

Study Guide — MI Unit 2: How to Screen Your Genes (End-of-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
Codon	A three-base mRNA sequence that specifies one amino acid or a stop signal.
Silent mutation	A base change that does not alter the amino acid (code redundancy).
Missense mutation	A base change that swaps one amino acid for another (e.g., sickle cell).
Nonsense mutation	A base change that creates a premature stop codon, truncating the protein.
Frameshift mutation	An insertion/deletion not a multiple of three that shifts the reading frame.
Restriction enzyme	A protein that cuts DNA at a specific recognition sequence.
RFLP	Restriction fragment length polymorphism; allele differences detected by altered cut-fragment sizes on a gel.
Sanger sequencing	Chain-termination method that reads one DNA fragment's base order at a time.
Next-generation sequencing (NGS)	Massively parallel sequencing of millions of fragments at once.
Microarray	A chip that tests many DNA sequences or expression levels simultaneously.
Newborn screening	Early post-birth testing for treatable conditions so therapy can start promptly.
Positive control	A sample with known target that should give a positive result, confirming the assay works.
Genetic privacy	Protection of a person's genetic information from unauthorized sharing or discrimination.

Worked examples (follow the reasoning)

Example 1. Translate the mRNA 5'-AUG CGU UAA-3' using the codon chart, then state what happens if the second codon mutates from CGU to UGA.

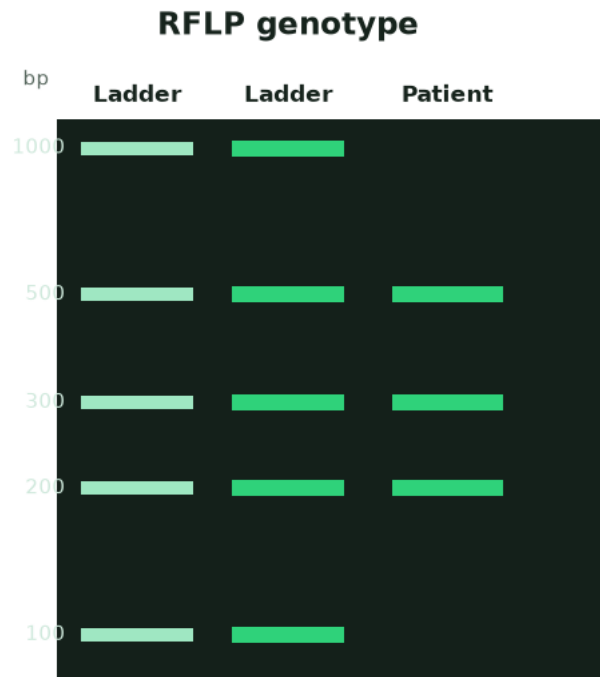
mRNA codon chart

UUU=Phe	UUC=Phe	UUA=Leu	UUG=Leu
UCU=Ser	UCC=Ser	UCA=Ser	UCG=Ser
UAU=Tyr	UAC=Tyr	UAA=Stop	UAG=Stop
UGU=Cys	UGC=Cys	UGA=Stop	UGG=Trp
CUU=Leu	CUC=Leu	CUA=Leu	CUG=Leu
CCU=Pro	CCC=Pro	CCA=Pro	CCG=Pro
CAU=His	CAC=His	CAA=Gln	CAG=Gln
CGU=Arg	CGC=Arg	CGA=Arg	CGG=Arg
AUU=Ile	AUC=Ile	AUA=Ile	AUG=Met
ACU=Thr	ACC=Thr	ACA=Thr	ACG=Thr
AAU=Asn	AAC=Asn	AAA=Lys	AAG=Lys
AGU=Ser	AGC=Ser	AGA=Arg	AGG=Arg
GUU=Val	GUC=Val	GUA=Val	GUG=Val
GCU=Ala	GCC=Ala	GCA=Ala	GCG=Ala
GAU=Asp	GAC=Asp	GAA=Glu	GAG=Glu
GGU=Gly	GGC=Gly	GGA=Gly	GGG=Gly

- AUG = Met (start).
- CGU = Arg.
- UAA = stop, so the original peptide is Met-Arg.
- If CGU -> UGA, the second codon becomes a STOP, so translation ends after Met.
- Replacing an amino-acid codon with a stop codon is a nonsense mutation, producing a shorter (truncated) protein.

Answer: Original peptide: Met-Arg (Met-Arg-Stop). The CGU->UGA change is a nonsense mutation that truncates the protein after Met.

Example 2. An RFLP test: the normal allele is cut into 300 bp + 200 bp; the mutant allele is uncut at 500 bp. A patient's gel lane shows bands at 500, 300, and 200 bp. What is the patient's genotype?



- The 300 + 200 bp bands come from a normal (cut) allele.
- The 500 bp band comes from a mutant (uncut) allele.
- Seeing both patterns means the patient carries one of each allele.

Answer: Heterozygous (carrier): one normal allele and one mutant allele.

Practice questions (with reasoning)

1. A base substitution changes codon GAA to GAG, but both still code glutamic acid. This is a:

mRNA codon chart

UUU=Phe	UUC=Phe	UUA=Leu	UUG=Leu
UCU=Ser	UCC=Ser	UCA=Ser	UCG=Ser
UAU=Tyr	UAC=Tyr	UAA=Stop	UAG=Stop
UGU=Cys	UGC=Cys	UGA=Stop	UGG=Trp
CUU=Leu	CUC=Leu	CUA=Leu	CUG=Leu
CCU=Pro	CCC=Pro	CCA=Pro	CCG=Pro
CAU=His	CAC=His	CAA=Gln	CAG=Gln
CGU=Arg	CGC=Arg	CGA=Arg	CGG=Arg
AUU=Ile	AUC=Ile	AUA=Ile	AUG=Met
ACU=Thr	ACC=Thr	ACA=Thr	ACG=Thr
AAU=Asn	AAC=Asn	AAA=Lys	AAG=Lys
AGU=Ser	AGC=Ser	AGA=Arg	AGG=Arg
GUU=Val	GUC=Val	GUA=Val	GUG=Val
GCU=Ala	GCC=Ala	GCA=Ala	GCG=Ala
GAU=Asp	GAC=Asp	GAA=Glu	GAG=Glu
GGU=Gly	GGC=Gly	GGA=Gly	GGG=Gly

- A. Silent mutation
- B. Missense mutation
- C. Nonsense mutation
- D. Frameshift mutation

Answer: A. The amino acid is unchanged due to code redundancy, so it is a silent mutation.

2. A lab needs to broadly screen one sample against thousands of disease-linked sequences at once. The best choice is a:

- A. Single PCR
- B. Microarray
- C. Karyotype
- D. Gram stain

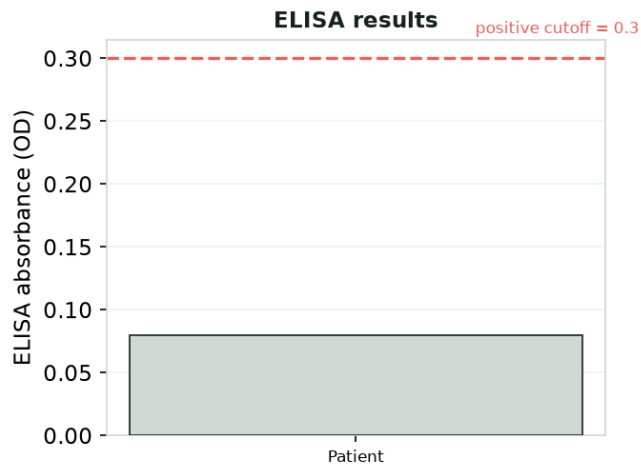
Answer: B. Microarrays test many sequences in parallel, ideal for broad screening.

3. In a PCR screen, a band appears in the no-template control. The correct action is to:

- A. Report results as valid
- B. Repeat the run because of contamination
- C. Ignore the controls
- D. Increase the cycle number

Answer: B. A band in the NTC indicates contamination, so the run is invalid and must be repeated.

4. SPIRAL (Unit 1): A patient's ELISA optical density is below the assay threshold. The result is interpreted as:



- A. Positive
- B. Negative (below threshold)
- C. Invalid
- D. A frameshift

Answer: B. An OD below the cutoff is a negative result for the target antibody/antigen.