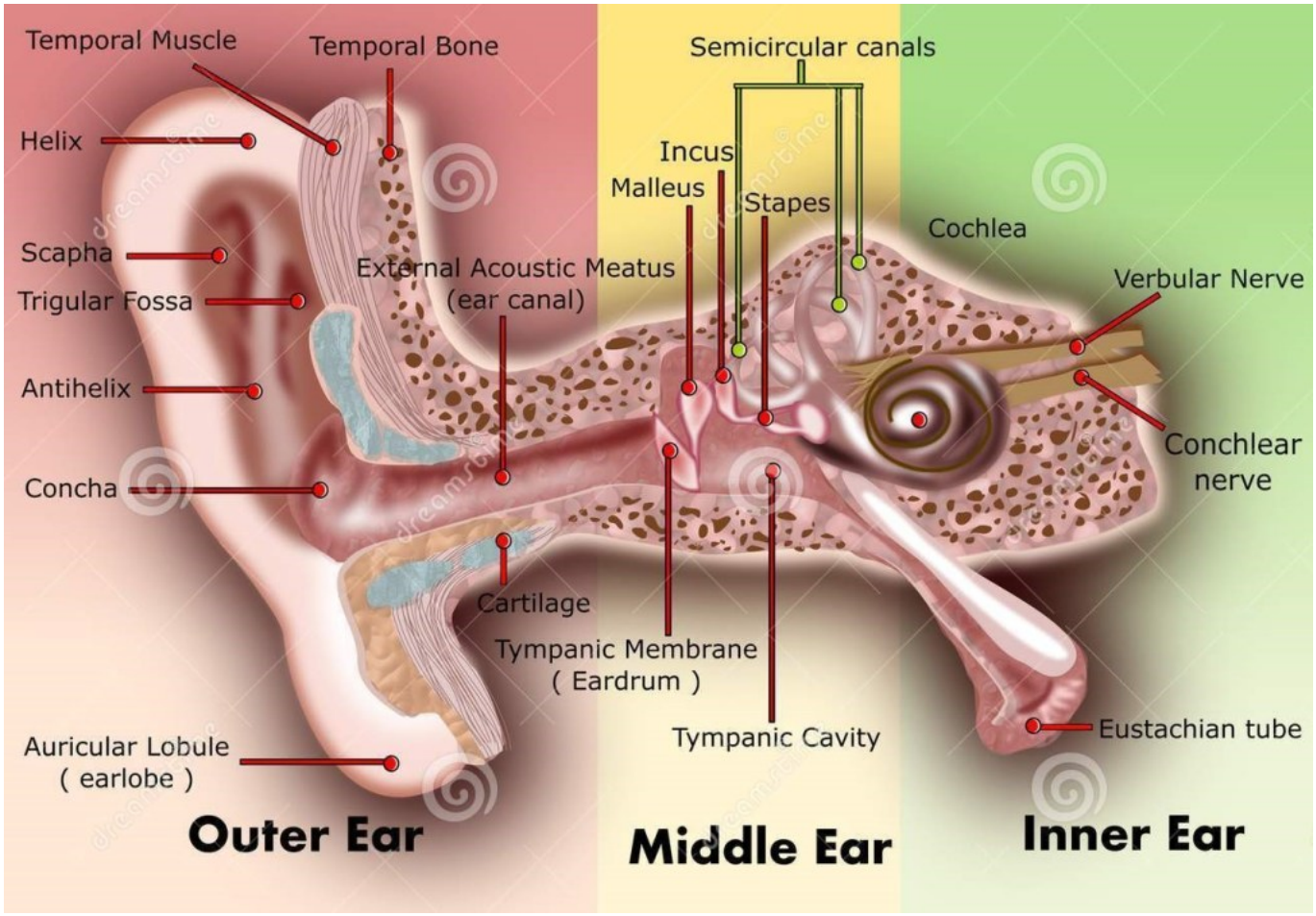
Congenital hearing loss (or hearing loss from birth) is when someone has a reduced ability to hear sounds like other people do, and can range from mild to moderate to severe. Hearing loss is one of the most common birth defects in newborns, affecting approximately 1 to 2 of every 1000 babies every year. People who have hearing loss at birth or before the development of speech are said to have “prelingual” hearing loss. It is important to note that not all prelingual hearing loss is congenital. As we age the number of people diagnosed with hearing loss (called prevalence) increases with about 65% of those older than 70 being affected. Those who develop hearing loss after the development of normal speech have “postlingual” hearing loss.

There are three different types of hearing loss:

- ***Conductive hearing loss*** occurs when the outer or middle ear is affected and sound is not able to be transmitted through the ear. This may be due to a malformation or obstruction (e.g. fluid in the ears).
- ***Sensorineural hearing loss (SNHL)*** occurs when the inner ear is affected, and may be due to differences with the hair cells (sensory hearing loss), central auditory pathway (central loss), or when there is an abnormality in how sound is transmitted from the inner ear to the brain (auditory neuropathy spectrum disorder).
- ***Mixed hearing loss*** occurs when there is both conductive and sensorineural hearing loss.



What causes hearing loss?

Hearing loss can be caused by both hereditary (genetic) and environmental (nongenetic) factors, or due to a combination.

For those with congenital hearing loss about 50-60% will have an underlying genetic cause, which means that they have a specific change in their genetic material that has caused the hearing loss. In developed countries, this increases to about 80% for those with congenital hearing loss. The chances that hearing loss is genetic is impacted by age of onset, family history, laterality (one or both ears), type of hearing loss, and whether there are other features or health problems.

Common environmental factors that cause hearing loss include exposure to specific infectious agents either during the pregnancy (prenatal) or after (postnatal), exposure to certain medications (called ototoxicity), chronic ear infections or fluid in the ears (chronic

otitis media), being born premature, significant noise exposure through occupation or recreation, injury to the ear, or aging. The most common infectious agents that cause congenital hearing loss include viruses called toxoplasmosis, rubella, *cytomegalovirus* (CMV), and herpes — all together these are called the TORCH "organisms." Of these, CMV is the most common, and affected individuals may present either at birth or later in infancy with hearing loss.

What are the genetic causes of congenital hearing loss?

Genetic causes of hearing loss may be classified as syndromic or non-syndromic. Syndromic means that the genetic cause is associated with a combination of other health problems or physical differences, such as eye, kidney, or skin abnormalities. More than 400 genetic syndromes are thought to be associated with hearing loss. Non-syndromic means that the genetic cause is only associated with hearing loss. It is important to note that some syndromic hearing loss may initially present with only hearing loss (so can mimic non-syndromic) but individuals develop other health concerns later in childhood or adulthood. This is one of the reasons a genetic evaluation with genetic testing is recommended for all children who have congenital hearing loss.

About 70-80% of congenital hearing loss is non-syndromic sensorineural hearing loss (SNHL), while the remaining 20-30% is syndromic. Genetic hearing loss may be inherited in a multiple ways. For non-syndromic hearing loss, it may be inherited in an autosomal recessive (75-80%), autosomal dominant (15-20%), X-linked (<2%), or mitochondrial pattern (<1%). The most common non-syndromic genetic cause is due to pathogenic variants in the GJB2 gene (also called Connexin 26). Other genes that can cause congenital hearing loss include GJB3 (Connexin 31), GJB6 (Connexin 30), and KCNQ4.

What type of genetic or other testing may be recommended?

For infants and children identified to have hearing loss it is important that they undergo a thorough evaluation. In addition to a complete hearing evaluation, a detailed review of their medical history (including prenatal), family history, and physical examination should also be done to help identify the underlying cause for their hearing loss which may aid in establishing appropriate management or treatment approaches.

Multiple organizations have published guidelines for the evaluation of infants and children found to have hearing loss — including the American College of Medical Genetics (ACMG), Joint Committee on Infant Hearing (JCIH), and International Pediatric Otolaryngology Group (IPOG). In summary, it is recommended that:

- CMV testing to be done on all with non-syndromic or sensorineural hearing loss.
- Temporal bone imaging should be considered.
- Genetics evaluation should be offered to families of infants and children with hearing loss in which a environmental cause is not known.
- Genetic testing (multigene panel or single gene testing) should be offered to all patients with sensorineural hearing loss.
- If a genetic syndrome is suspected, genetic testing should be targeted towards the suspected syndrome to confirm diagnosis.

Click here to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

Click here to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

Click here to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.