# Study Guide — MI Unit 2: How to Screen Your Genes (Pretest)

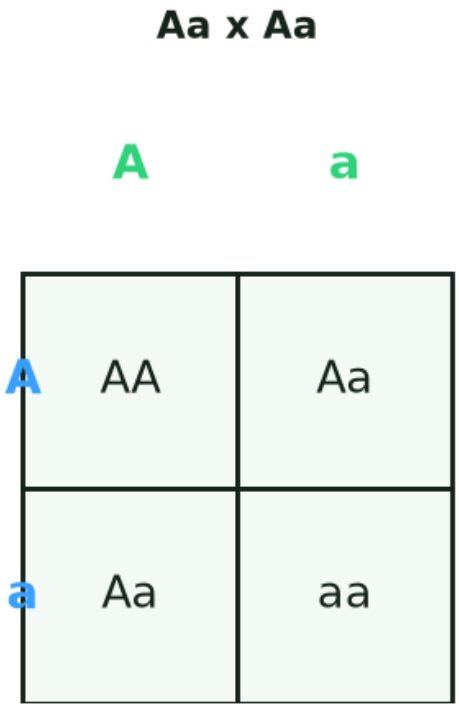
Medical Interventions (Genetics of Disease) · Unit 2 — How to Screen Your Genes · Modeled on the PLTW Guided Review format

## Vocabulary to know cold

| Term | Definition |
|---|---|
| Allele | One of the alternative versions of a gene at a given locus. |
| Autosomal recessive | Inheritance pattern needing two variant alleles to show the trait; can skip generations. |
| Autosomal dominant | Inheritance pattern in which one variant allele is enough to show the trait. |
| Carrier | A heterozygous person (Aa) who is unaffected but can pass the variant allele on. |
| Punnett square | A grid used to predict offspring genotype and phenotype probabilities. |
| Karyotype | An organized display of a cell's chromosomes used to detect number or structure changes. |
| Nondisjunction | Failure of chromosomes to separate in meiosis, leading to extra or missing chromosomes. |
| Trisomy 21 | Three copies of chromosome 21, the cause of Down syndrome. |
| Mutation | A change in the DNA base sequence. |
| PCR | Polymerase chain reaction; amplifies a specific DNA region into many copies. |
| Gel electrophoresis | Separates DNA fragments by size using an electric field; small fragments move farthest. |
| DNA sequencing | Determining the order of nucleotide bases in a DNA strand (Sanger or NGS). |
| Microarray | A chip that tests many DNA sequences or gene-expression levels at once. |
| Informed consent | Voluntary agreement to a procedure after understanding its risks, benefits, and alternatives. |

## Worked examples (follow the reasoning)

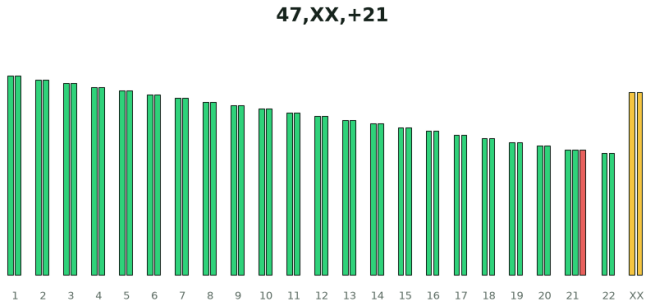
**Example 1.** Two carrier parents (Aa x Aa) plan a child. What is the chance the child is a carrier but unaffected?




- Fill the square: AA, Aa, Aa, aa.
- Carrier (heterozygous) genotypes are the two Aa boxes.
- 2 of the 4 boxes are Aa.

**Answer:** 50% chance the child is an unaffected carrier (Aa).

**Example 2.** A karyotype shows 47 chromosomes with three copies of chromosome 21. Name the condition and the meiotic error that usually causes it.




- Count: 47 chromosomes instead of 46 means an extra chromosome.
- The extra copy is at chromosome 21.
- An extra whole chromosome arises when a pair fails to separate in meiosis.

**Answer:** Down syndrome (trisomy 21), caused by nondisjunction.

## Practice questions (with reasoning)

**1.** A condition appears in every generation and an affected parent often has affected children. This pattern is MOST likely:

**A.** Autosomal recessive
**B.** Autosomal dominant
**C.** Carrier-only
**D.** Caused by aseptic error

**Answer: B.** Dominant traits typically appear in every generation because one allele is enough.

**2.** A scientist needs more copies of a small DNA region before testing it. Which technique should be used?

**A.** Gel electrophoresis
**B.** PCR
**C.** Microarray
**D.** Karyotyping

**Answer: B.** PCR amplifies the target region to provide enough DNA for analysis.

**3.** A base substitution changes one codon but the amino acid stays the same. This is a:

**A.** Silent mutation
**B.** Nonsense mutation
**C.** Frameshift mutation
**D.** Deletion

**Answer: A.** Because the genetic code is redundant, some substitutions do not change the amino acid (silent).