



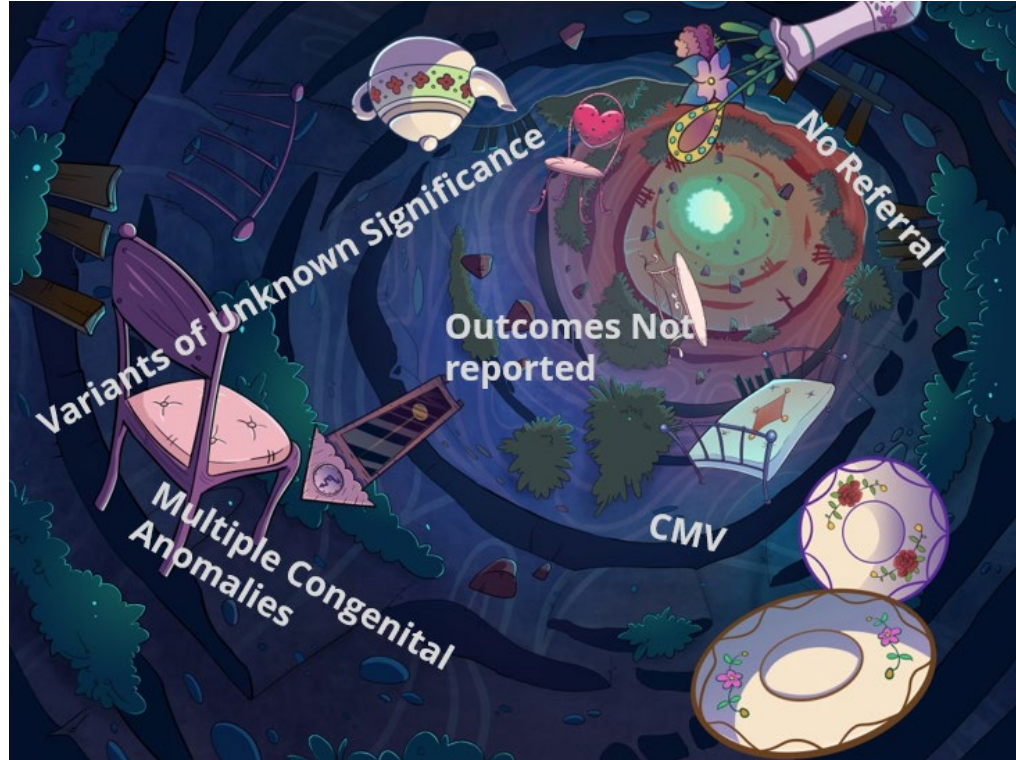
Enhancing Pathways: Streamlining Genetic Referrals for Hearing Loss to Regional Genetic Centers

Hilary Fryman, RN BSN

Disclosure

- I have no financial disclosures.

Disclaimer: The Rabbit Hole of Hearing Loss and Genetics





Genetics of Hearing Loss

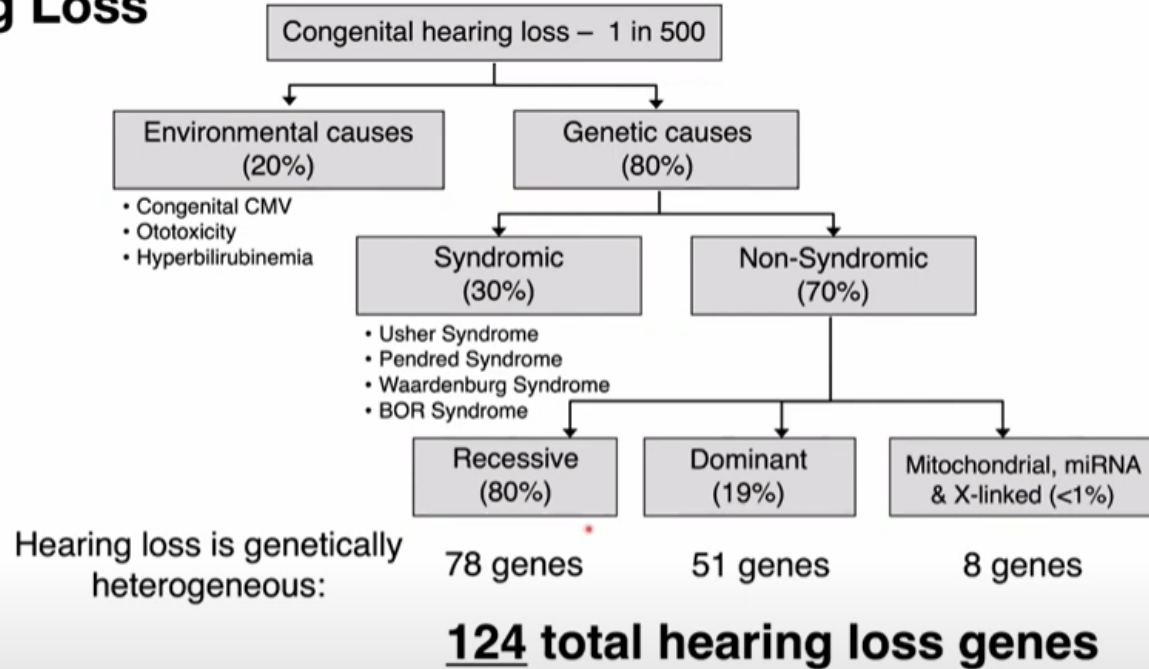
Why is this important?

“Hearing loss is not a diagnosis; rather, it is a symptom of an underlying pathological change to the auditory system.”

-Dr. Eliot Shearer

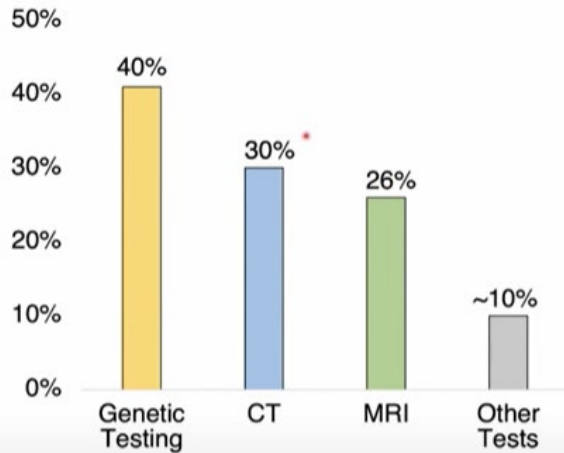
Genetics of Hearing Loss

Genetics of Hearing Loss



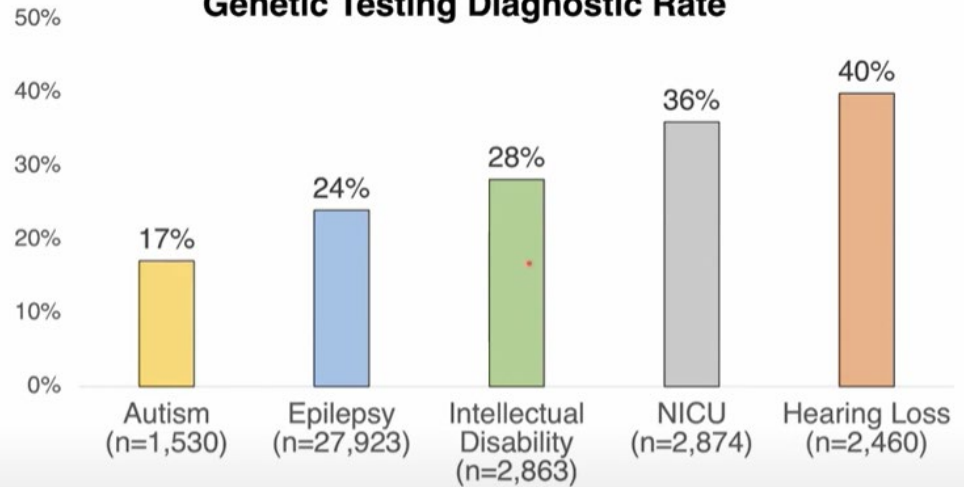
Single Most Effective Test

The Single Most Effective Test



Shearer et al 2019; Chen et al 2014; Kachniarz et al 2014

Genetic Testing Diagnostic Rate

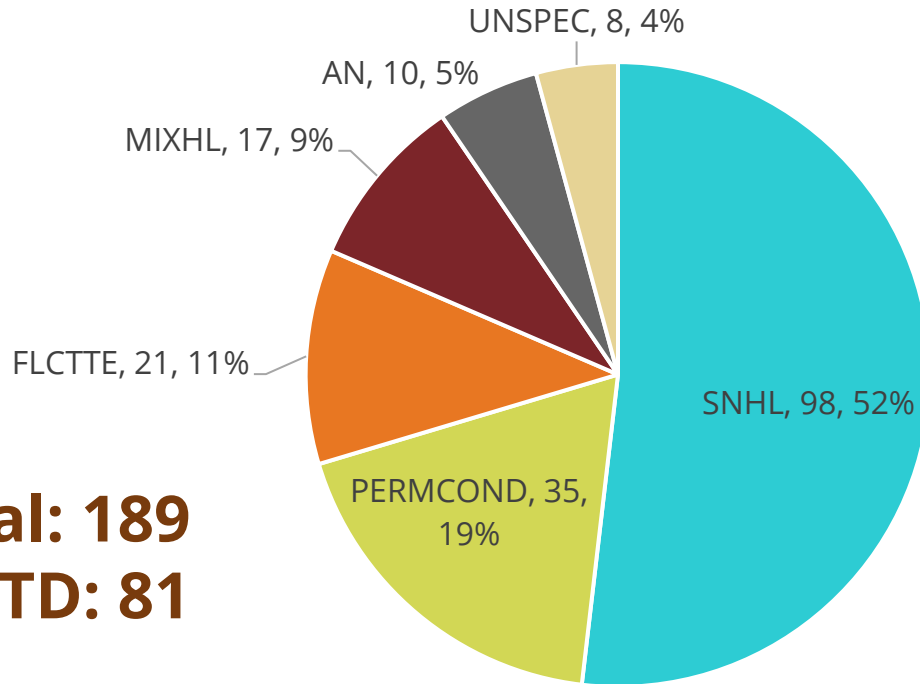


Sefanski et al 2020, Kingsmore & Cole 2022, Shearer et al 2019



Hearing Loss in Tennessee

Hearing Loss Trends in Tennessee, 2024 Births



2024 Total: 189
2025 YTD: 81

Hearing Loss Referrals in Tennessee

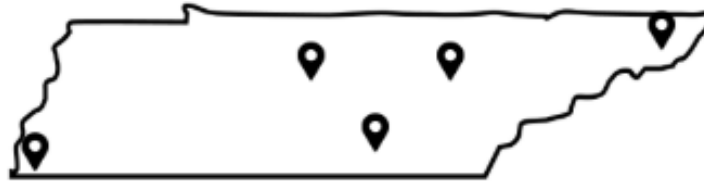


Automatic referrals made to:

- Early Intervention
- Children's Special Services
- Family Support



In 2023, began reviewing referral outcome data.



Genetic referrals began in March 2021.

Referral Notifications



Parents and Providers are sent a letter informing them that we have made referrals, and we provide them a flyer describing each referral and additional resources that they may qualify for.

TN Department of Health

Support for Children and Families Referral Information

The following early intervention resources are available for families caring for children in Tennessee. Diagnostic and financial eligibility criteria may be required to participate in these programs.

EARLY INTERVENTION IS KEY FOR INFANTS WHO HAVE RECEIVED A RECENT CONFIRMED DIAGNOSIS.

Children's Special Services (CSS)

- Provides financial help for medical care (if you qualify) up to age 21 after all other payment options have been used (Medicaid, private insurance, etc.).
- Families must meet certain requirements to be eligible.
- Supports families and helps manage care, including:
 - Making referrals, finding other resources in your neighborhood, working with schools, understanding insurance, medication and equipment help, and so much more

Tennessee Early Intervention System (TEIS)

- Offers therapy and other services to families of infants and young children with developmental delays or needs.
- Any child may be referred until age 3.
- Easy referrals - online, phone, email, and/or on the MyTN app (anyone can refer!)
- Services are free to families (if you qualify).
- Support focuses on daily routines around the home and helps families learn how to help their child learn throughout the day.

Family Voices of Tennessee

- Families helping families with emotional and educational support in caring for a child with special healthcare needs, chronic illnesses, or other needs.
- Connects families to local resources in your neighborhood.
- Helps families understand the healthcare system.
- Part of the TN Disability Coalition.

Regional Genetic Centers

- There are FIVE centers in Tennessee that can help families with labs and genetic counseling.
- Genetic Counseling is offered to families of children with hearing loss.

TN Department of Health

Additional Resources for Families

Community Health Access and Navigation in Tennessee (CHANT)

- Free care management in every county in Tennessee through the local health departments.
- Patient-focused, helping to understand the healthcare system, connecting families to social and medical support services.
- Connects families to local resources in your neighborhood.
- Easy referrals online (anyone can refer!).

Transportation Assistance for Appointments

Rides to and from appointments are available to TennCare members for TennCare covered services. **RIDE APPOINTMENTS MUST BE SCHEDULED AT LEAST 72 HOURS (3 DAYS) IN ADVANCE OF YOUR APPOINTMENT.**

- **Amerigroup Members - 1-866-680-0633**
 - Where's My Ride Option 5
 - Spanish Option 2
- **BlueCare Members - 1-855-735-4660**
 - Where's My Ride Option 3
 - Spanish Option 9
 - For trips over 90 miles, call 1-800-468-9698
- **TennCare Select Members - 1-866-473-7565**
 - Where's My Ride Option 3
 - Spanish Option 9
 - For trips over 90 miles, call 1-800-263-5479
- **United Health Care Members - 1-866-405-0238**
 - Where's My Ride Option 5
 - Spanish Option 2

Women, Infant, and Children (WIC) Services

- Helps provide healthy foods during pregnancy, after birth and for breastfeeding women, infants, and children through age 5.
- Offers breastfeeding help and referrals to other resources and services in your neighborhood.
- Families must meet certain requirements to qualify.
- To learn more, contact your local health department or call 1-800-342-5942

Hearing Loss Support

If your child has been diagnosed with hearing loss, there is support and options in how you choose to communicate with your child. To learn more, scan the QR code.

Department of Health, Administration No. 38023
The University publishes the proceedings of its own
conferences.

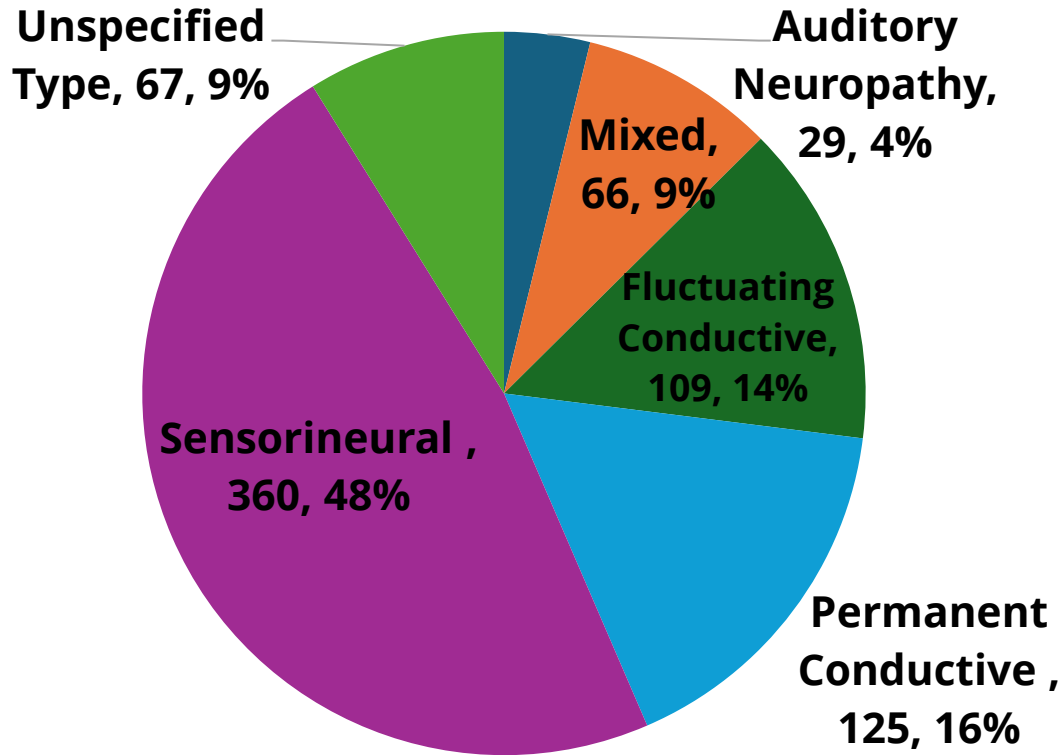
Genetic Follow-up on Hearing Loss Cases

- Aim: Evaluate incidence of genetic abnormalities among confirmed hearing loss cases referred to genetic tertiary centers.
- Design:
 - Each center was sent a spreadsheet that listed every case that was referred to them from March 2021 to April 2025 and report if the case had completed genetic testing (yes or no)
 - If yes, what were the results of the test.
 - If no, why was testing not completed.

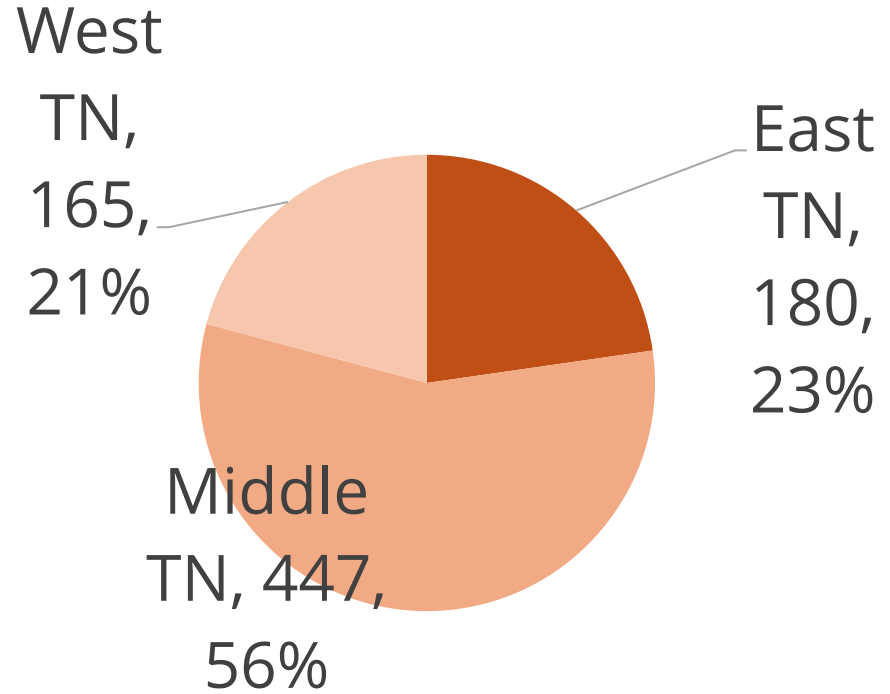
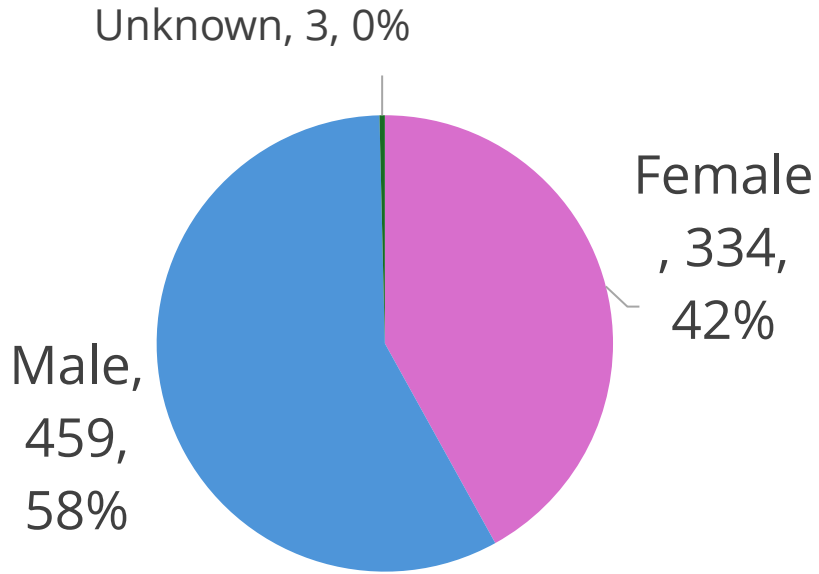
Results at a glance

- **796** cases were referred from March 2021 – April 2025
- **151** (19%) had a finite outcome reported
 - **89** (59%) were identified with a syndrome
 - **30** (20%) were non-syndromic
 - **32** (21%) Genetic testing not indicated

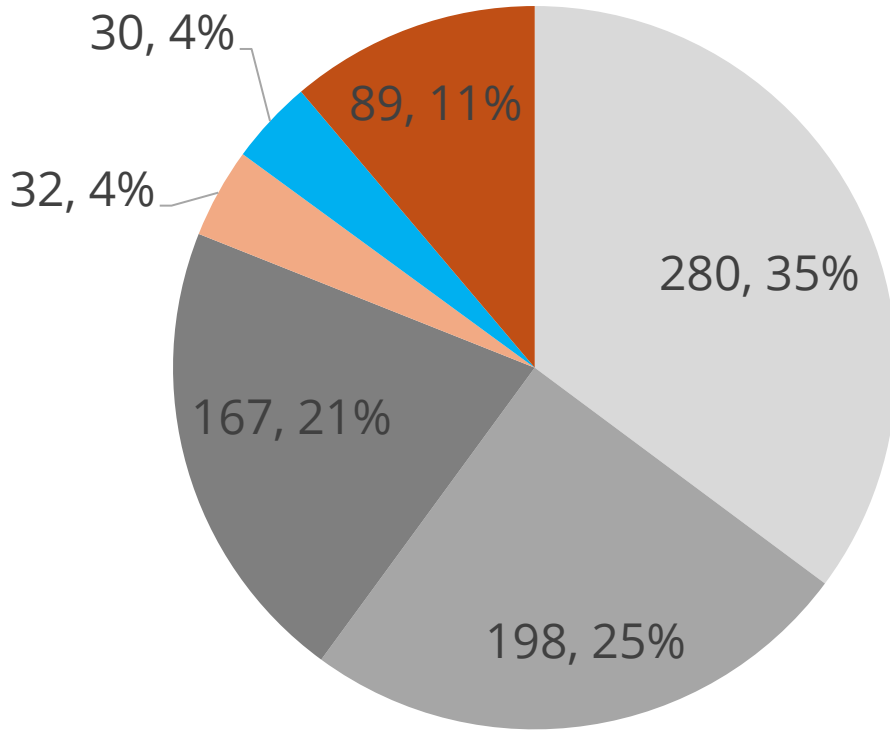
Referred cases by types of hearing loss



Referred cases by sex and grand division

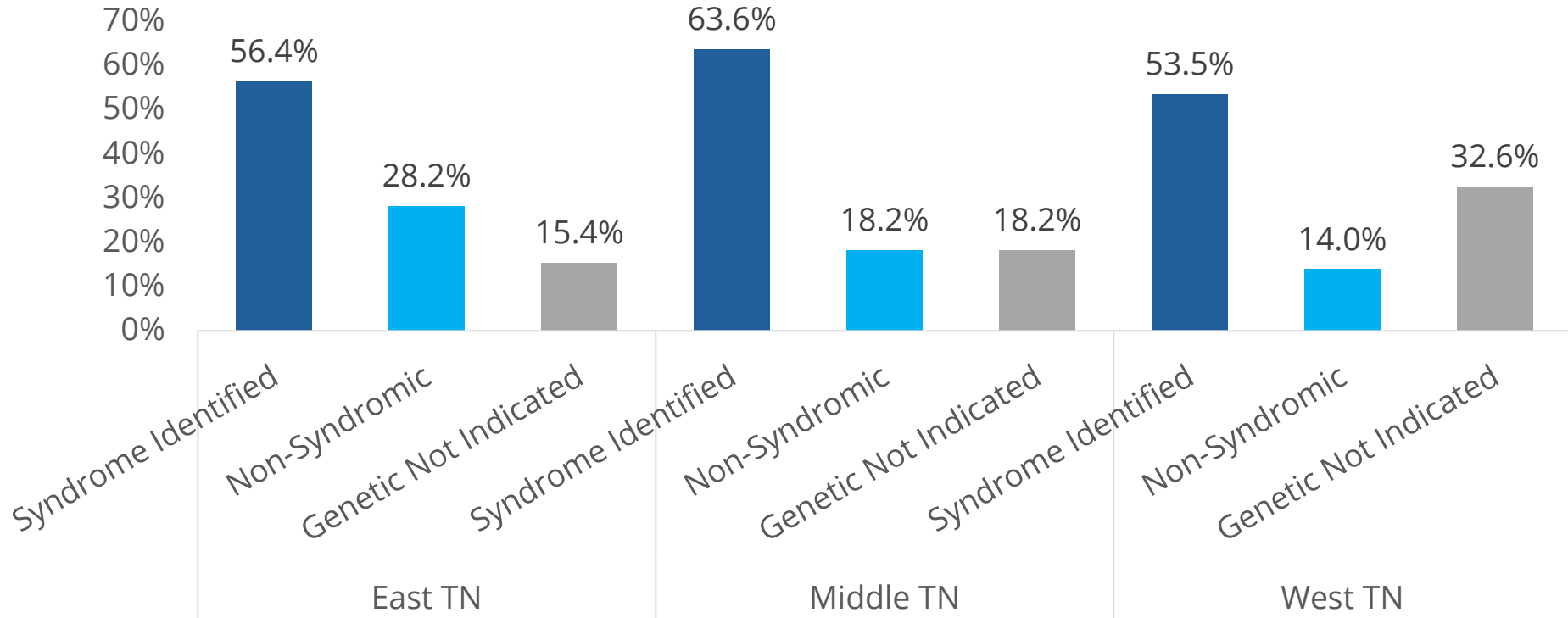


Outcome of genetic syndromes or reasons for not having an outcome

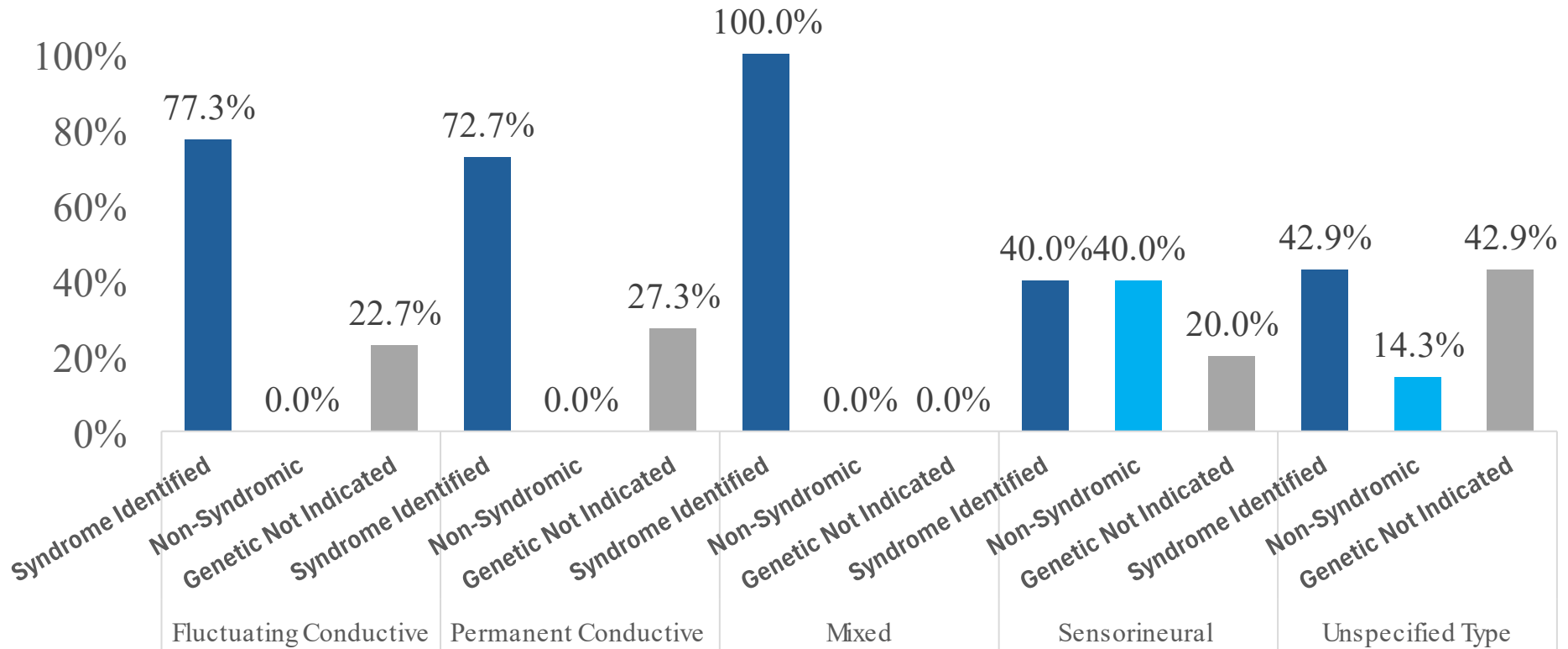


- No Information Received
- Expired/Refusal/Lost to Follow-up/HL Resolved
- Pending/Inconclusive
- Genetic Not Indicated
- Non-Syndromic
- Syndrome Identified

Genetic syndrome outcome by grand division



Genetic syndrome outcome by types of hearing loss



Challenges and Barriers



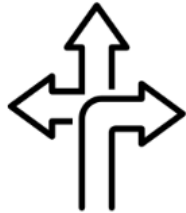
Insurance
Coverage



Limited
Testing



Communication



Guidance
Documents



Not a
Priority



Access to
Care

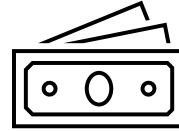
Thoughts for Implementation



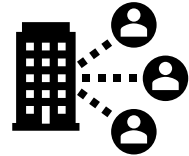
Reporting
Criteria



Information
Management



Costs



Collaborations
between EHDI and
DBS Follow-up.



Access to
Care



Capacity



Not all hearing
losses are
equal.

Future Projects



Collaboration with
Parent Guides



Guidance
Documents



Provider education



Update Referral
Forms

Take Aways



Complex Medical
Conditions



Hearing loss
has multiple
layers.



Education!
Education!
Education!

Contributors

- UT Lebonheur Genetics
 - Jessi Melvin
 - Dr. Nora Urraca
- Vanderbilt Genetics
 - Natalie Owen
 - Mackenzie Mosera
 - Eliza DeFore
- East Tennessee Children's Hospital Genetics
 - Sara Norton
- Erlanger Genetics
 - Jaclyn Smith
 - Leisa Haslerig
- East Tennessee State University Genetics
 - Stephanie Yoon
 - Jennifer Carver
- Dr. Yinmei Li

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