

Inherited disorders

4.6.1.7 · Inheritance, variation and evolution · revision notes

Polydactyly (caused by a dominant allele) and cystic fibrosis (caused by a recessive allele), how they are inherited, and the economic, social and ethical issues of embryo screening. Expect Punnett squares, carrier questions and 4 to 6 mark 'evaluate' questions using information you are given.

Key facts

- Polydactyly = extra fingers or toes; caused by a dominant allele
- Carrier = heterozygous for a recessive disorder: no disorder, but can pass the allele on
- $Dd \times dd \rightarrow 0.5$ polydactyly
- Economic = cost; social = effects on people and society; ethical = right or wrong
- Cystic fibrosis = a disorder of cell membranes; caused by a recessive allele
- $Ff \times Ff \rightarrow 0.25$ CF, 0.5 carrier, 0.25 unaffected non-carrier
- Embryo screening = testing embryo cells for the alleles that cause disorders

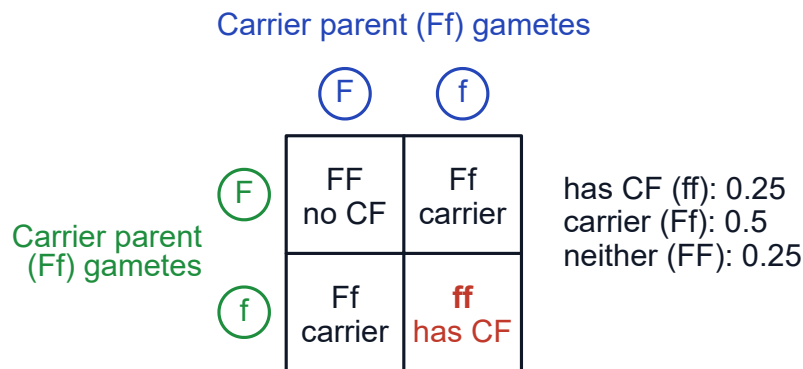
Notes

Two inherited disorders

- Some disorders are **inherited**: they are caused by the inheritance of certain alleles.
- **Polydactyly** (having extra fingers or toes) is caused by a **dominant** allele. One copy is enough to cause it, so there are no carriers.
- **Cystic fibrosis** (CF) is a disorder of **cell membranes**, caused by a **recessive** allele. A person must inherit two copies to have it.
- A person with one CF allele (heterozygous, e.g. Ff) is a **carrier**: they do not have the disorder but can pass the allele on.

Predicting the risk

- Two carriers ($Ff \times Ff$): probability of a child with CF = 0.25; of a carrier = 0.5; of a child who neither has CF nor carries it = 0.25.



Two carriers: a 1 in 4 chance of a child with cystic fibrosis, for every pregnancy.

- A parent with polydactyly (Dd) and a parent without it (dd): probability of a child with polydactyly = 0.5.
- The probability is the same for every pregnancy.

Embryo screening

- In **embryo screening**, cells from an embryo are tested for the alleles that cause a genetic disorder.
- Arguments for: it can prevent suffering from a serious disorder; it can save the long-term cost of treatment.
- Arguments against: it is expensive; embryos with the disorder are usually destroyed, which some people believe is wrong; some fear it could lead to choosing embryos for other characteristics ('designer babies'); some feel it suggests people with disorders are less valued.
- Economic** issues are about money, **social** issues are about the effects on people and society, and **ethical** issues are about what is right or wrong.
- Gene therapy, which aims to correct a faulty allele, is being researched as a possible future treatment.

How to answer each type of question

Explain carriers and allele types

1 to 2 marks

Grade 5

- Polydactyly: dominant allele, extra fingers or toes.
- Cystic fibrosis: recessive allele, disorder of cell membranes.
- Explain carriers using 'heterozygous' or 'one recessive allele'.

Example. Explain why a person can be a carrier of cystic fibrosis but cannot be a carrier of polydactyly. [2 marks]

Model answer. Cystic fibrosis is caused by a recessive allele, so a person with one copy (heterozygous) does not have the disorder but can pass it on (1). Polydactyly is caused by a dominant allele, so anyone with one copy has the condition (1).

Don't lose marks

- ✗ Saying cystic fibrosis is caused by a dominant allele, or polydactyly by a recessive one.
- ✗ Saying a carrier has a mild form of the disorder. A carrier does not have the disorder.
- ✗ Giving the probability of being a carrier when asked for the probability of having the disorder. Read the question carefully.
- ✗ In 'evaluate' questions, giving only arguments against (or only for), or giving no conclusion.

More tips

Memory tricks

- Poly = many, dactyl = digit: polydactyly means extra fingers or toes. D for Dominant, D for Digits.
- C for Cystic fibrosis, C for Carrier, C for Cell membranes: recessive, so carriers exist.
- Two carriers → 1 : 2 : 1 → a quarter affected, half carriers, a quarter clear.

Exam technique

- For ethics questions, use the information provided and quote figures from it.
- Label issues correctly if asked: economic = money, social = people and society, ethical = right or wrong.
- Use the words 'carrier' and 'heterozygous' when describing a person with one cystic fibrosis allele.

What each grade needs

What you need to be able to do, from the first marks up to the top grade.

Grade 3

Describe polydactyly and cystic fibrosis

Polydactyly is having extra fingers or toes; cystic fibrosis is a disorder of cell membranes.

Grade 4

State which allele causes each disorder

Polydactyly is caused by a dominant allele; cystic fibrosis by a recessive allele.

Grade 5

Explain what a carrier is

A heterozygous person with one recessive allele who does not have the disorder but can pass it on.

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