

The Short List: Ruling Out the Big Cleft Syndromes

Which specific syndromes must we rule out for Mateo, and what is the one red flag that flags each?

CARRY FORWARD A cleft is syndromic only when a pattern of additional findings supports that claim.	10-YEAR TAKEAWAY A red-flag checklist turns a broad syndrome question into specific findings that can be checked.
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Cells differentiate	Development sculpts	Cells move	Development can go wrong
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1. Observe the analogy before naming the biology

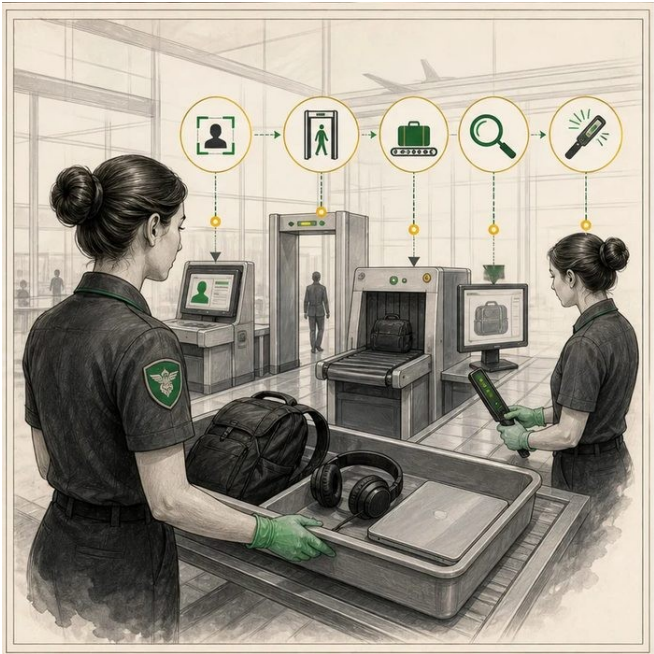


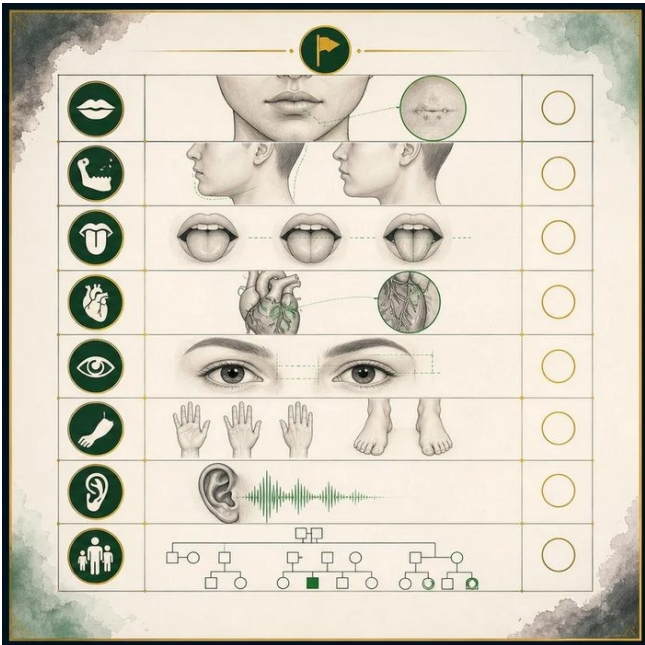
Illustration: Airport screening uses named checks

1. Why are the checkpoints named?
2. Which checks are visual and which need a tool?
3. What would happen if the screener relied on a general feeling?

2. Turn observations into rules

- 1. Rule 1: Break a broad risk into visible checks. Explain it in your own words.
 - 2. Rule 2: Match each concern to a specific sign. Explain it in your own words.
 - 3. Rule 3: A negative check narrows the list but does not erase all uncertainty. Explain it in your own words.
- Where the analogy stops: Patients are not security threats. The analogy only represents systematic screening.

3. Map the rules onto the biomedical example



Biomedical teaching illustration: Match named syndromes to named red flags

Analogy step	Biology step	How the relationship stays the same
Passport check	Family history and lip-pit check	
Bag scan	Heart, eye, jaw, and limb examination	
Secondary screening	Targeted referral or test when a flag is present	

DISEASE LESSON 4 | CASE FILE AND DECISION

4. Use the case evidence

ID	Finding supplied in the case	Why it matters
D04-E1	No lower-lip pits are seen.	A classic Van der Woude clue is absent.
D04-E2	Jaw size and breathing are normal.	The small-jaw and tongue-back pattern of Robin sequence is not present.
D04-E3	Heart exam, eye exam, and limb exam are unremarkable.	Several common syndromic red flags are not present on screening.

5. Make the clinical decision

You are the cleft team coordinator reviewing the red-flag matrix. The team has time for one next step before the family leaves.

- A. Complete the structured family history and genetics examination
- B. Assign a named syndrome from the cleft alone
- C. Stop all evaluation because the first exam is normal

Decision. Choose the next step and show how the red-flag evidence narrows, but does not close, the question.

Claim ceiling: You may say named red flags are absent on today's screen. You may not say every possible syndrome has been ruled out.

Claim. State the choice you can defend.

Evidence. Use these labeled IDs: D04-E1 + D04-E2 + D04-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. A red-flag checklist turns a broad syndrome question into specific findings that can be checked.

1. What is one red flag and the condition it may suggest?
2. Why does a normal screen narrow rather than end the evaluation?

Glossary

Term	Plain-English definition
Van der Woude syndrome	The most common syndromic, single-gene form of cleft lip and palate, usually caused by an IRF6 mutation and often marked by small lower-lip pits.
22q11.2 deletion syndrome	A condition caused by missing a small piece of chromosome 22, leading to heart defects, immune problems, and often cleft palate.
Stickler syndrome	An inherited collagen disorder that often pairs cleft palate with severe nearsightedness, joint problems, and hearing loss.
Pierre Robin sequence	A chain of features starting with a small lower jaw, which pushes the tongue back and blocks the airway and often comes with a cleft palate.
lip pits	Small paired depressions on the lower lip that serve as the hallmark sign flagging Van der Woude syndrome, a cleft-related disorder.
micrognathia	An unusually small lower jaw that can crowd the tongue backward and is sometimes linked with cleft palate and breathing trouble.
glossoptosis	Backward and downward displacement of the tongue, which can block the airway and is a key feature of Pierre Robin sequence.
velopharyngeal insufficiency	When the soft palate cannot fully seal off the nose during speech, letting air escape and making speech sound nasal.

Next lesson unlock

We now have a checklist of red flags, and Mateo trips none of them on inspection. But a checklist you eyeball by hand can miss the quiet ones: a tiny deletion of DNA causes no obvious sign, and subtle features hide. How does the team make the syndromic-versus-isolated call rigorous, with an exam protocol and a test, instead of just a confident glance?

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.