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Student Reference Sheet

Genetic Testing - The Facts

Genetic testing involves examining a person's genetic material, that is their chromosomes or DNA, or a particular protein coded for by their DNA. It is examined for some change that indicates the cause or the potential to cause disease or a disorder.

Large changes
Missing or added piece of a chromosome, or even whole chromosome.
Visible to cytogeneticists under a light microscope.
eg Down syndrome - extra chromosome 21.

Small changes
Extra, missing or altered DNA nucleotide at the molecular level.
Analysed by molecular geneticists.
eg cystic fibrosis - single amino acid missing.

Type of Genetic Test	Description and Application	Examples of Disease
1. Diagnostic	Confirms a diagnosis of a specific disorder in a person who already has symptoms and/or signs.	Fragile X Haemochromatosis
2. Presymptomatic • Predictive • Susceptibility	Determines the presence or absence of a mutation in a person with no features of the disorder at the time of testing, in order to predict the likelihood that the person will develop the disorder in the future.	Huntington disease Breast cancer
3. Carrier Testing	Determines the presence of an altered gene or chromosome abnormality which will not affect the person's health, but increase his/her chance of having children with a particular condition.	Thalassaemia Tay-Sachs disease
4. Prenatal	Usually diagnostic or presymptomatic, carried out on a developing fetus.	Duchenne muscular dystrophy
5. Preimplantation genetic diagnosis (PGD)	A form of presymptomatic test carried out on early embryos in the laboratory, with a view to transferring to the mother's uterus only those which will not develop the condition.	Cystic fibrosis
6. Screening	A screening test is one that is performed on individuals with no family history, symptoms or other reason to suggest an increased risk.	PKU newborn screening Maternal serum screening