

# CHARGE Syndrome

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## What is CHARGE syndrome?

**CHARGE syndrome, or CHARGE association,** is a genetic **syndrome** (a group of symptoms that are part of a medical disorder). Common features that make up CHARGE syndrome are:

- Coloboma of the eye (a hole or gap in a part of the eye)
- Heart defects (issues with the structures and function of the heart)
- Atresia of the choanae (blocked passages in the nose)
- Retardation of growth (delayed growth)
- Genital abnormalities (differences in size, shape, or location of the penis, vulva, testes, or ovaries)
- Ear abnormalities (differences in size, shape, location of parts of the ear)

People with CHARGE syndrome may have one, some, or all of these features.

Hearing loss is common in patients with CHARGE syndrome and ranges from mild to severe. It is also common for people with CHARGE syndrome to have inner ear (**cochlea**) issues, such as **cochlear nerve deficiency or CND** (when the cochlear nerve is too small, or when a person doesn't have a cochlear nerve).

## How is CHARGE syndrome diagnosed?

CHARGE syndrome is diagnosed through genetic testing. If you are diagnosed with CHARGE, it is important to follow up with other medical specialists if you have any other health issues.

A medical professional, typically an **otolaryngologist** (an ear, nose, and throat doctor), will use imaging tests like an MRI or a CT scan of your inner ear to figure out the cause of your hearing loss.

## **What are other conditions that someone might have if they have CHARGE syndrome?**

Inner ear **malformations** (when body parts develop in a way that is not normal) are common in patients with CHARGE. These may include a Mondini malformation, a narrow IAC (internal auditory canal), or cochlear nerve deficiency (CND). Other conditions that can happen with CHARGE include the following:

- Gastrointestinal reflux disease (GERD)
- Cranial nerve abnormalities
- Kidney abnormalities
- DiGeorge sequence
- Cleft lip/palate
- Scoliosis

## **How is CHARGE syndrome treated?**

- Depending on how much hearing loss you have, hearing aids or cochlear implants may help you hear speech (talking) and sounds in your environment better.
- Cochlear implants can help people with CHARGE if they have a cochlear nerve. However, cochlear implants usually don't work as well for people with cochlear nerve deficiency and a narrow IAC. It may work better for these patients to use a combination of speech and sign language for communication.

## Where can I find more information and resources about CHARGE syndrome?

The CHARGE Syndrome Foundation:

[www.CHARGEsyndrome.org](http://www.CHARGEsyndrome.org)



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