What to Expect at Your Child's Genetic Visit for Hearing Loss

COVERED IN THIS HANDOUT:

What is hearing loss?

Hearing loss is when your ability to hear sound is less than usual. It can affect one or both ears and can range in severity. A hearing test helps determine the extent of the hearing loss. Hearing loss can be mild, moderate, severe, or profound, depending on how much sound can still be heard.

Are there different types of hearing loss?

There are three common types of hearing loss: sensorineural, conductive, and mixed.

- Sensorineural hearing loss happens when the inner ear (cochlea) or the nerve pathways from the inner ear to the brain do not recognize sound in the usual way. Sensorineural hearing loss is sometimes called "nerve deafness", but that term is not exactly accurate.
- Conductive hearing loss happens when sound does not pass well from the outer ear to the ear drum and tiny bones of the middle ear.
- Mixed hearing loss happens when there is a combination of both conductive hearing loss and sensorineural hearing loss.

Why was my child referred for a genetics visit?

Your child was referred for a genetics visit to try to better understand why she/he has hearing loss. About half of the time, early childhood hearing loss is due to a genetic cause. A genetic cause is more likely when the hearing loss happens early in life and is sensorineural.

What can a genetics visit tell me about my child's hearing loss?

The main goal of a genetics visit is to try to understand the cause of your child's hearing loss. Some forms of genetic hearing loss can include medical concerns other than hearing, such as heart rhythm problems, vision problems, and kidney problems. A genetics visit can help find out if your child may be at risk for one or more of these other health problems. A genetics visit can also give you a better understanding of the chance for hearing loss in other family members (including future children).

There's no history of hearing loss in my family. Could my child still have a genetic hearing loss?

Most forms of genetic hearing loss happen unexpectedly, often for the first time in the family. So, even if you have no history of hearing loss in your family, your child might still have a genetic hearing loss.

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What happens at a genetic visit?

A medical geneticist and a genetic counselor will generally meet with you and your child. You will be asked about your family's medical history, either before the appointment by phone, or at the beginning of the visit. Your child's medical history is reviewed by the medical geneticist before your visit. During the visit, the medical geneticist will perform a physical exam of your child that includes measuring many different parts of the body, like the head, ears, and eyes. Much of the visit is spent discussing the possible causes for hearing loss in your child and answering your questions. In some cases, the medical geneticist may recommend additional testing, to be done after the visit. Most genetic visits last about one hour.

Can a cause for hearing loss always be found?

Not always. There are many different causes for hearing loss. About half of all childhood hearing loss is genetic, however the genetic cause is not always able to be found in these cases. Another 20 to 25% of cases are caused by environmental factors. That leaves about 25 to 30% of cases with an unknown cause.

What are environmental causes of hearing loss?

Environmental causes of hearing loss can include:

- Exposure to infections during pregnancy or after birth
- Premature birth
- Exposure to alcohol during pregnancy
- Exposure to medicines that can damage the auditory nerve

What are genetic causes of hearing loss?

Genetic causes of hearing loss can be sorted into non-syndromic and syndromic forms.

Non-syndromic forms of hearing loss affect only hearing and do not cause any other health issues. So far, over 65 different genes have been discovered that can cause non-syndromic hearing loss. Although there are many genes that can cause non-syndromic sensorineural hearing loss, genetic changes (mutations) in the GJB2 gene (also called connexin 26) account for nearly 50% of childhood sensorineural hearing loss.

Syndromic forms of hearing loss (also called genetic syndromes) are due to genes that can also cause other health problems or physical differences. There are over 300 different genetic syndromes that include hearing loss along with other physical differences or health problems.

Examples of syndromes with hearing loss (HL)	Medical findings other than hearing loss
Waardenburg syndrome (sensorineural HL)	→ Extremely pale blue eyes or eye colors that don't match; widely spaced eyes; white patch of hair or early graying
Branchio-oto-renal syndrome (all types of HL)	→ Unusual ear shape; pits or small skin tags in front of the ear; kidney problems
Stickler syndrome (sensorineural or conductive HL)	→ Extreme nearsightedness (myopia); other eye problems; small jaw; cleft palate (opening in the roof of the mouth); joint problems
Usher syndrome (profound, bilateral HL)	→ Decreased night vision and loss of side vision in childhood or early teens; balance problems
Pendred syndrome (severe-to-profound, bilateral HL)	→ Thyroid problems (goiter); balance problems

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What testing might be recommended for children with hearing loss?

The testing recommended for a child with hearing loss can vary from child to child and depends, in part, on a child's medical history.

Testing that might be considered at the time hearing loss is diagnosed includes:

- Eye exam by an ophthalmologist (eye doctor specialist)
- Electrocardiogram (EKG) - a test that monitors the heart beat
- Genetic testing

Some medical problems related to hearing loss may not happen until a child is older. For children five years old or older a pediatrician might consider ordering:

- Urinalysis with microscopy (a urine test)
- Thyroid function tests (a blood test)

And finally, some children with hearing loss may benefit by having imaging of the inner ear and the auditory nerve. Your child's otolaryngologist (ear doctor) will discuss the risks and benefits of imaging studies.

What are genetic tests?

Genetic tests are medical tests that can help find out whether or not a particular gene is working in the usual way. Genetic testing looks for changes in the genes called variants. Most genetic testing is done by taking a blood sample from the arm.

What kind of genetic testing is done for hearing loss?

Since there are so many genes that can cause hearing loss, it is important to know which gene (or genes) should be tested, before any testing is ordered. If a specific syndrome is being considered, genetic testing may look only at the gene that causes that syndrome. If there are no findings that suggest a syndrome, testing might include a panel of the most common genes that cause non-syndromic hearing loss. Genetic testing does not test for all types of genetic hearing loss, so the results do not always provide a explanation for hearing loss, even in cases that are likely genetic.

Will the doctor order genetic testing for my child?

Genetic testing is not done for every child with hearing loss, however, genetic testing may be offered if the results could help with your child's medical care or with family planning. You can talk with the medical geneticist about whether or not genetic testing would be helpful for your child.

Definitions:

Auditory nerve – the nerve that connects the ear to the brain; needed to hear sound
Gene – A single genetic instruction
Genetic counselor - A medical professional trained in genetics and counseling
Medical geneticist - A medical doctor with special training in conditions that are caused by differences in the genetic instructions

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