

Sample Part1 essay questions

Cytogenetic and molecular aberrations involving chromosome 17 have been observed in both constitutional and acquired disorders. Give examples of two constitutional and two acquired disorders involving chromosome 17. For each of your choices, briefly describe the disorder and the genetic mechanisms involved.

Describe the genetic tests that would be appropriate in the diagnosis of male infertility. How do the abnormalities found in these tests relate to the causes of infertility?

Discuss the current technical and scientific limitations of whole genome sequencing analysis within the field of rare disease. Include examples of application into practice. Discuss the implications of a negative result on future testing for the patient.

Discuss with examples how multi-cancer early detection genomics tests may be implemented in a health care setting, including a review of the advantages and disadvantages.

What is meant by the term mosaicism? Describe how mosaicism can contribute to disease, giving examples. What needs to be considered when testing for a mosaic disorder and when reporting a mosaic variant?