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CASE REPORT

Trisomy 18 With Multiple Congenital Anomalies: A Rare Case Report

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Abstract

The most common congenital anomalies, autosomal aneuploidies are linked to a variety of metabolic diseases, hormone imbalances, neurotransmitter abnormalities, and intellectual difficulties. Chromosomal segregation mistakes that occur during cell division are what cause trisomies. A number of studies have revealed that numerous disrupted cellular processes are linked to uncontrolled gene expression brought on by the triplication of chromosomes 13 and 18. Furthermore, the occurrence of embryonic abnormalities may be linked to a disrupted oxidative stress condition. These chromosomal aberrations generate many congenital abnormalities such as heart defects, gastrointestinal defects, tracheoesophageal abnormalities, endocrine disorders, vision, and hearing disorders, and limb and nervous system anomalies. However, there are many theories and mechanisms regarding this issue. In this study, we aimed to present a case with multiple congenital anomalies born from a 38-year-old mother.

Keywords: Trisomy 18, Congenital anomalies, Genetic

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Introduction

Trisomy 18 (Edward's) syndrome; is one of the most common chromosomal disorders 3 times more frequently in girls and is another frequent autosomal aneuploidy after Trisomy 21 (T21), affecting 1/6000 to 1/8000 live-birth fetuses. [1,2]. Trisomy 18 syndrome was first described in 1960 by Edwards, and genetic and environmental factors such as maternal and paternal germ cell dissociation and advanced maternal age are blamed [3-5].

T18 occurs most frequently as a result of a complete 18 trisomy due to a maternal meiotic nondisjunction, which is the most common form (94%) [6]. Mosaic trisomy 18 is the second cause corresponding to fewer than 5% of occurrences, and fewer than 2% of cases are caused by an

additional copy of long arm chromosome 18q [7]. These chromosomal aberrations generate many congenital abnormalities such as heart defects, gastrointestinal defects, tracheoesophageal abnormalities, endocrine disorders, vision and hearing disorders, and limb and nervous system anomalies [8-10]. Following the complexity of existing comorbidities, numerical chromosomal aberration, such as T13 and T18 are one of the main causes of miscarriage or stillbirth [11]. However, along with improvements in clinical management, an increased survival rate of patients with these syndromes has been reported [12, 13].

In this study, we aimed to present a case with multiple congenital anomalies born from a 38-year-old mother.

Case Description

A male baby born at the 33rd week of pregnancy weighing 1850 g was intubated in the delivery room due to his poor general condition and inability to maintain his saturations and was admitted to our intensive care unit with the diagnosis of neonatal respiratory distress, prematurity, and a dysmorphic baby. The patient was the 5th child born by cesarean section from the 6th pregnancy of a 38-year-old mother with polyhydramnios.

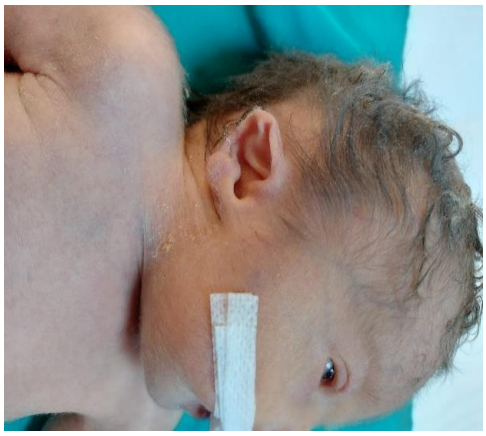

Figure 1: *Micrognathia and Microphthalmia*

The patient had micrognathia (fig.1), microphthalmia (fig.1), short mane neck, low ear appearance (fig.2), and left ankle was in extreme flexion posture (clubfoot, fig.3). Detailed examination of the patient revealed an artery and a vein in the umbilical cord, left choanal stenosis, bilateral clenched hand (fig.4), and unilateral undescended testis.

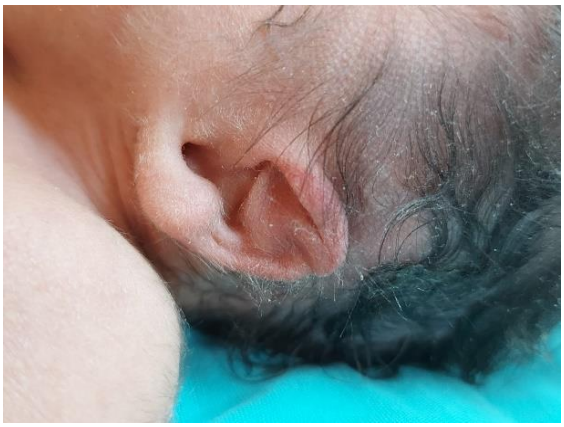

Figure 2: *Short Neck and Low Ear Appearance*

A chest X-ray was taken to the patient. The radiogram was compatible with RDS. The patient was treated with 1 dose of surfactant. And 6 hours later, the surfactant was applied again. Antimicrobial therapy was started in the patient who could not differentiate between RDS and congenital pneumonia.

In the laboratory findings of the patient, urea: 33.6 mg/dL and creatinine: 1,03 mg/dL. Hypocalcemia was also detected. Left kidney could not be observed in abdominal ultrasonography (USG) (hypoplasia). One cyst was observed in the hepatic intrahepatic bile ducts, not clinically significant. Asymmetrical dilatation in the 3rd ventricle was observed in the transfontanelle USG (Hydrocephaly?).

Positive inotropic therapy was initiated in the patient with circulatory disorder and cyanosis. Echocardiography revealed a double outlet right ventricle, large Ventricular septal defect (VSD) (subaortic), patent ductus arteriosus (PDA), and Secundum Atrial Septal Defect (ASD).

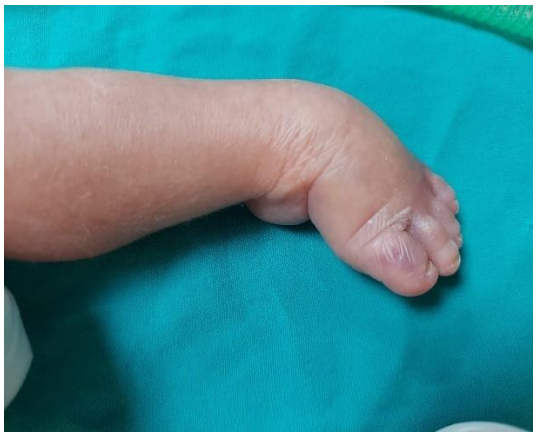

Figure 3: *Clubfoot*

The patient, who was followed up with the diagnosis of dysmorphic baby, underwent an eye examination. The diagnoses of right optic disc coloboma and Morning Plory Syndrome were suspected, but a clear diagnosis was not made.

The patient's TORCH tests were unremarkable. A diagnosis of trisomy 18 was made by performing chromosomal analysis in peripheral blood.

Figure 4: *Clenched hand*

Discussion

According to earlier research, 46–100% of newborns with trisomy 18 have some sort of congenital heart abnormality, with samples ranging from 13–114 infants [14, 15]. 34 newborns with trisomy 18 were included in the cohort of Embleton et al. Of these, an echocardiography or post-mortem testing was revealed [16]. Our patient was also on mechanical ventilator support and despite receiving 2 doses of surfactant and antimicrobial therapy, he could not maintain his saturation. According to the echocardiography, double outlet right ventricle, wide VSD, PDA, and ASD were observed in this patient.

In the etiology of trisomy 18, increased maternal age and faulty chromosome distribution are the most important factors [3,17]. All cases with trisomy 18 syndrome have low birth weight, characteristic facial appearance, and cardiac anomaly. In addition to these findings, there are also those with extremity deformity and renal anomalies [18]. In our case, we mentioned a case with micrognathia, microphthalmia, low ear, mane neck appearance, clubfoot deformity of the left foot, and bilateral clenched hand. The mother of our baby, who was born 1850 grams, was 38 years old.

Their mortality is due to sepsis, nutritional disorders, renal failure, and apnea due to cardiac anomalies [19]. When monitoring this patient with a trisomy 18 diagnosis, we have taken into account cardiac abnormal status, renal failure, and other grave health issues.

Trisomy 18 is a syndrome accompanied by multiple congenital anomalies and its definitive diagnosis is made by chromosome analysis [11]. We performed a genetic analysis as soon as possible.

In conclusion, As we mentioned above, chromosome analysis should be performed in patients with dysmorphic facial appearance, low ear micrognathia, microphthalmia, and mane neck. And the clinician should be alerted to accompanying disorders.

Conflict of interest:

The authors report no conflict of interest.

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No need for case reports

Contributions

Research concept and design: **HGÖ**
Data analysis and interpretation: **HGÖ**
Collection and/or assembly of data: **HGÖ**
Writing the article: **HGÖ**
Critical revision of the article: **HGÖ**
Final approval of the article: **HGÖ**

References

1. Jones K (Ed). Smith'sRecognizablePatterns of Human Malformation. Philadelphia: WB SaundersCompany; 2006: p. 13-17
2. Parker MJ, Budd JL, Draper ES, Young ID. Trisomy 13 andtrisomy 18 in a defined population: Epidemiological, geneticand prenatal observations. PrenatDiagn 2003; 23: 856-860.
3. Edwards JH, Harnden DG, Cameron AH, Crosse VM, Wolff OH. A newtrisomicsyndrome. Lancet 1960; 1: 787-790
4. Naguib KK, Al-Awadi SA, Bastaki L, et al. Clustering of Trisomy 18 in Kuwait: Geneticpredispositionorenvironmental? AnnSaudiMed 1999; 19: 197-200.
5. Nussbaum RL, Mclnnes RR, Willard HF. Principles of clinicalcytogenetics. In: McInnes RR (Ed). ThompsonandThompson Genetics in Medicine. Philadelphia: WB SaundersCompany; 2001: p. 140- 144.
6. Cereda A., Carey J.C. The trisomy 18 syndrome. *Orphanet J. Rare Dis.* 2012;7:81. doi: 10.1186/1750-1172-7-81.
7. Balasundaram P., Avulakunta I.D. *Edward Syndrome.* StatPearls Publishing; Treasure Island, FL, USA: 2021.
8. Pont S.J., Robbins J., Bird T., Gibson J.B., Cleves M.A., Tilford J.M., Aitken M.E. Congenital malformations among liveborn infants with trisomies 18 and 13. *Am. J. Med. Genet. Part A.* 2006;140A:1749–1756. doi: 10.1002/ajmg.a.31382.
9. Peterson J., Kochilas L.K., Catton K.G., Moller J.H., Setty S.P. Long-Term Outcomes of Children With Trisomy 13 and 18 After Congenital Heart Disease Interventions. *Ann. Thorac. Surg.* 2017;103:1941–1949. doi: 10.1016/j.athoracsur.2017.02.068.
10. Roberts W., Żurada A., Zurada-Zielińska A., Gielecki J., Loukas M. Anatomy of trisomy 18. *Clin. Anat.* 2016;29:628–632. doi: 10.1002/ca.22725.
11. Morris J.K., Savva G.M. The risk of fetal loss following a prenatal diagnosis of trisomy 13 or trisomy 18. *Am. J. Med. Genet. Part A.* 2008;146A:827–832. doi: 10.1002/ajmg.a.32220.
12. Anderson C.E., Punnett H.H., Huff V., De Chadarévian J.-P. Characterization of a Wilms tumor in a 9-year-old girl with trisomy 18. *Am. J. Med. Genet. Part A.* 2003;121A:52–55. doi: 10.1002/ajmg.a.20141.
13. Khan F., Jafri I. Characterization of a 16-Year-Old Long-Time Survivor of Edwards Syndrome. *Cureus.* 2021;13:e15205. doi: 10.7759/cureus.15205.
14. Rasmussen SA, Wong LY, Yang Q, May KM, Friedman JM. 2003. Population-based analyses of mortality in trisomy 13 and trisomy 18. Pediatrics 111:777–784.
15. Pont S.J., Robbins J., Bird T., Gibson J.B., Cleves M.A., Tilford J.M., Aitken M.E. Congenital malformations among liveborn infants with trisomies 18 and 13. *Am. J. Med. Genet. Part A.* 2006;140A:1749–1756. doi: 10.1002/ajmg.a.31382.
16. Embleton ND, Wyllie JP, Wright MJ, Burn J, Hunter S. 1996. Natural history of trisomy 18. Arch Dis Child Fetal Neonatal Ed 75:F38–F41.
17. Kelly M, Robinson BW, Moore JW. Trisomy 18 in a 20- year-old woman. Am J MedGenet 2002; 112: 397–399
18. Narlı N, Satar M, Süleymanova D. Trizomi 18 (Edward'ssendromu): Üç olgu sunumu. Klinik Gelişim Dergisi 1998; 11: 486-489
19. Nyberg DA, Kramer D, Resta RG. Prenatal sonographicfindings of trisomy 18: Rewiew of 47 cases. J UltrasoundMed 1993; 12:103.