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KLINEFELTER SYNDROME WITH AMBIGUOUS GENITALIA IN A CHILD

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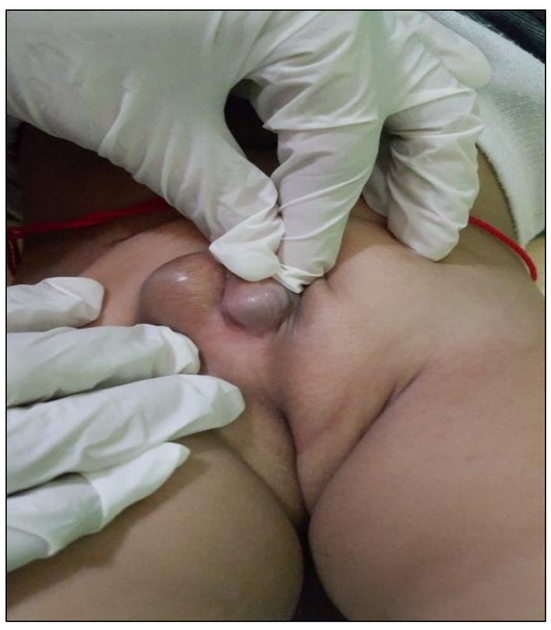
Background:
Klinefelter Syndrome (KS) is the most common sex chromosome disorder in males caused by additional X chromosome. It is characterized by progressive testicular failure. KS patient usually have complete male sexual differentiation without genital ambiguity. The prevalence of KS is 1 in 660 males which only 10% are detected before or during puberty, and about two third so fall men with X-chromosome polyploidies fail to be identified during their lifetime.

Objective:
To report a rare case of Klinefelter Syndrome (KS) with ambiguous genitalia in a 14 months old boy, especially for improving pediatrician awareness to recognize KS as early as possible.

Case:
Fourteen months old boy, BW 9.3 kg (WAZ < -2 SD); BH 76 cm (LAZ 0-(-2) SD); HC 44 cm (< -2 SD) visit pediatric endocrinology OPC with chief complain of small penile buried beneath scrotal and hypospadias. On physical exams, gonads were palpable before scrotal. The phallic length was 1.8 cm and diameter was 1 cm. Karyotyping showed 47, XXY. Genitography revealed contrast could pass through anterior and posterior urethra. Genital USG showed the testicles lies prescrotal. Bone age revealed equal to 14 months old boy.



Conclusion:
We reviewed the rare case of ambiguous genitalia associated with Klinefelter Syndrome (KS) in a child from endocrinology outpatient clinic Dr. Soetomo Hospital.

Keywords:
Klinefelter; 47, XXY; DSD

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The Topic: Sex Differentiation, Gynaecology or sex, 1st Author : Muhammad Faizi