

## Chapter 3 review questions

1. Down syndrome is caused by the presence of an extra copy of chromosome 21 and is also known as Trisomy 21.

a. Name the process that occurs in gamete formation to produce an abnormal number of chromosomes.

b. Name a prenatal test that a woman might have to detect the presence of genetic abnormalities.

c. Why is Down syndrome generally non-familial?

d. Why is it that mothers who are older than 40 have an much increased risk of having a child with Down syndrome?

2. Huntington disease and fragile X syndrome are caused by a section of DNA that is repeated many times.

a. Explain how this type of mutation can give rise to

i. genetic anticipation

ii. premutation

b. What are the consequences of genetic anticipation and premutation in the families of those with Huntington disease and fragile X?

Ans:

1.
  - a. non-disjunction
  - b. amniocentesis, chorionic villi sampling
  - c. non-disjunction is a spontaneous chromosome mutation and people with Down syndrome generally do not reproduce, hence all occurrences are newly arisen.
  - d. The completion of cell division in females only takes place on ovulation (not continuously as in males) after puberty and the activation of immature eggs after many years gives rise to imperfect separation of chromosomes.
  
2.
  - a.
    - i. The expanded repeat section of DNA is unstable and may increase between each generation resulting in more severe cases in later generations. This phenomenon is known as genetic anticipation
    - ii. A lower number of repeats than that necessary to produce the disorder is recognized in relatives of those with the disorder. This intermediate number of repeats between normal and the full expansion is known as a premutation.

b. For Huntington disease, genetic anticipation results in the appearance of HD at an earlier age and greater severity as it is passed on. For fragile X, the symptoms become more severe as it is passed on. Premutation carriers for both disorders are at greater than average risk of producing a child with the disorder