The occurrence of double aneuploidy; existence of two chromosomal anomalies in the same individual, is an uncommon phenomenon. Double trisomy may occur as autosomal with sex chromosome trisomy or double autosomal trisomy. This coincidence is very rare. Trisomy 18, that is first recognized as a specific entity in 1960 by discovery of the extra 18 chromosome, is characterized by intrauterine growth retardation, prominent occiput, micrognathia, rocker-bottom feet. Klinefelter’s syndrome, that is described in 1942, is the most common single cause of hypogonadism and infertility. Paternal meiosis errors account for about one half of Klinefelter syndrome while remainder are mostly due to maternal meiosis errors. Here, we present a newborn with trisomy 18 with Klinefelter syndrome (48 XXY, 18+), showing the clinical features of trisomy 18.

**Key Words:** Trisomy; klinefelter syndrome

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**ABSTRACT**

Double trisomy may occur as autosomal with sex chromosome trisomy or double autosomal trisomy. This coincidence is very rare. Trisomy 18, that is first recognized as a specific entity in 1960 by discovery of the extra 18 chromosome, is characterized by intrauterine growth retardation, prominent occiput, micrognathia, rocker-bottom feet. Klinefelter’s syndrome, that is described in 1942, is the most common single cause of hypogonadism and infertility. Paternal meiosis errors account for about one half of Klinefelter syndrome while remainder are mostly due to maternal meiosis errors. Here, we present a newborn with trisomy 18 with Klinefelter syndrome (48 XXY, 18+), showing the clinical features of trisomy 18.

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**ÖZET**


**Anahtar Kelimeler:** Trizomi; klinefelter sendromu

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CASE REPORT

The vaginally delivered term male patient was the first child of congenusinous 17 years old mother and 26 years old father. He admitted to the hospital due to cyanosis on the second day of birth. His weight was 2010 gr (<10 percentile) and height was 43 cm (<10 percentile). On physical examination he showed characteristics of trisomy 18 syndrome: he had a prominent occiput, micrognathia, rocker-bottom feet and overlapping of index fingers on second fingers, 4°/6° pan systolic murmur with thrill was detected. His both testes were palpated in the scrotum. He was hypoactive, newborn reflexes were normal. On echocardiographic examination, secundum atrial septal defect (6 mm), subaortic ventricular septal defect (3.5 mm), pulmonary atresia, patent ductus arteriosus (3 mm) were detected. On 3rd day of hospitalization he deteriorated and on the 5th day he died due to Klebsiella pneumonia sepsis in spite of intensive treatment. Chromosome analysis showed 48 XXY, 18+ (G banding), Figure 1. Parents have been informed about prenatal diagnosis, they did not give permission for autopsy.

DISCUSSION

Chromosome analysis of the patient who has the clinical features of trisomy 18 showed double trisomy of chromosomes X and 18. Autosomal abnormality predominates in clinical findings in double trisomy of sex and autosomal trisomy, as characteristic findings of sex chromosome abnormalities might not manifest themselves till puberty. In Klinefelter’s syndrome, maternal and paternal meiotic nondisjunction were contributed equally as an underlying mechanism.

This cooccurrence is very rare. For the first time, in 1963, Haylock et al. described the double trisomy of 48 XXY,18+ with failure to thrive, malformed ears, flexion deformities of extremities, congenital heart disease. In 1967, Zellweger and Abbo, and Cohen and Bumbalo described two different patients of 48 XXY,18+. In a literature survey of Taylor, he reported 3 out of 153 cases of trisomy 18 also had Klinefelter’s syndrome. Jia-Woei et al described a case with the longest survival of >15 months. All these cases had clinical manifestations typical for trisomy 18. Begam et al described another case diagnosed prenatally with amniocentesis and cytogenetic analysis due to prenatal abnormal ultrasonography findings. Only two of the defined cases till now were diagnosed prenatally. The mean maternal age at diagnosis was 31.4 ± 9.8 years. Although an association between maternal age and double trisomy was not observed in the review of the cases, our case was the youngest mother in all of them.

In conclusion, other chromosomal anomalies can accompany to trisomy 18 and chromosome analysis should be done even though phenotype is typical.

REFERENCES