Genes and disease

Description Genes in Disease Essay.

Genes name Disease name HFE. Hemochromatosis Introduction (~400 words). • Brief introduction to your gene and its function in health. Using figures it should be possible to address the following points from in this section: Its chromosomal location. How long is the gene? Does the gene have any introns? How long is the protein? How many transcripts have been described and what is the size of your chosen transcript? Main body (~1000 words). • Brief introduction to your disease. • Explain how the mutation you have chosen impacts on normal gene function. • Briefly state the inheritance pattern of your chosen gene/disease pairing. • Explain how genetic tests can be used to diagnose your chosen disease. • Explain how the disease can be treated/managed. • Explain the prognosis for your disease. Conclusion (~100 words). Summarize the findings of your report. No new information should be introduced

BRIEF

* You are required to produce a 1500 word essay on the role of a single gene in disease.
* Your report should consider the following:
  + What is the gene’s normal function?
  + What is the role of the gene in a named disease?
  + How do mutations contribute to the pathogenesis of the disease?
  + How is the disease managed/treated.
* Which gene/disease should you choose?

HOW DO GENES CAUSE DISEASE

* Genes contain the instructions for the production of our proteins.
  + Amongst other things!
* Defects in these instructions can be catastrophic.
  + Mutations.
* Mutations can come in many forms:
  + Single nucleotide polymorphisms.
  + Insertions.
  + Deletions.
* Where do these mutations happen and what impact do they have?

EXONS

* These are the part of the gene which give rise to the protein sequence.
* Mutations within the exon have the potential to impact protein structure/function.
* Exonic polymorphisms can be classed as:
  + Synonymous
  + Non-synonymous