STUDENT CASE FILE | DISEASE DOMAIN | LESSONS 1 TO 20

The Baby Mateo Case

One observed structural difference. Twenty evidence updates. One bounded clinical synthesis.

Composite patient snapshot

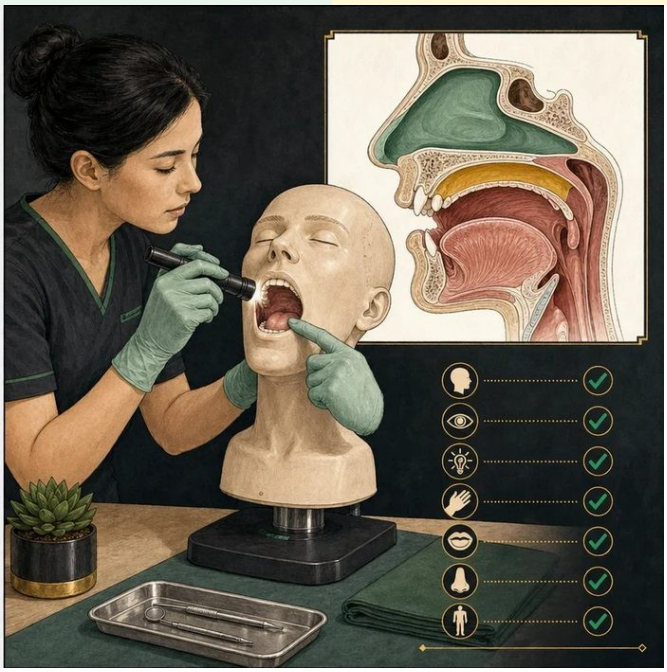
Patient	Mateo, a composite teaching case. No real patient data.
Birth	Term newborn. Breathing and tone are stable.
Observed structure	Complete unilateral left cleft lip and palate.
Initial whole-body exam	No additional congenital anomalies identified on the first structured examination.
Inquiry target	Is the cleft isolated or part of a syndrome? What evidence supports the cause model and care plan?
Evidence rule	Use only the labeled findings supplied in the lesson. Do not invent history, tests, symptoms, or family details.

EVIDENCE SUPPLIED

Every required finding has a stable ID.

EVIDENCE NOT SUPPLIED

Do not invent missing symptoms, tests, exposures, or family history.

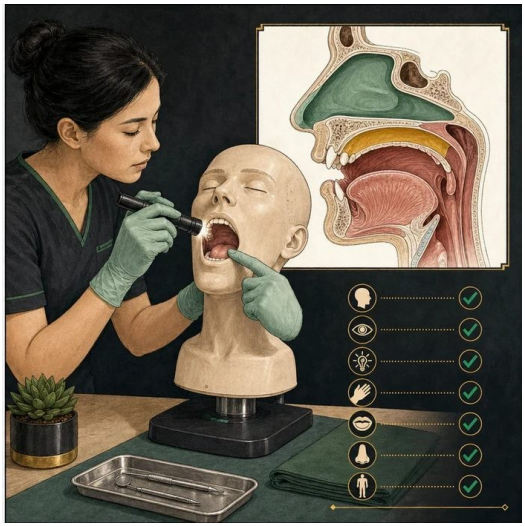


Illustration, not a clinical photograph. Composite craniofacial teaching model.

EVIDENCE UPDATE 1 OF 20 | PEDIATRICIAN (CLEFT TEAM COORDINATOR)

Day One: Recognizing and Communicating a Cleft at Birth

Day-one cleft care begins with a complete exam and clear, respectful communication.



Biomedical teaching illustration: Map the inspection onto Mateo's newborn exam

ID	Finding supplied in the case	Why it matters
D01-E1	The upper lip has a visible gap on the left side.	The lip finding is left-sided and unilateral.
D01-E2	The palate gap is seen with a light and confirmed by palpation.	The palate finding is real and should not be missed by inspection alone.
D01-E3	Breathing, tone, heart sounds, eyes, ears, hands, feet, and jaw size are otherwise unremarkable.	The chart must include important normal findings while the team keeps the cause open.

Decision in this update: Which response will you give, and which two evidence IDs make it the safest choice?

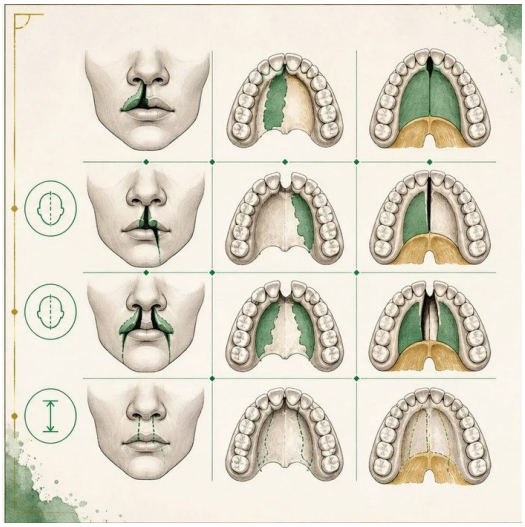
Claim ceiling: You may describe the structural finding. You may not claim a genetic cause or final isolated-versus-syndromic classification yet.

Handoff to the next update: We saw and named Mateo's cleft, but 'a cleft' is not precise enough for a chart or a surgical plan. Exactly which structures are split, on which side, and how completely? We need the team's shared language for describing it.

EVIDENCE UPDATE 2 OF 20 | PEDIATRICIAN (CLEFT TEAM COORDINATOR)

Describing the Cleft: Type, Side, and How Complete

A useful cleft description names the structures, the side, and the completeness as three separate choices.



Biomedical teaching illustration: Sort Mateo on three clinical axes

ID	Finding supplied in the case	Why it matters
D02-E1	The gap involves both the lip and the palate.	Structure category: cleft lip and palate.
D02-E2	Only the left side of the lip is involved.	Laterality: unilateral, left.
D02-E3	The lip gap reaches the nostril and the palate is open along its length.	Completeness: complete.

Decision in this update: Choose the registry description and cite one evidence ID for each word that narrows the description.

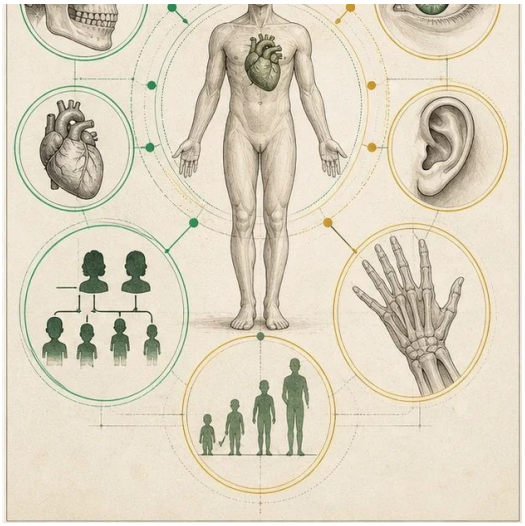
Claim ceiling: You may classify anatomy. You may not infer a syndrome from the cleft pattern alone.

Handoff to the next update: We can describe Mateo's cleft exactly: complete, unilateral, left, lip and palate. But a precise description still does not tell us WHY. A cleft can travel alone, or it can be one clue to a bigger condition. Could Mateo's cleft be a sign of a larger syndrome?

EVIDENCE UPDATE 3 OF 20 | GENETICIST / DYSMORPHOLOGIST (CLEFT TEAM)

Is the Cleft a Clue? The Syndromic Question

A cleft is syndromic only when a pattern of additional findings supports that claim.



Biomedical teaching illustration: Ask whether the cleft travels alone

ID	Finding supplied in the case	Why it matters
D03-E1	Mateo has a complete unilateral left cleft lip and palate.	This confirms the structural finding but does not classify the cause.
D03-E2	No jaw, heart, limb, eye, ear, or tone abnormality was found on the newborn exam.	The first exam does not show a broader pattern.
D03-E3	Family history and targeted syndrome checks are not yet complete.	The classification must remain provisional.

Decision in this update: Choose the most defensible chart phrase and explain why the other two go beyond the evidence.

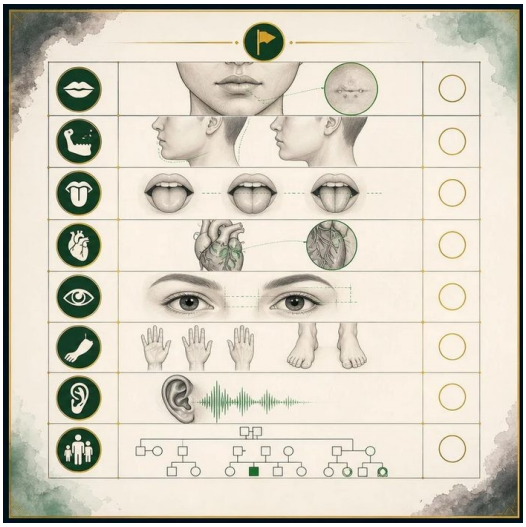
Claim ceiling: You may state that no red flags have been found so far. You may not call the cleft isolated or nonsyndromic until the evaluation is complete.

Handoff to the next update: We learned that a cleft can ride alone or come bundled with other features, so the only honest answer right now is 'we do not know yet, we have to check.' But 'check for a syndrome' is too vague to act on. Which specific syndromes must a cleft team actually rule out, and what red flags point to each one?

EVIDENCE UPDATE 4 OF 20 | GENETICIST / DYSMORPHOLOGIST (CLEFT TEAM)

The Short List: Ruling Out the Big Cleft Syndromes

A red-flag checklist turns a broad syndrome question into specific findings that can be checked.



Biomedical teaching illustration: Match named syndromes to named red flags

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.

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

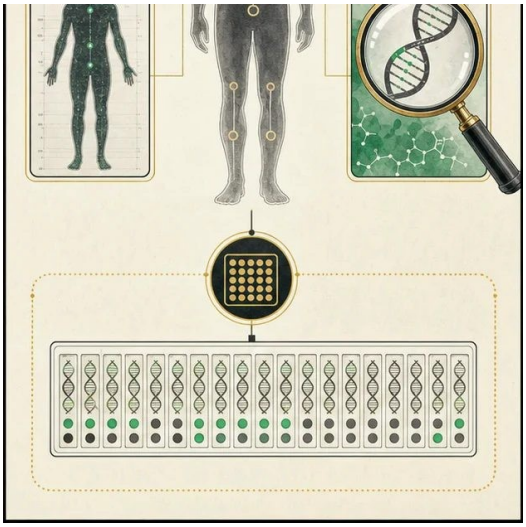
Claim ceiling: You may say named red flags are absent on today's screen. You may not say every possible syndrome has been ruled out.

Handoff to the next update: 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?

EVIDENCE UPDATE 5 OF 20 | GENETICIST / DYSMORPHOLOGIST (CLEFT TEAM)

The Exam and the Test That Sort Syndromic from Isolated

Testing is most useful when the history and exam create a question the test can answer.



Biomedical teaching illustration: Let the exam choose the genetic test

ID	Finding supplied in the case	Why it matters
D05-E1	Mateo's structured exam shows the cleft but no additional congenital anomalies.	The current pattern is more consistent with an isolated finding than a multiple-anomaly pattern.
D05-E2	The three-generation family history shows no repeated cleft pattern or matching syndrome features.	There is no clear single-syndrome target from family history.
D05-E3	Chromosomal microarray is best at detecting missing or extra DNA segments, not every possible DNA change.	A broad test has a defined scope and a normal result has limits.

Decision in this update: Choose a testing plan, cite the case evidence, and state one thing your plan cannot rule out.

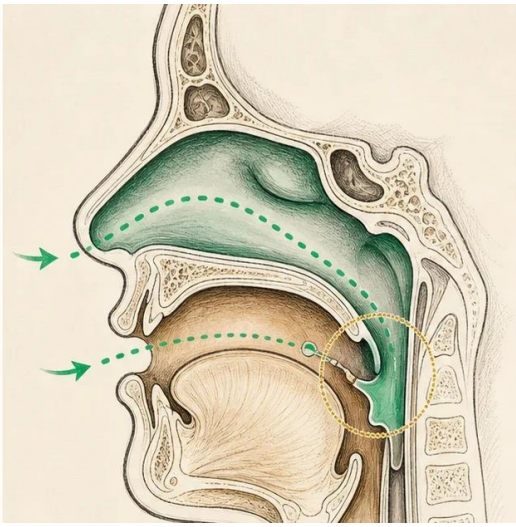
Claim ceiling: You may justify a measured testing plan. You may not say a normal microarray proves there is no genetic contribution.

Handoff to the next update: We sorted the exam and tests that separate a syndrome from a lone cleft, and Mateo's structured exam and testing point toward an isolated cleft. That workup takes time to complete, and meanwhile a two-day-old still has to eat. So how do we keep Mateo fed and growing while the rest of the picture comes together?

EVIDENCE UPDATE 6 OF 20 | FEEDING SPECIALIST

Keeping Mateo Fed and Growing

An open palate limits suction, so feeding support replaces suction with flow control and positioning.



Biomedical teaching illustration: Replace missing suction with controlled assistance

ID	Finding supplied in the case	Why it matters
D06-E1	Mateo tires during feeding and cannot maintain suction on a standard bottle.	The open palate is interfering with pressure generation.
D06-E2	Breathing is calm and oxygen level remains normal during the observed feed.	The immediate problem is feeding mechanics rather than respiratory failure.
D06-E3	With an assisted-delivery bottle, upright position, and pauses, intake improves.	The modified plan addresses the mechanism.

Decision in this update: Choose the discharge plan and connect each part to the evidence and pressure model.

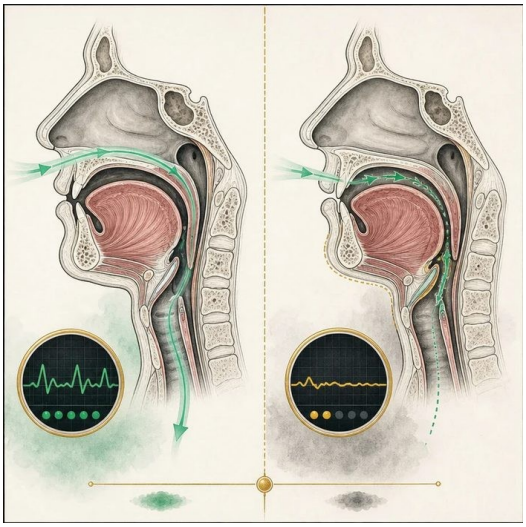
Claim ceiling: You may recommend the observed safe strategy. You may not assume every infant with a cleft has the same feeding ability.

Handoff to the next update: We answered today's question: Mateo can be fed with the right bottle and position. But most is not all. A few cleft babies struggle not just to eat but to breathe, especially when the jaw is small and the tongue falls back. When is a cleft an actual airway emergency?

EVIDENCE UPDATE 7 OF 20 | NEONATOLOGIST

When a Cleft Is an Airway Emergency

In Robin sequence, the small jaw lets the tongue fall back and block the airway; the palate gap itself is not the blockage.



Biomedical teaching illustration: Trace the Robin sequence mechanism

ID	Finding supplied in the case	Why it matters
D07-E1	Mateo has normal jaw size and no tongue-back position on examination.	The key Robin sequence mechanism is absent.
D07-E2	Breathing is quiet, oxygen level is normal, and there are no retractions.	There is no current sign of airway distress.
D07-E3	A comparison infant has micrognathia, glossoptosis, noisy breathing, and oxygen drops when lying flat.	This pattern requires urgent airway action.

Decision in this update: Choose the infant who needs escalation and trace the mechanism in order.

Claim ceiling: You may rule on the current airway signs. You may not claim that Mateo can never develop a breathing problem later.

Handoff to the next update: Mateo's airway is safe, his cleft is isolated, and feeding is handled. Step back: how common is a cleft like his, and in whom does it happen most?

EVIDENCE UPDATE 8 OF 20 | EPIDEMIOLOGIST

How Common Is Mateo's Cleft, and in Whom?

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

Biomedical teaching illustration: Read population patterns without overclaiming

ID	Finding supplied in the case	Why it matters
D08-E1	Cleft lip with or without cleft palate occurs in roughly 1 in 1,000 births in current U.S. CDC estimates.	The condition is uncommon but not rare in population terms.
D08-E2	Unilateral clefts are more common than bilateral clefts, and left-sided unilateral clefts are more common than right-sided ones.	Mateo fits common laterality patterns.
D08-E3	Mateo is one patient with a specific history.	Population rates cannot identify his individual cause.

Decision in this update: Write the two sentences and label which one is population-level and which one is patient-level.

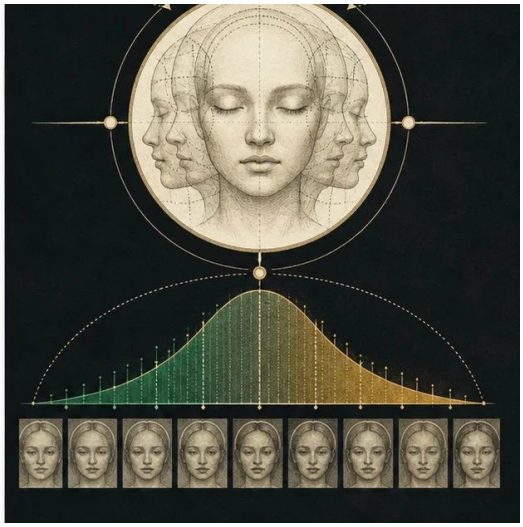
Claim ceiling: You may describe prevalence and pattern. You may not use group data to claim why Mateo's cleft happened.

Handoff to the next update: Mateo's cleft fits the single most common pattern, which means it is not rare and not random. So what actually caused it: his genes, something in the pregnancy, or both?

EVIDENCE UPDATE 9 OF 20 | PEDIATRICIAN / GENETICIST (CLEFT TEAM)

What Caused Mateo's Cleft?

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



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

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.

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

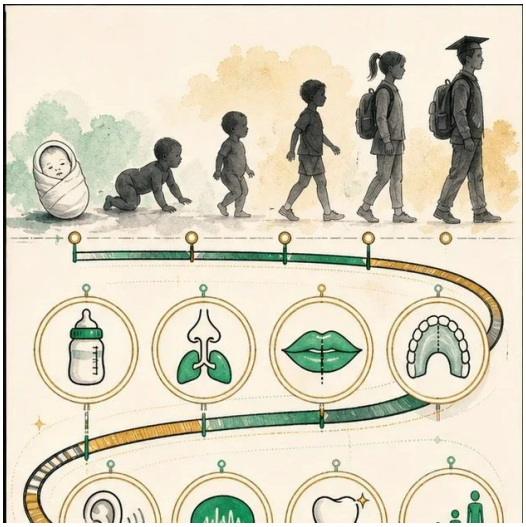
Claim ceiling: You may explain multifactorial risk. You may not claim a specific exposure or variant caused Mateo's cleft.

Handoff to the next update: 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?

EVIDENCE UPDATE 10 OF 20 | CLEFT TEAM COORDINATOR

The Plan for the Next 18 Years

Cleft care follows developmental windows, so the right action depends on what the child needs next.



Biomedical teaching illustration: Build Mateo's care around developmental windows

ID	Finding supplied in the case	Why it matters
D10-E1	Mateo needs feeding and growth follow-up now, before surgery.	Immediate needs come first in the sequence.
D10-E2	Lip and palate operations are staged at different times for different purposes.	One operation cannot solve every problem.
D10-E3	Hearing, speech, dental growth, psychosocial health, and facial growth require repeated checks over years.	The plan must extend beyond the first repair.

Decision in this update: Choose the plan structure and place each evidence item in the correct time band.

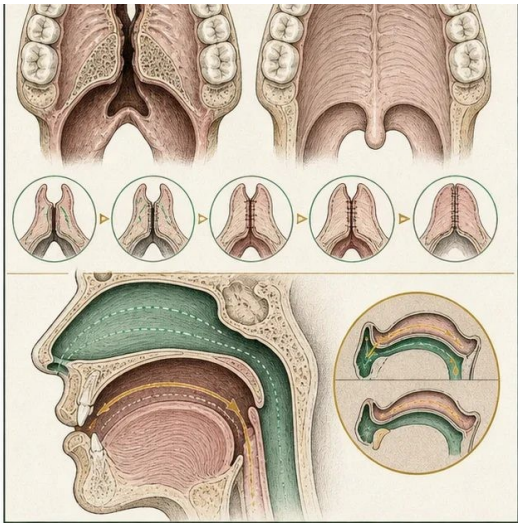
Claim ceiling: You may provide typical windows and ownership. You may not promise exact dates before individual evaluation.

Handoff to the next update: We have an 18-year care plan for Mateo, but what do the operations on that plan actually fix? What do the surgeries accomplish clinically?

EVIDENCE UPDATE 11 OF 20 | PLASTIC SURGEON (CLEFT TEAM)

What the Surgeries Are Actually For

Lip repair restores form, while palate repair separates spaces and supports speech; different jobs require staged operations.



Biomedical teaching illustration: Match each operation to its clinical job

ID	Finding supplied in the case	Why it matters
D11-E1	Mateo's parents want the lip and nose to have continuous form.	This concern maps to lip repair.
D11-E2	The open palate connects the mouth and nose and cannot support normal oral pressure for speech.	This functional problem maps to palate repair.
D11-E3	Speech sounds begin developing during the same period in which palate repair is commonly planned.	Timing protects a developmental window.

Decision in this update: Choose the explanation and match each evidence ID to the operation it supports.

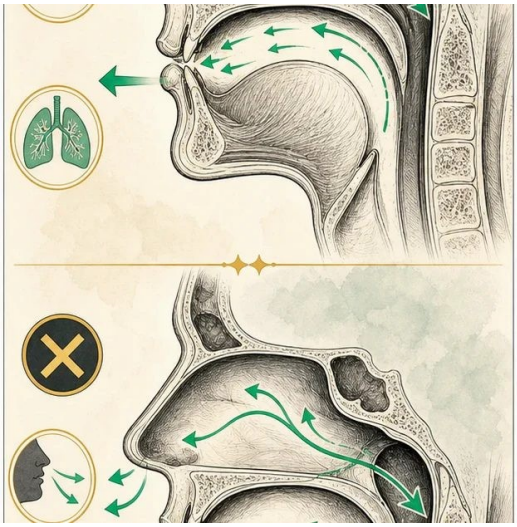
Claim ceiling: You may explain typical goals and timing. You may not promise a specific operation date or outcome.

Handoff to the next update: We answered why the palate is repaired early to protect speech. But that raises a new question: how exactly does a repaired palate let Mateo make clear speech sounds, and what happens if it still does not seal well?

EVIDENCE UPDATE 12 OF 20 | SPEECH-LANGUAGE PATHOLOGIST (CLEFT TEAM)

How a Repaired Palate Lets Mateo Talk

Clear pressure sounds need the soft palate to move upward and seal the mouth from the nose.



Biomedical teaching illustration: Watch the soft palate act as a moving seal

ID	Finding supplied in the case	Why it matters
D12-E1	Mateo's voice sounds overly nasal during connected speech.	Resonance suggests air and sound are entering the nose at the wrong time.
D12-E2	The consonants p, b, t, d, k, and g sound weak and nasal air is visible on a mirror.	Pressure consonants are affected by a seal problem.
D12-E3	Vowels and low-pressure sounds are easier to produce.	The problem is patterned rather than random.

Decision in this update: Choose the next step and use the pattern across the three evidence IDs.

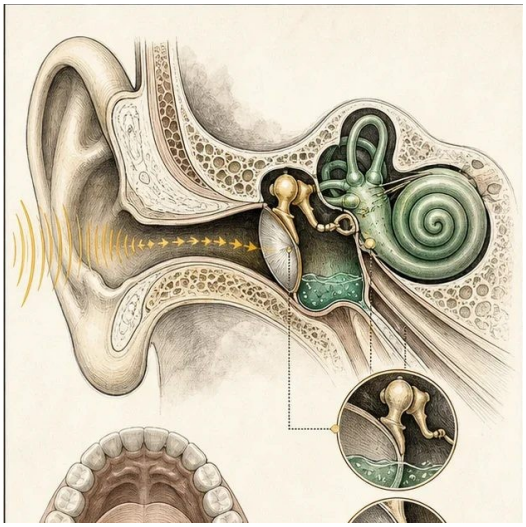
Claim ceiling: You may identify a pattern consistent with VPI. You may not diagnose the exact structural cause without further evaluation.

Handoff to the next update: We answered how the palate lets Mateo make clear speech and what VPI is. But the same little muscles near the palate do another job: they help drain the middle ear. Mateo keeps getting ear infections and does not always turn to soft sounds. How do we protect his hearing?

EVIDENCE UPDATE 13 OF 20 | AUDIOLOGIST (CLEFT TEAM)

Protecting Mateo's Hearing

Cleft-related middle-ear fluid can reduce hearing, so repeated hearing checks protect speech development.



Biomedical teaching illustration: Connect palate muscles to middle-ear drainage

ID	Finding supplied in the case	Why it matters
D13-E1	Mateo passed the newborn hearing screen.	Hearing was adequate at that moment, not guaranteed for all later months.
D13-E2	At follow-up, both eardrums show reduced movement and middle-ear fluid.	Sound conduction may now be reduced.
D13-E3	Mateo sometimes does not turn toward soft speech sounds.	The functional observation supports prompt audiology testing.

Decision in this update: Choose the plan and explain why the newborn result does not cancel the new evidence.

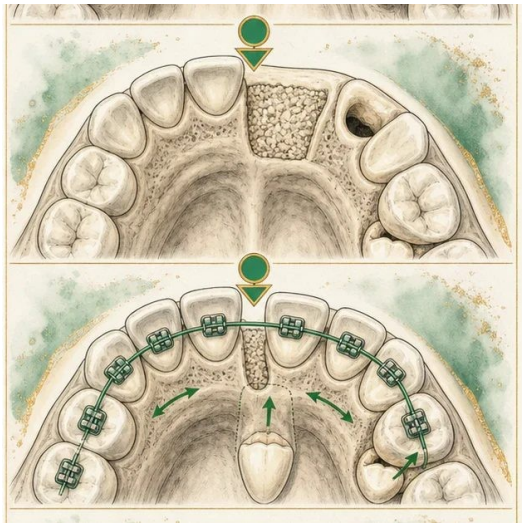
Claim ceiling: You may say hearing is at risk and testing is needed. You may not state the degree of loss before audiology results.

Handoff to the next update: We answered why the cleft palate threatens hearing and how the team protects it. But Mateo's first teeth are coming in, and his parents notice some are crooked and one looks unusually small. How do we care for his teeth and his bite over the years ahead?

EVIDENCE UPDATE 14 OF 20 | DENTIST / ORTHODONTIST (CLEFT TEAM)

Caring for Mateo's Teeth and Bite

Dental and orthodontic care is staged because teeth, bone, and facial growth change on different schedules.



Biomedical teaching illustration: Sequence care through the tooth-bearing cleft

ID	Finding supplied in the case	Why it matters
D14-E1	Mateo's cleft passes through the left tooth-bearing alveolar ridge.	Teeth and supporting bone near the cleft are at risk for predictable differences.
D14-E2	One lateral incisor is missing and nearby teeth are rotated.	The dental pattern matches the location of the cleft.
D14-E3	The permanent canine has not yet reached the usual graft-planning stage.	The team must monitor growth and eruption before choosing the next procedure.

Decision in this update: Choose the staged plan and identify what happens now versus what waits for a developmental marker.

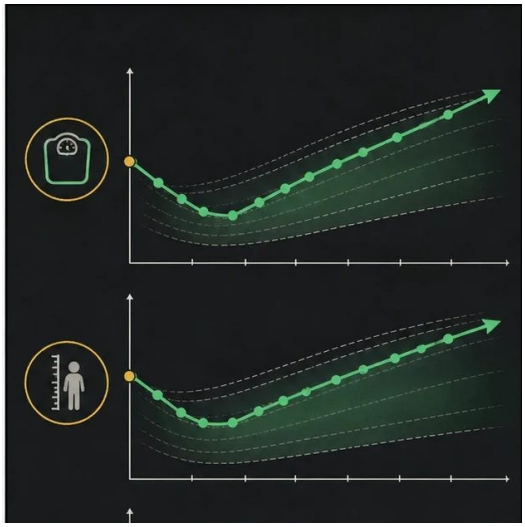
Claim ceiling: You may outline the sequence. You may not set a surgery date without current imaging and eruption assessment.

Handoff to the next update: We answered why the cleft changes Mateo's teeth and bite and why dental care is staged. But now Mateo has a lot going on at once: surgeries, speech therapy, ear care, and dental visits. With all of that, how do we make sure the most basic thing is still on track, that he is actually growing and developing well overall?

EVIDENCE UPDATE 15 OF 20 | PEDIATRICIAN (CLEFT TEAM)

Is Mateo Growing Well?

Growth decisions come from trends across time, not one measurement on one day.



Biomedical teaching illustration: Read Mateo's growth as a longitudinal pattern

ID	Finding supplied in the case	Why it matters
D15-E1	Mateo's weight dropped during the first feeding week but rose steadily after the assisted-feeding plan began.	The intervention appears to improve intake.
D15-E2	Length and head circumference continue along their prior percentile bands.	Overall physical growth remains proportionate.
D15-E3	Motor, language, and social milestones are appropriate for the visit.	Developmental progress supports continued monitoring rather than an emergency conclusion.

Decision in this update: Choose the plan and describe the trend across all three evidence IDs.

Claim ceiling: You may judge the current trend. You may not promise that later growth or facial development will stay normal.

Handoff to the next update: We answered how the team keeps Mateo growing and developing well. His body and growth are well cared for. But a child is more than a body. Mateo and his parents face stares, worry, the stress of many appointments, and questions about how he will feel about himself. How do we support Mateo and his family beyond the body?

EVIDENCE UPDATE 16 OF 20 | PSYCHOLOGIST / SOCIAL WORKER (CLEFT TEAM)

Supporting Mateo and His Family Beyond the Body

Psychosocial care belongs in the treatment plan because health includes confidence, relationships, school, and family well-being.

Biomedical teaching illustration: Assess the whole child, not only the repaired anatomy

ID	Finding supplied in the case	Why it matters
D16-E1	Mateo's family reports stress from repeated appointments and worry about other people's reactions.	Family burden is part of the current health picture.
D16-E2	Mateo participates in class but avoids speaking during a new peer group activity.	A context-specific concern deserves a private, nonjudgmental check-in.
D16-E3	No crisis signs are reported, and Mateo identifies one trusted adult and one supportive friend.	The plan can build on existing strengths while monitoring needs.

Decision in this update: Choose the support plan and separate the evidence from assumptions about how Mateo feels.

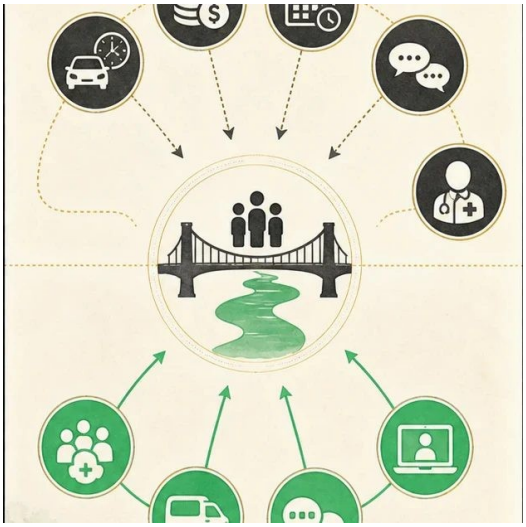
Claim ceiling: You may identify needs reported in the case. You may not assign feelings, diagnoses, or stigma experiences that Mateo did not report.

Handoff to the next update: We answered what Mateo's family needs beyond the body. But we have assumed his team will provide all of this. Does every child like Mateo actually get the same care, whether they are born in a big city, a rural town, or a low-income country?

EVIDENCE UPDATE 17 OF 20 | CLEFT TEAM COORDINATOR (HEALTH-EQUITY LENS)

Does Every Child Like Mateo Get the Same Care?

Equal treatment is not always equitable care; the plan must remove the barriers that block access to the same health goal.



Biomedical teaching illustration: Redesign the route to coordinated cleft care

ID	Finding supplied in the case	Why it matters
D17-E1	The family missed two visits because the caregiver could not leave work and the clinic is across the city.	Scheduling and transportation are documented barriers.
D17-E2	The next required services are hearing and speech follow-up, both available through the hospital network.	The clinical goal is clear and should remain unchanged.
D17-E3	The network offers combined appointments, transportation support, and telehealth check-ins when clinically appropriate.	Available supports match the named barriers.

Decision in this update: Choose the recovery plan and match each support to one documented barrier.

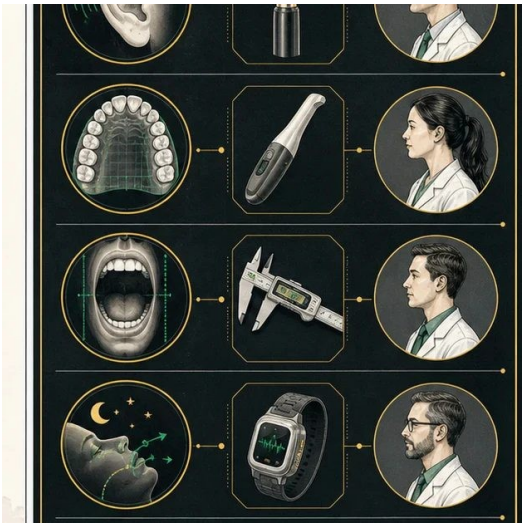
Claim ceiling: You may respond to the documented barriers. You may not invent income, immigration, language, or family details not in the case.

Handoff to the next update: We answered that access is unequal, and that Mateo is fortunate to have a full team. But even with full access, the work is not finished. A repaired cleft is not a cured cleft. What problems can arise later for Mateo, even years after surgery, and how would the team catch them?

EVIDENCE UPDATE 18 OF 20 | CLEFT TEAM COORDINATOR (LONG-TERM SURVEILLANCE)

A Repaired Cleft Is Not a Cured Cleft

A repaired cleft still needs surveillance because speech, hearing, teeth, airway, and growth can change later.



Biomedical teaching illustration: Match each possible complication to its detector

ID	Finding supplied in the case	Why it matters
D18-E1	Mateo has new nasal air escape during fast speech.	Speech and velopharyngeal function need reassessment.
D18-E2	Hearing is stable, and middle-ear fluid is not present today.	Hearing surveillance continues, but it is not the immediate problem.
D18-E3	The dental team notes a narrowing upper arch during growth.	Orthodontic monitoring needs a separate plan.

Decision in this update: Build the routing plan and cite one evidence ID for each action.

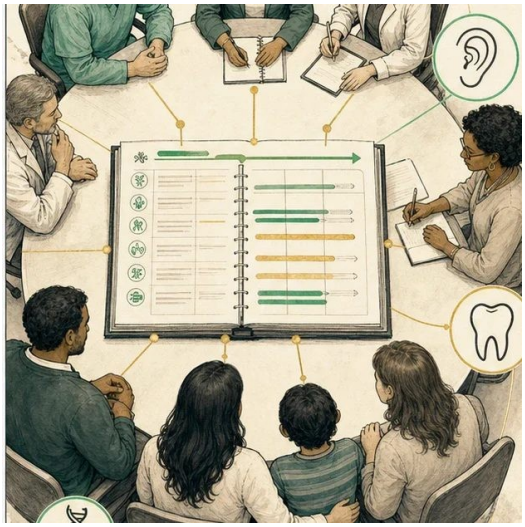
Claim ceiling: You may identify the next evaluation. You may not diagnose the cause of a new problem before the specialist assessment.

Handoff to the next update: We answered what can still go wrong and who catches it. But Mateo now needs a surgeon, a speech-language pathologist, an audiologist, an orthodontist, and more, all watching different things across many years. How does one team deliver all of this care without anything falling through the cracks?

EVIDENCE UPDATE 19 OF 20 | CLEFT TEAM COORDINATOR (TEAM DESIGN)

How a Team Delivers Care Without Gaps

A cleft team works when specialists share one plan, one record, and clear ownership across time.



Biomedical teaching illustration: Coordinate many specialists around one child

ID	Finding supplied in the case	Why it matters
D19-E1	Mateo has three follow-up actions due in different clinics within six months.	Uncoordinated scheduling creates a risk that one action will be missed.
D19-E2	The speech finding affects surgical planning, and hearing status affects speech interpretation.	The specialists' decisions depend on each other's evidence.
D19-E3	The family asks for one contact person and one written plan.	Coordination must be visible and usable to the family.

Decision in this update: Choose the coordination plan and show where each evidence item changes the plan.

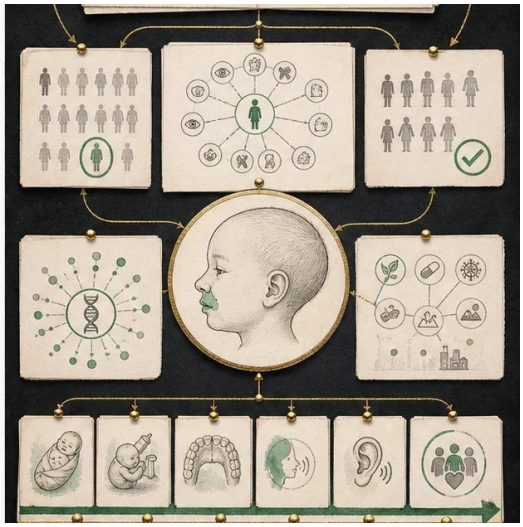
Claim ceiling: You may assign ownership and sequence. You may not make a specialist's clinical decision without that specialist and the family.

Handoff to the next update: We answered how the team holds it all together: shared records, a coordinator, and continuity across 18 years. That completes the machinery of Mateo's care. So now, with everything from his first newborn exam to his lifelong team in hand, we can finally ask the biggest question of all: what is Mateo's complete clinical story, and what, after eighteen lessons of evidence, is his actual diagnosis?

EVIDENCE UPDATE 20 OF 20 | THE WHOLE CLEFT TEAM

Mateo's Complete Clinical Story (and His Diagnosis)

Mateo's record supports an isolated, nonsyndromic cleft lip and palate with multifactorial risk and lifelong coordinated care.



Biomedical teaching illustration: Build Mateo's final clinical synthesis

ID	Finding supplied in the case	Why it matters
D20-E1	The anatomy is a complete unilateral left cleft lip and palate.	The structural diagnosis is specific and stable.
D20-E2	Repeated exam, family history, and follow-up show no consistent additional syndrome pattern.	The record supports an isolated, nonsyndromic classification at this time.
D20-E3	No single proven cause is identified, and research supports many contributing gene and environment influences.	A multifactorial risk model fits the evidence.
D20-E4	Mateo needs staged feeding, surgical, speech, hearing, dental, growth, psychosocial, and coordination follow-up.	The diagnosis includes a longitudinal care plan, not only a label.

Decision in this update: Choose the synthesis, cite at least three evidence IDs, and name one new finding that would require revision.

Claim ceiling: You may state the best current synthesis. You may not claim certainty about one causal variant or exposure.

Handoff to the next update: We answered the question we have chased since day one: Mateo has isolated, nonsyndromic cleft lip and palate, multifactorial, managed over a lifetime. But Mateo is not only the disease team's patient. The genetic, developmental, anatomical, and experimental teams have studied the same patient from their own angles. Five lenses, one Mateo, one diagnosis: bring your Disease Domain Report to the joint conference and see how your answer matches theirs.