Kinds of Typos in DNA

What kinds of typos can occur in DNA?

CARRY FORWARD

Variant interpretation combines clinical assertions, population frequency, molecular consequence, and evidence quality.

10-YEAR TAKEAWAY

DNA variants change proteins in different ways, so variant type is evidence about mechanism, not a verdict by itself.

1. Observe the analogy before naming the biology

Illustration: Edit one instruction manual four different ways

1. Which edit changes one item but keeps the rest?

2. Which edit stops the message early?

3. Which edit shifts every group after it?

2. Turn observations into rules

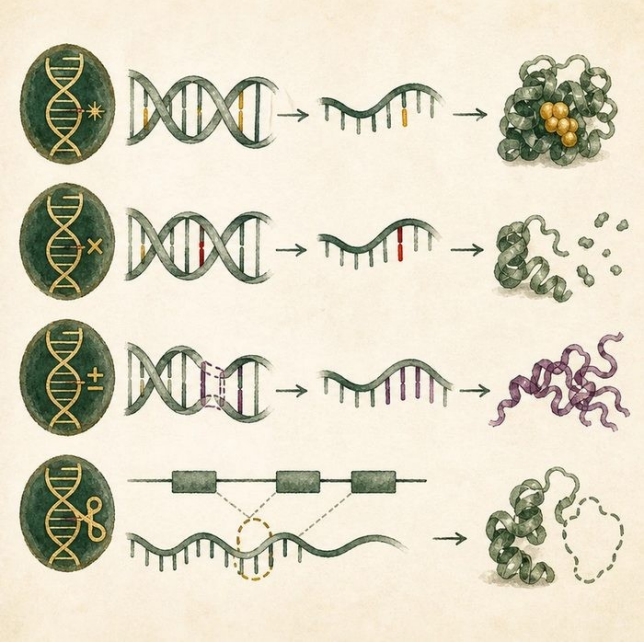
1. Rule 1: Missense swaps one amino acid. Explain it in your own words.

2. Rule 2: Nonsense and frameshift variants can truncate protein. Explain it in your own words.

3. Rule 3: Splice variants can change which RNA sections remain. Explain it in your own words.

Where the analogy stops: Protein consequences depend on location and biology, not only the editing label.

3. Map the rules onto the biomedical example



Biomedical teaching illustration: Translate four IRF6 variant types

Analogy step	Biology step	How the relationship stays the same
Swapped word	Missense variant	_____
Early period	Nonsense variant	_____
Shifted spacing or page join	Frameshift or splice-site variant	_____

4. Use the case evidence

ID	Finding supplied in the case	Why it matters
GEN07-E1	A missense variant replaces one amino acid.	The rest of the reading frame can remain intact.
GEN07-E2	A nonsense or frameshift variant can create a premature stop.	The protein may be shortened or its RNA degraded.
GEN07-E3	A splice-site variant can alter exon joining.	The mature mRNA can lose or gain sequence.

5. Make the clinical decision

You are annotating three IRF6 lab reports. Report A is missense, B is an early nonsense, and C is a splice-site change near an exon boundary.

A. Predict different possible consequences and request location-specific evidence.

B. Call all three identical.

C. Call every missense benign.

Decision. Choose the annotation strategy and cite two distinct mechanisms.

Claim ceiling: You may predict molecular consequences. You may not classify pathogenicity from variant type alone.

Claim. State the choice you can defend.

Evidence. Use these labeled IDs: GEN07-E1 + GEN07-E2.

Reasoning. Explain why the evidence supports the claim and stays inside the claim ceiling.

6. Reduce the lesson to one lasting idea

Take-home. DNA variants change proteins in different ways, so variant type is evidence about mechanism, not a verdict by itself.

1. Which variant type shifts later codons?

2. Why is variant type not a final classification?

Glossary

Term	Plain-English definition
missense	A DNA change that swaps one amino acid in a protein, leaving it full length but possibly altering how it works.
nonsense	A DNA change that creates an early stop codon, halting translation so the protein is cut short and usually nonfunctional.
frameshift	An insertion or deletion not a multiple of three that re-groups every codon after it, scrambling the rest of the protein.
splice-site variant	A change at an intron and exon boundary that disrupts mRNA splicing, so the wrong pieces are joined and the protein is altered.
reading frame	The set of three-letter codons the ribosome reads in order along messenger RNA; a frameshift mutation throws this grouping off.

Next lesson unlock

We can sort IRF6 typos by type. But IRF6 causes two different diseases, a milder one and a more severe one. Why would one gene cause two diseases?

The journal links on the lesson page are optional. Everything required for this guided-notes set is already supplied here and in the case file.