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Commentary

Diagnosis and Treatment of Turner's Syndrome

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DESCRIPTION

Turner Syndrome (TS), also known as 45, X or 45, X0, is an X chromosomal deficiency in females. The affected people exhibit a variety of signs and symptoms. At birth, it's common to see a short, webbed neck, low-set ears, and a low hairline at the nape of the neck, short height, and swollen hands and feet. Without hormone therapy, women who are impacted typically don't get menstrual cycles, and they can't get pregnant without modern reproductive technology. Diabetes, low thyroid hormone, and heart abnormalities are more common in people with the disease.

The majority of Turner Syndrome patients have average intelligence, but difficulty with spatial visualisation, which may be necessary to learn mathematics. Additionally, vision and hearing issues are more frequent than normal. Turner syndrome is not usually inherited; rather, it develops during the production of a parent's reproductive cells or during the first cell division stage of development. A chromosomal abnormality in which all or even a portion of one of the X chromosomes is absent or mutated is the cause of Turner syndrome.

People with TS typically have 45 chromosomes in part or all of its cells, compared to the average of 46. When the chromosomal abnormality only affects a small number of cells, it is referred to as TS with genetic mutations. Typically, there are fewer symptoms in these situations.

Physical symptoms and genetic testing are used to make the diagnosis. One in 2,000 to one in 5,000 female infants have Turner syndrome. Nearly all religions and geographic areas are impacted. Generally patients with TS have shorter life spans because of diabetes and heart issues.

Causes

Turner syndrome is caused by the absence of one complete or partial copy of the X chromosome in some or all the cells. The abnormal cells may have only one X (monosomy) (45,X) or they may be affected by one of several types of partial monosomy like a deletion of the short p arm of one X chromosome (46,X,del(Xp)) or the presence of an isochromosome with two qarms (46,X,i(Xq)) Turner syndrome has distinct features due to the lack of pseudoautosomal regions, which are typically spared from X-inactivation. In mosaic individuals, cells with X monosomy (45,X) may occur along with cells that are normal (46,XX), cells that have partial monosomies, or cells that have a Y chromosome (46,XY).

Diagnosis

Diagnosis to Turner Syndrome is done in 2 stages. They are Prenatal and post natal stage.

Prenatal: During pregnancy, chorionic villus sampling or amniocentesis can be used to identify Turner syndrome. Generally, abnormal ultrasound results can detect infants with Turner syndrome (i.e., heart defect, kidney abnormality, cystic hygroma, ascites). Abnormalities on ultrasonography were found in 67.2% of Turner syndrome cases that were prenatally diagnosed. One anomaly was found in 69.1% of patients, and two or more anomalies were found in 30.9% of instances. A triple or quadruple maternal serum screen that is abnormal may also point to a higher risk of Turner syndrome.

Compared to those diagnosed based on ultrasonographic abnormalities, foetuses detected by positive maternal serum screening are more frequently found to have a mosaic karyotype, and inversely, those with mosaic karyotypes are less likely to have associated ultrasound abnormalities.

Postnatal: During the Diagnosis Physical symptoms and markers are first recognised, and then a genetic test is performed. Physical symptoms and markers are first recognised, and then a genetic test is performed to confirm the diagnosis. Cardiovascular issues, particularly aortic or pulmonary stenosis, and feeding issues in babies are physiological signs that frequently confirm a WS diagnosis. Developmental delays are frequently perceived as the initial sign of the syndrome.

Treatment

As a chromosomal condition, there is no cure for Turner syndrome. However, much can be done to minimize the symptoms. While most of the physical findings are harmless, significant medical problems can be associated with the syndrome. Most of these significant conditions are treatable with surgery and other therapies including hormonal therapy.

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