

8. Audiological Evaluation

The external ear of the child with Progeria

Behavioral testing for assessing hearing thresholds

Objective electrophysiologic tests of auditory function

Summary



This section describes the typical hearing profile of children with Progeria, as well as a guide for the audiological evaluation and potential management strategies.

The external ear of the child with Progeria

In the typical ear, the pinna and lateral 1/3 of the ear canal are comprised of cartilage that is compliant, and subcutaneous fat allows the transducers used in behavioral and electrophysiological tests of hearing to fit snugly and comfortably in the ear. The status of the external ear in children with Progeria poses a special difficulty in conducting hearing assessment, as they are characterized by loss of compliance of the cartilage and loss of skin flexibility. The result is that the ear can be markedly sensitive to pressure applied by transducers applied to the pinna (such as the supra-aural earphones often used for air-conduction testing) and to the ear canal (such as tympanometry probes for performing acoustic immittance or insert earphones used for air-conduction testing or otoacoustic emissions). See Figures 1 and 2 (next page) for photographs of ears of two children with Progeria. To the touch, the pinna is obviously more rigid than are the pinnae of children who don't have Progeria. Those engaged in hearing testing should manually apply pressure to the pinna and ask the patient if that pressure causes discomfort before placing TDH supra-aural earphones.

Children with Progeria often develop low frequency conductive hearing loss. In general this does not lead to functional impairment.

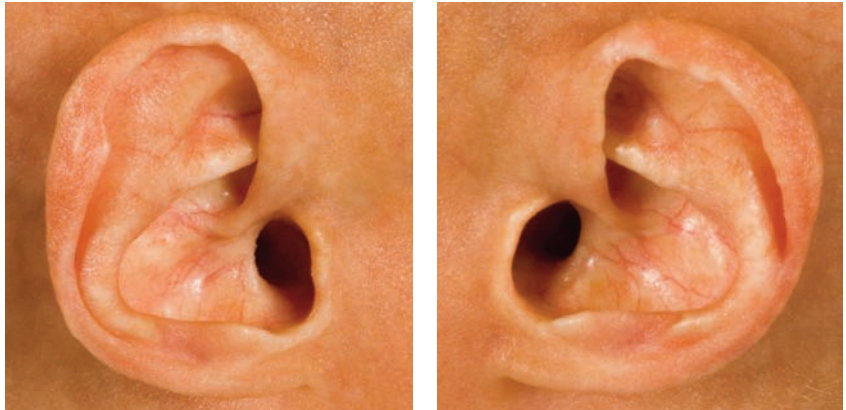


Figure 1. The right and left ears of children with Progeria. Note the large size of the entrance of the external auditory canal relative to the pinna.



Figure 2. The right and left ears of a child with Progeria. Note the obvious stenosis at the cartilaginous-bony juncture of the ear canal, most easily seen in the left ear.

The cartilaginous portion of the ear canal often has an appearance of a general loss of cartilage, resulting in a caliber significantly larger than the bony portion that comprises the medial 2/3 of the ear canal. This difference in the size of the cartilaginous and bony portion of the ear canal can be confusing when attempting to place an earphone in the canal. Usually, an earphone or tympanometry probe tip is coupled manually to the cartilaginous portion of the ear canal. The significant size discrepancy can make it difficult to obtain a hermetic seal when attempting tympanometry and middle-ear muscle reflex testing. While potentially easier than obtaining a seal by coupling the eartip to the bony portion of the canal, that part of the canal is very sensitive in anyone, and so it may be difficult for the patient with Progeria to tolerate placement of eartips for audiometric testing. Engage the patient in the testing process by introducing them to the next test and explaining that the eartips are manufactured with assumptions (that is, the appropriate size and rigidity of the eartips) that don't necessarily apply to a child with Progeria. Children should also be uniformly given full license to suspend any test at any time, which may also increase their trust in the examiners and perhaps their tolerance of mild discomfort.

Cerumen impaction is often reported by families to be problematic in children with Progeria. The earwax is often very dry and adheres to the ear canal wall at the bony-cartilaginous juncture. Children with Progeria should routinely be seen by a physician to examine ears for cerumen impaction and follow physician recommendations for using liquid solutions to try to soften wax prior to manual extraction by a physician.

Behavioral testing for assessing hearing thresholds

Measuring pure-tone hearing detection thresholds by behavioral audiometry is the gold-standard for the clinical assessment of hearing function.

Patients with Progeria are, by-and-large, cognitively typical for age, so their language is appropriate for a child their age. A child's language age is a good indicator for which behavioral test technique is most appropriate for determining pure-tone hearing thresholds, or if the child can be tested behaviorally at all. Given that this disorder presents around age 18 to 24 months, hearing can be assessed in children with Progeria at the earliest age of diagnosis by visual reinforcement audiometry; this pediatric test technique is valid for typically-developing children ages 8 months to roughly 30 months. Children with Progeria ages 2 to 5 years can usually be tested by conditioned play audiometry. Children ages 5 years and older can usually be tested by conventional "hand-raising" audiometry.

Children with Progeria almost uniformly have some degree of low-frequency conductive hearing loss. See Figure 3 for a typical audiogram (hearing test results) in a child with Progeria. Hearing loss is not always bilateral, nor is it always symmetrical when hearing loss exists in both ears. The configuration is the same, however, when hearing loss existed: low-to-mid frequency upsloping to better (and perhaps normal) hearing in the higher frequencies.

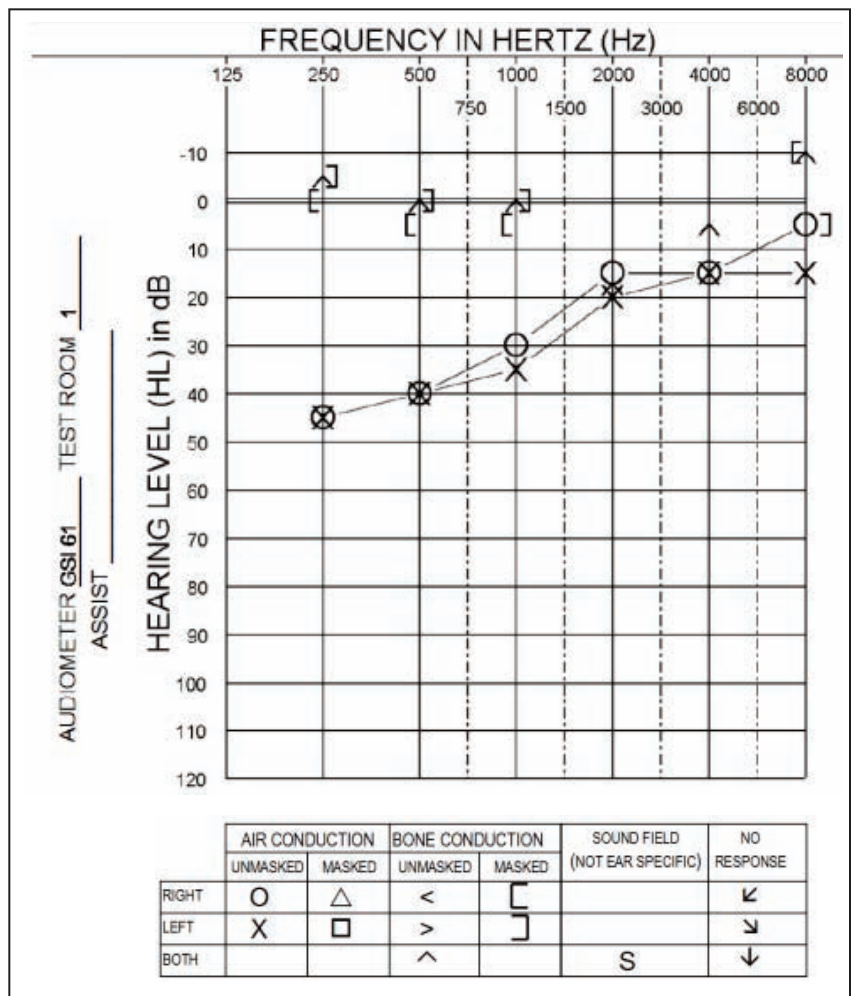


Figure 3. Typical audiogram of a child with Progeria.

Objective electrophysiologic tests of auditory function

> Tympanometry

Tympanometry is a test to assess the gross function of the middle-ear. It is performed by manually applying or inserting a rubber tipped probe that is intended to hermetically seal the ear canal. A low frequency tone (226 Hz) is presented in the ear canal while air pressure is changed from +200 daPa to -400 daPa. This change in air pressure is quite gentle and usually is completed in seconds. The change in the sound pressure level of the low frequency tone in the ear canal is a result of sound being transmitted more or less efficiently through the middle-ear system as a function of the air pressure in the ear canal. There are normative data for equivalent ear canal physical volume, peak pressure, static compliance, and tympanic width. Findings on tympanometry are essentially normal in many children (regardless of hearing test results). When abnormal, static compliance is usually reduced and tympanic width is consequently wider than normal. Otologic examination in a few patients by a pediatric otolaryngologist did not reveal middle-ear effusion in any of these patients with reduced static compliance. The reason for the abnormal tympanometry findings remains unclear at this time.

> Acoustic reflex (middle-ear muscle reflex) threshold

Middle-ear muscle reflex threshold is a gross measure of middle-ear function that incorporates a reflex arc ascending from the 8th cranial nerve to the level of the superior olivary complex and descending the 7th cranial nerve both ipsilateral and contralateral to the stimulus. The test is conducted much the same way as tympanometry, making use of the same probe tip used in tympanometry. A hermetic seal is necessary to complete this testing, which can usually be completed within a few minutes. A low frequency probe tone (226 Hz) is presented in the ear canal and the ear canal air pressure is kept stable. A stimulating tone of varying frequencies (typically 500 Hz, 1000 Hz, and/or 2000 Hz) is presented in the ear canal at relatively high intensity (normal reflex thresholds are 85-90 dB HL). A stimulating tone sufficient to engage the middle-ear muscle reflex causes the stapedius muscle to contract, stiffening the middle-ear system. This stiffening can be detected in much the same way as it is with tympanometry. When there is middle-ear dysfunction, middle-ear muscle reflexes are typically elevated (> 90 dB HL) or absent (no reflex elicited using a maximum stimulus of 110 dB HL). Children with Progeria almost uniformly have elevated or absent middle-ear muscle reflexes, regardless of findings on tympanometry.

> Otoacoustic emissions

Otoacoustic emissions are a measure of the functional integrity of the cochlea, up to the level of the outer hair cell. These “ear sound” emissions are thought to arise from the electromotility of healthy outer hair cells, and so are a by-product of the normal hearing mechanism. People with sensorineural hearing loss, such as that caused by age (“presbycusis”) or noise (“noise-induced hearing loss”), have absent otoacoustic emissions. They can be evoked by an external sound stimulus, such as a click or a pair of pure tones, and the resulting response from the cochlea can be measured in the ear canal with a very sensitive microphone if the ambient noise (in the room as well as from the patient) is quiet enough that the emission can be measured. This test, then, requires the placement of an earphone in the ear canal, which houses both a transducer for generating sound as well as recording sound. It does not require a hermetic seal, but a reasonably good coupling to the walls of the ear canal are necessary so that sound does not leak out of the ear. Children with Progeria almost uniformly have normal otoacoustic emissions in the mid to high frequencies. It is known that otoacoustic emissions are typically affected (are either reduced or absent) by conductive transmission loss in the middle-ear due to middle-ear dysfunction. Otoacoustic emissions in children with Progeria are typical of what one would expect based on their audiogram: At frequencies where a conductive hearing loss exists (in these patients, usually low-to-mid frequencies), the otoacoustic emissions are reduced or absent. Of specific note, high frequency otoacoustic emissions (as high as 10k Hz) are uniformly present in children with Progeria as long as the conductive hearing loss does not extend to these higher frequencies. It would then seem that the cochlea of a child with Progeria does not age prematurely.

> Auditory Brainstem Response (also known as Brainstem Auditory Evoked Response)

Auditory Brainstem Response measures the far field electrical potentials evoked by a sound stimulus from the auditory brainstem nuclei through the level of the lateral lemniscus. Testing is typically used to estimate hearing thresholds in children too young or developmentally impaired who cannot participate in behavioral audiometry, or in cases where there is suspicion for a lesion of the ascending auditory neural pathway (such as a tumor on the 8th cranial nerve). As this test requires the passive participation of the patient, sleep is often desired during this testing (either natural or through the use of sedation). Similar concerns regarding placement of a transducer in the ear canal continue here, as the transducers used for Auditory Brain-

stem Response are the same as those used in behavioral audiometry. An additional concern is that the evoked response is recorded far field, using three or four scalp electrodes which must have low ($< 5k$ ohm) and well-balanced skin impedance (all within $5k$ ohm). Usually, a mild abrasive is used to exfoliate the skin and remove dead skin cells. Given the truly thin skin of the patient with Progeria, care must be taken to not compromise skin integrity should this testing be conducted.

Summary

Children with Progeria have low-to-mid frequency conductive hearing loss that is usually mild, but can be moderate (or greater) in degree. The pathophysiology of this hearing loss is not clear at this time. Some children have grossly abnormal tympanometry with hearing thresholds that were relatively normal, while in other patients with normal tympanometry their hearing thresholds could be significantly elevated (hearing significantly impaired). Cerumen and middle ear effusion are not usually factors contributing to the hearing losses recorded. Middle-ear muscle reflexes were almost uniformly elevated or absent. Otoacoustic emissions are almost uniformly normal at frequencies where the conductive hearing mechanism is normal to near normal (in the mid-to-high frequencies). The site-of-lesion then would appear to be some dysfunction in the middle-ear system unrelated to an ear infection/middle-ear effusion. This dysfunction results in a stiffening of the system and thus loss of sound transmission properties of the middle-ear.

A patient with a mild low frequency hearing loss has little functional impairment with communication. Consequently, parents usually report that their child with Progeria hears very well; often a low-frequency hearing loss was found that was not previously diagnosed. Audiological interventions were usually limited to annual monitoring of hearing for progressive worsening of hearing into the speech frequencies, or perhaps preferential seating in the classroom. Occasionally, based on parent report of the child with low-frequency hearing loss having difficulty attending to the teacher's voice, FM educational amplification is recommended to help the child hear the teacher's voice preferentially over the ambient sound in the classroom. Given the anatomical changes of the external ear described earlier in this document, coupling a hearing aid to the ear via personal custom earmold could be challenging. Prognosis for use of hearing aids is very good as the type of hearing loss is conductive and there is no expected loss of clarity of the signal, as there can be when there is greater than a moderate degree of cochlear (i.e., sensorineural) hearing loss.