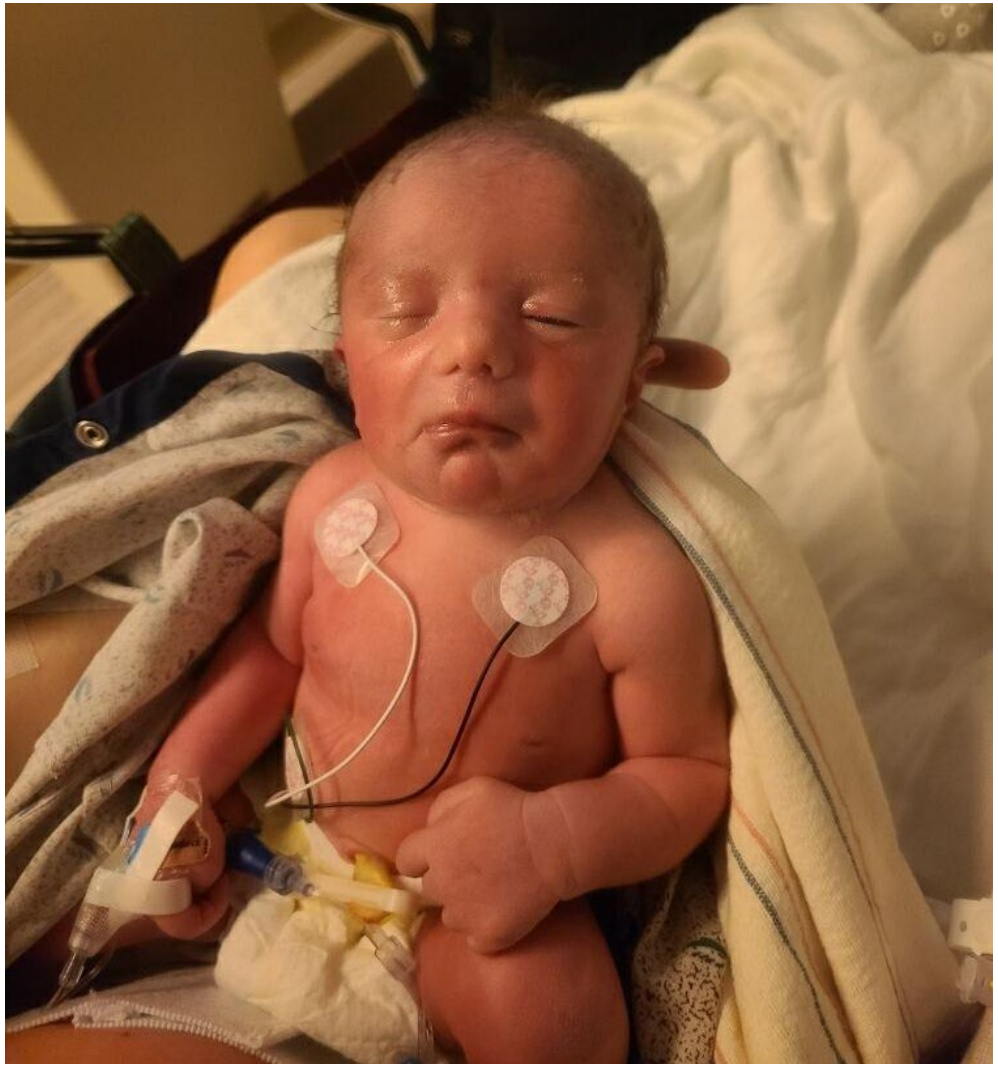
We've Come a Long Way: Amalia's CHARGE syndrome journey

Born December 2020



Initial Evaluation

- Facial asymmetry- MRI brain to rule out stroke
- Feeding difficulties and aspiration
  - Need for respiratory support again
  - Placed a nasal feeding tube for nutrition
- Low muscle tone
- Heart murmur
- Fluid in the kidneys
- Early genetic testing

First Christmas

When everything changes...

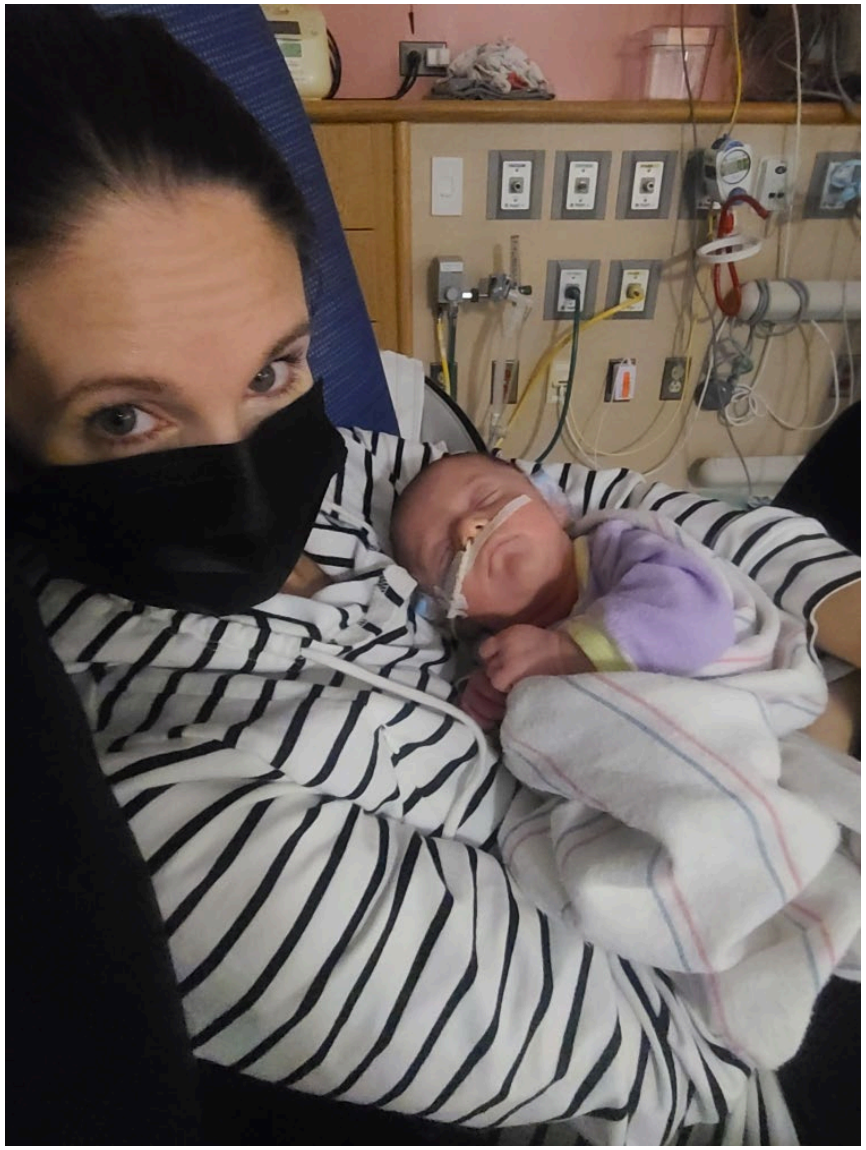
Transfer to Children's Hospital of Philadelphia (CHOP)

CHARGE syndrome

C: coloboma or cranial nerve
H: heart defects
A: atresia of choanae
R: retardation of growth or development
G: genital/urinary abnormalities
E: ear malformation and/or hearing loss

Genetics: CHD-7 gene most common gene affected
Most frequently, de novo mutation

Hospital Days



And more hospital days…

5 weeks old
Her first surgery

Finally going home after 42 days

“Sleeping” ABR

Pennsylvania Early Intervention Program

- PT
- Special Instruction-Hearing
- SLP for feeding
- ASL course at local community college
- Eventually, added OT and special instruction for vision

Family Connections for Language and Learning Pennsylvania State

- Anne Gaspich, Program Director
- Parent Mentor
- Deaf Mentor
- Facebook group with resources, education and events
- Virtual classes
- Reading materials and articles
- ASL courses and references
- Introduced to American Society for Deaf Children

Life with Amalia

CT scan of her middle ear

- Absent left oval window and underdeveloped stapes
- No semicircular canals bilaterally
- CT findings consistent with CHARGE syndrome
  - Still no genetic diagnosis
  - Genetics were not willing to give her the diagnosis with lack of genetic confirmation
- Given lack of answers, reached out to genetics at Cincinnati Children's Hospital CHARGE center

Bone conduction hearing aid

Diagnosis

- Epigenetic testing confirmed CHARGE syndrome diagnosis at 13 months old
- Evaluation in Cincinnati Children's CHARGE center at 18 months old

Oral Eating

Choanal Atresia Surgery

Daycare

Nico Arrives

Attended Pennsylvania Great Start Conference July 2023

- Network with other families
  - Amalia met other toddlers with hearing loss and wearing technology
- Parent Cafe
- Meet with many parent mentors from all over the state
  - Learn about ways to advocate for Amalia
- Learned of more online resources
  - Parent to Parent of PA
- Met our deaf mentor in-person
- Paid for which makes it accessible to all families despite financial resources

Recently…

- Getting ready to transition to pre-school in fall
- Learning and using more ASL and spoken language
- Just entered the 3-5 year old school-based EI program
- Enrolled in tumbling class
- Drinking thin liquids by mouth

Challenges for families of deaf plus children…

- Parents to medically complex children are juggling multiple diagnoses, consultants and follow ups
- Hearing loss is sometimes overshadowed by other medical problems
- Follow up may be delayed when parents have many appointments to attend and coordinate
- Lack of access to specialized, multidisciplinary care
- Many times, a hearing loss diagnosis is made later in kids with multiple other medical problems
- Language or learning struggles can be attributed to other conditions rather than untreated hearing loss
- Harder to gain the information from ABR when child is more wakeful, or results are considered unreliable
- Families do not have health literacy
- Families aren’t made aware of state programs or opportunities

How to support families…

- Educate families
  - Testing expectations
  - Importance of early detection and intervention
  - Importance of early access language
  - Communication options
  - Device options
- Coordinate with other services
  - Clumping appointments together
- Refer parents to support groups to gain experience and advice from other families
- Make sure the child is receiving services from early intention
- Be flexible- approach each child with his or her uniqueness in mind
- Provide resources for both medical and educational communities

Questions?