

ENT NEWS

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Updated on Evaluation and Management of Children with Sensorineural Hearing Loss (SNHL)

After a newborn baby or a child has had an abnormal auditory brainstem response (ABR), the physician needs to be sure that this test has been done with a normal middle ear condition. More specifically, if there has been fluid in the middle ear, the fluid will need to be removed either spontaneously, with medication or with ventilation tubes before another ABR or otoacoustic emission (OAE) would be done to determine if there is truly a persistent abnormal hearing condition. If the ABR or OAE continues to be abnormal, then the question becomes what to do with a child who has a nonsyndromic SNHL. (i.e. SNHL without other significant anomalies.)

The incidence of SNHL is approximately 1 in 1,000 births. This is a significant problem in healthcare since this would involve 40,000 children born in the United States each year who have a significant hearing loss and including 4,000 children who would be profoundly hearing impaired (essentially have no functional hearing).

Once a history and physical examination have been done to rule out any specific syndrome(s), one is left with determining the cause of the hearing loss in these children. In the past, a large battery of tests were routinely done on everyone, however, this still left no specific diagnosis in 40-50% of these children. With increased molecular testing, a different paradigm has evolved (and will probably continue to evolve as more specific molecular testing becomes commercially available). Commercial molecular genetic testing is a relatively new diagnostic tool in the diagnosis of children with SNHL. There are hundreds of mutations and dozens of genes that have been found to be associated with hearing loss. There are more than 60 loci for genes associated with nonsyndromic hearing loss, but with current technology, it is not practical to test for mutation of all of these loci. However, recently the mutation in a single gene GJB2 which encodes a Connexin protein (Cx26), ordered at Spectrum Health Butterworth Campus, has been shown in approximately 40% of children with severe to profound nonsyndromic SNHL. (Recently we had a patient who has a mild to moderate SNHL with a positive Cx26) This is a recessive mutation to show the phenotype of SNHL. In the midwest, the carrier rate for all Cx26 mutations is 3%. Also up to this time, the children with positive Cx26 have an exceedingly low probability of having other congenital abnormalities. Therefore, this one blood test has a much higher yield than any one or combination of the tests that have previously been done for the etiology of SNHL. It is still too early to use Cx26 to predict the future progression of hearing loss (whether it would stay the same or would be getting worse) and the outcome of cochlear implantation. The studies have shown that the false positive rate is at 2.6% and the false negative rate is at 3.1% with these children. It has also been recommended that if there is a positive Cx26 test, then the laboratories would proceed to code the entire gene as compared to looking at the specific sites that have previously been found to be abnormal. Genetic testing for SNHL is still in its infancy but in the future a single blood test that now screens for one gene may be able to screen for dozens of genes and hundreds of mutations.

At this time if Cx26 is positive, no further tests are being done except to do a CT scan (when the child is a candidate for cochlear implant, or is older and does not need a general anesthetic for the CT Scan). With a positive Cx26, we would also recommend an eye examination to rule out retinitis pigmentosa. If Cx26 is negative, then the tests that should be done include a urinalysis (proteinuria for Alport's Syndrome), an EKG for prolonged QT interval (Jervell and Lang-Nielsen Syndrome), ophthalmology evaluation (retinitis pigmentosa), and a CT scan of the temporal bone (enlarged vestibular aqueduct and/or incomplete development of the cochlea).

Other tests that could be included on a case by case basis would include the following:

- CBC (anemia, sickle cell anemia, macrothrombocytopenia)
- Thyroid function test (hypothyroidism – isolated or Pendred Syndrome – which now also has a testable gene (DFMB4)
- Sedimentation rate (autoimmune disease)
- Renal Function tests/electrolytes – (renal failure, Alport Syndrome)
- Cholesterol, triglycerides – Hyperlipidemia
- FTA – Syphilis
- TORCH titers (congenital infection – Toxoplasmosis, rubella, cytomegala virus, herpes simplex)
- Consultation (genetics, ophthalmology – various syndromes and retinitis pigmentosa)
- EKG – prolonged QT interval (Jervell & Lange – Nielsen Syndrome)
- CT scan of the temporal bone (enlarged vestibular aqueduct & incomplete development of the cochlea)

Also included is a algorithm which hopefully will be helpful in determining the test pattern that you may undertake or be of assistance in counseling your patients if these patients are seen by an otolaryngology practice. There is also a Web site that may be helpful www.uia.ac.be/dnalab/hhh.