Study Guide — MI Unit 2: How to Screen Your Genes (Mid-Unit Test)

Medical Interventions (Genetics of Disease) · Unit 2 — How to Screen Your Genes · Modeled on the PLTW Guided Review format

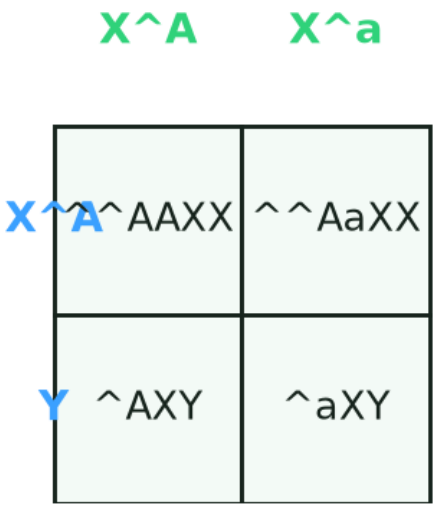
Vocabulary to know cold

Term	Definition
Genotype	The allele combination an organism carries (e.g., Aa).
Phenotype	The observable trait produced by the genotype.
Homozygous	Having two identical alleles (AA or aa).
Heterozygous	Having two different alleles (Aa); often an unaffected carrier for recessive traits.
X-linked recessive	A trait on the X chromosome; affects more males, passed by carrier mothers, no male-to-male transmissio
Mitochondrial inheritance	Trait passed only through the mother to all of her children.
Pedigree	A diagram of family relationships used to track inheritance of a trait.
Punnett square	A grid predicting offspring genotype/phenotype ratios from parent gametes.
Monosomy	A missing chromosome (e.g., 45,X Turner syndrome).
Trisomy	An extra chromosome (e.g., 47,+21 Down syndrome).
Nondisjunction	Failure of chromosomes to separate during meiosis, causing aneuploidy.
No-template control (NTC)	A PCR control with no DNA added; a band in it signals contamination.
Sensitivity	The fraction of true cases a test correctly identifies as positive.

Worked examples (follow the reasoning)

Example 1. A woman who is a carrier for an X-linked recessive disorder (X^A X^a) has children with an unaffected man (X^A Y). Predict the affected fraction of sons and of daughters.

Carrier mother x unaffected father



- Mother's gametes: X^A and X^a. Father's gametes: X^A and Y.
- Offspring: X^A X^A, X^A X^a, X^A Y, X^a Y.
- Sons (got Y from dad): X^A Y unaffected, X^a Y affected -> 1/2 of sons affected.
- Daughters (got X^A from dad): X^A X^A and X^A X^a, both unaffected -> 0 affected (half are carriers).

Answer: 1/2 of sons affected; 0 daughters affected (1/2 of daughters are carriers).

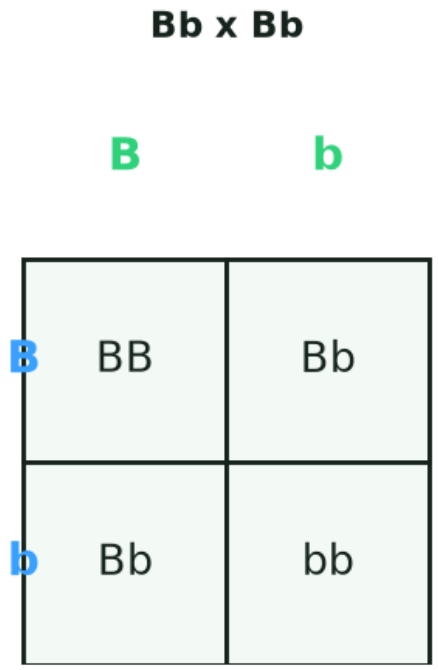
Example 2. Interpret this pedigree: two unaffected parents have one affected and one unaffected child. State the most likely inheritance pattern and the parents' genotypes.

- Both parents are unaffected but produced an affected child -> trait is recessive and 'hidden' in parents.
- An affected child means each parent carried one variant allele.
- Therefore both parents are heterozygous carriers (Aa).

Answer: Autosomal recessive; both parents are carriers (Aa x Aa).

Practice questions (with reasoning)

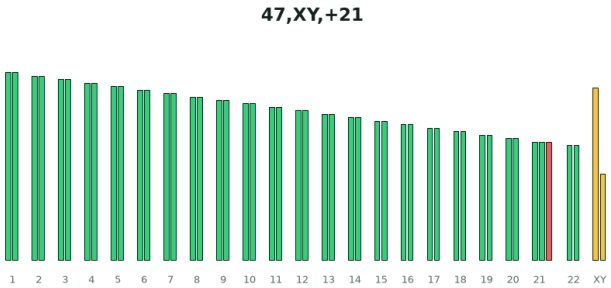
1. Two carrier parents (Bb x Bb) for a recessive disease have three unaffected children. The probability their NEXT child is affected is:



A. 0%
B. 25%
C. 50%
D. 75%

Answer: B. Each pregnancy is independent; the recessive (bb) outcome remains 25%.

2. A karyotype reads 47,XY,+21. The patient has:



A. Turner syndrome
B. Down syndrome
C. A normal male karyotype
D. Monosomy 21

Answer: B. An extra chromosome 21 (47,+21) is Down syndrome.

3. A PCR plate shows a band in the no-template control well. The run should be:

A. Reported as normal
B. Rejected and repeated because of contamination
C. Used only for the patient lanes
D. Diluted and rerun without a control

Answer: B. A band in the NTC means contamination, invalidating the run; it must be repeated.