A Newborn with Klinefelter and Trisomy 18 Syndrome: Case Report

Klinefelter ve Trizomi 18 Birlikteliği Olan Bir Yenidoğan Olgusu

Ferda ÖZLÜ,[a]
Hacer YAPICIOĞLU,[a]
Mehmet SATAR,[a]
Nejat NARLI,[a]
Kenan ÖZCAN,[a]
Nilgün TANRIVERDİ,[b]
Sevcan ERDEM[c]

Departments of
[a]Neonatology,
[b]Medical Biology and Genetics,
[c]Pediatric Cardiology,
Çukurova University Faculty of Medicine, Adana

Geliş Tarihi/*Received:* 10.12.2010
Kabul Tarihi/*Accepted:* 03.05.2011

This case report was presented as a poster at 40th Congress of the Turkish Pediatric, 24-25 January 2004, İstanbul.

Yazışma Adresi/*Correspondence:*
Ferda ÖZLÜ
Çukurova University Faculty of Medicine, Department of Neonatology, Adana, TÜRKİYE/TURKEY
ferdaozlu72@yahoo.com

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.

Key Words: Trisomy; klinefelter syndrome

ÖZET Çift trizomi, otozomal ve seks kromozom trizomi olarak ya da çift otozomal trizomi olarak ortaya çıkabilir. Birlikteliği oldukça nadirdir. İlk defa 1960 yılında fazla bir 18 kromozomun gösterilmesi ile tarif edilmiş olan Trizomi 18 intrauterin gelişme geriliği, belirgin oksiput, mikrognati, çıkık topuk ile karakterizedir. 1942'de tarif edilen Klinefelter sendromu hipogonadiz ve infertilitenin en sık tek nedenidir. Paternal mayoz hataları Klinefelter sendromunun yarısını oluştururken, kalan kısmın çoğundan maternal mayoz bölünme hataları sorumludur. Burada trizomi 18 ve klinefelter sendromu birlikteliği olan trizomi 18 klinik bulguları gösteren bir yenidoğan olgusu sunulmuştur.

Anahtar Kelimeler: Trizomi; klinefelter sendromu

Turkiye Klinikleri J Pediatr 2012;21(3):173-4

The occurrence of double aneuploidy; existence of two chromosomal anomalies in the same individual, is an uncommon phenomenon. Double trisomy may occur as autosomal and sex chromosome trisomy or double autosomal trisomy.[1] The occurrence of double trisomy in the same individual is relatively rare. Trisomy 18, that is first recognized as a specific entity in 1960 by discovery of the extra 18 chromosome and characterized by intrauterine growth retardation, prominent occiput, micrognathia, rocker-bottom feet is the second most common trisomy and has an incidence of 3/1000 newborn.[2]

On the other hand Klinefelter's syndrome is the most common single cause of hypogonadism and infertility affecting 1/600 males.[3] Paternal meiosis errors account for about one half of Klinefelter syndrome while remainder are due to maternal meiosis errors The co occurrence of these two anomalies is very rare and to our knowledge only 8 cases of XXY and trisomy 18 have been reported.[1,4-8]

Here we present a newborn with trisomy 18 with Klinefelter syndrome after having permission from parents.

CASE REPORT

The vaginally delivered term male patient was the first child of congansinous 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.

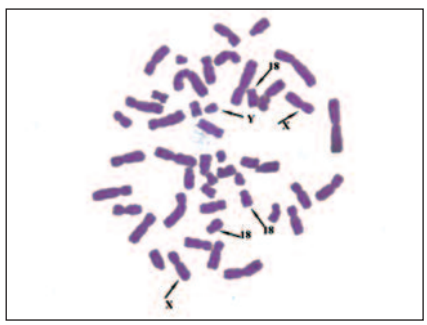

FIGURE 1: The chromosome analysis of the patient showing additional X chromosome and 18th chromosome; 48 XXY, 18+.
(See for colored form http://pediatri.turkiyeklinikleri.com/)

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 mieotic nondisjunction were contributed equally as an underlying mechanism.

This cooccurence 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.[4] In 1967, Zellweger and Abbo, and Cohen and Bumbalo described two different patients of 48 XXY,18+.[5,6] In a literature survey of Taylor,[9] 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.[10] 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.[11] Only two of the defined cases till now were diagnosed prenatally. The mean maternal age at diagnosis was 31.4 ± 9.8 years.[1] 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

1. Nielsen J, Vetner M, Holm V, Askjaer AS, Reske-Nielsen E, Zizka J. Klinefelter's syndrome and trisomy 18 in a newborn boy. Clin Genet 1978;13(3):259-64.
2. Jones KL. Trisomy 18 syndrome. Smith's Recognizable Patterns of Human Malformation. 6thed. Philadelphia: Elsevier Saunders Company; 2006. p.13-7.
3. Jones KL. XXY syndrome, Klinefelter syndrome. Smith's Recognizable Patterns of Human Malformation. 6th ed. Philadelphia: Elsevier Saunders Company; 2006. p.68-9.
4. Haylock JM, Fitzgerald DD, Dennett X. Two cases of the 18-trisomy syndrome, one in combination with an XXY karyotype. Hum Chromos Newsl 1963;1(9):15-6.
5. Zellweger H, Abbo G. Double trisomy and double trisomic mosaicism. Am J Dis Child 1967;113(3):329-37.
6. Cohen MM, Bumbalo TS. Double aneuploidy: trisomy-18 and Klinefelter's syndrome. Am J Dis Child 1967;113(4):483-6.
7. Henchman DC, Grey J, Campbell JB, Nance S. Klinefelter's syndrome with mosaicism trisomy-18. Aust Paediatr J 1970;6(3):142-5.
8. Bach C, Toublanc JE, Gautier M. [Case of double aneuploidy: trisomy 18 and XXY]. Ann Genet 1973;16(1):61-6.
9. Taylor AI. Autosomal trisomy syndromes: a detailed study of 27 cases of Edwards' syndrome and 27 cases of Patau's syndrome. J Med Genet 1968;5(3):227-52.
10. Hou JW. Double aneuploidy: trisomy 18 and XXY in a boy. Chang Gung Med J 2006;29(4):6-12.
11. Begam M, Bekdache GN, Murthy SK, Mirghani HM. Double aneuploidy of trisomy 18 and Klinefelter syndrome: prenatal diagnosis and perinatal outcome. J Perinat Med 2010;38(5):565-6.