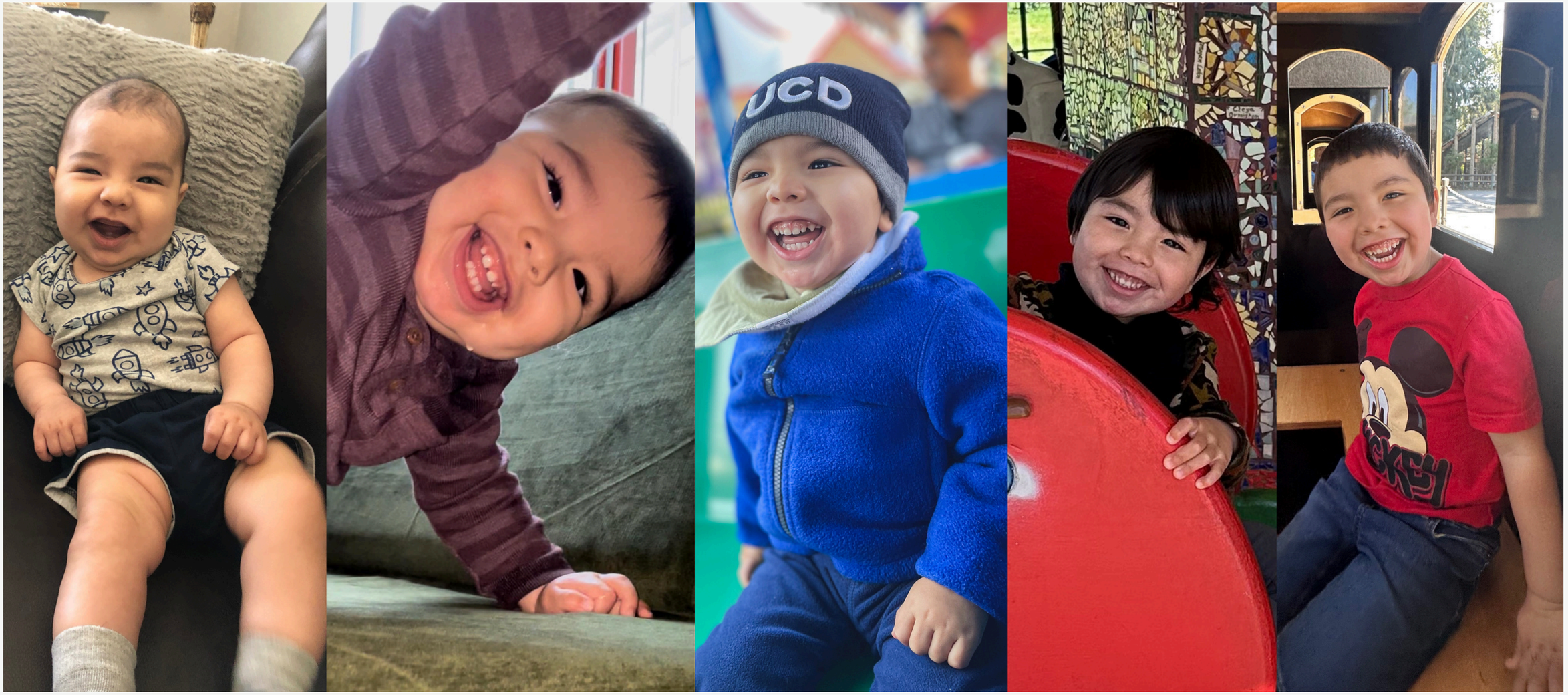
PURA Syndrome Awareness Day

October 23, 2025



Five lessons in awareness from five years of loving Benny

Lessons From Age 0-1:

Early Development & Advocacy

Benny was born at 9 lbs 11 oz after a long labor followed by induction at 41 weeks and 6 days. From the beginning, something felt wrong. He looked healthy but was very sleepy and hard to wake for feedings during his first 3 months. He lost a concerning amount of weight after birth, but his pediatrician wasn't too worried because he began slowly regaining weight.

At 6 months, he had a head tilt and still wasn't sitting up or engaging like other babies. I brought up my concerns to multiple people, but they were often brushed off with, "He may develop at his own pace. Let's watch and see." I knew something wasn't right.

At 8 months, we finally got referred to a neurologist and a geneticist, but insurance denied genetic testing due to *lack of medical necessity*. This year, **new guidelines** released by the AAP recommend exome/genome sequencing as a first-tier test for any child displaying global developmental delays, highlighting the importance of early action.

> "Looking back, the worst part was realizing I couldn't fully enjoy my happy, loving son in my pursuit for answers and validation."

Take Aways:

- Trust your instincts. *You* know your child best.
- Early genetic testing can provide answers and community.
- Listen to parents. Validation can make all the difference.

Do you know someone in need of genetic testing? **Genome Medical** | **Project FIND OUT** | **Start Genetic**

Lessons From Age 1-2:

Early Intevention & Support Systems

This was the year of baby steps. Every clap, every point, every moment Benny used his hands or showed curiosity felt *huge*. Through early intervention, I learned how to play with Benny. I learned to slow down, be intentional, follow his lead, and celebrate the small things.

It was also the year I felt the pain of comparison. Watching other children run, talk, and climb while I sat with so many unknowns was hard. I remember asking his PT if she thought Benny would ever be able to run. Looking at how well Benny is doing now makes the question sound silly, but at that time, she didn't even have a definite answer. It was hard to relate, to explain, and to be understood.

At 22 months, Benny began blossoming as he started at the UC Davis Early Childhood Lab School (ECLS). He still had poor balance and could not verbally communicate, but he had a teacher who believed in him, supported him, and did her best to understand him.

I remember making a communication table for his teacher, and she embraced it fully. By the next year, Benny was speaking enough that we didn't need it. That's what support looks like: meeting a child where they are and helping them grow. Thank you, Teacher Faith — and all staff at the ECLS.

> Support by believing in them, not doing it for them.

Take Aways:

- Early intervention changes trajectories.
- Comparison steals joy; celebrate progress instead.
- Inclusion and belief build confidence.

Lessons From Age 2-3:

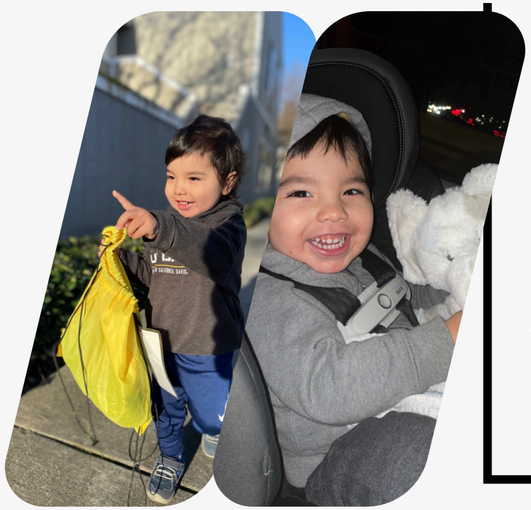
Communication Comes in Many Forms

This was the year Benny's words began to bloom slowly, but surely. Still, compared to other children, his speech delays became more obvious.

It was also the year I came to realize that Benny had always understood so much more than he could say. Receptive language — understanding what others say — is a quiet strength of many individuals diagnosed with PURA Syndrome. Their expressive language often comes later, comes differently, or may not come at all.

Many people equate verbal ability with intelligence, but Benny, along other individuals with PURA Syndrome, remind me every day that words are just one way of communicating. His eyes, his laughter, and his gestures have always been loud.

And no — we still did not have a diagnosis.



> "Just because you can't hear the words doesn't mean there's nothing being said."

Take Aways:

- Speech is not the only form of communication.
- Don't underestimate someone because they don't speak.
- Receptive language often outpaces expressive — especially in PURA Syndrome.

Lessons From Age 3-4:

Diagnosis & The Spectrum of Rare Diseases

At three and a half, after years of uncertainty, we finally received Benny's diagnosis: **PURA Syndrome**.

The genetic counselor delivered the news with tears in her eyes. I remember thinking, "Is this degenerative or life-threatening?" I was prepared for a neurodevelopmental disorder diagnosis but I didn't understand why it would make her so sad to give Benny — who was sitting beside me, smiling, engaged, full of joy (photo on the right) — this diagnosis.

When I searched online, I understood. The published information painted a very severe picture — the only picture. While Benny's symptoms were in line with PURA Syndrome, the severity of them was not. I wondered how Benny could be so rare, within an already rare population.

Over time, I have come to realize that there is a spectrum for many rare diseases that goes unseen and unpublished because they are often undiagnosed, misdiagnosed, or not a part of the population who needs urgent medical treatment.

> A diagnosis gave us a name, not a limit.

Take Aways:

- A diagnosis brings clarity, not limitation.
- Every child with PURA Syndrome is different.
- Don't shy away from disabilities — ask, learn, include.

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Lessons From Age 4-5:

Inclusion, Advocacy & Understanding Communication Differences

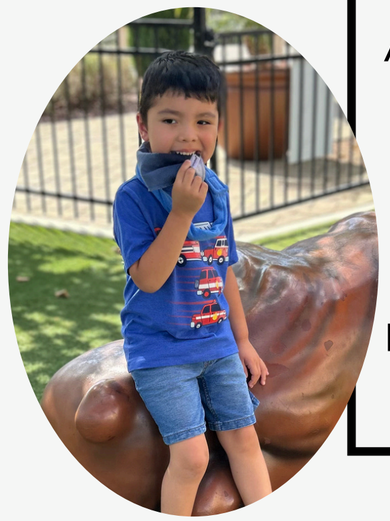
This year, Benny became more aware of his differences. He's self-conscious about his drooling and wearing chewy necklaces and doesn't like when people comment on it.

When filling out his PURA awareness cards this year, I asked him what he finds most challenging. He responded: "Play."

He loves playing and being around other kids, but when they don't understand him or he can't understand their way of playing, I can see him pull back and watch.

I know he's trying so hard to connect. And I know how much it hurts when others don't take the time to truly understand what he's saying.

Rare diseases are individually rare, but collectively common. Over **300 million** people live with a rare disease worldwide. **89.%** of childhood rare disease affect the nervous system, including PURA Syndrome.



> Awareness brings understanding but understanding requires listening.

Take Aways:

- Be patient and take time to really understand.
- Every child deserves to be included.
- Awareness can change lives.

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Benny loves:

- **“**Anything that goes” — specifically trains and tractors!
- His family
- Being silly/hiding
- Rides
- Interacting one-on-one with others

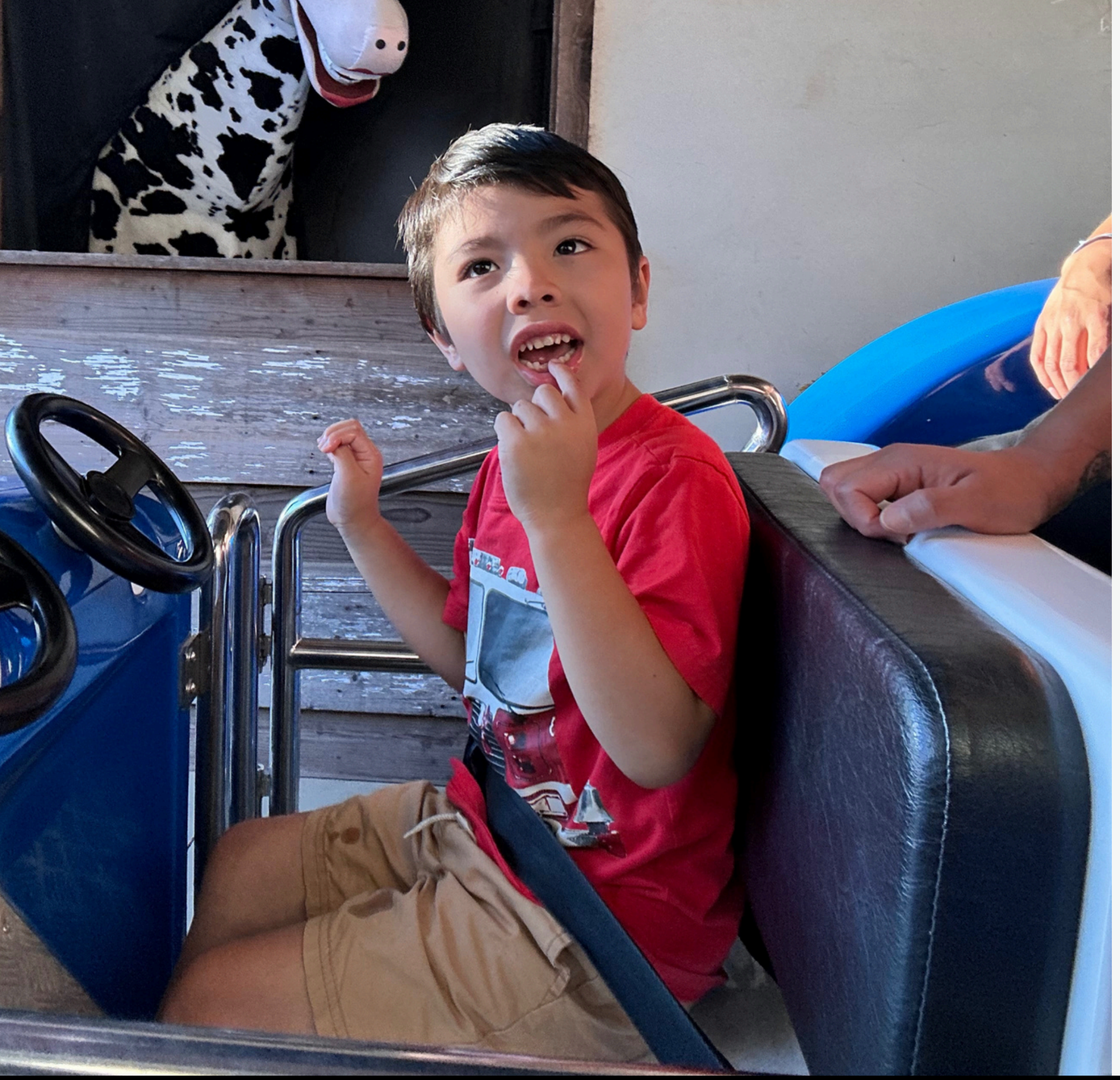
Benny is:

- Happy
- Loving
- Silly
- Funny
- Sassy
- Stubborn
- As I tell him, “The best son I could ever ask for.”

It is hard for Benny to:

- Articulate his words
- String words together to form longer sentences
- Close his mouth/stop drooling
- Play with multiple children at once
- Understand games with abstract rules
- Use fine motor skills (although this has gotten so much better recently!)
- Coordinate movements
- Regulate emotions when he’s frustrated that something is too hard for him; he does not like pressure or to be tested

I am so lucky to be his mom.



Five years ago, I hadn't planned for a life of unknowns — of therapies, tests, questions that may never be answered.

A year and a half ago, I didn’t know what PURA Syndrome was.

Today, it gives us a name and a community, but not a limit.
Through Benny, I’ve leaned patience, compassion, and purpose.

Awareness isn’t just about knowing a condition’s name.

It’s about seeing the people behind it.

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To learn and support:

PURA Syndrome FOUNDATION

To follow Benny's journey:

To read about my unique perspective on gene therapies for rare neurodevelopmental disorders:

*All icons are links