

## Recognition of genital ambiguity as an unusual presentation of Klinefelter syndrome in childhood

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### Abstract

Klinefelter syndrome (KS) is a sex chromosome aneuploidy caused by the presence of one or more supernumerary X chromosomes. Common clinical phenotypes of KS are comprised of tall stature with a feminine body type, gynecomastia, small testes and infertility. Cases of KS with genital abnormalities as the main evaluation in childhood are barely disclosed. We report four children presented to our clinic for assessment of ambiguous genitalia who were ultimately diagnosed with KS. The first patient was a 4 months male baby who presented with phenoscrotal hypospadias, bifid scrotal and small testes. Endocrine studies suggested a normal hypothalamic-pituitary-gonadal axis. The second patient was a 3 weeks baby born with the concern for ambiguous genitalia. He was evaluated at birth for bifid scrotum, small testes and glanular hypospadias. The third and fourth patients were three and seven years old boys with severe hypospadias, bifid scrotal and small testes. Hormonal analysis showed a low level of testosterone with normal level of FSH and LH. The chromosome analysis was 47, XXY for all of the patients confirming the diagnosis of KS. Individuals with KS have a highly varied phenotype comprising a range of physical features, however, genital anomalies are rarely described as characteristics feature of the syndrome in childhood. Clinicians need to be aware of the phenotypic variability of KS and recognize KS as one of the causes of abnormal genitalia at birth. This finding, along with appropriate genetic counselling, suggest that early detection of KS is important in monitoring potential development problems; such as hypogonadism, gynecomastia and gender dysphoria in the future.

**Keywords:** : Klinefelter syndrome; genital; chromosome

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