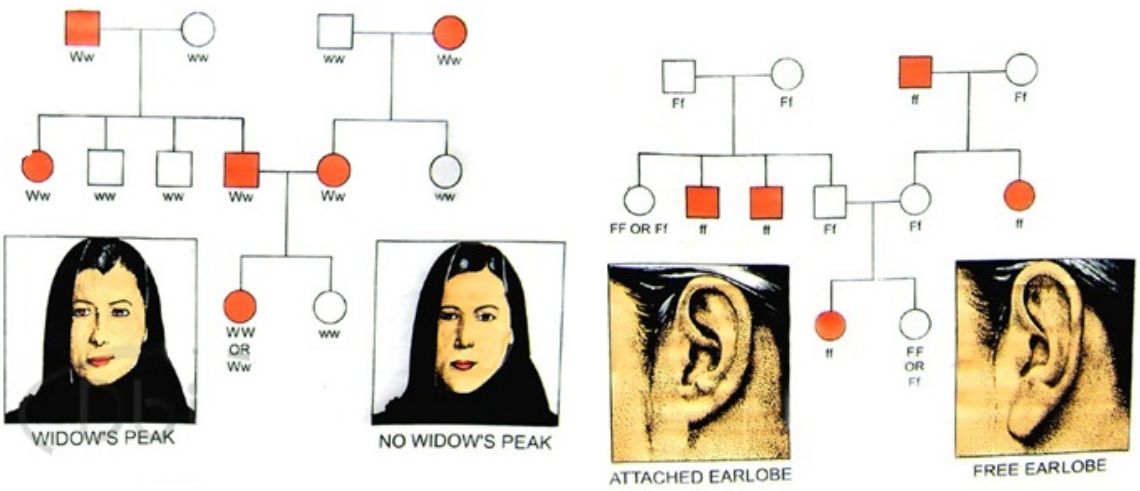


Inheritance of Recessive Human Genetic Disorders

These conditions range from relatively mild (albinism) to life-threatening (cystic fibrosis).

Cystic Fibrosis:

The normal allele for this gene codes for a membrane protein that transports Cl- between cells and extracellular fluid. If these channels are defective or absent, there are abnormally high extracellular levels of chloride. This causes the mucus coats of certain cells to become thicker and stickier than normal. This mucus builds up in the pancreas, lungs, digestive tract, and causes poor absorption of nutrients, chronic bronchitis and bacterial infections.

Sickle-Cell Disease

This is caused by the substitution of a single amino acid in haemoglobin. When oxygen levels in the blood of an affected individual are low, sickle-cell haemoglobin aggregate into long rods that deform RBCs into a sickle shape. The sickle cells clump and clog capillaries throughout the body.

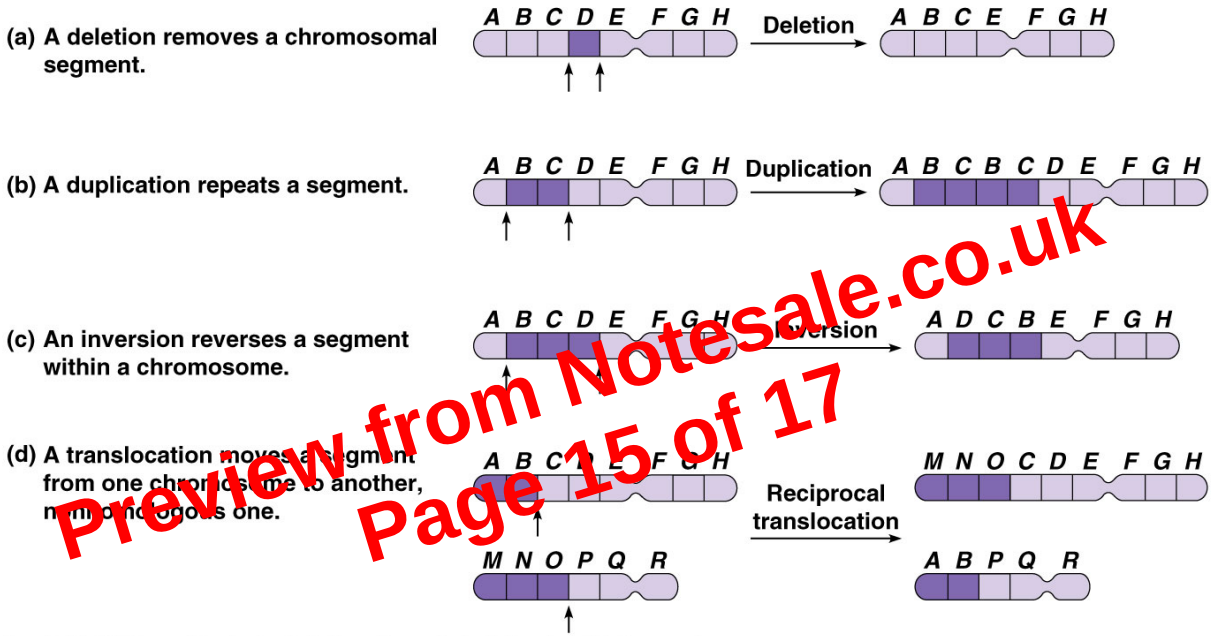
Individuals with sickle-cell allele have increased resistance to malaria. Therefore carriers of sickle-cell disease are relatively free of both.

Inheritance of Dominant Human Genetic Disorders

An example of this is achondroplasia (form of dwarfism). Lethal dominant alleles are much less common than lethal recessives. If a lethal dominant kills an offspring before it can mature and reproduce, the allele will not be passed on to future

Alterations of chromosome structure

- **Deletion**: a chromosome fragment lacking a centromere is lost during cell division, it would be missing certain genes
- **Duplication**: a fragment becomes attached to an extra segment to a sister chromatid. Alternatively, a detached fragment may attach to a nonsister chromatid of a homologous chromosome.
- **Inversion**: a chromosomal fragment reattaches to the original chromosome, but in the reverse orientation.
- **Translocation**: a chromosomal fragment joins a nonhomologous chromosome.



Deletions and duplications are especially likely to occur during meiosis. Homologous chromatids may break up and rejoin at incorrect places during crossing over, so that one chromatid loses more genes than it receives.

Duplications and translocations are typically harmful.

Human disorders due to chromosomal mutations

Surviving individuals of aneuploid conditions have a set of symptoms – a syndrome-characteristic type of aneulpoidy. This can be diagnosed before birth by fetal testing.

Down syndrome

This is due to three copies of chromosome 21 or trisomy 21. They have characteristic facial features, short stature, heart defects, susceptibility to respiratory