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 penis 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