

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 sent to a comprehensive genetic panel with the results summarized in **Figure 1**.

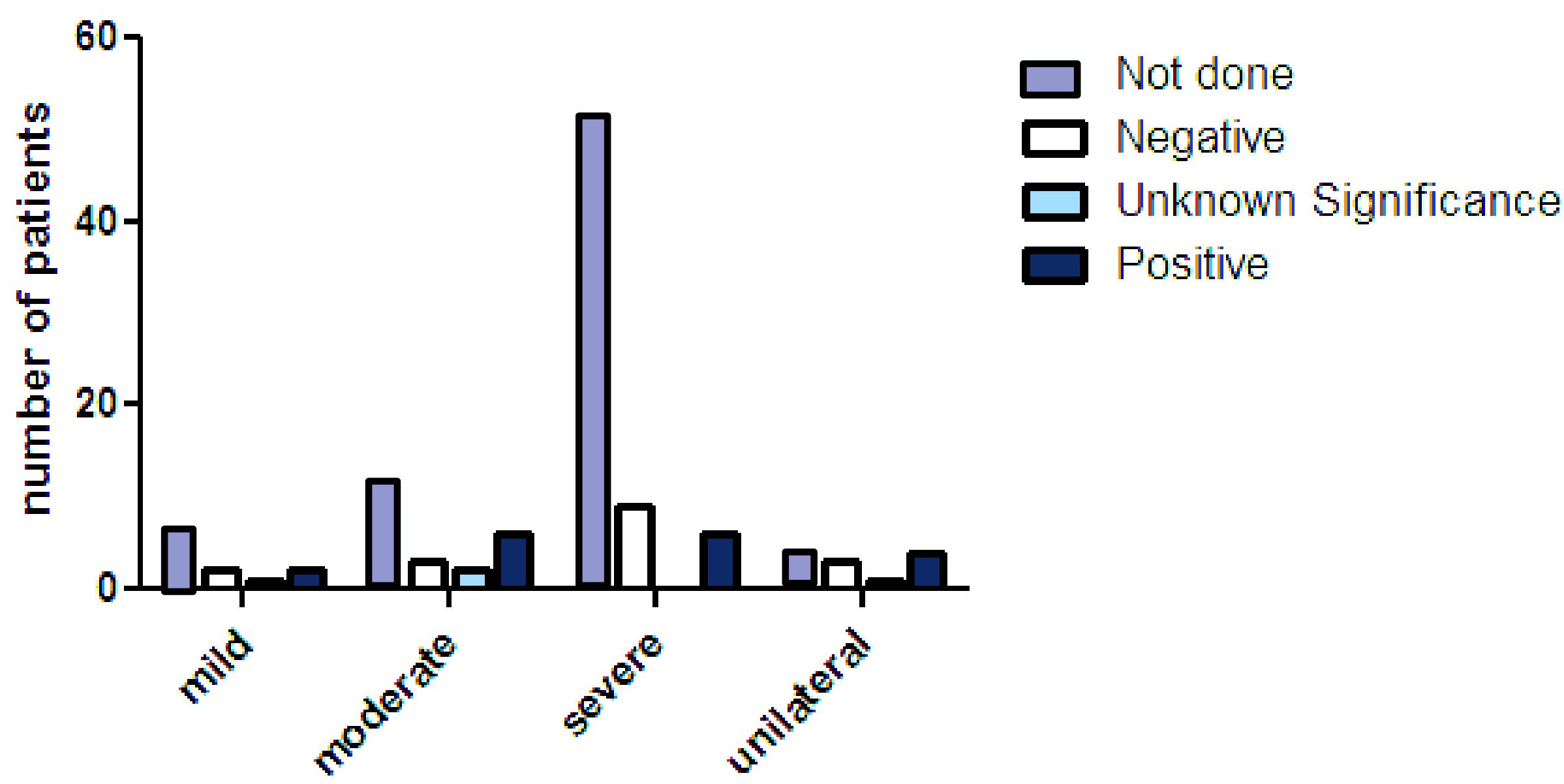


Figure 1: Comprehensive Gene Testing at Children's National

- The diagnostic yield for patients who underwent CGP:
 - Mild hearing loss was 60%
 - Moderate hearing loss was 73%
 - Severe hearing loss was 40%
 - Unilateral hearing loss was 44%
- 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%)

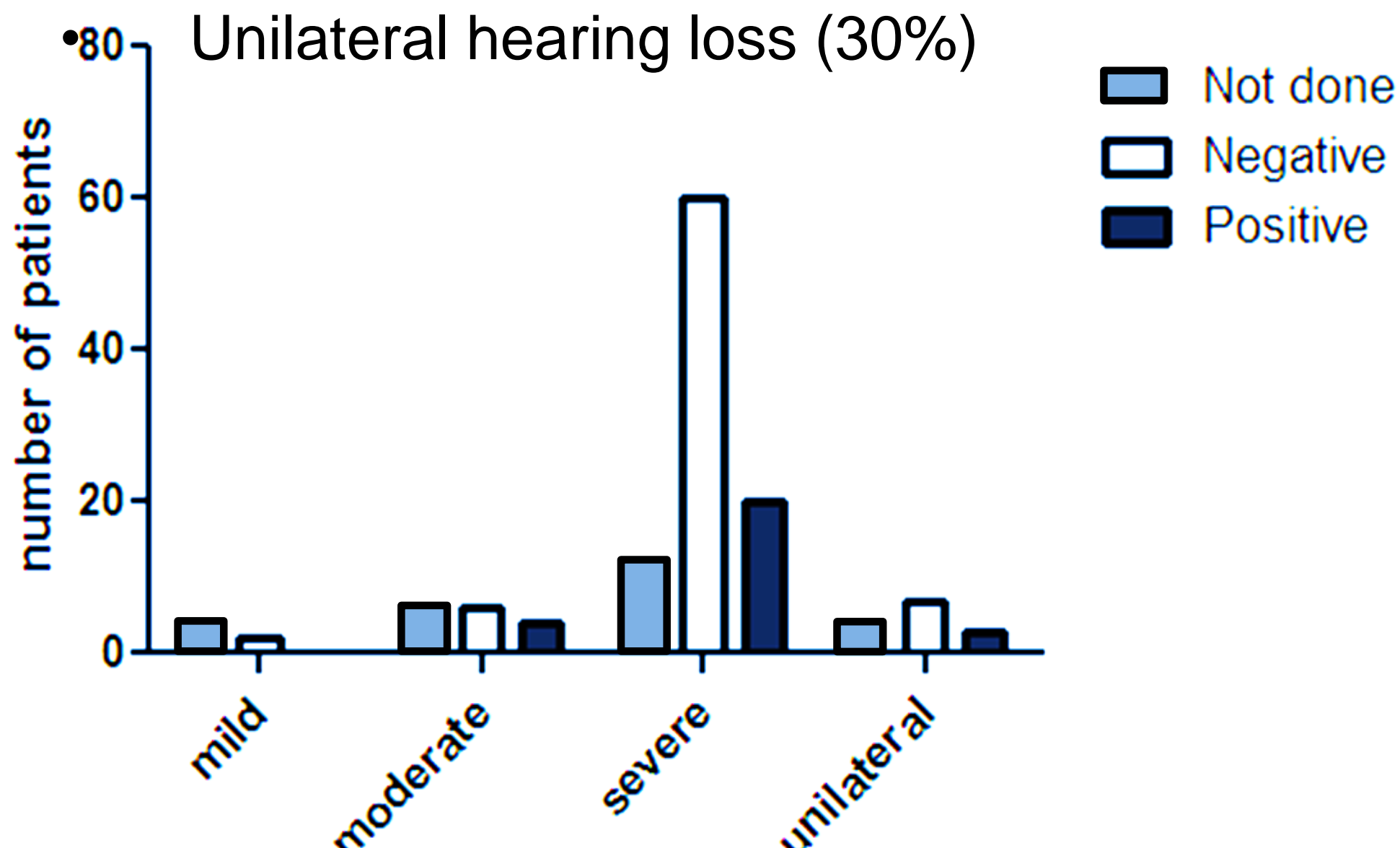


Figure 2: Radiographic Imaging Diagnostic Yield

Radiographic Imaging Compared to CGP

Using chi-square analysis, a statistical difference ($p < 0.0001$) was detected between the diagnostic yield of imaging compared to the diagnostic yield of CGP is summarized in **Figure 3**. Only four patients had both positive CGP and imaging.

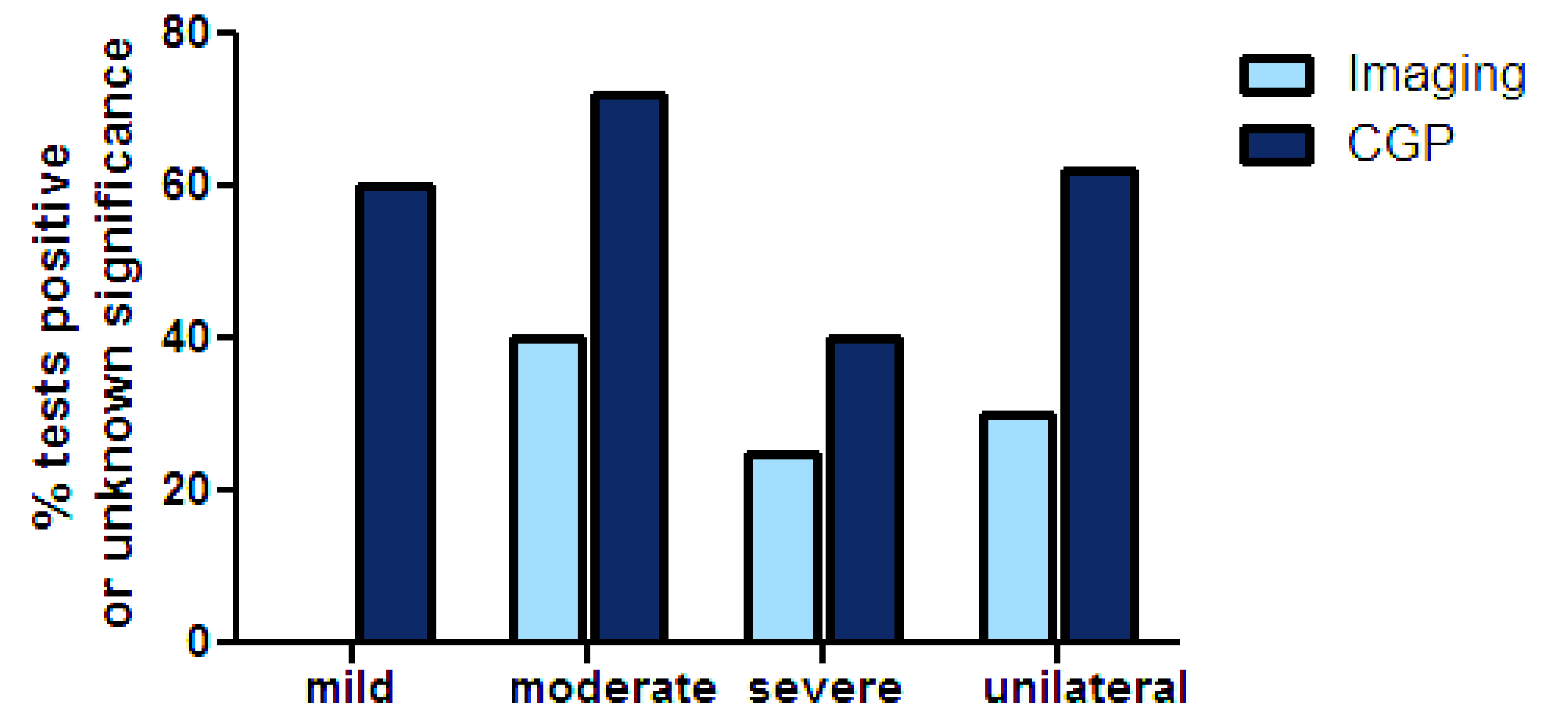


Figure 3: Overall Diagnostic Yield Imaging vs CGP

Cost Analysis

Cost Analysis was performed using the diagnostic yields obtained in this study and calculated per 100 patients. Total cost saving is represented in **Figure 4**.

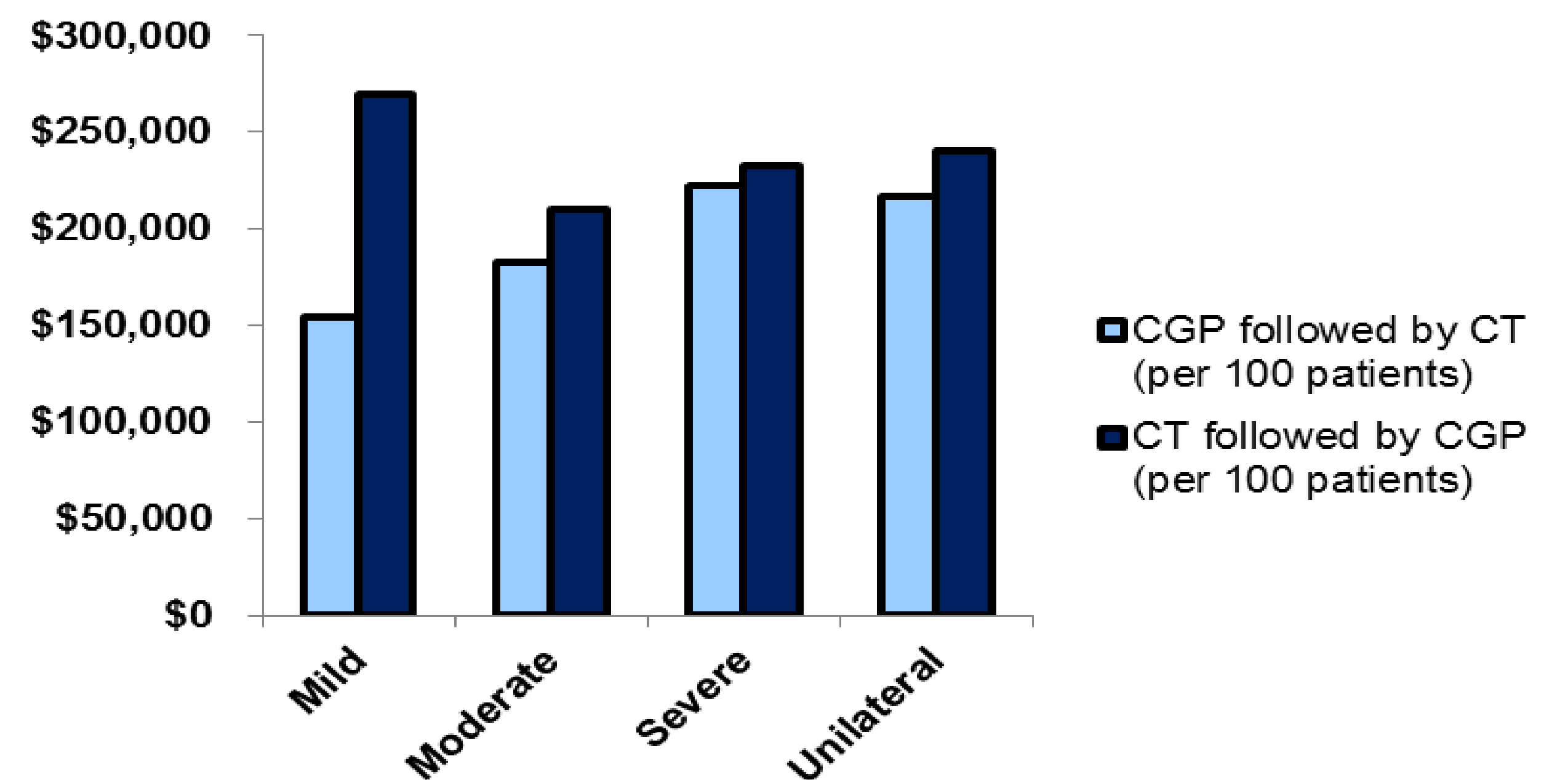


Figure 4: Cost Analysis performed per 100 Patients Comparing a CGP first diagnostic vs a CT first diagnostic paradigm

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**)

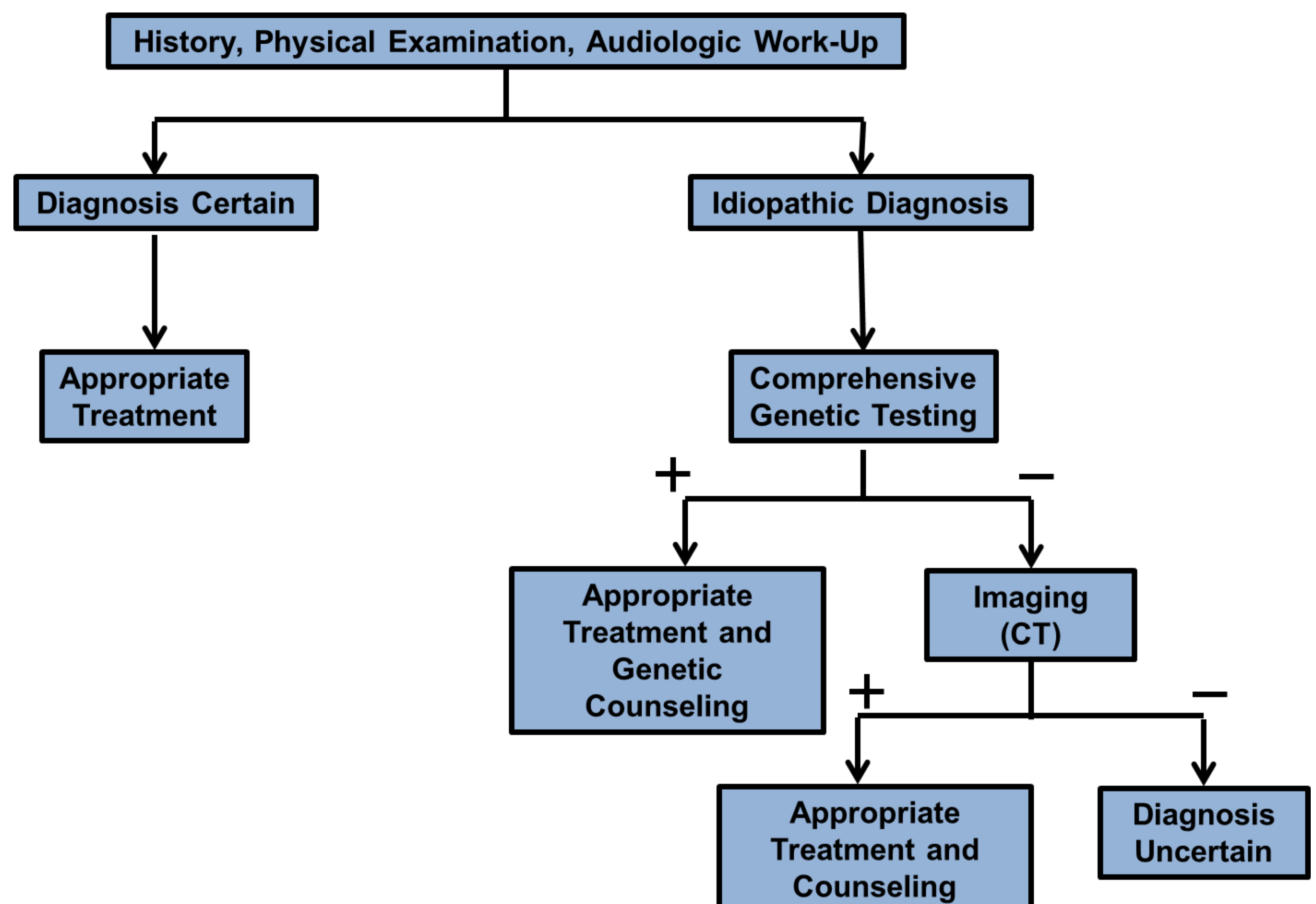


Figure 5: Proposed Diagnostic Paradigm

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