Journal of Genetic Syndromes & Gene Therapy

OPEN ACCESS Freely available online

Commentary

Genetic Abnormalities Caused due to Noonan Syndrome

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DESCRIPTION

Relatively frequent developmental in Noonan syndrome exhibits undersize growth, distinctive facial dysmorphism, hypertrophic cardiomyopathy, variable degrees of cognitive impairment, and defects in skeletal, ectodermal, and hematologic development. It was established that genetically diverse and heterozygous mutations in nine genes (*PTPN11, SOS1, KRAS, NRAS, RAF1, BRAF, SHOC2, MEK1,* and *CBL*) are responsible for Noonan syndrome. A tiny percentage of NS patients have recently been found to have KRAS gene mutations. All fetuses with poly hydramnios, pleural effusions, edema, and high nuchal fluid and a normal karyotype should be assessed for NS utilizing blood, chorionic villi, and amniotic fluid samples for mutations. With special care and counseling, the majority of children with NS will mature and function normally in adulthood.

Diagnosis

Skeletal abnormalities, hypertrophic cardiomyopathy, and Congenital Heart Defects (CHD) are common indicators of facial dysmorphic traits are the symptoms of NS include cognitive deficits, cryptorchidism, and ectodermal abnormalities. Noonan syndrome's high phenotypic variety can be partially ascribed to molecular anomalies. Although the prevalence of the distinguishing qualities among those who are affected is depending on the patient's age, the diagnosis of NS is based on the clinical symptoms. Making the diagnosis can be quite difficult, especially for adults. There is a great deal of variance in expression, and the phenotype is less noticeable as people get older. Numerous rating methods have been developed to help the diagnostic process. Skeletal abnormalities, hypertrophic cardiomyopathy, and Congenital Heart Defects (CHD) are common indicators of facial dysmorphic traits are the symptoms of NS include cognitive deficits, cryptorchidism, and ectodermal abnormalities. Noonan syndrome's high phenotypic variety can be partially ascribed to molecular anomalies. Although the prevalence of the distinguishing qualities among those who are affected is depending on the patient's age, the diagnosis of NS is based on the clinical symptoms. Making the diagnosis can be quite difficult, especially for adults. There is a great deal of variance in expression, and the phenotype is less noticeable as people get older. Numerous rating methods have been developed to help the diagnostic process.

Prognosis

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.

Symptoms

Noonan syndrome symptoms can range widely from moderate to severe depending on the individual certain traits might be connected to the particular gene where the mutation occurred.

Facial traits

One of the primary clinical characteristics that support a diagnosis of Noonan syndrome is facial appearance characteristics which may be more obvious in infants and young children but they alter as people get older. These distinctive characteristics get softer as people age.

- Droopy lids and wide-set, downward-slanting eyes. Irises are a light blue or green color.
- Rotated rearward and low-set ears.
- Wide base and a bulbous tip of the nose, which is sunken at the top.
- The upper lip has a huge peak and the mouth has a deep groove between the nose and the mouth. With time, tit increase that

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Received: 02-Jun-2022, Manuscript No. JGSGT-22-18683; **Editor assigned:** 06-Jun-2022, Pre QC No. JGSGT-22-18683 (PQ); **Reviewed:** 20-Jun-2022, QC No. JGSGT-22-18683; **Revised:** 27-Jun-2022, Manuscript No. JGSGT-22-18683 (R); **Published:** 04-Jul-2022, DOI: 10.35248/ 2157-7412.22.13. 369.

Citation: Poole H(2022) Genetic Abnormalities Caused due to Noonan Syndrome.Genet Syndr Gene Ther.13:369.

connects the corner of the lips to the edge of the nose develops a deep groove. The lower jaw may be tiny, the palate may be strongly arched, and the teeth may be misaligned.

- While facial characteristics may initially seem coarse, they become crisper with age. The face could seem lifeless and drooping.
- The head may look big, with a high forehead and a low hairline on the back of the head.
- As we age, our skin may appear transparent and thin.
- Noonan syndrome can interfere with usual growth. Many kids with Noonan syndrome don't grow as fast as they should. There may be the following problems:
- The weight at birth is probably usual, although growth decreases with age.
- Difficulties eating might lead to poor nutrition and weight gain.
- Low growth hormone levels could exist.
- The growth surge that typically occurs throughout adolescence may be postponed.
- By adulthood, some persons with Noonan syndrome may attain average height, but low stature is more prevalent. However, growth sometimes continues into the late adolescent years since this illness causes a delay in bone maturation.

Causes

Noonan syndrome is the outcome of a genetic mutation. Numerous genes may experience these alterations because of the errors in these genes, and permanently active proteins are produced. The constant activation of proteins disrupts the normal process of cell growth and division because these genes are important in the formation of many tissues throughout the body.

The following mutations can result in Noonan syndrome:

- Inherited A child has a 50% risk of having Noonan syndrome if one of their parents has the disorder (autosomal dominant) and has the faulty gene.
- Random when a novel mutation occurs in children without a genetic predisposition for Noonan syndrome, the condition can arise.

CONCLUSION

Recent discoveries derived from a massive disease gene hunting effort have established that NS, one of the most common developmental disorders in man, is caused by genetic lesions promoting up regulation of RAS signaling. These discoveries also documented that aberrant activation of RAS signaling, particularly through the MAPK cascade, underlies several clinically related disorders that today are grouped within the NCFCS family also known as RASopathies. The disruption of this signaling system also constitutes one of the most frequent occurrences impacting developmental processes, based on the relatively high prevalence of several of these illnesses.