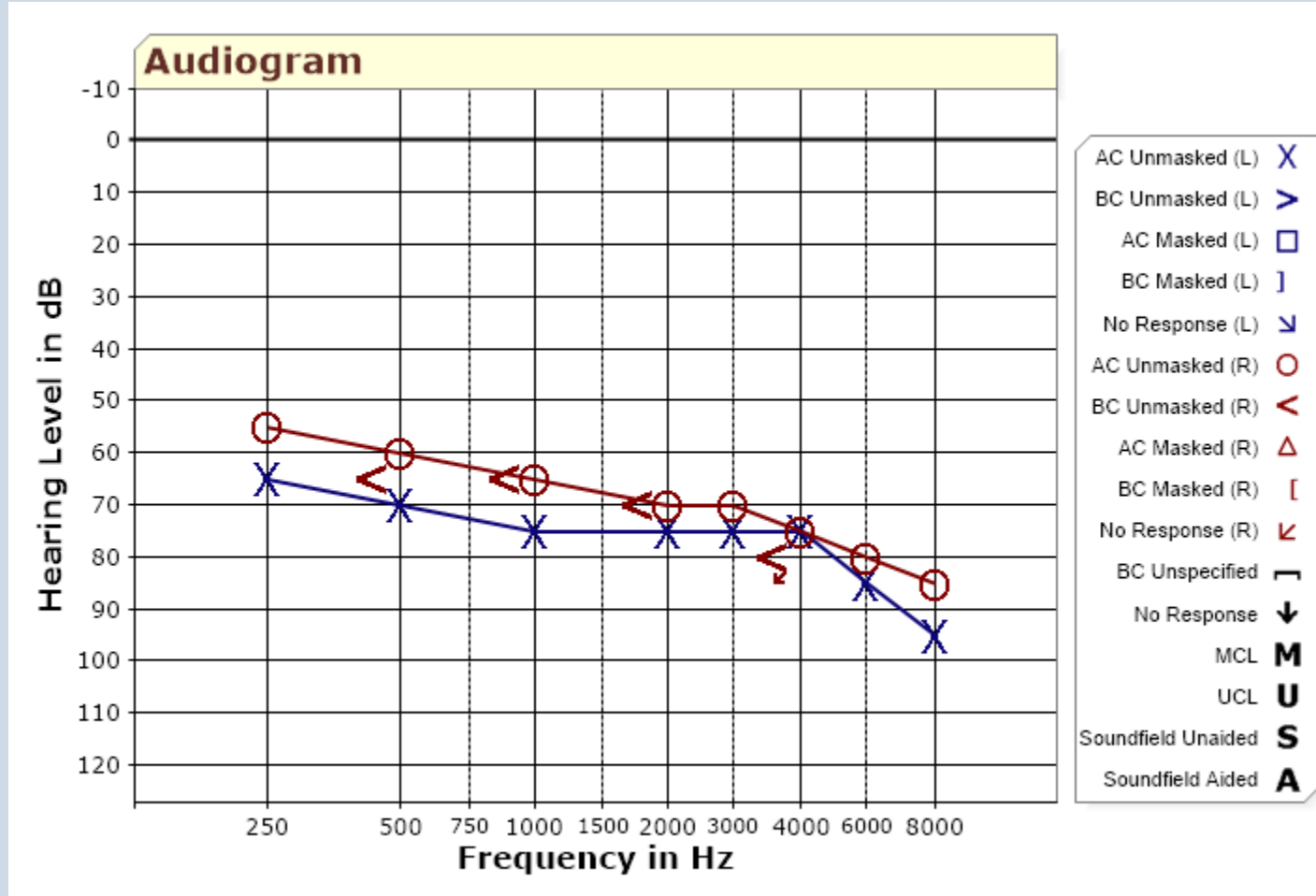

UNEXPECTED SURPRISE

Jana Brown, AuD

PATIENT PRESENTATION

- Pt. presented to clinic with his wife
- Stroke in 1998 resulted in
 - *Imbalance with episodes of vertigo lasting minutes*
 - *Right paresis*
 - *Difficulty speaking*
 - *Expressively aphasic*
- Financial difficulties

AUDIOGRAM



WORD RECOGNITION

- Mono-syllabic picture boards were used for testing
- Pt was unable to point to the correct picture frequently when using monitored live voice
- Pt became very frustrated with this task and teared up several times
- His word recognition was poor AU with auditory cues only
- When the patient was allowed to lipread, he was able to point to the correct pictures about a third of the time.

- Pt. not surgical candidate for CI due to cardiovascular issues
- Wife said he would not use amplification
- Obvious great difficulty communicating due to aphasia and hearing loss

Sooooo...

- Counseled pt extensively on importance of hearing and stimulating his brain
- Agreed to EMIs
- Pt. and our office looked for financial help - None found
- Made appointment for “follow up” Hee hee!

- The following video is not an endorsement
- We use multiple hearing aid manufacturers

“FOLLOW UP” APPOINTMENT



REAL “FOLLOW UP”

- Pt. has done well but had to increase gain slowly due to length of time with no auditory stimulation
- Still doing well but health is declining rapidly now
- Pt. and family are extremely grateful for our help.
- Great reminder of why we do what we do!

- Thank you!



Kentucky Sound
Foundations

Benefits of Auditory Training in patients with Auditory Processing Disorder

Casey Rutledge Roof, AuD., CCC-A
Audiologist/Owner
Kentucky Sound Foundations

Objectives

- The listener will understand who may benefit from auditory training
- The listener will observe several elements and programs within auditory training to target specific listening skills



Financial Disclosures

- No financial disclosures
- Sole owner, LLC

The what, why, who, and how

- What is auditory training?
 - Training of auditory discrimination
 - Hearing and tolerance in noise
 - Expanding short-term verbal memory capacity
 - Musical discrimination of tones, contour, rhythm, instruments and chord trains
 - Under the larger umbrella of Aural Rehabilitation
 - Auditory training is for people with hearing loss and/or auditory processing difficulties



The Aural Rehabilitation Umbrella



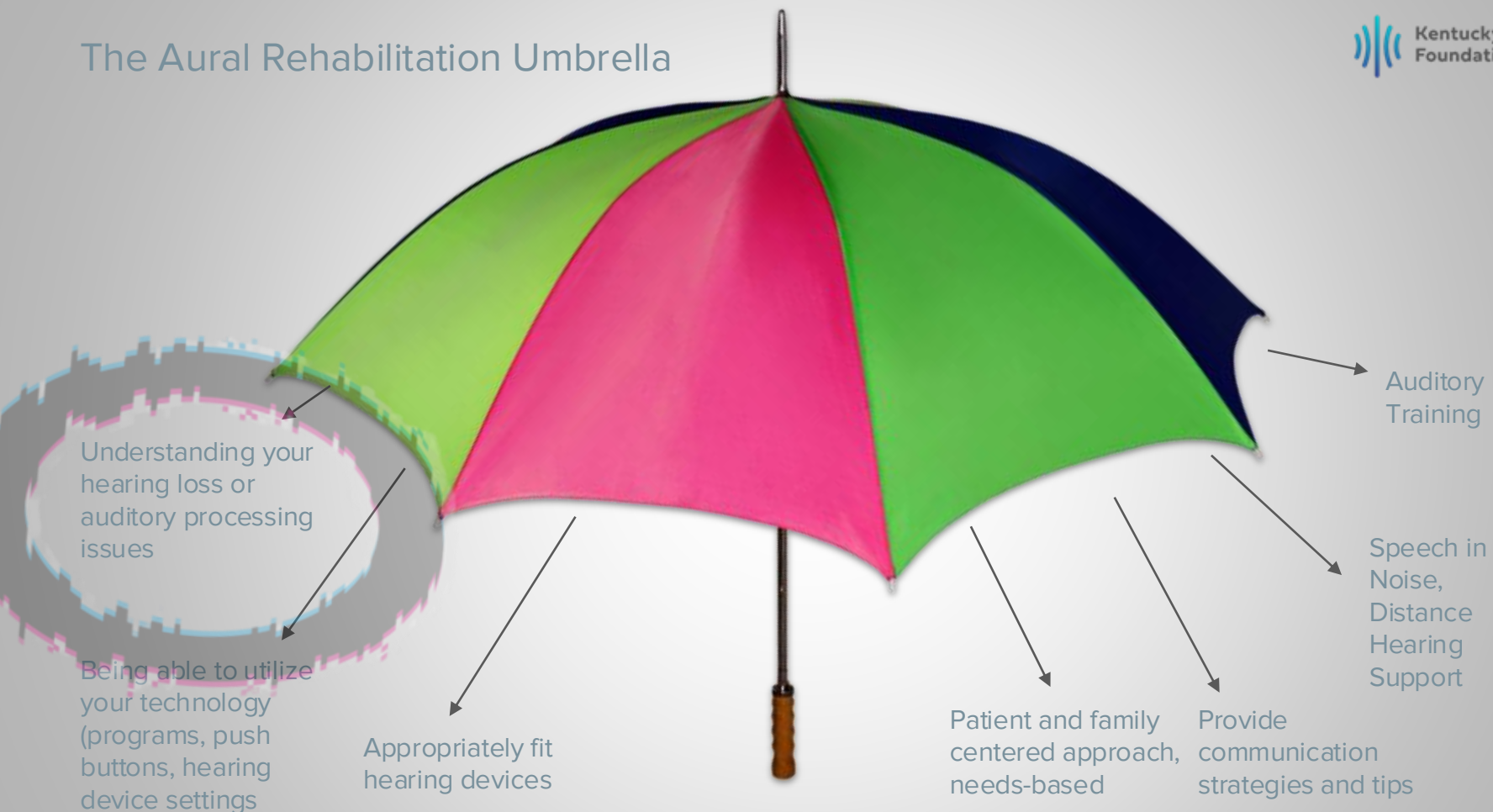
The Aural Rehabilitation Umbrella



The what, why, who, and how

- Why would you refer patients for auditory training?
 - Overall improvement in objective scores and functional outcomes relative to their personal COSI goals and questionnaires
 - Increase confidence in auditory abilities
 - Reading and spelling
 - Improve auditory processing abilities
 - Can help the patient with hearing loss conceptualize where their listening difficulties may arise, relate to current performance issues/complaints, and how to improve performance with devices

The Aural Rehabilitation Umbrella



The what, why, who, and how

- Who would you refer for auditory training?
 - Those suspected to have auditory processing issues (that would be evaluated)
 - Patients with hearing loss and complaints regarding discrimination or hearing in noise that are not ready to pursue amplification
 - Patients struggling, even with appropriately-fit hearing technology/cochlear implantation, etc.
 - Non-descriptive hearing complaints

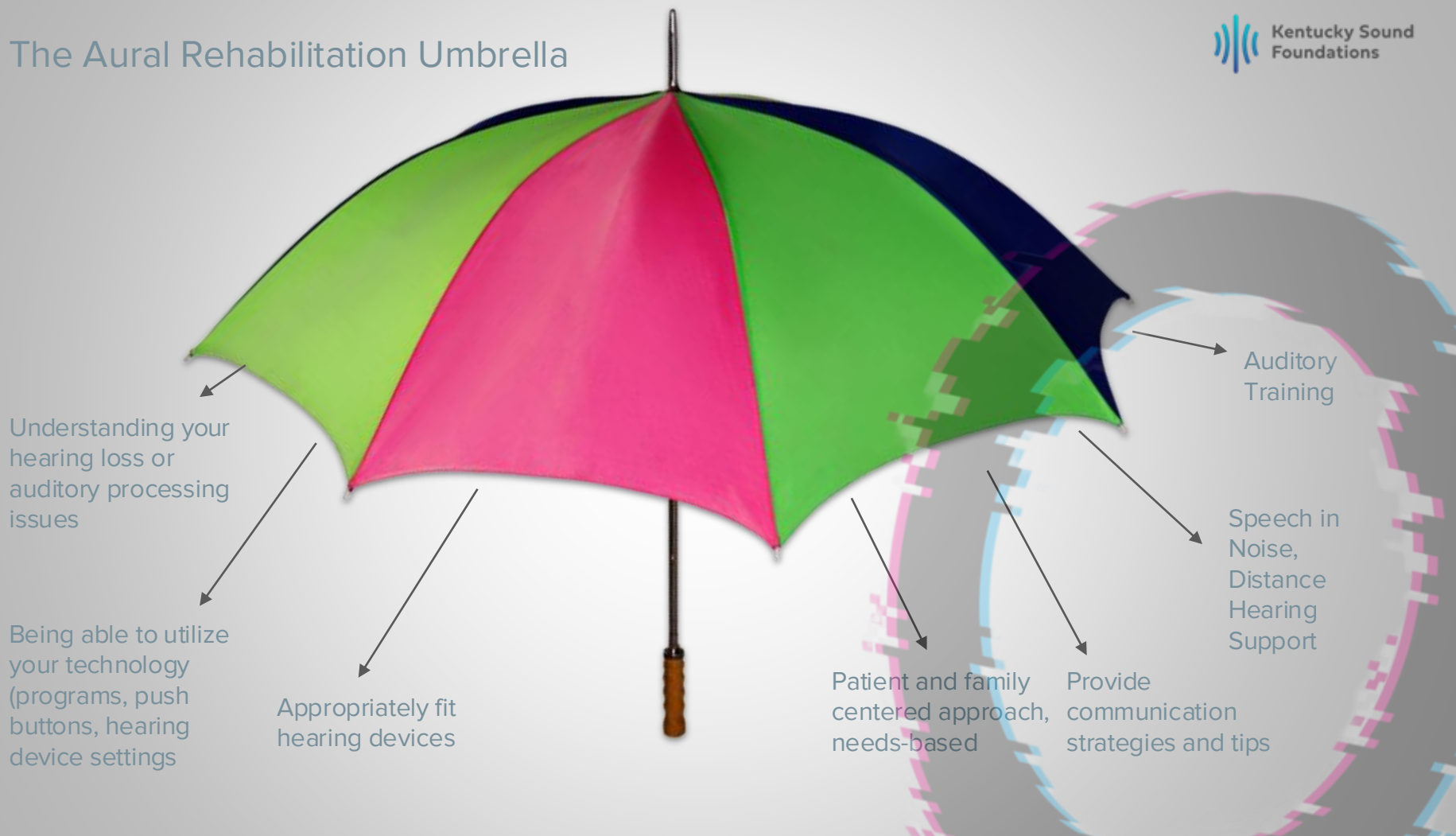
The Aural Rehabilitation Umbrella



The what, why, who, and how

- How do you complete auditory training?
 - Program is formulated from results of the evaluation and completed in rounds of therapy
 - Most people need 1.5-2 rounds of therapy
 - Teletherapy completed once or twice per week

The Aural Rehabilitation Umbrella



The how

- Formulating a therapy plan
 - Buffalo Model Framework
 - Bellis-Ferre Model
 - Neurophysiological Model



Patient 1: 31 year-old male

Subjective:

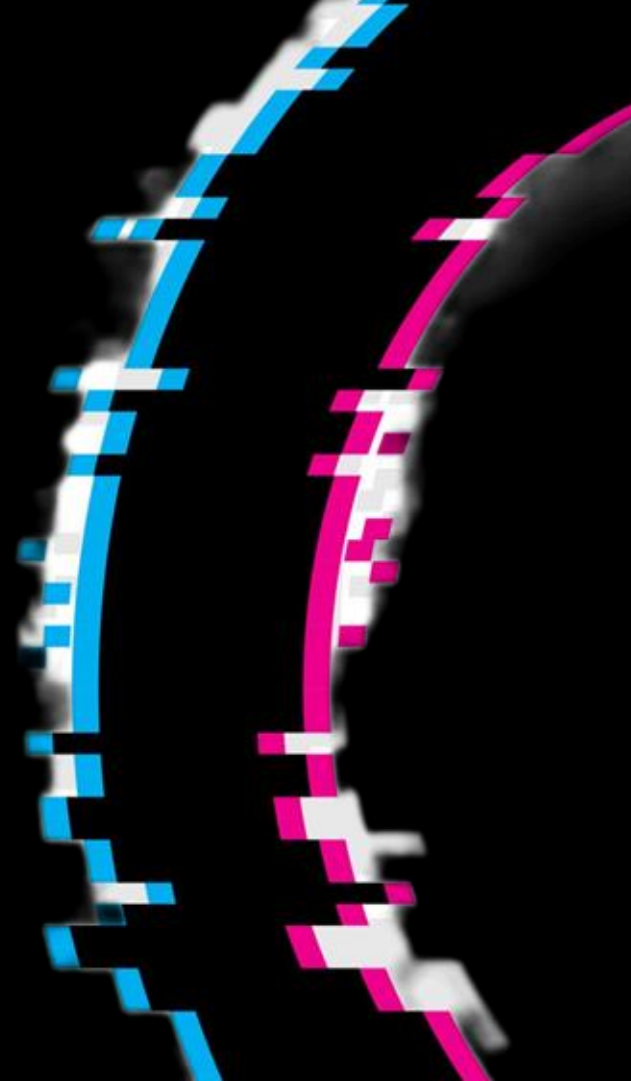
- Issues understanding and hearing verbal directions
- Difficulty maintaining auditory attention
- Trouble following conversations, especially in background noise
- Socially frustrated and withdraws from larger social situations aside from family
- Trouble spelling
- Relies on visual cues, supports, etc.
- Difficulty hearing and understanding work conference calls
- Modified work environment (home office)



Patient 1: 31 year-old male

Subjective:

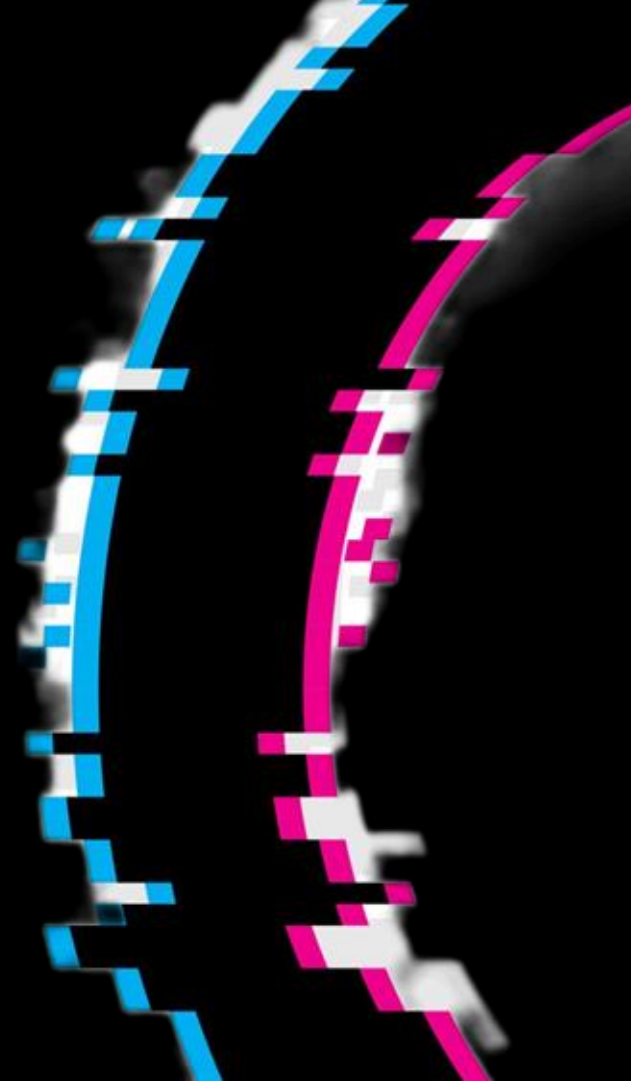
- Tinnitus bilaterally
- Generally clumsy
- Describes himself to have “auditory dyslexia”
- Issues with depth perception
- Asymmetric vision (left eye > than right eye)
- Right-handed
- Diet-controlled DMT2



Patient 1: 31 year-old male

Pre-Test Findings

- Asymmetric hearing thresholds, mild HF SL R, borderline normal hearing LE
- SRT 20dB HL R, 10dB HL L
- Fit with bilateral Phonak Audeo Sphere Infinio RIC hearing devices



Patient 1: 31 year-old male

Test Findings

W22	Right Ear	Left Ear	Binaural	RNC	RC	LNC	LC
Quiet	92%	92%					
Noise	36%	44%					
Phonemic Synthesis Test			32%				
SSW				Abn	Abn	Abn	Abn

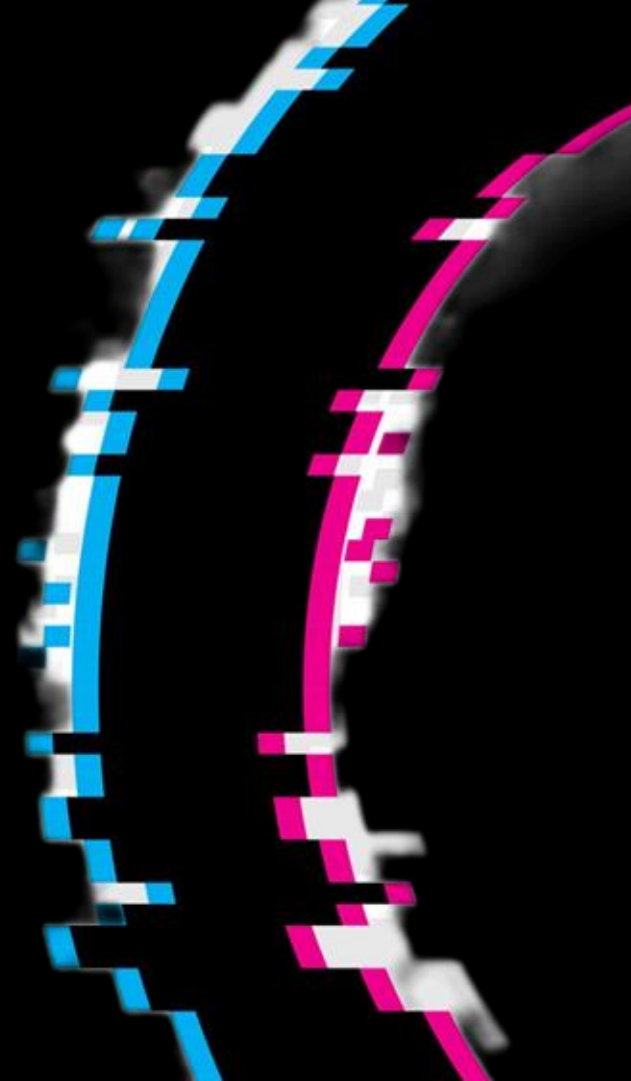
Patient 1: 31 year-old male

DigiSpan	Forward recall below normal limits	Reverse recall at the bottom range of normal	
DDdT	Normal dichotic advantage, RE advantage (free recall and directed), directed advantage	Diotic- low end of normal	Dichotic Free recall (R/L, total)-abnormal Dichotic Directed (L)- abn
LiSN-S	Talker, Spatial, Total Advantage Score WNL	Abnormal speech in noise scores not attributed to spatial processing disorder	
SCAN-A	Time Compressed Sentences- abn	Filtered Words- borderline WNL	Competing Sentences- ABN

Patient 1: 31 year-old male

Conclusions

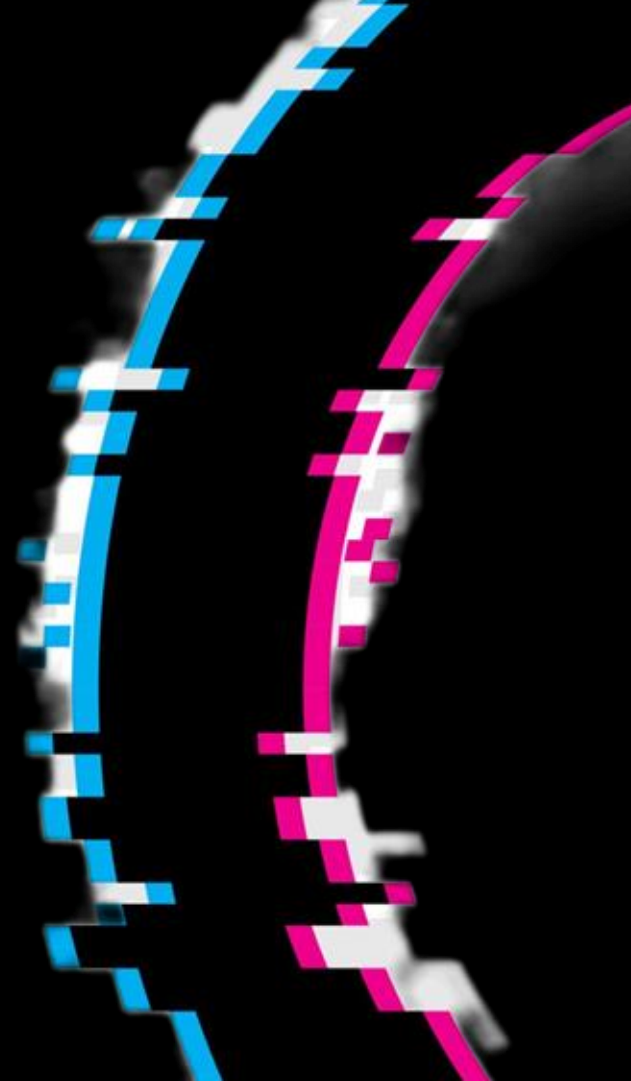
- Asymmetric hearing (R poorer than L)
- Decoding, Tolerance Fading Memory, Organizational Issues within Buffalo Model Framework/categorization
- Negative for Spatial Processing Disorder
- >2 SD across several tests within the test battery
- Would benefit from auditory training and strategies/accommodations



Patient 1: 31 year-old male

Treatment Plan

- Auditory training to facilitate proper encoding of the auditory stimulus (phonemic training)
- Auditory memory, Noise Tolerance and improved speech understanding in noise
- Improved auditory organizational abilities
- 15 weeks, 1x per week



Patient 1: 31 year-old male

Phonemic Training Program

- Each session focuses on a subset of phonemes
- Repetition, repetition, repetition

Auditory Memory/Speech in Noise

- Scaffolding digits, words, phrases, sentences
- Words in Noise Training, Other Speech in Noise trainings

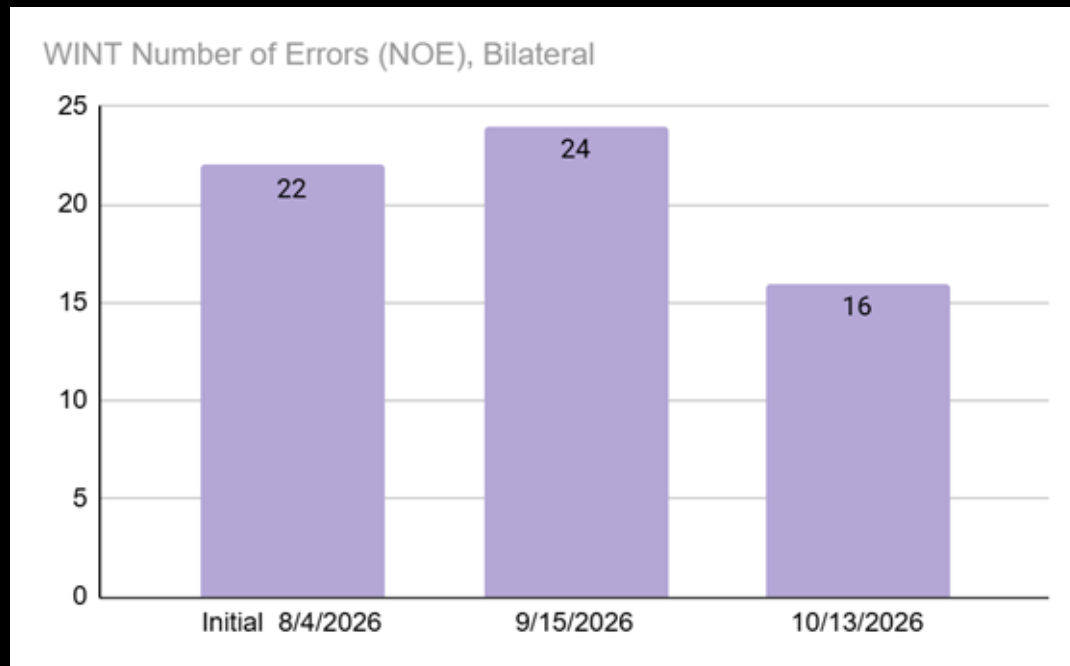
Skills Remediation

- Auditory closure
- Phonemic manipulation, substitution
- Automaticity

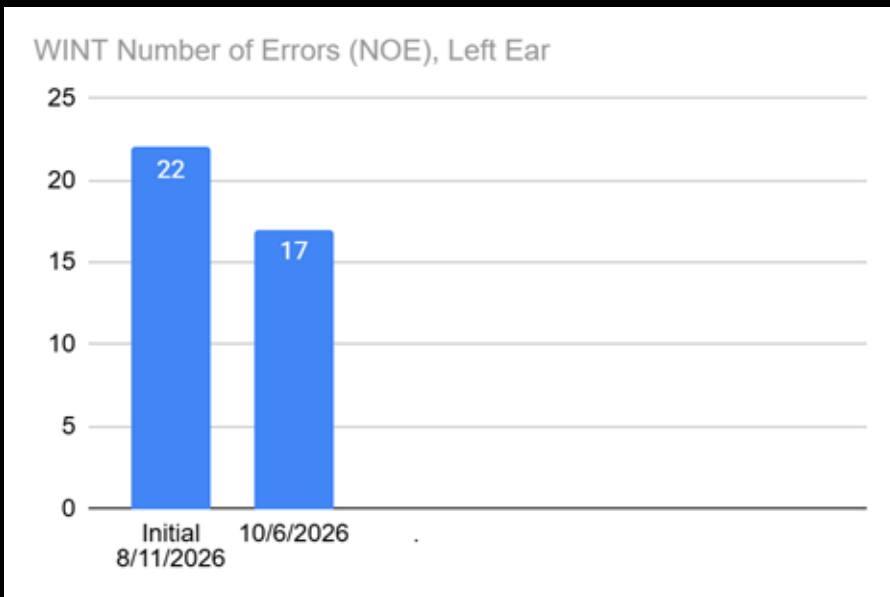
Auditory Training Schedule



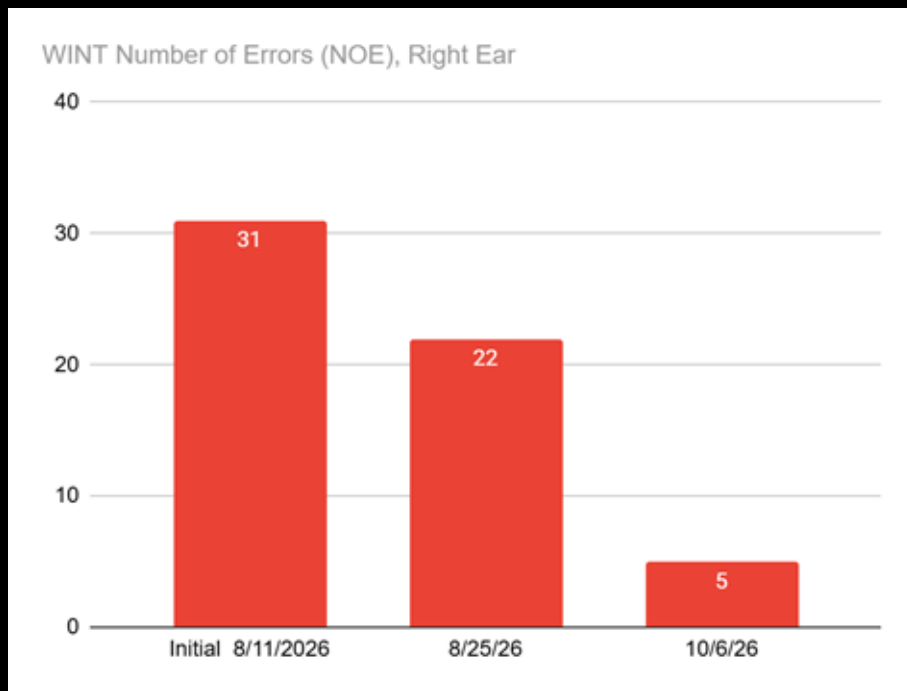
Patient 1: 31 year-old male



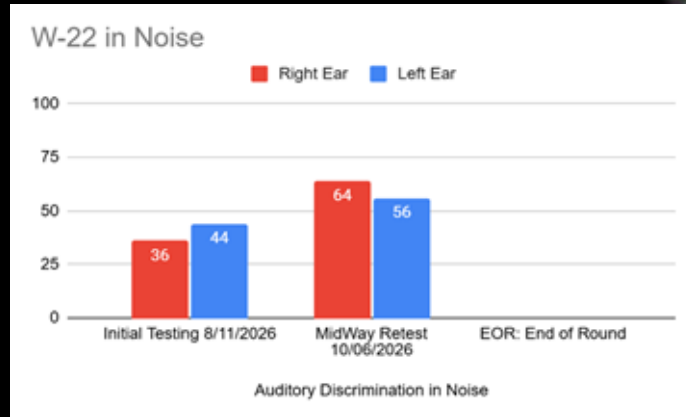
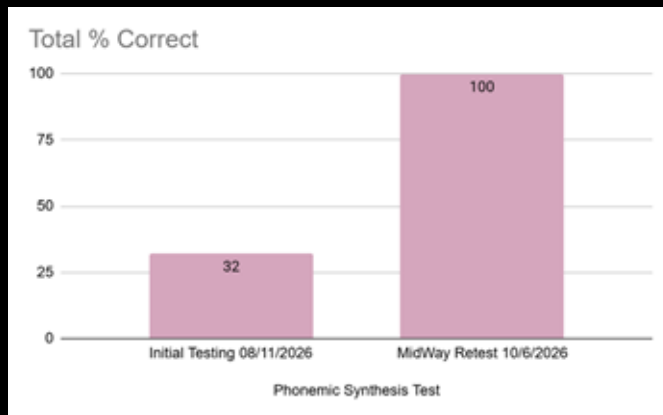
Patient 1: 31 year-old male



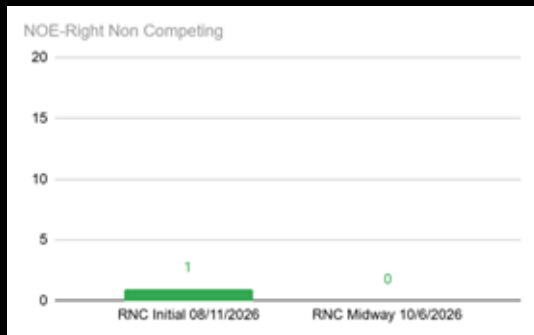
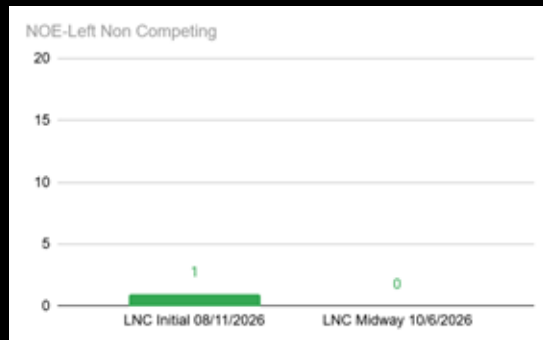
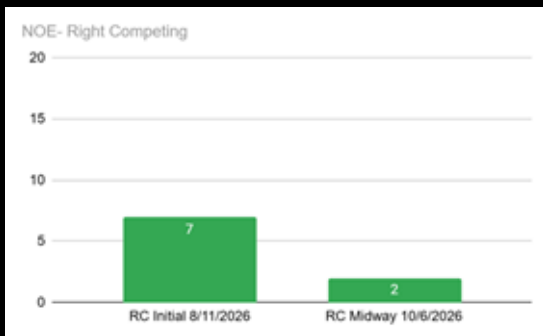
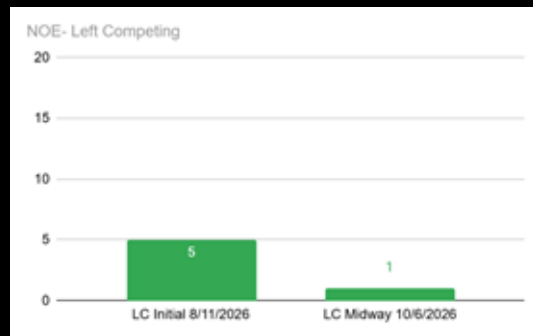
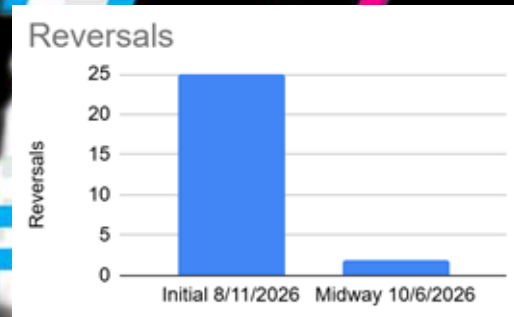
Patient 1: 31 year-old male



Patient 1: Retest 1 (midway)



Patient 1: Retest 1 (midway)-SSW



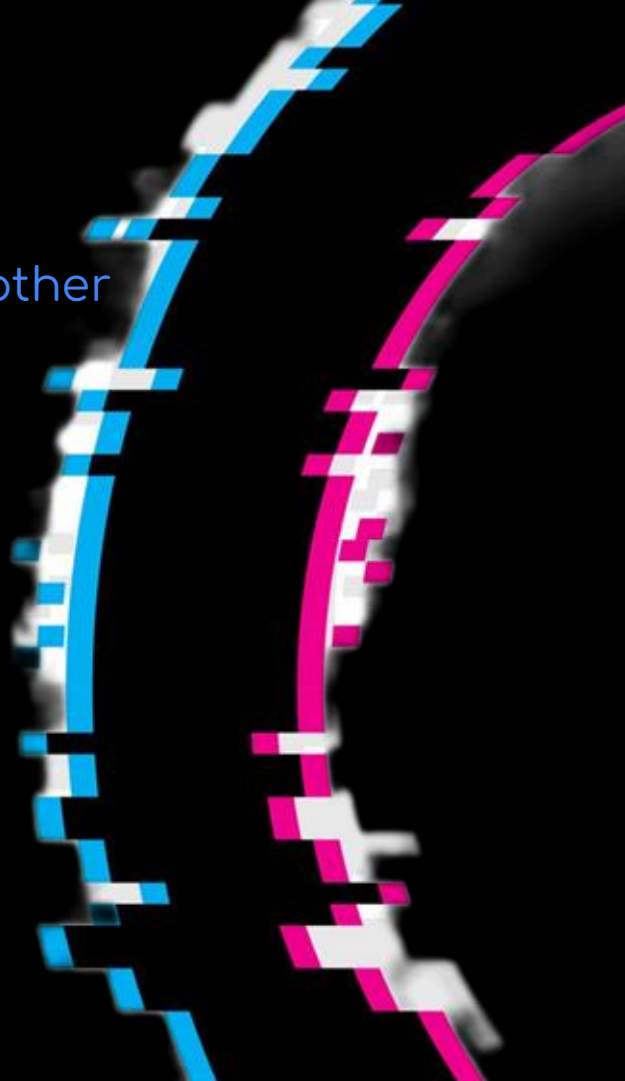
Patient 1: Therapy Plan revision

- Will finish up phonemic training
- Dichotic training
- Musical training

Patient 2: 7 year-old male

Subjective:

- Patient previously diagnosed with CAPD by another provider in 2024
 - Speech in Quiet abn left ear
 - 92% right ear, 76% left ear
 - Speech in Noise abn AU-
 - 52%
 - Phonemic Synthesis -
 - 1 SD below normal
 - Competing Sentences
 - Abn R, normal L
 - Low Pass filtered speech
 - Abn AU



Patient 2: 7 year-old male

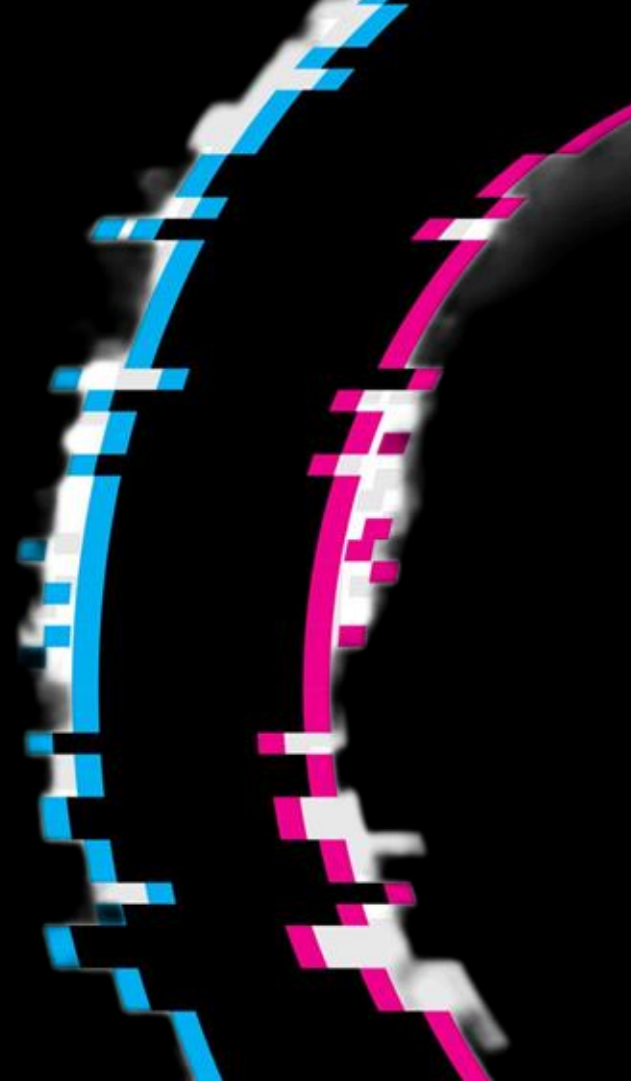
Eval,

- SSW
 - Normal R/L NC
 - Abn R/L C
- TAPS
 - Abn # memory forward, word memory, sentence memory
- Screening of phonological skills
 - Abn rhyming
 - Syllable and phonemic deletion
 - Blending syllables and phonemes

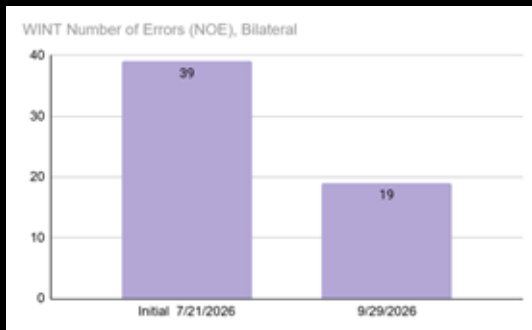
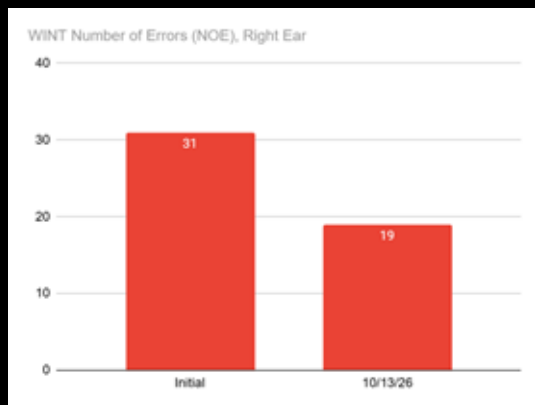
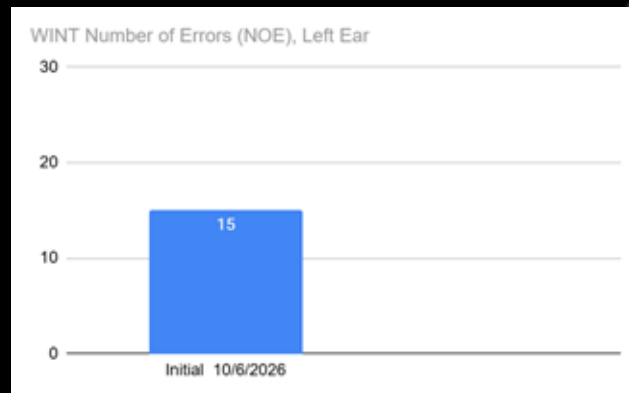


Patient 2: 7 year-old male

- Plan
 - Phonemic training
 - Auditory attention
 - Phonemic deletion, substitution via auditory prompts
 - Auditory vigilance training
 - Words in Noise
 - Temporal processing training
 - Dichotic training
 - Auditory-Visual Integration (IM)



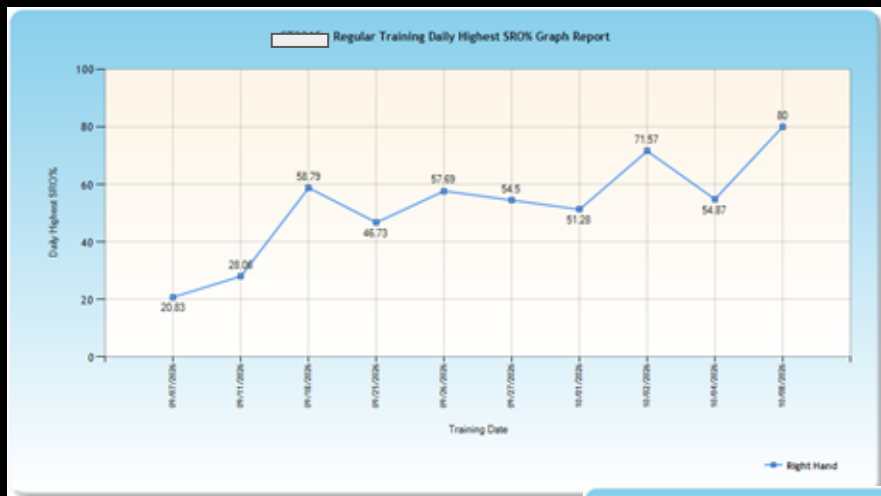
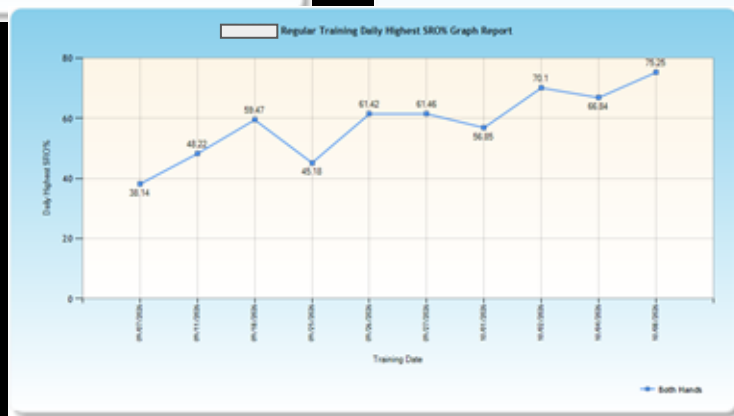
Patient 2: 7 year-old male



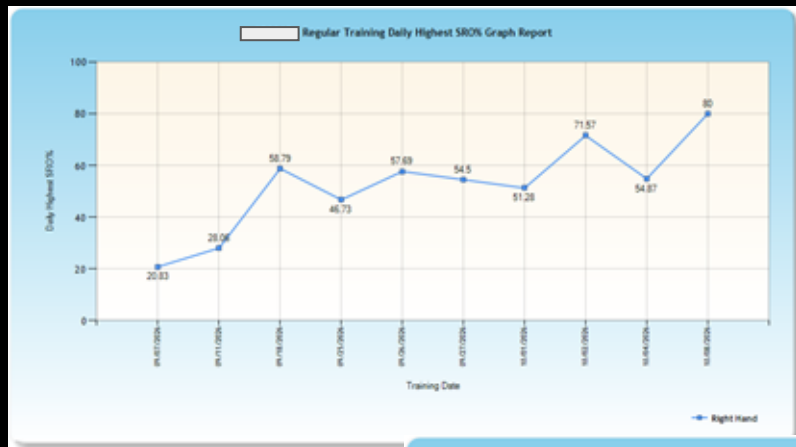
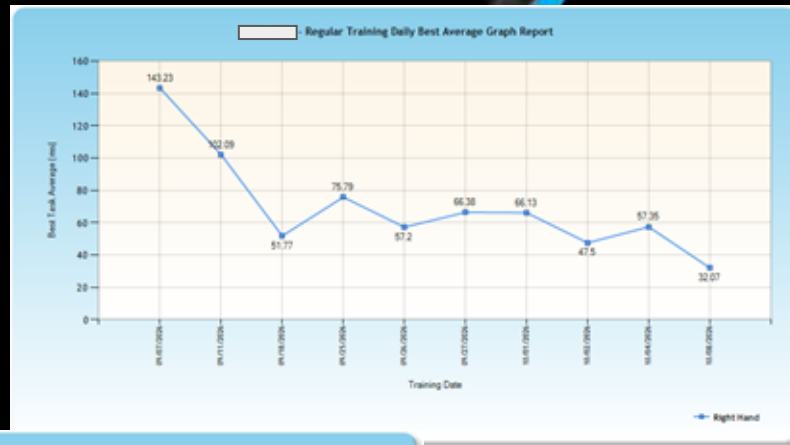
1

[illegible]

Patient 2: 7 year-old male



Patient 2: 7 year-old male



Thank you!

www.kentuckysoundfoundations.com

A decorative graphic at the bottom of the slide featuring two curved, pixelated lines. The upper line is blue and the lower line is pink, both with a jagged, digital edge effect.

Casey Rutledge Roof, AuD., CCC-A
Audiologist/Owner
caseyroof@kentuckysoundfoundations.com
502-401-1616

When Genes Don't Tell the Whole Story:

A Tale of 2 Brothers



dreamstime.com

ID 20633546 © Anna Velichkovsky

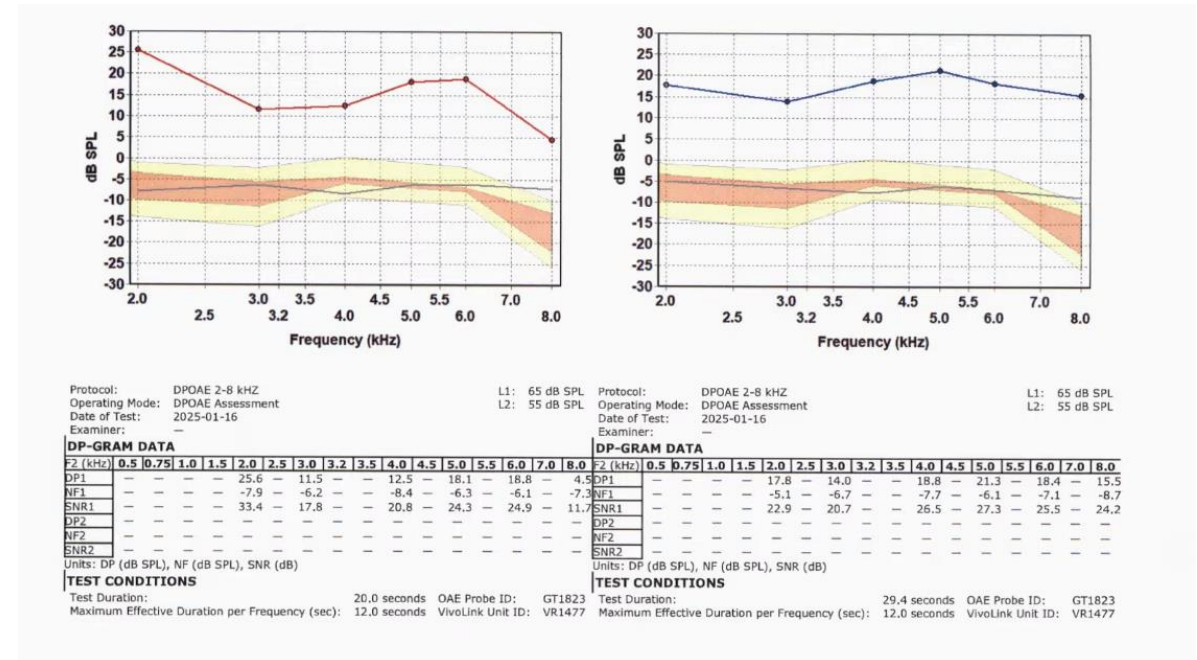
Newborn hearing screening

- Born 39 weeks gestation-mother with gestational diabetes but no other risk factors
- Failed hearing screening twice on each ear in hospital; CMV screening was negative
- Seen in Audiology at 14 days of life for diagnostic hearing evaluation



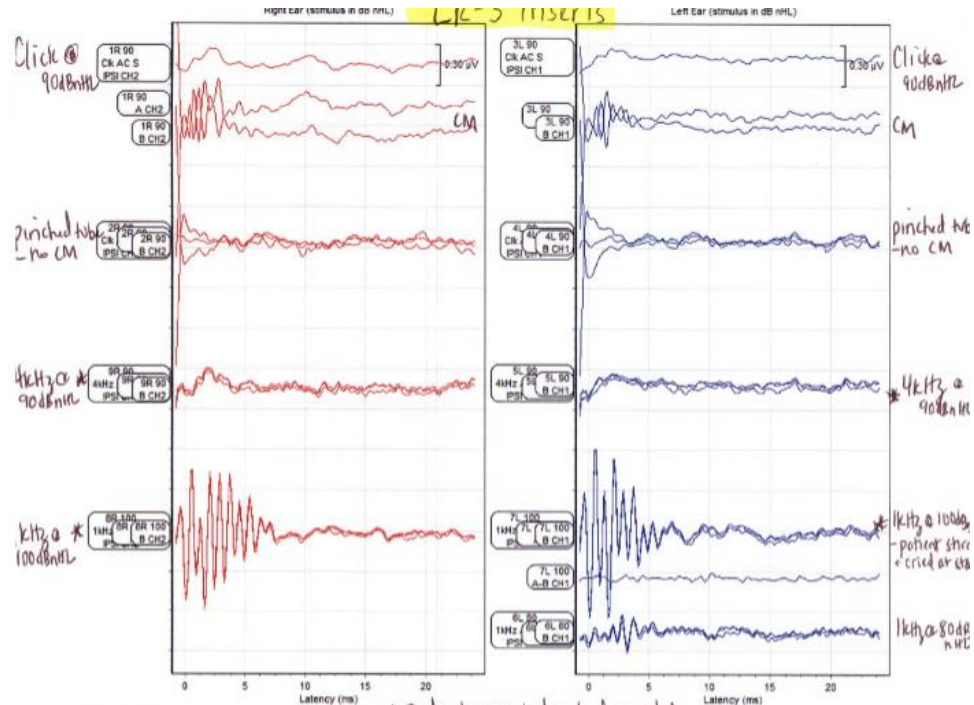
First Follow Up – 2 weeks of age

- Mom reports no significant medical history
- Brother failed NBHS when he was born but had normal follow-up diagnostic eval at another hospital
- Normal tympanometry
- Robust OAEs



.....A good start!

First Follow Up



ABR WAVEFORM DATA

* Did not correct due to degree hL.

Corr. Coef.	RN (µV)	SNR	Time (ms)												Amp (µV)		Ratio		BS	G													
			I	F	II	III	IV	V	V'	VII	III-V	IV-V	V'	V-V'	V-V'/[I-V]																		
1 R 90 CH2	0.065	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
2 R 90 CH2	0.029	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
3 L 90 CH1	0.046	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
4 L 90 CH1	0.029	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
5 L 90 CH1	0.013	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
6 L 90 CH1	0.013	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
7 L 100 CH1	0.013	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
8 R 100 CH2	0.011	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
9 R 90 CH2	0.010	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—

Corr. Coef. = Correlation Coefficient, RN = Residual Noise

KEY TEST CONDITIONS

Protocol	Stimulus Level	Stimulus Type	Stimulus Rate (Hz)	Polarity	Mask Level	* Noise Adj. Score
1 R R ABR AC Click 37.7	90 dB nHL	click	37.7	Alt.Split	—	CH1:5432
2 R R ABR AC Click 37.7	90 dB nHL	click	37.7	Alt.Split	—	CH1:5534
3 R L ABR AC Click 37.7	90 dB nHL	click	37.7	Alt.Split	—	CH1:2257
4 R L ABR AC Click 37.7	90 dB nHL	click	37.7	Alt.Split	—	CH2:2266
5 R L ABR AC 4000 Hz TB 37.7	90 dB nHL	4000 Hz	37.7	Rare/fac.	—	CH1:5636
						CH2:5666
						CH1:2430
						CH2:2543
						CH1:2808
						CH2:2767

* Using the SOAP-Kalman Weighted algorithm, 2000 "noise adjusted sweeps" is comparable to 2000 "accepted sweeps" used in conventional averaging.

- Abnormal ABR with no synchronous waveforms
- Cochlear microphonic present
- Again, reviewed case history with mom and no risk factors for AN reported

Minimal Response Levels (dB eHL)

ABR minimal response levels are an estimate of peripheral hearing sensitivity and can be expected to be within +/- 5 dB of behavioral thresholds.

	1000 Hz	4000 Hz
Right AIR	NR at 100dB eHL	NR at 90dB eHL
Left AIR	NR at 100dB eHL	NR at 90dB eHL

ABR Summary:

Right Ear: Present cochlear microphonic with absent waves - results consistent with ANSD

Left Ear: Present cochlear microphonic with absent waves - results consistent with ANSD

DPOAE (2000-8000 Hz)

DPOAE Equipment: Vivosonic

Right ear: Present at all test frequencies

Left ear: Present at all test frequencies

Present OAEs indicate normal cochlear outer hair cell function.

SUMMARY

- Today's test results indicate a sensorineural hearing loss in both ears. Results are consistent with bilateral ANSD.
- This is likely a communicatively-significant hearing loss. However, behavioral testing and parental feedback regarding observed responses to sounds are recommended prior to any treatment recommendations.

RECOMMENDATIONS

1. Repeat ABR Evaluation in 1-2 months. This was scheduled on 2/20/25.
2. Referral to Ear & Hearing Center. (Family will be contacted to schedule)

Causes of ANSD

Most commonly at birth or shortly after birth.

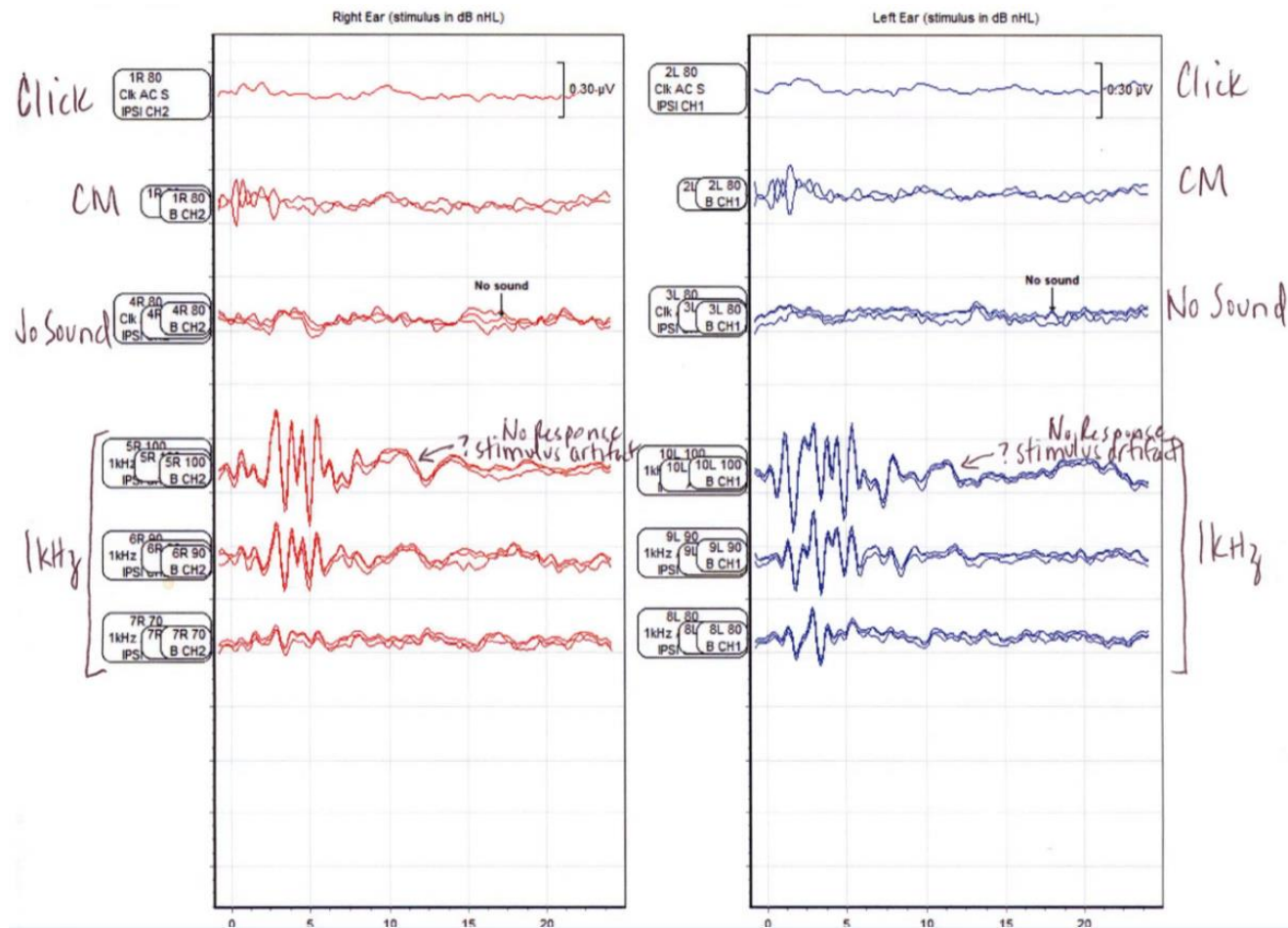
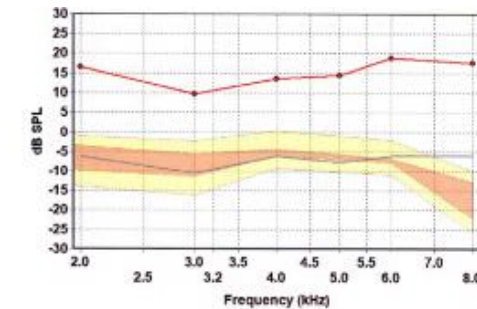
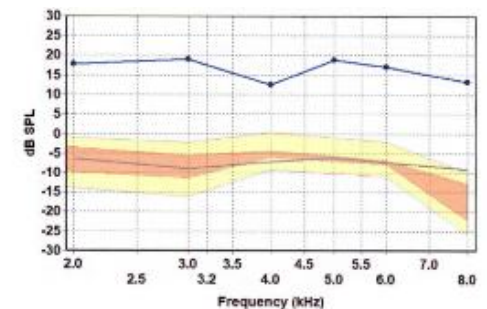
Common causes at birth:

- Premature birth
- Hyperbilirubinemia
- Genetics
- Neurological conditions

- Prevalence among hearing impaired populations is reported as 0.5 to 15%
 - Davis and Hirsh (1979)-0.5%
 - Krause et al. (1984)-15%
 - Rance et al. (1999)-11%
- 30% of patient with ANSD will have another disability/diagnosis

Second Follow Up – 6 weeks of age

Mother reports no concerns: startles and turns head to sounds



Protocol: DPOAE 2-B kHz
Operating Mode: DPOAE Assessment
Date of Test: 2025-02-17
Examiner: —

L1: 65 dB SPL
L2: 55 dB SPL

Protocol: DPOAE 2-B kHz
Operating Mode: DPOAE Assessment
Date of Test: 2025-02-17
Examiner: —

L1: 65 dB SPL
L2: 55 dB SPL

DP-GRAM DATA

F2 (kHz)	0.5	0.75	1.0	1.5	2.0	2.5	3.0	3.2	3.5	4.0	4.5	5.0	5.5	6.0	7.0	8.0
DP1	—	—	—	16.5	—	9.6	—	13.5	—	14.3	—	18.9	—	17.0	—	13.2
NF1	—	—	—	-6.1	—	-10.5	—	-6.2	—	-7.8	—	-8.1	—	-6.1	—	-9.1
SNR1	—	—	—	22.6	—	20.0	—	19.6	—	22.1	—	25.0	—	23.8	—	22.3
DP2	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
NF2	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—
SNR2	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—	—

Units: DP (dB SPL), NF (dB SPL), SNR (dB)

TEST CONDITIONS

Test Duration: 13.5 seconds OAE Probe ID: GT1823
Maximum Effective Duration per Frequency (sec): 12.0 seconds VivoLink Unit ID: VR1477

Test Duration: 19.6 seconds OAE Probe ID: GT1823
Maximum Effective Duration per Frequency (sec): 12.0 seconds VivoLink Unit ID: VR1477

MRI Results

PROCEDURE COMMENTS: MRI of the brain and IACs was performed without intravenous contrast.

FINDINGS:

The ventricles and extra-axial spaces are normal in size and configuration.
There is no mass or evidence of intracranial hemorrhage.
Parenchymal signal and morphology are normal.
There are normal flow-voids in the intracranial vessels.

Dedicated imaging of the posterior fossa and internal auditory canals demonstrates no additional abnormalities.

The cochlea, vestibule, and semicircular canals appear normal.
There is no lesion impinging upon cranial nerves VII and VIII.
There are no foci of diffusion restriction.

IMPRESSION

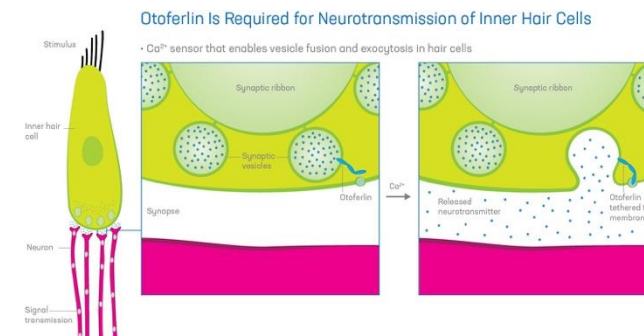
Normal exam.

Genetics Results

Genetic Significance

Gene	HGVS	Zygosity	OMIM Phenotype	Classification
OTOF	Chr2(GRCh37):g.26681086C>T NM_001287489.2:c.5816G>A p.(Arg1939Gln)	Heterozygous	Auditory neuropathy, autosomal recessive, 1 (AR) Deafness, autosomal recessive 9 (AR)	Pathogenic
<p>HGMD: CM033480 ClinVar: 548986 (Pathogenic) NCBI: rs201326023 Global gnomAD: 0.004 % Max gnomAD: 0.027 % In-silico predictions: suggest a damaging effect on protein function</p> <p>This variant is also known as NM_194323.3:c.3515G>A (p.Arg1172Gln) in the literature. It has been reported in the homozygous and compound heterozygous states in multiple individuals with auditory neuropathy or nonsyndromic hearing loss (PMID 16097006; PMID 14635104; PMID: 24053799; PMID: 26632695; PMID: 22575033; PMID: 34424407; PMID 34097718; PMID 34536124; PMID 34403091; PMID: 34335733; PMID: 35248088). This variant has been classified pathogenic by other clinical laboratories (ClinVar ID: 548986).</p>				

- Otoferlin
 - Otoferlin is expressed in inner hair cells
 - Often causes profound hearing loss in impacted patients
 - Non-syndromic recessive deafness



https://www.audiology.org/wp-content/uploads/2021/04/AT333_Main_Images_OnTrends-DecibelTherapeutics-min.jpg

What does this mean?

The important finding

- The detected OTOF variant has been well documented in the literature and is considered **disease-causing (pathogenic)**. However, the key point is that the individual has **only one pathogenic variant identified** ("heterozygous").

OTOF-related hearing loss is autosomal recessive

- OTOF-related hearing loss and auditory neuropathy are inherited in an **autosomal recessive** manner.
 - This means a person typically needs **two pathogenic OTOF variants** (one inherited from each parent) to be affected by classic OTOF-related hearing loss.



Determining the cause of the ANSD helps direct treatment plan and cause of ANSD.

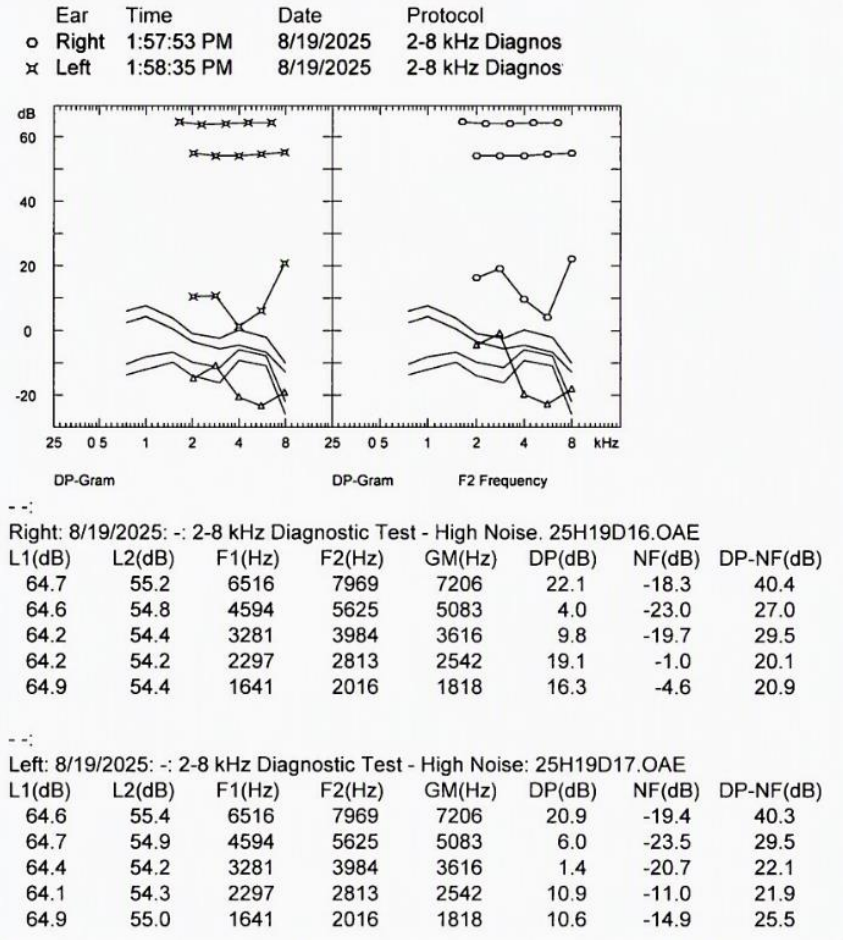
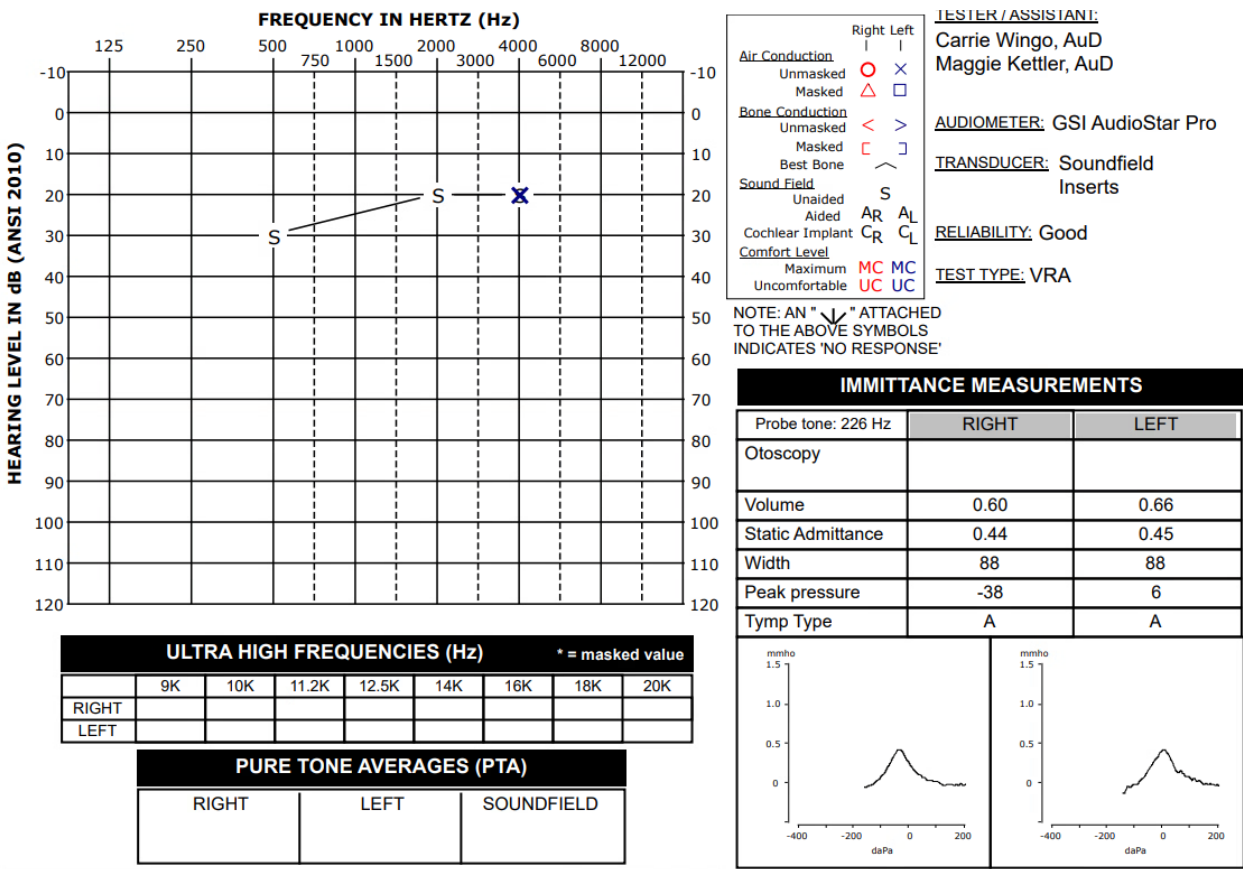


Patients with ANSD often do poorly with hearings aids and have poorer speech perception testing



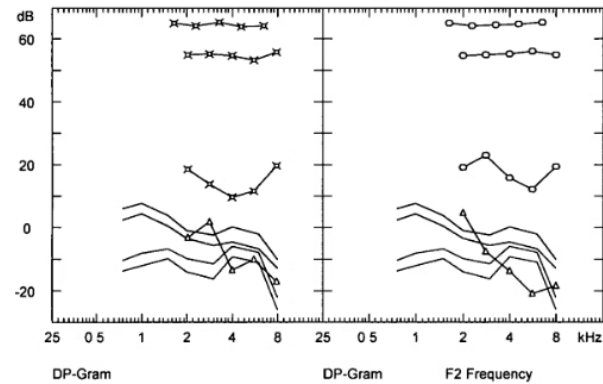
Some will do very well with a cochlear implant or gene therapy

Behavioral Assessment – 7 months of age



Behavioral Assessment – 10 months of age

Ear Time Date Protocol
 o Right 8:12:44 AM 11/4/2025 2-8 kHz Diagnostic Test - High Noise
 x Left 8:13:28 AM 11/4/2025 2-8 kHz Diagnostic Test - High Noise

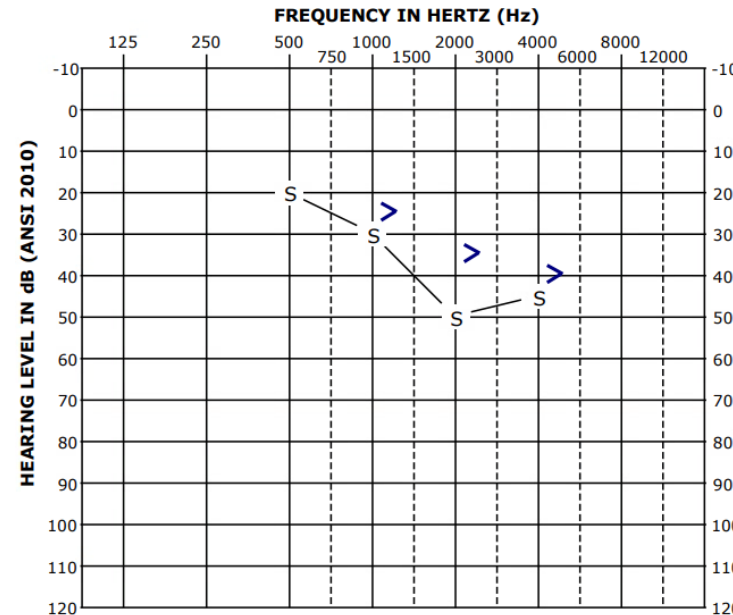


Right: 11/4/2025: -: 2-8 kHz Diagnostic Test - High Noise: 25K04D13.OAE

L1(dB)	L2(dB)	F1(Hz)	F2(Hz)	GM(Hz)	DP(dB)	NF(dB)	DP-NF(dB)
65.3	55.0	6516	7969	7206	19.5	-18.5	38.0
64.8	56.1	4594	5625	5083	12.2	-20.9	33.1
64.5	55.5	3281	3984	3616	15.9	-13.6	29.5
64.4	55.0	2297	2813	2542	23.0	-7.5	30.5
65.1	54.9	1641	2016	1818	19.1	4.6	14.5

Left: 11/4/2025: -: 2-8 kHz Diagnostic Test - High Noise: 25K04D14.OAE

L1(dB)	L2(dB)	F1(Hz)	F2(Hz)	GM(Hz)	DP(dB)	NF(dB)	DP-NF(dB)
64.4	56.0	6516	7969	7206	19.7	-17.0	36.7
64.1	53.3	4594	5625	5083	11.7	-10.2	21.9
65.4	54.9	3281	3984	3616	9.8	-13.3	23.1
64.3	55.4	2297	2813	2542	13.9	1.9	12.0
65.0	55.0	1641	2016	1818	18.7	-3.1	21.8



ULTRA HIGH FREQUENCIES (Hz)								* = masked value
	9K	10K	11.2K	12.5K	14K	16K	18K	20K
RIGHT								
LEFT								

PURE TONE AVERAGES (PTA)		
RIGHT	LEFT	SOUNDFIELD
		Air 33

TESTER / ASSISTANT: Maggie Kettler, AuD

AUDIOMETER: GSI AudioStar Pro

TRANSDUCER:

RELIABILITY:

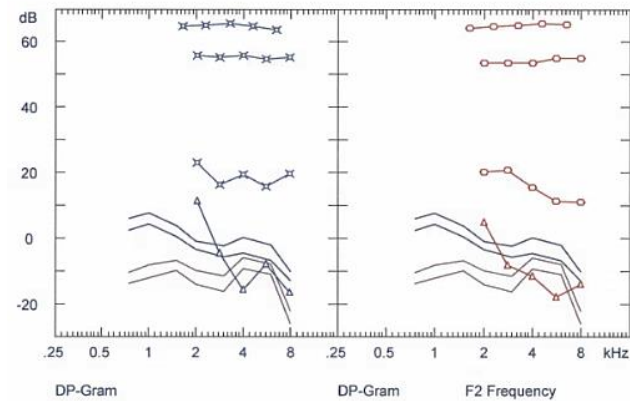
TEST TYPE:

NOTE: AN "X" ATTACHED TO THE ABOVE SYMBOLS INDICATES 'NO RESPONSE'

IMMITTANCE MEASUREMENTS		
Probe tone: 226 Hz	RIGHT	LEFT
Otoscopy		
Volume	0.59	0.63
Static Admittance	0.36	0.31
Width	121	114
Peak pressure	-33	-14
Tymp Type	A	A

Behavioral Assessment – 13 months of age

Ear Time Date Protocol
 x Left 8:10:59 AM 2/11/2026 2-8 kHz Diagnostic Test - High Noise
 o Right 8:11:40 AM 2/11/2026 2-8 kHz Diagnostic Test - High Noise



--:

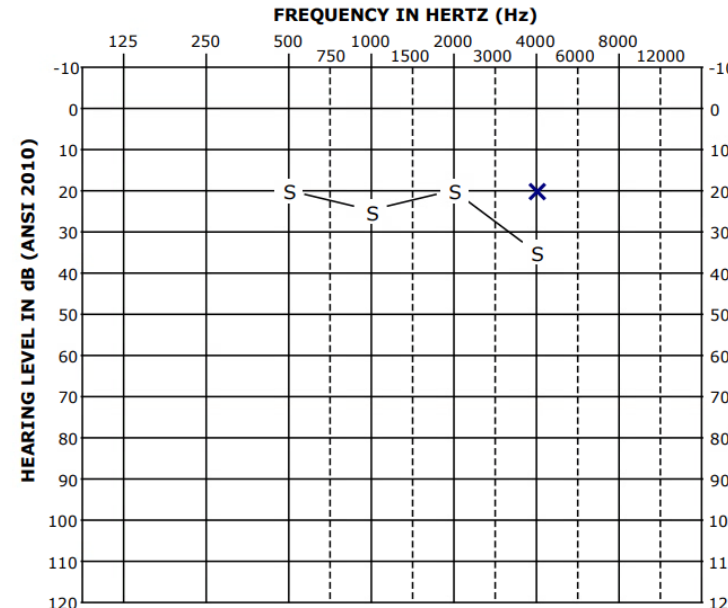
Left: 2/11/2026: -: 2-8 kHz Diagnostic Test - High Noise: 26B11D13.OAE

L1(dB)	L2(dB)	F1(Hz)	F2(Hz)	GM(Hz)	DP(dB)	NF(dB)	DP-NF(dB)
63.8	55.3	6516	7969	7206	19.8	-16.6	36.4
64.8	54.9	4594	5625	5083	15.9	-7.8	23.7
65.8	55.9	3281	3984	3616	19.5	-15.7	35.2
65.1	55.4	2297	2813	2542	16.5	-4.6	21.1
64.8	55.8	1641	2016	1818	23.0	11.3	11.7

--:

Right: 2/11/2026: -: 2-8 kHz Diagnostic Test - High Noise: 26B11D14.OAE

L1(dB)	L2(dB)	F1(Hz)	F2(Hz)	GM(Hz)	DP(dB)	NF(dB)	DP-NF(dB)
65.4	55.1	6516	7969	7206	11.1	-14.1	25.2
65.7	55.2	4594	5625	5083	11.5	-18.0	29.5
65.2	53.6	3281	3984	3616	15.5	-11.6	27.1
64.8	53.7	2297	2813	2542	20.7	-8.1	28.8
64.4	53.7	1641	2016	1818	20.3	5.0	15.3



ULTRA HIGH FREQUENCIES (Hz) * = masked value

	9K	10K	11.2K	12.5K	14K	16K	18K	20K
RIGHT								
LEFT								

PURE TONE AVERAGES (PTA)

RIGHT	LEFT	SOUNDFIELD
		Air 22

TESTER / ASSISTANT:
Maggie Kettler, AuD
Carrie Wingo, AuD

AUDIOMETER: GSI AudioStar Pro

TRANSDUCER: Inserts

RELIABILITY: Good

TEST TYPE: VRA

NOTE: AN "*" ATTACHED TO THE ABOVE SYMBOLS INDICATES 'NO RESPONSE'

IMMITTANCE MEASUREMENTS

Probe tone: 226 Hz	RIGHT	LEFT
Otoscopy		
Volume	0.58	0.58
Static Admittance	0.33	0.41
Width	112	96
Peak pressure	13	-5
Tym Type	A	A

mmho

daPa

Brother

- Failed newborn hearing screening both ears; history of speech delay
- ABR at another center was normal
- Normal head CT at 18 months completed for non-related issue which later resolved
- Slightly delayed speech development but by age 3 is hitting all speech and language milestones



Symptoms of ANSD

Hearing sounds but not understanding words.

Trouble hearing in noisy environments.



Speech delay in children.

Inconsistent responses to sound.

Brother: Genetic Results

Result Summary

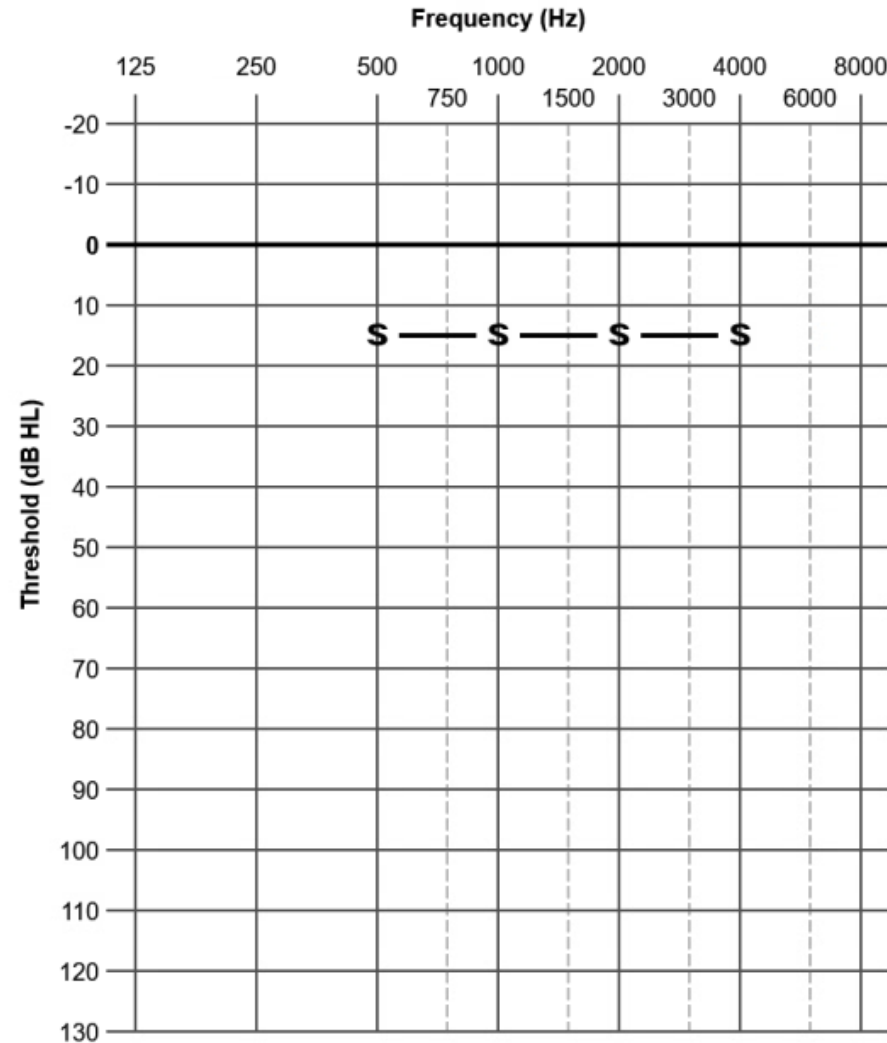
Gene	HGVS	AA Change	Zygosity	Classification
OTOF	NM_001287489.2:c.5816G>A	p.(Arg1939Gln)	Heterozygous	Pathogenic
OTOF	NM_001287489.2:c.5971G>C	p.(Val1991Leu)	Heterozygous	Uncertain significance

If this patient has an ANSD phenotype, this report would make me suspicious that the VUS may eventually prove to be disease-causing because:

1. There is already one confirmed pathogenic OTOF variant.
2. A second rare OTOF variant is present.
3. The variants are located close together in the protein.
4. OTOF is one of the most common causes of congenital ANSD.

This is not proof, but it increases the likelihood that the VUS may be clinically relevant.

Brother: Behavioral Assessment





- What causes a child with a known genetic cause of AN and an ABR that shows AN to present with normal hearing?
- Will hearing change over time?
- Will speech and language development be impacted?
- Will they present with hearing difficulty in background noise?
- What does this mean for candidacy of gene therapy or traditional CI surgery?

Recommendations

- Monitor both children audiologically and with speech-language evaluations
- Consider sedated ABR to see if AN has resolved
 - Currently waiting for other sedation opportunities.

Hearing aids-Traditional
hearing aids are fit to the
loss that the patient shows
during testing; less
effective for patients with
poor speech perception
testing

Cochlear implants-Should
be considered regardless
of hearing thresholds if
speech perception is
decreased; outcomes vary
greatly depending on cause
or location of lesion

Assistive listening devices-
Can be utilized with other
devices or alone if patient
has good speech
perception testing

Gene Therapy-Very new
treatment option being
investigated

Thank you!

Questions?

Kentucky Audiology & Tinnitus Services



Tinnitus

Tinnitus Evaluation Workflow



Background



Case
History

+ 56 Year Old Female

Permanent Residence is in Colorado

+ Ramsay Hunt Syndrome - Jan 2025

Triggered initial onset of tinnitus

+ Hypogammaglobulinemia

Autoimmune disorder

+ Previously used Lenire Tinnitus Treatment

Tinnitus Ratings

+ THI: 100/100

Catastrophic Handicap of Tinnitus

+ Daily Impact

Severe Disruption to Sleep
Emotional Disregulation

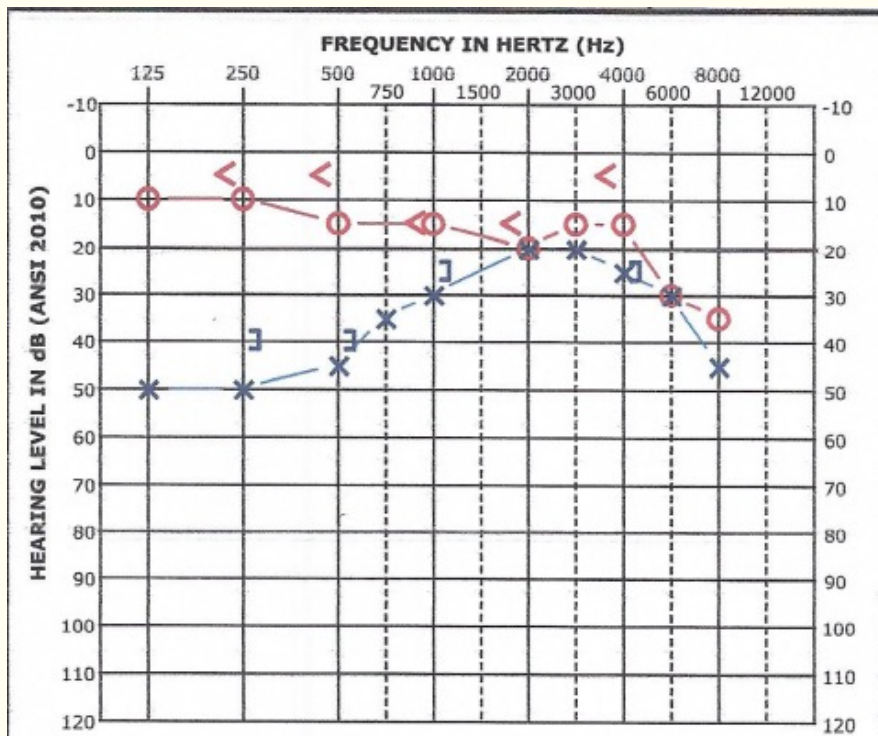
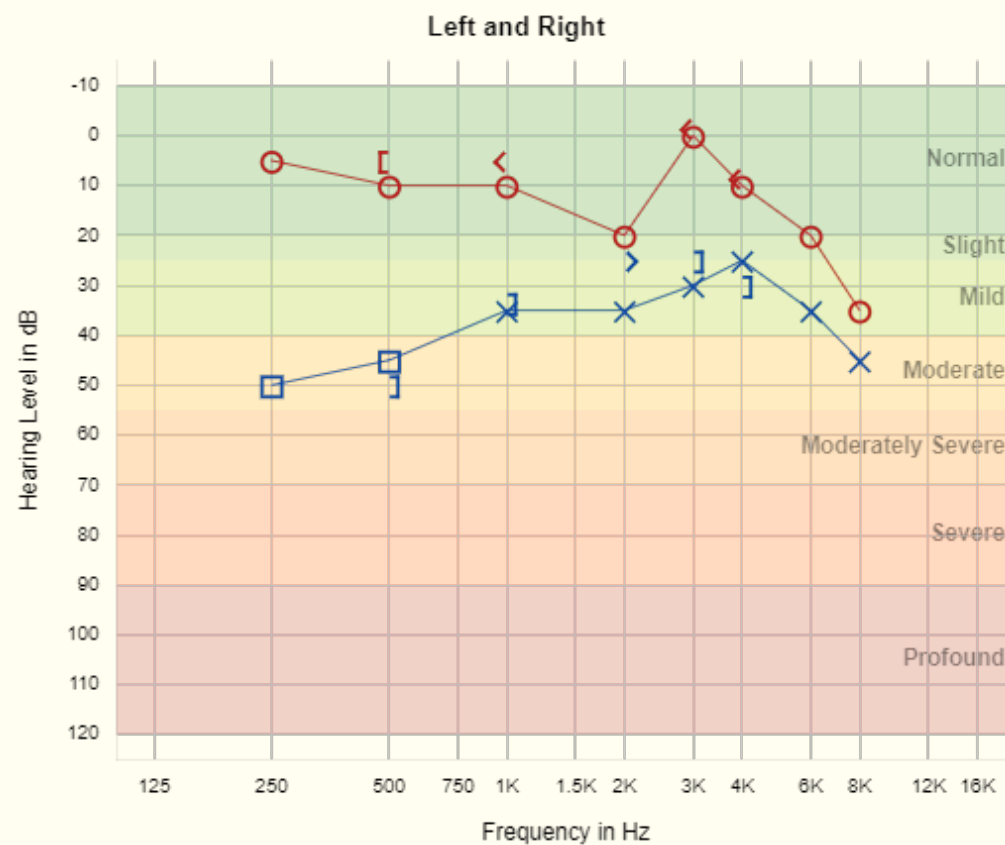
+ Awareness and Annoyance Ratings

AW: 100%, AN: 100%, S: 10/10, AN: 10/10, EL: 10/10

+ Suicidal Ideations

"Get better or seek accompanied suicide in Switzerland"

Case
History



**Audiometric
Evaluation**

Next Steps...



Sound Therapy - Ear Level Devices

Can be used 24/7, often beneficial for improved sleep



Lenire Tinnitus Treatment Device



Follow up TRT Appointment



Recommendation to seek CBT/Counseling

**Customized
Treatment
Plan**

***Two days later the patient
needed an immediate referral
due to a mental health crisis.***

Office Suicide Plan



Train Your Staff

Do you have a plan?

Do you have a motive?

Do you have the means to carry out your plan?



Ask The Right Questions

If telehealth, where is the patient? Who else is home? Do they have weapons in the house?



Same or Next Day Scheduling

Treat this like SSNHL - this is an **EMERGENCY**



Referral Versus Hospitalization

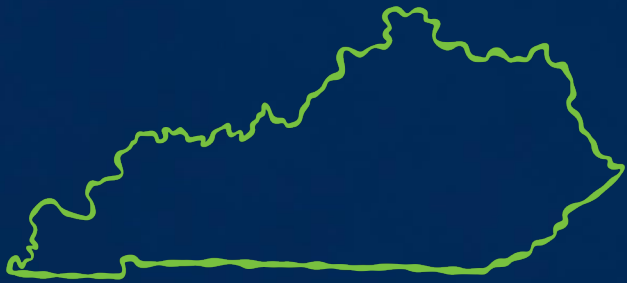
Have referral sources for local counselors ready - online or in person.

Know the local hospitals/mental health centers in your area.



Create a 'What If' Plan with Your Patient

Local Resources



+ Suicide Hotline - 988

+ Eastern State Hospital - Lexington
The Ridge - Lexington
Central State Hospital - Louisville
UofL Peace Hospital - Louisville
The Brook - Louisville
Western State Hospital - Hopkinsville
Lincoln Trail Behavioral Health System
- Radcliff
SUN Behavioral Health - Erlanger

+ Local Therapists

**One
Week
Later...**

**Patient called my office to
troubleshoot her loaner devices.**

Returned back home to Colorado.

Noticed an immediate improvement in symptoms - both hearing and tinnitus
"Reduced from a level 100 to a level 5"

Decided to start managing anxiety.

.

***She then thanked me - for
caring enough to send her
for help.***

Clinical Timeline

Initial Visit - August 31

Sent to Local Hospital - Sept 2

Check in - Sept 3

Device Troubleshooting -
Sept 11

TRT Follow Up - Sept 22

Less than one month...

Tinnitus Ratings



THI: 36/100

Mild Handicap of Tinnitus



Sound Therapy

Wearing ear level devices daily



Daily Impact

Able to attend local music festival and is sleeping more consistently



Suicidal Ideations

Still mentioned this idea in reference to future tinnitus spikes

Tinnitus
Retraining
Therapy
(TRT)

Kentucky Audiology & Tinnitus Services



Sound Sensitivity

Types of Sound Sensitivity

Hyperacusis

Aversion to all sounds above a certain **decibel level**.

General overactivity in the central auditory system.

Misophonia

Aversion to **very specific sounds**, usually low level repetitive sounds.

Often related to the quality of the sound, rather than the volume.

Sound Sensitivity Evaluation Workflow



Background



+ 12 Year Old Male

Accompanied to appointment by Mom

+ Triggers started around 9-10 y/o

Computer mouse clicking, computer mouse on mouse pad, whispering, chewing, feet moving against floor

Misophonia or Hyperacusis?

+ Both emotional and physical reactions

+ Attention Deficit Hyperactivity Disorder (ADHD)

Case
History

Sound Sensitivity Ratings

+ **MAQ: 16/63**

+ **HADS: 5/42**

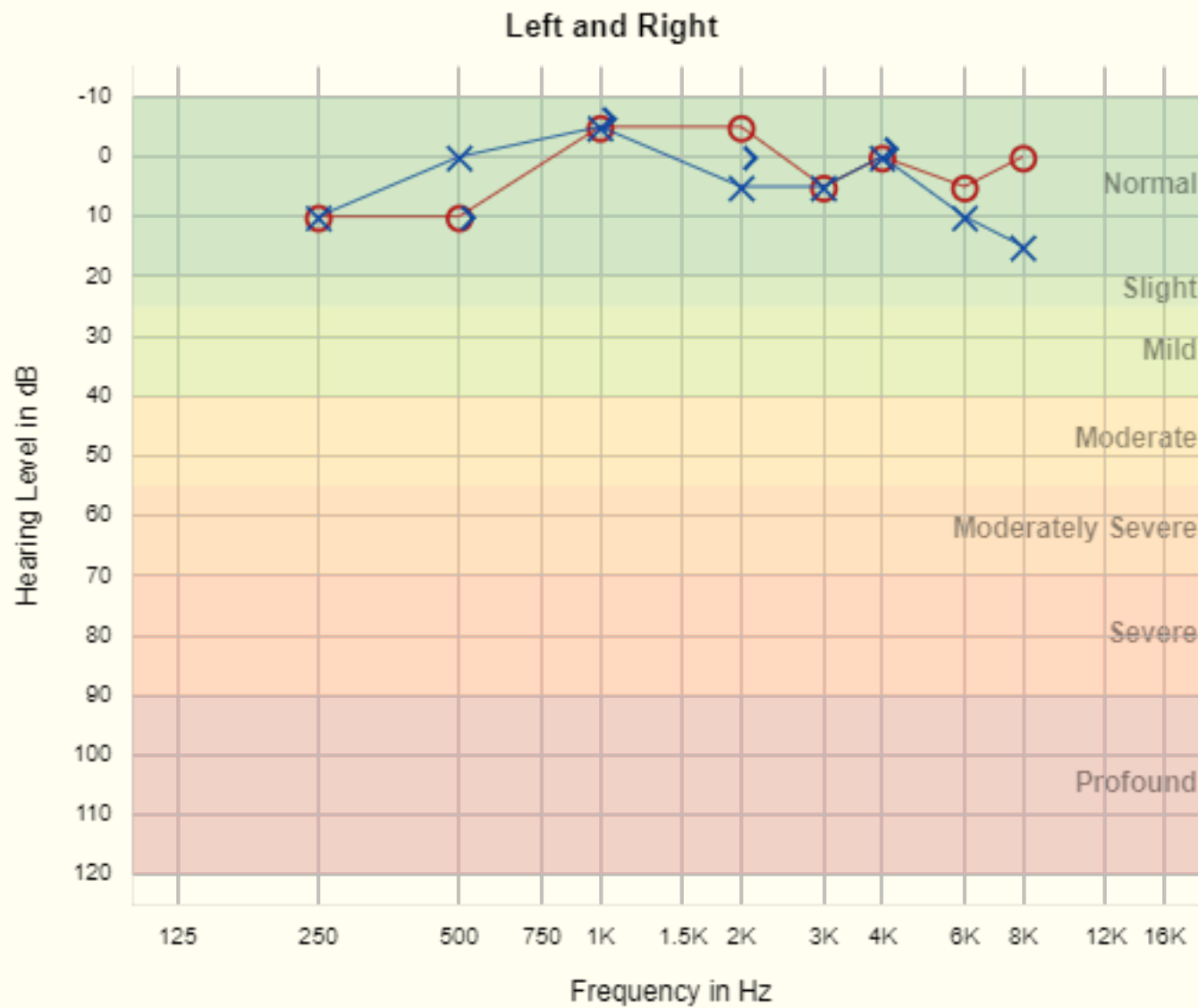
No concerns for anxiety or depression

+ **MKHQ: 34/100**

+ **Trigger Sound Ratings**

Mouse clicking 8/10, Mouse on mouse pad 6/10,
whispering 4/10, chewing 4/10, feet on floor 0.1/10

Case
History



**Audiometric
Evaluation**

Next Steps...



Sound Therapy - Computer

Play sound while using computer



Music Therapy



Relaxation Exercises

Jumping on trampoline, legos



Rearrange daily schedule

Limit time on computer

**Tinnitus
Retraining
Therapy
(TRT)**

**Customized
Treatment
Plan**

Six Weeks Later...

Mom and Patient returned to my office.

MAQ: 2/63

Report from Patient: *"things are going much better".*

Purchased silent mouse.

Using YouTube videos for sound therapy while on the computer.

Report from Mom: Big reduction in reaction.

Currently on summer break - spending more time outside.

Transferring to new school in Fall, with smaller class sizes and less computer work.

.

Customized
Treatment
Plan

**Discussed Possible
IEP/504 Plan**

Three Months Later...

Phone call with Mom to discuss school transition.

Overall great transition.

Noticing maturity outside of sound triggers - trying new foods.

Able to tolerate fire drill at school.

Still struggling at home while playing action video games on his computer.

Customized
Treatment
Plan



sgarrard@kytinnitustreatment.com

Thank You!