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Perspective

DiGeorge Syndrome: Effect on 22nd Chromosome in Children

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DESCRIPTION

A chromosomal condition called DiGeorge Syndrome (DGS), it's commonly affects the 22nd chromosome. Poor body system development can results in a variety of medical issues, such as behavioral issues, a cleft palate, and heart defects. DiGeorge syndrome is also known as 22q11.2 deletion syndrome. A minor deletion at q11.2 on the 22nd chromosome is present in about 90% of those who have the disorder. Other names for the condition include CATCH22, conotruncal syndrome, Shprintzen syndrome, and velocardiofacial syndrome. One in 4,000 people are affected to the DiGeorge syndrome. The syndrome typically begins during fertilization either on the maternal or paternal side. It may happen during the time of fetal development.

The common signs and symptoms may include behavior problems, breathing problems, delayed growth, poor muscle tone, cleft palate, respiratory problems, frequent infections, cyanosis, thymus gland abnormalities, hearing impairment, visual abnormalities, autoimmune diseases, parathyroid gland abnormalities, schizophrenia, altered kidney functions etc.

Numerous body systems emerge as a result of the deletion of chromosome 22 in the DiGeorge syndrome. As a result, the disease may lead to a number of developmental mistakes in the foetus. The complications which include in DiGeroge syndrome are hypothyroidism, heart defects, cleft palate, thymus gland dysfunction, psychiatric disorders, and other problems such as kidney impairment, visual problems, short stature etc.

Diagnosis

DiGeorge syndrome (22q11.2 deletion syndrome) is generally diagnosed through a laboratory test that can find the deletion in chromosome 22. Based on clinical observation of numerous symptoms in various organs, DGS is frequently identified from birth or infancy. A genetic test can be done to confirm the diagnosis.

Treatment

The treatment and therapy for the DiGeroge syndrome may include interventions for:

Hypo-parathyroidism: Calcium and vitamin D supplements can be used to treat hypoparathyroidism.

Heart problems: The majority of 22q11.2 deletion syndrome-related to heart abnormalities, an immediate postnatal surgery to repair the heart and increase the flow of oxygen-rich blood.

Restricted thymus gland activity: If child has some thymic activity, infections could be common but not always serious. The same treatment is typically given to children with these diseases such as colds and ear infections. Most of kids with impaired thymic function receive their vaccines according to schedule. Immune system performance often gets better with age for kids with mild thymus dysfunction.

Severe thymus dysfunction: In this case, children's are at risk for a number of serious infections if the thymus is severely impaired or absent. A transplant of thymus tissue, specific bone marrow cells or specialized disease-fighting blood cells is necessary for treatment.

Cleft palate: A cleft palate or other abnormalities of the palate or lips can be surgically corrected.

Overall development: A variety of therapies such as speech therapy, occupational therapy and developmental therapy which helps to treat the children's.

Psychiatric treatment: If children's is diagnosed with depression, autism spectrum disorder, Attention-Deficit/Hyperactivity Disorder (ADHD) or any other mental health or behavioral difficulties, the psychiatric treatment may be advised.

Management of other conditions: These could involve taking care of challenges with eating and growth, hearing or vision issues, as well as other medical disorders.

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