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Perspective

Overview on 22q11.2 Deletion Syndrome: Understanding a Complex Genetic Disorder

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DESCRIPTION

22q11.2 deletion syndrome, also known as DiGeorge syndrome or velocardiofacial syndrome, is a genetic disorder caused by a small missing piece of chromosome 22. This condition affects approximately 1 in 2,000 to 4,000 people, making it one of the most common genetic disorders worldwide. Despite its prevalence, the syndrome is not well known, and many people remain undiagnosed or misdiagnosed.

Symptoms

The symptoms of 22q11.2 deletion syndrome can vary greatly from person to person, making it a complex condition to diagnose and manage. Some common symptoms include heart defects, cleft palate, feeding difficulties in infants, learning disabilities, speech and language delays and immune system problems. Individuals with the syndrome may also experience psychiatric conditions such as anxiety, depression and schizophrenia.

Diagnosis

Diagnosing 22q11.2 deletion syndrome can be challenging, as the symptoms can be nonspecific and overlap with other conditions. Genetic testing is the most reliable method of diagnosis and can confirm the presence of the deletion on chromosome 22. However, some cases may be missed if the deletion is small or if not all cells have the deletion. Clinical evaluation, including a physical exam and assessment of developmental and cognitive function, is also essential in identifying the syndrome.

Treatment

All children with 22q require frequent examinations with a team of specialists on a regular basis. This group frequently comprises doctors with specialized training in: bones (orthopedic surgery) (orthopedic surgery), issues with the ears, nose and throat (otolaryngology) problems with the eyes (ophthalmology), Genetics, development, and learning (developmental pediatrics), hearing (audiology) (audiology), heart issues (cardiology), mineral and hormone balance (endocrinology), infection-fighting (immunology) (immunology), kidney issues (nephrology, urology), problems with the brain and nervous system (neurology and neurosurgery), psychological well-being (psychology and psychiatry), problems with conduct (behavioral psychology), concerns about speech, language, and voice (speech-language pathologist), difficulty with learning and cognitive impairments (neuropsychology). Some typical issues that may necessitate therapy include:

Low calcium: This is typical in children with the syndrome, especially when they are young. Yet, it can also occur at periods of stress, such as puberty or following surgery. Calcium and vitamin D supplements may be necessary for your youngster.

Development problems: Young children with 22q11.2DS may experience delays in meeting developmental milestones. Parents should explore Physical Treatment (PT), Occupational Therapy (OT), and speech therapy for their child, according to the International 22q11.2 Deletion Syndrome Foundation.

Management

Management of 22q11.2 deletion syndrome requires a multidisciplinary approach and depends on the individual's specific symptoms and needs. Early intervention is crucial, as many of the conditions associated with the syndrome can be treated or managed with appropriate medical and educational interventions. Treatment options may include surgery for heart defects or cleft palate, speech therapy, occupational therapy, and medication for psychiatric conditions.

Education and awareness

Despite being one of the most common genetic disorders, 22q11.2 deletion syndrome remains largely unknown to the general public and even some healthcare professionals. Increased education and awareness about the condition can help improve early diagnosis and appropriate management, leading to better outcomes for individuals with the syndrome.

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CONCLUSION

22q11.2 deletion syndrome is a complex genetic disorder that can affect many aspects of a person's health and development. Diagnosis and management require a multidisciplinary approach, and early intervention is crucial to improving outcomes. Increased education and awareness about the syndrome can help ensure that individuals with the condition receive appropriate care and support. While there is no cure for 22q11.2 deletion syndrome, with the right interventions and support, individuals with the condition can lead healthy, fulfilling lives.