DIAGNOSTIC EFFICIENCY AND COST EFFECTIVENESS OF COMPREHENSIVE GENE PANEL TESTING VERSUS IMAGING IN EVALUATION OF PEDIATRIC SENSORINEURAL HEARING LOSS

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Objectives

1. Determine the diagnostic yield for comprehensive genetic testing (CGP) versus imaging in children with SNHL
2. Evaluate the cost effectiveness of each diagnostic modality
3. Assist the clinician in diagnosis of SNHL

Materials and Methods

- Retrospective chart review
- Data collected from 1/1/2010 – 12/21/2015
- Patients with SNHL evaluated with genetic testing included
- Cost analysis of testing modalities performed

Results

Genetic Testing

- 137 patients with SNHL were evaluated with genetic testing
  - 98 patients had severe bilateral SNHL (72%)
  - 17 patients had moderate bilateral SNHL (13%)
  - 7 patients had mild bilateral SNHL (5%)
  - 8 patients had unilateral SNHL (6%)
  - 6 patients had unknown level of SNHL (4%)
- 31 patient samples were went to a comprehensive genetic panel with the results summarized in Figure 1.

- Most frequent mutations detected were GJB2 (8%), and GPR98 (5%)

Radiographic Imaging

- 105 of the 136 patients were evaluated with imaging
  - CT was performed in 92 patients
  - MRI was performed in 46 patients
  - Most frequent abnormalities detected were cochlear dysplasia (14) and vestibular dysplasia (8)
- Diagnostic yield for patients were: (Figure 2)
  - Mild hearing loss (0%)
  - Moderate hearing loss (40%)
  - Severe hearing loss (28%)
  - Unilateral hearing loss (30%)

Discussion

- CGP has a higher diagnostic yield and cost effectiveness in patients with mild, moderate, severe, and unilateral hearing loss compared to imaging diagnostic yield and cost across the same levels of hearing loss.
- A new diagnostic paradigm has been developed (Figure 5)

Conclusion

- CGP should be obtained prior to imaging in the evaluation of hearing loss in patients with congenital SNHL due to its superior diagnostic yield and cost effectiveness