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

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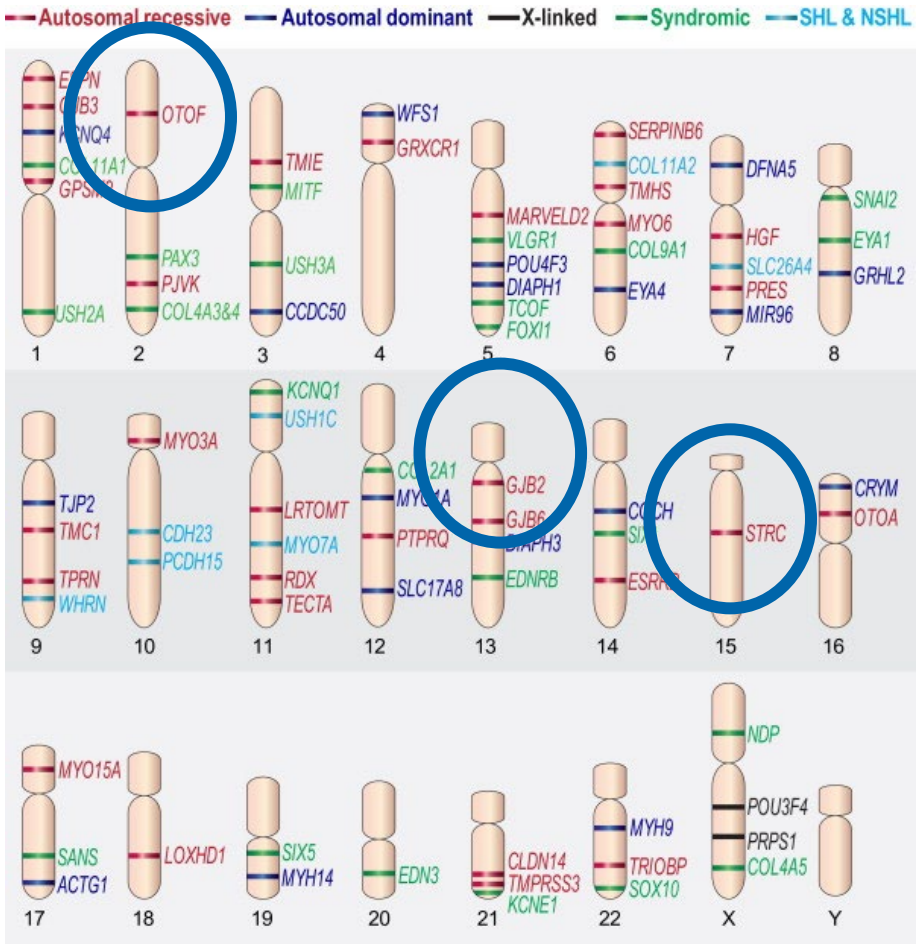
Genetic Protein Deficiencies Cause Most Congenital Hearing Loss



Raviv et al. (2010) Ann NY Acad Sci

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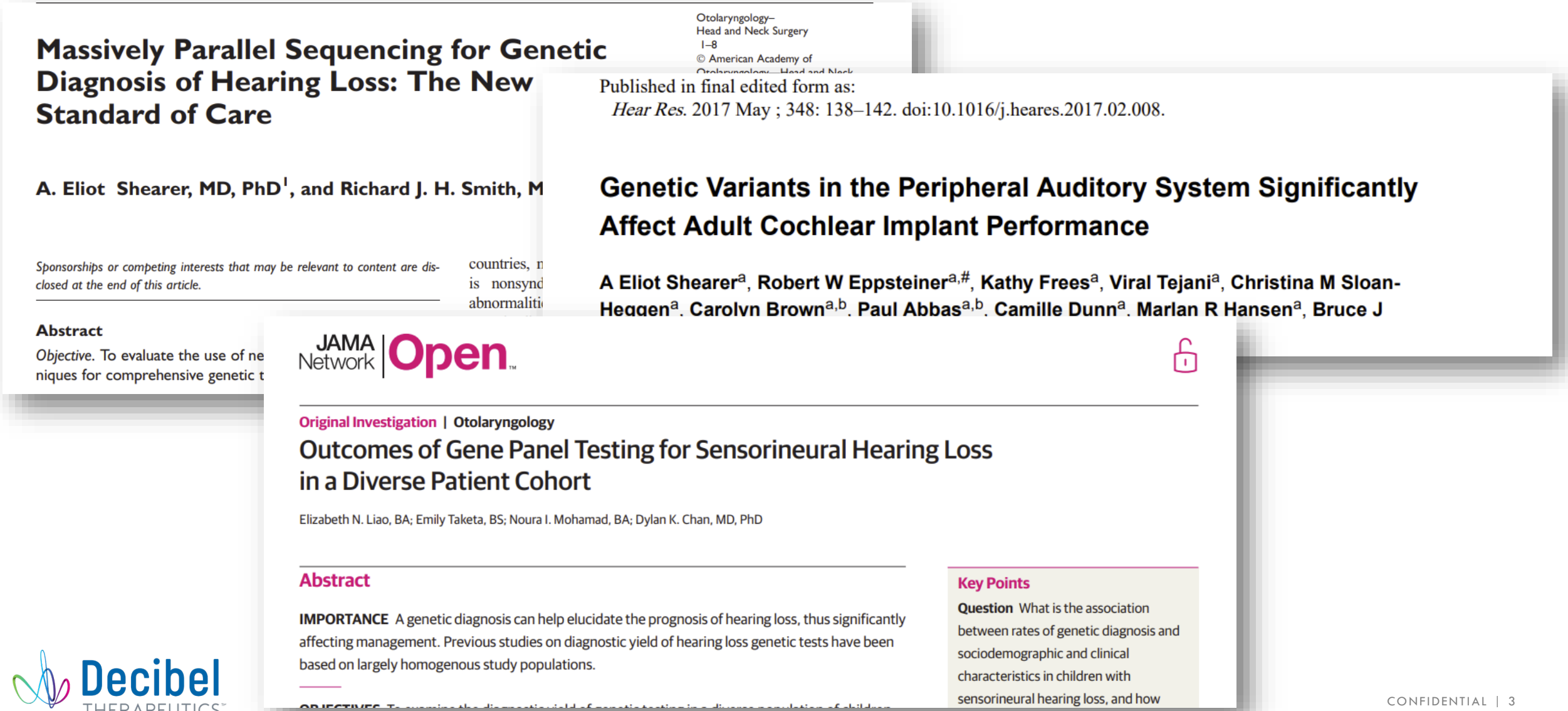
Current evidence shows genetic testing is broadly useful in improving clinical management of children with hearing loss



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1–8
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Massively Parallel Sequencing for Genetic Diagnosis of Hearing Loss: The New Standard of Care

A. Eliot Shearer, MD, PhD[1], and Richard J. H. Smith, M

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

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Abstract

Objective. To evaluate the use of ne
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Genetic Variants in the Peripheral Auditory System Significantly Affect Adult Cochlear Implant Performance

A Eliot Shearer[a], Robert W Eppsteiner[a,#], Kathy Frees[a], Viral Tejani[a], Christina M Sloan-Heggen[a], Carolyn Brown[a,b], Paul Abbas[a,b], Camille Dunn[a], Marlan R Hansen[a], Bruce J

JAMA Network | Open

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.

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

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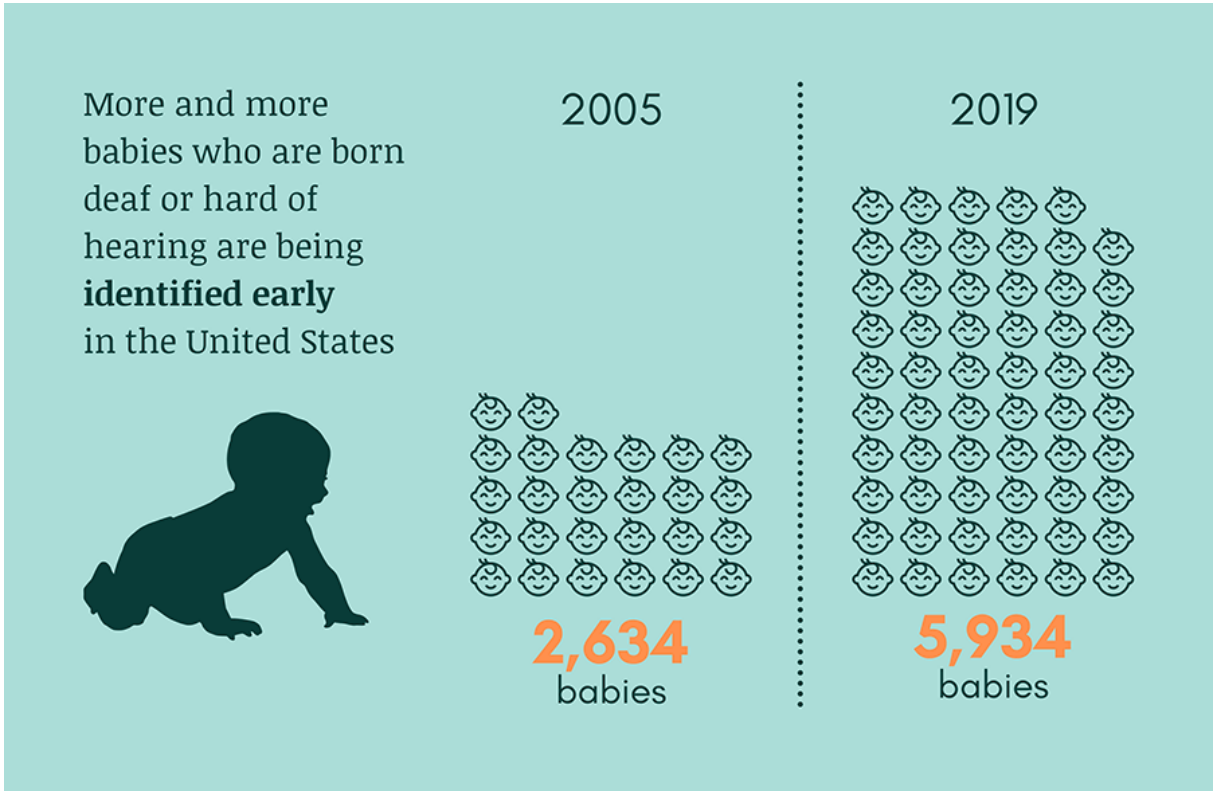
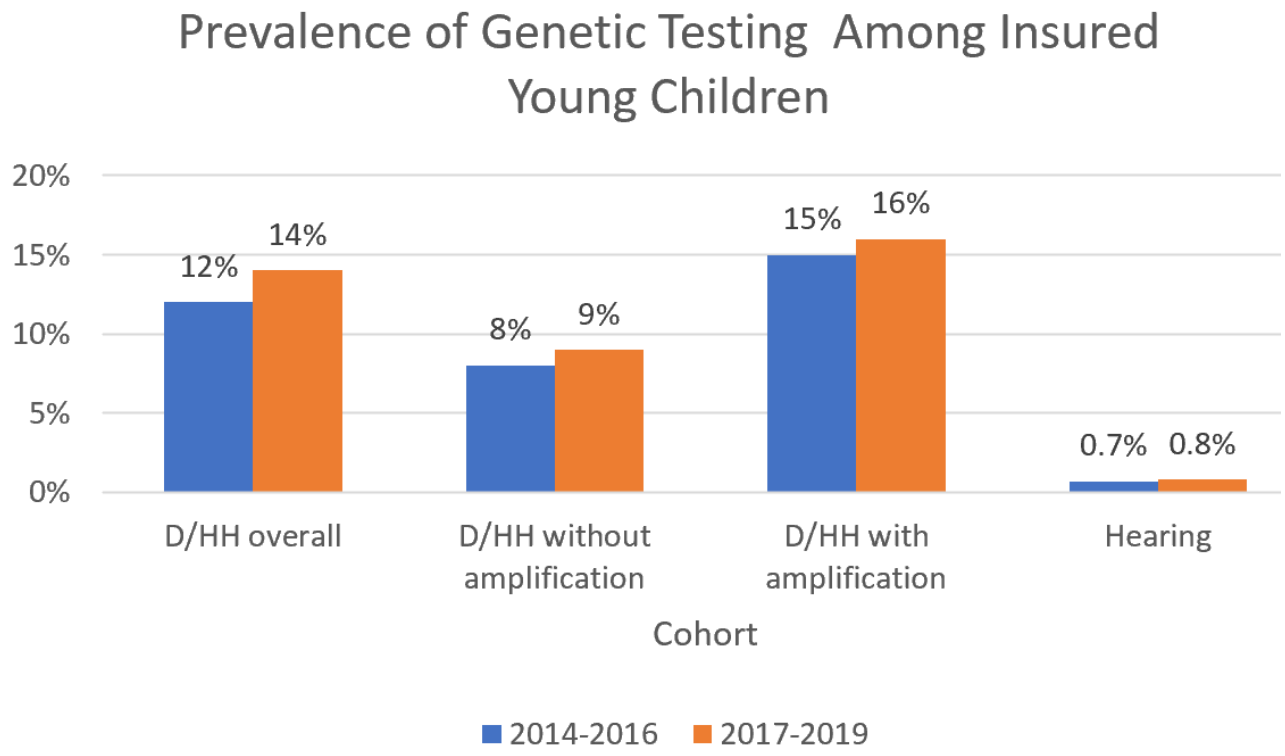
Early Identification is Key to Catalyzing Novel Therapeutics

Massive Progress Over the Last 2 Decades



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

Genetic Diagnosis is a Work in Progress



S. Charania, S. Grosse, K. Dundon EHDI 2022 Poster Study

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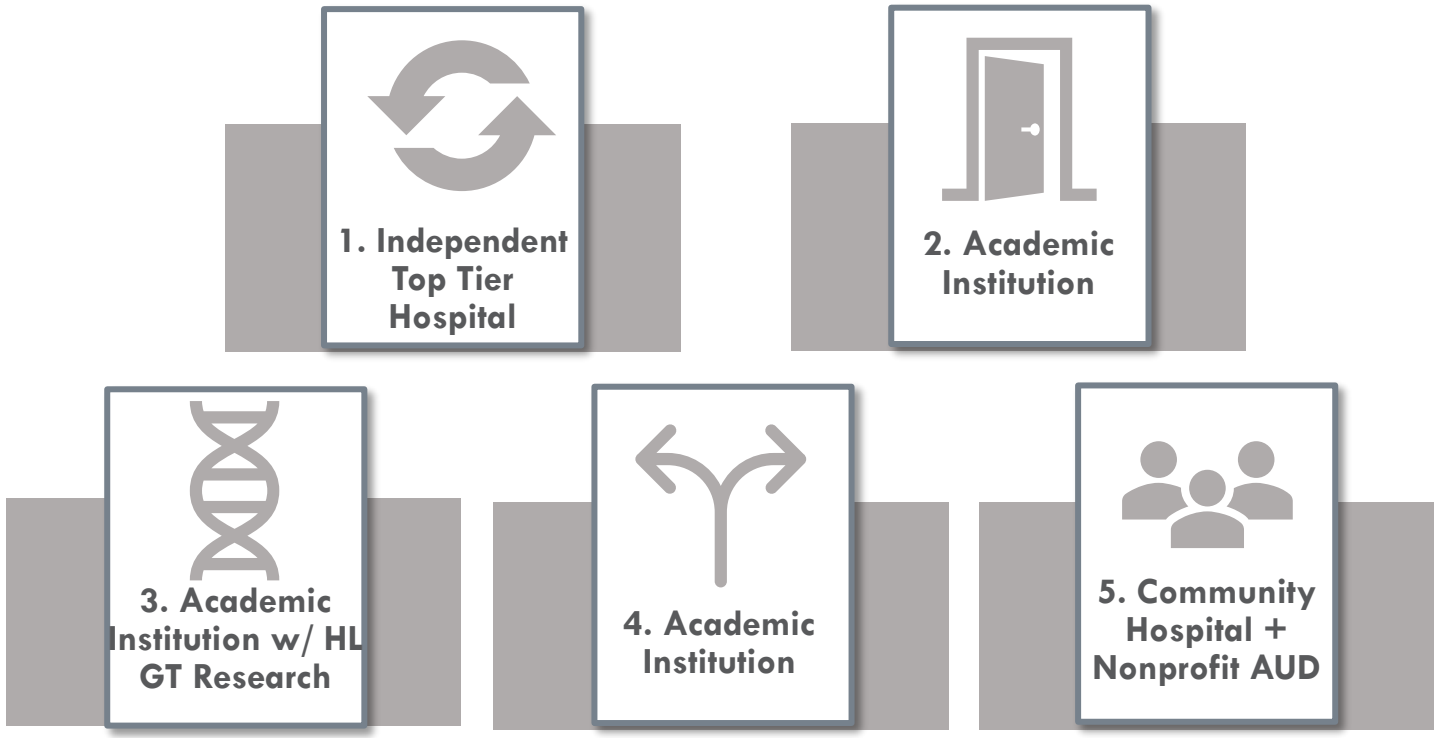
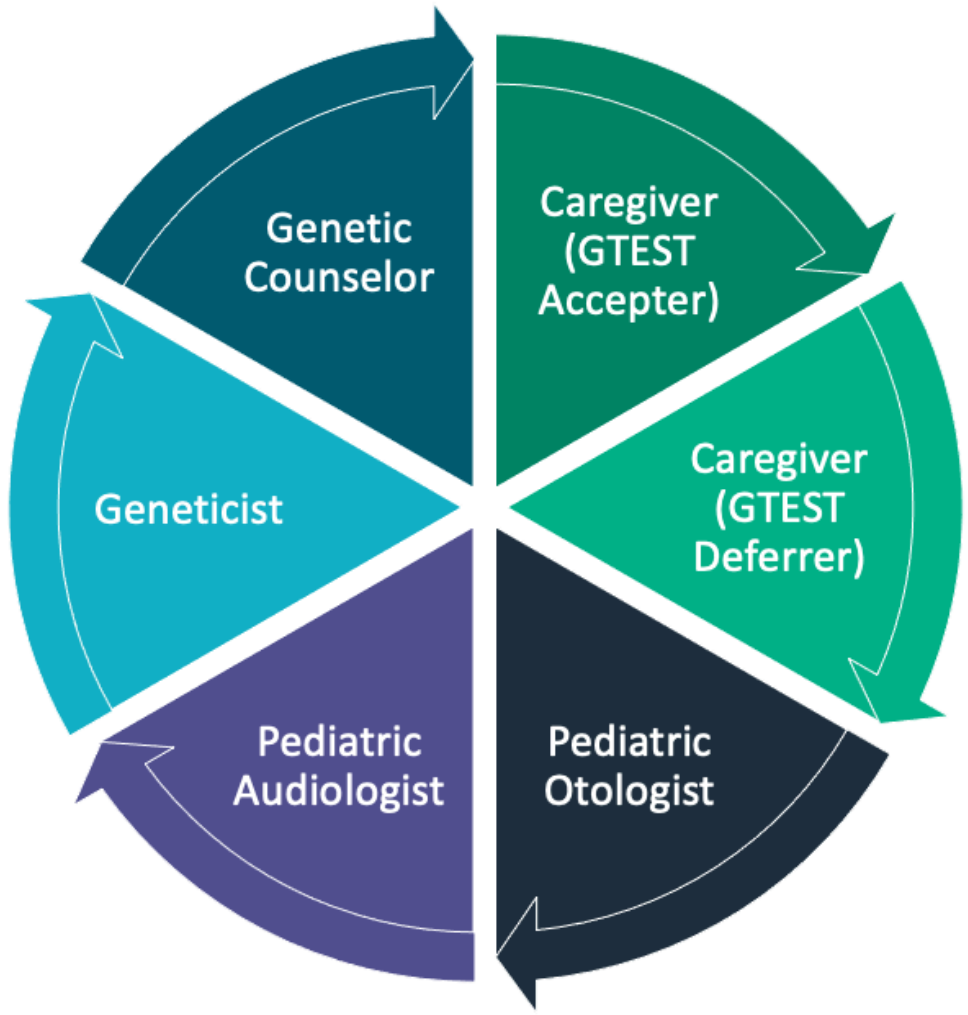
Clinicians and Caregivers Selected from 5 Different Children's Hospitals

All 5 Otologists interviewed treated at least one caregiver in sample





Source: Independent, blinded, market research conducted by Auditory Insight

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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

SOURCE: Auditory Insight analysis

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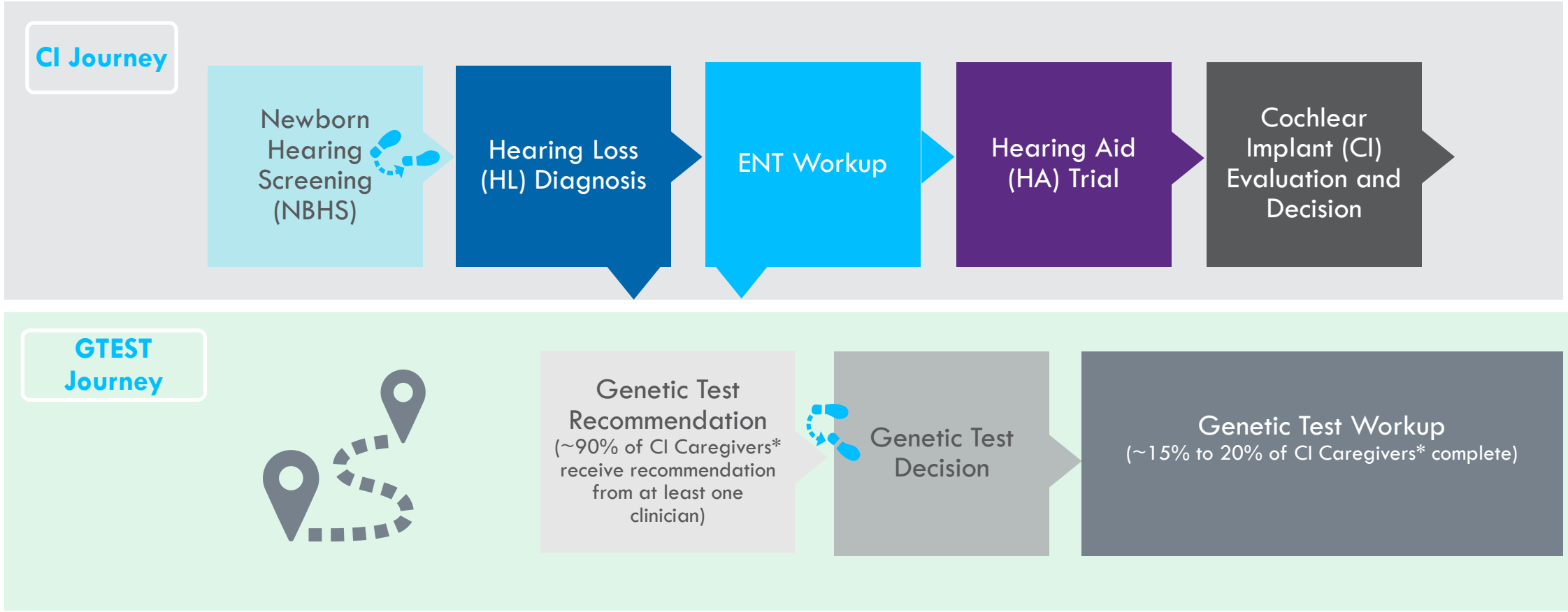
Hearing loss treatment and genetic testing are typically concurrent, but often not integrated



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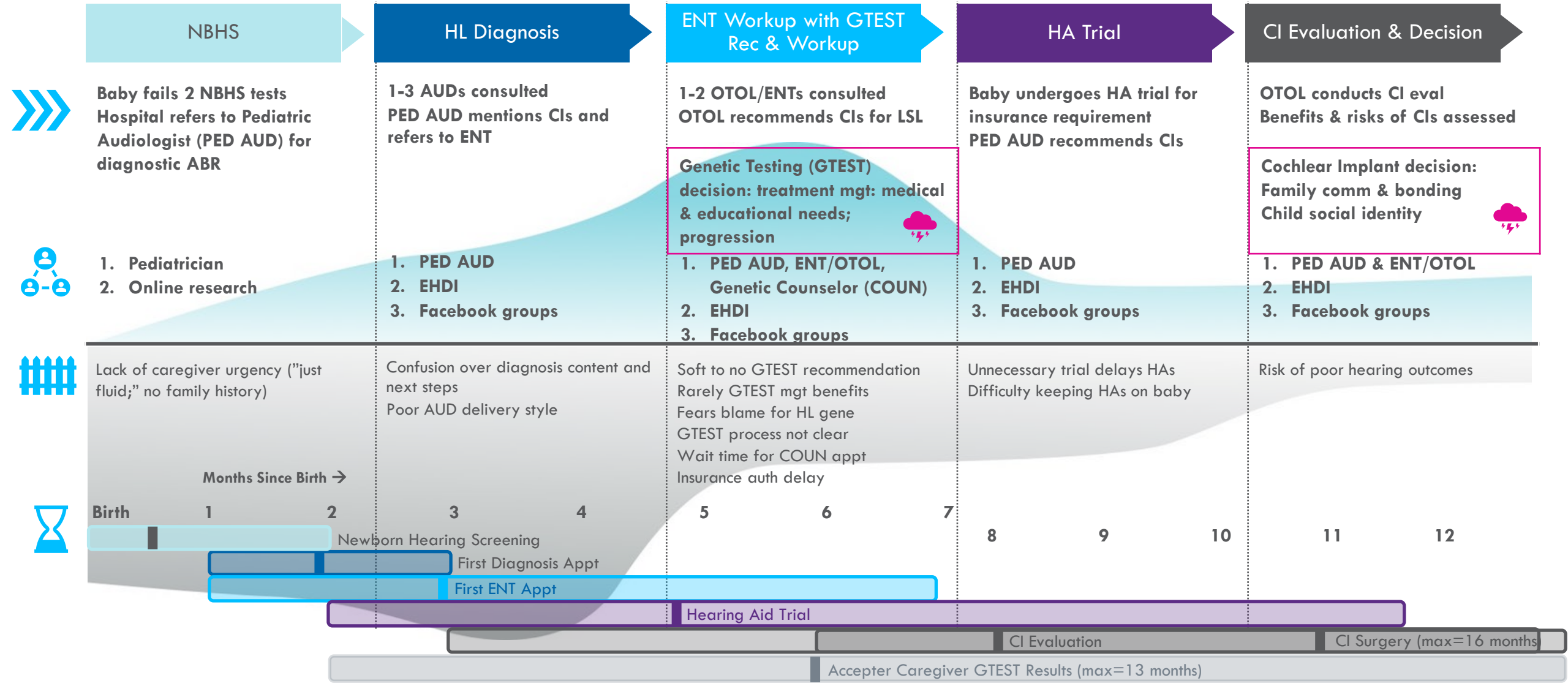

KEY:

MIN AVG MAX

KEY:

Positive emotions which can be amplified

Negative emotions which function as a barrier

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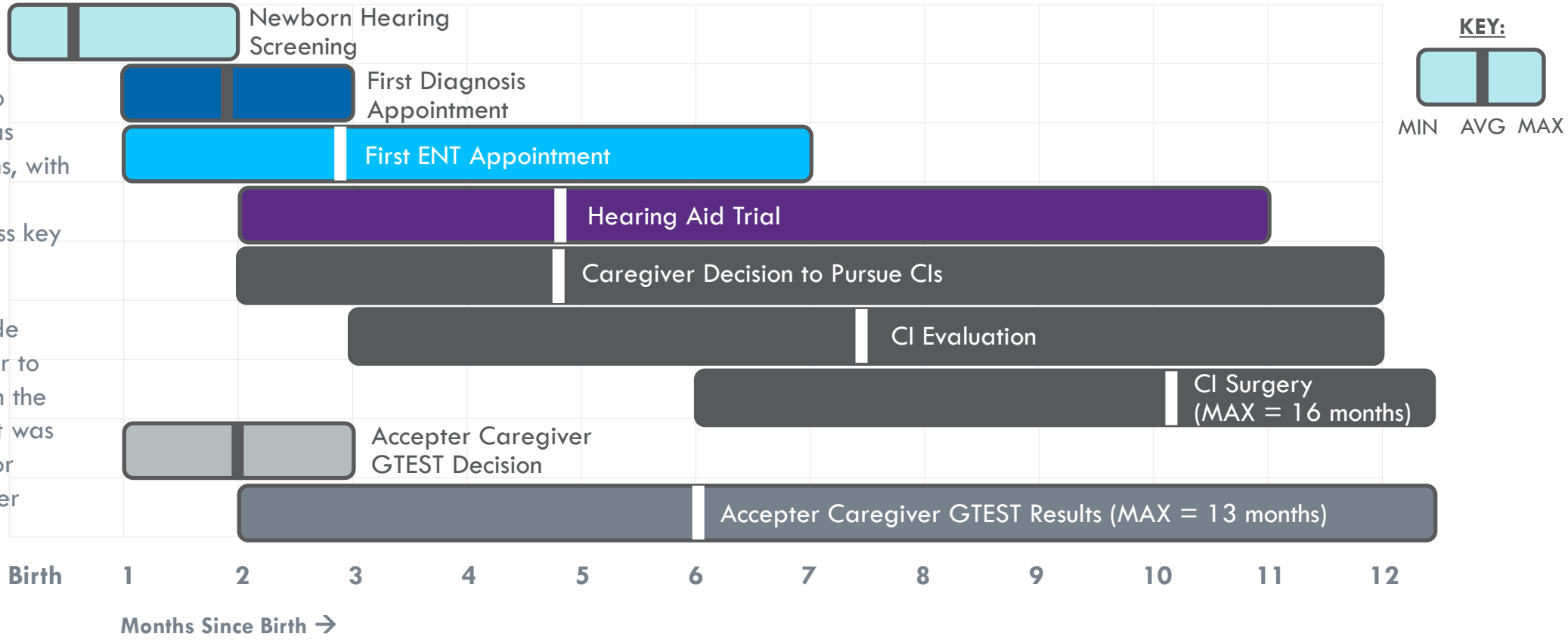
Time Intervals, by Journey Stage

- 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.

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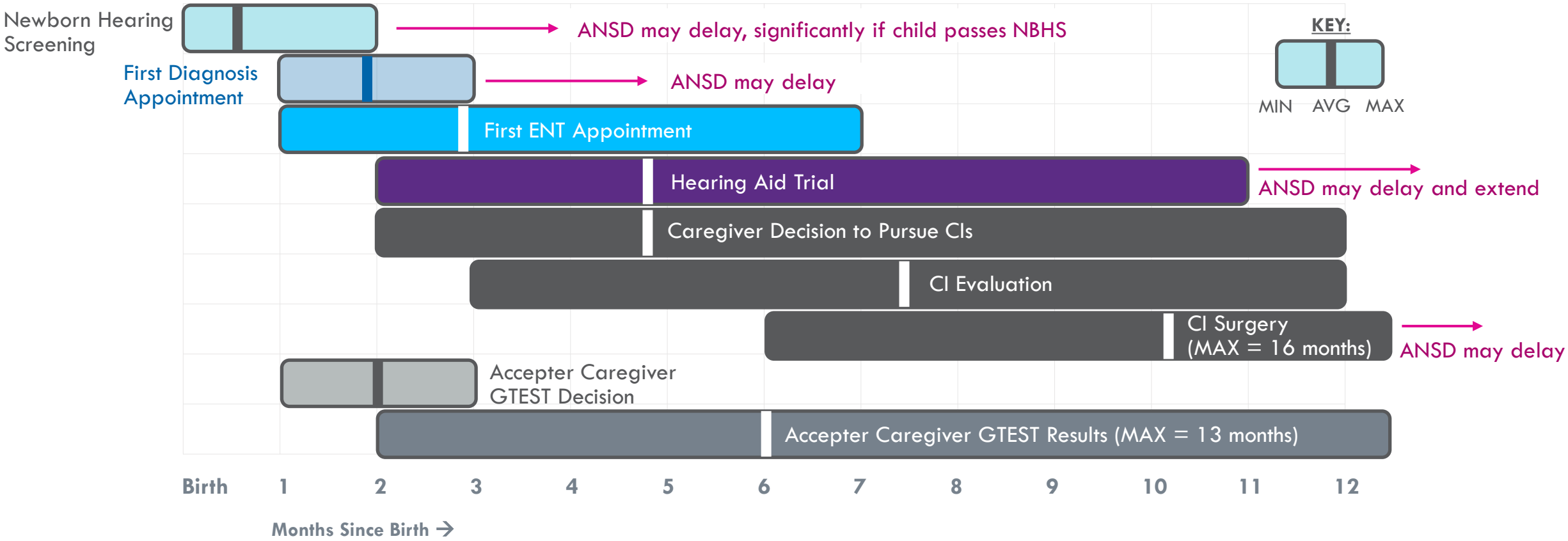
Auditory Neuropathy Spectrum Disorder (ANSD) may Extend Patient Journey in Multiple Stages



CI Caregivers* Emotions, by Journey Stage

	Newborn Hearing Screening	HL Diagnosis	GTEST Decision & ENT/GTEST Workup	HA Trial	CI Evaluation and Decision
Intensity of Emotions	6	13	27	8	9
Positive emotions which can be amplified	Intuition/observations of HL Sees ASL as option	Encouraged and relieved Focused on treatment Unconditional love Diagnosis sinks in	Acceptance of diagnosis Reassurance Desire to give child LSL Focused on workup Focused on ASL option Supported by care team Hopeful for CIs Eager for info from GTEST Relieved by GTEST results Optimistic Happy Excited	Acceptance of diagnosis Hopeful HAs will help Determined Happy; Excited	Relieved eligible for CI Eager and hopeful Excited
Negative emotions which function as a barrier	Trying not to worry Disbelief Denial: observations child can hear Denial: focused on joys of newborn Concerned Worried, nervous Sadness, upset, crying Overwhelmed Grieving Shocked Guilt Alone and unsupported Scared; fear of unknown Terrified	Disbelief Confused Concerned Distracted (can't retain information) Nervous Worried At a loss (don't know anyone with HL) Sad, upset, crying Overwhelmed, overloaded Afraid Grieving Shocked Guilt Alone and unsupported Traumatized	Sad; nervous Afraid; unfamiliar w/CIs Overwhelmed Guilt (something during pregnancy caused HL and or may have passed on HL gene)	Struggling with diagnosis Nervous and worried Angry at insurance HA trial requirement	Worried Afraid (surgical risks) Overwhelmed
	35	40	17	15	6

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.

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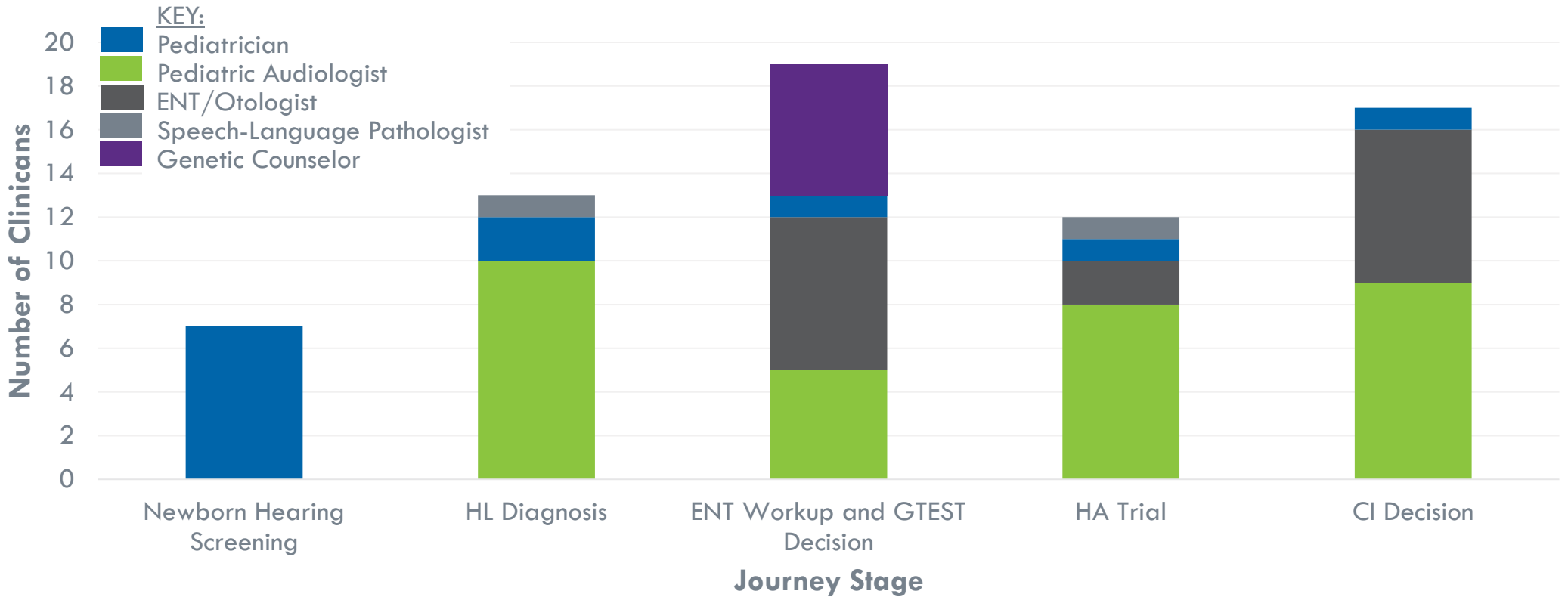
Audiologists are the most influential provider to caregivers across the journey

Pediatrician's influence rapidly wanes after NBHS

Providers Who Influence Caregivers* by Journey Stage



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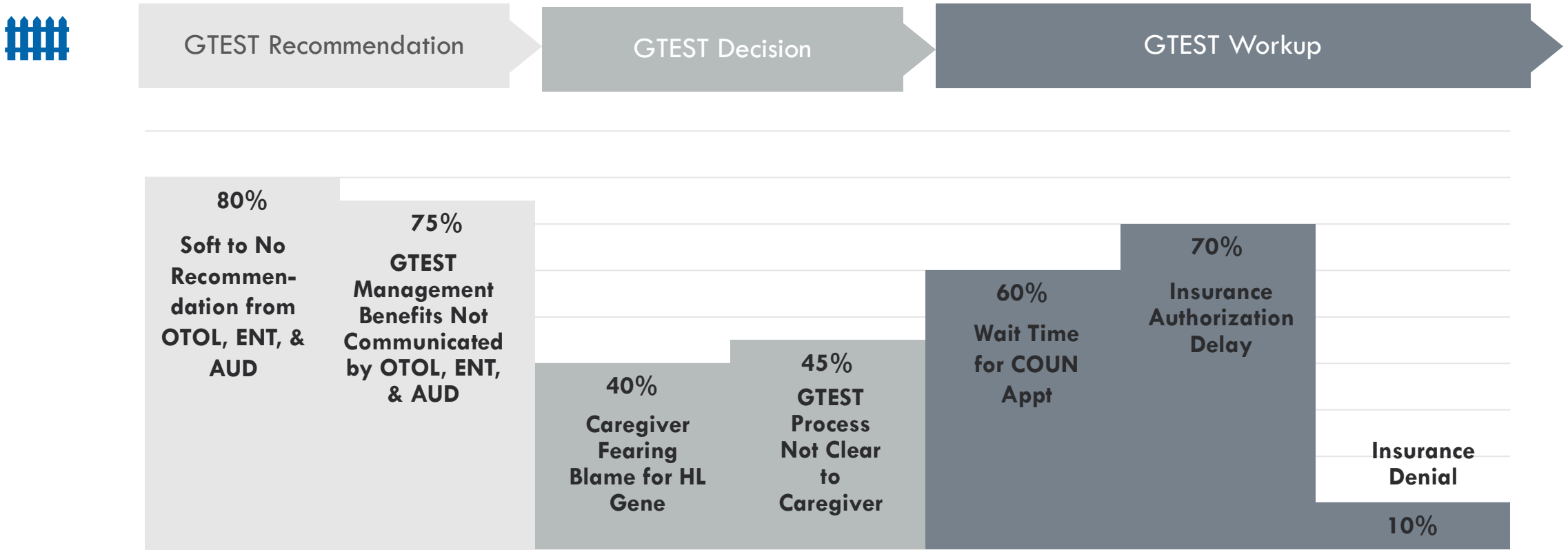
Genetic Testing Hierarchy of Barriers

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



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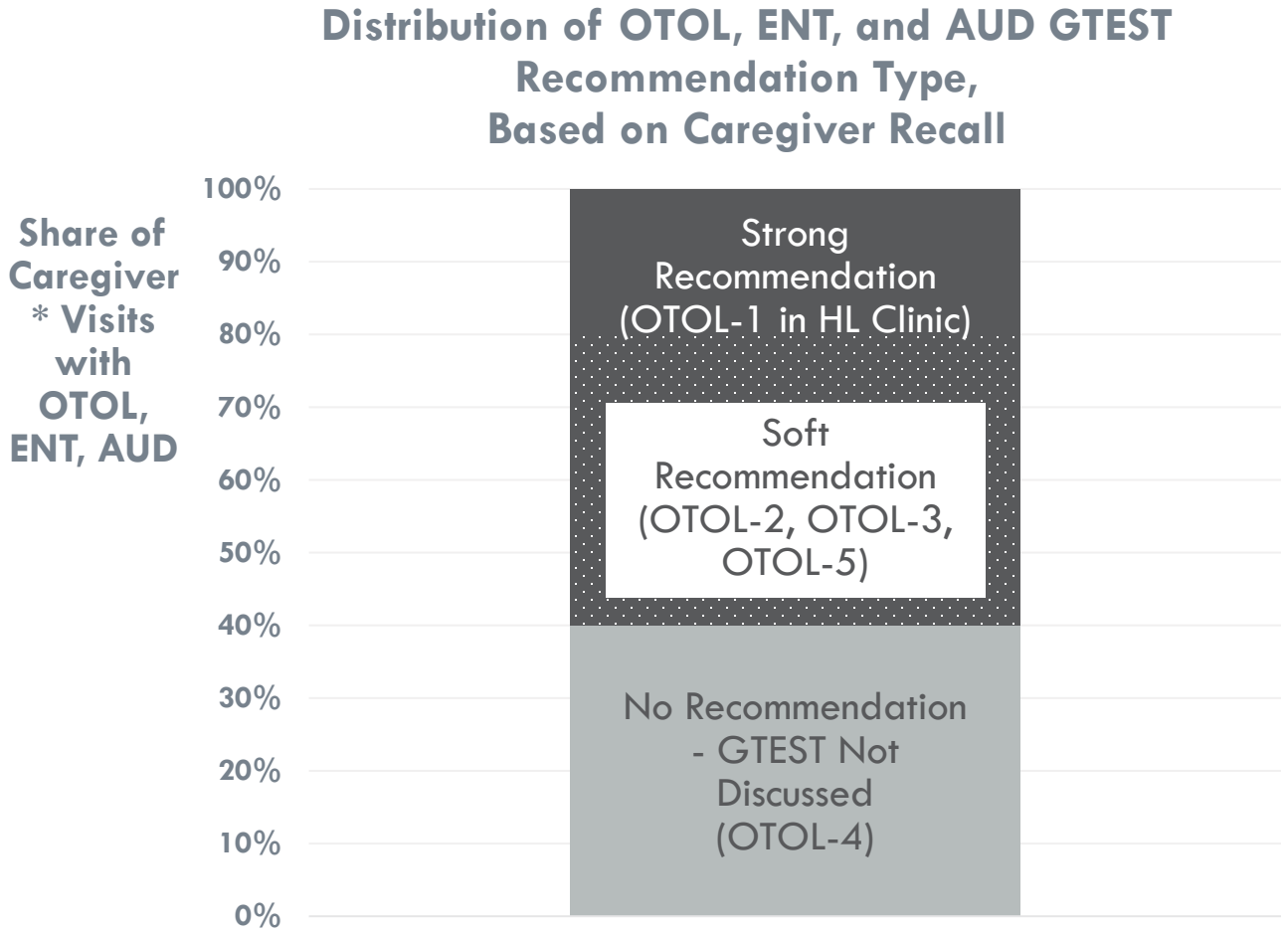
Clinicians don't make a strong recommendation for genetic testing most of the time



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

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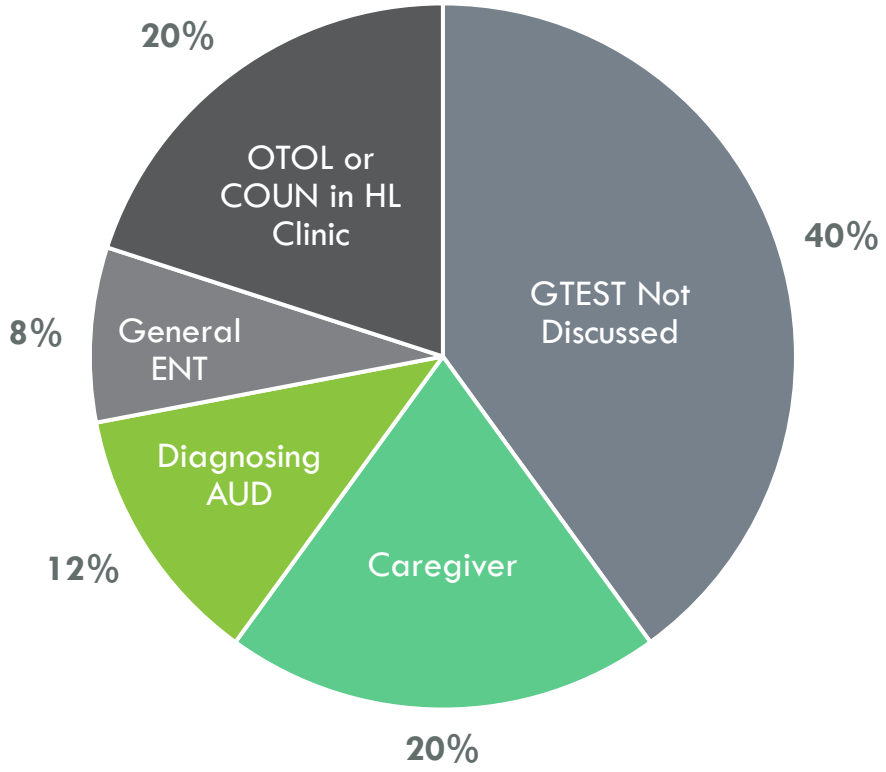
Caregivers Were Frequent Initiators of Genetic Testing Conversation

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

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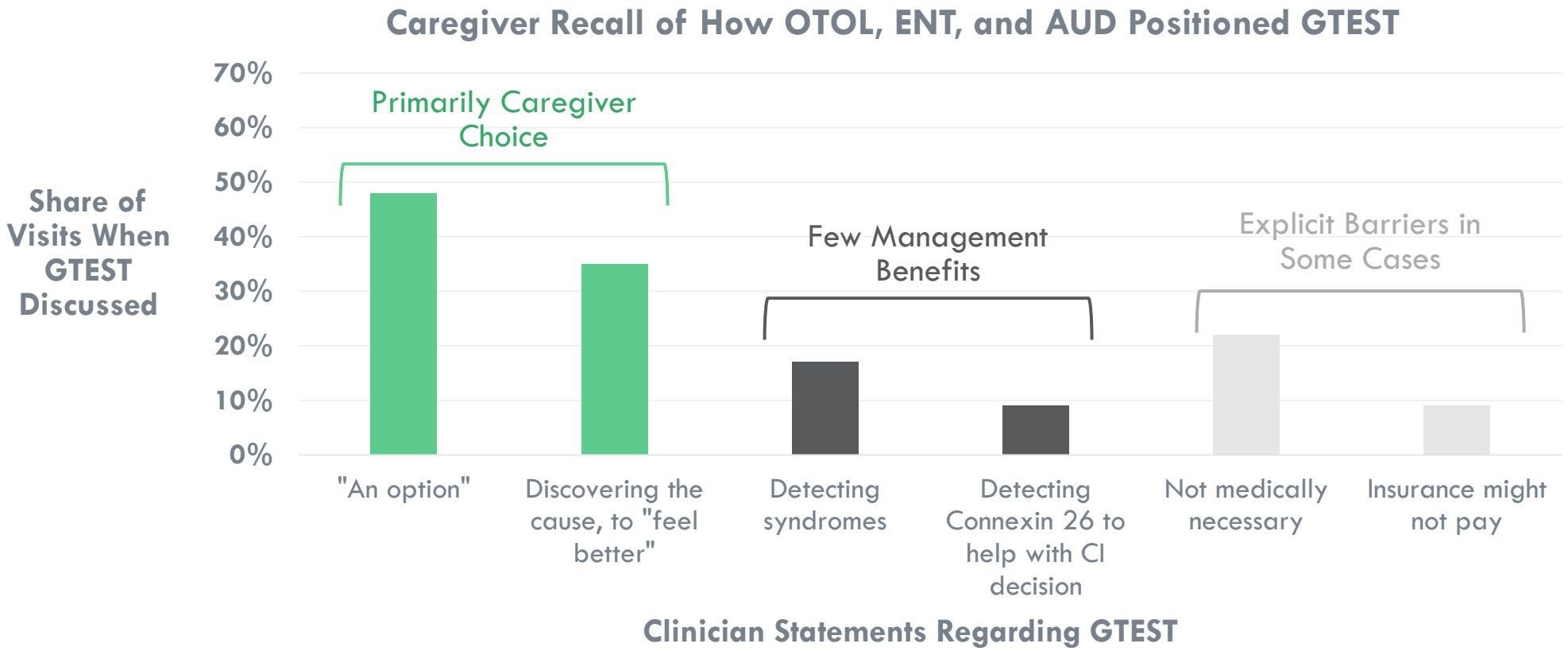
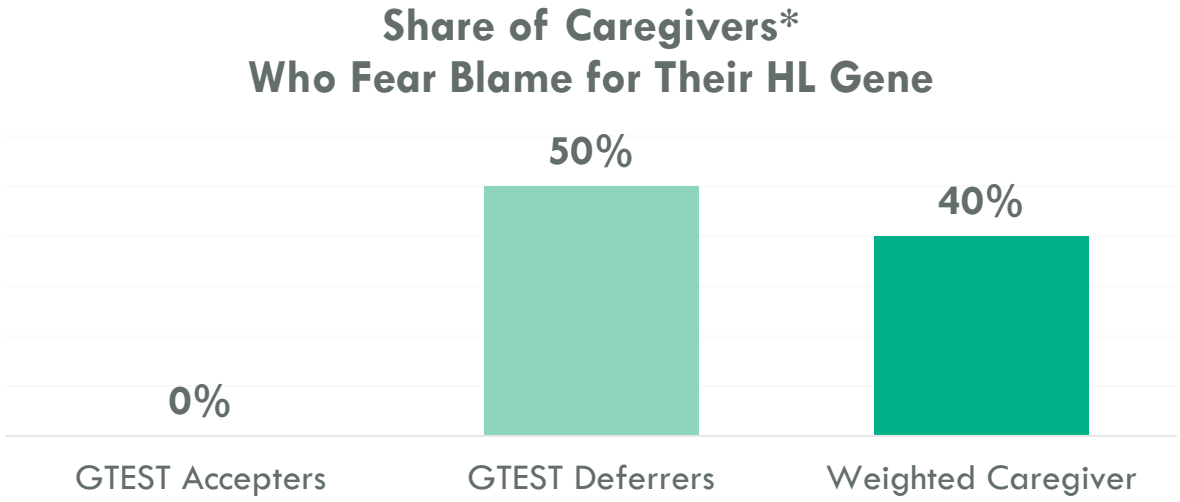
Clinicians are positioning genetic testing as an option, to find the cause to "feel better"

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



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



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 Accepter

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

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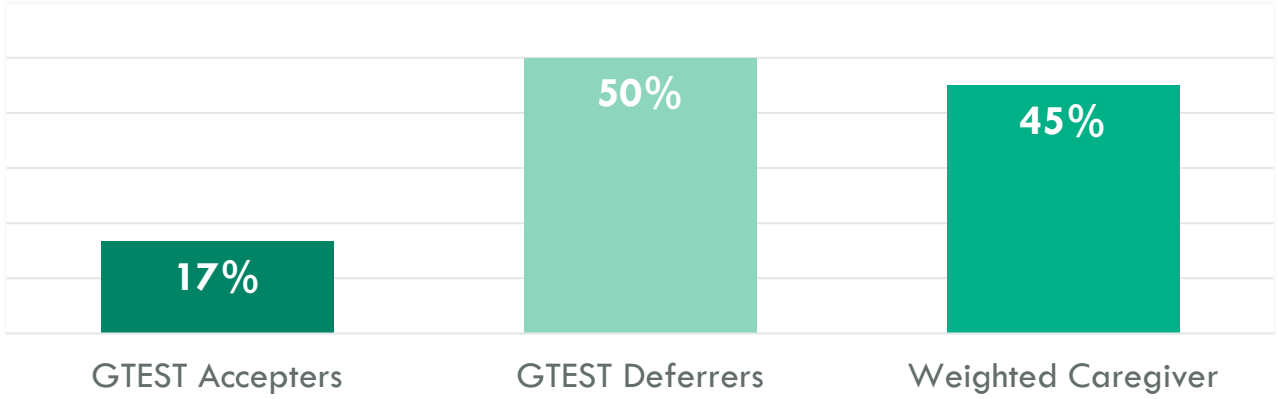
Deferrers struggled to understand how GTEST process might work

Share of Caregivers* for Whom GTEST Process Unclear



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

Conclusions

- Half of Deferrers did not feel that they received clear direction on the process, whereas only 1 Accepter 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

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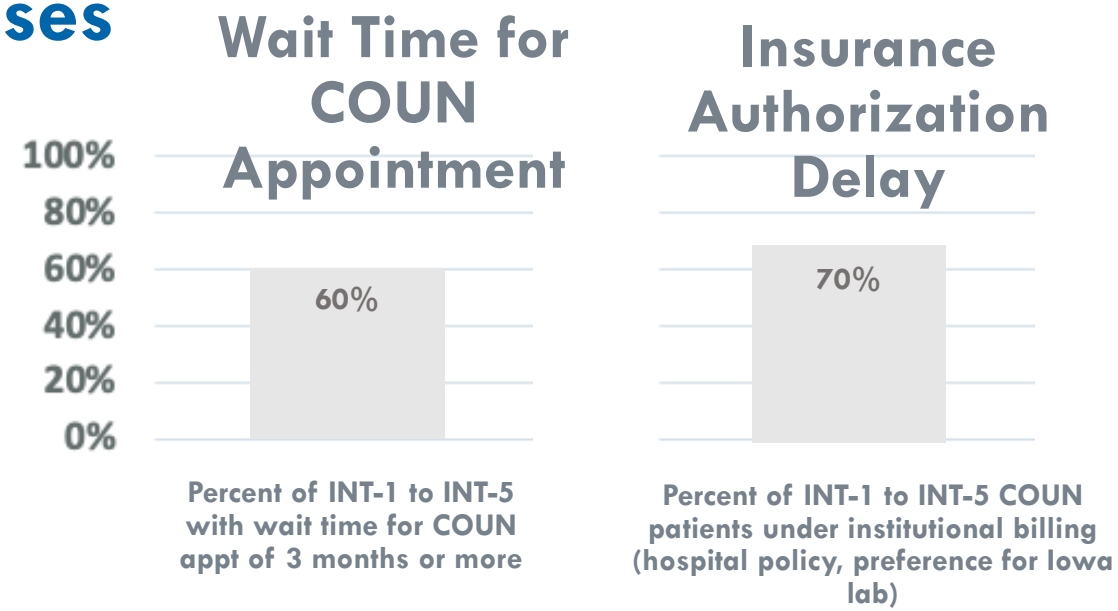
Caregivers' experiences with genetic testing backs up these two barriers determined by clinicians in 2 hospital's operational processes



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

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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

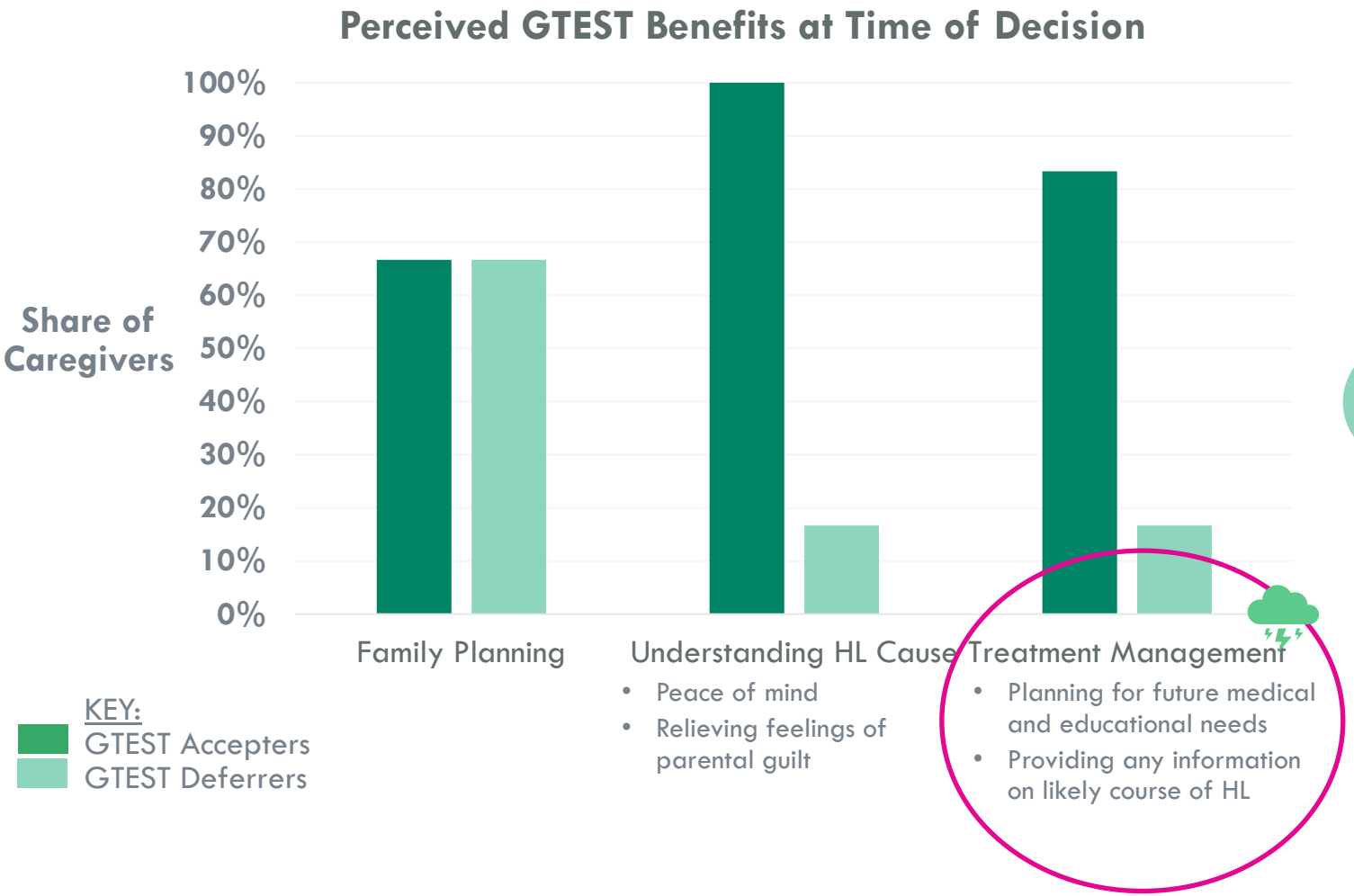
Caregiver Core Beliefs

- 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



- Peace of mind
- Relieving feelings of parental guilt

- Planning for future medical and educational needs
- Providing any information on likely course of HL

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)

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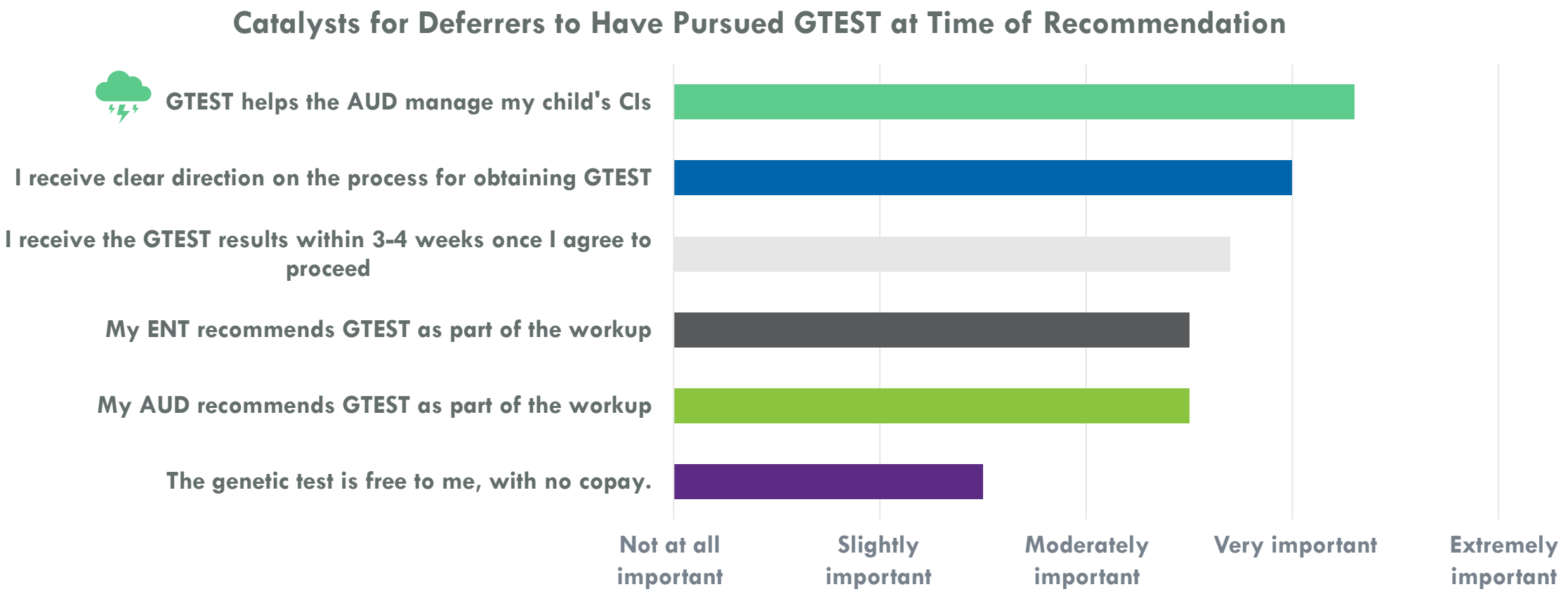
Catalysts ranked by those who did not pursue genetic testing



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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

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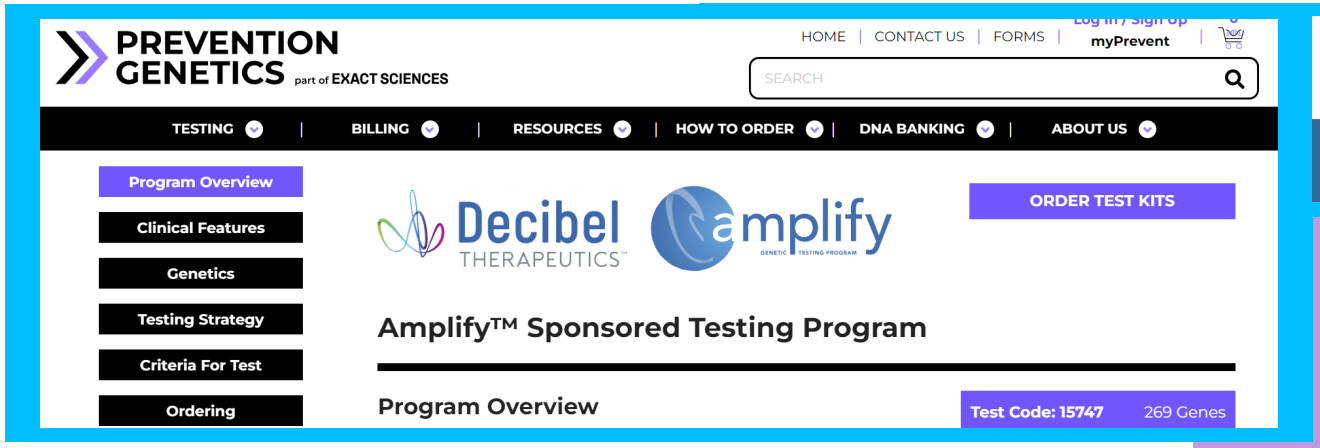
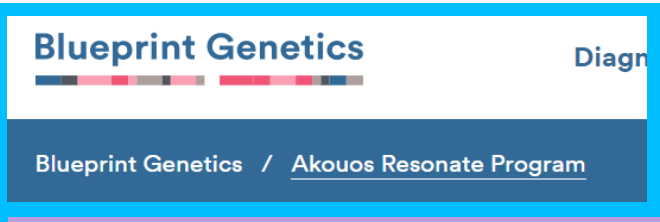
Comprehensive Genetic Hearing Loss Panels are Now Available to Diagnose Inner Ear Protein Deficiencies

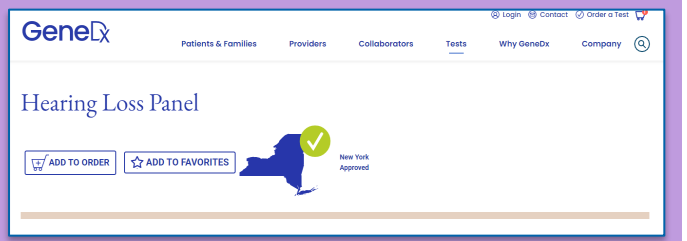
Sponsored

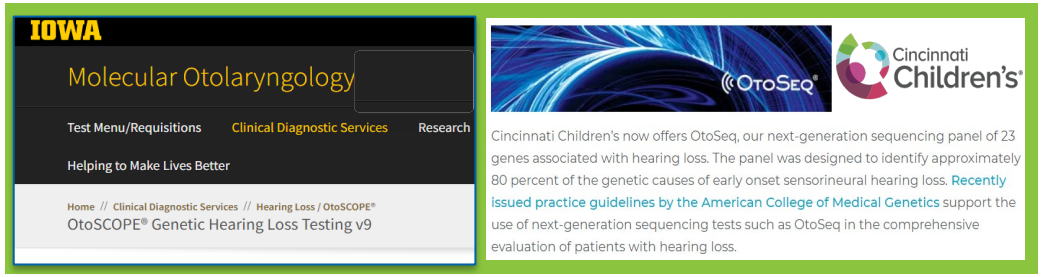
Academic

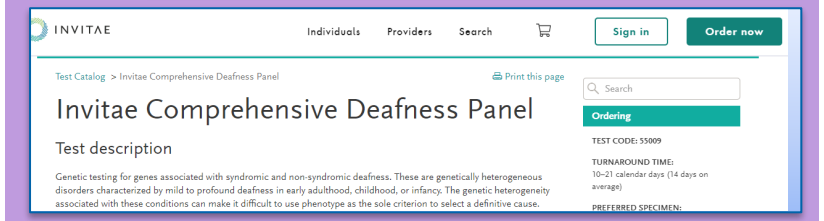
Commercial

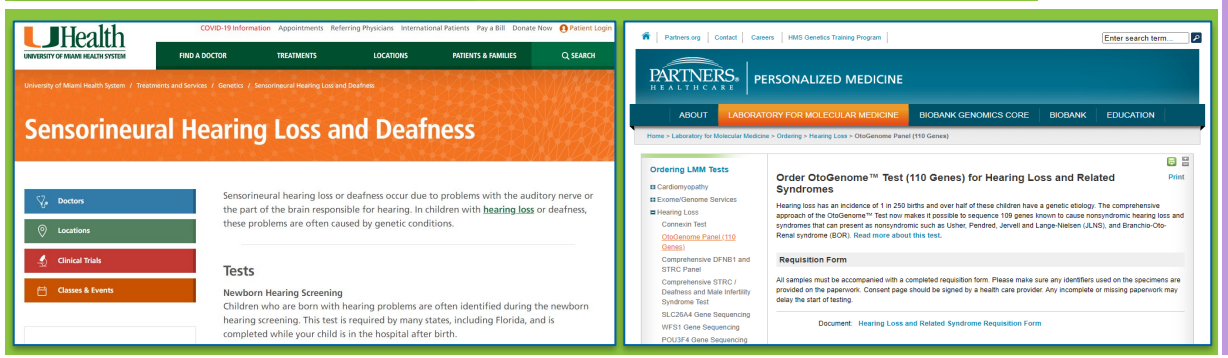


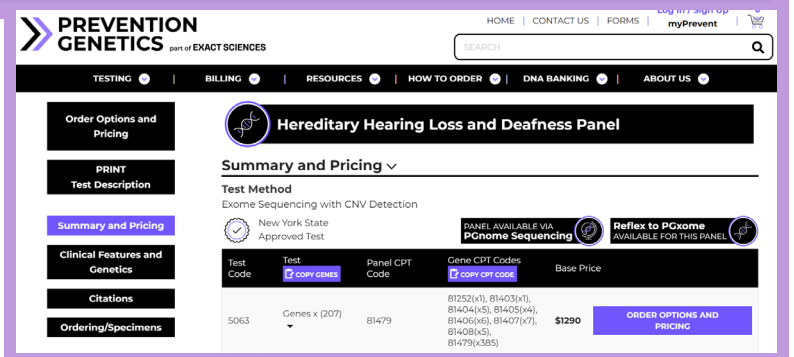












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amplify GENETIC TESTING PROGRAM

PREVENTION GENETICS part of EXACT SCIENCES

Hereditary Hearing Loss and Deafness Panel

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

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