Utilization of Genetics Services in the Diagnosis of Newborn Hearing Loss in the State of Ohio

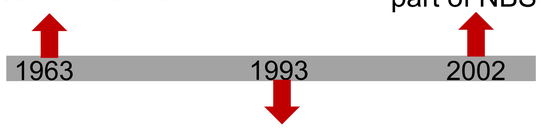
THE OHIO STATE UNIVERSITY
WEXNER MEDICAL CENTER

Cara Barnett MS LGC[1], Elizabeth Jordan MMsc LGC[1], Prashant Malhotra MD[2], Allyson VanHorn MPH MEd[3], John Myers MS[1]
[1]*The Ohio State University,* Columbus, OH, [2]*Nationwide Children's Hospital,* Columbus, OH [3]*The Ohio Department of Health,* Columbus, OH

Background

Newborn Screening (NBS) becomes a public health initiative[1]

Ohio implements Newborn Hearing Screening (NHS) as part of NBS[3]



National Institutes of Health (NIH) Consensus Statement focuses on early identification of Hearing Loss (HL) and creation of Universal Newborn Hearing Screening (UNHS)[2]

Causes of Newborn Hearing Loss[7-8]

- Acquired (environmental) or genetic
- 50-60% of newborn HL is genetic (new studies suggest up to 80% is genetic)
- Most common cause: Autosomal Recessive Non-Syndromic HL (*GJB2*)
- On average, 41% of patients who have genetic testing receive a genetic diagnosis

National Guidelines for Clinical Evaluation

- Involve a multidisciplinary team
- Genetics is a key part of this team
- All diagnostic evaluations should occur by 3 months of life

Aims

Aim 1: Evaluate provider practice patterns, beliefs, and knowledge of the role of genetics in the medical diagnosis of newborn HL

Aim 2: Assess parent experience and knowledge of the role of genetics in the diagnostic process for their newborn with HL

National Guidelines Referenced

- American Academy of Pediatrics (AAP)
- AAP
- American College of Medical Genetics (ACMG)
- American Journal of Audiology
- International Pediatric Otolaryngology Group (IPOG)

Comprehensive table outline and comparison of national guidelines available upon request

Methods

Populations surveyed

- Providers: Otolaryngologists (ENTs) and audiologists (AuD)
- Parents of children with newborn HL identified by the Ohio NHS

Cross-sectional survey instruments

- Non-validated created based on comprehensive literature review and guidelines evaluation
- Validated genetic knowledge measure

Data analysis

- Descriptive statistical analysis to determine thematical trends

Results

Demographics

Providers

- 113 responses, 95 included in analysis
- 14 (14.74%) ENT and 81 (85.26%) AuD
- Metropolitan/suburban pediatric practice

Parents

- 45 responses, 39 included in analysis
- All biological parents of child with HL

Results

Multidisciplinary care

- All providers favored a multidisciplinary care model
- Parents reported receiving multidisciplinary care
- Most frequently reported members of the care team:

Provider	*Parent*
1. AuD	1. AuD
2. ENT	2. Pediatrician
3. EI specialist	3. ENT

Genetics referral process

- Who is most responsible for referring/who placed your child's referral?

Provider	*Parent*
1. ENT	1. ENT
2. AuD	2. Pediatrician

Beliefs about genetics

Provider

- 95.51% (n=85) strongly believe/believe it is important families are presented with a genetics evaluation
- 76.4% (n=68) strongly believe/believe families are interested in a genetics evaluation

Parent

- 53% (n=17) with a child who was not previously evaluated were likely/very likely to pursue a genetics evaluation
- 25% (n=8) were unsure
- 22% (n=7) were unlikely/very unlikely

Parent reported genetic testing outcomes:

Cause of Hearing Loss	Frequency	Percent
***GJB2* mutations, Connexin 26**	3	21.43
Syndromic	5	35.71
Other	1	7.14
No cause determined	5	35.71

Results

Selected parent free text responses

"We are thankful for newborn screening because we were able to get him aided Before he was three months old"

"It would be nice if this was covered by insurance so we could test her genetics to see how this will impact her, if any other issues will arise, and what her future of a family would look like. However, the testing is not affordable so we will continue to focus on her treatments and whatever other issues arise"

Conclusions

- Multiple guidelines exist with varying detail
- **No clear workflow regarding who should discuss genetics**
- Collaborative, multidisciplinary team-based approach is needed
- **Parents are interested in genetics information**
- Providers are interested in CEUs on the genetics of newborn HL

Acknowledgements

- Funding:
  - National Society of Genetic Counseling (NSGC) Pediatric/Clinical Special Interest Group (SIG)
  - NSGC Public Health SIG
  - OSU CCTS Core Services Voucher

References

1. American College of Medical Genetics Newborn Screening Expert Group. (2006). Newborn screening: Toward a uniform screening panel and system--executive summary. *Pediatrics*, *117*(5 Pt 2), S296-307. https://doi.org/10.1542/peds.2005-2633I
2. Early identification of hearing impairment in infants and young children. (1993). *NIH Consensus Statement*, *11*(1), 1–24.
3. Ohio revised code 3701.504
4. Linden Phillips, L., Bitner-Glindzicz, M., Lench, N., Steel, K. P., Langford, C., Dawson, S. J., Davis, A., Simpson, S., & Packer, C. (2013). The future role of genetic screening to detect newborns at risk of childhood-onset hearing loss. *International Journal of Audiology*, *52*(2), 124–133. https://doi.org/10.3109/14992027.2012.733424
5. Yoshinaga-Itano PhD, C., Coulter BA, D., & Thomson MA, V. (2000). The Colorado Newborn Hearing Screening Project: Effects on Speech and Language Development for Children With Hearing Loss. *Journal of Perinatology*, *20*(S1), S132–S137. https://doi.org/10.1038/sj.jp.7200438
6. Yoshinaga-Itano, C. (2004). Levels of evidence: Universal newborn hearing screening (UNHS) and early hearing detection and intervention systems (EHDI). *Journal of Communication Disorders*, *37*(5), 451–465. https://doi.org/10.1016/j.jcomdis.2004.04.008 Wroblewska-Seniuk, K. E., Dabrowski, P., Szyfter, W., & Mazela, J. (2017). Universal newborn hearing screening: Methods and results, obstacles, and benefits. *Pediatric Research*, *81*(3), 415–422. https://doi.org/10.1038/pr.2016.250
7. Morton, C. C., & Nance, W. E. (2006). Newborn Hearing Screening—A Silent Revolution. *New England Journal of Medicine*, *354*(20), 2151–2164. https://doi.org/10.1056/NEJMra050700
8. Bowl, M. R., & Brown, S. D. M. (2018). Genetic landscape of auditory dysfunction. *Human Molecular Genetics*, *27*(R2), R130–R135. https://doi.org/10.1093/hmg/ddy158

Outline of professional organizations published guidance on genetics in newborn HL

Organization*	Publication Year	Member of the multidisciplinary team	Age care should be received from team	Hearing provider** role in genetic risk assessment	Components of family history	Pre-test discussion components	Benefits of genetic testing	Drawbacks of genetic testing	First-tier genetic testing
Topic		**Recommendations**							
AAP Joint Committee on Infant Hearing (JCIH)	2007	• Otolaryngologist • Genetics • Ophthalmologist • Developmental pediatrics • EHI/FSS+ • Social work	• 3 months	• Obtain family history • Refer for genetics evaluation	• HL-targeted +++	• Obtain informed consent	• Provide a genetic diagnosis • Prognostic information • Recurrence risk • Guide clinical management***		
AAP Clinical Report	2009	• Otolaryngologist • Audiologist • SLP++ • Genetics • EHI/FSS+					• Provide a genetic diagnosis • Recurrence risk		• *GJB2* and *GJB6* sequencing +/- del/dup†
ACMG ACT Sheet	2010	• Multidisciplinary team • Genetic specialist	• 3 months • Early inter-vention by 6 months						
ACMG	2014	• Otolaryngologist • Clinical geneticist • Genetic counselors • Audiologists • SLP++ • EHI/FSS+ • Other appropriate specialists		• Obtain family history • Refer for genetics evaluation	• Three-generation • HL-targeted+++ • Ancestry • Parental consanguinity/related-ness	• Pretest genetic counseling should be provided • Obtain informed consent	• Provide a genetic diagnosis • Identify need for additional evaluations • Guide clinical management*** • Prognostic information • Recurrence risk • Psychosocial benefits††		• *GJB2* and *GJB6* sequencing +/- del/dup†
AJA	2015			• Refer for genetics evaluation	• Three-generation • HL targeted+++ • Parental consanguinity/related-ness	• Benefits of genetic testing	• Provide a genetic diagnosis • Prognostic information • Recurrence risk • Psychosocial benefits††	• A negative genetic test does not rule out a genetic cause	
IPOG	2016			• Order first tier genetic testing • Consider referral for genetics evaluation		• Utilize a shared decision-making model • Diagnostic rates • Result implication • Discussion of patient goals	• Identify need for additional evaluations	• A negative genetic test does not rule out a genetic cause	• Multi-gene HL panel • If panel unavailable: *GJB2* and *GJB6* sequencing +/- del/dup†

**Hearing provider refers to otolaryngologists and/or audiologists
***A genetic diagnosis can change clinical management for the patient by indicating the need for further evaluation by additional specialists
+ Early Hearing Intervention and Family Support Services (EHI/FSS)
++ Speech Language Pathologist
+++Family history questions targeted towards childhood-onset permanent hearing loss
† +/- del/dup refers to with or without deletion/duplication analysis
†† Psychosocial benefits can include knowing the cause of their child's hearing loss, dispelling misinformation, and facilitating referral for unrelated hereditary conditions