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A Comprehensive and Clinical Review of Chromosome 14 Mosaic Trisomy Syndrome

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Abstract

Chromosome 14 mosaic trisomy syndrome is a rare chromosomal disorder in which chromosome number 14 appears three times (trisomy) instead of twice in some cells of the body. Symptoms and physical findings associated with chromosome 14, Trisomy Mosaic may depend on the percentage of cells containing an extra chromosome 14. Trisomy mosaic syndrome of chromosome 14 may also be characterized by abnormal prenatal growth retardation (prenatal growth retardation) and failure to grow and gain weight at an expected rate (growth spurt) in infancy.

Keywords: Chromosome 14 Mosaic Trisomy Syndrome; Rare Chromosomal Disorder; Extra Chromosome 14; Developmental Delay; Mental Retardation; Distinct Craniofacial Anomalies

Abbreviations: CVS: Chorionic Villus Sampling; EKG: Electrocardiography.

Overview of Chromosome 14 Mosaic Trisomy Syndrome

Chromosome 14 mosaic trisomy syndrome is a rare chromosomal disorder in which chromosome number 14 appears three times (trisomy) instead of twice in some cells of the body. The term "mosaic" indicates that some cells contain an extra chromosome 14, while others have a normal chromosome pair [1]. This disorder may be characterized by prenatal growth retardation (intrauterine growth retardation), failure to grow and gain weight as expected (growth spurts) in infancy, delay in acquiring skills that require coordination of mental and physical abilities. (psychomotor delays) and mental retardation. Affected babies also have specific abnormalities in the head and face area (skull, face) such as a prominent forehead, deep-set and widely spaced eyes; wide nose bridge; and the ears are small and misshapen. Additional craniofacial abnormalities may include an unusually small lower jaw (micrognathia), large mouth, and plump lips; and incomplete closure or abnormal high arch of the roof of the mouth (palate). Many affected infants also have structural heart abnormalities (eg, tetralogy of Fallot). In some cases, additional physical abnormalities may also be present [1].

Clinical Signs and Symptoms of Mosaic Trisomy Syndrome of Chromosome 14

Symptoms and physical findings associated with chromosome 14, Trisomy Mosaic may depend on the percentage of cells containing an extra chromosome 14. However, this syndrome is usually characterized by developmental delay, mental retardation, distinct

craniofacial anomalies, or other physical abnormalities [1]. Before birth, there may be too much fluid in the amniotic sac (polyhydramnios), which is the thin-walled membrane that surrounds the developing fetus during pregnancy. In addition, some babies with chromosome 14, Trisomy Mosaic may be born before 37 weeks of pregnancy (premature baby). (Pregnancy refers to the period of development from conception to birth. "Full-term" babies are born between 37 and 42 weeks of pregnancy) [1] (Figure 1).

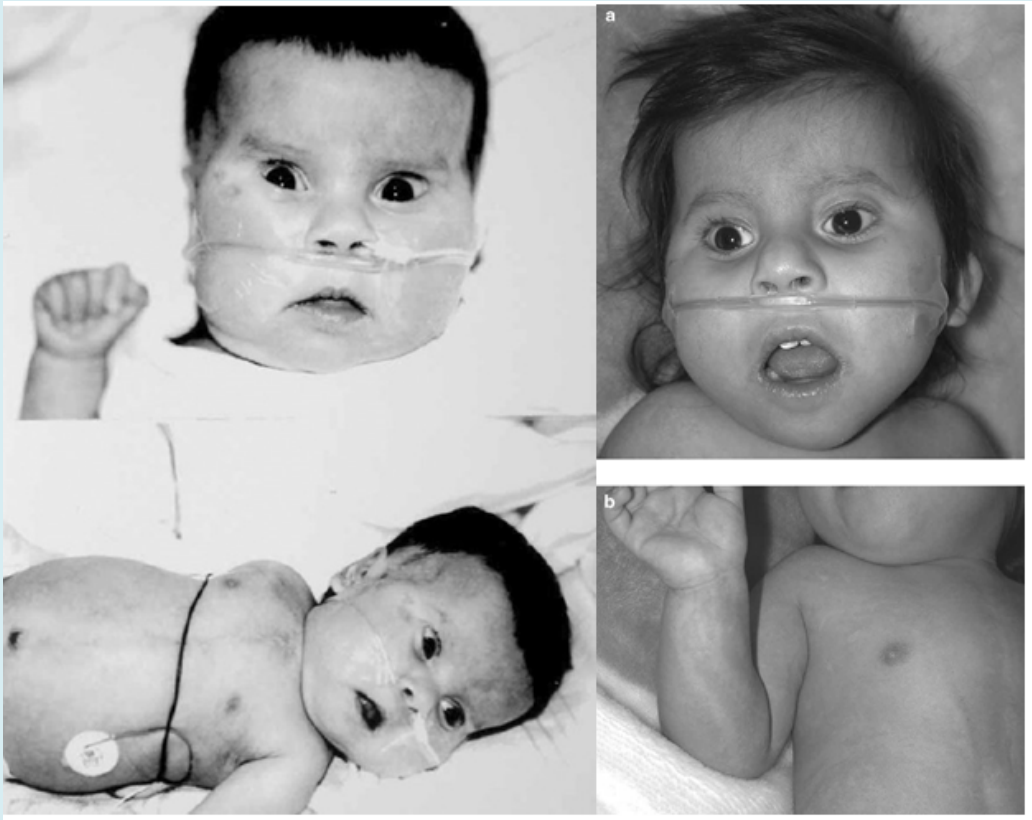

Figure 1: Images of children with mosaic trisomy syndrome of chromosome 14 [1].

Trisomy mosaic syndrome of chromosome 14 may also be characterized by abnormal prenatal growth retardation (prenatal growth retardation) and failure to grow and gain weight at an expected rate (growth spurt) in infancy. In addition, affected individuals typically have psychomotor delays and mental retardation. This syndrome is also associated with characteristic craniofacial abnormalities, resulting in a distinctive appearance. Such features may include a prominent forehead, broad nose; an unusually long vertical groove in the center of the upper lip (philtrum); Prominent upper jaw (upper jaw); and a small and receding mandible (microretrognathia). Various eye abnormalities may also be present, such as deep-set eyes or large distance between the eyes (eye hypertelorism), abnormal reduction in eye size (microphthalmia); Narrow eyelid folds (palm clefts); slight drooping of the upper eyelid (ptosis); or a transparent layer on the eye. Affected infants may also have small, maladjusted and abnormally folded ears, a wide mouth, and plump lips; Incomplete closure (cleft) or abnormally high arch of the roof of the mouth (palate); or have a short and wide neck. Additionally, in some cases, the face may look slightly different from one side to the other (facial asymmetry) [1] (Figure 2).

Many babies with mosaic trisomy syndrome of chromosome 14 also have structural abnormalities of the heart that are present at birth (congenital heart defects), especially tetralogy of Fallot. This birth defect is characterized by a combination of distinct heart (cardiac) abnormalities. These include an abnormal hole in the partition (septum) that separates the two lower chambers (ventricles) of the heart, obstruction of the proper outflow of blood from the lower right ventricle due to a narrowing of the opening between the ventricle and the pulmonary artery (pulmonary stenosis), displacement of the aorta, which causes oxygen-depleted blood to flow from the right ventricle to the

aorta, and thickening (hypertrophy) of the heart muscle (myocardium) of the right ventricle (the pulmonary artery carries oxygen-depleted blood from the right ventricle to the lungs, where the exchange of oxygen and carbon dioxide occurs. The aorta, the body's main artery, originates in the left ventricle and supplies oxygen-rich blood to most people. vessels.) Associated symptoms and findings may include oxygenation insufficient to body tissues (hypoxia), bluish discoloration of the skin and mucous membranes (cyanosis), shortness of breath, feeding problems, lack of growth or other abnormalities. In some cases, mosaic trisomy syndrome of chromosome 14 may be associated with other heart defects. In severe cases, congenital heart disease may lead to potentially life-threatening complications.

In some people with this syndrome, additional physical abnormalities may also be present. For example, according to reports in the medical literature, many babies with mosaic trisomy syndrome of chromosome 14 have areas of abnormally increased skin pigment (hyperpigmentation) that may appear linear, spiral (wheel), patchy, or reticular (reticular). Additional physical findings associated with this syndrome include genital abnormalities in affected men, including undescended testicles or an abnormally small penis, body asymmetry, such as dissimilarity in the length of the arms or legs (limb length asymmetry); or other skeletal abnormalities, such as hip dislocation, overlap of some fingers or toes, or other features [1].

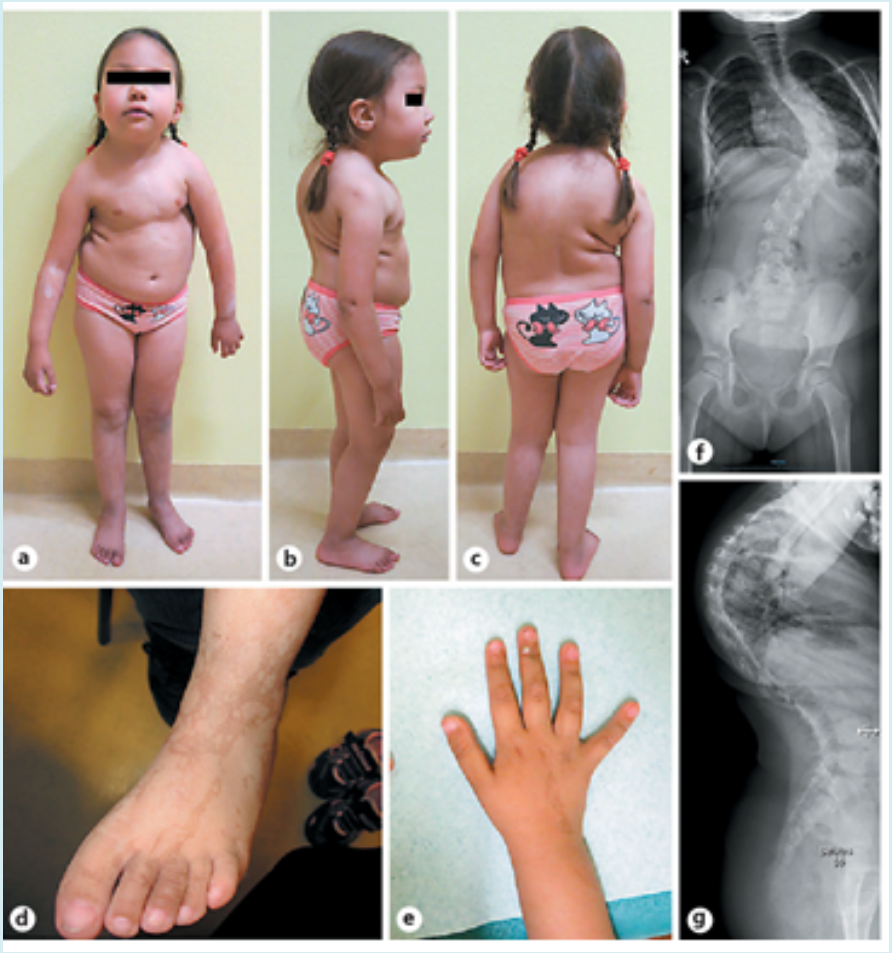

Figure 2: Another view of the types of disorders related to trisomy mosaic syndrome of chromosome 14 with spinal disorder in the form of spinal curvature and brown skin spots on the legs of a child with this syndrome [1].

Etiology of Chromosome 14 Mosaic Trisomy Syndrome

In people with mosaic trisomy 14 syndrome, chromosome number 14 is present three times (trisomy) instead of twice in some of the body's cells (mosaicism). The same chromosomal arrangement (i.e. karyotype) is usually present in all cells of the body. ("Karyotype" refers to the complete set of chromosomes in the nucleus of a cell.) However, those with mosaicism have two or more cell lines

that are karyotype distinct. In people with mosaic trisomy 14 syndrome, trisomy 14 is present in a percentage of cells, while other cells have a normal chromosomal makeup. The extra chromosome is responsible for the symptoms and physical findings that characterize the syndrome. People with a low percentage of damaged cells (low mosaicism) may have fewer and more severe symptoms than people with a high percentage of damaged cells (high mosaicism) [1,2] (Figure 3).

Trisomy mosaic syndrome of chromosome 14 appears to result from chromosomal segregation errors (disjunction) during reproductive cell division in one parent (parental meiosis) or during cell division after fertilization (embryonic mitosis). There are some reports in which this syndrome appears to occur due to uniparental disomy or the formation of an isochromosome (isochromosome 14q). Uniparental disomy is a chromosomal abnormality in which affected individuals inherit both copies of a chromosome pair instead of one copy from each parent. An isochromosome is an abnormal chromosome with identical arms on each side of the centromere [1,2].

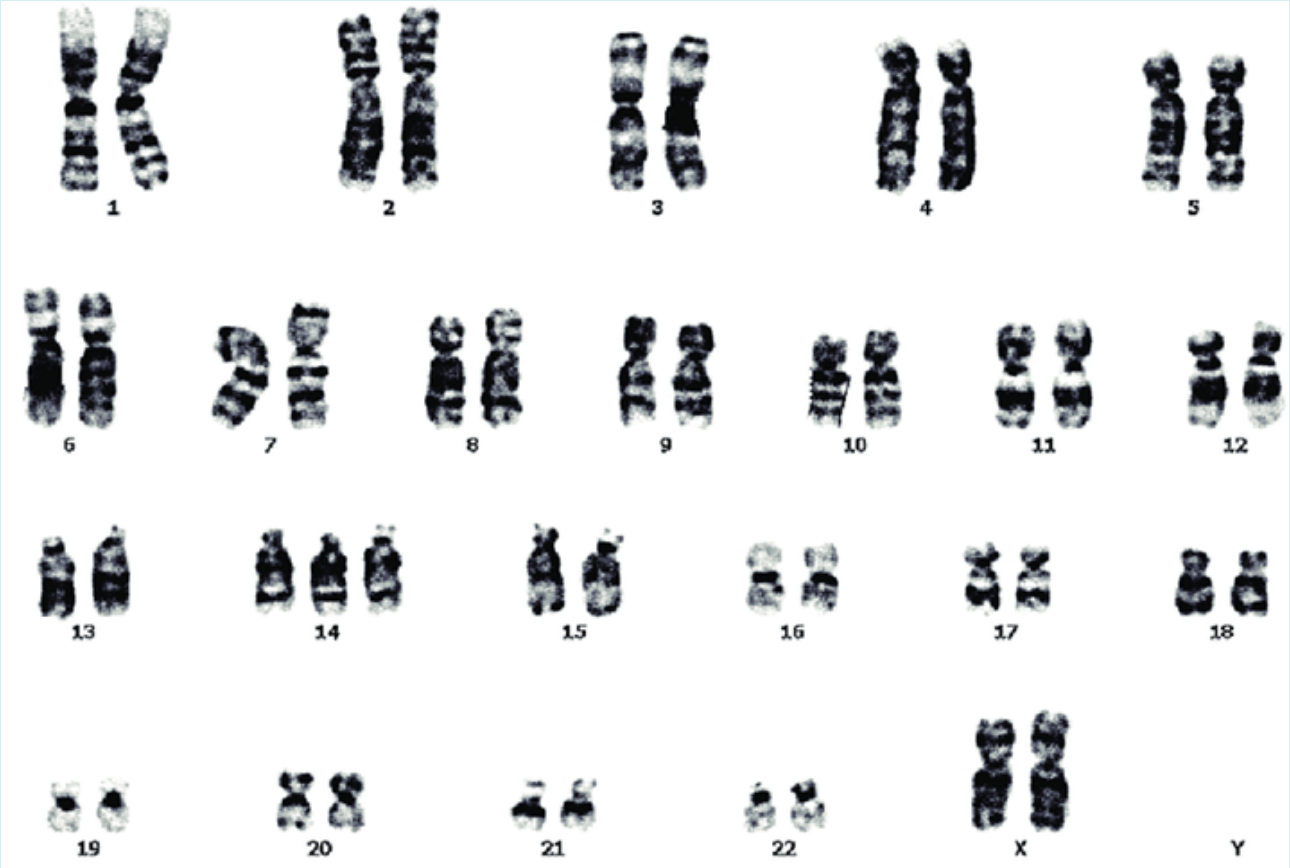

Figure 3: Schematic of the chromosomal karyotype of a human with mosaic trisomy of chromosome 14 [1].

Frequency of Mosaic Trisomy Syndrome of Chromosome 14

In observed cases, trisomy mosaic syndrome of chromosome 14 appears to affect females more than males. So far, more than 20 cases of this syndrome have been reported in the medical literature [1,3].

Disorders Associated with Trisomy Mosaic Syndrome of Chromosome 14

Other chromosomal disorders may present with symptoms and findings similar to those potentially associated with trisomy mosaic syndrome of chromosome 14. Chromosomal testing is necessary to confirm the specific chromosomal abnormality present (Figure 4).

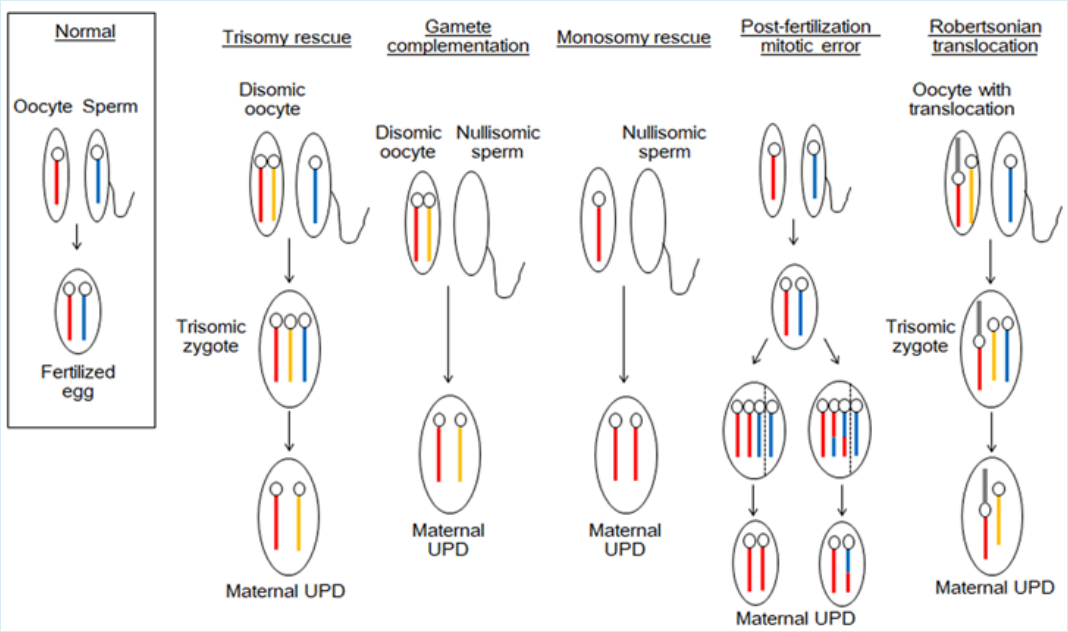

Figure 4: Schematic of the uniparental disomy inheritance pattern that mosaic trisomy syndrome of chromosome 14 follows this pattern [1].

Diagnosis of Chromosome 14 Mosaic Trisomy Syndrome

Diagnosis of mosaic trisomy syndrome of chromosome 14 may be suggested before birth (fetal) with specialized tests such as ultrasound, amniocentesis or chorionic villus sampling (CVS). During fetal ultrasound, reflected sound waves are used to create an image of the developing fetus. An ultrasound may show specific findings that indicate mosaic trisomy syndrome of chromosome 14, such as excessive fluid in the membranous sac around the growing fetus (polyhydramnios), growth retardation, or certain physical abnormalities (such as cleft palate). Palate During 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 a fluid or tissue sample may reveal the presence of mosaic trisomy syndrome of chromosome 14 [1,3].

The diagnosis may also be made or confirmed after birth based on a thorough clinical evaluation, diagnosis of specific physical findings, and chromosomal analysis. In addition, specialized tests may also be done to identify or characterize certain abnormalities that may be associated with the disorder, such as congenital heart defects. A cardiac evaluation may include a clinical examination with a stethoscope to assess heart and lung sounds. X-ray studies; Tests that record the electrical activity of the heart muscle (electrocardiography [EKG]). A technique in which a sound wave is directed towards the heart and allows the evaluation of the movement and structure of the heart (echocardiogram). Or other procedures (eg, cardiac catheterization) [1,4].

Paths for the Treatment of Chromosome 14 Mosaic Trisomy Syndrome

Treatment of mosaic trisomy syndrome of chromosome 14 is directed towards 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 heart abnormalities (cardiologists); or other health care professionals [1,4].

If affected infants and children with tetralogy of Fallot have periods in which the cyanosis suddenly worsens, treatment may include administration of oxygen, morphine, sodium bicarbonate, or other necessary treatments to help improve oxygen concentration. Additionally, in people with tetralogy of Fallot, surgical procedures may be necessary to help relieve symptoms and correct heart abnormalities (eg, systemic pulmonary artery shunt, open-heart corrective surgery). Because people with tetralogy of Fallot may be prone to bacterial infection of the inner lining of the heart (endocarditis), antibiotics should be prescribed before and after dental visits and surgical procedures. Respiratory infections should also be treated intensively and promptly [1,4].

In some infants and children with mosaic trisomy syndrome of chromosome 14, treatment may include surgical repair of certain craniofacial abnormalities, genitalia, or other potential abnormalities associated with the disorder. The surgical procedures performed depend on the severity of the anatomical abnormalities, their associated symptoms, and other factors. Other treatment is symptomatic and supportive. Early intervention may be important in ensuring that children with trisomy mosaic syndrome 14 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 affected people and their families [1,4].

Investigational Treatments

Research into birth defects and their causes continues. The National Institutes of Health (NIH) sponsors the Human Genome Project, which aims to map every gene in the human body and learn why they are malfunctioning. It is hoped that this new knowledge will lead to the prevention and treatment of chromosomal disorders in the future [1,4].

Discussion and Conclusion

Symptoms and physical findings associated with chromosome 14, Trisomy Mosaic may depend on the percentage of cells containing an extra chromosome 14. However, this syndrome is usually characterized by developmental delay, mental retardation, distinct craniofacial anomalies, or other physical abnormalities. In people with mosaic trisomy 14 syndrome, chromosome number 14 is present three times (trisomy) instead of twice in some of the body's cells (mosaicism). The same chromosomal arrangement (i.e. karyotype) is usually present in all cells of the body. ("Karyotype" refers to the complete set of chromosomes in the nucleus of a cell). Diagnosis of mosaic trisomy syndrome of chromosome 14 may be suggested before birth (fetal) with specialized tests such as ultrasound, amniocentesis or chorionic villus sampling (CVS). During fetal ultrasound, reflected sound waves are used to create an image of the developing fetus. If affected infants and children with tetralogy of Fallot have periods in which the cyanosis suddenly worsens, treatment may include administration of oxygen, morphine, sodium bicarbonate, or other necessary treatments to help improve oxygen concentration. Additionally, in people with tetralogy of Fallot, surgical procedures may be necessary to help relieve symptoms and correct heart abnormalities (eg, systemic pulmonary artery shunt, open-heart corrective surgery) [1,4].

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