

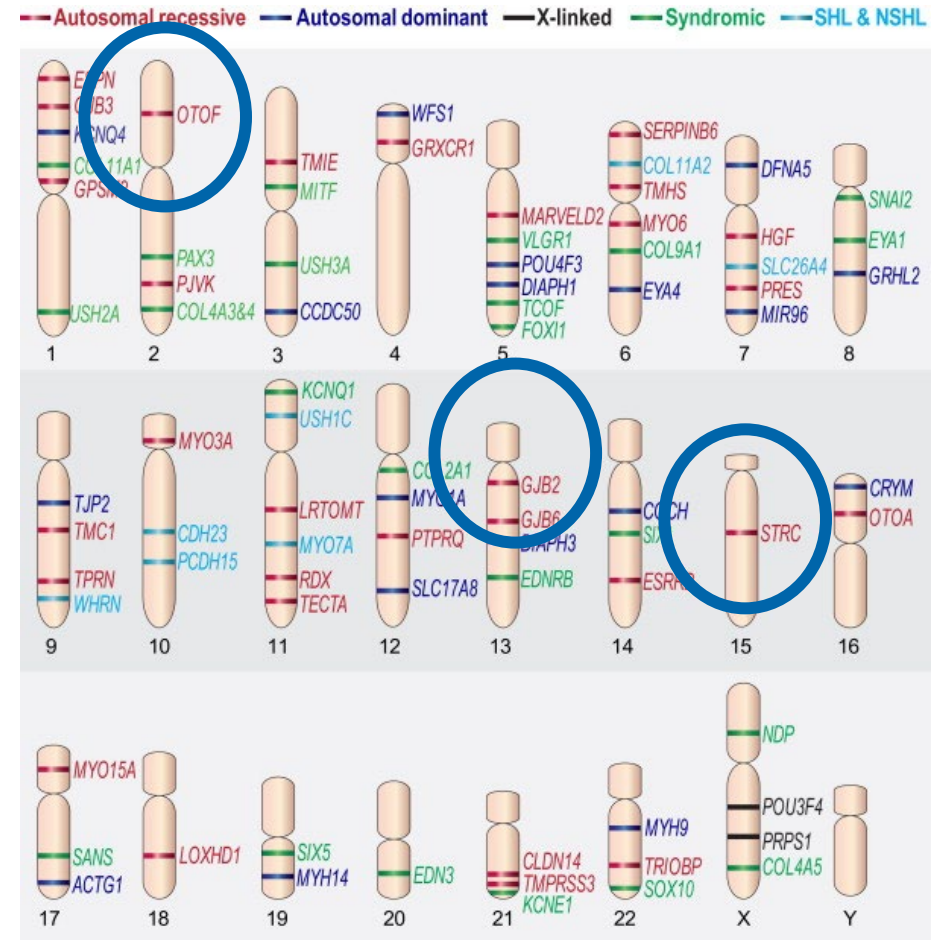
# Barriers to Genetic Testing for Hearing Loss: A Real World Survey

Tera Quigley, AuD, CCC-A  
Associate Director, Medical Affairs



**Decibel**  
THERAPEUTICS™

# Genetic Protein Deficiencies Cause Most Congenital Hearing Loss



Raviv et al. (2010) Ann NY Acad Sci

# Current evidence shows genetic testing is broadly useful in improving clinical management of children with hearing loss

## Massively Parallel Sequencing for Genetic Diagnosis of Hearing Loss: The New Standard of Care

A. Eliot Shearer, MD, PhD<sup>1</sup>, and Richard J. H. Smith, MD

Sponsorships or competing interests that may be relevant to content are disclosed at the end of this article.

### Abstract

**Objective.** To evaluate the use of new techniques for comprehensive genetic testing

countries, and is nonsyndromic abnormality

Otolaryngology—  
Head and Neck Surgery  
1–8  
© American Academy of  
Otolaryngology—Head and Neck

Published in final edited form as:

*Hear Res.* 2017 May ; 348: 138–142. doi:10.1016/j.heares.2017.02.008.

## Genetic Variants in the Peripheral Auditory System Significantly Affect Adult Cochlear Implant Performance

A Eliot Shearer<sup>a</sup>, Robert W Eppsteiner<sup>a,#</sup>, Kathy Frees<sup>a</sup>, Viral Tejani<sup>a</sup>, Christina M Sloan-Heaen<sup>a</sup>, Carolyn Brown<sup>a,b</sup>, Paul Abbas<sup>a,b</sup>, Camille Dunn<sup>a</sup>, Marlan R Hansen<sup>a</sup>, Bruce J

JAMA  
Network | **Open**<sup>™</sup>



Original Investigation | Otolaryngology

## Outcomes of Gene Panel Testing for Sensorineural Hearing Loss in a Diverse Patient Cohort

Elizabeth N. Liao, BA; Emily Taketa, BS; Noura I. Mohamad, BA; Dylan K. Chan, MD, PhD

### Abstract

**IMPORTANCE** A genetic diagnosis can help elucidate the prognosis of hearing loss, thus significantly affecting management. Previous studies on diagnostic yield of hearing loss genetic tests have been based on largely homogenous study populations.

**OBJECTIVES** To examine the diagnostic yield of genetic testing in a diverse population of children

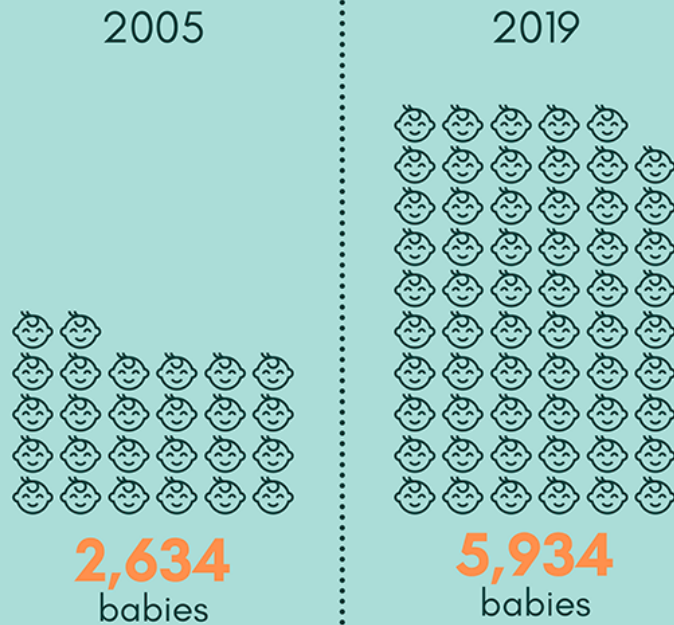
### Key Points

**Question** What is the association between rates of genetic diagnosis and sociodemographic and clinical characteristics in children with sensorineural hearing loss, and how

# Early Identification is Key to Catalyzing Novel Therapeutics

## Massive Progress Over the Last 2 Decades

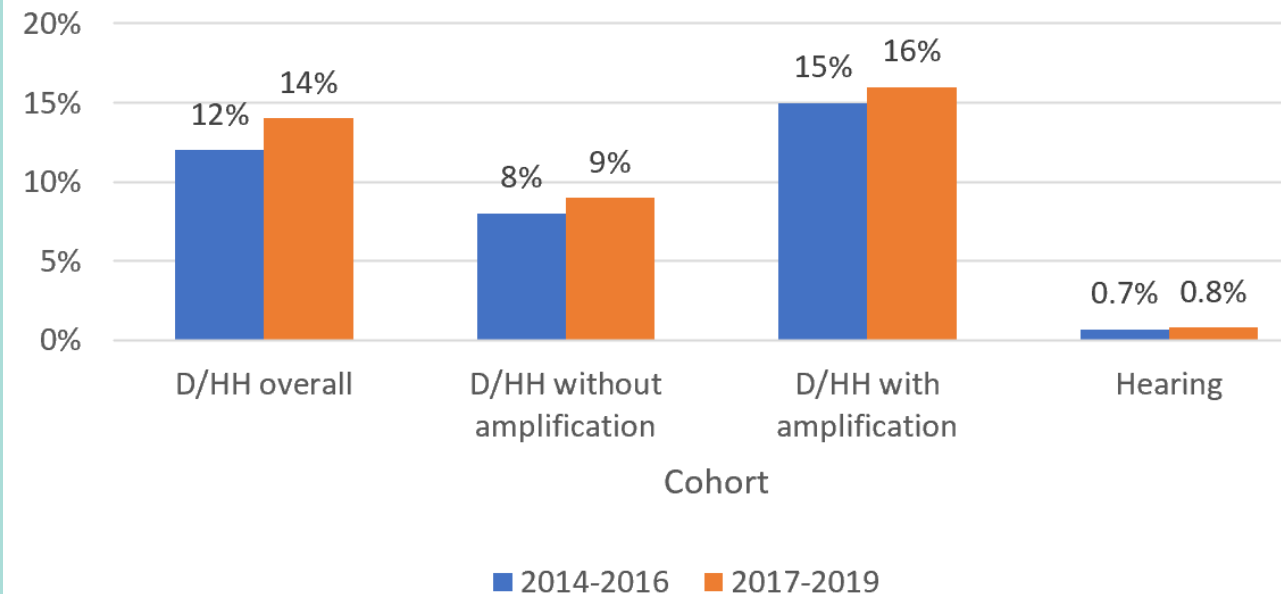
More and more babies who are born deaf or hard of hearing are being **identified early** in the United States



CDC: <https://www.cdc.gov/ncbddd/hearingloss/ehdi-data.html>

## Genetic Diagnosis is a Work in Progress

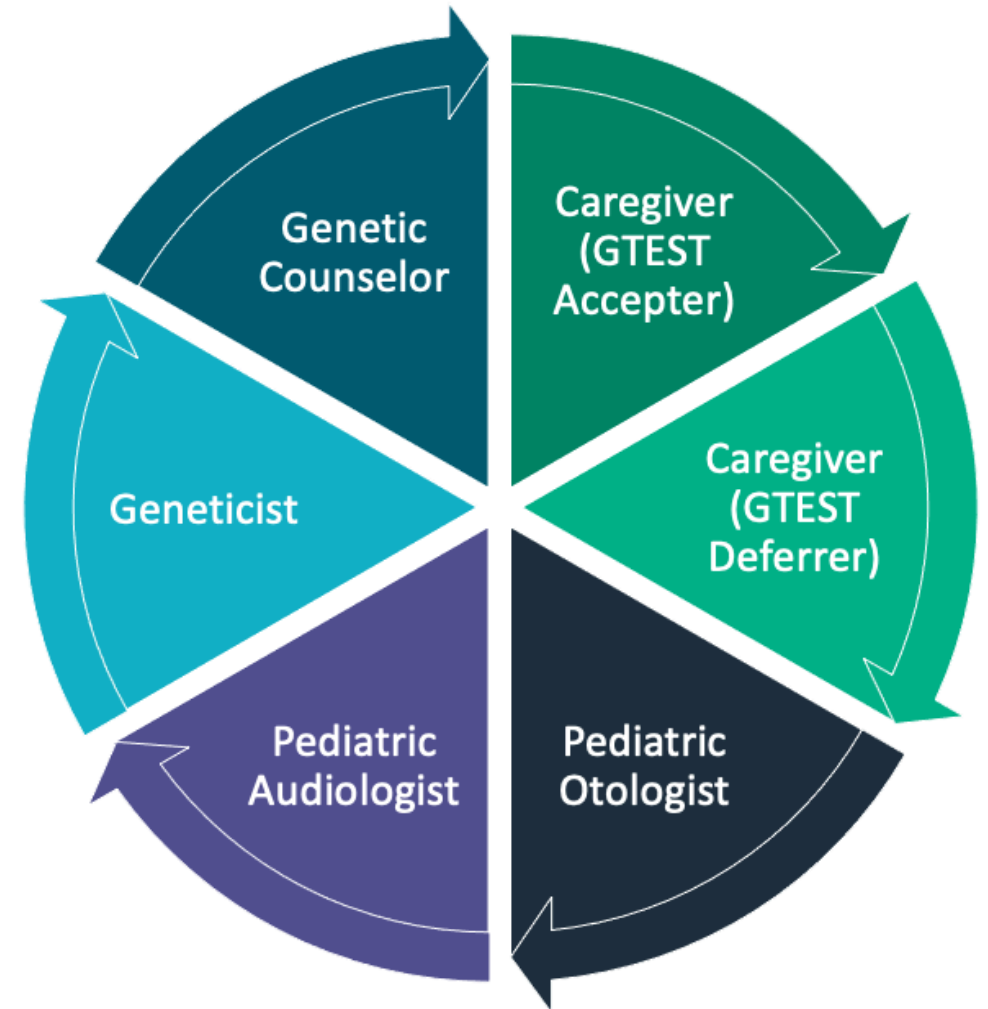
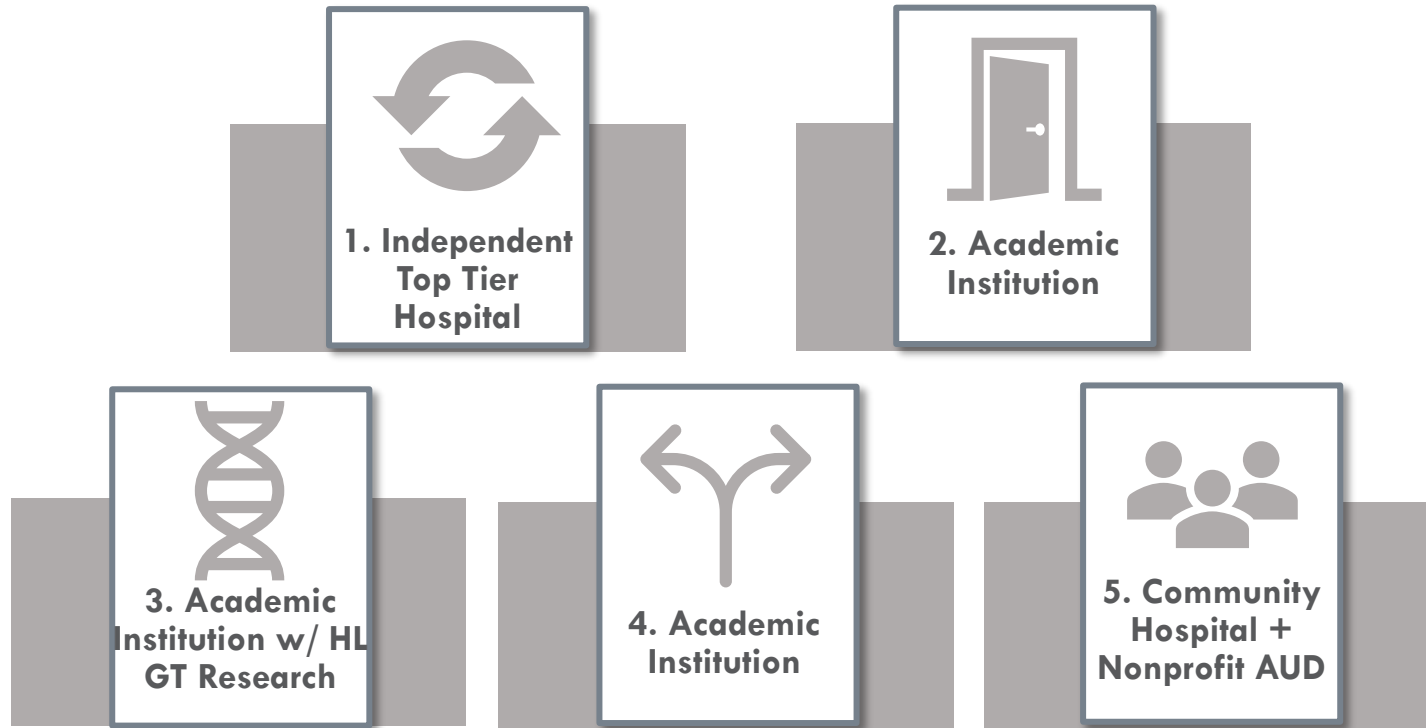
### Prevalence of Genetic Testing Among Insured Young Children



[S. Charania, S. Grosse, K. Dundon EHDI 2022 Poster Study](#)

# Clinicians and Caregivers Selected from 5 Different Children's Hospitals

All 5 Otologists interviewed treated at least one caregiver in sample



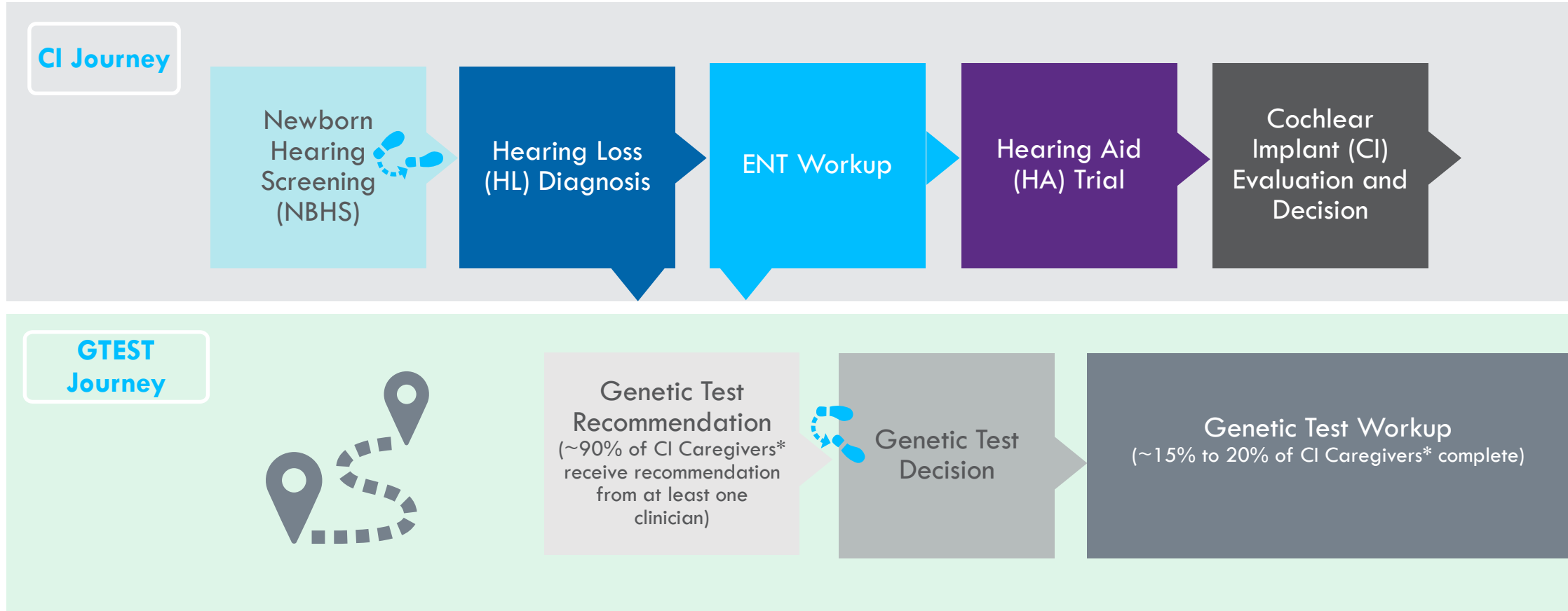
# This study focuses on the CI-indicated and compliant segment, which has the highest likelihood of success in navigating the caregiver CI Journey

Segments of Children Diagnosed with Bilateral SNHL in First Year of Life				
Compliance:	Hearing Loss Severity			
	Mild	Moderate	Severe	Profound
Lost to follow up after failing NBHS (e.g., children with ANSD who pass with OAE)				
Elect not to proceed with indicated treatment (HAs or CIs)				
Elect to proceed with indicated treatment (HAs or CIs)			Focus of this study	

### Patient Characteristics

- Severe to profound, sensorineural, nonsyndromic, bilateral HL
- CI candidates
- Diagnosed in first year of life, within last several years
- Caregivers accordingly described as “CI Caregivers\*” throughout this deck
- Half have undergone genetic test, while half have not

# Hearing loss treatment and genetic testing are typically concurrent, but often not integrated







### NBHS

Baby fails 2 NBHS tests  
Hospital refers to Pediatric Audiologist (PED AUD) for diagnostic ABR

- 1. Pediatrician
- 2. Online research

### HL Diagnosis

1-3 AUDs consulted  
PED AUD mentions CIs and refers to ENT

- 1. PED AUD
- 2. EHDI
- 3. Facebook groups

### ENT Workup with GTEST Rec & Workup

1-2 OTO/ENTs consulted  
OTOL recommends CIs for LSL

**Genetic Testing (GTEST) decision: treatment mgt: medical & educational needs; progression**

- 1. PED AUD, ENT/OTOL, Genetic Counselor (COUN)
- 2. EHDI
- 3. Facebook groups

### HA Trial

Baby undergoes HA trial for insurance requirement  
PED AUD recommends CIs

- 1. PED AUD
- 2. EHDI
- 3. Facebook groups

### CI Evaluation & Decision

OTOL conducts CI eval  
Benefits & risks of CIs assessed

**Cochlear Implant decision: Family comm & bonding  
Child social identity**

- 1. PED AUD & ENT/OTOL
- 2. EHDI
- 3. Facebook groups

Lack of caregiver urgency ("just fluid;" no family history)

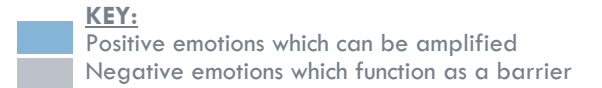
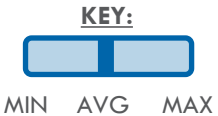
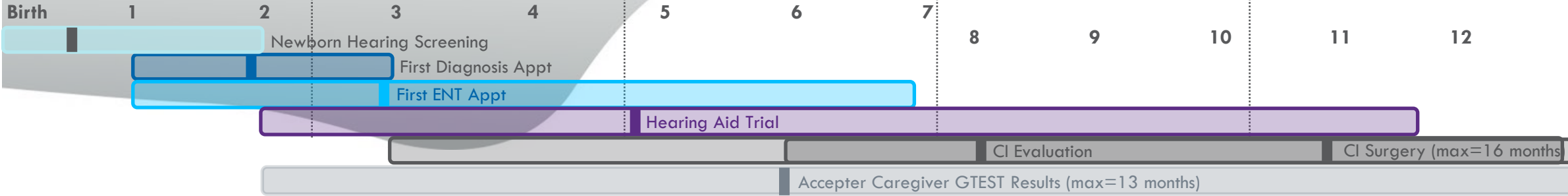
Confusion over diagnosis content and next steps  
Poor AUD delivery style

Soft to no GTEST recommendation  
Rarely GTEST mgt benefits  
Fears blame for HL gene  
GTEST process not clear  
Wait time for COUN appt  
Insurance auth delay

Unnecessary trial delays HAs  
Difficulty keeping HAs on baby

Risk of poor hearing outcomes

Months Since Birth →



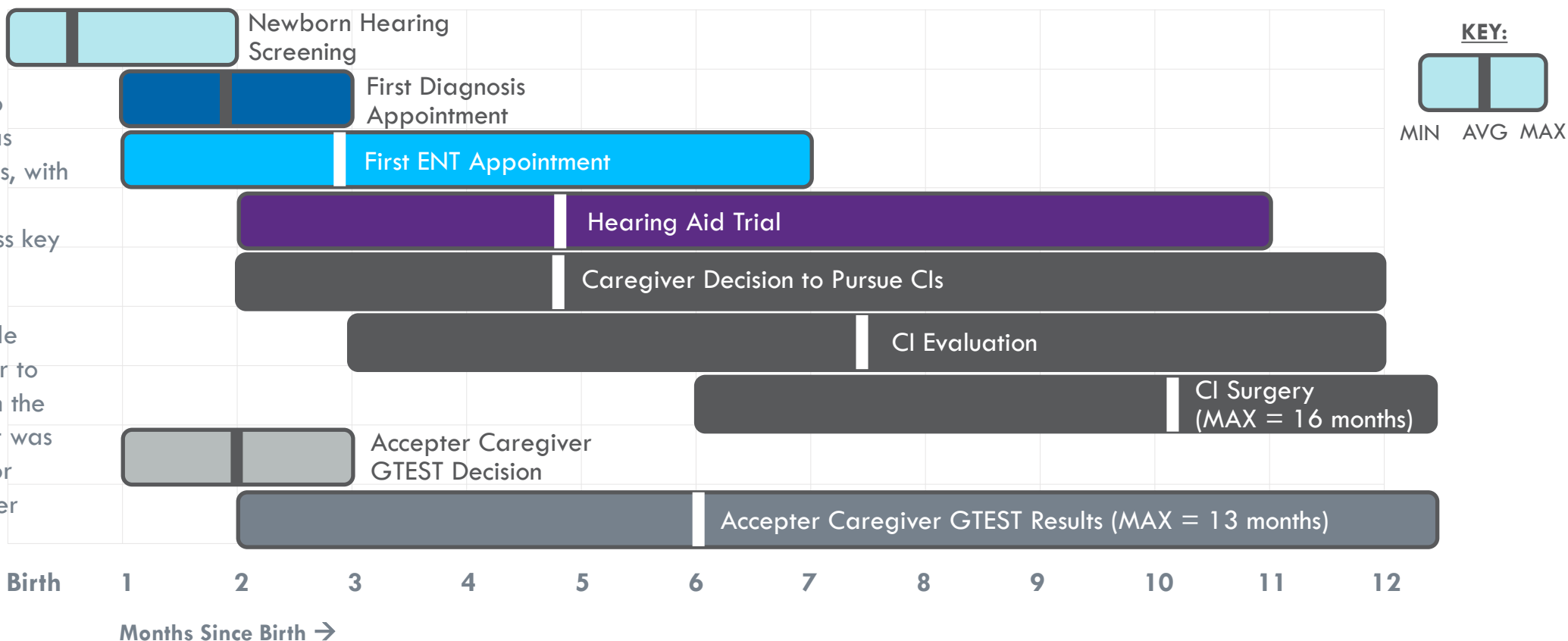




# Time Intervals, by Journey Stage

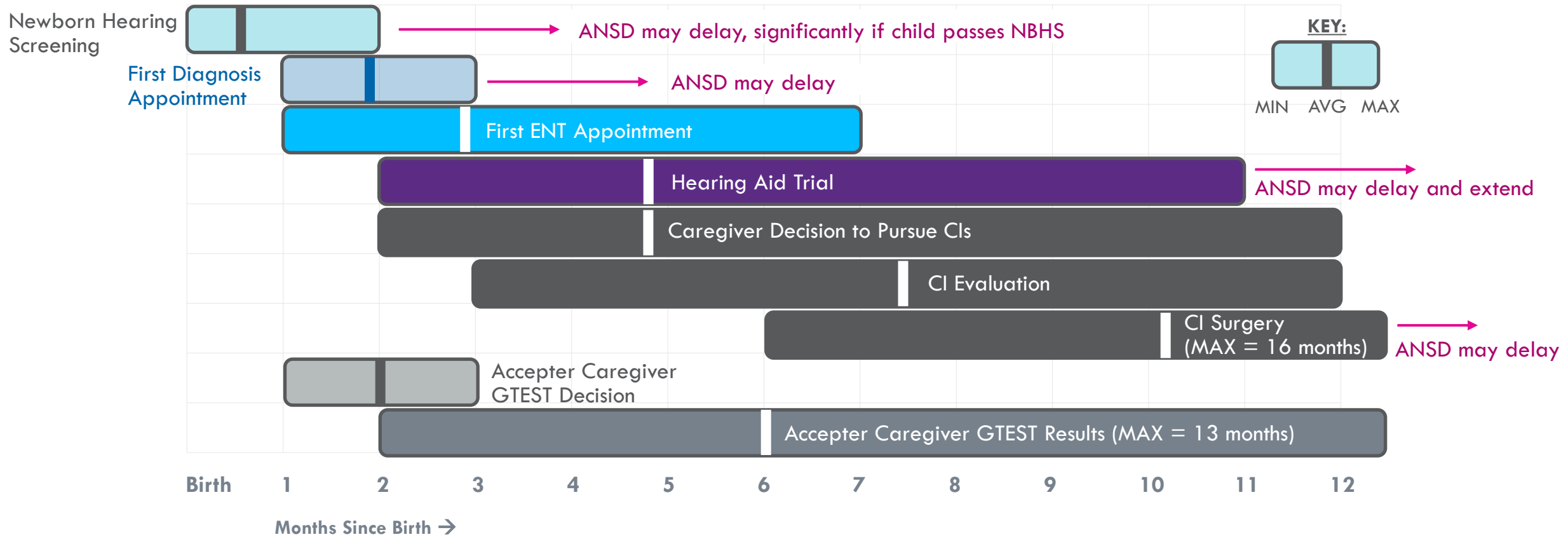
- Average time to implantation was about 10 months, with considerable variability across key decision points

- Caregivers made decision whether to pursue GTEST in the first visit when it was recommended or shortly thereafter



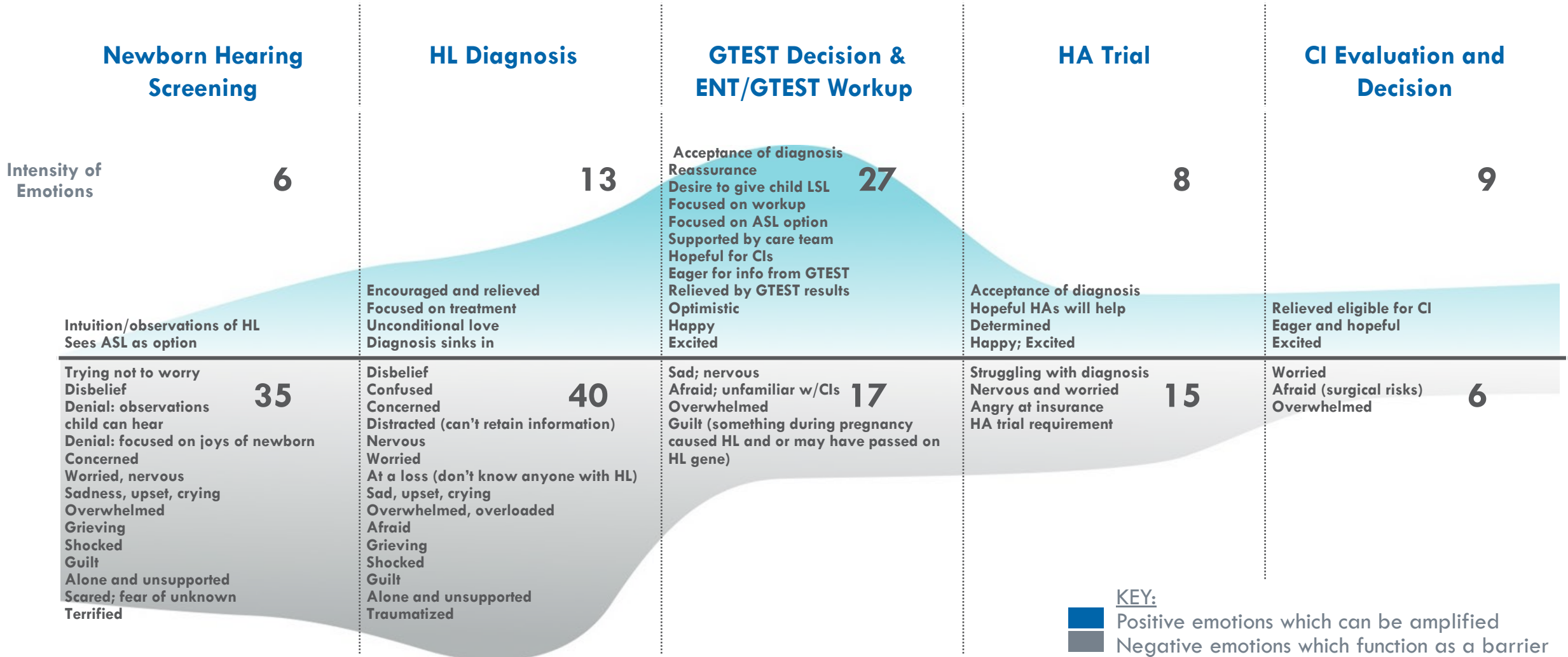
Note that one caregiver took a year to get the HL Diagnosis because her baby did not undergo NBHS since it was not covered by insurance, according to caregiver. This outlier data excluded from chart above but is an important example of barriers faced by caregivers.

# Auditory Neuropathy Spectrum Disorder (ANSD) may Extend Patient Journey in Multiple Stages





# CI Caregivers\* Emotions, by Journey Stage



**KEY:**  
■ Positive emotions which can be amplified  
■ Negative emotions which function as a barrier

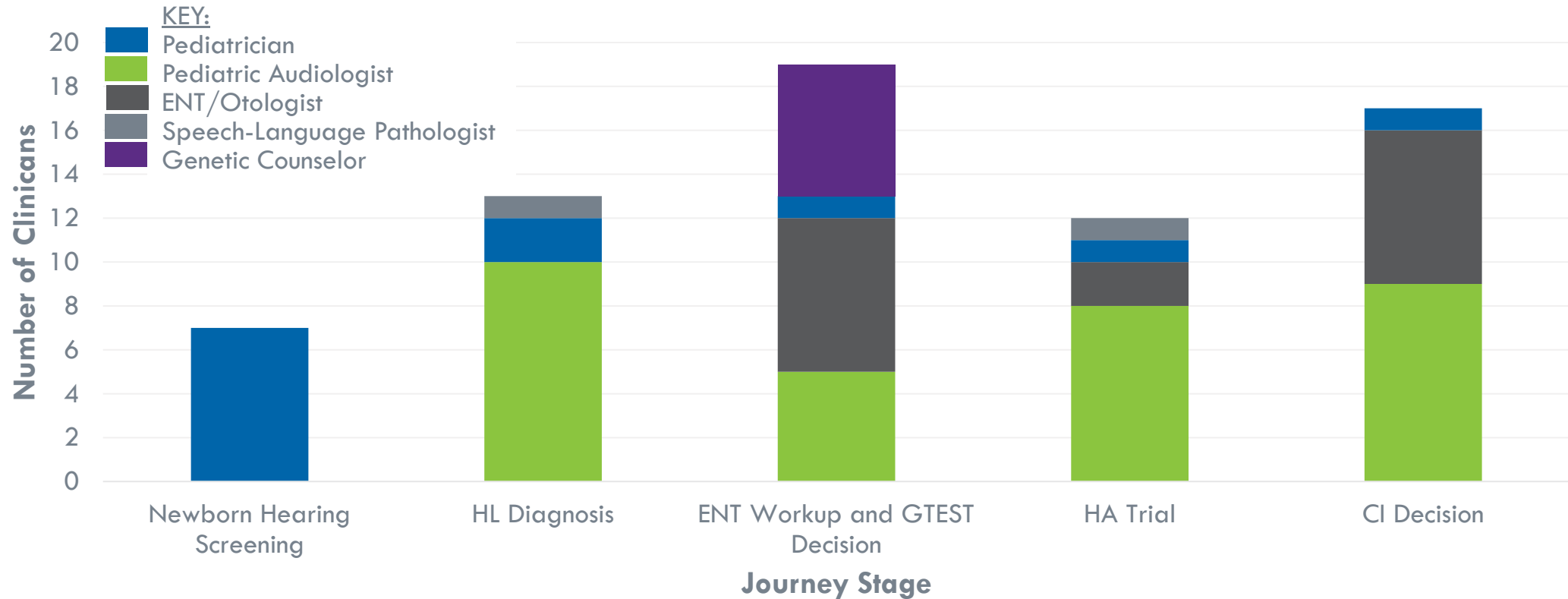
Intensity of emotions = emotions experienced for each stage multiplied by number of people experiencing each emotion.

# Audiologists are the most influential provider to caregivers across the journey

Pediatrician's influence rapidly wanes after NBHS



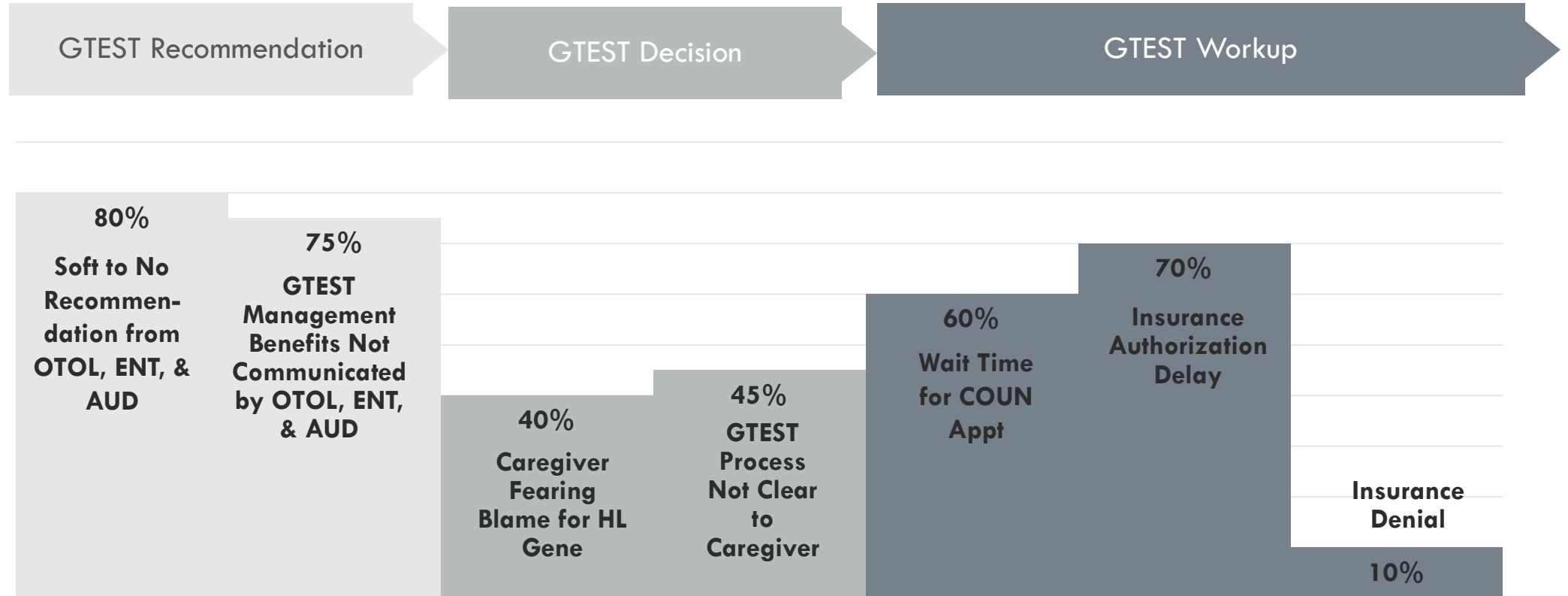
## Providers Who Influence Caregivers\* by Journey Stage





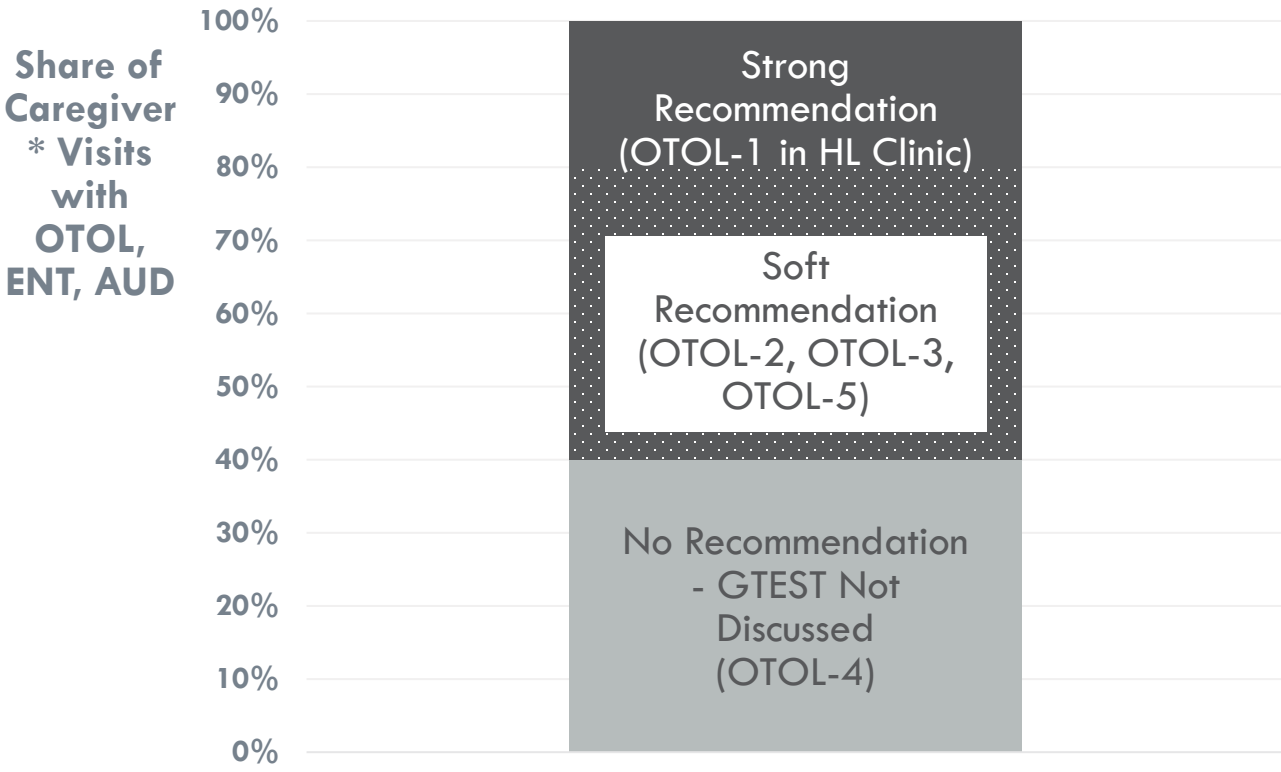
# **Genetic Testing Hierarchy of Barriers**

# Clinicians make a soft to no genetic testing recommendation most of the time



# Clinicians don't make a strong recommendation for genetic testing most of the time

Distribution of OTOL, ENT, and AUD GTEST Recommendation Type, Based on Caregiver Recall



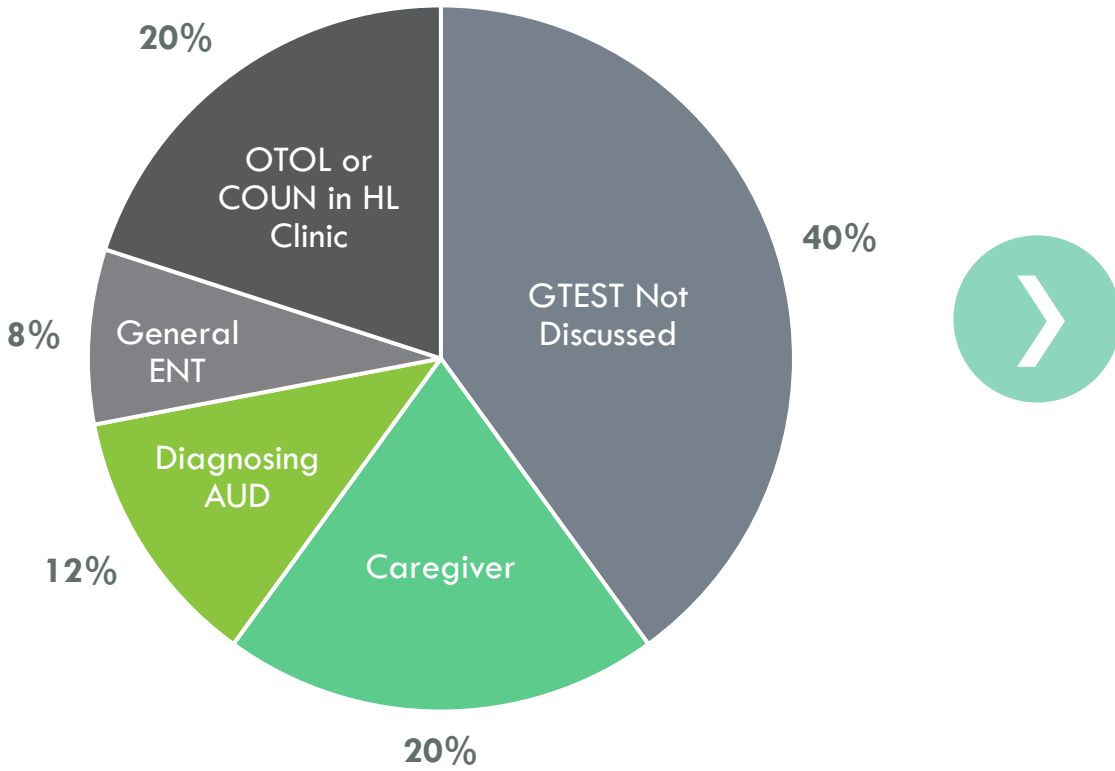
### Conclusions

- Most of the time (80%), OTOLs, ENTs, and AUDs deliver a soft to no recommendation for GTEST, based on caregiver recall
- Deferrers were marginally more likely not to receive any GTEST recommendation (matter not discussed)
- As gatekeepers, clinicians influence caregivers' sense of urgency around GTEST



# Caregivers Were Frequent Initiators of Genetic Testing Conversation

Initiator of GTEST Discussion with CI Caregivers\*, as Share of Total Visits, Based on Caregiver Recall



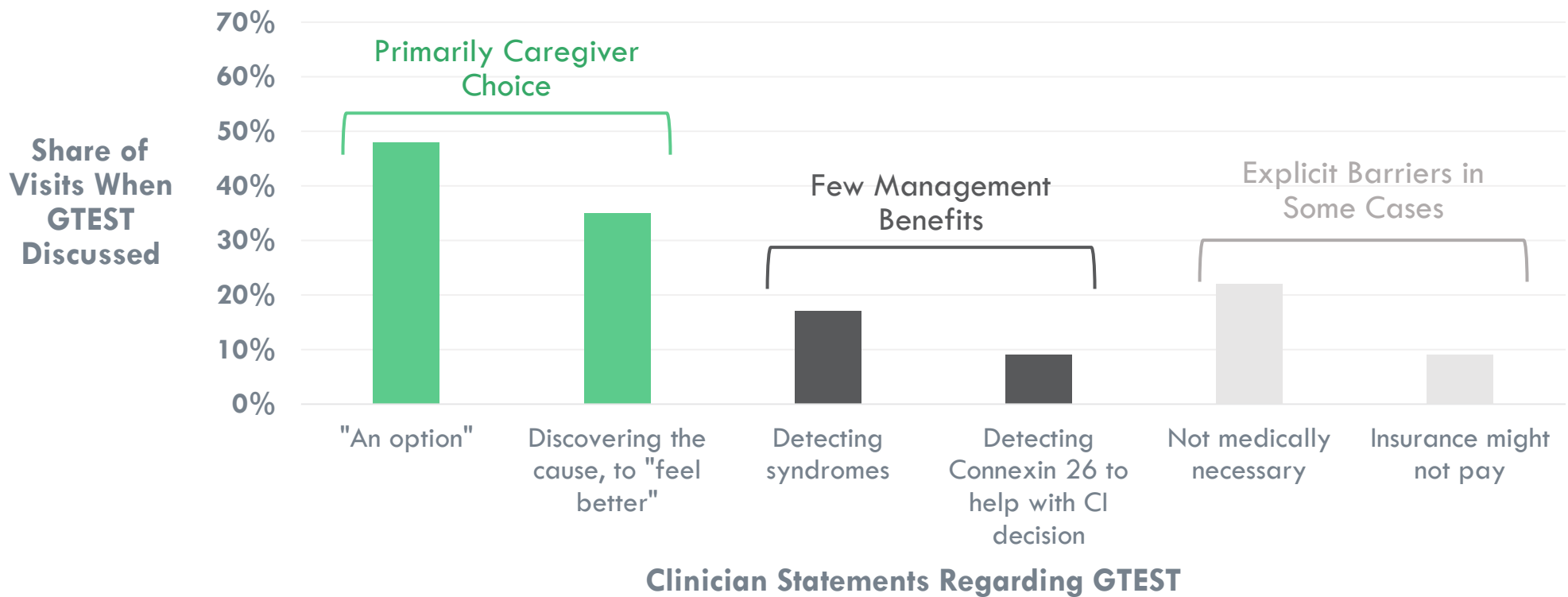
### Conclusions

- Caregivers are just as likely as OTOLs to bring up GTEST
- Caregivers learn about GTEST from EHDI hearing therapist, other parents, Facebook CI group, pediatrician, GTEST for older child, and profession as a teacher
- General ENTs and Diagnosing AUDs who bring up GTEST play an important role in supplementing OTOLs to create awareness

# Clinicians are positioning genetic testing as an option, to find the cause to “feel better”

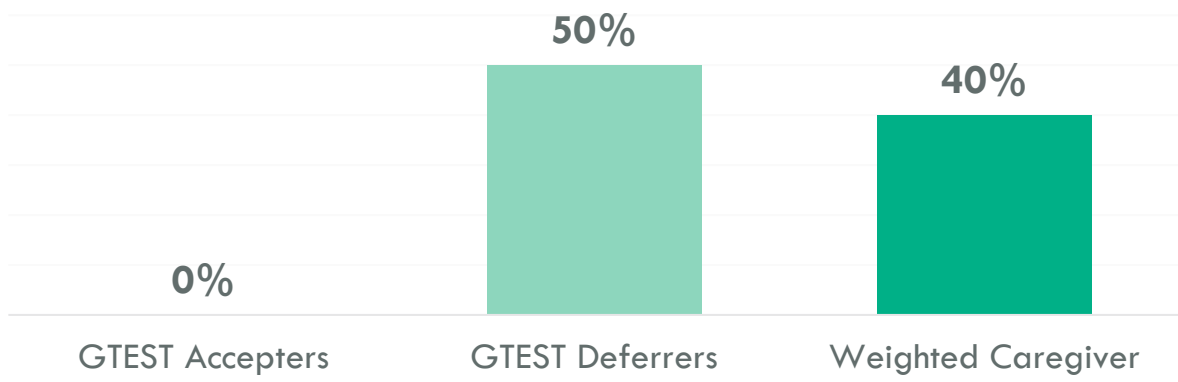
In 75% of the visits when genetic testing was discussed, management benefits were not mentioned

Caregiver Recall of How OTOL, ENT, and AUD Positioned GTEST



# Fear of being blamed for their genes was powerful barrier for Deferrers

Share of Caregivers\*  
Who Fear Blame for Their HL Gene



### Quotes

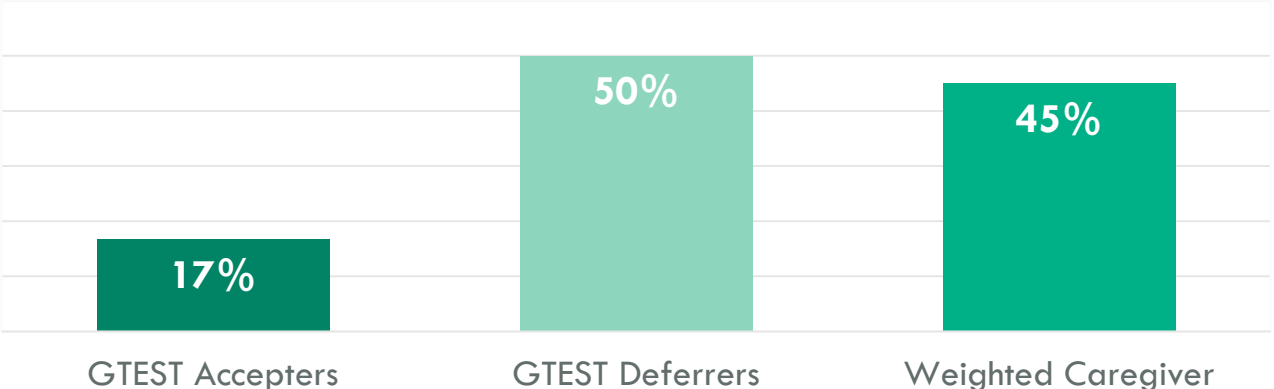
- *“It’s one of the things I wanted left in the dark. Because I was blaming myself enough during that period. So, I definitely didn’t want to go that route and be like, ‘I was the carrier of it.’” - GTEST Deferrer*
- *“I think just everyone benefits. My husband benefits, his brother benefits. Our future children. I just think having that answer helps. I benefit from stop blaming myself.” - GTEST Acceptor*

### Conclusions

- Deferrers worried that specifically THEIR genes (as opposed to their partner) “caused” the child’s HL
- Positioning genetic testing as finding the cause of HL will trigger potential Deferrers, causing them to shut down, with little recourse
- In contrast, Accepters viewed genetic testing as a way of freeing them from guilt that something they did wrong during pregnancy caused the child’s HL
- AUDs and Genetic Counselors anticipated this barrier, while OTOLs did not

# Deferrers struggled to understand how GTEST process might work

Share of Caregivers\* for Whom GTEST Process Unclear



## Conclusions

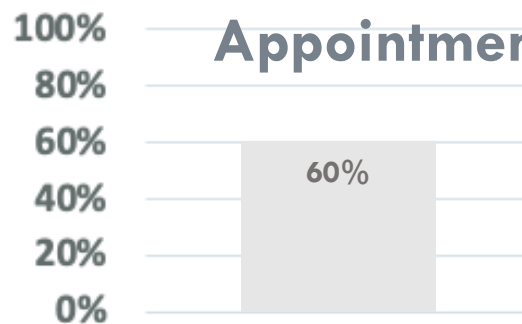
- Half of Deferrers did not feel that they received clear direction on the process, whereas only 1 Acceptor felt that way
- With lower SES, Deferrers may have less context for understanding how the process would unfold
- **Implication is that clinicians should outline process, stressing ease, while delivering the genetic testing recommendation**
- Genetic counselors underestimated this barrier

### Quotes

*“I still don't even know what we would do, honestly, to get the ball rolling on that, if we wanted to....I think if we had been at the ENT, and they said, ‘We schedule it. You just need to go here on this date and then you'll have your results in a couple weeks.’ We probably would've been a lot more likely to say, ‘Sure, let's just go ahead, why not?’” - GTEST Deferrer*

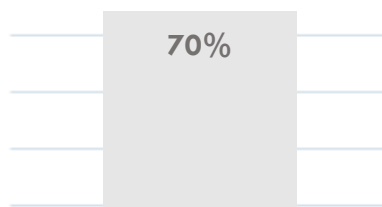
# Caregivers' experiences with genetic testing backs up these two barriers determined by clinicians in 2 hospital's operational processes

## Wait Time for COUN Appointment



Percent of INT-1 to INT-5 with wait time for COUN appt of 3 months or more

## Insurance Authorization Delay



Percent of INT-1 to INT-5 COUN patients under institutional billing (hospital policy, preference for Iowa lab)



## Conclusions

- These structural barriers are more difficult to address
- Long wait times and complicated processes can deter caregivers who go to make an appointment – too much of a hassle compared to the benefit
- Accepters of genetic testing were very unlikely to receive care via time-consuming path of waiting for appointment with general genetics clinic plus institutional billing

## Quotes

- *“I wanted to do the genetic testing. Actually, I already started a process. It's just that the clinic that we called, the one that our pediatrician recommended, they said they don't have availability until next year.” - GTEST Deferrer*

# This barrier currently not significant but needs to be monitored ongoing

## Phase I research pegged this barrier as low

- Genetic counselor's workarounds at all five INTs keep this barrier low
- Genetic counselors find self-pay deals and financial assistance programs from commercial labs and even seek assistance from charities for caregivers whose insurance denies genetic testing

## Caregiver research confirms Phase I findings

- Genetic testing Accepters had low OOP ranging from \$0 to \$300, confirming reports from genetic counselors in Phase I interviews that few patients who want to pursue GTEST have a financial barrier
- Further confirmation comes from a CI center with a high volume of HL GTEST, in which 8% did not pursue because insurance denied from 2018 -2022

## The barrier should be monitored ongoing

- This barrier is currently low, with a finalized estimate of 10%
- However, the barrier could increase if growth in GTEST volumes strain genetic counselor resources for self-pay workaround, especially for hospital centers requiring institutional billing
- An important next step is to educate clinicians on this size

# Four barriers hypothesized by clinicians were minimal to nonexistent for caregivers of children with severe to profound hearing loss

## Caregiver Blood Test Concern

- While a few (3 of 12) caregivers said blood draw made them uncomfortable for a baby, only 1 reported it was a contributing factor to deferring GTEST
- A couple of caregivers (2 of 12) offered that blood draw was a positive factor since simple and noninvasive
- Clinicians overestimated this barrier

## Caregiver Worry Re Medical Record

- None of the caregivers expressed any concern that GTEST would impact child's future health insurance, life insurance, or school record
- AUDs overestimated this barrier

## Caregiver Core Beliefs

- Only 1 of 12 caregivers reported GTEST conflicted with their moral or religious beliefs: "science can go too far"
- No caregivers felt GTEST implied child needed to be fixed (although no Deaf caregivers in sample)
- No caregivers wanted to wait until child was old enough to make the GTEST decision, perhaps because of this severe to profound population
- OTOLs and AUDs overestimated this barrier

## Caregivers Need to Discuss

- No caregivers reported needing to discuss GTEST with each other before proceeding
- Often, they made their decision on the spot after clinician proposed GTEST
- Clinicians correctly estimated this barrier as small



# Genetic test results help personalize patient care, understand possible outcomes

Perceived GTEST Benefits at Time of Decision

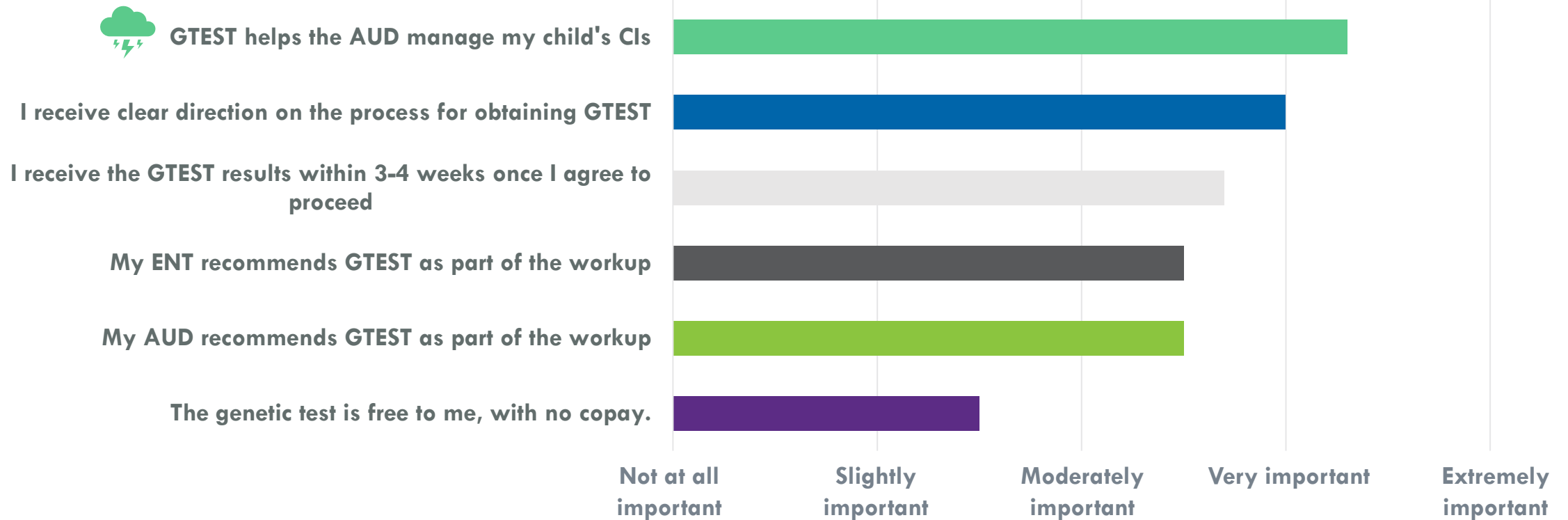


## Conclusions

- At time of decision, two-thirds of Accepters and Deferrers saw GTEST benefit of family planning
- At time of decision, Accepters were far more likely to see benefit in understanding HL cause
- The catalyst for two-thirds of Accepters was treatment management, getting clarity on the child’s health for the present and preparing for the future
- GTEST Deferrers, however, did not perceive a treatment management benefit at time of decision (due to lack of education)

# Catalysts ranked by those who did not pursue genetic testing

## Catalysts for Deferrers to Have Pursued GTEST at Time of Recommendation



# Providing education and support to clinicians and caregivers can help reduce barriers to genetic testing for patients with congenital hearing loss

- Highlight **clinical evidence** showing **benefits of early genetic testing**
- Address **process issues and structural constraints** to obtaining a genetic test

# Comprehensive Genetic Hearing Loss Panels are Now Available to Diagnose Inner Ear Protein Deficiencies

**PREVENTION GENETICS** part of EXACT SCIENCES

HOME | CONTACT US | FORMS | myPrevent

TESTING | BILLING | RESOURCES | HOW TO ORDER | DNA BANKING | ABOUT US

Program Overview  
Clinical Features  
Genetics  
Testing Strategy  
Criteria For Test  
Ordering

**Decibel THERAPEUTICS** **amplify** GENETIC TESTING PROGRAM

**Amplify™ Sponsored Testing Program**

Program Overview

Test Code: 15747 269 Genes

ORDER TEST KITS

Sponsored

**Blueprint Genetics** Diagn

Blueprint Genetics / Akouos Resonate Program

GeneDx

Hearing Loss Panel

ADD TO ORDER ADD TO FAVORITES

New York Approved

**IOWA** Molecular Otolaryngology

Test Menu/Requisitions Clinical Diagnostic Services Research

Helping to Make Lives Better

Home // Clinical Diagnostic Services // Hearing Loss / OtoSCOPE® OtoSCOPE® Genetic Hearing Loss Testing v9

**OtoSeq** Cincinnati Children's

Cincinnati Children's now offers OtoSeq, our next-generation sequencing panel of 23 genes associated with hearing loss. The panel was designed to identify approximately 80 percent of the genetic causes of early onset sensorineural hearing loss. Recently issued practice guidelines by the American College of Medical Genetics support the use of next-generation sequencing tests such as OtoSeq in the comprehensive evaluation of patients with hearing loss.

Academic

INVITAE Individuals Providers Search Sign in Order now

Test Catalog > Invitae Comprehensive Deafness Panel

Invitae Comprehensive Deafness Panel

Test description

Genetic testing for genes associated with syndromic and non-syndromic deafness. These are genetically heterogeneous disorders characterized by mild to profound deafness in early adulthood, childhood, or infancy. The genetic heterogeneity associated with these conditions can make it difficult to use phenotype as the sole criterion to select a definitive cause.

TEST CODE: 55009

TURNAROUND TIME: 10-21 calendar days (14 days on average)

PREFERRED SPECIMEN:

Commercial

UHealth UNIVERSITY OF MIAMI HEALTH SYSTEM

COVID-19 Information Appointments Referring Physicians International Patients Pay a Bill Donate Now Patient Login

FIND A DOCTOR TREATMENTS LOCATIONS PATIENTS & FAMILIES Q SEARCH

University of Miami Health System / Treatments and Services / Genetics / Sensorineural Hearing Loss and Deafness

**Sensorineural Hearing Loss and Deafness**

Doctors Locations Clinical Trials Classes & Events

Sensorineural hearing loss or deafness occur due to problems with the auditory nerve or the part of the brain responsible for hearing. In children with hearing loss or deafness, these problems are often caused by genetic conditions.

Tests

Newborn Hearing Screening

Children who are born with hearing problems are often identified during the newborn hearing screening. This test is required by many states, including Florida, and is completed while your child is in the hospital after birth.

PARTNERS HEALTHCARE PERSONALIZED MEDICINE

ABOUT LABORATORY FOR MOLECULAR MEDICINE BIOBANK GENOMICS CORE BIOBANK EDUCATION

Home > Laboratory for Molecular Medicine > Ordering > Hearing Loss > OtoGenome Panel (110 Genes)

Ordering LMM Tests

- Cardiomyopathy
- Exome/Genome Services
- Hearing Loss
- Genetics Test
- OtoGenome Panel (110 Genes)
- Comprehensive DFNB1 and STRC Panel
- Comprehensive STRC / Deafness and Male Infertility Syndrome Test
- SLC26A4 Gene Sequencing
- WFS1 Gene Sequencing
- POU3F4 Gene Sequencing

Order OtoGenome™ Test (110 Genes) for Hearing Loss and Related Syndromes

Hearing loss has an incidence of 1 in 250 births and over half of these children have a genetic etiology. The comprehensive approach of the OtoGenome™ Test now makes it possible to sequence 109 genes known to cause nonsyndromic hearing loss and syndromic that can present an nonsyndromic such as Usher, Pendred, Jervell and Lange-Nielsen (JLNS), and Branchio-Oto-Renal syndrome (BOR). Read more about this test.

Requestion Form

All samples must be accompanied with a completed requisition form. Please make sure any identifiers used on the specimens are provided on the paperwork. Consent page should be signed by a health care provider. Any incomplete or missing paperwork may delay the start of testing.

Document: Hearing Loss and Related Syndrome Requisition Form

**PREVENTION GENETICS** part of EXACT SCIENCES

HOME | CONTACT US | FORMS | myPrevent

TESTING | BILLING | RESOURCES | HOW TO ORDER | DNA BANKING | ABOUT US

Order Options and Pricing

PRINT Test Description

Summary and Pricing

Clinical Features and Genetics

Citations

Ordering/Specimens

**Hereditary Hearing Loss and Deafness Panel**

Summary and Pricing

Test Method

Exome Sequencing with CNV Detection

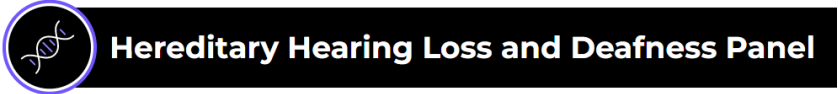
New York State Approved Test

PANEL AVAILABLE VIA PGenome Sequencing REFLEX TO PGenome AVAILABLE FOR THIS PANEL

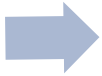
Test Code	Test	Panel CPT Code	Gene CPT Codes	Base Price
5063	Genes x (207)	81479	81252(x1), 81403(x1), 81404(x5), 81405(x4), 81406(x6), 81407(x7), 81408(x5), 81479(x385)	\$1290

ORDER OPTIONS AND PRICING





Clinician determines eligibility



Buccal swab sent to PG



Results available in 18 days



Genetic counseling available for Patient/CG



**Decibel receives deidentified data:**

- Overall testing activity per site
- Number of positive variants per site



Prevention Genetics' Genetic Counselors available to ordering clinician throughout entire process

