

This is the **Higher Combined** version — includes Higher Tier content. Some Separate-only details are omitted.

Some genetic disorders are caused by faulty alleles and can be passed from parents to offspring through sexual reproduction.

- Polydactyly: extra fingers or toes. Caused by a **DOMINANT** allele (D). Only **ONE** copy needed to show the condition.
- If one parent has Dd: 50% of children will have polydactyly. If Dd × Dd: 75% chance of polydactyly.
- Cystic fibrosis (CF): thick, sticky mucus in lungs, digestive system and reproductive system. Caused by a **RECESSIVE** allele (f).
- **TWO** copies needed (ff) to have cystic fibrosis. Parents can be carriers (Ff) without showing symptoms.
- Carrier parents (Ff × Ff): 25% chance of affected child (ff), 50% carriers (Ff), 25% unaffected (FF).
- Embryo screening: testing embryos produced by IVF for genetic disorders before implantation. Can select against affected embryos.
- ★ **HT** Sickle cell anaemia: recessive disorder. Red blood cells have abnormal shape → block capillaries, reduced **O₂** carrying. Heterozygotes (sickle cell trait) have some resistance to malaria.
- ★ **HT** Genetic counselling: helps families understand risk of inherited conditions and options available.

Key Terms

Polydactyly	Inherited condition — extra digits — caused by dominant allele (D)
Cystic fibrosis	Inherited condition causing thick mucus — caused by recessive allele (f) — both copies needed to be affected
Carrier	A heterozygous individual (Ff) who carries a recessive allele but does not show the condition
Embryo screening	Testing embryos for genetic disorders before implantation in IVF — ethical controversy
Sickle cell anaemia	Recessive blood disorder — abnormal haemoglobin — heterozygotes resistant to malaria

■ **Exam Tip:** **DOMINANT** = needs only **ONE** copy (polydactyly). **RECESSIVE** = needs **TWO** copies (cystic fibrosis). This distinction is fundamental — learn it. Carriers of recessive conditions are **HETEROZYGOUS** (Ff) — they have the allele but not the condition.