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Review Article

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A Comprehensive Review of Chromosome 6, Partial Trisomy 6q Syndrome

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Abstract

Partial trisomy syndrome of chromosome 6q is a very rare chromosomal disorder in which part of chromosome number six (6q) is present three times (trisomy) instead of twice in the body's cells. Many people with partial trisomy syndrome of chromosome 6q also have characteristic neck abnormalities. In people with partial trisomy syndrome of chromosome 6q, all or part of the distal end (q) arm of chromosome number 6 is present three times (trisomy) instead of twice in the cells of the body.

Keywords: Chromosome 6, Partial Trisomy 6q Syndrome, Human Chromosomal Abnormality, Genetic Disorders.

Overview of Chromosome 6, Partial Trisomy 6q Syndrome

Partial trisomy syndrome of chromosome 6q is a very rare chromosomal disorder in which part of chromosome number six (6q) is present three times (trisomy) instead of twice in the body's cells. Associated symptoms and findings may vary in range and severity from case to case. However, many infants and children with slow physical growth (developmental delay), mental retardation; Malformations of the skull and facial area (craniofacial); neck unusually short and reticulated; They have abnormal bending (flexion) or extension of certain joints in fixed positions (joint contractures) or other physical abnormalities. In most cases, partial trisomy syndrome of chromosome 6q is the result of a balanced translocation in one of the parents.[1]

Clinical Signs and Symptoms of Chromosome 6, Partial Trisomy 6q Syndrome

As mentioned above, the symptoms and physical findings associated with partial trisomy syndrome of chromosome 6q may be variable. However, in many cases, this disorder is associated with developmental delay before and after birth, severe to profound mental retardation, delay in acquiring skills that require coordination of muscle and mental activity (psychomotor retardation), abnormalities Distinctive craniofacial region (craniofacial), musculoskeletal abnormalities or additional physical features are identified.1 Characteristic craniofacial abnormalities may include a small head (microcephaly), abnormal flatness of the face and back of the head (occipital), bulging and widely spaced "almond-shaped" eyes (ocular hypertelorism), or slanted eyelid folds. be down (palm slits). Affected individuals may have a small, bow-shaped mouth with thin lips, a small jaw (micrognathia), and incomplete closure of the roof of the mouth (cleft palate), a large and flat nose, misshapen ears, or thin, bow-shaped eyebrows. In some cases, the fibrous joints (i.e., coronal and sagittal sutures) between certain bones at the front and sides of the skull (frontal and parietal bones) may close prematurely (craniosynostosis), causing the head to grow upward (toricephaly). As a result, the head may appear unusually long, narrow, and pointed at the top, and the forehead may be unnaturally prominent [1].

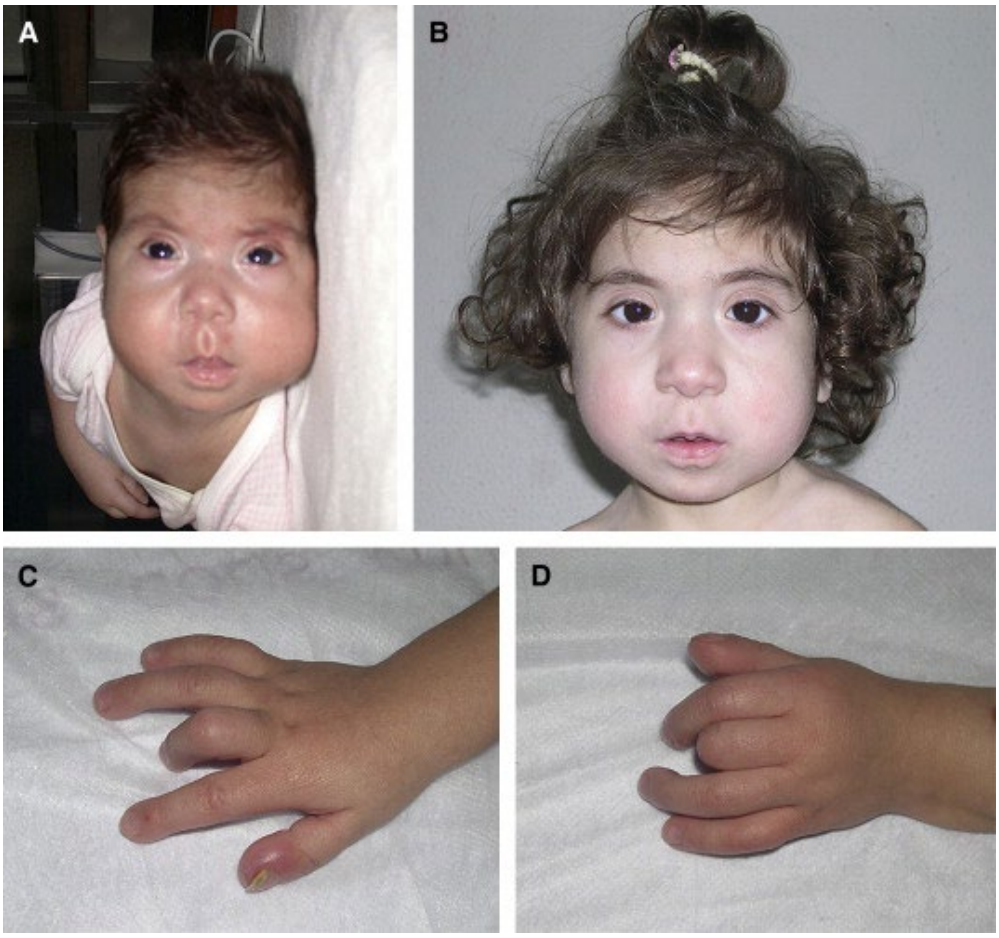

Figure 1: Images of Children with Chromosome 6, Partial Trisomy 6q Syndrome.1

Many people with partial trisomy syndrome of chromosome 6q also have characteristic neck abnormalities. The neck may be unusually short and wide, with abnormal folds at the front (anterior) or sides (lateral), potentially limiting jaw and neck movement. Additionally, the hairline may be unnaturally low on the back of the neck.1 Partial trisomy 6q syndrome is also often associated with abnormal bending (flexion), extension, and stabilization of certain joints (contractions), such as the fingers, wrists, or other areas (eg, elbows, knees, hips), causing Limitation of movement and abnormal postures. Affected individuals may also have adhesions or fusion of fingers or certain toes (syndactyly), deformities in which the hands or feet are twisted out of shape or position (knife hands or club feet), abnormal curvature of the spine. Vertebrae (scoliosis) decrease in chest diameter; or nipples with a long distance [1].

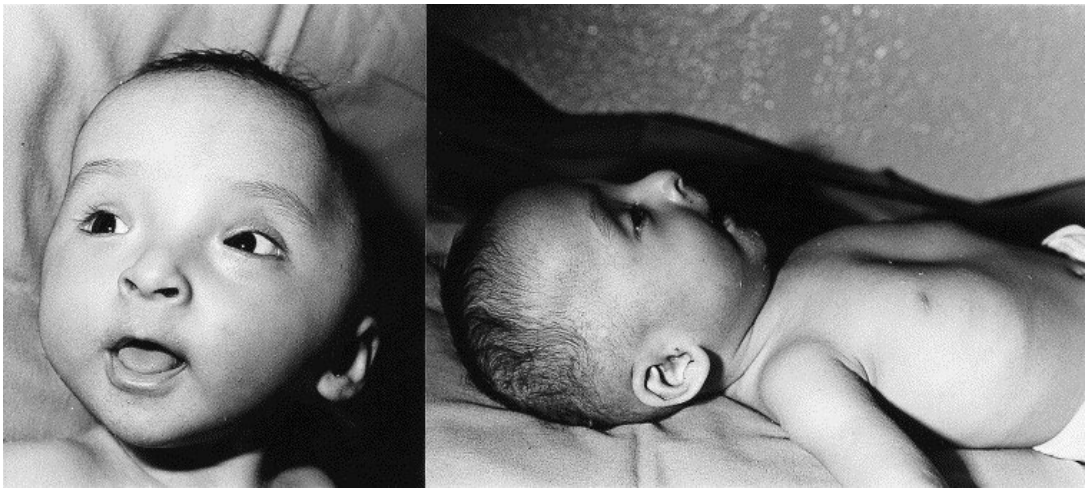

Figure 2: Images of a Baby with Chromosome 6, Partial Trisomy 6q Syndrome.1

Genital abnormalities may also be present. In affected women, skin folds around the vaginal opening (hypo plastic labia) may be underdeveloped. In affected men, genital abnormalities may include an abnormally small penis (micropenis), underdevelopment of the scrotum, abnormal position of the urethra (hypospadias), such as the underside of the penis, or undescended testicles. In rare cases, people with partial trisomy syndrome of chromosome 6q may also have various internal organ abnormalities. These may include cardiac, intestinal, renal, or brain abnormalities [1, 2].

Etiology of Chromosome 6, Partial Trisomy 6q Syndrome

In people with partial trisomy syndrome of chromosome 6q, all or part of the distal end (q) arm of chromosome number 6 is present three times (trisomy) instead of twice in the cells of the body. Reports show that in people in this disorder, the repetitive portion of 6q2 begins at various points (i.e., breakpoints) between bands 6q21 to 6q26 and may extend to the end (or "terminus") of chromosome 6q (qter). The range and severity of associated symptoms may depend on the specific length and location of the 6q repeat.[1, 2] In the majority of reported cases, partial trisomy syndrome of chromosome 6q results from a balanced chromosomal rearrangement in one parent, usually of maternal origin. However, paternal chromosomal rearrangements have been reported in rare cases. The parental chromosomal rearrangement was usually a "balanced translocation". Translocations occur when parts of certain chromosomes break and rearrange, causing the genetic material to move around and change the set of chromosomes. If a chromosomal rearrangement is balanced, meaning it consists of an altered but balanced set of chromosomes, it is usually harmless to the carrier. However, such chromosomal rearrangements may be associated with an increased risk of abnormal chromosomal development in carrier offspring [1, 2].

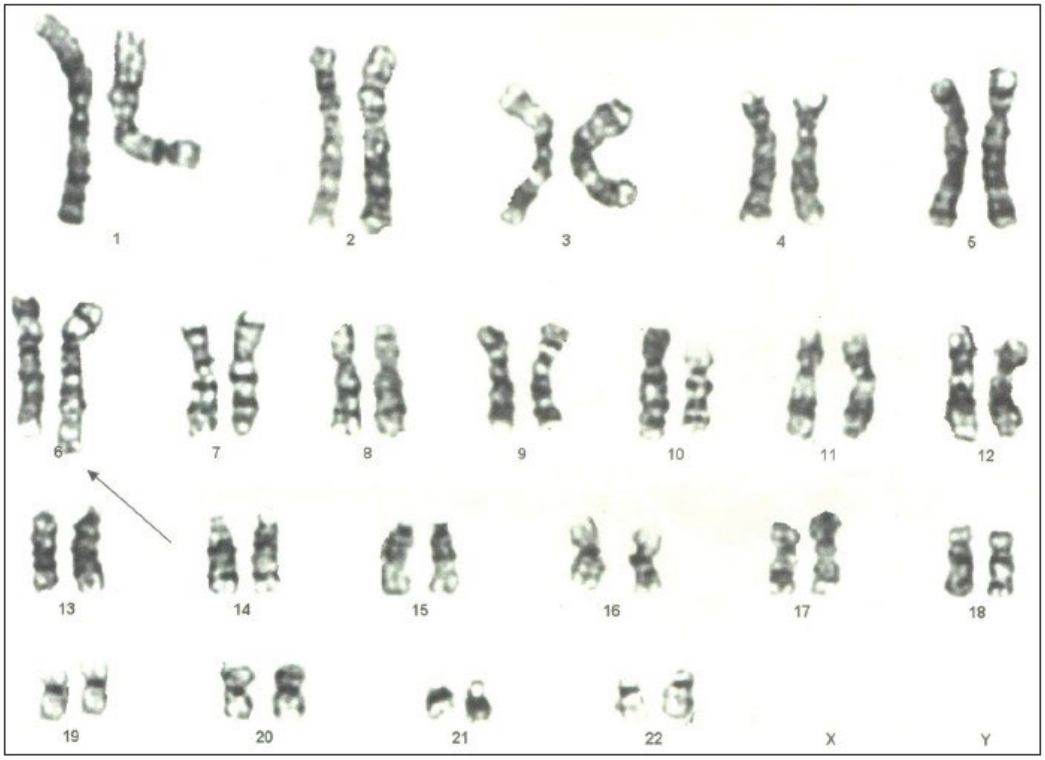

Figure 3: Schematic of the karyotype of a human with Chromosome 6, Partial Trisomy 6q Syndrome.[1]

Rare cases have also been reported in which the chromosomal rearrangement of the parents was reversed. Inversion is characterized by breaking a chromosome in two places and reuniting the segment in the opposite direction. There have also been rare cases where partial trisomy syndrome of chromosome 6q appears to be caused by spontaneous (de novo) errors early in embryonic development. In such new cases, the parents of the affected child usually have normal chromosomes and have a relatively low risk of having another child with chromosomal abnormalities. Chromosomal analysis and genetic counseling are usually recommended for parents of an affected child to help confirm or rule out the presence of a balanced translocation or other chromosomal rearrangement in one parent [1, 3].

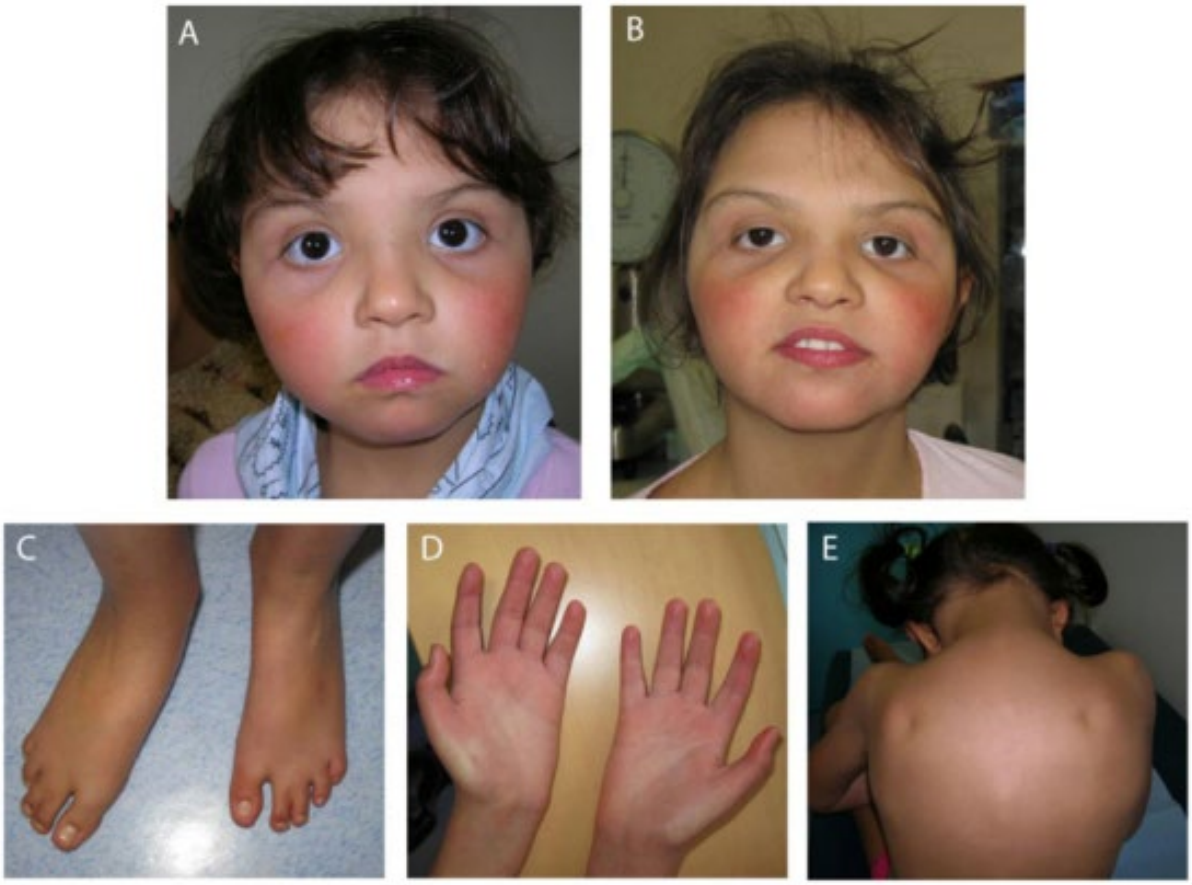

Figure 4: Images of Children with Chromosome 6, Partial Trisomy 6q Syndrome with Related Disorders.[1]

Frequency of Chromosome 6, Partial Trisomy 6q Syndrome

Partial trisomy syndrome of chromosome 6q is a very rare chromosomal disorder that appears to affect males and females equally. Approximately 30 cases have been reported in the medical literature [1, 3].

Disorders Associated With Chromosome 6, Partial Trisomy 6q Syndrome

Additional chromosomal abnormalities may have features similar to those potentially associated with partial trisomy syndrome of chromosome 6q. Chromosomal testing is necessary to confirm the specific chromosomal abnormality present [1, 4].

Diagnosis of Chromosome 6, Partial Trisomy 6q Syndrome

In some cases, partial trisomy syndrome of chromosome 6q may be suggested prenatally (fetal) with specialized tests such as ultrasound, amniocentesis, or chorionic villus sampling (CVS). During fetal ultrasound, reflected sound waves create an image of the developing fetus, potentially revealing certain findings that indicate a chromosomal disorder or other abnormalities. With amniocentesis, a sample of the fluid that surrounds the developing fetus is removed and analyzed, while CVS involves taking tissue samples from part of the placenta. Chromosomal studies performed on such fluid or tissue samples may reveal the presence of partial trisomy syndrome of chromosome 6q. [1, 4]. Partial trisomy syndrome of chromosome 6q may also be diagnosed or confirmed after birth with a thorough clinical evaluation, characteristic physical findings, and chromosomal analysis. Additional specialized tests may also be performed to help identify or characterize specific abnormalities that may be associated with the disorder [1, 4].

Treatment Pathways for Chromosome 6, Partial Trisomy 6q Syndrome

Treatment of partial trisomy syndrome of chromosome 6q is di-

rected towards the specific symptoms that are evident in each individual. Such treatment may require the coordinated efforts of a team of medical professionals such as pediatricians, surgeons, physicians who specialize in disorders of the skeleton, muscles, joints, and related tissues (orthopedics), physical therapists, or other health care professionals. [1, 5]. For some affected individuals, doctors may recommend surgical correction of some craniofacial, limb, genital, or internal organ abnormalities associated with this disorder. In addition, physical therapy, the use of special orthopedic devices, or additional orthopedic techniques, including surgery, may be recommended to help manage musculoskeletal abnormalities, such as joint contractures and scoliosis. The surgical procedures performed depend on the severity of the anatomical abnormalities, their associated symptoms, and other factors. [1, 5]. Early intervention may be important in ensuring that affected children reach their potential. Special services that may be helpful include special education or other medical, social, or vocational services. Genetic counseling will also be useful for the families of affected people. Another treatment of this disorder is symptomatic and supportive [1, 5].

Discussion and Conclusion

Partial trisomy 6q syndrome is also often associated with abnormal bending (flexion), extension, and stabilization of certain joints (contractions), such as the fingers, wrists, or other areas (eg, elbows, knees, hips), causing Limitation of movement and abnormal postures. Genital abnormalities may also be present. In affected women, skin folds around the vaginal opening (hypo plastic labia) may be underdeveloped. In affected men, genital abnormalities may include an abnormally small penis (micropenis), underdevelopment of the scrotum, abnormal position of the urethra (hypospadias), such as the underside of the penis, or undescended testicles. In the majority of reported cases, partial trisomy syndrome of chromosome 6q results from a balanced chromosomal rearrangement in one parent, usually of maternal origin. However, paternal chromosomal rearrangements have been reported in rare cases. The parental chromosomal rearrangement was usually a "balanced translocation". Treatment of partial trisomy syndrome of chromosome 6q is directed towards the specific symptoms that are evident in each individual. For some affected individuals, doctors may recommend surgical correction of some craniofacial, limb, genital, or internal organ abnormalities associated with this disorder [1, 5].

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