, point mutation, or inversion) and where is it (nucleotide number)?

Once you have scanned the sequence and come to your conclusion, head to the volunteers and report your result as if you were informing the daughter. Be prepared for some counter questions.