Mosaic double trisomy: Down's syndrome and XYY

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Objective

Trisomy is the most commonly identified chromosomal abnormality, occurring in at least 4% of all clinically recognized pregnancies. Most of the trisomies are associated with a single additional chromosome, although two other types can be observed, those with double trisomy and those with both a normal and trisomic cell line or mosaic trisomies. The co-occurrence of two numerical chromosomal abnormalities in same individual (double aneuploidy) is relatively rare but mosaic double aneuploidy are extremely rare and the clinical presentations are variable depending on the predominating aneuploidy or a combination effect of both.

Methods

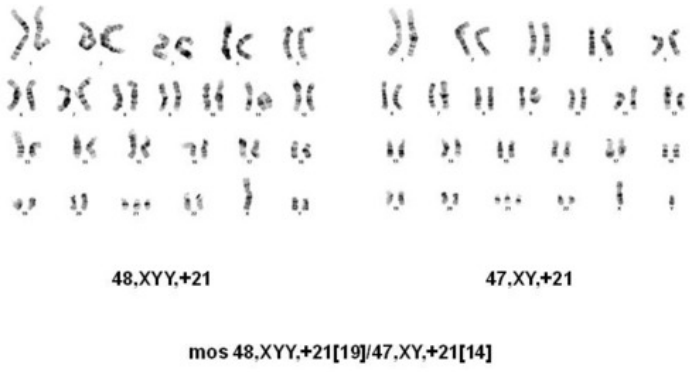
We report an infant with Down syndrome who had mosaicism for XYY and was diagnosed prenatally by chorionic villus sampling performed at the 12 week of pregnancy due to the increased nuchal translucency. There is only two other cases in the literature but they were diagnosed postnatally.

Results

A 25 year-old primigravid woman was referred to our clinic for increased nuchal translucency (NT) at 12th week of gestation. Transvaginal ultrasonography revealed a single live fetus compatible with gestational age and a NT of 5 mm. Chorionic villus sampling was performed and cytogenetic analysis showed a mosaicism for a double aneuploidy, Down syndrome and XYY. The karyotype was 48, XYY, +21[19]/47, XY, +21[14] (Figure-1). The parents refused the termination of pregnancy. At 22 weeks of gestation ultrasonography and fetal echocardiography revealed perimembranous ventricular septal defect and pelvic kidney. A male infant weighing 2980 g was vaginally delivered at 39 weeks of gestation and the phenotypic features of the neonatal were typical for Down syndrome (Figure-2).

Conclusion

Double aneuploidy was first described in a patient with both Down and Klinefelter (48, XXY, +21) syndromes. This is also the most commonly described double aneuploidy. Fewer than 40 cases of Down syndrome with XYY have been reported until date, only two of which have mosaicism for XYY. Aneuploidies are due to non-disjunction at meiosis or post zygotic mitosis. Double aneuploidy results when there is non-disjunction involving two chromosome pairs in one or both parents. Non-disjunction during maternal meiosis I is the most common cause of trisomy 21. The XYY occurs when 24YY spermatozoa are formed due to non-disjunction either at paternal meiosis II or mitosis. Unlike Down syndrome, the XYY is not associated with increased parental age. The presence of an associated sex chromosome abnormality in children with Down syndrome may not be clinically evident until puberty. The only consistent phenotypic feature associated with the XYY syndrome is tall stature, which becomes evident at about 5-6 years of age. These children may have learning difficulties, attention deficits, hyperactivity and increased aggressiveness. Therefore, it is important to recognize the XYY abnormality at the earliest so that these children can be evaluated periodically and given appropriate care and interventions for learning and behavioral needs.



48,XYY,+21

47,XY,+21

mos 48,XYY,+21[19]/47,XY,+21[14]