

What Caused Mateo's Cleft?

What caused Mateo's cleft: his genes, something in the pregnancy, or both?

CARRY FORWARD

Epidemiology describes patterns in groups; it does not identify the cause of one child's cleft.

10-YEAR TAKEAWAY

Most isolated clefts reflect many small genetic and environmental influences crossing a developmental threshold.

Cells differentiate

Development sculpts

Cells move

Development can go wrong

1. Observe the analogy before naming the biology



Illustration: Many small drops can fill a threshold cup

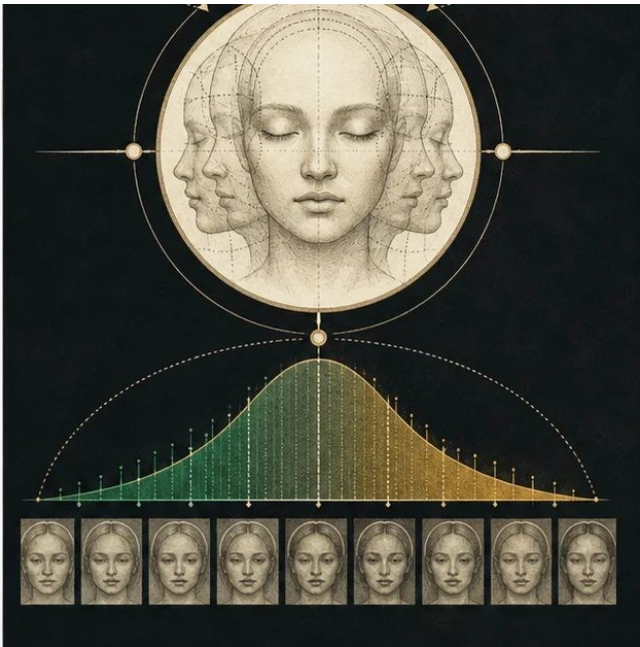
1. Does one drop fill the cup?
2. Could different combinations reach the same line?
3. Can you identify one guilty drop after the cup overflows?

2. Turn observations into rules

- 1. Rule 1: Many small influences can add together. Explain it in your own words.
- 2. Rule 2: Different combinations can cross the same threshold. Explain it in your own words.
- 3. Rule 3: Risk is not certainty and association is not blame. Explain it in your own words.

Where the analogy stops: Biologic risk is not a literal liquid sum. The model simplifies interacting influences into a threshold idea.

3. Map the rules onto the biomedical example



Biomedical teaching illustration: Use the threshold model for an isolated cleft

Analogy step	Biology step	How the relationship stays the same
Small drops	Small genetic variants and environmental exposures	
Fill line	Developmental threshold for disrupted fusion	
Overflow	A cleft forms during the developmental window	

4. Use the case evidence

ID	Finding supplied in the case	Why it matters
D09-E1	Mateo has no clear syndrome pattern or single-gene family pattern.	A simple one-gene explanation is not supported by the case.
D09-E2	Research finds many genetic regions associated with common nonsyndromic clefts.	Multiple small genetic influences can contribute.
D09-E3	Smoking and some other pregnancy exposures are associated with higher risk, but no exposure in Mateo's case proves causation.	Environmental factors change risk without assigning blame for an individual case.

5. Make the clinical decision

You are answering Mateo's parents when they ask, 'What did we do to cause this?' Your answer must be accurate, clear, and free of blame.

- A. Explain multifactorial risk and that no single cause can be identified in this case
- B. Name one exposure as the cause
- C. Say genes always determine the outcome

Decision. Choose the explanation and use all three evidence IDs to set the limits of the claim.

Claim ceiling: You may explain multifactorial risk. You may not claim a specific exposure or variant caused Mateo's cleft.

Claim. State the choice you can defend.

Evidence. Use these labeled IDs: D09-E1 + D09-E2 + D09-E3.

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

6. Reduce the lesson to one lasting idea

Take-home. Most isolated clefts reflect many small genetic and environmental influences crossing a developmental threshold.

1. Why can two families reach the same outcome through different combinations?
2. What is the difference between a risk factor and a cause?

Glossary

Term	Plain-English definition
multifactorial	Caused by several genes acting together with environmental factors, not by a single gene alone.
twin concordance	How often both members of a twin pair share a trait, used to compare identical and fraternal twins and gauge genetic influence.
teratogen	An agent such as a drug, chemical, or infection that can disrupt development and cause birth differences if exposure happens during pregnancy.
risk factor	Anything that raises a person's chance of developing a disease, such as smoking, family history, or high blood pressure, without guaranteeing it.
odds ratio	A number comparing the odds of having a factor in affected versus unaffected people; above 1 suggests the factor is associated.
threshold model	The idea that a trait appears only when a person's combined liability from genes and environment crosses a set line.

Next lesson unlock

Mateo's cleft came from many small influences working together, not one fixable thing. So if no single cause can be removed, what is the actual plan for the next 18 years of his care?

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.