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 × dd → 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 × Ff → 0.25 CF, 0.5 carrier, 0.25 unaffected non-carrier
- Embryo screening = testing embryo cells for the alleles that cause disorders

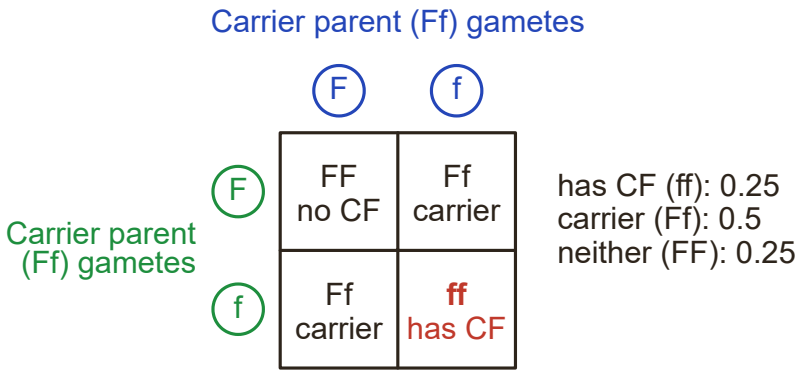
Notes

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 × 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; parents can decide whether to continue the pregnancy or prepare for a child with the disorder.
- Arguments against: it is expensive; it can harm the embryo or cause a miscarriage; the embryo cannot consent; embryos with the disorder are usually destroyed, which some people believe is wrong.
- Some fear screening could lead to choosing embryos for other characteristics ('designer babies'), or 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** aims to correct a faulty allele. Like embryo screening, it may relieve suffering but raises ethical issues.

How to answer each type of question

Explain carriers and allele types

1 to 2 marks | Grade 5

1. Polydactyly: dominant allele, extra fingers or toes.
2. Cystic fibrosis: recessive allele, disorder of cell membranes.
3. 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).

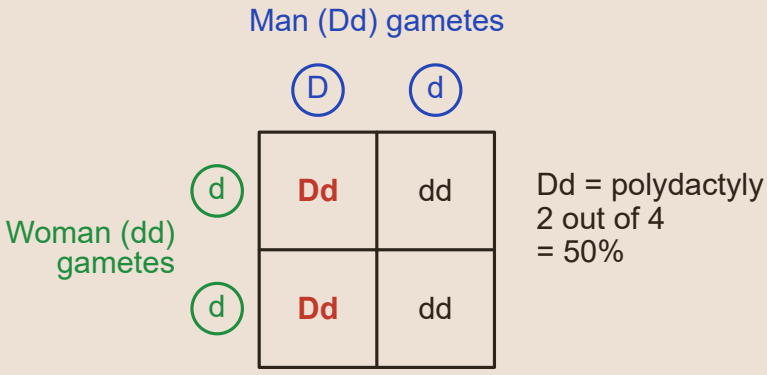
Use a Punnett square for a disorder

3 to 4 marks | Grade 6

1. Write the parents' genotypes (a CF carrier is Ff; a person with polydactyly is usually Dd).
2. Show the gametes and complete the square.
3. Answer exactly what is asked: affected, carrier, or unaffected.

Example. A man with polydactyly (Dd) and a woman without polydactyly (dd) plan to have a child. Use a Punnett square to find the percentage chance that their child will have polydactyly. [3 marks]

Model answer.



Gametes D and d from the man, d and d from the woman (1). Offspring Dd, Dd, dd, dd (1). 2 out of 4 have polydactyly, so 50% (1).

Evaluate embryo screening

4 to 6 marks | Grade 8

1. Use the information given; examiners reward points taken from it.
2. Give arguments for (preventing suffering, cost of lifelong treatment).
3. Give arguments against (cost, destroying embryos, 'designer babies').
4. End with a conclusion that follows from your points.

Example. A couple are both carriers of cystic fibrosis. They are considering IVF with embryo screening, so that only embryos without cystic fibrosis are placed in the mother's uterus. Evaluate the use of embryo screening for this couple. [6 marks]

Model answer.

Marked by levels; a top answer covers both sides and reaches a conclusion. For example:
Each embryo has a 0.25 probability of having CF (1). Screening means their child will not have CF, so it prevents suffering from a lifelong disorder (1). It could also save the cost of long-term medical treatment (1). However, IVF and screening are expensive (1). Embryos with CF would be destroyed, which some people believe is wrong (1). Some people worry it could lead to choosing embryos for other characteristics (1). Conclusion: because the couple have a high risk of a child with a serious disorder, screening is justified if they accept the ethical issues, but it should be their own choice.

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.

Grade 5

Discuss the issues of embryo screening

Give economic, social and ethical arguments for and against, using the information provided.

Grade 6

Predict disorder risks using Punnett squares

For example, two carriers of cystic fibrosis have a 0.25 probability of an affected child.

Grade 8

Evaluate embryo screening with a justified conclusion

Weigh the benefits against the costs and concerns, and make a judgement supported by the information.

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