

# Family Perspectives on Navigating a CHARGE Syndrome Diagnosis

EHDI Conference 2024

Monday, March 18 | 10:35 AM - 11:00 AM | Mineral Hall D/E

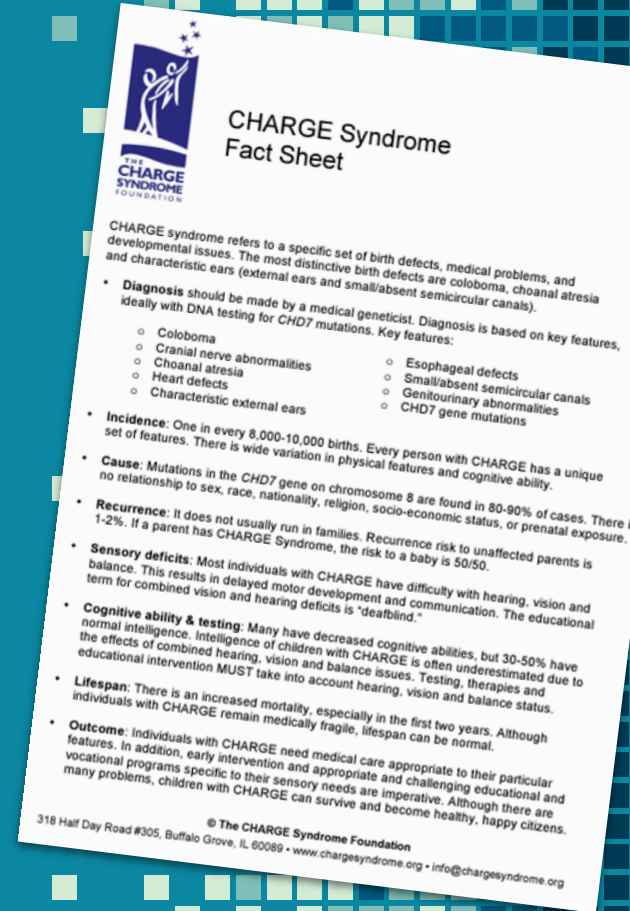
“Babies with CHARGE syndrome are often born with life-threatening birth defects. They spend many months in the hospital and undergo many surgeries and other treatments. Swallowing and breathing problems make life difficult even when they come home. Most have hearing loss, vision loss, and balance problems that delay their development and communication. Despite these seemingly insurmountable obstacles, children with CHARGE syndrome often far surpass their medical, physical, educational, and social expectations.”


*([chargesyndrome.org/about-charge/overview/](https://chargesyndrome.org/about-charge/overview/))*

# CHARGE Syndrome Overview

1. Low incidence (1 in 8-10,000 births)
2. Characteristic ear malformations  
Inner ear: monodini, semicircular canals malformed  
Middle ear: malformed ossicles  
Outer ear: distinct shape
3. Coloboma of the iris, retina, optic nerve occur in 80-90%
4. Choanal Atresia or Stenosis present in 50-60%
5. Cranial nerve abnormalities
6. Various congenital heart conditions occur in 75%
7. Feeding and swallowing in early years complicated

*([chargesyndrome.org/overview/signs-symptoms/](https://chargesyndrome.org/overview/signs-symptoms/))*



 **CHARGE Syndrome Fact Sheet**

CHARGE syndrome refers to a specific set of birth defects, medical problems, and developmental issues. The most distinctive birth defects are coloboma, choanal atresia and characteristic ears (external ears and small/absent semicircular canals).

- **Diagnosis** should be made by a medical geneticist. Diagnosis is based on key features, ideally with DNA testing for *CHD7* mutations. Key features:
  - Coloboma
  - Cranial nerve abnormalities
  - Choanal atresia
  - Heart defects
  - Characteristic external ears
  - Esophageal defects
  - Small/absent semicircular canals
  - Genitourinary abnormalities
  - *CHD7* gene mutations
- **Incidence:** One in every 8,000-10,000 births. Every person with CHARGE has a unique set of features. There is wide variation in physical features and cognitive ability.
- **Cause:** Mutations in the *CHD7* gene on chromosome 8 are found in 80-90% of cases. There is no relationship to sex, race, nationality, religion, socio-economic status, or prenatal exposure.
- **Recurrence:** It does not usually run in families. Recurrence risk to unaffected parents is 1-2%. If a parent has CHARGE Syndrome, the risk to a baby is 50/50.
- **Sensory deficits:** Most individuals with CHARGE have difficulty with hearing, vision and balance. This results in delayed motor development and communication. The educational term for combined vision and hearing deficits is "deafblind."
- **Cognitive ability & testing:** Many have decreased cognitive abilities, but 30-50% have normal intelligence. Intelligence of children with CHARGE is often underestimated due to the effects of combined hearing, vision and balance issues. Testing, therapies and educational intervention **MUST** take into account hearing, vision and balance status.
- **Lifespan:** There is an increased mortality, especially in the first two years. Although individuals with CHARGE remain medically fragile, lifespan can be normal.
- **Outcome:** Individuals with CHARGE need medical care appropriate to their particular features. In addition, early intervention and appropriate and challenging educational and vocational programs specific to their sensory needs are imperative. Although there are many problems, children with CHARGE can survive and become healthy, happy citizens.

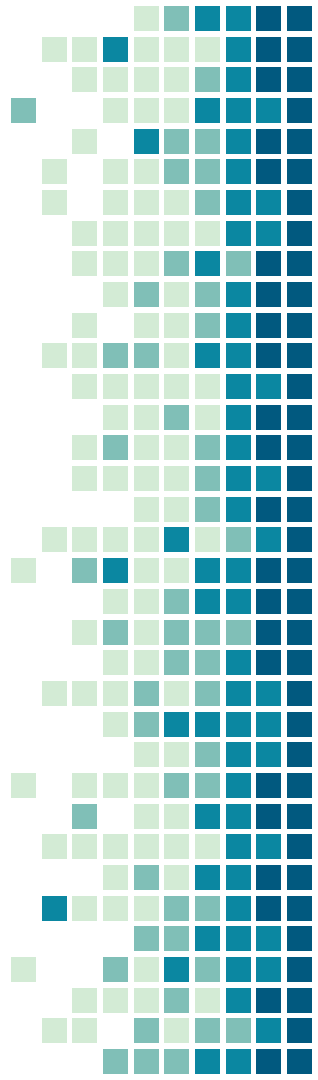
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318 Half Day Road #305, Buffalo Grove, IL 60089 • [www.chargesyndrome.org](http://www.chargesyndrome.org) • [info@chargesyndrome.org](mailto:info@chargesyndrome.org)

# And yet we THRIVE!



# Iris Filkins

Colorado State Liaison  
CHARGE Syndrome  
Foundation



# Meet Remi



- A soon to be 3 year old!
- Bilateral cochlear implants
- Loves Elmo & Sesame Street
- Learning ASL
- 50+ expressive signs
- Attends a toddler program at the Anchor Center for Blind Children
- Planning to attend preschool in the fall

# A Warrior from Day One

- Born at 34 weeks
- 7 week NICU stay resulting in three surgeries
- . . . and what felt like diagnosis after diagnosis



# CHARGE Diagnosis

- Colobomas
- Microphthalmia
- Choanal atresia
- Ear abnormalities
- Cranial nerve differences
- Malrotation
- Silent aspiration
- Facial palsy







# Hearing Loss Diagnosis

- Transfer to Level 4 NICU
- 2 rounds of CT & MRI imaging
- 3 Sleeping ABRs
- 1 Sedated ABR during ear tube placement at 6 months old
- Trialing hearing aids for 9+ months
- Second opinion for cochlear implants
- Bilateral cochlear implants
  - Right side at 13 months old
  - Left side at 21 months old

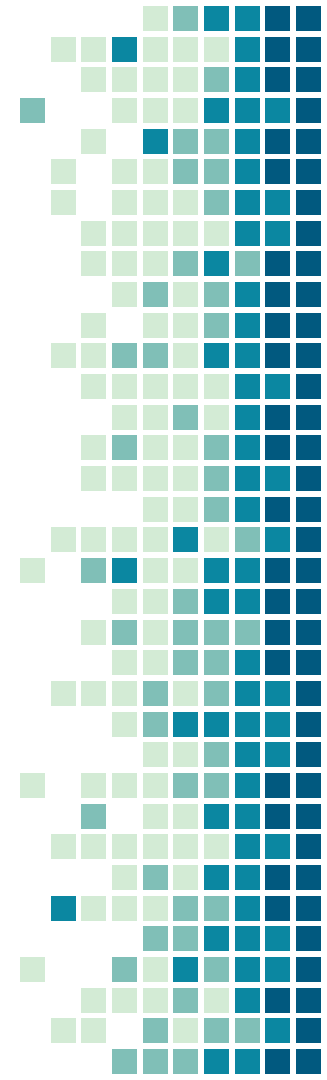
# Total Communication Approach

- Sign Language
  - ASL lessons through Early Intervention
  - Community intervener for Deaf-Blind children
- Spoken Language
  - Lots of narrating!
- Touch Cues
- Starting to introduce an AAC device



If I could go back,  
what is one thing I would tell myself?

Find other medical parents - it will be crucial to  
navigating this journey.



# Staci Petralba

Audiologist  
Mother of 2 year old with  
CHARGE Syndrome



# Meet Benjamin

- Will turn 3 in May
- Hearing aid in the right ear, cochlear implant left ear
- Total communication, mostly vocalizations
- 20-30 expressive words and signs
- Loves reading books, bubbles, pandas, rocking and swinging



# Benjamin's Grand Entrance

- Normal pregnancy, born at 38 weeks
- 8 weeks in
  - 3 hospitals
  - 2 states
  - 3 ambulance trips
  - 2 flights
- 4 surgeries before coming home

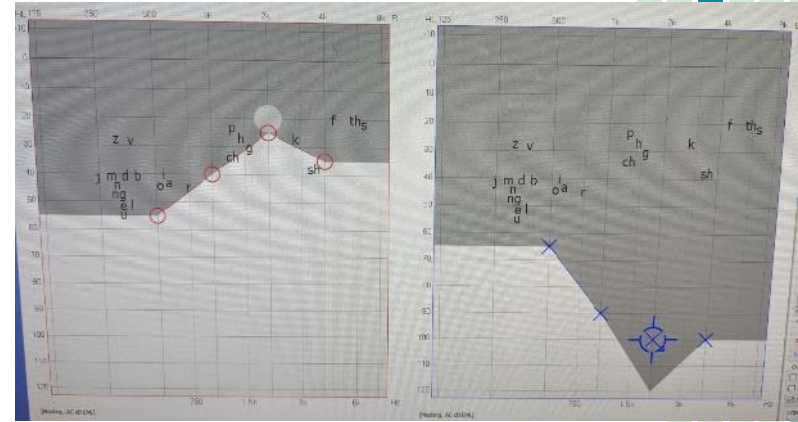


# CHARGE Diagnosis

- CHARGE was immediately suspected
- Has all of the major diagnostic criteria
- Choanal atresia + Colobomas
- Genetics confirmed diagnosis at 1.5 months old



# Hearing Loss Diagnosis

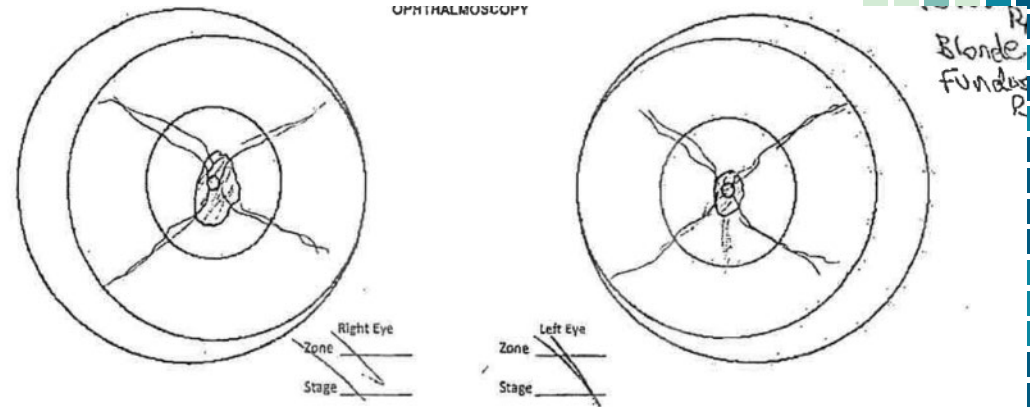


- Right ear: Mild to moderate hearing loss
- Left ear: Severe to profound hearing loss



# Choosing Communication Approach

- Total communication
- Unsure of functional vision status
- Poor support from early intervention with ASL
- Vocal cord paralyzed



# Hearing Technology Journey



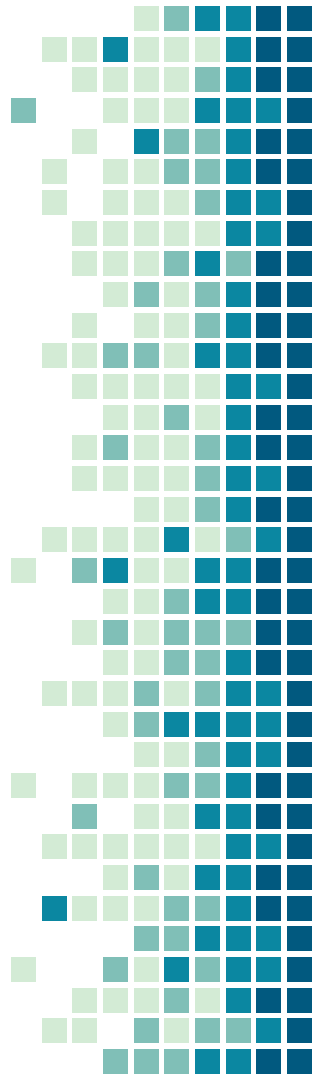
Hearing aids  
at 4 months  
old

Cochlear  
implant in the  
left ear at 2.5  
years old



If I could go back,  
what is one thing I would tell myself?

Don't second guess yourself, trust your instinct



# Barriers families face in meeting 1-3-6:

1. Length of hospital stays and family separation
2. Travel for specialized medical care and procedures
3. Coordinating care between multiple systems
4. Difficult to obtain quality audiological diagnostics and confirm hearing levels, need to test multiple times
5. Navigating insurance coverage and/or medicaid waivers requires additional time to get treatment
6. Complicated ear anatomy takes time to understand and create a plan of action



## CHARGE Syndrome Foundation Resources:

- 1. Full-time Outreach and Engagement Manager**  
Lourdes Quintana Baez ([lourdes@chargesyndrome.org](mailto:lourdes@chargesyndrome.org))
- 2. Medical Travel Assistance**  
Reimbursement for expenses not covered by insurance for travel over 200 miles
- 3. EWRAP: Ethan Wolfe Recreational Assistance Program**  
Adaptive recreation equipment & program costs not covered by insurance
- 4. Mountains of information for professionals!**  
([chargesyndrome.org/for-professionals/](https://chargesyndrome.org/for-professionals/))  
--\$50K Scientific Research Grant Program  
--\$25K Clinical Research Grant Program
- 5. Attend our biennial conference in 2025!**  
“The Grand Adventure,” Phoenix, AZ , July 24–27, 2025.

# Family-to-Family Support Matters

1. **Find your state's CSF liaison!**  
Each keeps a list of state specific resources  
Organizes an annual meet-up for our Walk & Roll
2. **Join the CSF Birth to Age Three P2P Support Group!**  
3rd Saturday Monthly, 7:00p ET
3. **Connect with State Deafblind Project!**  
[www.nationaldb.org/state-deaf-blind-projects/](http://www.nationaldb.org/state-deaf-blind-projects/)  
Family Engagement Coordinators work closely with CSF
4. **Attend our 2025 Conference!**  
Only place we feel 'at home'  
Generous scholarships available



CHARGE SYNDROME FOUNDATION  
**Birth to Age 3 Parent-to-Parent Support Group**  
January 20, 2024  
NEW TIME!  
4:00 PM PT, 5:00 MT, 6:00 CT, 7:00 ET



Scan the code to register



**THE GRAND ADVENTURE**  
RISING FROM CHALLENGES TOGETHER  
16th International CHARGE Syndrome Conference  
Phoenix, Arizona • July 24-27, 2025

JOIN US at the

2024

# Walk & Roll for CHARGE

Saturday, May 18, 2024

- Free virtual registration
- Liaisons are hosting fun meetups all over the country
- Raising awareness and funds
- More info at [www.chargesyndrome.org/](http://www.chargesyndrome.org/)

## Walk and Roll for CHARGE

5th Annual Walk and Roll for CHARGE – May 18, 2024



 Sierra Strong \$1,000 of \$1,000 Reached <a href="#">View</a>	 CHARGE on Tucker \$750 of \$1,000 Reached <a href="#">View</a>	 Zach Attack the Whale \$700 of \$1,000 Reached <a href="#">View</a>	 Victoria Strong \$660 of \$1,000 Reached <a href="#">View</a>
 Carm Farm \$650 of \$1,000 Reached <a href="#">View</a>	 Massachusetts and New Hampshire Families CHARGE On Together \$618 of \$1,000 Reached <a href="#">View</a>	 Parkins School for the Blind - Staff and Families \$550 of \$1,000 Reached <a href="#">View</a>	 NEVADA Friends and Family \$500 of \$1,000 Reached <a href="#">View</a>
 Team Christian Hall \$475 of \$1,000 Reached <a href="#">View</a>	 CHARGE FLORIDA \$320 of \$1,000 Reached <a href="#">View</a>	 CHARGE On Colorado \$265 of \$1,000 Reached <a href="#">View</a>	 NY CHARGE \$250 of \$1,000 Reached <a href="#">View</a>
 Iowa CHARGES Onward \$127 of \$1,000 Reached <a href="#">View</a>	 CHARGE on for James \$125 of \$1,000 Reached <a href="#">View</a>	 Carter's Crew \$123 of \$1,000 Reached <a href="#">View</a>	 Kansas CHARGE's \$100 of \$1,000 Reached <a href="#">View</a>
 Team MtCushes \$76 of \$1,000 Reached <a href="#">View</a>	 CHARGE Washington \$50 of \$1,000 Reached <a href="#">View</a>	 CHARGE Illinois \$50 of \$1,000 Reached <a href="#">View</a>	 CHARGE Oregon \$50 of \$1,000 Reached <a href="#">View</a>

Together, be active, and support our CHARGE syndrome and all funds raised will help

NOW

ARGE! All across the country the CHARGE community centers, community centers, and shopping malls to the Foundation. Whether it's a large state-wide event, the goal remains the same: to be active while raising awareness.

# Stay Connected

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