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CHARGE SYNDROME CLASSIFICATION, ETIOLOGY AND TREATMENT

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Annotation: Charge syndrome CS is a genetic condition. It is caused by a change or variant in at least one gene. However, we do not find a genetic change in all individuals with features of CS. Most of the time, the genetic change is present in the child but not in the parents. This means that it is a new change in the individual with CS and not inherited. These changes occur with each pregnancy. Because they happen with each pregnancy, it is only important for a person if they happen in a gene that can cause a medical condition. These are not caused by anything that a parent does or does not do.

Key words: Genetic , children , charge syndrome .

It is very rare to have a family history of CS, but families with the condition are known. It is not common for families to have more than one child with CS. It can rarely happen with about a 1% chance that a future child would be impacted by CS. Individuals with CS have a 50% risk of having a child also impacted by CS.

The *CHD7* gene is the most common gene affected in individuals with CS. About two-thirds of individuals show a misspelling (or variant) of the *CHD7* gene. A variant or misspelling in a gene changes the way the body uses that particular gene. Genes can be thought of as recipes for machines the body uses to make the body and do everything the body needs to do on a daily basis. The *CHD7* gene is an important recipe the body uses from the time of conception and throughout a person's life. The recipe that makes the *CHD7* protein or machine can be changed by a variant or misspelling. If the recipe is changed, the recipe is put together differently. If the recipe is put together differently, the machine that it makes will work differently.

If an individual has features that are suspicious for CS, a genetic test can be performed to confirm the diagnosis. This genetic test is helpful to find out if a child has CS. There are other tests that are also done if this test does not show the gene mutation. Knowing the gene change is important for an individual with features of CS because other conditions can cause the same symptoms. The point of genetic testing is always to fully understand a patient's diagnosis. If we confirm the diagnosis, we can make sure an individual is getting the proper and proactive care they need.

Due to the wide range of findings in children with CS, the diagnosis can be hard to make. Every individual with CS is unique, even within families where CS was inherited or happened in another pregnancy. The below symptom lists were defined to improve the ability to detect and diagnose CS.

Symptoms common to CHARGE syndrome include:

- Eyes: A coloboma (missing piece of the eye) in some parts of the eye (including the iris or colored part of the eye), small eyes or missing eye(s) can impact vision
- Throat: Passages that go from the back of the nose to the throat can be narrow (stenosis) or blocked (atresia). This can cause problems with swallowing, breathing and choking.
- Nose: Decreased or no sense of smell
- Face: Paralysis or weakness on one or both sides of the face.

- Ears: Changes to the way the ear was put together can lead to hearing loss and balance problems.

Even with the above symptom lists, it can be hard to make a definitive diagnosis of CS. Genetic tests for CHD7 mutations are not always available. Also not all patients with CS will have CHD7 changes.

Best practice for CS includes a coordinated team evaluation to assess a wide range of medical and developmental conditions. This helps to make a correct diagnosis and a management plan. A medical geneticist familiar with CS should be part of this process.

Early on, infants and toddlers with CS often need medical care and therapies. This is due to major health issues (such as: airway, breathing, heart defects, other birth defects, feeding issues). As an infant's medical conditions stabilize, a focus on hearing, vision, and development is vital. This will help the child to communicate and learn. Without the ability to effectively communicate, children with CS often have challenging behaviors. This is often due to frustration or used as another way to communicate. Some of these problems likely stem from chronic health issues and challenges with vision and hearing. As the care of children with CS improves, the goal is to provide the right treatment as early as possible. It can be overwhelming to think about all of an individual's needs at once. It can help to think of all the ways that day-to-day events can be used to build the child's skills. Reading and interacting with children with CS promotes development and learning with each interaction.

Development of effective communication skills is a common challenge for children with CS. This can affect their day-to-day life. Most babies with CHARGE syndrome have hearing and vision challenges that can delay their development. Balance and orientation in space is also often a challenge. Further, understanding a person with CS's needs throughout the lifespan is vital. Having a care plan in place that changes as a person with CS grows can ensure quality of life and optimal outcome. For example, all individuals with CS should be monitored for feeding problems like aspiration or things going into the lungs or windpipe when a person eats or swallows their spit. This is important to know as soon as possible. Our team or your care team should monitor for this as a person grows.

As we have seen, care and the journey of an individual with CS is a complex and unique experience. Individuals often need long-term medical and educational support. Timely support helps them meet their potential. Early and effective care and support helps children with CS participate meaningfully at home, school, and in their community. Families know their loved one best, and their priorities and goals should stay at the forefront of care. Other important care should be provided, and our team is here to help you make a proactive plan moving forward.

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