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Opinion Article

Clinical Symptoms and Diagnosis of Jacobsen Syndrome

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DESCRIPTION

Jacobsen Syndrome (JBS) is a rare contiguous gene syndrome whereas 85%–92% of cases has a *de novo* origin monosomy 11q, also known as Jacobsen syndrome and brought on by a portion of chromosome 11's long arm being deleted. About 56% of people with the 11q terminal deletion between 7 and 20 Mb who also have CHD exhibit growth retardation, intellectual impairment, and recognizable facial dysmorphism. Most common clinical features include skull deformities, hypertelorism, ptosis, coloboma, downslanting palpebral fissures, epicanthal folds, broad nasal bridge, short nose, v-shaped mouth, small ears, and posteriorly rotated ears. Prenatal and postnatal physical growth retardation, psychomotor impairment, and distinctive facial dysmorphism are also common. Pancytopenia, thrombocytopenia, and abnormal platelet function are frequently present at birth. Malformations of the heart, kidney, gastrointestinal system, genitalia, central nervous system, and bones are common in patients. Immunological, hormonal, ocular, and hearing issues could also be present. About 5 to 10 percent of all 11q-patients get HLHS Approximately 20% of infant deaths occur in the first two years of life, with CHD complications and bleeding brought on by thrombocytopenia being the most frequent causes of death in JBS that increase the range of molecular and clinical problems in this syndrome and improves our understanding of the variety in the mutation locations in this area.

Symptoms

Children's deviant and sluggish growth is observed in both the womb and after delivery, which is one of the early indications of JB syndrome many Jacobsen syndrome sufferers will be shorter than average when they reach adult height. Additionally, they could have macrocephaly or larger-than-normal heads.

Physical symptoms include distinctive facial features. These include:

- Wide-set eyes with droopy eyelids.
- Tiny and drooping ears
- Broad nasal bridge.
- Rounded lip corners.
- Short lower jaw
- Narrow upper lip

The inner corners of the eyes are concerned with skin folds.

Jacobsen syndrome is frequently followed by cognitive impairment which may result in delayed development, and affect the acquisition of speech and motor abilities in some babies who have trouble eating. A significant number will also have learning impairments. The Jacobsen syndrome also manifests as behavioral issues. These might be connected to addictive behavior, being easily distracted, having a short attention span, and people suffering from ADHD and Jacobsen syndrome.

Causes

Deletion of chromosomal 11 genetic materials causes Jacobsen syndrome. Each affected person has a different type of deletion, with the majority losing 5 million to 16 million DNA-building pieces. Chromosome 11's tip is added in the deletion of all affected individuals when compared to lesser deletions, larger deletions produce more severe indications and symptoms. Jacobsen syndrome does not typically run in families. Only 5 to 10% of cases include a child inheriting the illness from a parent who is not affected. Chromosome 11 contains their parents' genetic information, which is rearranged but remains as a balanced translocation. Many of these genes are still poorly understood. However, the genes in this area are essential for the suitable growth of numerous bodily organs, such as the heart, brain, and facial features only a less number of genes have been identified as potential causes of the distinctive characteristics of Jacobsen syndrome.

Treatment

There is no known treatment for Jacobsen syndrome; instead, each patient's specific indications and symptoms are addressed. The coordinated efforts of a group of different professionals may be necessary throughout treatment. Regular monitoring is needed for those with thrombocytopenia, or low platelet counts. There may be a need for blood or platelet transfusions before or

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during operations many drugs can be used to treat the consequences of various congenital cardiac defects. Surgery may be necessary to correct some of the abnormalities of the condition.

Those with specific cardiac defects would require antibiotics before any operation due to the possibility of bacterial infection of the heart lining and valve eye disorder can be treated with surgery, eyeglasses, contact lenses, or other treatments for vision problems. Orthopedic treatment may be used to treat abnormalities of the bones, muscles, tendons, joints, and other body parts, possibly in conjunction with surgery. Physical therapy's benefits might improve balance and movement. Early intervention is essential if children with disabilities are to reach their full potential.

CONCLUSION

Infancy and adolescence are common in Jacobsen Syndrome (JBS), a rare chromosomal disorder with variable phenotypic expressivity, which is discovered through clinical examination, hematological tests, and cytogenetic studies. The diagnosis of JBS during pregnancy and fetal ultrasonography abnormalities are uncommon.