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A Comprehensive Review of Chromosome 7, Partial Monosomy 7p Syndrome

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Abstract

Partial monosomy syndrome of chromosome 7p is a rare chromosomal disorder characterized by the deletion (monosomy) of part of the short arm (p) of chromosome number 7 (7p). Associated symptoms and findings may be variable and may depend on the size and specific location of the 7p deleted segment. However, in many cases, there is premature closure of the fibrous joints (cranial sutures) between some of the bones of the skull (craniosynostosis), leading to an abnormal head shape. For example, depending on the specific sutures involved, the forehead may appear unusually "triangular" (trigonocephaly) or the head may appear abnormally long and narrow with a pointed or conical point (toriscephaly). Partial monosomy syndrome of chromosome 7p is also usually characterized by premature closure of one or more fibrous joints (cranial sutures) between certain bones in the skull (craniosynostosis), potentially leading to a deformed skull and abnormal head shape. In people with partial monosomy syndrome of chromosome 7p, there is a deletion (monosomy) of part of the short arm (p) of chromosome number 7. Based on reports in the medical literature, there is considerable variability in the size and location of the 7p deleted segment, potentially affecting the range and severity of symptoms and associated findings. Reported cases include variable "distal" deletions that extend to the terminal band (eg, from 7p13-pter to 7p21,p22-pter) and various "interstitial" deletions (eg, from 7p13 or 7p15 to 7p21).

Keywords: Partial Monosomy Syndrome of Chromosome 7p; Pregnancy Disorder; Chromosomal Disorder; Children's Diseases

Abbreviations: CSF: Cerebro Spinal Fluid; CVS: Chorionic Villus Sampling; VSD: Ventricular Septal Defect.

Overview of Chromosome 7, Partial Monosomy 7p Syndrome

Partial monosomy syndrome of chromosome 7p is a rare chromosomal disorder characterized by the deletion (monosomy) of part of the short arm (p) of chromosome number 7 (7p). Associated symptoms and findings may be variable and may depend on the size and specific location of the 7p deleted segment. However, in many cases, there is premature closure of the fibrous joints (cranial sutures) between some of the bones of the skull (craniosynostosis), leading to an abnormal head shape. For example, depending on the specific sutures involved, the forehead may appear

unusually "triangular" (trigonocephaly) or the head may appear abnormally long and narrow with a pointed or conical point (toriscephaly). Also, affected infants and children may have additional abnormalities in the skull and face (craniofacial). Such abnormalities may include an unusually small head (microcephaly), close-set or large eyes (ocular hypotelorism or hypertelorism), downward folds of the eyelids (cleft palms), or other findings [1]. Partial monosomy syndrome of chromosome 7p may also be characterized by additional physical features such as growth failure, musculoskeletal abnormalities, genital defects, structural abnormalities of the heart present at birth (congenital heart defects), or other abnormalities. In addition, some affected individuals may have varying degrees of mental retardation and delays in acquiring skills that require coordination of mental and motor activities (psychomotor delays) and normal intelligence has also been reported in this syndrome. In most cases, partial monosomy syndrome of chromosome 7p appears to result from spontaneous (de novo) errors in early embryonic development that occur for unknown reasons [1].

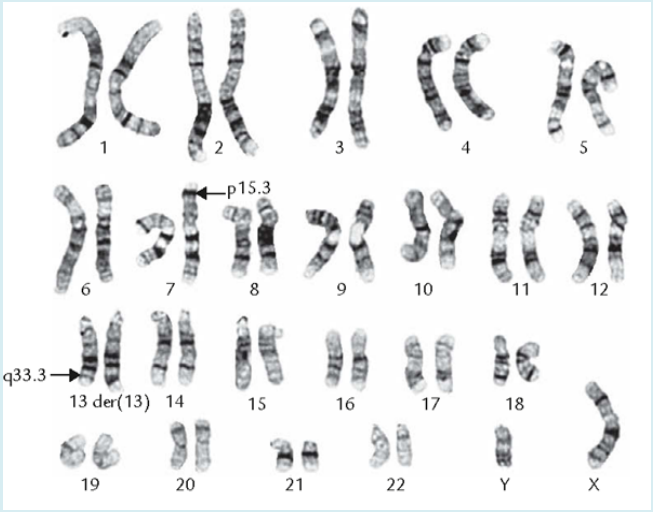

Figure 1: Schematic of the Partial Monosomic Karyotype of Chromosome 7p [1].

Clinical Signs and Symptoms of Chromosome 7, Partial Monosomy 7p Syndrome

As mentioned above, the symptoms and physical findings associated with partial monosomy syndrome of chromosome 7p may vary in range and severity from case to case. However, many affected individuals have developmental delays before and after birth (prenatal and postnatal growth retardation). This syndrome may also be associated with varying degrees of psychomotor retardation and mental retardation. However, as mentioned earlier, some affected individuals may have normal intelligence [1,2].

Partial monosomy syndrome of chromosome 7p is also usually characterized by premature closure of one or more fibrous joints (cranial sutures) between certain bones in the skull (craniosynostosis), potentially leading to a deformed skull and abnormal head shape. The degree and severity of craniosynostosis may vary depending on the specific sutures of the skull involved. For example, according to reports in the medical literature, partial monosomy syndrome of chromosome 7p may be associated with different types of craniosynostosis, such as trigonocephaly or toriscephaly [1,2].

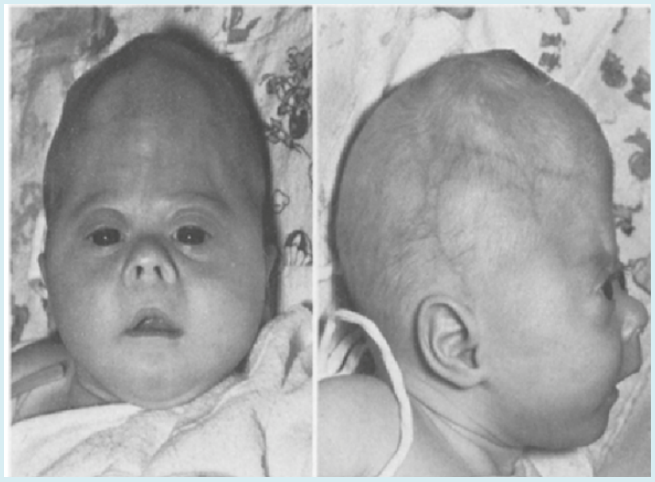

Figure 2: Image of a Baby with Partial Monosomy Syndrome of Chromosome 7p with Related Disorder [1].

In trigonocephaly, premature closure of the suture between the bones that make up the forehead (i.e., the metopic suture) may cause the forehead to appear abnormally narrow, pointed, and "triangular" or "rod-shaped," with an abnormally narrowing of the distance between the eyes (ocular hypotelorism) Torsicephaly (also known as oxycephaly or acrocephaly) is characterized by premature fusion of the suture (i.e., coronal suture) between the bones that make up the forehead and the top of the skull (frontal and parietal bones), and possibly other sutures. It makes the head appear unusually long and narrow, conical or pointed. Additionally, the back of the head (occiput) may appear flat and the forehead unusually prominent. In some cases, other craniofacial features associated with variable craniosynostosis may include an unusually small head (microcephaly), widely spaced eyes (ocular hypertelorism), or other findings. The skull may also look slightly different from one side to the other (cranial asymmetry) [1,2].

In addition, in some cases, craniosynostosis, especially involving two or more cranial sutures, may lead to certain

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neurological complications. Such complications may include hydrocephalus and abnormally increased pressure inside the skull (intracranial pressure).

Hydrocephalus is a condition in which obstruction of the flow or absorption of cerebrospinal fluid (CSF) results in an abnormal accumulation of cerebrospinal fluid, usually under increased pressure. CSF is the watery protective fluid that flows through the cavities (ventricles) of the brain, the canal containing the spinal cord (spinal canal) and the space between the protective membrane layers (meninges) surrounding the brain and spinal cord (i.e. the subarachnoid space). Depending on age of onset and other factors, associated symptoms may include sudden episodes of uncontrolled bioelectrical activity in the brain (seizures), irritability, vomiting, headache, loss of coordination, worsening mental function, or other findings. In severe cases, potentially life-threatening complications may occur [2].

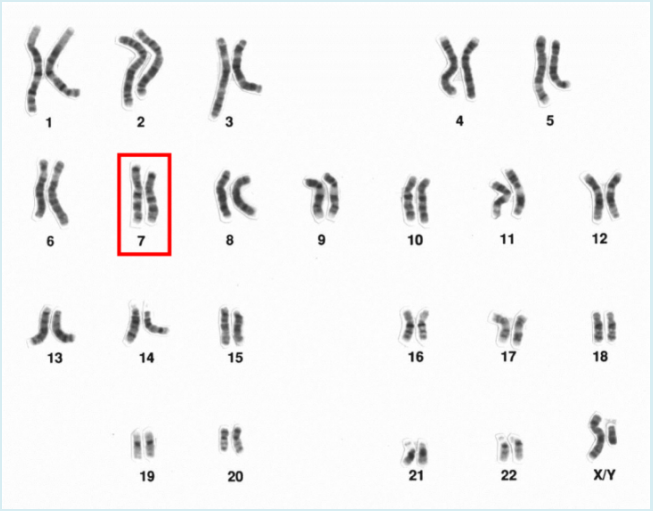

Figure 3: Schematic of Normal Human Male Chromosomal Karyotype [1].

In some affected individuals, partial monosomy syndrome of chromosome 7p may be associated with additional craniofacial abnormalities. Such features may include downward-sloping eyelid folds (palmar clefts), vertical skin folds that may cover the inner corners of the eyes (epicanthal folds); Drooping of the upper eyelid (ptosis); small, maladjusted, misshapen (dysplastic) ears; sunken nasal bridge ("saddle nose"); or other abnormalities [3]. In some cases, partial monosomy syndrome of chromosome 7p may also be characterized by musculoskeletal abnormalities. Reported features include permanent bending of one or more fingers (camptodactyly), abnormally short hands; deformities of thumbs; A deformity in which the top of the foot is elevated and the heel is turned outward ("clubfoot" [i.e., heel talipes]), limited range of motion of certain joints; or other findings [3]. According to some reports, up to 50% of affected individuals may have congenital heart defects. Such heart defects may include an abnormal opening in the partition (septum) that separates the two lower chambers (ventricles) of the heart (Ventricular Septal Defect [VSD]), a hole in the septum that separates the two upper chambers of the heart. It separates (atrial septal defect) or other heart abnormalities, allowing some oxygen-rich blood to circulate in the lungs, potentially leading to increased blood pressure in the lungs (pulmonary hypertension).

Associated symptoms and findings may vary depending on the size, nature, or combination of cardiac abnormalities present and other factors. For example, in some cases, such as those with small isolated VSDs, no symptoms may be apparent (asymptomatic). However, in other cases, such as those with larger VSDs, associated symptoms and findings may include feeding problems, poor growth, difficult or labored breathing (dyspnea), excessive sweating, increased susceptibility to respiratory infections, impaired In the heart's ability to pump blood effectively to the lungs and the rest of the body (heart failure), an enlarged heart, or other abnormalities. In severe cases, congenital heart disease may lead to potentially life-threatening complications [3].

Partial monosomy syndrome of chromosome 7p may also be characterized by additional physical features. Some affected individuals may have abnormal skin bump patterns on the palms. Other reported findings include a highly arched roof of the mouth (palate) or incomplete closure (cleft) of the palate (cleft palate), retraction or downward displacement of the tongue (glossoptosis); Underdevelopment (hypoplasia) of the external genitalia; kidney (renal) defects, such as renal hypoplasia; abnormally small colon (microcolon); or other defects [3].

Etiology of Chromosome 7, Partial Monosomy 7p Syndrome

In people with partial monosomy syndrome of chromosome 7p, there is a deletion (monosomy) of part of the short arm (p) of chromosome number 7. Based on reports in the medical literature, there is considerable variability in the size and location of the 7p deleted segment, potentially affecting the range and severity of symptoms and associated findings. Reported cases include variable "distal" deletions that extend to the terminal band (eg, from 7p13-pter to 7p21,p22-pter) and various "interstitial" deletions (eg, from 7p13 or 7p15 to 7p21). "Distal" refers to farther or farther from a specific reference point, meaning the centromere of a chromosome; "interstitial" means actual, as in between other regions of a chromosome. Evidence from some researchers

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suggests that craniosynostosis is associated with a partial monosomy syndrome. Chromosome is 7p. Partial monosomy syndrome of chromosome 7p appears to be due to partial or complete deletion of 7p21-p22 or, rarely, monosomy 7p13-p14. Further research is needed to learn more about the specific region(s) that may be responsible for the expression of characteristic symptoms and findings in individuals with chromosomal syndromes [4].

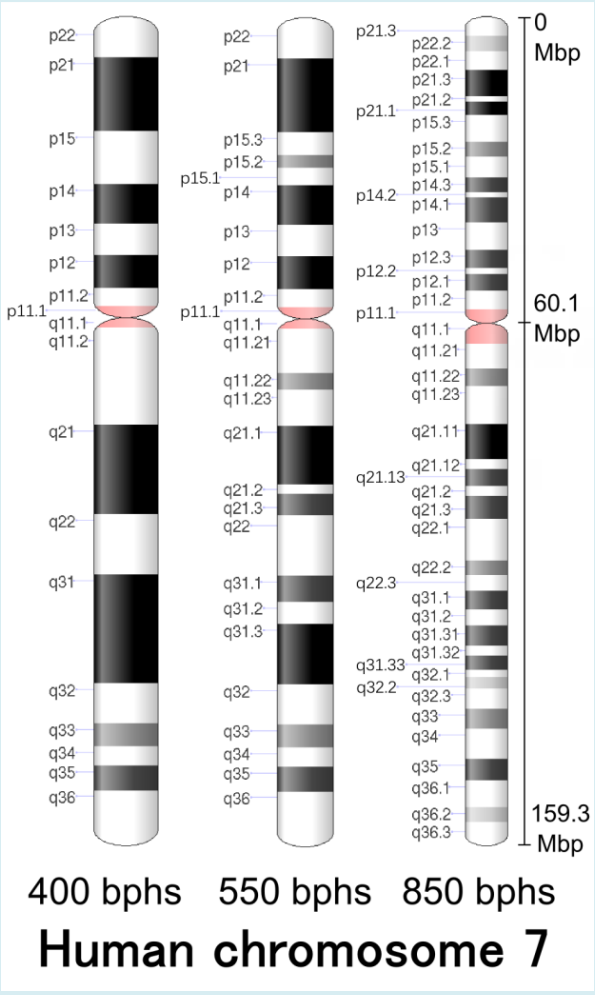

Figure 4: Schematic of physical map of chromosome number 7.

In most cases, partial monosomy syndrome of chromosome 7p appears to result from spontaneous (de novo) errors during early fetal development that occur for unknown reasons. In such cases, the parents of the affected child usually have normal chromosomes and have a relatively low risk of having another child with the chromosomal abnormality [1,4]. Rare cases have also been reported that appear to result from balanced chromosomal rearrangements in one parent. If a chromosomal rearrangement is balanced, meaning that 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. 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 chromosomal rearrangement involving chromosome 7 in one parent [4].

Frequency of Chromosome 7, Partial Monosomy 7p Syndrome

Partial monosomy syndrome of chromosome 7p is a rare chromosomal disorder that appears to affect males and females in relatively equal numbers. More than 30 cases have been reported in the medical literature [1,5].

Disorders Associated with Chromosome 7, Partial Monosomy 7p Syndrome

Additional chromosomal abnormalities may present with symptoms and findings similar to partial monosomy syndrome of chromosome 7p. Chromosomal testing is necessary to confirm the specific chromosomal abnormality present [5].

Diagnosis of Chromosome 7, Partial Monosomy 7p Syndrome

In some cases, the diagnosis of partial monosomy syndrome of chromosome 7p may be suggested before birth (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 characteristic 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 analysis performed on such a fluid or tissue sample may reveal the presence of partial monosomy syndrome of chromosome 7p [5].This syndrome may be diagnosed or confirmed after birth by thorough clinical evaluation, identification of characteristic physical findings, and chromosomal analysis. The diagnostic evaluation may include various studies, including advanced imaging techniques, to help identify or characterize specific abnormalities that may be associated with the syndrome (eg, specific craniofacial defects, musculoskeletal abnormalities). In addition, a complete cardiac evaluation may be recommended to detect any cardiac abnormalities that may be present.

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Such evaluation may include a thorough clinical examination, assessment of heart and lung sounds through the use of a stethoscope, and specialized tests that enable doctors to evaluate the structure and function of the heart (eg, X-ray studies, electrocardiography [ECG], echocardiography) [1,5].

Treatment Pathways for Chromosome 7, Partial Monosomy 7p Syndrome

Treatment of partial monosomy syndrome of chromosome 7p is directed 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; Doctors who diagnose and treat disorders of the skeleton, joints, muscles, and related tissues (orthopedics), cardiologists, doctors who diagnose and treat nerve disorders (neurologists) [1,5].

For infants with craniosynostosis, depending on the number and type of cranial suture(s) involved and other factors, early surgery may be recommended to prevent abnormal head formation, increase cranial capacity, or prevent possible neurological complications. Surgery may also be recommended for some additional craniofacial abnormalities, musculoskeletal defects, or other physical abnormalities associated with this syndrome. In addition, for people with congenital heart defects, treatment may require the administration of certain drugs, surgical intervention, or other procedures. The surgical procedures performed depend on the size, nature, severity, and composition of the anatomical abnormalities, their associated symptoms, and other factors [5]. Early intervention services may also be important in ensuring that disadvantaged children reach their potential. Special services that may be beneficial include special therapy training, physical therapy, or other medical, social, or vocational services. Genetic counseling will also be useful for affected people and their families. Another treatment of this disorder is symptomatic and supportive [5].

Discussion

Partial monosomy syndrome of chromosome 7p is also usually characterized by premature closure of one or more fibrous joints (cranial sutures) between certain bones in the skull (craniosynostosis), potentially leading to a deformed skull and abnormal head shape. The degree and severity of craniosynostosis may vary depending on the specific sutures of the skull involved. For example, according to reports in the medical literature, partial monosomy syndrome of chromosome 7p may be associated with different types of craniosynostosis, such as trigonocephaly or toriscephaly. In addition, in some cases, craniosynostosis, especially involving two or more cranial sutures, may lead to certain neurological complications. Such complications may include hydrocephalus and abnormally increased pressure inside the skull (intracranial pressure). Hydrocephalus is a condition in which obstruction of the flow or absorption of cerebrospinal fluid (CSF) results in an abnormal accumulation of cerebrospinal fluid, usually under increased pressure. CSF is the watery protective fluid that flows through the cavities (ventricles) of the brain, the canal containing the spinal cord (spinal canal) and the space between the protective membrane layers (meninges) surrounding the brain and spinal cord (i.e. the subarachnoid space). According to some reports, up to 50% of affected individuals may have congenital heart defects. Such heart defects may include an abnormal opening in the partition (septum) that separates the two lower chambers (ventricles) of the heart (Ventricular Septal Defect [VSD]), a hole in the septum that separates the two upper chambers of the heart. It separates (Atrial Septal Defect) or other heart abnormalities, allowing some oxygen-rich blood to circulate in the lungs, potentially leading to increased blood pressure in the lungs (pulmonary hypertension).

Associated symptoms and findings may vary depending on the size, nature, or combination of cardiac abnormalities present and other factors. For example, in some cases, such as those with small isolated VSDs, no symptoms may be apparent (asymptomatic). In most cases, partial monosomy syndrome of chromosome 7p appears to result from spontaneous (de novo) errors during early fetal development that occur for unknown reasons. In such cases, the parents of the affected child usually have normal chromosomes and have a relatively low risk of having another child with the chromosomal abnormality. This syndrome may be diagnosed or confirmed after birth by thorough clinical evaluation, identification of characteristic physical findings, and chromosomal analysis. The diagnostic evaluation may include various studies, including advanced imaging techniques, to help identify or characterize specific abnormalities that may be associated with the syndrome (eg, specific craniofacial defects, musculoskeletal abnormalities). For infants with craniosynostosis, depending on the number and type of cranial sutures involved and other factors, early surgery may be recommended to prevent abnormal head formation, increase cranial capacity, or prevent possible neurological complications. Surgery may also be recommended for some additional craniofacial abnormalities, musculoskeletal defects, or other physical abnormalities associated with this syndrome. It is worth mentioning that children with Pfeiffer syndrome, Alperts syndrome and Crouzon syndrome also have bone abnormalities in the skull in the form of craniosynostosis, similar to this syndrome [1-10].

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