Brief Communication

Unusual presentation of Klinefelter syndrome

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ABSTRACT

Introduction: Klinefelter syndrome usually presents in the puberty and adulthood with its characteristic features. We report a boy who had Klinefelter syndrome with hypospadias and hydrocele. **Case Note:** Six and half year old boy had complaints of genitourinary problem in the form of hypospadias, small phallus and hydrocele. Karyotyping showed 47,XXY. **Conclusion:** This case illustrates that Klinefelter syndrome was presented in the infancy with hypospadias and hydrocele which are very uncommon presentation of the disease

Key words: Klinefelter syndrome, Hypospadias, hydrocele, genitourinary, Karyotyping

Introduction

Infants with Klinefelter syndrome may manifest micropenis, hypospadias, cryptorchidism or developmental delay. However, hypospadias and hydrocele are rare presenting features of this commonest chromosomal disorder. In this case report, we describe a boy who had hypospadias and right sided hydrocele along with 47, XXY chromosomal disorder.

Case Report

A 6½-year-old boy attended to our endocrine out-patient department with the complaints of misdirection of urine and small phallus since birth. He was born in non-consanguineous family. His mother was a primigravida and it was a booked case. It was also a normal vaginal institutional delivery at term. The meconium and urine was passed normally after birth. On examination, his weight and height was 16.2 kg (>50th to <75th percentile) and 121 cm (>50th to <75th percentile) respectively. Upper segment and lower segment ratio was 1.01 and arm span and blood pressure were 119 cm and 96/60 mmHg respectively. There was no gynecomastia. Examination of the external genitalia revealed, small phallus (phallic length −2.5 cm), labioscrotal folds were separated. The boy also had penile type of hypospadias [Figure 1]. There was vaginal hydrocele of the right side [Figure 1]. The left testes were firm and the volume was ~2.0 ml. The right testes could not be palpated due to the presence of hydrocele. Routine laboratory investigations including complete hemogram, serum electrolyte, urea, creatinine, plasma glucose were within the normal limits. Ultrasonography of the abdomen revealed no abnormality, but ultrasography of testes showed right sided hydrocele and the left testes size was 1.75 cm × 1.2 cm. LH and FSH were high (11.8 IU/L and 15.0 IU/L) and he had undetectable serum testosterone level. Serum 17 (OH) progesterone was normal. Karyotyping was advised, which revealed 47, XXY [Figure 2].

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Quick Response Code:	Website: www.ijem.in
	DOI: 10.4103/2230-8210.123567

Discussion

This was a case of Klinefelter syndrome with hypospadias and hydrocele. Klinefelter syndrome is the most common chromosomal disorder with prevalence of 1 in 500-640 men.[1] It is estimated that more than 50% of men are not diagnosed and that 90% of those identified are diagnosed

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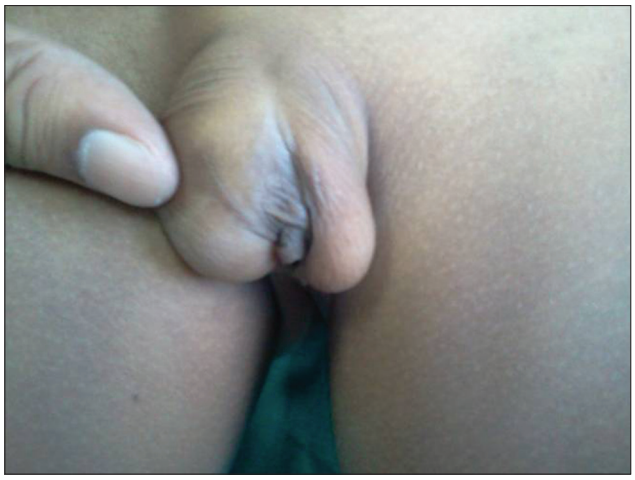

Figure 1: Right sided hydrocele, small phallus

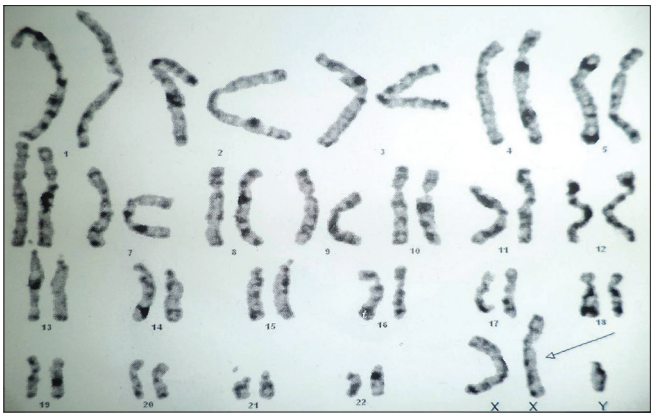

Figure 2: Karyotyping showing an extra X chromosome (47, XXY)

only post-pubertally.[2] Prenatal detection of the Klinefelter syndrome karyotype is the first of three peaks in the diagnosis. Second, in childhood, small testes and phallus[3] and a tall, thin body habitus are clues to the diagnosis of Klinefelter syndrome; although, these findings overlap with findings in the normal boys. Poor school performances and behavioral problems also may occur.[5] The boys may also manifest clinodactyly, hypertelorism, gynecomastia, elbow dysplasia, high arched palate, hypotonia, language delay or learning and reading disabilities requiring therapy.[4] Finally, the diagnosis in adulthood results from presentation with similar complaints, but increasingly made during the evaluation for infertility. At puberty, the testes fail to increase in size and become firm due to a progressive loss of germ cells and seminiferous tubule hyalinization and fibrosis Genitourinary problems like hypospadias as a presenting feature is very rare. Serum FSH and LH are increased uniformly in men with Klinefelter syndrome, thus indicating Leydig cell and seminiferous tubule dysfunction. Mean serum testosterone levels are reduced, but as many as one-third of patients have total testosterone levels within the low-normal range.

Conclusion

In this case, the Klinefelter syndrome has been presented unusually in infancy and the presenting features are hypospadias and hydrocele. Although, the genitourinary problems are rare in Klinefelter syndrome, it should be considered and thoroughly investigated so that they can be managed appropriately.

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Cite this article as: Das C, Sahana PK, Sengupta N, Roy M, Dasgupta R. Unusual presentation of Klinefelter syndrome. Indian J Endocr Metab 2013;17:S683-4.

Source of Support: Nil, **Conflict of Interest:** None declared.