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Review Article

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A Comprehensive Review of Chromosome 3, Monosomy 3p Syndrome

Aida Parsa Sadat and Shahin Asadi*

Medical Genetics Director of the Division of Medical Genetics and Molecular Pathology Research. Division of Medical Genetics and Molecular Pathology Research, USA

***Corresponding author:** Shahin Asadi, Medical Genetics Director of the Division of Medical Genetics and Molecular Pathology Research. Division of Medical Genetics and Molecular Pathology Research, Center of Complex Disease, U.S.A, ORCID CODE:0000-0001-7992-7658.

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Abstract

Chromosome 3, Monosomy 3p Syndrome is a rare chromosomal disorder in which the distal (distal) part of the short arm (p) of chromosome number 3 is missing (deleted or monosomic). The range and severity of symptoms and findings may vary. As mentioned above, the symptoms and physical findings associated with monosomy 3p syndrome may vary in range and severity from case to case. However, according to reports in the medical literature, many individuals with low birth weight, significant postnatal growth retardation (postnatal growth retardation); severe to profound mental retardation; Severe delay in acquiring skills that require coordination of mental and motor activities (psychomotor retardation). Chromosome 3p monosomy syndrome is a rare chromosomal disorder in which there is a deletion (monosomy) of the distal (distal) arm of chromosome number 3.

Keywords: Chromosome 3p monosomy syndrome, Chromosome abnormality disorders

Overview of the Chromosome 3, Monosomy 3p Syndrome

Chromosome 3p monosomy syndrome is a rare chromosomal disorder in which the distal (distal) part of the short arm (p) of chromosome number 3 is missing (deleted or monosomic). The range and severity of symptoms and findings may vary. However, associated features often include prenatal and postnatal growth retardation (prenatal and postnatal growth deficiency). severe to profound mental retardation; distinctive malformations of the skull and facial region (craniofacial); eyebrows that grow together (synophry); or excessive hair growth (hypertrichosis). Additional physical abnormalities may also be present. In many cases, the 3p monosomy syndrome appears to occur spontaneously (de novo) for unknown reasons [1].

Clinical Signs and Symptoms of Chromosome 3, Monosomy 3p Syndrome

As mentioned above, the symptoms and physical findings associated with monosomy 3p syndrome may vary in range and severity from case to case. However, according to reports in the medical literature, many individuals with low birth weight, significant postnatal growth retardation (postnatal growth retardation); severe to profound mental retardation; Severe delay in acquiring skills that require coordination of mental and motor activities (psychomotor retardation). excessive hair growth (hypertrichosis); or have specific abnormalities of the skull and facial area (skull face) [1].

Many affected babies have an abnormally small head (microcephaly) that may appear unusually short and wide (brachycephaly), flat back of the head (occiput); or an abnormally long, narrow, and prominent forehead. Additional craniofacial abnormalities may include a triangular face, arched eyebrows that grow together (synophrys); broad and straight nose; an unusually long vertical groove in the middle of the upper lip (philtrum); thin lips; or an abnormally small lower jaw (lower jaw). Affected

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individuals may also have their mouths down. wide-spaced eyes (ocular hypertelorism); vertical skin folds that cover the inner corners of the eyes (epicanthal folds); upward slanting eyelid folds (palm clefts); Drooping of the upper eyelid (ptosis); Or small and misshapen ears are other symptoms of this syndrome [1] (Figure 1).

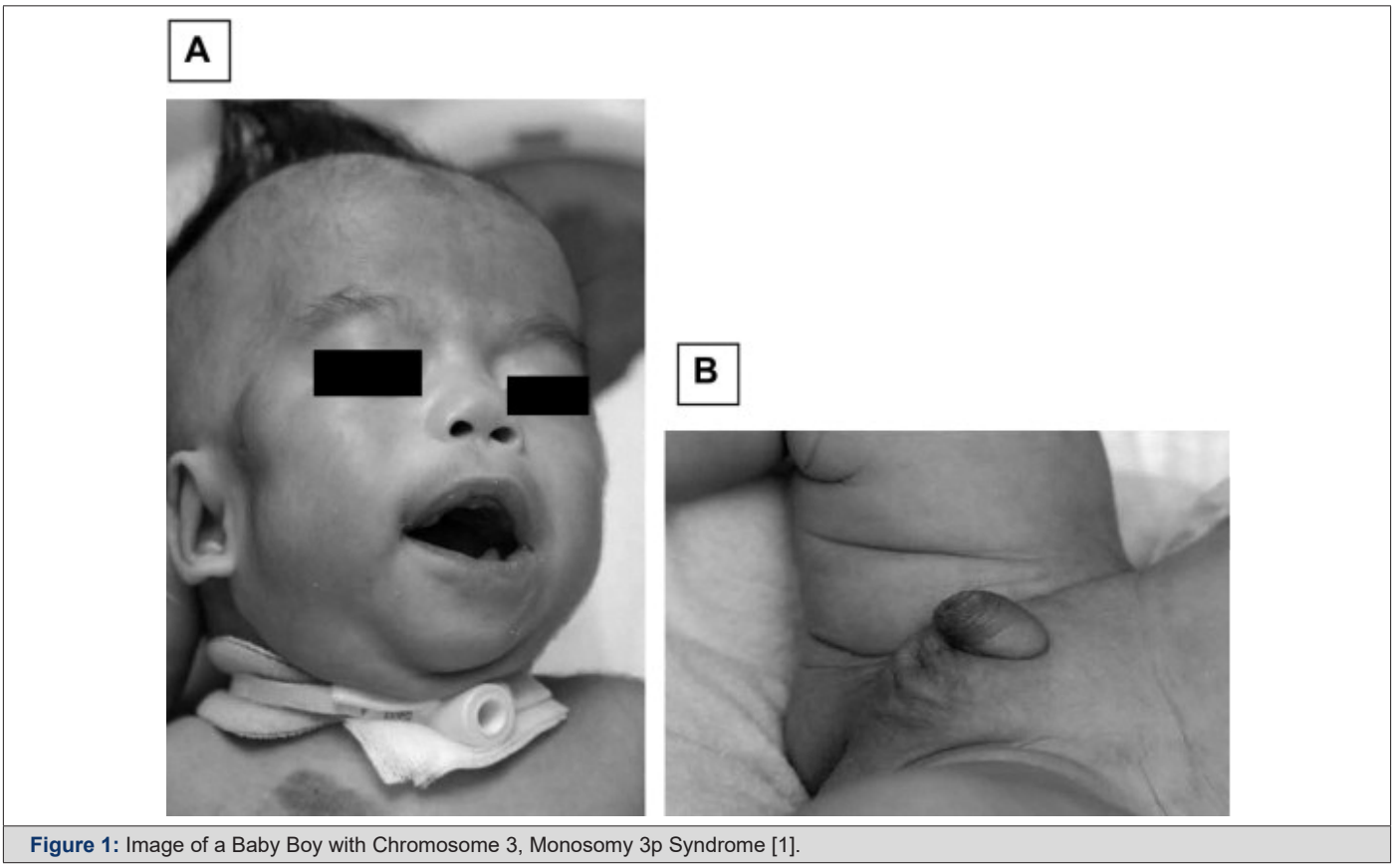

Figure 1: Image of a Baby Boy with Chromosome 3, Monosomy 3p Syndrome [1].

Chromosome 3p monosomy syndrome is also usually associated with an abnormal number of fingers or toes (polydactyly). In many cases, extra fingers may also be present, especially duplication of the pinky or fifth finger (psaxial polydactyly). In addition, in some cases, affected infants may experience duplication of certain toes, especially the fifth toe [1,2].

Reports show that some affected people may have hearing loss or visual impairment. In addition, this disorder may be accompanied by additional physical features, such as an abnormal anterior (anterior) position of the anus, undescended testes into the scrotum in affected men, kidney (renal) defects, structural abnormalities of the heart (congenital heart defects); or have other abnormalities [1,2].

Etiology of the Chromosome 3, Monosomy 3p Syndrome

Chromosome 3p monosomy syndrome is a rare chromosomal disorder in which there is a deletion (monosomy) of the distal (distal) arm of chromosome number 3. The researchers show that the characteristic symptoms and findings of this syndrome result from a deletion of chromosomal material that extends from band 25 on the short arm of chromosome 3 (the breakpoint) to the end or "terminal" of 3p (3p25–>pter) [1,3].

In most reported cases, the monosomy syndrome of chromosome 3p appears to result from spontaneous (de novo) errors very early in embryonic development. 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. All but one case of Monosomy 3p were new (de novo) chromosomal alterations. In one case, the deletion was inherited from a mother who had the same 3p deletion. Potentially, monosomy 3p can result from a translocation or inversion of the parents [1,3].

Translocation occurs when regions of certain chromosomes break apart and are swapped, causing genetic material to move around and changing sets of chromosomes. If such chromosomal rearrangements are "balanced," all chromosomal material is present in two copies but in different locations. Balanced displacements are usually harmless to the carrier. However, such chromosomal rearrangements may be associated with an increased risk of abnormal chromosomal development in the carrier's offspring if inherited in an "unbalanced" state. An unbalanced translocation

occurs when only one of the two chromosomes involved in the translocation is inherited from the carrier parent. The result is either too much (duplication or trisomy) or too little (deletion or monosomy) chromosomal material. An inversion occurs when a chromosome breaks in two places on a single chromosome and the segment between the breaks rejoins the chromosome in the reverse order. Chromosomal analysis and genetic counseling are usually recommended for parents of an affected child to help confirm or rule out a chromosomal rearrangement in one parent [1,3].

Frequency of Chromosome 3, Monosomy 3p Syndrome

Chromosome 3p monosomy syndrome appears to affect males and females in relatively equal numbers. Since the disorder was first reported in 1978 (Verjaal M), approximately 34 cases have been described in the medical literature [1,4] (Figure 2).

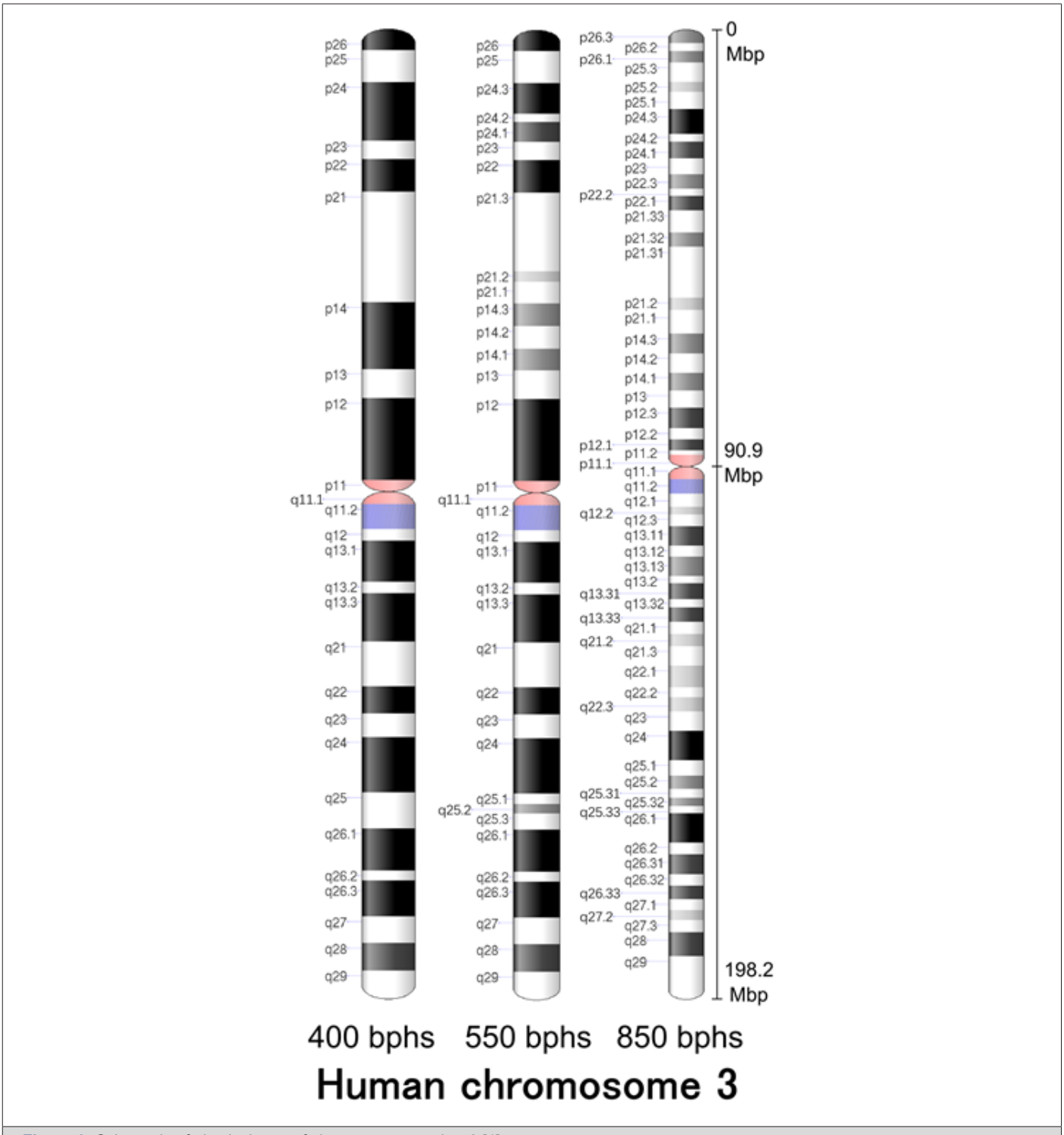

Figure 2: Schematic of physical map of chromosome number 3 [1].

Disorders associated with Chromosome 3, Monosomy 3p Syndrome

Additional chromosomal disorders may have features similar to monosomy 3p syndrome. Chromosomal analysis is necessary to confirm the specific chromosomal abnormality present [1,4].

Diagnosis of Chromosome 3, Monosomy 3p Syndrome

In some cases, monosomy 3p syndrome 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 findings that indicate a chromosomal disorder or other developmental abnormality in the fetus. 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 Monosomy 3p [1,4].

The disorder may also be diagnosed or confirmed after birth based on thorough clinical evaluation, diagnosis of characteristic findings (such as developmental delay, mental retardation, psychomotor retardation, craniofacial abnormalities, etc.) and chromosomal analysis. to be Specialized tests may also be done to help identify or characterize specific abnormalities that may be associated with the disorder [1,4].

Treatment pathways for Chromosome 3, Monosomy 3p Syndrome

Treatment of monosomy 3p syndrome is directed toward 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; neurologists; Doctors who diagnose and treat heart abnormalities (cardiologists); audiologists; or other health care professionals need. In some cases, doctors may recommend surgical repair of certain abnormalities associated with this disorder. The specific surgical procedures performed depend on the severity and location 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, physical therapy, or other medical, social, or vocational services. Genetic counseling will also be useful for families of affected children. Another treatment of this disorder is symptomatic and supportive [1,5].

Discussion

Chromosome 3p monosomy syndrome is a rare chromosomal disorder in which the distal (distal) part of the short arm (p) of chromosome number 3 is missing (deleted or monosomic). The range and severity of symptoms and findings may vary. However, associated features often include prenatal and postnatal growth retardation (prenatal and postnatal growth deficiency). Many affected babies have an abnormally small head (microcephaly) that may appear unusually short and wide (brachycephaly), flat back of the head (occiput); or an abnormally long, narrow, and prominent forehead. Additional craniofacial abnormalities may include a triangular face, arched eyebrows that grow together (synophrys); broad and straight nose; an unusually long vertical groove in the middle of the upper lip (philtrum); thin lips; or an abnormally small lower jaw (lower jaw). In most reported cases, the monosomy syndrome of chromosome 3p appears to result from spontaneous (de novo) errors very early in embryonic development. 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. All but one case of Monosomy 3p were new (de novo) chromosomal alterations. The disorder may also be diagnosed or confirmed after birth based on thorough clinical evaluation, diagnosis of characteristic findings (such as developmental delay, mental retardation, psychomotor retardation, craniofacial abnormalities, etc.) and chromosomal analysis. Treatment of monosomy 3p syndrome is directed toward 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; neurologists; Doctors who diagnose and treat heart abnormalities (cardiologists); audiologists; or other health care professionals need. In some cases, doctors may recommend surgical repair of certain abnormalities associated with this disorder [1-5].

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