**Genetic and chromosomal disorders**

What methods are used to obtain DNA samples?

|  |  |  |  |
| --- | --- | --- | --- |
| Disorder | Cause | Progression | Diagnostic tests |
| Cystic Fibrosis (CF) |  |  |  |
| Huntington’s |  |  |  |
| Haemophilia |  |  |  |
| Down’s Syndrome |  |  |  |

*When writing about the* ***cause****, you will need to take into account the following 1. Is the disorder caused by a gene mutation or a chromosomal addition? 2. If it is caused by a gene mutation, is the mutation on the dominant or recessive allele? 3. If it is caused by a gene mutation, is the mutated allele found on the sex (X) chromosome?*

**Cancer** is the uncontrolled division of cells.

An accumulation of cells can cause a tumour:

|  |  |  |
| --- | --- | --- |
|  | Benign tumour | Malignant tumour |
| Growth rate |  |  |
| Capacity to spread (to different tissues of the body) |  |  |
| Capacity to cause damage |  |  |

**The role of mutations in development of cancer**

Some genes code for proteins that activate cell division; some genes code for proteins that stop cell division. It is important that the expression of these genes is closely regulated, so cell division is activated when necessary, but also inactivated, to prevent the accumulation of cells.

Proto-oncogenes code for proteins that ……….……….…. cell division.

What effect would a mutated proto-oncogene have on cell division?

Tumour suppressor genes code for proteins that ………………… cell division.

What effect would a mutated tumour-suppressor gene have on cell division?

**Genetic disorders and diagnosis**

**Definitions**

|  |  |
| --- | --- |
|  | Definition |
| Allele |  |
| Dominant |  |
| Recessive |  |
| Genotype |  |
| Phenotype |  |
| Heterozygous |  |
| Homozygous |  |
| Sex linkage |  |
| Carrier |  |