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Genetic Pedigrees & Modes of Inheritance

Study Guide — Pedigrees

High-school/pre-med-level questions on interpreting pedigrees and identifying modes of inheritance (autosomal vs sex-linked, dominant vs recessive, mitochondrial, and common probability tricks).

30 items — Study Guide with Answers

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1 **In a large family, an inherited disease appears in every generation. Both males and females are affected in roughly equal numbers. An affected father can transmit the disease to both his sons and daughters. Unaffected parents never have affected children (ignoring rare new mutations). Which mode of inheritance best fits this description?**

A **Autosomal dominant** ✓

B Autosomal recessive

C X-linked recessive

D X-linked dominant

E Mitochondrial

▶ **Explanation:** The trait appears in every generation (dominant), affects both sexes equally, and shows male-to-male transmission, all consistent with an autosomal dominant pattern.

2 **In another family, an inherited disorder appears only in siblings (brothers and sisters) of the same generation. Parents are phenotypically normal but may be related (first cousins). Both males and females can be affected. The trait seems to 'skip' generations. Which mode of inheritance is most consistent with this pattern?**

A Autosomal dominant

B **Autosomal recessive** ✓

C X-linked recessive

D Y-linked

E Mitochondrial

▶ **Explanation:** Unaffected parents with affected children of both sexes, especially with consanguinity and generation skipping, is classic for autosomal recessive inheritance.

3 **In a pedigree, an inherited disease affects mainly males. Affected males are usually born to unaffected mothers. There is no father-to-son transmission, but affected males are often related through carrier females. Which mode of inheritance is most likely?**

A Autosomal recessive

B Autosomal dominant

C **X-linked recessive** ✓

D X-linked dominant

E Mitochondrial

▶ **Explanation:** X-linked recessive traits often affect mainly males, are transmitted by carrier females, and show no male-to-male transmission.

4 **In a family, all children (sons and daughters) of an affected woman are affected. However, none of the children of an affected man are affected, unless their mother is also affected. Which inheritance pattern best explains this?**

A Autosomal dominant

B Autosomal recessive

C X-linked dominant

D Y-linked

E **Mitochondrial** ✓

▶ **Explanation:** Mitochondrial DNA is inherited from the mother only. Thus affected mothers pass the trait to all children; affected fathers do not transmit it (unless the mother is also affected).

5 **In a pedigree, an unusual trait appears only in males. Every affected male has an affected father, and all sons of an affected male are affected. Females are never affected. Which mode of inheritance is most consistent?**

A Autosomal dominant

B Autosomal recessive

C X-linked recessive

D **Y-linked** ✓

E Mitochondrial

▶ **Explanation:** Y-linked traits appear only in males and show strict father-to-son transmission.

6 **In a three-generation pedigree, an affected father has no affected sons but all of his daughters are affected. Affected females pass the trait to about half of their sons and half of their daughters. Which inheritance pattern best fits this description?**

A Autosomal dominant

B Autosomal recessive

C X-linked recessive

D **X-linked dominant** ✓

E Y-linked

▶ **Explanation:** In X-linked dominant inheritance, affected males pass the trait to all daughters (they receive his X) and no sons, while affected heterozygous females pass it to about half of all children of either sex.

7 **An autosomal recessive disease is caused by the allele a. Two phenotypically normal parents have had one affected child (aa). Assuming no new mutations, what are the parents' most likely genotypes?**

A AA and AA

B AA and aa

C **Aa and Aa** ✓

D aa and aa

E Aa and aa

▶ **Explanation:** For an autosomal recessive disorder, unaffected parents of an affected child must both be heterozygous carriers (Aa).

8 **For the couple in the previous question (both Aa for an autosomal recessive disease), what is the probability that their next child will be affected by the disease?**

A 1/2

B 1/3

C **1/4** ✓

D 3/4

E 0

▶ **Explanation:** A Punnett square for Aa × Aa gives 1/4 aa (affected), 1/2 Aa (carriers), and 1/4 AA (unaffected non-carriers).

9 **The same carrier couple (Aa × Aa) for an autosomal recessive disease already have one affected child (aa). What is the probability that their next TWO children will both be unaffected?**

A 1/4

B 3/4

C **9/16** ✓

D 1/2

E 3/8

▶ **Explanation:** Each child independently has a 3/4 chance of being unaffected (AA or Aa). For two children: $(3/4) \times (3/4) = 9/16$.

10 **In an autosomal recessive disease (allele a), two carrier parents (Aa × Aa) have an unaffected child. What is the probability that this unaffected child is a carrier (Aa), given that they are known to be phenotypically normal?**

A 1/4

B 1/2

C **2/3** ✓

D 3/4

E 0

▶ **Explanation:** From Aa × Aa, unaffected genotypes are AA and Aa in a 1:2 ratio. Among unaffected children, 2 out of 3 are carriers (Aa).

11 **A woman is a carrier for an X-linked recessive disease (X X). Her partner is completely unaffected (X Y). What is the probability that a SON of this couple will be affected?**

A 0

B 1/4

C **1/2** ✓

D 3/4

E 1

▶ **Explanation:** A son receives Y from his father and either X (normal) or X (mutant) from his mother, with equal probability. Thus 1/2 of sons are affected.

12 **The same couple as in the previous question (carrier mother for an X-linked recessive disease and unaffected father) ask about their DAUGHTERS. What is the probability that a daughter will be affected by the disease?**

A **0** ✓

B 1/4

C 1/2

D 3/4

E 1

▶ **Explanation:** Daughters receive a normal X from their father and either X or X from their mother, making them either normal (X X) or carriers (X X), but not affected in a simple X-linked recessive condition.

13 **An affected man with an X-linked recessive disease (X Y) has children with a completely normal woman (X X). What proportion of their SONS will be affected?**

A **0%** ✓

B 25%

C 50%

D 75%

E 100%

▶ **Explanation:** Sons inherit Y from their father and X from their mother, so all sons are X Y (unaffected). Affected fathers do not transmit X-linked recessive disorders to their sons.

14 In the same cross (affected father X Y × normal mother X X), what proportion of their DAUGHTERS will be carriers?

A 0%

B 25%

C 50%

D **100%** ✓

E It depends on the sex ratio

▶ **Explanation:** All daughters inherit the father's mutant X and the mother's normal X , making them obligate carriers (X X).

15 Why is the term 'carrier' usually NOT used for males in X-linked recessive conditions?

A Because males do not inherit X-linked traits

B Because males always have two X chromosomes

C **Because males have only one X chromosome, so if they carry the mutant allele they are affected** ✓

D Because males cannot transmit X chromosomes to offspring

E Because male pedigrees are not used in genetics

▶ **Explanation:** Males are hemizygous for X-linked genes: having one mutant allele means they express the disease, so they are not 'silent' carriers.

16 **A heterozygous man for an autosomal dominant disease (Aa) has children with an unaffected woman (aa). Assuming full penetrance, what is the probability that a child will inherit the disease?**

A 0

B 1/4

C **1/2** ✓

D 3/4

E 1

▶ **Explanation:** The cross Aa × aa gives 1/2 Aa (affected) and 1/2 aa (unaffected). Each child has a 50% risk.

17 **In the autosomal dominant cross Aa × aa from the previous question (full penetrance), what is the probability that an UNaffected child is a carrier of the disease allele?**

A **0** ✓

B 1/2

C 2/3

D 3/4

E 1

▶ **Explanation:** In a fully penetrant autosomal dominant trait, any individual with allele A is affected. Thus an unaffected child must be aa and cannot carry A.

18 **In a pedigree for a rare disease, two unaffected parents have an affected child. Which of the following inheritance patterns is LEAST likely if there is no evidence of new mutation or reduced penetrance?**

A Autosomal recessive

B X-linked recessive (affected son, carrier mother)

C **Autosomal dominant** ✓

D New mitochondrial mutation in the mother

E Autosomal recessive with consanguinity

▶ **Explanation:** Unaffected parents with an affected child is typical of recessive or X-linked recessive inheritance, or a new mutation. It is inconsistent with a normal fully penetrant autosomal dominant trait.

19 **A pedigree shows many more affected males than females. Affected males are related through females, and there is no male-to-male transmission. Sometimes the trait 'skips a generation' through unaffected females. Which inheritance is most likely?**

A Autosomal recessive

B Autosomal dominant

C **X-linked recessive** ✓

D X-linked dominant

E Mitochondrial

▶ **Explanation:** The pattern of mostly affected males, no male-to-male transmission and 'skipping' via carrier females is typical of X-linked recessive inheritance.

20 **A pedigree shows an inherited disease that affects males and females equally. Affected individuals always have at least one affected parent, and there are numerous examples of affected fathers with affected sons. Which inheritance pattern is LEAST likely?**

A Autosomal dominant

B **Autosomal recessive with new mutations in every affected individual** ✓

C Autosomal recessive with one parent affected and one carrier

D Autosomal recessive with both parents affected

E Mitochondrial

▶ **Explanation:** The described pattern strongly suggests an autosomal dominant trait. Explaining it by recessive inheritance would require highly unlikely coincidental genotypes for many couples.

21 **In a family with a suspected mitochondrial disorder, which observation would argue AGAINST mitochondrial inheritance?**

A An affected mother has several affected sons and daughters

B All children of affected mothers are affected

C **An affected father has several affected sons but an unaffected daughter** ✓

D Affected individuals are present in every generation

E Both males and females are affected

▶ **Explanation:** In mitochondrial inheritance, affected fathers do not pass the disease to their children through sperm mitochondria. Father-to-child transmission (especially to sons only) argues against a purely mitochondrial pattern.

22 **A disease appears only in males in a pedigree. Affected fathers never have affected daughters, but ALL their sons are affected. Which inheritance pattern is most consistent?**

A X-linked recessive

B X-linked dominant

C **Y-linked** ✓

D Autosomal recessive with male-limited expression

E Mitochondrial

▶ **Explanation:** Strict father-to-son transmission confined to males indicates Y-linked inheritance.

23 **Two healthy parents from a small, isolated village (where marriages between relatives are common) have three children, two of whom have the same rare recessive disease. What is the MOST likely explanation?**

A Autosomal dominant disease with incomplete penetrance

B **Autosomal recessive disease with both parents as carriers** ✓

C X-linked dominant disease inherited from the father

D Mitochondrial disease inherited from the father

E A Y-linked trait

▶ **Explanation:** Consanguinity increases the chance that both parents are heterozygous for the same rare autosomal recessive allele.

24 **A new dominant mutation arises in the sperm of a healthy man. He and his healthy partner have one child who is affected by an autosomal dominant disease; there is no prior family history. Which statement is TRUE if this child grows up and has children with an unaffected partner?**

A The disease will not be transmitted further because it was a new mutation

B **Each child has a 50% chance of inheriting the disease allele** ✓

C All of their children will be affected

D None of their children will be affected

E Only their daughters will be affected

▶ **Explanation:** Once the new dominant mutation is present in the child's germline (Aa), it behaves like any other autosomal dominant allele: each child has a 50% risk.

25 **In a pedigree for a rare trait, affected individuals are equally likely to be male or female. When two affected parents have a child, the child is always affected. Unaffected parents never have affected children. Which inheritance pattern is most consistent?**

A **Autosomal dominant** ✓

B Autosomal recessive

C X-linked recessive

D Mitochondrial

E Y-linked

▶ **Explanation:** Equal sex ratio and no skipping of generations with affected × affected producing only affected offspring is characteristic of autosomal dominant inheritance (assuming AA and Aa genotypes are viable).

26 **A pedigree shows equal numbers of affected males and females. An affected male has affected sons and affected daughters. The disease sometimes skips a generation when two carriers have only unaffected children. Which mode of inheritance is most consistent?**

A X-linked recessive

B X-linked dominant

C **Autosomal recessive** ✓

D Mitochondrial

E Y-linked

▶ **Explanation:** Equal sex involvement and generation skipping with affected individuals sometimes born to unaffected parents indicate autosomal recessive inheritance.

27 **A woman is heterozygous for an X-linked dominant disorder. Her partner is unaffected. What proportion of their CHILDREN (regardless of sex) are expected to be affected?**

A 0%

B 25%

C **50%** ✓

D 75%

E 100%

▶ **Explanation:** An affected heterozygous mother passes the mutant X to half of her children, regardless of sex, in an X-linked dominant trait.

28 **Which observation in a pedigree would strongly suggest X-linked DOMINANT rather than autosomal dominant inheritance?**

A Equal numbers of affected males and females

B Unaffected parents having affected children

C **Affected fathers transmit the trait to all daughters but to none of their sons** ✓

D Affected mothers transmit the trait to all sons and no daughters

E Trait appears only in males

▶ **Explanation:** In X-linked dominant inheritance, an affected male passes his mutant X chromosome to all daughters and none of his sons, a pattern not seen in autosomal dominant traits.

29 **In a large pedigree for a rare disease, affected individuals appear in both sexes. The proportion of affected males and females is roughly equal. Parents of affected individuals are usually unaffected. There is no obvious sex bias. Which inheritance pattern is most likely?**

A **Autosomal recessive** ✓

B X-linked recessive

C X-linked dominant

D Y-linked

E Mitochondrial

▶ **Explanation:** Equal affected numbers in males and females with unaffected parents is highly suggestive of autosomal recessive inheritance rather than X-linked recessive.

30 **In a family with a known X-linked recessive disease, a woman has one affected son and one unaffected daughter. The father is unaffected. The mother is therefore an obligate carrier. What is the probability that the unaffected daughter is a carrier?**

A 0

B 1/4

C **1/2** ✓

D 2/3

E 1

▶ **Explanation:** With an unaffected father (X Y) and carrier mother (X X), daughters are either X X (normal) or X X (carriers) in equal proportions, so an unaffected daughter has a $1/2$ chance of being a carrier.