49 XXXXY SYNDROME, AN INFANT PRESENTING WITH AMBIGUOUS GENITALIA

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INTRODUCTION

- The 49, XXXXY syndrome is a rare sex chromosome aneuploidy disorder, first reported in 1960 by Fraccaro *et al.*
- Incidence of 1 in 85,000-100,000 male births. The "classical triad" of 49, XXXXY syndrome consists of Mental retardation, radioulnar synostosis and hypogonadism.
- Other phenotypic features include low birth weight, slow growth with retarded bone age, craniofacial anomalies, abnormal genitals, multiple skeletal deformities with joint laxity, cardiac deformities and mental retardation.
- Previously, this syndrome was thought to be a variant of the Klinefelter syndrome (49, XXY). However children with the 49, XXXXY syndrome have distinct facial features, body habitus, multiple skeletal anomalies & cardiac defects, not found in Klinefelter.

CASE REPORT

- Nine month old infant brought to OPD with suspected ambiguous genitalia.
- The infant was born at term to a 21 year old primigravida following an uneventful normal pregnancy of non-consanguineous marriage.
- At birth the neonate weighed 1700 gms, also he was diagnosed to have congenital talipes equinus varus of right foot (CTEV) & ambiguous genitalia. A cast was applied to the foot to treat the talipes equinovarus deformity.
- On examination of the infant, he had developmental delay in milestones.
- Genital examination revealed severe hypospadias with prepenile scrotum, left undescended testes and normally descended right testes. The child had CTEV of the right foot.
- Chest x-ray showed cardiomegaly and 2-D echo revealed PDA. Karyotype analysis by G-banding showed 49 XXXXY chromosomes at a band resolution of 500.
- The parents were counseled regarding the child's health and prognosis.
- The genital reconstruction was planned at a later date.

CONCLUSIONS

- ❖ Infants of 49 XXXXY syndrome commonly present with a various anomalies and delayed developmental milestones which is confirmed on chromosome karyotyping.
- ❖ Additional X chromosome results in not only infertility, but also hypoplastic and undervirilised genitalia.
- ❖ Mental retardation remains a major problem in these patients. Proper counseling is required.

REFERENCES

❖ Fraccaro M, Kaijser K, Lindsten J. A child with 49 chromosomes. *Lancet* 1960; **22:899**-902.
❖ Peet J, Weaver DD, Vance GH. 49, XXXXY: a distinct phenotype. Three new cases and review. *J Med Genet* 1998; **35:420**-4.