We present a two-level system of aetiological investigation that is clinically practical. Patients with ANSD sufficiently severe to consider cochlear implantation are generally identified at an early age. Although prematurity and jaundice are the most commonly identified aetiological factors in ANSD, aetiological investigation is important to guide prognosis and identify comorbidity. Imaging findings identify important prognostic factors in a significant minority. An important minority may have genetic and syndromic diagnoses that require further management.

Objectives

Auditory neuropathy spectrum disorder (ANSD) is an audiological diagnosis typified by the presence of intact outer hair cell function in the cochlea. ANSD is thought to account for 7-10% of all childhood permanent hearing impairment, and can be secondary to a range of pathological processes. This paper describes the rationale, methods and findings from this aetiological investigation.

Methods

Retrospective audit from four cochlear implant programmes.

Results

97 patients were identified. 79% of patients were identified before the age of one. Prematurity and jaundice were the most frequently identified aetiological factors. 33 patients had cochlear nerve deficiency on imaging. Genetic diagnoses include otoferlin mutation, SX010 gene change, connexin 26 mutation and A1FM1 gene mutation. ANSD was seen as part of a number of syndromes including Kallman’s syndrome, CHARGE syndrome, X-linked deafness, SOTOS syndrome, Brown Violettio Van Laere syndrome, and CAPOS syndrome.

Table 1: Guidelines for the investigation of children with ANSD

<table>
<thead>
<tr>
<th>Level</th>
<th>Criteria for investigation of ANSD</th>
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<tbody>
<tr>
<td>1</td>
<td>Hearing history and examination, audiometric testing, and auditory brainstem responses.</td>
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<tr>
<td>2</td>
<td>Advanced diagnostic imaging, including magnetic resonance imaging (MRI) of the inner ear and brainstem auditory evoked potentials (BAEPs).</td>
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T2 weighted MRI demonstrates right sided cochlear nerve deficiency with a narrow IAC.

The next step in our research is long term follow-up of patients with ANSD to determine factors that predict outcome from hearing rehabilitation strategies.