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Grand Rounds: Pediatric Audiology - Exploring the Uniqueness of Every Child to Achieve Best Outcomes,

in partnership with Phoenix Children's Hospital

Lynn Eyde, AuD
Maddie McNamee, AuD
Allie Sayer, AuD
Alissa Nickerson, AuD
Ashley Geske, AuD
Deborah Flynn, AuD

Wendy Steuerwald, AuD

Allie Sayer, AuD

Allie Sayer, AuD, CCC-A graduated from Arizona State University with a Bachelor of Science degree in Speech and Hearing Sciences; she also graduated from Barrett, the Honors College at ASU after completing an honors thesis studying vocabulary differences in children with mild to moderate hearing loss in comparison to their normal hearing peers. She continued at Arizona State University to obtain her Doctorate in Audiology degree, after completing her hospital-based externship at St. Louis Children's Hospital. Dr. Sayer worked in the private sector with both children and adults before coming to Phoenix Children's Hospital. She is a member of and certified through the American Speech, Language, and Hearing Association. Her clinical areas of specialty include diagnostics, hearing aids, and cochlear implants.

Alissa Nickerson, AuD

Alissa Nickerson, AuD, CCC-A, is a clinical audiologist at Phoenix Children's specializing in the diagnosis and management of childhood hearing loss. Alissa enjoys working with children of all ages and has a special interest in pediatric amplification. She is a collaborating member of "Stop CMV AZ," an organization which works to educate and advocate for those impacted by CMV in the state of Arizona. Prior to joining the staff at Phoenix Children's, Alissa was employed in private practice in Chicago, Illinois. She has publications in the areas of post-baccalaureate audiology training and amplification profiles of pediatric cochlear implant users.

Ashley Geske, AuD, CCC-A

Ashley Geske, AuD, CCC-A graduated from Arizona State University with a Bachelor of Science in Speech and Hearing Sciences and a clinical Doctorate of Audiology. Prior to joining Phoenix Children's Hospital in August of 2022, she worked at Providence Speech and Hearing Center partnered with the Children's Hospital of Orange County where she worked with pediatric patients in both outpatient and inpatient hospital settings. Her clinical areas of specialty include diagnostics, hearing aids, and cochlear implants.

Deborah Flynn, AuD

Deborah Flynn is the Audiology Lead for Cochlear Implants at Phoenix Children's Hospital. She has extensive experience in identification, intervention, and management of infants, children, and teens with hearing loss. Her involvement in the Audiology Team includes cochlear implants, hearing aids, pediatric audiological evaluations, ototoxic monitoring, evoked potentials, assistive listening technology, and newborn hearing screenings. Her clinic interests include a multidisciplinary team approach for children with cochlear implants and hearing aids. She presents on various audiological topics throughout Arizona. She has been an adjunct professor at AT Still University, Arizona School of Health Sciences and enjoys teaching students.

Lynn Eyde, AuD, CCC-A

LynnMarie Eyde, AuD, CCC-A is the Manager of Audiology Services at Phoenix Children's Hospital (PCH). She graduated from the University of Arizona in 2014 with her Doctorate in Audiology. In 2016, Lynn started working as a clinical audiologist at Phoenix Children's and within that time she has worked closely to establish and maintain the PCH Newborn Hearing Screening program.

Maddie McNamee, AuD

Madeline McNamee is a clinical audiologist at Phoenix Children's Hospital specializing in the diagnosis and management of childhood hearing loss. Madeline has a special interest in electrophysiological assessment. She loves that every day in a pediatric setting is unique!

Wendy Steuerwald, AuD

Wendy Steuerwald is the Director of Audiology at Phoenix Children's. She works with audiologists to create protocols and an environment where exceptional patient care is provided.

Disclosures

- **Presenter Disclosure**: Financial: Allie Sayer is employed by Phoenix Children's Hospital. Non-financial: Allie Sayer has no relevant non-financial relationships to disclose. Financial: Alissa Nickerson is employed by Phoenix Children's Hospital. Non-financial: Alissa Nickerson has no relevant non-financial relationships to disclose. Financial: Ashley Geske is employed by Phoenix Children's Hospital. Non-financial: Ashley Geske has no relevant non-financial relationships to disclose. Financial: Deborah Flynn is employed by Phoenix Children's Hospital. Non-financial: Deborah Flynn has no relevant non-financial relationships to disclose. Financial: Lynn Eyde is employed by Phoenix Children's Hospital. Non-financial: Lynn Eyde has no relevant non-financial relationships to disclose. Financial: Maddie McNamee is employed by Phoenix Children's Hospital. Non-financial: Maddie McNamee has no relevant non-financial relationships to disclose. Financial: Wendy Steuerwald is employed by Phoenix Children's Hospital. Non-financial: Wendy Steuerwald has no relevant non-financial relationships to disclose. In lieu of accepting an honorarium, a donation has been made to Phoenix Children's Hospital.
- **Content Disclosure**: This learning event does not focus exclusively on any specific product or service.
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Learning Outcomes

After this course, participants will be able to:

- Describe how a multidisciplinary approach can help facilitate best patient care.
- Discuss and analyze several complex audiologic cases.
- Describe how complicating factors can come together to support the patient experience.

Arizona: The Grand Canyon State!

What makes Phoenix Children's (PCH) unique?

- "PCH is the sixth largest children's hospital in the fifth largest city in the country, with the fourth largest pediatric population" Mandy Marcinko https://www.mysunwest.com/blog/this-is-phoenix-childrens-hospital
- Part of a statewide pediatric health system
- 2 additional hospitals under construction
- Wonderfully collaborative care team!
- PCH ENT is the EHDI Chapter Champion
- State of Arizona new born screening contract
- PCH provides Newborn Hearing Screening services at 4 local hospitals
- BAER (brainstem auditory evoked response) = ABR
- Generous, clinically inquisitive Audiologists

Introducing Sophia

A case of failed newborn hearing screening

Lynn Eyde, Au.D., CCC-A

Newborn Hearing Screening (NBHS) Review

- NBHS is not a test of normal hearing sensitivity
  - The test does not capture the frequency range of hearing or rule out slight or mild hearing losses
  - A pass does not indicate normal hearing sensitivity
- A pass is when both ears pass during the same testing session
- A fail is when one or both ears did not pass during the same testing session
- Stop and start is when a test is stopped before the test has completed. This is considered an incomplete (INC) test.

Sophia's Case History

- Failed inpatient NBHS at an outside facility
- Failed outpatient hearing NBHS at PCH Audiology
- Uncomplicated pregnancy and birth history
  - Mother diagnosed with COVID-19 during 2nd trimester of pregnancy
- No family history of hearing loss in childhood
- Father is in Mexico and has not been able to obtain immigration status
- Home language is Spanish (mom does speak English and Spanish)

Sophia's Journey

- Inpatient newborn hearing screening (NBHS) results:

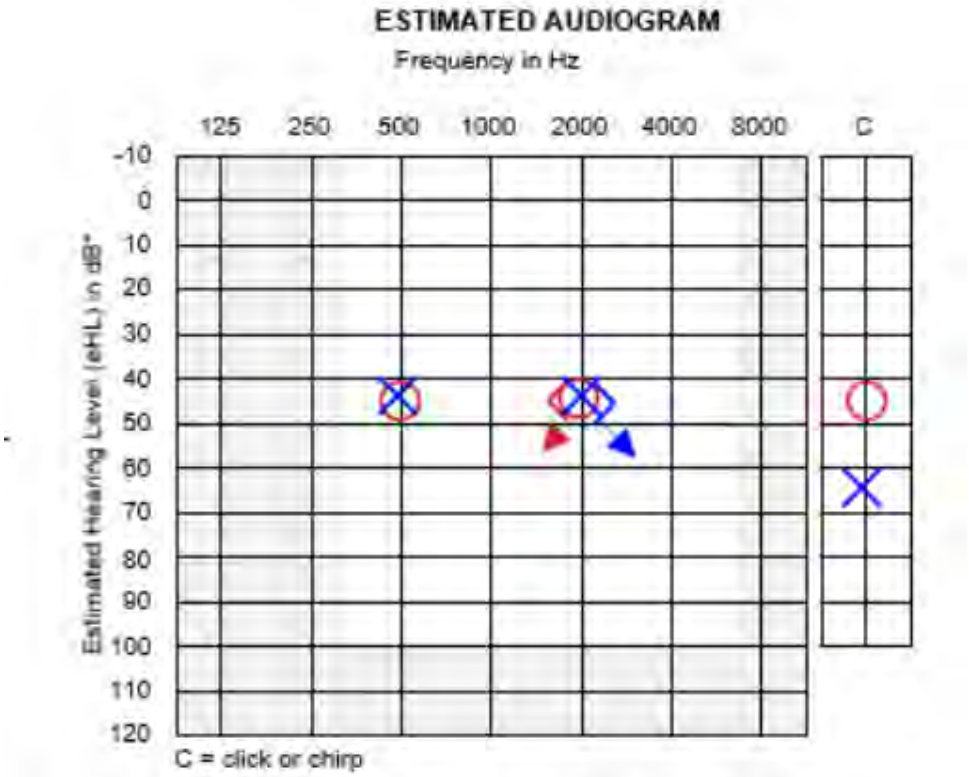
Date	Left Ear	Right Ear
01/01/2022	11 INC tests; 0 tests completed.	Did not test
01/02/2022	6 INC tests, then a passing result	4 INC tests, then a failed results

  - PCP referred to PCH for an outpatient newborn hearing screening
- Outpatient NBHS results:

Date	Left Ear	Right Ear
01/17/2022	Failed	Failed

  - Diagnostic brainstem auditory evoked response (BAER) study appointment scheduled prior to leaving outpatient appointment

01/20/2022: BAER Study #1

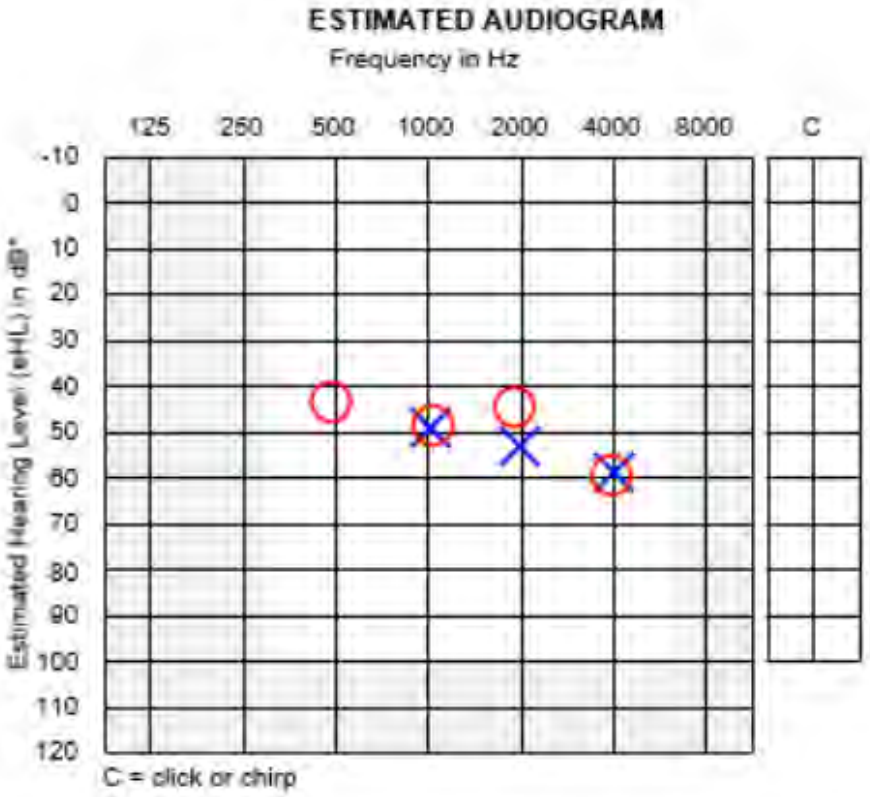


- 1000 Hz Tympanometry:
  - Peaked tympanogram, bilaterally.
- Distortion-product otoacoustic emissions:
  - Absent, bilaterally
- BAER study results:
  - Moderate sensorineural hearing loss, bilaterally

Counseling

- The patient having a “pass” in one ear on the newborn hearing screening confused the mother
- I explained that the newborn hearing screening does not indicate normal hearing sensitivity.
- The passing result in this case is related to over-screening of the patient
- The mother acknowledged the information and started to ask about next steps
- The mother also told me that the father was in Mexico and had not been granted immigration status
- I offered to include the father in the discussion and relay the results through an interpreter. The mother declined at that time.

02/08/2022: BAER Study #2



- BAER study results:
  - Moderate sensorineural hearing loss, bilaterally
- Patient was scheduled to come back on 02/16/2022 for a hearing aid consult with the managing audiologist

Otolaryngology and Audiology

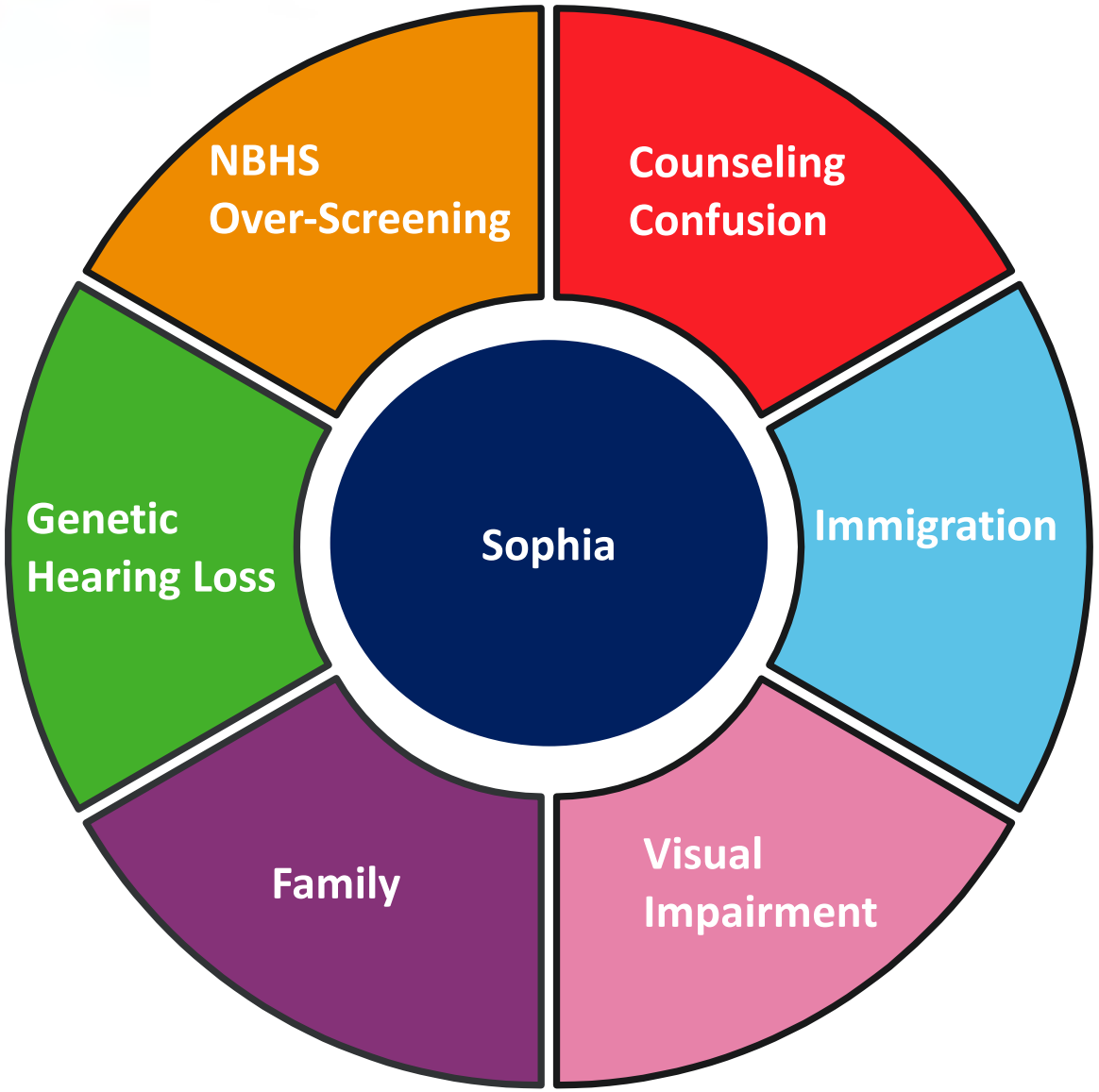
- 01/25/2022: Established care with Otolaryngology
  - Ordered MRI, bloodwork, Genetics consult, and Ophthalmology consult
- 02/16/2022: Patient was seen for a hearing aid consult
- 03/03/2022: MRI completed; normal results
- 03/11/2022: Patient was fit with binaural behind-the-ear hearing aids and custom earmolds

Ophthalmology and Genetics

- 03/2022: Ophthalmology consult indicated no ocular complications. Recommended follow-up in one year
- 07/2022: Consult with Genetics
  - Hearing loss gene panel was ordered
- 10/2022: Genetic cause confirmed as USH2A gene (autosomal recessive): Usher syndrome type 2A
  - Associated with moderate to severe congenital hearing loss, progressive retinal degeneration
- Following Genetics visit; Sophia and her mother consulted with a retinal specialist.
  - Exam was reportedly normal at that time.

Audiology Progress

- Sophia has had inconsistent hearing aid usage
  - Datalogging has been consistently 2-3 hours each day
  - Patient is watched by her grandmother while the mother works
  - Her grandmother does not feel comfortable with putting in the hearing aids
- Sophia does receive services through Early Intervention and the Arizona School for the Deaf and Blind (ASDB)
- Mother had requested a letter be written to try to get immigration granted for the patient's father
  - A letter was written by a few different providers
  - He has yet to be granted this and this creates difficulty for Sophia and her mother


NBHS
Over-Screening
Counseling
Confusion
Genetic
Hearing Loss
Sophia
Immigration
Family
Visual
Impairment

Introducing Walter

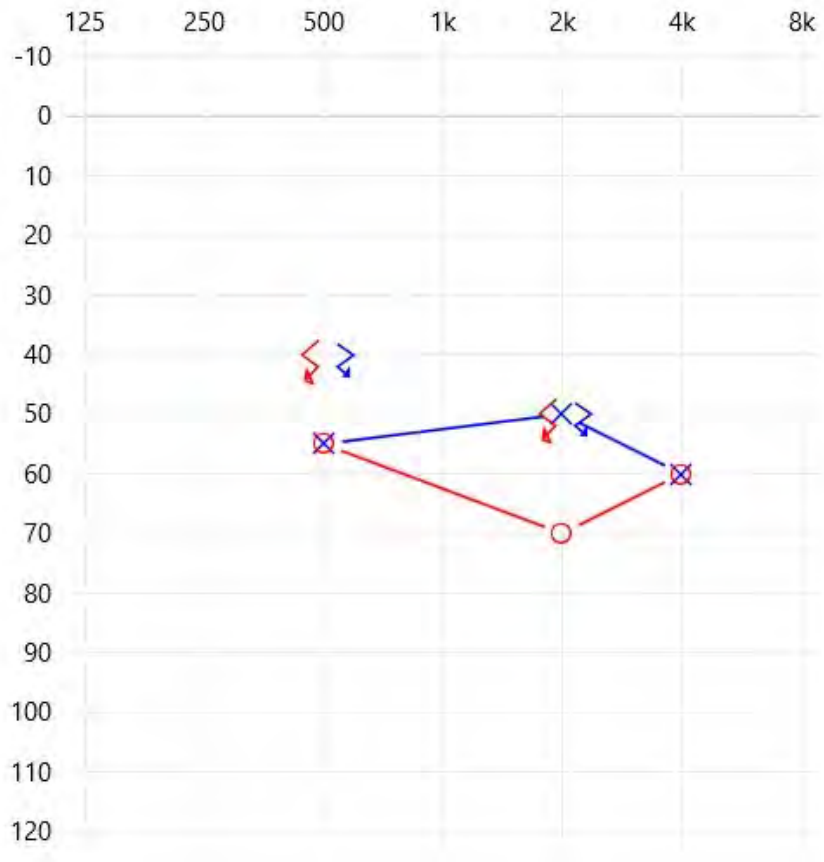
A case of unexpected test results

Maddie McNamee, Au.D., CCC-A

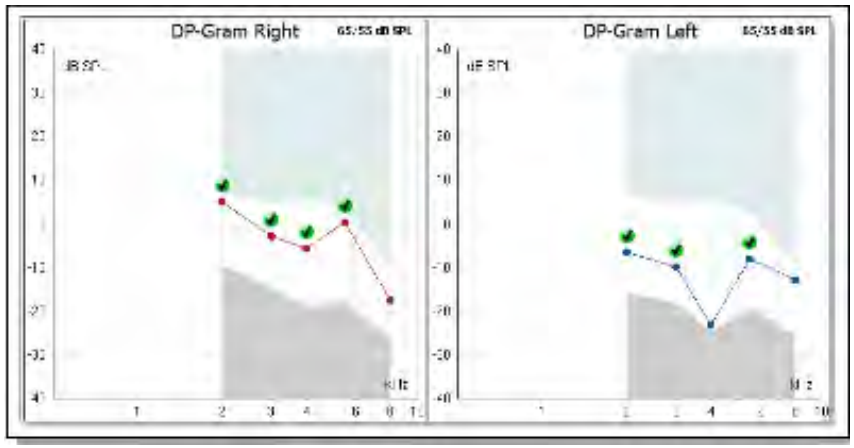
Walter's Journey

- 3/10/2021: Seen at 12-days-old by our NBHS program following failed inpatient screening.
  - Referred on A-ABR, bilaterally.
- Then referred for diagnostic testing

BAER Testing from 3/17/2021

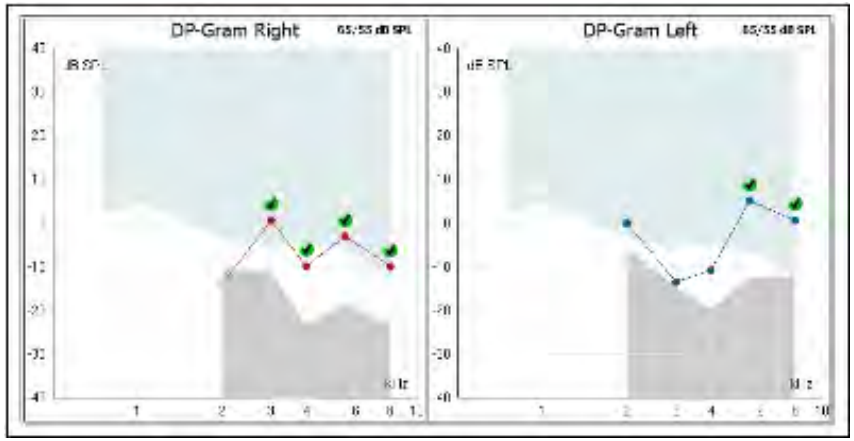


- Tympanometry with 1,000 Hz probe-tone revealed a flat tympanogram, bilaterally.
- High level click ruled out ANSD



BAER Testing from 4/22/2021

- Tympanometry with 1,000 Hz probe-tone revealed a peaked right tympanogram and a flat left tympanogram.



Walter's Journey Continued

- 5/28/2021: Fit with binaural hearing aids.
- Received services from AzIEP (early intervention) and Hands & Voices.
- Became an excellent hearing aid user.
  - Datalogging routinely 8+ hours even during infancy.

Medical Work-Up

- 4/8/2021: Established care with PCH ENT
  - At that time, it was reported that:
    - Mom tested positive for COVID-19 when she was 7 weeks pregnant.
    - CMV testing was obtained and was negative.
- 4/19/2021: Established care with PCH Ophthalmology
  - Hypermetropia noted. Otherwise, unremarkable. F/U in a year.
- 6/16/2021: Established care with PCH Genetics & Metabolism
  - A hearing loss gene panel was ordered

Genetic Results

- 9/22/2021: Genetics reviewed results with family
  - Hearing loss panel revealed variants of unknown significance in GIPC3 and OTOGL.
    - Each of these genes is associated with autosomal recessive/non-syndromic hearing loss disorders
- A renal ultrasound was ordered.
  - Completed on 10/18/2021 and indicated normal kidney structures.

GIPC3

- GIPC3 mutation = mechanical conduction disorders in the inner ear.
- GIPC3 causes SNHL and acoustic seizures.
- Researchers found stereocilia bundle deficits of OHC and IHC.
  - The hair bundle of IHC appeared less rigid and sparse.

Charizopoulou, N., Lelli, A., Schraders, M., Ray, K., Hildebrand, M. S., Ramesh, A., Srisailapathy, C. R., Oostrik, J., Admiraal, R. J., Neely, H. R., Latoche, J. R., Smith, R. J., Northup, J. K., Kremer, H., Holt, J. R., & Noben-Trauth, K. (2011). GIPC3 mutations associated with audiogenic seizures and sensorineural hearing loss in mouse and human. *Nature Communications*, *2*(1). https://doi.org/10.1038/ncomms1200

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OTOGL

- Researchers found those with this mutation presented with mild/moderate sensorineural hearing loss.

Pan, C., Li, J., Wang, S., Shi, C., Zhang, Y., & Yu, Y. (2022). Novel heterozygous mutations in the Otogelin-like (OTOGL) gene in a child with bilateral mild nonsyndromic sensorineural hearing loss. *Gene*, *808*, 146000. https://doi.org/10.1016/j.gene.2021.146000

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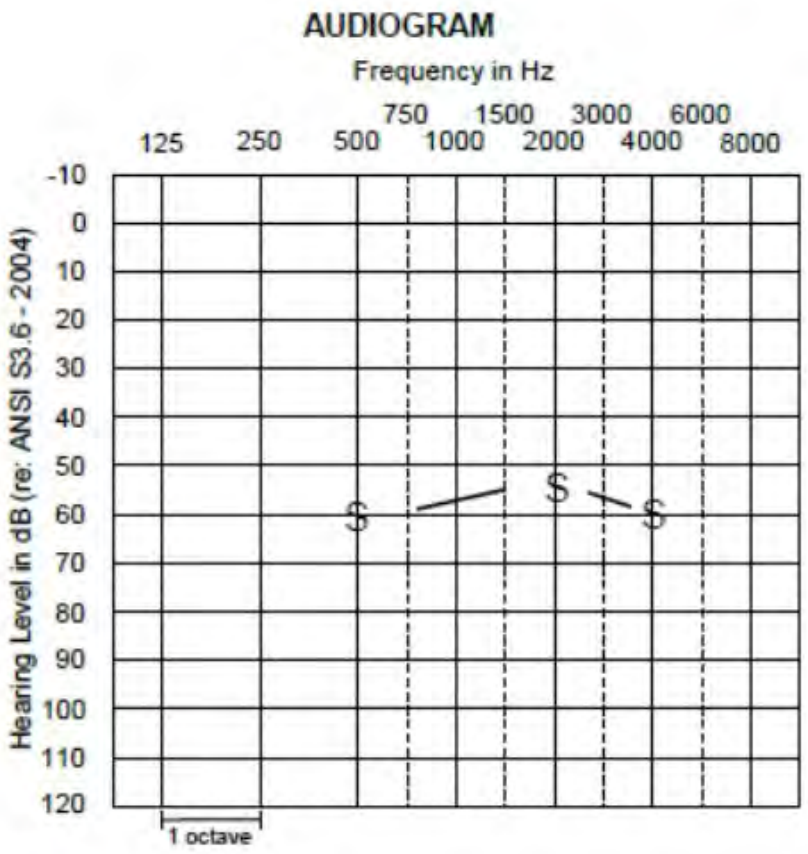
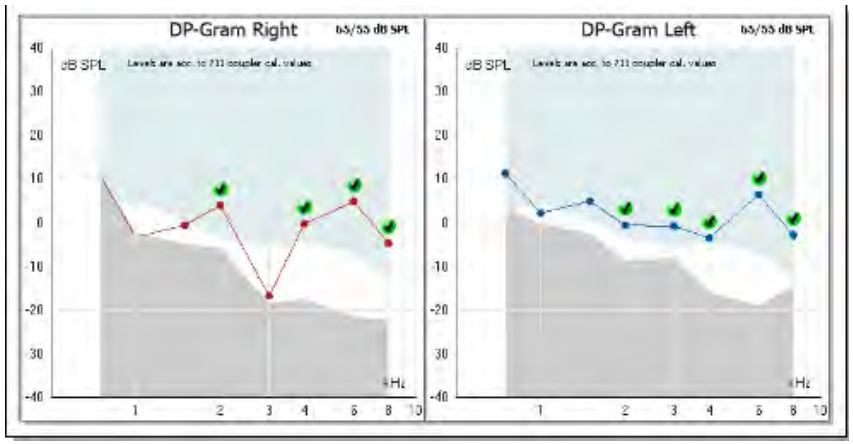
Otologic Follow-Up

- Throughout this time, Walter continued to have middle ear dysfunction.
- He underwent PE tube placement on 2/15/2022
  - A repeat BAER study was completed following PE tube placement.
    - Severe rising to moderately-severe SNHL.
    - Decrease in thresholds
    - Recommended we monitor closely and attempt behavioral testing at our next follow-up.

Testing from 3/17/2022

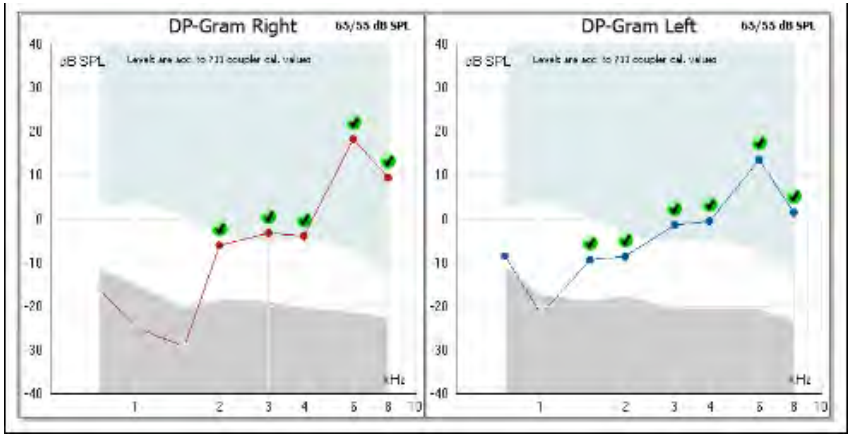


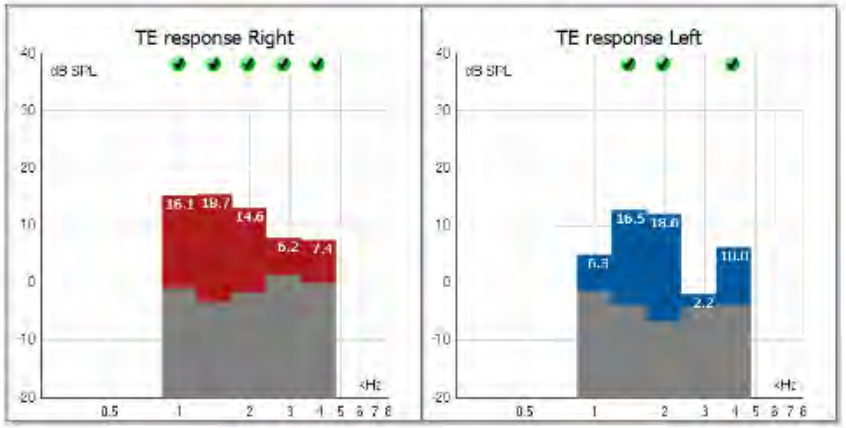
- Tympanometry = large ear canal volumes, consistent with patent PE tubes.

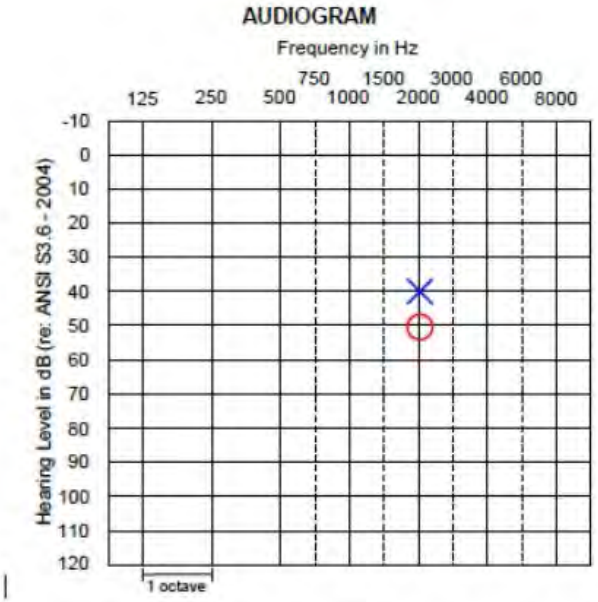


Testing from 3/17/2023

- Tympanometry = large ear canal volumes, consistent with patent PE tubes.
- Single tester CPA paradigm used
- Supra-Aural Headphones







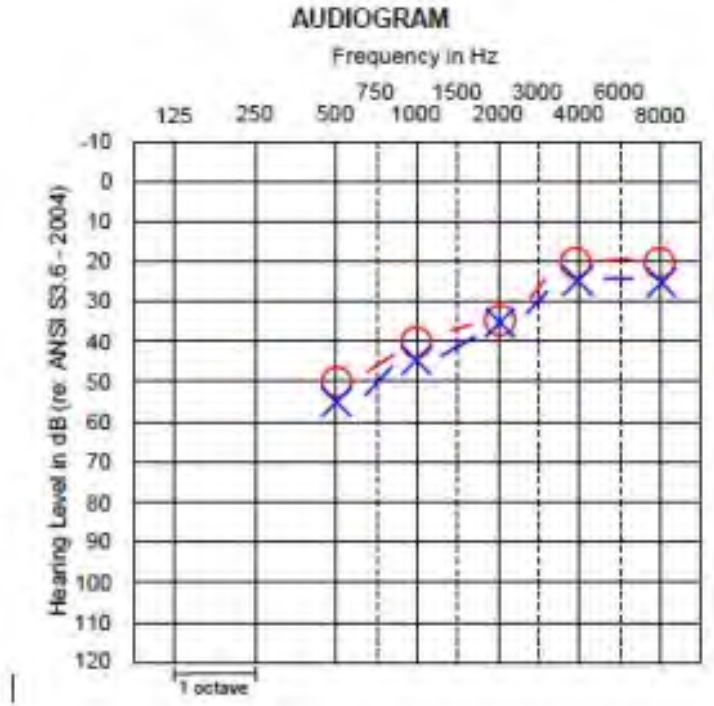
Speech Audiometry:

Date	Side	Test	Percentage Correct/Threshold	Choice	Word List Used	Condition
03-17-2023	Right Ear	SRT	50 dB	HL	Body Part Identification	Unaided
	Left Ear	SRT	40 dB	HL	Body Part Identification	Unaided

Visit from 6/23/2023

- Mother reported that Walter had recently been rejecting his hearing aids and stating he "doesn't want them."
- Historically, he had been a very consistent hearing aid user, and the mother was curious if a change in hearing has contributed to this.

Testing from 6/23/2023



- Tympanometry = large ear canal volumes, consistent with patent PE tubes.
- Two-tester CPA paradigm used
- Supra-Aural Headphones

Speech Audiometry:

Date	Side	Test	Percentage Correct/Threshold	Choice	Word List Used	Condition	Stimulus
06-23-2023	Right Ear	SRT	30 dB	HL	Body Part ID	Unaided	MLV
	Left Ear	SRT	35 dB	HL	Body Part ID	Unaided	MLV

What did we do?

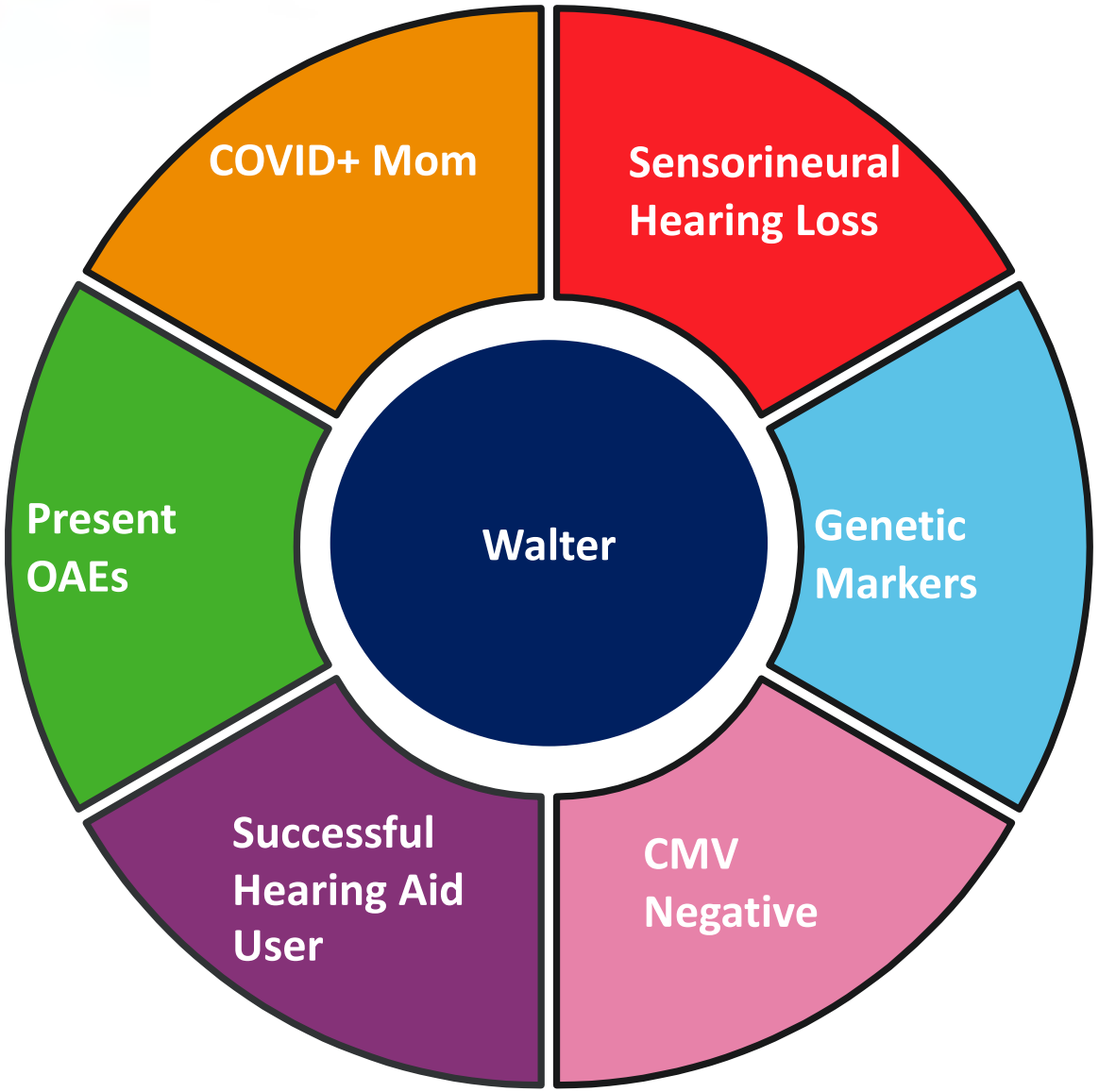
- Reviewed the results with the parents.
- Noted the significant improvement in hearing thresholds.
- Re-programmed the hearing aids.
- Parents to monitor Walter's progress.
- Re-test in 3 months to expand and confirm testing.

Visit from 10/6/2023

- Walter had been doing well since hearing aid re-programming in June.
- Behavioral testing was repeated, with the addition of bone-conduction.
  - Consistent with June's results.
  - SRT completed with recorded picture spondees.
- Hearing aid programming was not adjusted.
- Agreed to continue to monitor Walter closely.

Take-Aways

- Every child/case is going to be different.
- We need to adjust our care plan to meet their needs and support their growth and development.


COVID+ Mom
Sensorineural Hearing Loss
Present OAEs
Walter
Genetic Markers
Successful Hearing Aid User
CMV Negative

References

Charizopoulou, N., Lelli, A., Schraders, M., Ray, K., Hildebrand, M. S., Ramesh, A., Srisailapathy, C. R., Oostrik, J., Admiraal, R. J., Neely, H. R., Latoche, J. R., Smith, R. J., Northup, J. K., Kremer, H., Holt, J. R., & Noben-Trauth, K. (2011). GIPC3 mutations associated with audiogenic seizures and sensorineural hearing loss in mouse and human. *Nature Communications*, *2*(1). https://doi.org/10.1038/ncomms1200

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Introducing Maria

A case of many hurdles to proper care

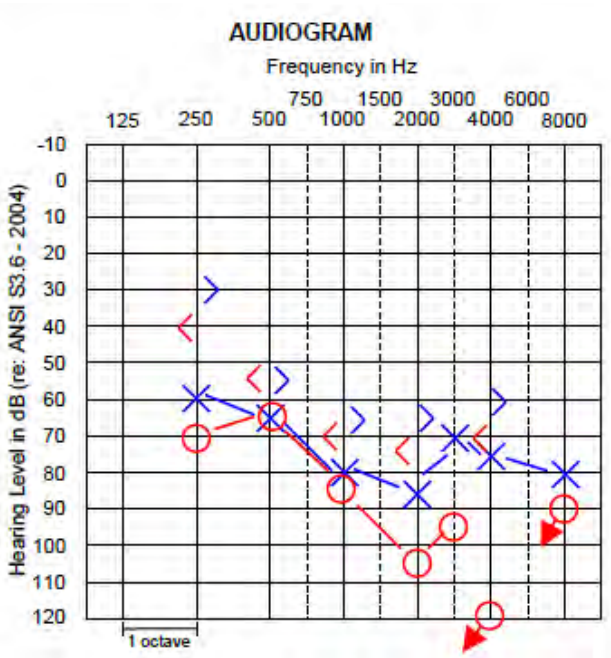
Allie Sayer, Au.D., CCC-A

Maria's First Visit

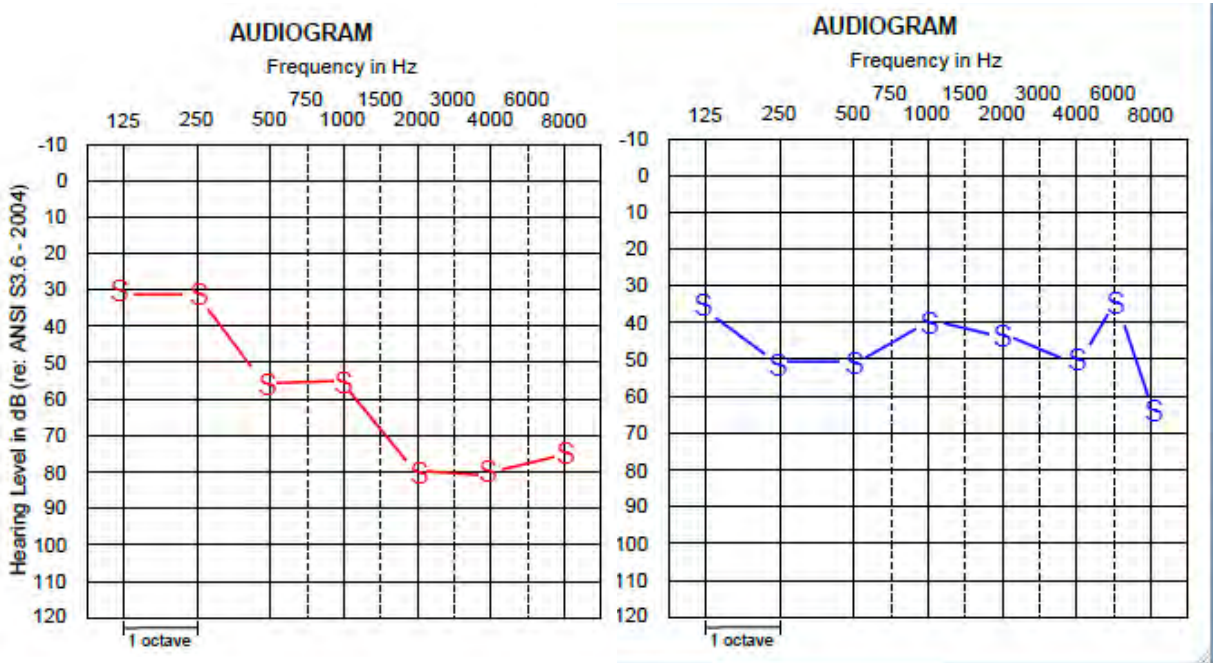
- Seen in November 2019 at 11 years of age.
- Recently relocated from Florida, where she first received care in the U.S. after moving from Venezuela
- No insurance due to immigration status (asylum)

Outside Records - Venezuela

1/13/2014

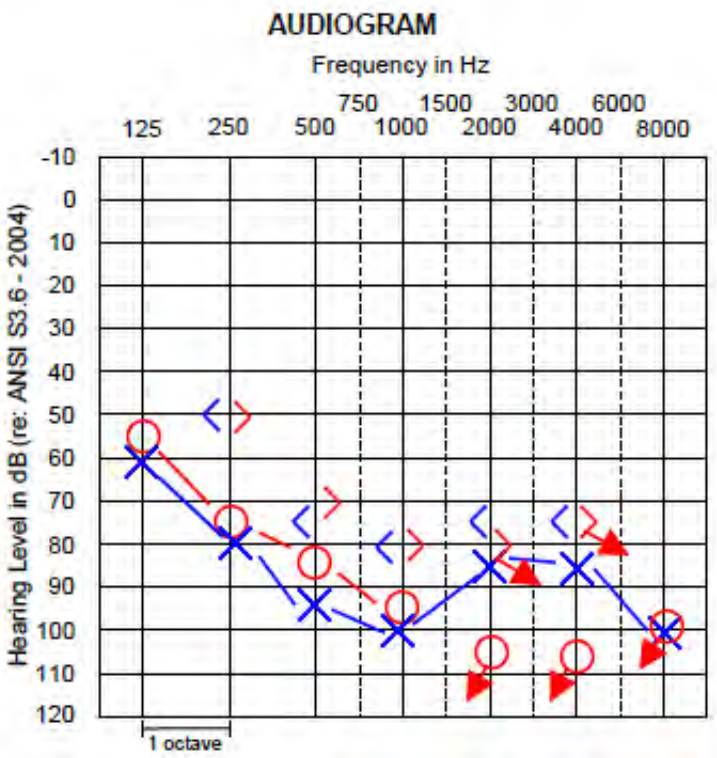


3/26/2015

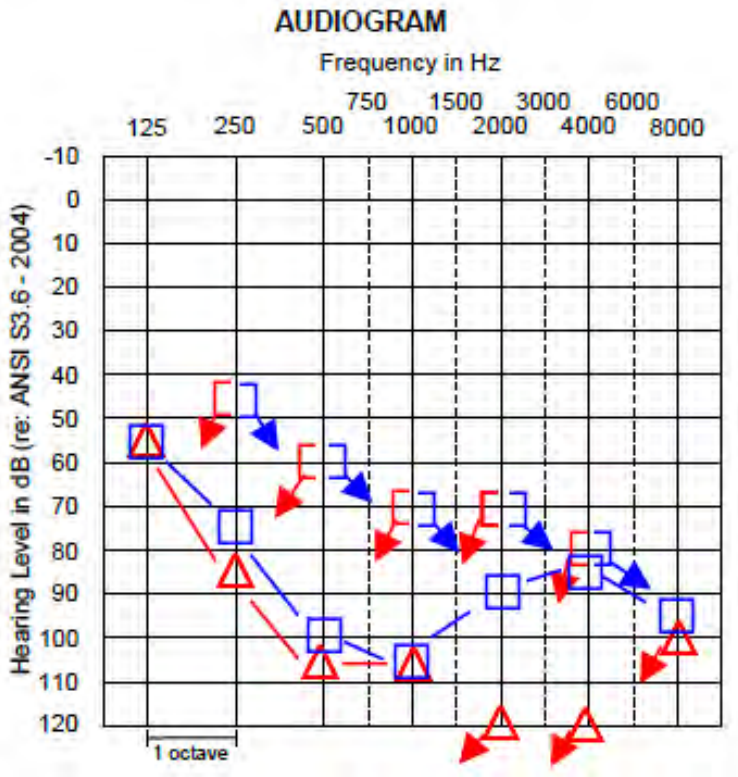


Outside Records - Venezuela

12/12/2016

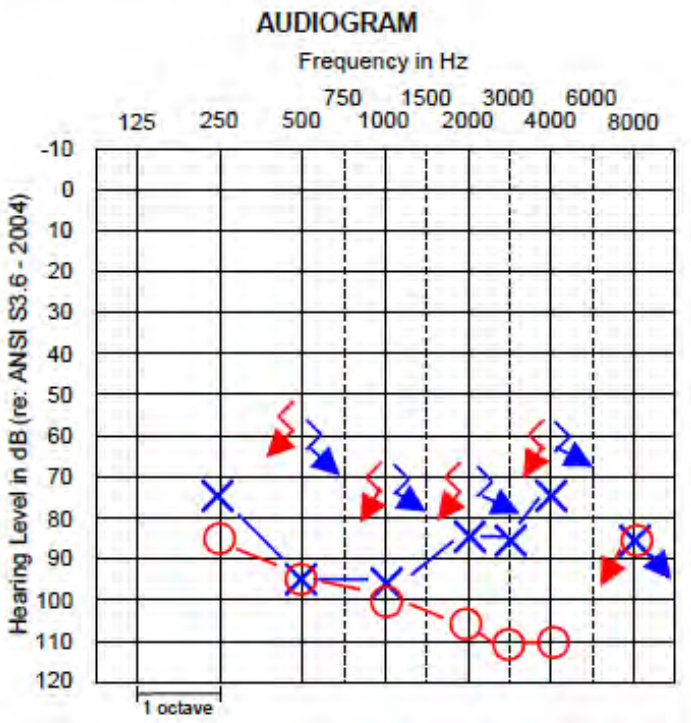


3/20/2018



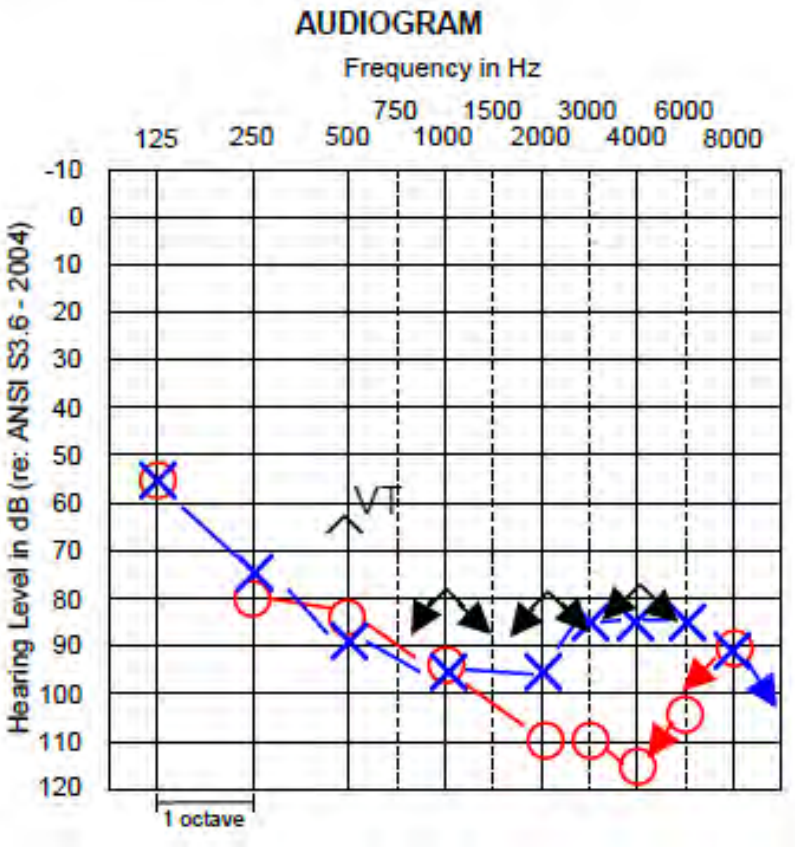
Outside Records - Florida

3/26/2019



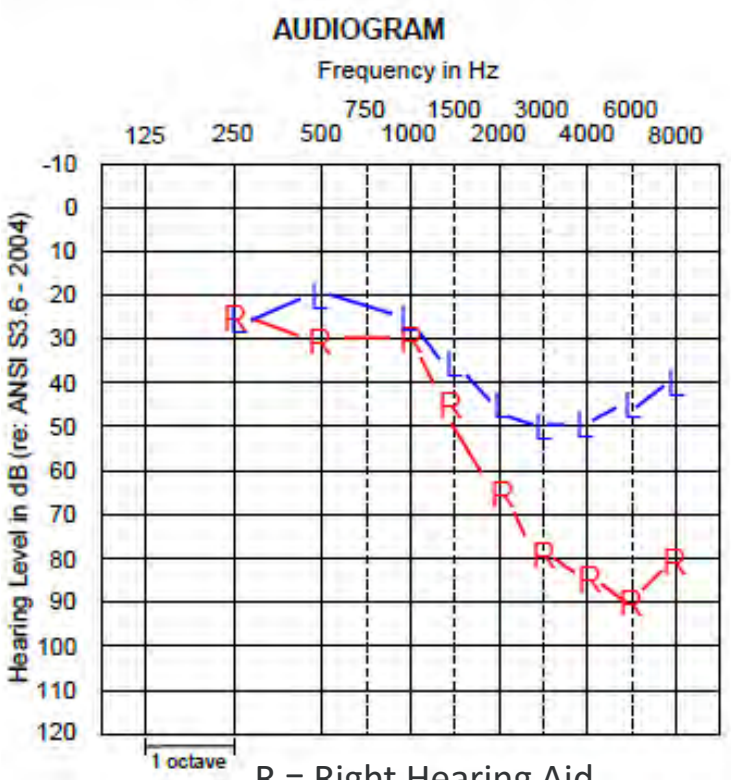
- Family requesting evaluation for cochlear implants.
- Wearing a single, slimtube BTE in the left ear with closed dome.

Our First Test Results



- Fit with loaner high-power behind the ear (BTE) hearing aids
  - Complained of poor sound quality from right
  - Otherwise very happy with the new hearing aids

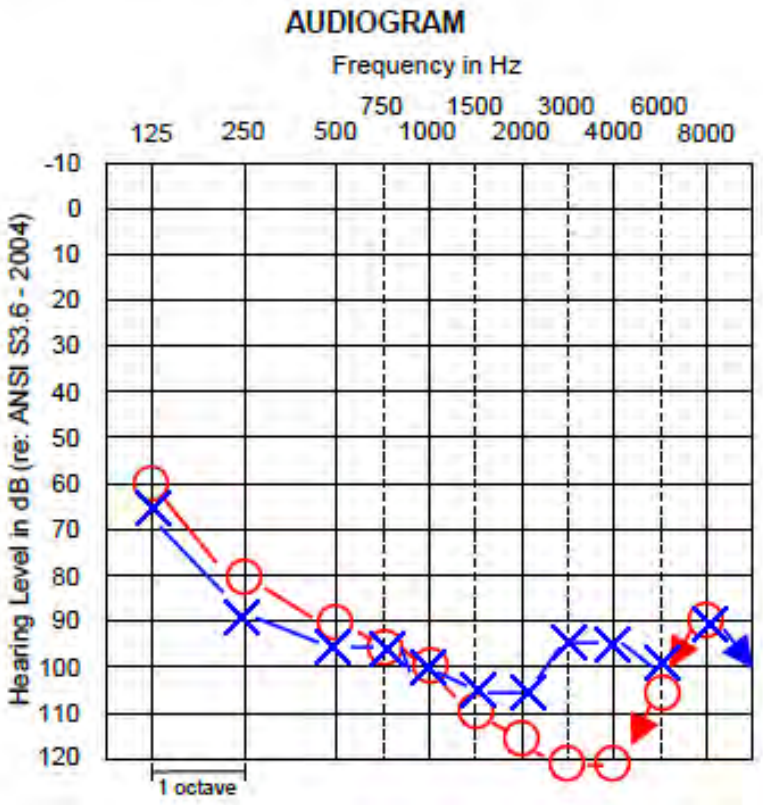
Follow-up



R = Right Hearing Aid
L = Left Hearing Aid

- Spanish WRS Testing
  - Listas de Palabras Bisilabicas Para Ninos at 60 dB SPL
  - Right Aided: 24%
  - Left Aided: 64%
- Concerns:
  - MLV presented by mother
  - Scoring

Follow-up



- August 2020
  - Decrease in hearing
  - Online school due to pandemic
  - Hearing aids reprogramed
- November 2020
  - New onset tinnitus
  - Hearing essentially stable
  - Unable to increase hearing aid gain
  - NOW HAS INSURANCE!

CI Evaluation

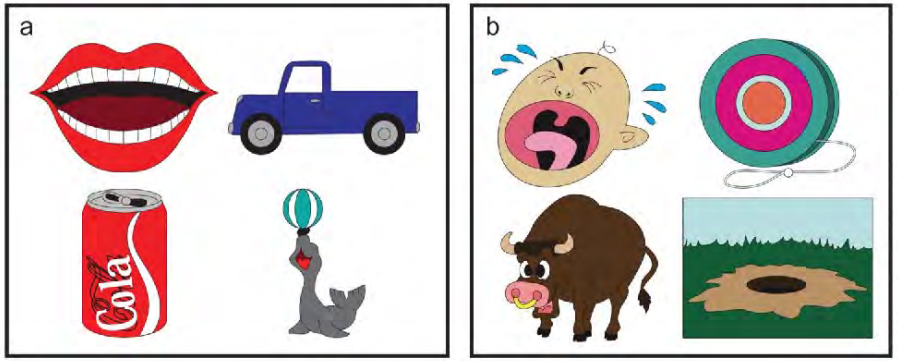
- Team Workup
  - ENT consult
  - CT scan
  - Psychology consult
  - Speech and language evaluation
- Audiology Testing
  - MLV for speech testing with mom's voice

	Unaided	Aided
Right	20%	48%
Left	52%	64%

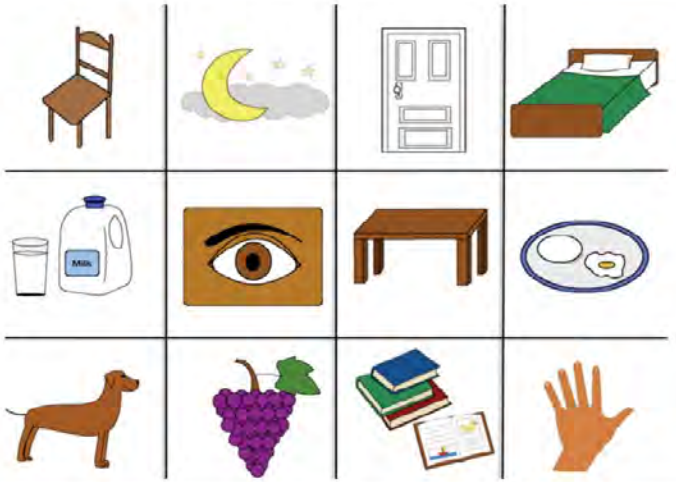
Recorded Spanish Materials

- Standard Lists
  - Spanish Auditory Tests
    - Trisyllables for SRT
    - Bisyllable WRS Lists
    - Monosyllable WRS Lists
  - AZ Bio en Espanol
- Pediatric Tests
  - Spanish Pediatric Identification Test (SPPIT)
  - Spanish Pediatric Speech Recognition Threshold Test (SPSRT)

SPPIT (Mendel et al., 2020)



SPSRT (Mendel et al., 2019)



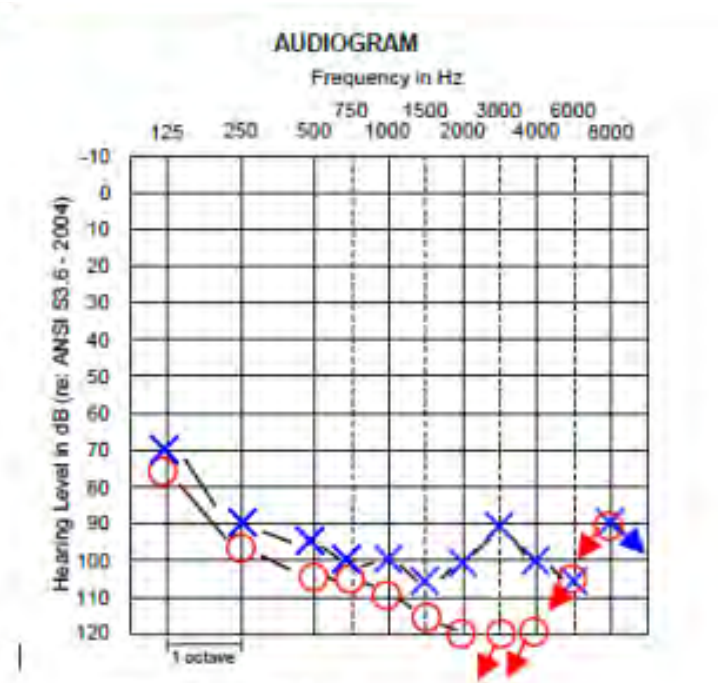
Re-evaluation with Recorded

	Right Ear	Left Ear
MLV	48%	64%
Recorded	**0%**	**12%**

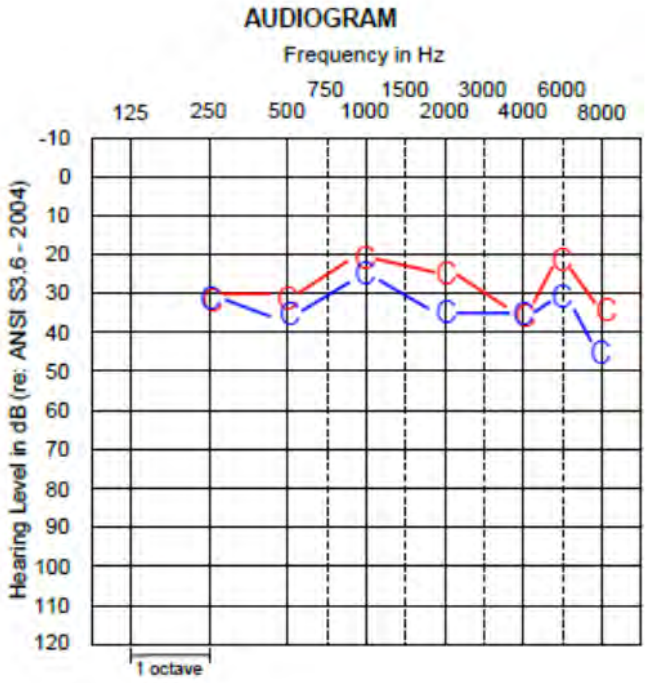
- Team now agreed with bilateral simultaneous implantation
- Surgery in April 2021
- Activated 2 weeks post-operatively in May 2021
- Immediately compliant with full-time use, and speech therapy attendance.

Outcomes

- 3 Weeks Post-Activation
  - Communicating with video interpreter
  - Reporting subjective easier speech understanding than with her hearing aids
- Residual Hearing

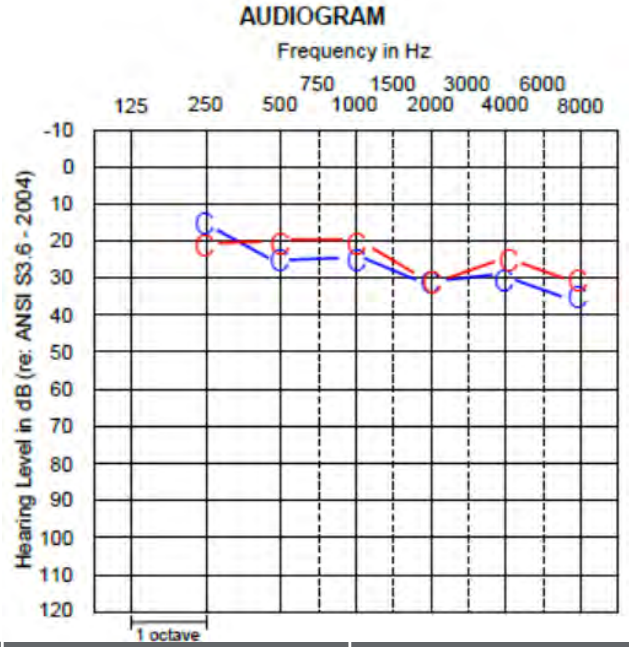


7 Weeks Post-Activation



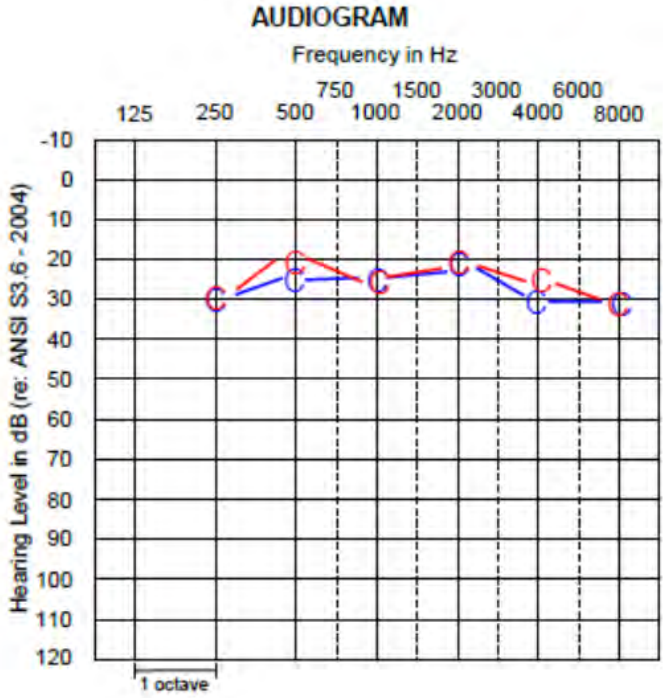
	Right	Left	Bilateral
Pre-CI (UP HAs)	0%	12%	DNT
CI	**40%**	**40%**	**44%**

6 Months Post-Activation



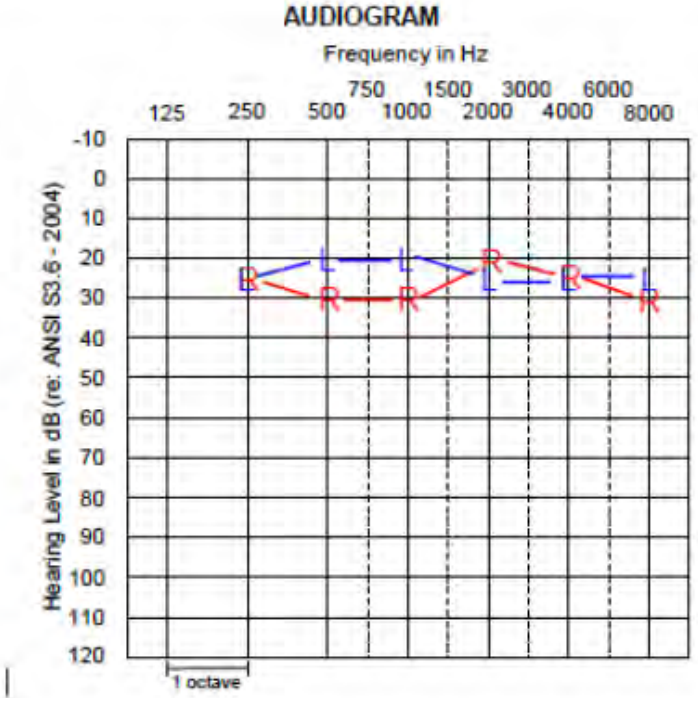
	Right	Left	Bilateral
Pre-CI (UP HAs)	0%	12%	DNT
CI 7 Weeks Post	40%	40%	44%
CI 6 Months Post Spanish	**44%**	**60%**	**DNT**
CI 6 Months Post English	**32%**	**60%**	**DNT**

1 Year Post-Activation



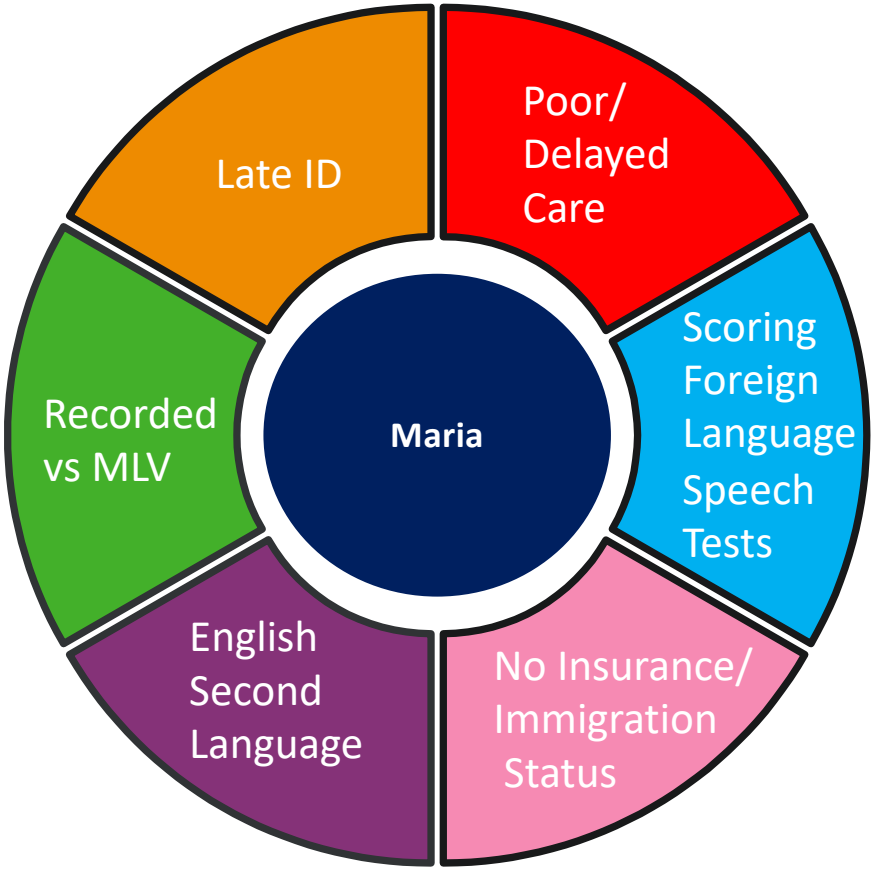
	PBK ½ Lists
Right Ear	40%
Left Ear	68%

Now



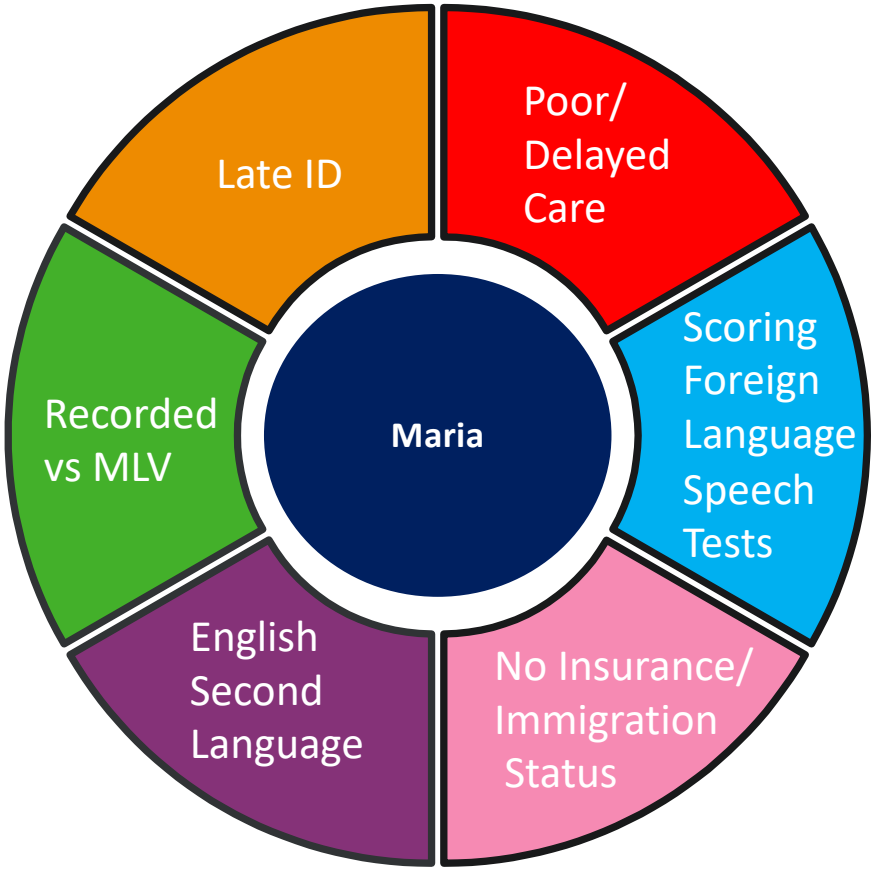
	Right	Left	Bilateral
English	60%	76%	72%
Spanish	44%	84%	64%

The Uniqueness of Maria



Take Aways

- Language barriers are extremely complicated in evaluation and treatment of patients with hearing loss.
- Use of recorded materials is important.
- Success looks different for every patient.



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Introducing Jacob

A Case of NF2

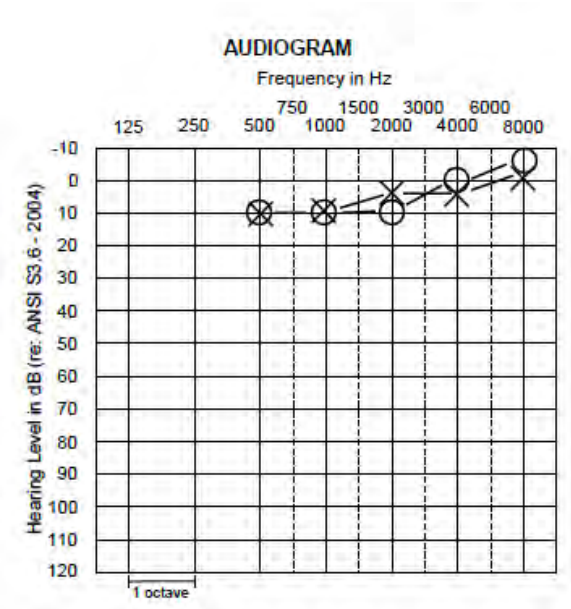
Alissa Nickerson, Au.D., CCC-A

Jacob

- "Jacob," is a *now* 16-year-old male with Neurofibromatosis Type 2.
- At the current time, he has multiple intracranial tumors, including bilateral vestibular schwannomas.
- His symptoms onset around age 5, when he developed an abdominal soft tissue lesion.
- Biopsy through external dermatology group revealed a histopathology consistent with a schwannoma.

Initial Work-up: 2018

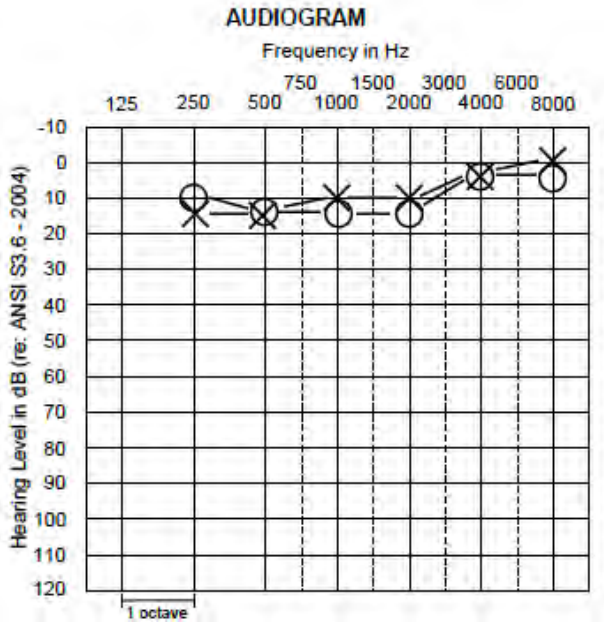
- **MRI 5/21/2018**: Multiple intracranial lesions consistent with manifestations of neurofibromatosis type 2. Bilateral right (10mm) greater than left (4mm) vestibular schwannomas.
- **Audiologic Evaluation 8/2/2018**:
  - Present distortion product otoacoustic emissions (DPOAES) AU
  - Ipsilateral acoustic reflexes (AR) present AU 500-4000 Hz
  - Audiologic testing within normal limits (WNL) 500-8000 Hz
  - Word recognition score (WRS) excellent AU at soft input level, 40 dB HL



Fast Forward to 2020...

- **MRI 1/24/2020:** Increased size of the bilateral vestibular schwannomas.

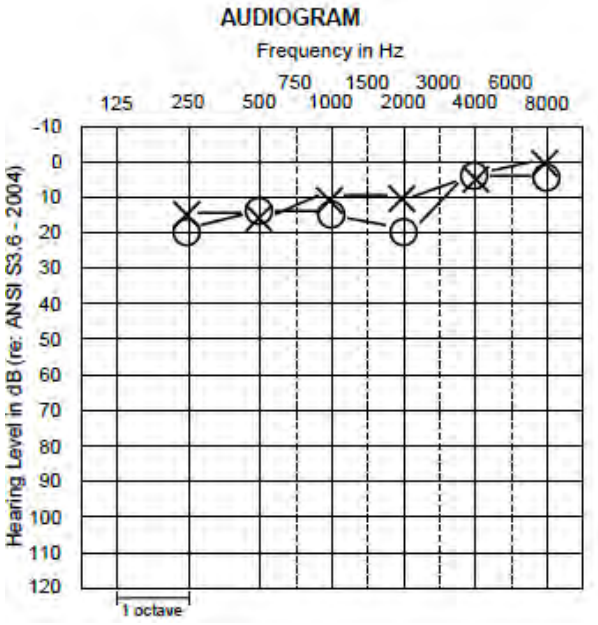
- **Audiologic Evaluation 4/24/2020:**
  - Overall present DPOAEs AU
  - Ipsilateral AR present left, absent right
  - Audiologic testing WNL 250-8000 Hz
  - WRS excellent AU at soft input level, 50 dB HL



Early 2021

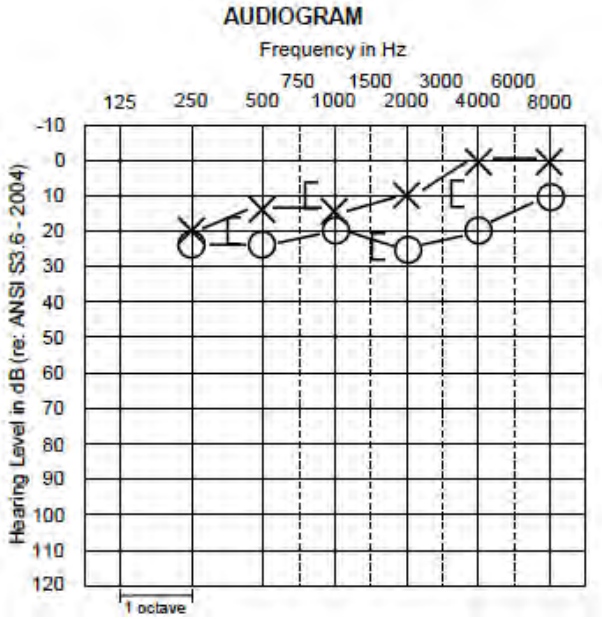
- **MRI 1/11/2021:** Yet again, increased size of bilateral vestibular schwannomas.

- **Audiologic Evaluation 2/2/2021:**
  - Present DPOAEs AU, but reduced on the right
  - Ipsilateral AR present left, absent right
  - Audiologic testing WNL 250-8000 Hz
  - WRS excellent AU at soft input level, 50 dB HL
    96% right, 100% left
  - WRS good right and excellent left at loud input level, 80 dB HL
    80% right, 92% left



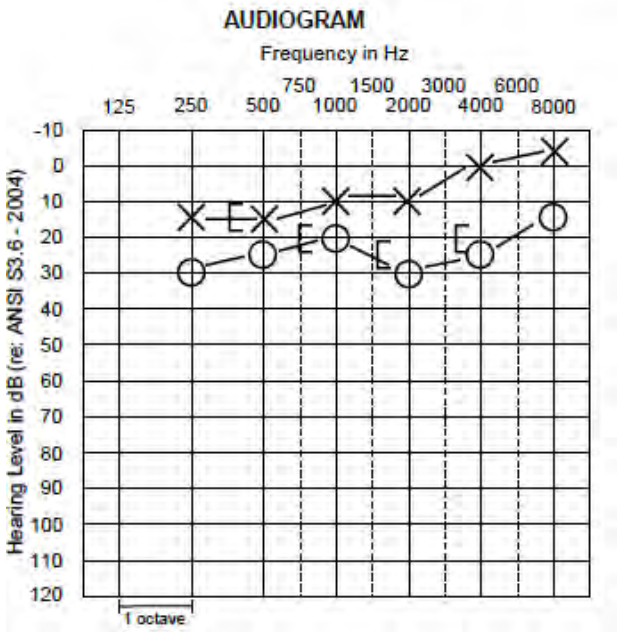
Late 2021

- **MRI 12/3/2021:** The bilateral IAC vestibular schwannomas were stable.
- **Audiologic Evaluation 8/10/2021:**
  - DPOAEs reduced <3kHz right, present overall left
  - Ipsilateral AR 1kHz present left, absent right
  - Audiologic testing revealed mild/slight SNHL
    right and normal hearing sensitivity in the left
  - WRS excellent bilaterally
    (60 dB HL right, 50 dB HL left)



2022 – Something New...

- **MRI 5/9/2022:** Demonstrate size stability of larger/dominant masses in the bilateral internal auditory canals.
- **Audiologic Evaluation 2/1/2022:**
  - DPOAEs absent bilaterally
  - Ipsilateral AR present left, absent right
  - Audiologic testing reveals slight worsening of right mild SNHL with normal hearing sensitivity left
  - **WRS very poor at average (36%) and loud (16%) input levels right and excellent at average input level (92%) left.**



Avastin Therapy (bevacizumab)

- Huang et al (2018) show improvement in patient-reported speech understanding and auditory quality with therapy.
- Plotkin et al (2009) suggest that treatment can be associated with hearing improvement, tumor-volume reduction, or both, in some patients with NF2.

- In Jacob's case, although his hematology/ oncology provider introduced this as an option in April of 2020, it was not recommended until after the February 2022 evaluation showed diminished WRS.
- He started Avastin therapy after his abdominal surgery in March 2022.

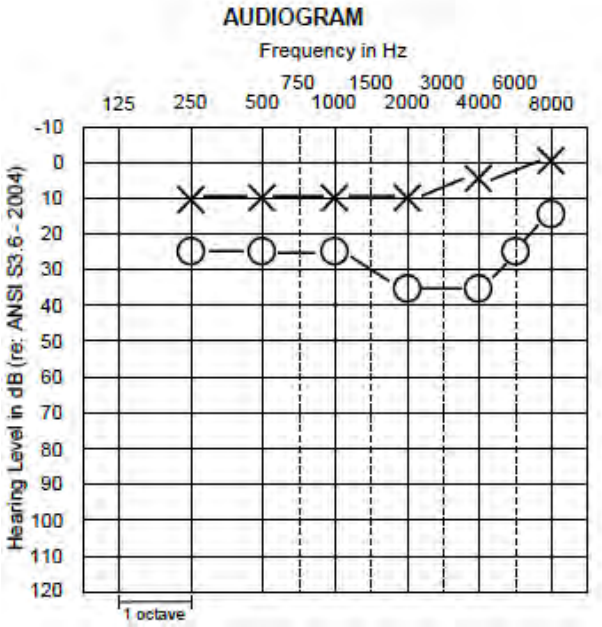
Huang V, Bergner AL, Halpin C, Merker VL, Sheridan MR, Widemann BC, Blakeley JO, Plotkin SR. Improvement in Patient-reported Hearing After Treatment With Bevacizumab in People With Neurofibromatosis Type 2. Otol Neurotol. 2018 Jun;39(5):632-638. doi: 10.1097/MAO.0000000000001781. PMID: 29649040; PMCID: PMC6642810.

Plotkin SR, Stemmer-Rachamimov AO, Barker FG 2nd, Halpin C, Padera TP, Tyrrell A, Sorensen AG, Jain RK, di Tomaso E. Hearing improvement after bevacizumab in patients with neurofibromatosis type 2. N Engl J Med. 2009 Jul 23;361(4):358-67. doi: 10.1056/NEJMoa0902579. Epub 2009 Jul 8. PMID: 19587327; PMCID: PMC4816642.

June 2022 – Did we turn a corner?

- **MRI 8/9/2022:** Size stability

- **Audiologic Evaluation 6/7/2022:**
  - DPAOES present (but somewhat reduced) right and present overall left
  - Ipsilateral AR absent right, present left
  - Audiologic testing revealed mild hearing loss right ear with normal hearing sensitivity in the left ear.
  - WRS good in the right ear (88%) and excellent in the left ear (100%)
    - 50 dB HL left, 60 dB HL right



Summary of "Jacob"

- Individuals impacted by NF2 generally develop hearing loss.
- Hearing improvement and tumor-volume reduction can be achieved with Avastin therapy in *some* patients.
- Data on long-term maintenance therapy is limited.
- Where is Jacob today? Doing well!


Surveillance
Minor Change,
Big Impact
Patient
report
Jacob
Counseling
Observation
Future Planning

Introducing Noah

A Late Identification of Hearing Loss

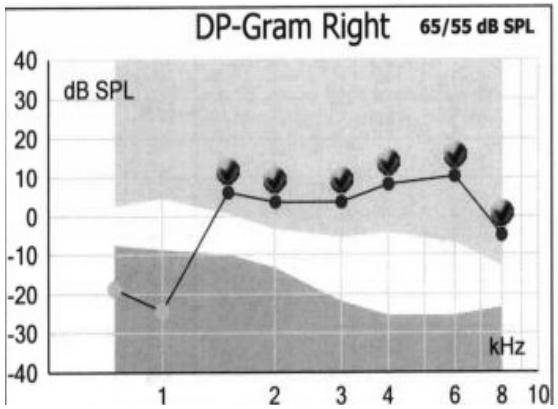
Ashley Geske, Au.D, CCC-A

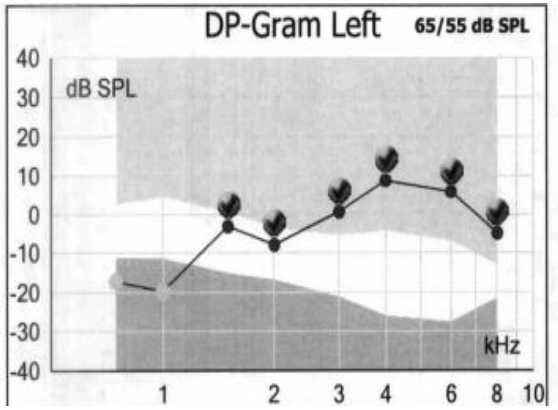
Noah

- Presented to our clinic at 8 ½ years old.
- Referred from an outside clinic due to inability to obtain behavioral testing.
- Parents reported significant concerns for hearing.
- Multiple failed hearing screenings since kindergarten, however, always passed OAE screening.
- Per mother previous testing has always been inconclusive.

1st Audiogram

- Tympanometry showed Type A tympanograms bilaterally
- OAEs were present/reduced 1.5-8kHz bilaterally
- Ipsilateral acoustic reflexes were essentially elevated/absent bilaterally



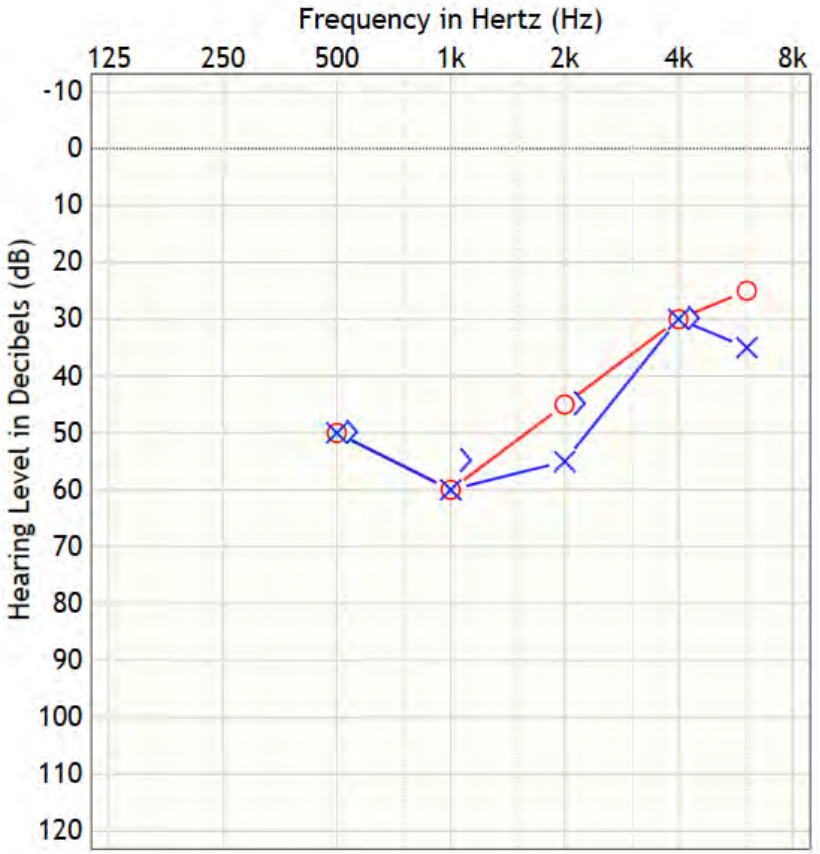


ACOUSTIC REFLEX TESTING (dB HL)

Sound in Right Ear					Sound in Left Ear			
0.5 kHz	1 kHz	2 kHz	4 kHz		0.5 kHz	1 kHz	2 kHz	4 kHz
				Contra				
95	105	RA		Ipsi	100	RA	RA	

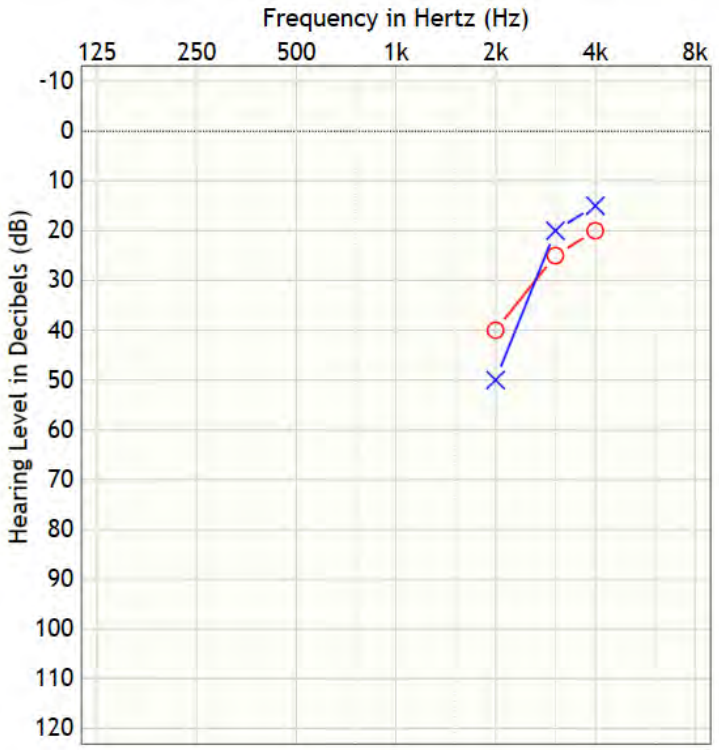
1st Audiogram

- Good Reliability
- Attempted PBK however could not tell if errors were due to articulation or hearing
- Completed a WIPI at 80dBHL
  - Right: 80%
  - Left: 100%
- Recommended BAER study and repeat audiogram



2nd Audiogram

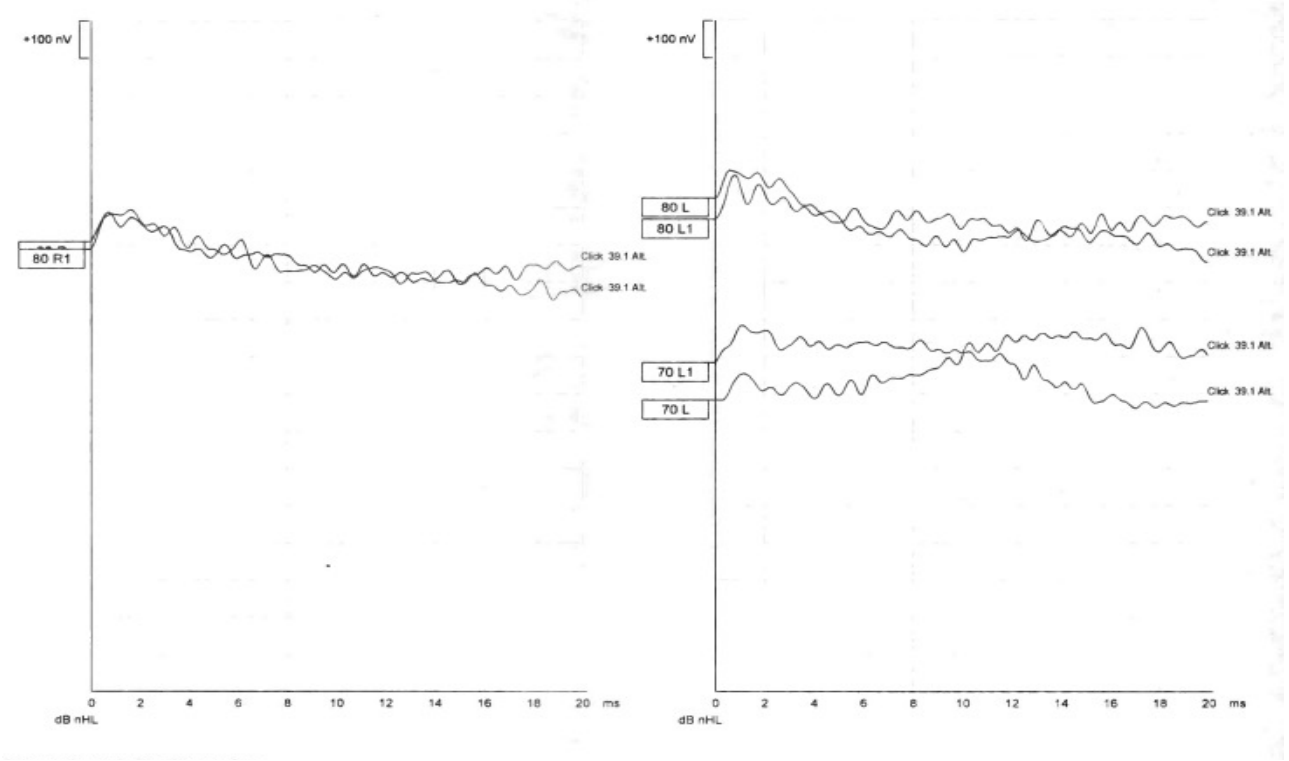
- Completed multiple levels of WRS to cross check audiogram reliability.
- Thresholds that were obtained were consistent with previous testing



Side	Test	Presentation Level	Percentage Correct/Threshold	Word List Used	Stimulus
Right Ear	SRT		40dBHL*	Picture pointing spondees	Recorded
Left Ear	SRT		45dBHL*	Picture pointing spondees	Recorded
Right Ear	WRS	45dBHL	24%	WIPI	Recorded
Left Ear	WRS	45dBHL	8%	WIPI	Recorded
Right Ear	WRS	70dBHL	64%	WIPI	Recorded
Left Ear	WRS	70dBHL	80%	WIPI	Recorded

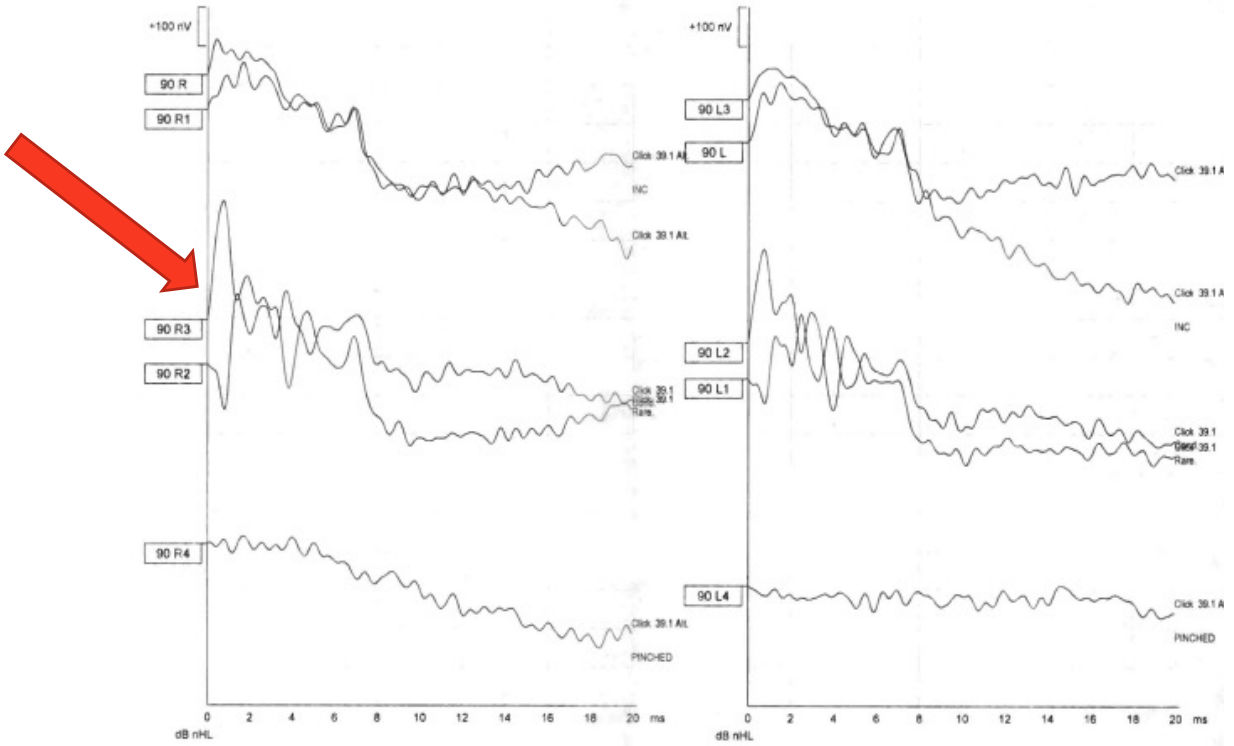
BAER Study

- Click at 70 and 80dBnHL
- No identifiable wave V waveforms observed



BAER Study

- Large cochlear microphonic observed at high intensities.
- Possible wave V observed at 90dBnHL however, disappeared at 80dB nHL.

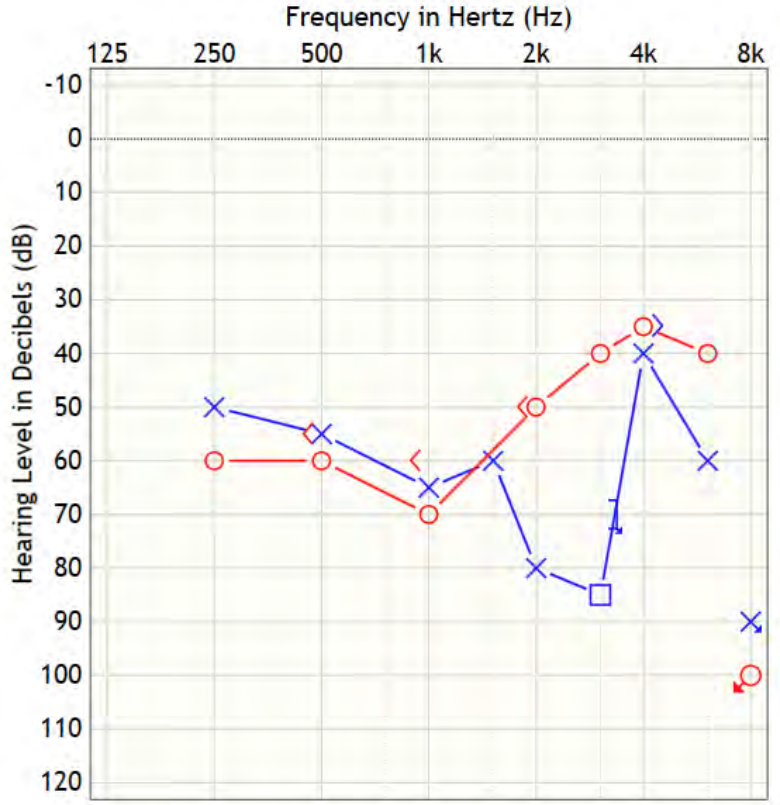


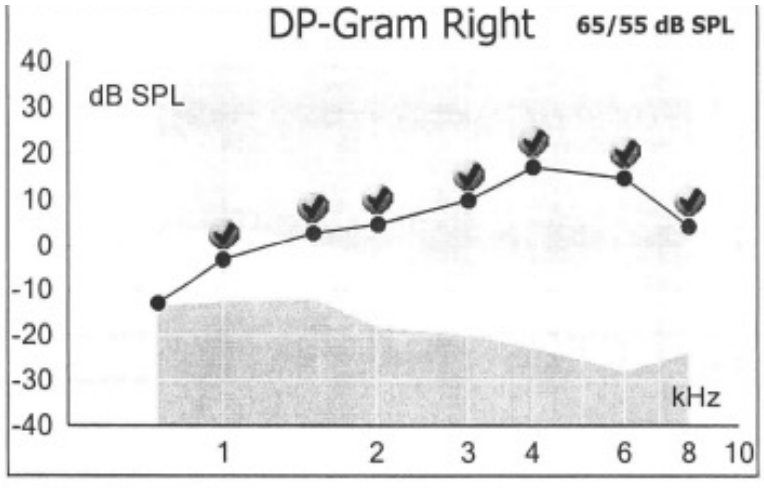
Hearing Aid Trial

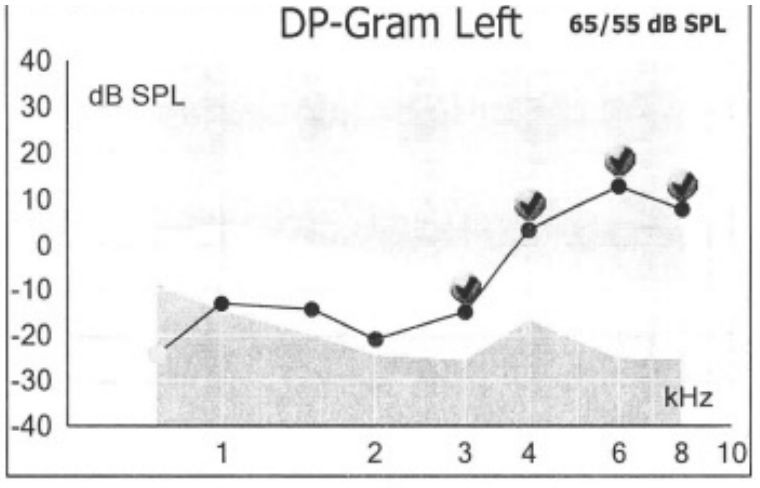
- 6-month hearing aid trial to assess benefit.
- Fit with loaner devices in both ears.
- Noah liked the hearing aids and data logging was 11+ hours a day.
- Parent and teachers report he does better with devices on however is still relying on visual cues.
- Still struggles in background noise and when speaker is further away.
- Had formal psychological evaluation and was diagnosed with autism level 1.

CI Evaluation

- Progression from 2-3kHz in the left ear.
- Progression also noted in his OAEs.







Cochlear Implant Evaluation

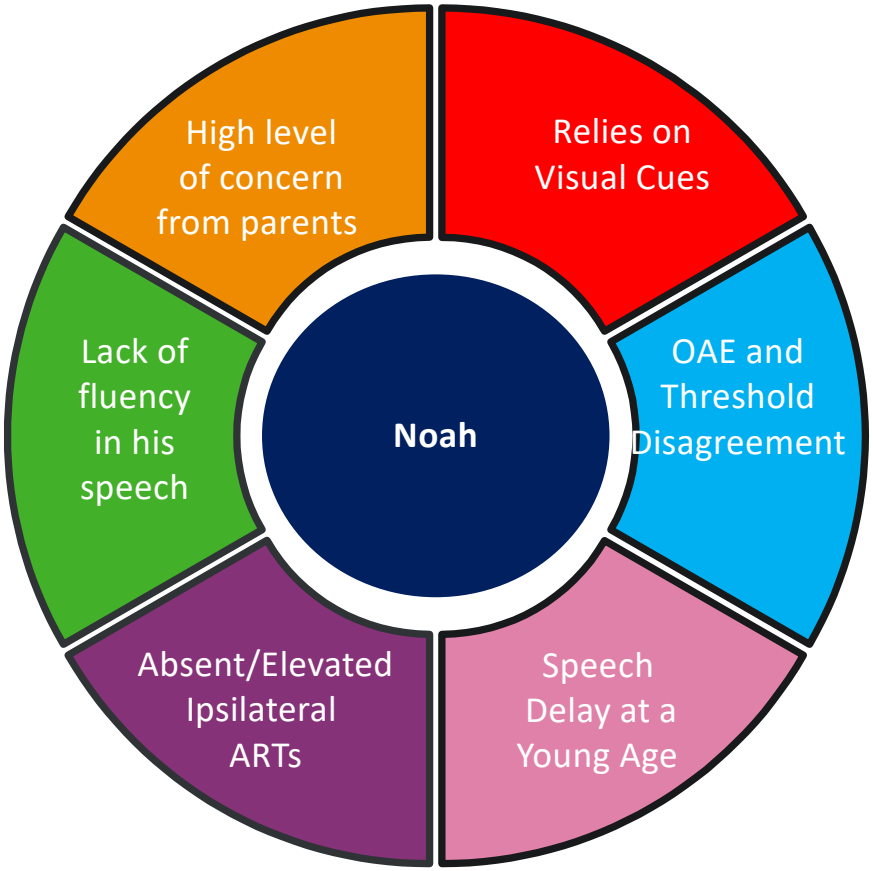
- Aided monaural and binaural WRS indicated very poor speech understanding in both ears.
- Speech in noise testing with 0dB and +5dB SNR was 16% and 29% respectively.

Aided Word Recognition:

Date	Side	Device	Presentation Level	dB Type	Percentage Correct (Words)	Percentage Correct (Phonemes)	Word List	Stimulus	Condition	Signal-to-Noise Ratio
10-12-202	Right SF	HA	60	dBA	16%	37%	CNC List 5	Recorded	quiet	
10-12-202	Left SF	HA	60	dBA	20%	53%	CNC List 2	Recorded	quiet	
10-12-202	Bin SF	HA	60	dBA	36%	65%	CNC List 1	Recorded	quiet	
10-12-202	Bin SF	HA	60	HL	16%	N/A	Peds AZ Bio List	Recorded	noise	0dB
10-12-202	Bin SF	HA	60	HL	29%	N/A	Peds AZ Bio List	Recorded	noise	+5dB

To Be Continued

- Still pending discussion with cochlear implant team.
- Still pending MRI/CT scan.
- Genetic ANSD Panel
  - Pathogenic variant in IDUA gene, does not explain his hearing loss.
- Etiology is still not determined meaning:
  - Site of lesion is unknown.
  - Anticipated outcomes are unknown.


Noah
Relies on Visual Cues
OAE and Threshold Disagreement
Speech Delay at a Young Age
Absent/Elevated Ipsilateral ARTs
Lack of fluency in his speech
High level of concern from parents

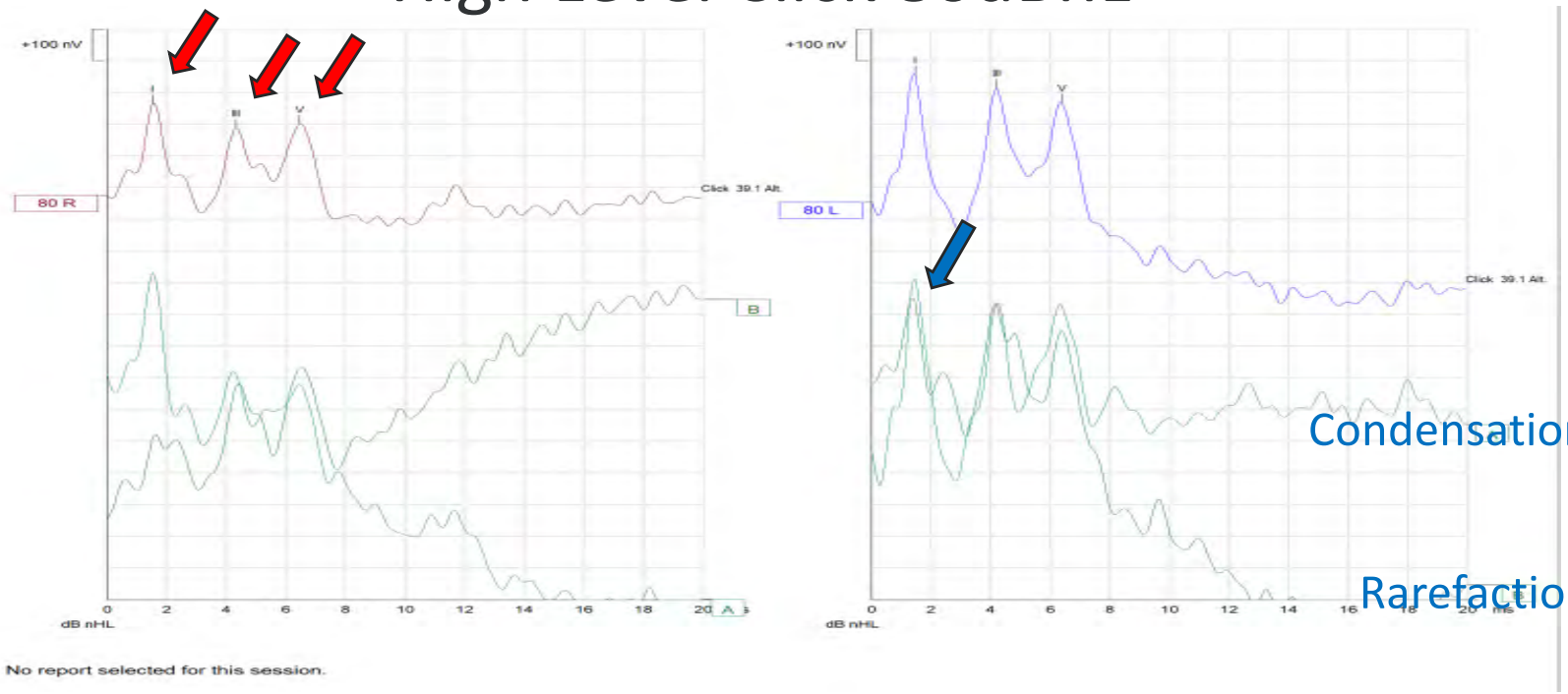
Introducing Ethan

Case of the Abnormal Auditory Brainstem Response Test

Deborah Flynn, Au.D, CCC-A

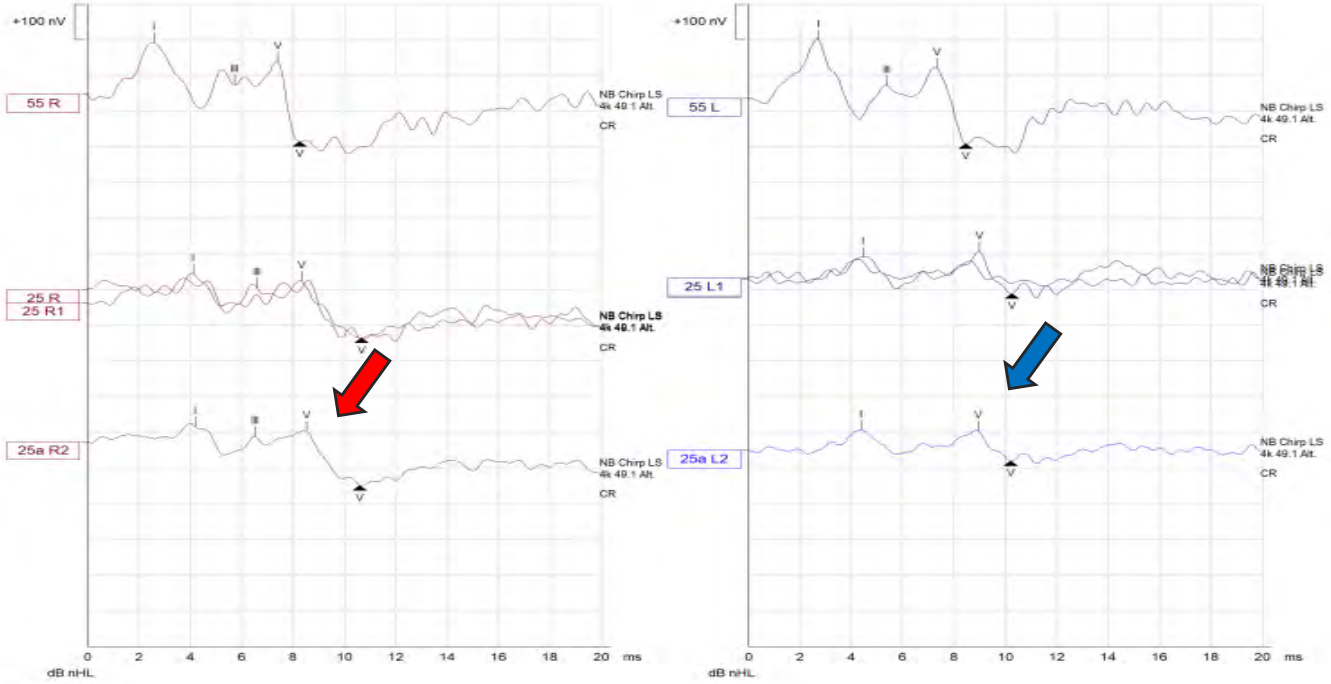
Normal ABR Results

High Level Click 80dBnL



Look for Waves I, III, V
Latency of each wave
How the waveform responses to different stimuli

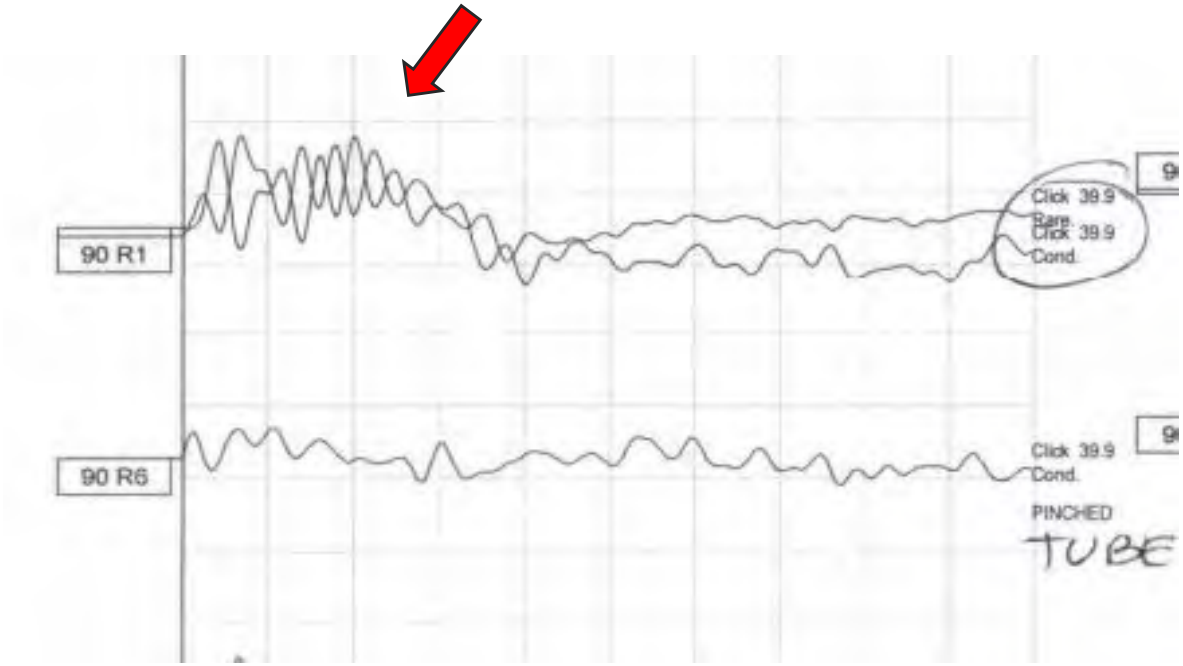
Threshold ABR at 4000Hz



Searching for threshold of Wave V at each frequency

ABR-Auditory Neuropathy

- Reversal of early components of the Wave form



Rarefaction and Condensation

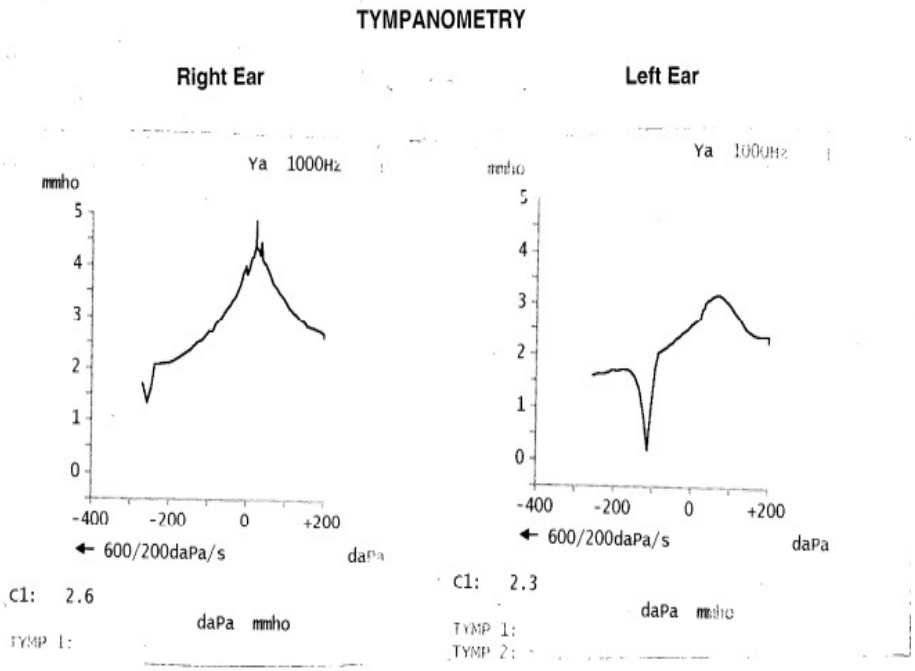
Pinched tube

Ethan's History

- Ethan initially presented at 53-days for an MRI and ABR/BAER study.
  - Born full term without complications
  - One other healthy sibling
  - No family history of hearing loss
  - Failed two inpatient and one outpatient newborn hearing screening.
  - Congenital nystagmus noted at 16 days of life
  - Nystagmus was horizontal, present in all directions of gaze and present at all times

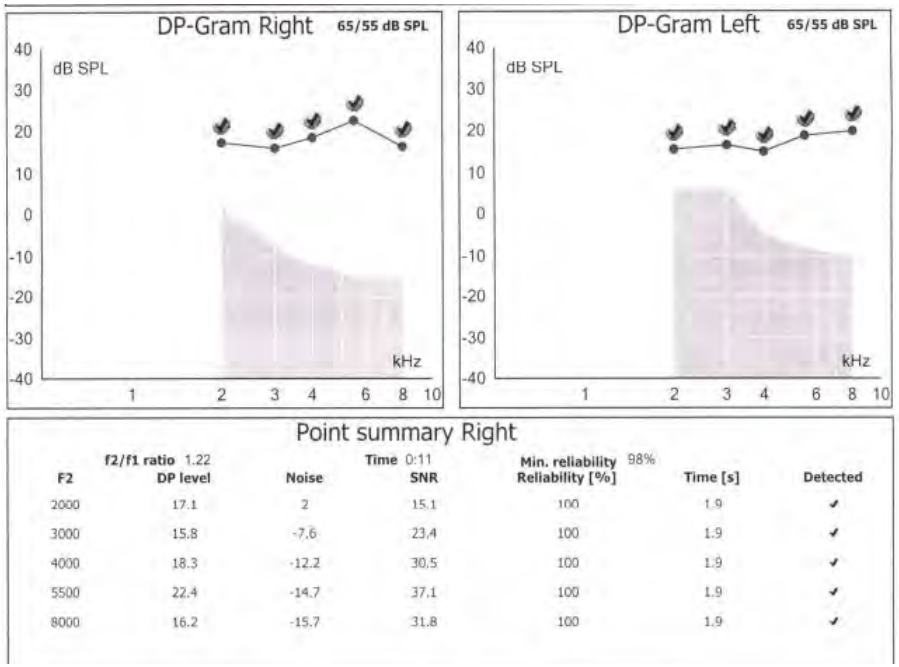
Tympanometry and OAE

1000Hz Probe tone



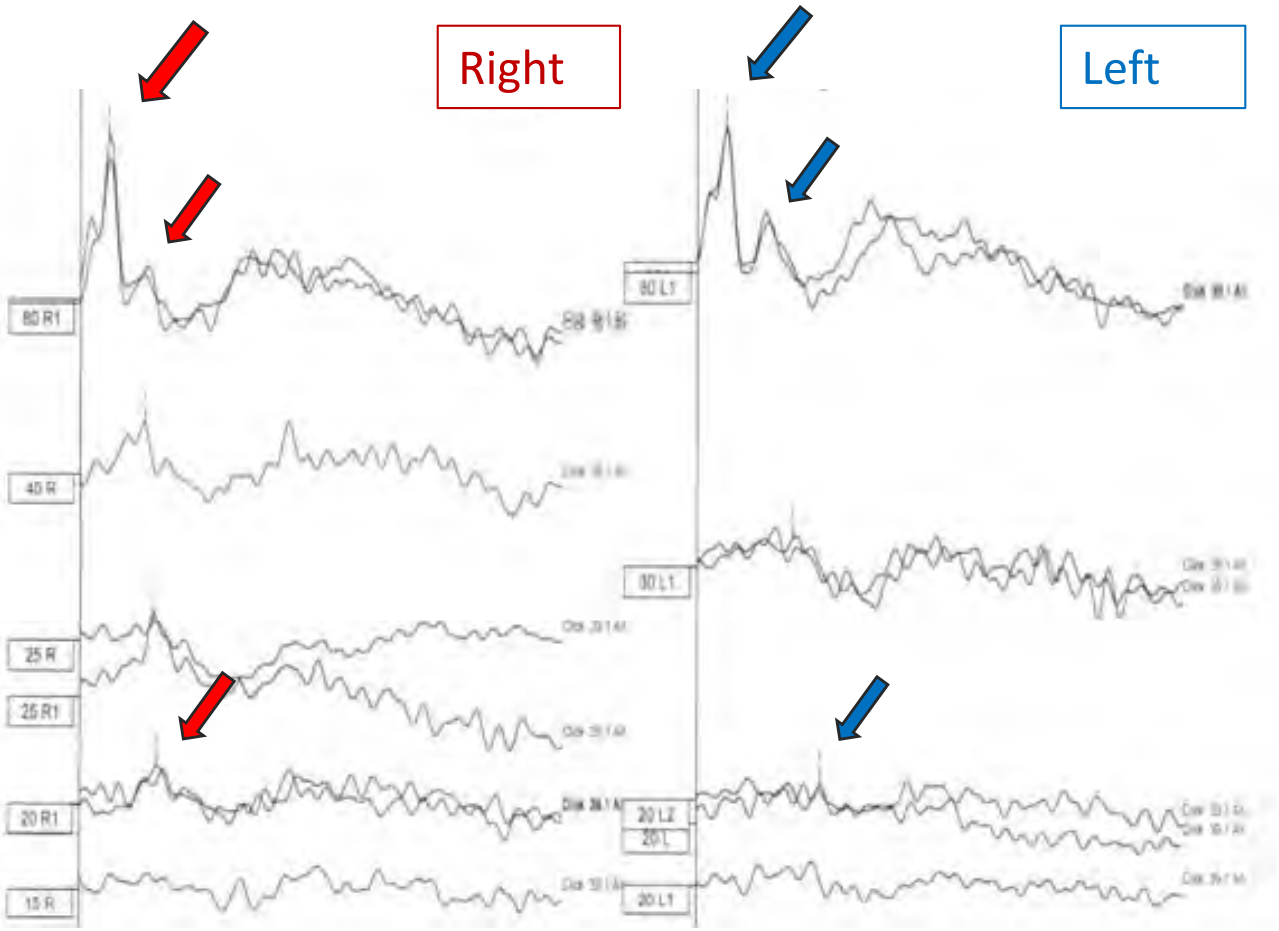
Peaked tympanograms, AU

Distortion Product Otoacoustic Emissions

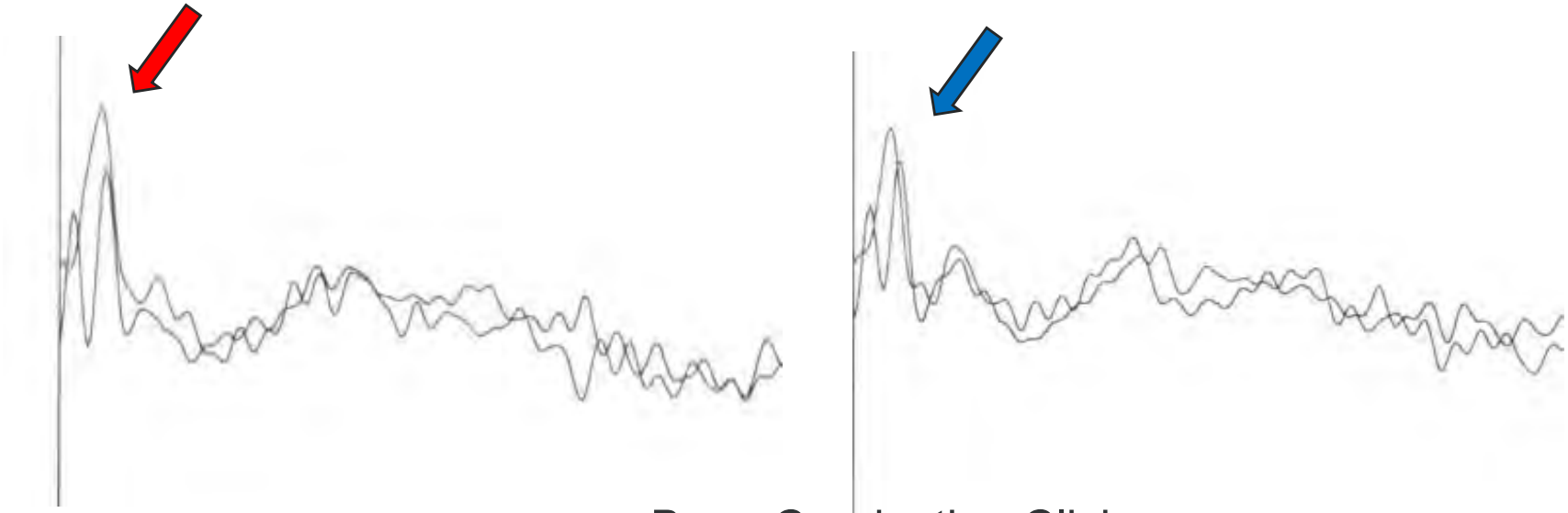


Present otoacoustic emissions

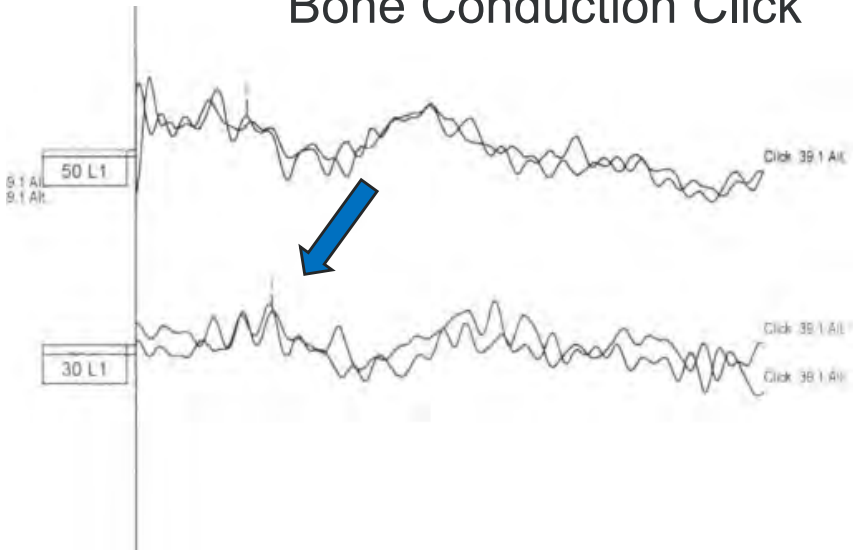
ABR RESULTS
Click Stimulus



Rare and Con tracings 80dBnHL Click



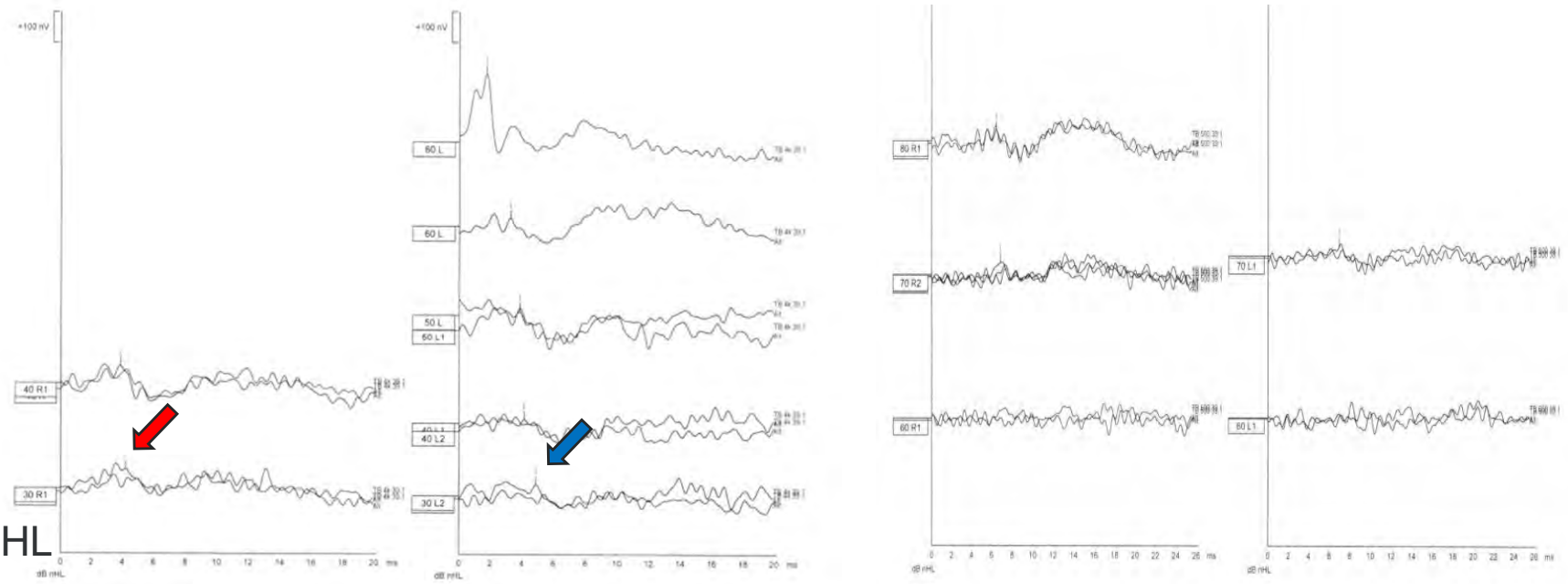
Bone Conduction Click



BAER Study
Tone Bursts

4000Hz

500Hz



30dBnHL

30dBnHL=20dBeHL

Wave I present, measured to threshold

MRI Results

- MRI of the brain without contrast
- Brain:
  - The signal of the brain parenchyma was normal.
  - The myelination pattern was normal for age.
  - There was no mass effect, midline shift, or basal cistern effacement.
  - The ventricles and sulci were normal size and configuration.
  - There was no abnormal fluid collection.
  - There is a fully formed corpus callosum.

MRI Results continued

- Temporal Bone Results
  - External auditory canal was patent.
  - The middle ear cavity and mastoid air cells was clear. The vestibulo-cochlear nerves appeared normal.
  - The inner ear structures, including the semicircular canals, vestibule, and cochlea were unremarkable.
  - The vestibular aqueduct was not enlarged.

  - RADIOLOGIST IMPRESSIONS: **Unremarkable MRI of the brain and temporal bones.**

Reviewing Results with Family

- Difficult diagnosis given the inability to predict auditory function
- MOC was confident that patient could hear noises and startled to sounds.
- This was cautiously promising; along with present OAEs and Wave I measured down to 20dBeHL
- Briefly discussed difference between detection and discrimination.

ABR Report

- **Brainstem Auditory Evoked Responses are abnormal**. The later component waves(Wave III and V) are absent while the earlier components (Waves I and II) are present and measured down to 30dBnHL.
- The overall pattern of findings is consistent with a **disorder at the level of the auditory brainstem pathway**, which limits the application of this test to assess patient's hearing sensitivity.
- There was no extended, reversible cochlear microphonic.
- Distortion product otoacoustic emissions were present, bilaterally.
- **Neural Hearing Loss**

Recommendations

- Repeat ABR in 3 months
- Referral to ENT
- Recommendation for ophthalmology
- Submitted a referral to AZEIP
- Encouraged Hands and Voices/Guide By Your Side
- Neurology

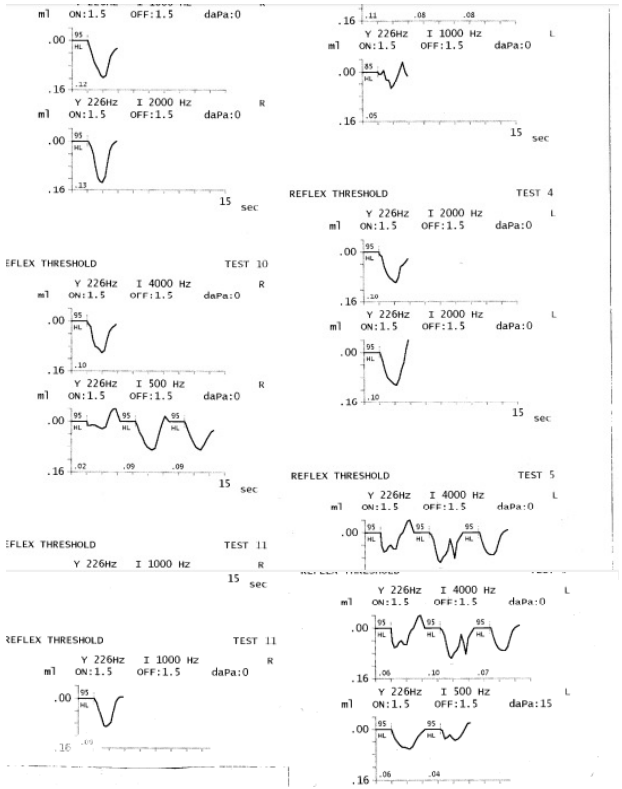
3-month follow-up ABR test

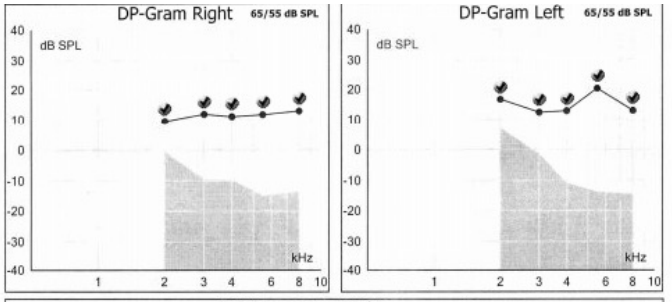
- Pt. Returned for a non-sedated ABR
- Interim History:
  - **ENT**-recommended fup BAER as per audiology recommendations (3 months)
  - **Ophthalmology**- No sign of amblyopia or AHP. Amblyopia suspected, both eyes, secondary to congenital nystagmus: No need for treatment at this time.
  - **Neurology-** Normal MRI, recommended fup in 1 year

Follow-up ABR Study

- 1000Hz tympanograms-peaked
- Acoustic reflexes- present

OAEs-present





Point summary Right

f2/f1 ratio 1.22 | Time 0:14 | Min. reliability 98%

F2	DP level	Noise	SNR	Reliability [%]	Time [s]	Detected
2000	9.5	-0.5	10	100	5.5	✔
3000	11.8	-9.6	21.4	100	1.9	✔
4000	11	-9.8	20.8	100	1.9	✔
5500	11.6	-14.9	26.6	100	1.9	✔
8000	12.8	-13.5	26.4	100	1.9	✔

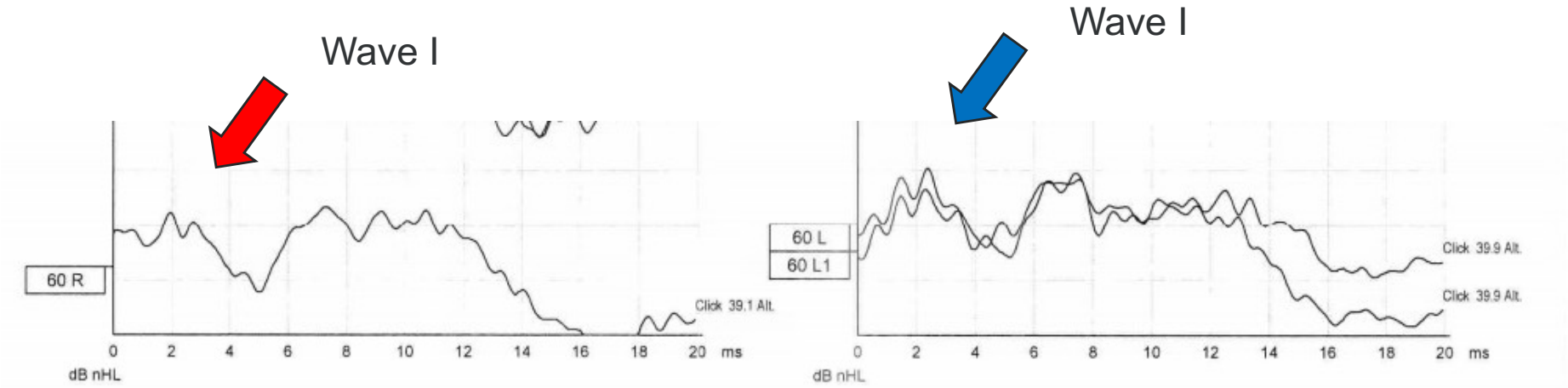
Point summary Left

f2/f1 ratio 1.22 | Time 0:11 | Min. reliability 98%

F2	DP level	Noise	SNR	Reliability [%]	Time [s]	Detected
2000	16.6	6.9	9.6	100	2.6	✔
3000	12.2	-1.5	13.7	100	1.9	✔
4000	12.7	-10.9	23.6	100	1.9	✔
5500	20.3	-13.8	34	100	1.9	✔
8000	12.8	-14.5	27.3	100	1.9	✔

Started testing at 60dBnHL

Click



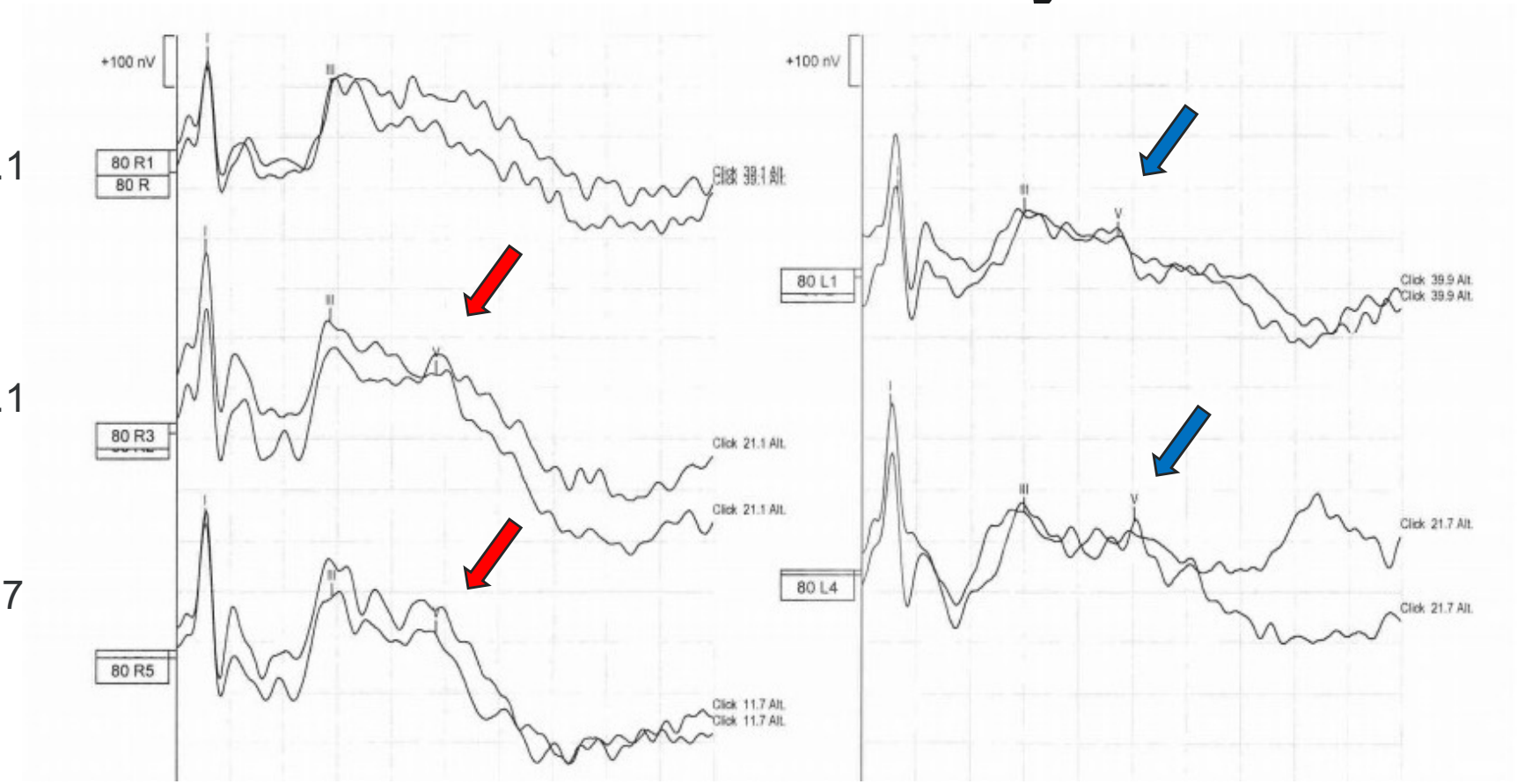
Decided to complete a Rate Study

Rate Study

39.1

21.1

11.7

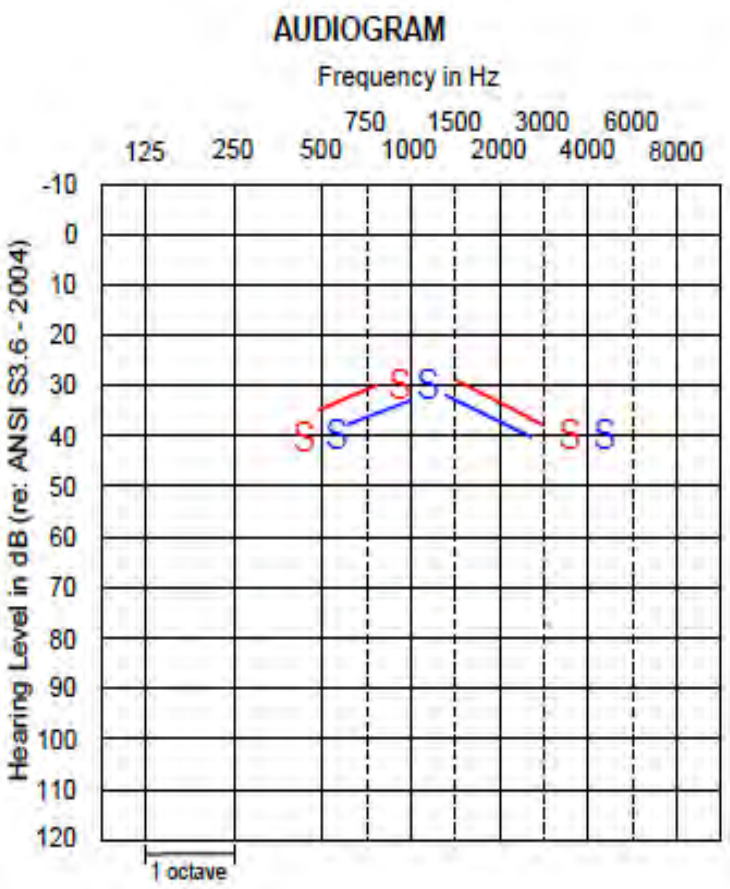


Pt. Awoke/CNT 11.7

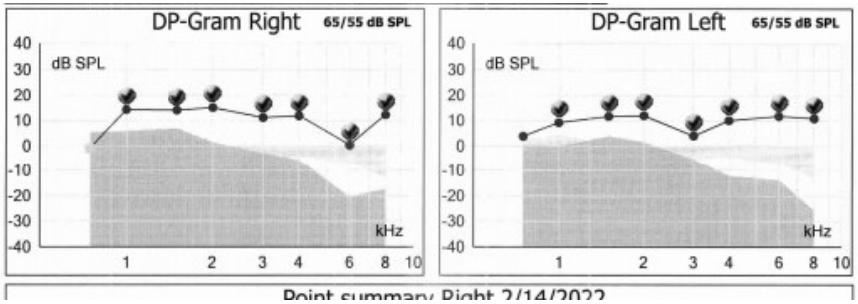
Behavioral Testing

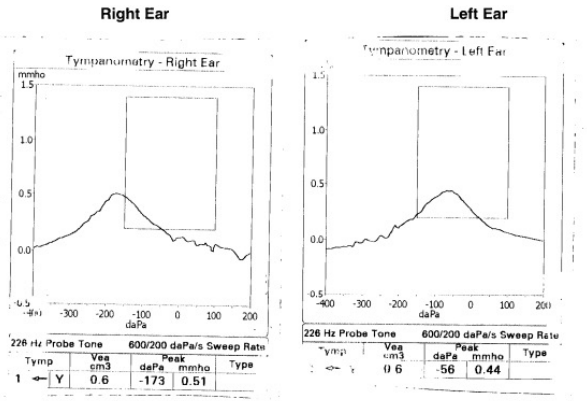
- Pt is now 8 months
  - Alert, attentive, sitting with support but good head and neck strength
  - Responds to environmental sounds
  - Responds to family members, particularly mother
  - If television is on and Ethan can't see, he will turn his head to find and see it

Test Results



*Localized both right and left
*Startle Response at 500, 1000, and 4000Hz
*Minimal Response Levels





Recommendations

- 2nd opinion Neurology consult
- Genetics
- Speech Evaluation
- Ongoing monitoring by ENT
- Ongoing monitoring by Audiology
- AZEIP services (PT, OT, SLP, Development)

2nd Opinion Neurology consult

- Sent to Genetics
- Recommended Serial MRI
- Follow-up with Audiology and ENT
- Follow-up with Ophthalmology
- Ongoing AZEIP services

Genetics Results

- Genetic panel: Hearing loss panel (possible carriership for Usher Syndrome), Heterozygous for VUS on **USH2A**, c.497 A>C, p.(E166A)-**undetermined significance**
- Invitae Cerebral Palsy panel showing a pathogenic variant and a VUS in the **GJC2 gene**. GJC2 is associated with autosomal recessive hereditary spastic paraplegia and **hypomyelinating leukodystrophy.**
- Both parents were tested GJC2 c.-167A>G (Non-coding); Heterozygous; Pathogenic; maternally inherited

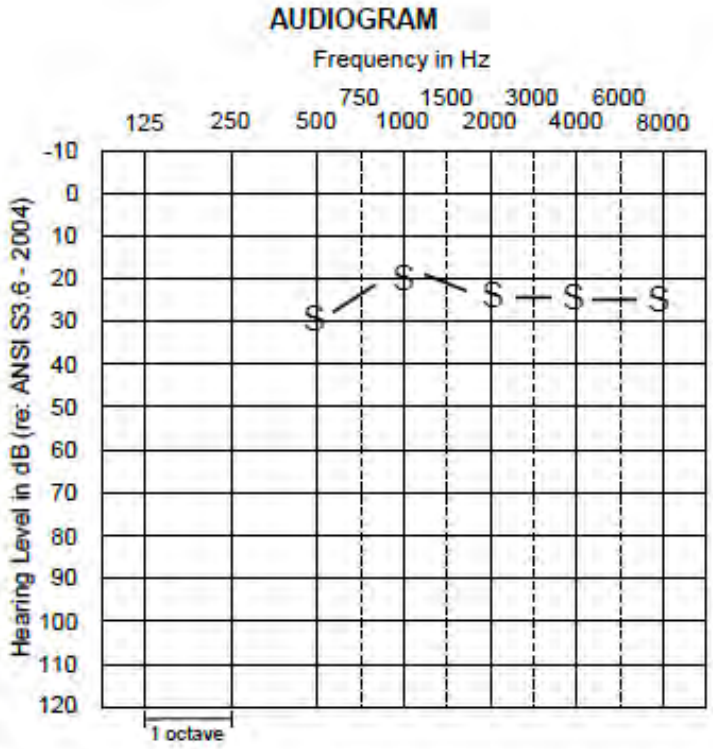
Genetics

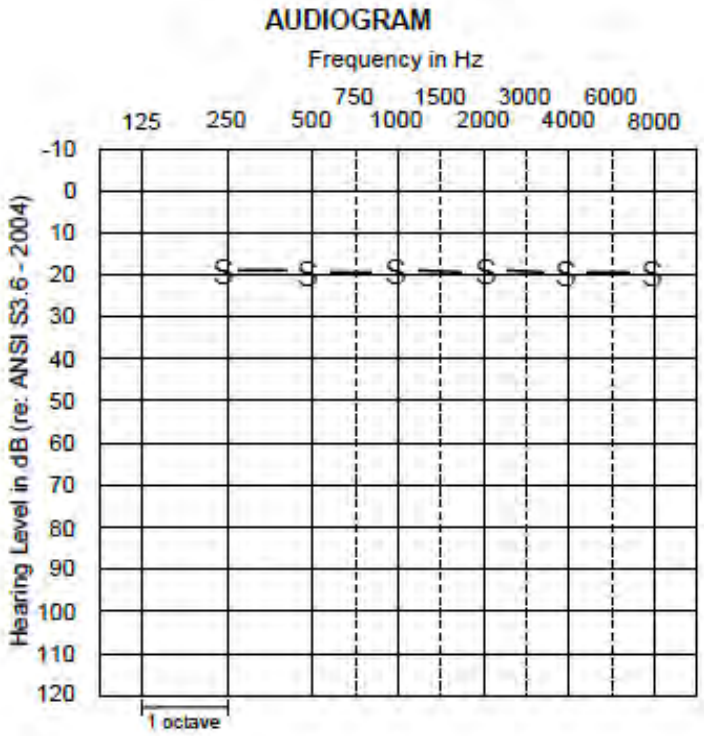
- GJC2 has been associated with **Pelizaeus-Merzbacher-like disease**/hypomyelinating
- slowly progressive leukodystrophy that typically presents during the neonatal or early-infantile period
- leukodystrophy-2 (HLD2). The presentation is usually with nystagmus, motor delays, ataxia, choreoathetotic movements, dysarthria, and progressive spasticity.
- Mild peripheral neuropathy may be present.
- Hearing loss and optic atrophy are observed in rare cases.

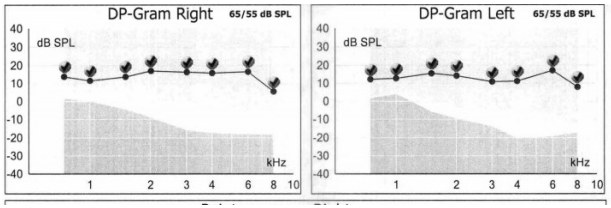
Follow-up MRI Results
18 months and 2 years

- Myelination is distinctly **abnormal** in this patient
- **Hypomyelination** in the posterior hemispheres, affecting occipital and parietal lobes, and posterior temporal lobes.
- Partial **hypomyelination of the corpus callosum**
- Predominantly posterior white matter T2/FLAIR hyperintensity and **volume loss**.

Behavioral testing over time



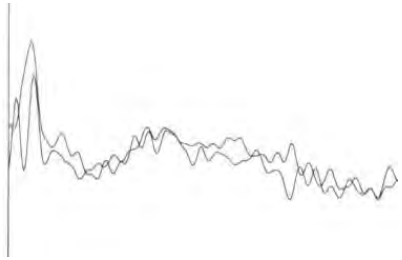


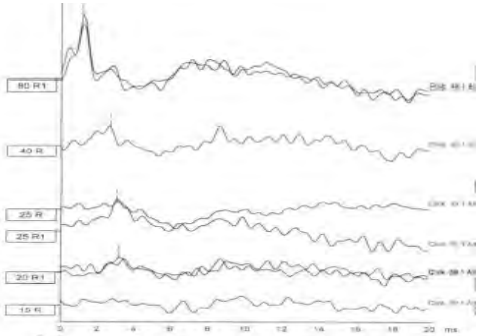


*Type A tympanograms
*Localized both right and left

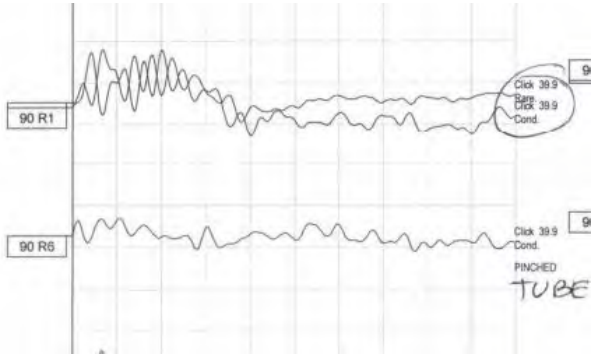
Neural Hearing Loss vs. ANSD

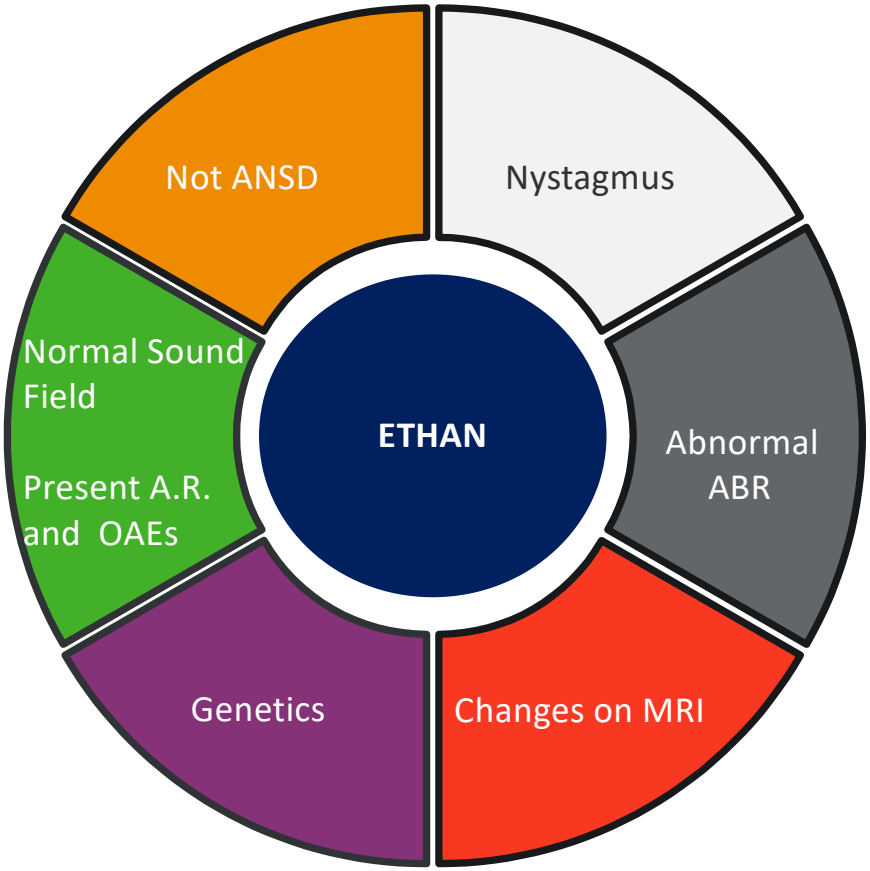
- BAER Differences
  - Neural H.L.- Wave I does not reverse with Rare and Con stimuli, may be able to trace Wave I down using lower intensity levels





  - ANSD-does reverse with Rare and Con stimuli




Not ANSD
Nystagmus
Normal Sound Field
Present A.R. and OAEs
ETHAN
Abnormal ABR
Genetics
Changes on MRI

References

- (1) Hobson GM, Garbern JY. Pelizaeus-Merzbacher disease, Pelizaeus-Merzbacher-like disease 1, and related hypomyelinating disorders. Semin Neurol. 2012 Feb;32(1):62-7. doi: 10.1055/s-0032-1306388. Epub 2012 Mar 15. PMID: 22422208.
- (2) Nahhas N, Conant A, Orthmann-Murphy J, et al. Pelizaeus-Merzbacher-Like Disease 1. 2017 Dec 21 [Updated 2019 Jan 17]. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2023. Available from: https://www.ncbi.nlm.nih.gov/books/NBK470716/

Thank you!

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