Double aneuploidy-a rare congenital anomaly of down's syndrome with klinefelter syndrome

Introduction: The coincidence rate of both Down and a Klinefelter syndromes in the same individual is estimated to lie in the range 0.4 to 0.9 per 10,000 male births. The cause of double aneuploidy chromosomal abnormalities may arise from two meiotic non-disjunctional events, but up to date the mechanisms by which these arise, have not been well studied. The prevalence of Down syndrome is one in 770 live births and is increased with advanced maternal age. It is one of the best-recognized and most common chromosome disorders caused by the presence of all or part of a third copy of chromosome 21. Klinefelter syndrome is a chromosomal condition that affects male physical and cognitive development. Its signs and symptoms vary among affected individuals. The prevalence of Klinefelter syndrome is one in 800 male births. Males with Klinefelter syndrome are not usually detected in the newborn period. These individuals are generally normal in appearance before puberty. Due to rarity of this case, we are reporting this unusual case of double aneuploidy showing trisomy 21 and Klinefelter syndrome together.

Case Report: Baby presented at day of life three. The chief complaint was yellowish discoloration of skin up to legs. The neonate was of birth order sixth and last born from non consanguineous parents, aged 45 years for the father and 40 years for the mother at the time of his birth. On physical examination, the patient presents typical features for Down syndrome with up slanted palpebral fissures, sandal gap and mongoloid face, small nose and flat nasal bridge, uncurled hair, short neck and hyperlaxity of joints. The present case of 48, XXY, +21 showed features typical of Down syndrome alone. This is only expected, as features characteristic of Klinefelter syndrome are not apparent until the post-pubertal stage.

Conclusion: Down-Klinefelter syndrome is a rare occurrence. It presents trisomy 21 characteristics early in life and Klinefelter syndrome features after 10 months of age. Pediatric cardiologists are familiar with screening of babies with Down's syndrome for congenital heart disease. However, in children diagnosed with Klinefelter syndrome, a congenital heart disease has only rarely been reported. Reports on congenital heart disease with double aneuploidy of Down's syndrome and Klinefelter syndrome are scarce.

Biography
Neha Raghava has completed her MBBS in year 2013 from Manipal University, India and her MD in Pediatrics in 2017 from Meerut, India. Currently she is working as a Senior Resident in the Department of Pediatrics in Safdarjung Hospital New Delhi.