



A case of double aneuploidy: Trisomy 21 With XYY

Sal H, Comert EH, Guvendag Guven ES, Guven S

Karadeniz Technical University, School of Medicine, Department of Obstetrics and Gynecology, Trabzon, Turkey., TRABZON, Turkey

Objective

Chromosomal abnormalities are seen in nearly 1% of live born infants. Trisomy 21 is the most common aneuploidy seen in infants. Despite the fact that 1 in 1000 boys have the karyotype 47, XYY (XYY), approximately 85% or more of males with XYY are never diagnosed. Some infants with typical phenotypic features of Down syndrome may have rarely an additional aneuploidy (e. g. XXY, XXX, XYY).

Methods

We report a 19-year-old pregnancy with Down syndrome + XYY in the first trimester.

Results

Pregnancies with 48 XYY + 21 can be diagnosed at first trimester. This will enable early intervention to provide the adequate supportive care and management.

Conclusion

Down and Klinefelter syndrome(48, XXY, +21) is the most commonly described double aneuploidy and the first seen is double aneuploidy. Down syndrome and XYY association are very rare, as in our case. Fewer than 50 cases of Down syndrome with XYY have been reported until date. Non-disjunction at meiosis or post zygotic mitosis, cause aneuploidies. Non-disjunction involving two chromosome pairs in one or both parents play a role at pathophysiology of double aneuploidy. Non-disjunction during maternal meiosis I cause trisomy 21. The XYY occurs when 24YY spermatozoa are formed due to non-disjunction either at paternal meiosis II or mitosis. The disease and prognosis of fetus with double aneuploidy should explain family and termination option should be offered. In cases that do not accept the termination, common information with the pediatrician should be provided. Boys with XYY are diagnosed postnatal because of tall stature and behavioral issues. These infants can be evaluated periodically and given appropriate care for learning and behavioral needs. So it is important to recognize the XYY abnormality at the earliest.