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A Sri Lankan child with 49,XXXXY syndrome

Vajira H. W. Dissanayake,[1,2] Palinda Bandarage,[1] Christeen R. J. Pedurupillay,[2] and Rohan W. Jayasekara[1]

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Abstract

Pentasomy 49,XXXXY is a rare sex chromosome disorder usually presenting with ambigous genitalia, facial dysmorphism, mental retardation and a combination of cardiac, skeletal and other malformations. The incidence of the condition is estimated to be 1 in 85,000 male births. Previously, this condition was identified as a Klinefelter variant. The condition is suspected in a patient, by a combination of characteristic clinical findings, and the diagnosis is confirmed by chromosome culture and karyotyping. In the case we report here, the main presentation of ambiguous genitalia led to a suspicion of a sex chromosome aneuploidy which was subsequently confirmed by chromosomal analysis.

Keywords: Ambiguous genitalia, sex chromosome aneuploidy, XXXXY syndrome