49XXXXY Syndrome: A case report of a rare genetic disorder

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49XXXXY is a rare genetic disorder. It is estimated to occur in one in 85,000 live male births. Boys with 49XXXXY have an additional three X chromosomes, giving them a total of 49 chromosomes. The most common features of 49XXXXY syndrome include intellectual disability, speech difficulties, hypotonia, underdeveloped sex organs, recurrent infections (Autoimmune disease) and infertility. The diagnosis can be made by karyotyping in cell samples taken from placental tissue by chorionic villus sampling or from amniocentesis. CGH array is another method for diagnosis of 49XXXXY.

Case Summary: T.O. is an 11 months old male infant born at 36 weeks' gestation by elective C/S for severe IUGR, prematurity and breech. His mother is a 26 years old Irish lady and she had no antenatal issues. His birth weight was 2.28 Kg. He was admitted to NICU after delivery for mild respiratory distress. On examination he was hypotonic and had micropenis. On day of life (DOL) 2 his sodium level was found to be 147 and continued to be raised until DOL 3 with high serum osmolality. He had a cranial USS on DOL 3 which showed several sub-ependymal cysts in the frontal lobe suspicious for a congenital cyst formation. He also had a renal USS which was normal. At 4 weeks of age he had an MRI Brain which showed connatal cysts adjacent to the frontal horns of both lateral ventricles. He was discharged on DOL 16 from the NICU. He had 4 monthly pediatric outpatients follow up as well as physiotherapy follow up. T.O. had recurrent chest infections requiring four admissions. On his admission at 8 months of age he had genetic and metabolic work up and a diagnosis of 49XXXXY was reached. An early intervention referral was made for his persistent central/peripheral hypotonia and his developmental delay.

Conclusion: Rare diagnosis like 49XXXXY can often be missed or diagnosed late due to lack of sufficient features. Features such as underdeveloped genitalia and hypotonia should always raise concerns of underlying cause.

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