# Questions

## Three Clinical Cases, Identify Variation and Analyse Variants

1. If a patient is referred to clinical genetics, it is usually because they are suspected to be at risk of a genetic disorder
   1. What is a genetic disorder (briefly)?
   2. Why might a symptom or collection of symptoms that someone is experiencing be thought to have a genetic cause (as opposed to being caused by something else)?
   3. How might a person benefit from knowing that their condition is genetic? What problems might they encounter from knowing this? (hint- think about potential legal and social issues too)
   4. Who ultimately decides whether or not a genetic test should be performed?
2. Laboratory genomics also tests for genetic changes that are not inherited, but are acquired, such as those in some types of cancers. These variants are often referred to as somatic variants (this is in contrast to germline variants).
   1. What could be some of the benefits to the patient of testing for these?

## Resources

<https://www.nice.org.uk/guidance/cg164/ifp/chapter/genetic-counselling-and-genetic-testing>

<https://www.bsgm.org.uk/media/678746/consent_and_confidentiality_2011.pdf>

<https://pathways.nice.org.uk/pathways/familial-breast-cancer>

<https://scienceblog.cancerresearchuk.org/2011/11/21/our-stratified-medicine-programme-what-is-it-and-how-will-it-work/?_ga=2.188493291.2105290516.1552571033-313784797.1505992911>

<http://www.wales.nhs.uk/sites3/page.cfm?orgid=457&pid=25400>