What Caused my Child's Hearing Loss?

This question can be one of the most pressing for parents when they learn that their child is Deaf or Hard of Hearing.

This section answers many of the questions that families have about the causes of hearing loss, including genetic causes.

The exact cause of a child's hearing loss can be difficult to pinpoint.

About 1 in 500 infants has hearing loss during early childhood. Hearing loss has many causes: some are **genetic** (that is, caused by a baby's genes) or non-genetic, outside factors, often called **environmental factors** (like injuries, illness or certain medications).

For many babies, the cause of hearing loss is unknown. In approximately 25% of all children, it is not possible to determine the cause of hearing loss.

Environmental Factors

Environmental factors account for about 25% of **congenital** hearing loss (hearing loss is present at birth). Let's first take a look at some non-genetic factors that can cause congenital hearing loss:

- maternal infections, such as rubella (German measles), cytomegalovirus, or herpes simplex virus. Now that there is a vaccine for rubella, the most common non-genetic cause of hearing loss at birth is cytomegalovirus.
- prematurity
- low birth weight
- birth injuries
- toxins including drugs and alcohol consumed by the mother during pregnancy

- complications associated with jaundice
- maternal diabetes
- lack of oxygen (anoxia)

Hearing loss can occur at any time in one's life, as a result of an illness or injury. Below are some environmental factors that can cause hearing loss after birth (**acquired hearing loss**):

- ear infections (very common in children)
- medications that are toxic to the ear
- meningitis
- measles
- encephalitis
- chicken pox
- flu
- mumps
- head injury
- noise exposure

Genetic Hearing Loss

Almost 60% of hearing losses are caused by genetics. Genetics is the process of a parent passing certain **genes** to their children. Genes tell the cells of the body how to grow and work. Genetic hearing loss is caused by changes in genes**.** A person's appearance - height, hair color, skin colour, and eye colour - is determined by genes. Other characteristics affected by heredity are:

- likelihood of getting certain diseases
- mental abilities

- natural talents

A trait that is passed down through families (inherited) may:

- have no effect on your health or well-being - for example, the trait might just cause a white patch of hair or an earlobe that is longer than normal
- have only a minor effect - for example, colour blindness
- have a major effect on your quality or length of life

For some genetic disorders, genetic testing can pinpoint the cause.

There are many genes that are involved in hearing. Sometimes, a gene does not form in the way it should. When this happens, it is called a mutation. Some mutations cause **syndromic hearing loss** and others cause **nonsyndromic hearing loss**. Scientists are working to find all of the genes involved in hearing loss.

Sometimes, both genes and environment work together to cause hearing loss. For example, there are some medicines that can cause hearing loss, but only among people who have certain mutations in their genes.

Why is it Important to Know the Cause of Hearing Loss?

Where possible, it is helpful to know the cause for medical reasons. For example, if cytomegalovirus is the cause, then parents will need to be watching for **progressive** hearing loss (hearing loss can get worse over time). In addition, parents can be given information that might prevent or reduce the likelihood of progressive hearing loss. For example, minor head trauma can lead to hearing loss progression in children with enlarged vestibular aqueducts. Avoidance of contact sports may reduce this risk.

Sometimes, knowing the cause can help families to know how well different communication approaches will work. For example, children with profound hearing loss because of a Connexin 26 mutation (a common genetic cause) typically do very well with cochlear implants. This diagnosis may help a family

decide to pursue cochlear implant technology. (This does not mean that a family *should* pursue cochlear implants if there is a Connexin 26 mutation. If they *did* decide to pursue cochlear implants, the outcome is typically positive in terms of developing spoken language.)

Doctors begin by looking at a person's physical features, medical history, and family history. Based on this, they classify the hearing loss in the ways described earlier:

- congenital or acquired
- prelingual or postlingual
- progressive or nonprogressive
- conductive or sensorineural
- syndromic or nonsyndromic
- familial or sporadic

The classifications often point to certain causes. The doctors might ask for more medical tests to look for signs of **syndromic** hearing loss, and they might ask for genetic tests.

What are GJB2 and Connexin 26?

The GJB2 gene contains the instructions for a protein called Connexin 26; this protein plays an important role in the functioning of a part of the ear called the cochlea. The cochlea is a very complex and specialized part of the body that needs many instructions to guide its development and functioning. These instructions come from genes such as the GJB2, GJB3, and GJB6. Changes in any one of these genes can result in hearing loss.

About 40% of newborns with a **genetic** hearing loss (who do not have a syndrome) have a mutation in the GJB2 gene. There are many different mutations in this gene that can cause hearing loss.

Most of the GJB2 mutations are recessive, meaning that a person can have one usual copy of the gene and one copy of the mutation and will have full hearing function. (Everyone has two copies of the GJB2 gene, one from each parent). However, a child who has two copies of a gene with a mutation, one mutation inherited from each parent, will have hearing loss. This means that if both parents have a copy of the gene with a mutation, they can have a child with hearing loss, even though both parents can hear. In fact, most babies with hearing loss are born to parents with typical hearing.

About Genetic Testing

What Is Genetic Testing?

A genetic test involves looking to see if certain mutations are present. A sample usually is taken from one of two different sources: (1) a small sample of a person's blood, or (2) cheek cells from a person's mouth. To get the cheek cells, a small, toothbrush-like swab is rubbed inside a person's mouth. The cheek swab is easy and painless, but the sample obtained from this method is sometimes unstable and might not be usable.

What Are the Benefits of Genetic Testing?

If a mutation is found, it might explain why the person has a condition such as hearing loss. In some cases, knowing what mutation a person has will allow doctors to predict how severe the condition might become and what other symptoms can be expected. Then, the person can get any other medical care that might be needed. Also, knowing the cause of a person's condition will let him or her know what the chances are of passing the condition on to his or her children. It also lets other family members know the chances that they might have a child with the same condition.

What Are the Limits of Genetic Testing?

- Not all of the genes that cause conditions are known. So, even if a condition runs in a family, it might not be possible to find the mutation that causes it.

- Some tests are hard to do. For example, the bigger a gene is, the harder it is to study the whole gene.
- Sometimes, it is not possible to tell if a mutation is the cause of a condition, or just a coincidence.

What Are the Risks of Genetic Testing?

Some people have strong feelings when they get the results of a genetic test. Some people feel angry, sad, or guilty if they find out that they or their child has a mutation. **It is important to remember that everyone carries mutations of some kind, and that a person's genes are no one's "fault."**

Genetic tests are different from other medical tests in that the results provide information about other members of the family, and not just the person being tested. Some family members do not want to know that a mutation runs in their family. Also, because children get their genes from their parents, genetic tests that involve several family members can reveal personal information, such as a child having been adopted or having a different biological father.

Sometimes, people are concerned about keeping the results of their genetic tests private. For example, they do not want their friends, relatives, or co-workers to find out. Genetic testing results are kept private. Test results cannot be seen by anyone who is not involved in the testing unless the person tested or his or her parents or guardians give permission.