Saethre–Chotzen Syndrome

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Abstract- *Saethre–Chotzen syndrome (SCS) is an inherited autosomal dominant genetic disorder associated with craniosynostosis caused by microdeletion of 7p21 chromosome resulted from novel mutations and abnormalities may be influenced by parental age. It is characterized by droopy eyelids (ptosis), widely separated eyes (hypertelorism), and modest hand and foot deformities (syndactyly). It is diagnosed prenatal period using aminocentesis and chorionic vili sampling. The overall diagnosis of SCS is primarily based on clinical findings and observations based on dysmorphology examination and radiographic evaluation. Cranioplasty is the only management for this disorder, after cranial reconstruction surgery child may be needed to wear a moulding helmet or other kind of head protection until the cranial bones have settled into position.*

Keywords- Saethre–Chotzen syndrome (SCS), chromosomal defect, cranioplasty

I. DEFINITION

Saethre–Chotzen syndrome (**SCS**), also known as **acrocephalosyndactyly type III**, is a rare congenital disorder associated with craniosynostosis (premature closure of one or more of the sutures between the bones of the skull). The shape of the head and face is affected, resulting in a cone-shaped head and asymmetrical face. SCS patients also exhibit droopy eyelids (ptosis), widely separated eyes (hypertelorism), and modest hand and foot deformities (syndactyly).

Mild to moderate intellectual or learning difficulties may be present in people with more severe forms of SCS. Some people with SCS may require medical or surgical treatment, depending on the severity of their condition.

Causes:

SCS is inherited as an autosomal dominant characteristic in most cases. However, children with a 7p21 microdeletion (chromosome harbouring the SCS locus) occasionally acquire additional problems and, in most cases, display substantial neurological impairments. The emergence of novel mutations and abnormalities may be influenced by parental age.

Clinical manifestations:

SCS manifests itself in a variety of ways. Because to undeveloped eye sockets, cheekbones, and lower jaw, the majority of people with SCS have uneven facial characteristics and a somewhat flat face. In addition to the physical defects, patients with SCS suffer from growth delays, resulting in a small stature. Although the majority of people with SCS are intelligent, some may experience mild to moderate mental impairments. When numerous cranial sutures close prematurely, more severe cases of SCS occur, with more devastating facial abnormalities.

Characteristics of Saethre–Chotzen syndrome (**SCS)**

The hand of a 4-year-old boy with syndactyly

- Flat, asymmetric head and face
- Head is typically cone-shaped (acrocephaly) or flat (brachycephaly) but can also be long and narrow (dolichocephaly)
- Head is short from front to back
- Lopsided face
- Low-set hairline causing forehead to appear tall and wide

Defects of the hands and feet

- Webbing (syndactyly) between the second and third finger and between the second and third toes
- Short fingers and toes (brachydactyly)
- Broad thumb and/or a broad hallux (big toe) with a valgus deformity (outward angulation of the distal segment of a bone/joint)
- Hands have a single palmer flexion crease

Ocular defects

A diagram showing a complete cleft lip and palate

- Unevenly positioned eyes that may be crossed (strabismus) or wide-set (hypertelorism)
- Vision problems due to abnormal facial anatomy, which causes mechanical disturbances of the extraocular muscles, resulting in strabismus (crossed eyes)

- Tear duct stenosis (narrowing of the tear duct)
- Drooping eyelids (ptosis)
- Downward slanting palpebral fissures (separation between upper and lower eyelids)
- Nearsightedness (myopia)
- Epicanthal folds (skin folds of the upper eyelid covering the inner corner of the eye)
- Blepharophimosis (bilateral ptosis with reduced size of eyelid)
- Optic atrophy
- Refractory errors

Ear, nose, and mouth defects

• Small, low-set ears with a pronounced (bulging) pinna that can be rotated somewhat backwards
• A somewhat off-center beaked nose with a deviated septum (slightly curved downward at tip).
• Dental anomalies such as enamel hypoplasia (thin enamel due to incomplete production), hyperdontia (extra teeth), and peg teeth are linked to malocclusion (small, abnormally shaped teeth)
• Cleft palate with a prominent arch

Less common defects

- Short stature
- Vertebral fusions
- Congenital heart problems
- Speech problems
- Anal atresia (malformed rectum)
- Undescended testes (cryptorchidism)
- Renal (kidney) abnormalities
- Personality disorders

II. DIAGNOSTIC METHOD

Prenatal diagnosis

Prenatal testing is commonly done between 15 and 18 weeks of pregnancy, and amniocentesis is used to collect DNA from the fetus's cells. During weeks 10–12, chorionic villus sampling (CVS) can be used to obtain DNA from the foetus for prenatal testing. There has recently been a surge in interest in using ultrasound equipment to diagnose embryonic skull anomalies caused by immature cranial suture fusion..

Clinical diagnosis

Clinical findings and observations based on dysmorphology examination (assessing structural flaws) and radiographic evaluation are used to make the overall diagnosis of SCS (X-rays, MRIs, and CT scans).

Molecular/genetic diagnosis

A clinical diagnosis of SCS can be confirmed by utilising DNA analysis, such as sequence analysis, deletion/duplication analysis, and cytogenetics/FISH analysis, to look for mutations in the TWIST1 gene (the only gene known to cause SCS).

Exon 1 (the TWIST1 coding area) sequence analysis is an effective tool for finding the frequency of mutations in the TWIST1 gene. Nose, missense, splice site mutations, and intragenic deletions/insertions are examples of these mutations.

Deletion/duplication analysis detects alterations in the TWIST1 gene that would be missed by sequence analysis. PCR, multiplex ligation-dependent probe amplification (MLPA), and chromosomal microarray are all common approaches (CMA).

Gene/chromosome problems can also be studied through cytogenetic testing and direct gene testing. Using fluorescent in situ hybridization (FISH) and/or comparative genomic hybridization, cytogenetic testing examines chromosomes to detect gains or losses of chromosomes or chromosome segments (CGH). To discover genetic abnormalities, direct gene testing involves blood, hair, skin, amniotic fluid, or other tissues. Direct gene testing can be used to determine if someone has SCS by looking for mutations in the TWIST1 gene in their blood.

III. TREATMENT

Cranioplasty

Cranioplasty is a surgical technique that corrects cranial bones that have fused prematurely. The procedure involves reconstructing and repositioning the bones and sutures to allow for proper growth.

Early childhood midfacial surgery may be required to repair breathing abnormalities, dental malocclusion, and swallowing difficulties. Surgery is also used to treat a cleft palate, which may include the use of tympanostomy tubes. After facial growth is complete, an individual may require orthognathic and/or orthodontic therapy if necessary.

A child may be needed to wear a moulding helmet or other kind of head protection until the cranial bones have

settled into position after cranial reconstruction surgery. This usually takes three months, depending on the age of the child and the severity of the disease.

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