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A Novel Case of Live Born with 49, XXXY, + 10 Karyotype: Implications of Autosomal Trisomies Other Than 13, 18 and 21

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ABSTRACT Trisomy occurs in at least 4% of pregnancies and is the most common chromosomal abnormality seen in humans. Double trisomies are extremely rare, and most of them involve the sex chromosomes combined with either trisomy 13, 18 or 21. We have identified a novel case with 49, XXXY, +10 karyotype. A ten-month-old boy, with delayed developmental milestones was referred for cytogenetic evaluation. The child was 3rd in birth order delivered vaginally to a consanguineous couple at full term. At birth the only phenotypic abnormalities noticed were low set ears and wide spaced eyes. A suspicion of some abnormality arose when the child failed to attain head holding by 5 months. On detailed evaluation at presentation features observed included short stature, bilateral undescended testis with phallic length of 12mm, the respiratory system had bilateral crepts with wheeze, hepatosplenomegaly was present and D/Q assessment gave a developmental age of 5 months. There was no evidence of mosaicism as all the metaphases analyzed had 49 chromosomes and the buccal epithelium showed cells with a double sex chromatin body. Parental karyotypes were normal and the age of the father and mother were 30 and 25 years respectively, ruling out advanced maternal age as a cause for non-disjunction. No other identifiable factor responsible for inducing double trisomy could be identified, hence the origin of this karyotype is an enigma. The mild phenotypic dysmorphisms seen in this child is surprising and he represents a novel case of a survivor with double trisomy involving sex chromosomes and chromosome 10.