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Triploidy syndrome: A rare fatal anomaly in an Arab child; A case report

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Introduction: Triploidy is a rare chromosomal abnormality of an additional set of chromosomes in the cell for a total of 69 chromosomes rather than the normal 46 chromosomes per cell. The extra set of chromosomes originates either from the father or the mother during fertilization. Infants with triploidy usually are miscarried early in the pregnancy or die within the first days of life. We are reporting a case of this rare fatal anomaly from Kuwait.

Case report: A girl baby was born to unrelated Syrian parents in Kuwait at 35 weeks by LSCS. Mother was 25-year primigravida without antenatal care. Pregnancy was uneventful except preterm labor at 35 w. Multiple dysmorphic features noted were- IUGR with relative macrocephaly, parieto-occipital prominence, microphthalmia, small palpebral fissure, flat large low set ears, beaked and deviated nose, micrognathia, thin lips, triangular face, camptodactyly with partial syndactyly 3,4,5 fingers, prominent calcaneum, rocker bottom feet, camp to-partial syndactyly feet (figure 1-5). US head: Prominent choroids plexus. ECHO: ASD and PDA. US abdomen: Agenesis left kidney. Karyotype: 69xxx Baby died after 40 days with cardiorespiratory failure.

Discussion: Triploidy accounts for 1-3 percent of all pregnancies. This disorder does not run in families and is not associated with maternal or paternal age. Apart from multisystem anomalies, they have abnormal facies and syndactily of third and fourth fingers of the hands and the second and third toes of the feet. Antenatal diagnosis possible by karyotyping by amniocentesis or chorionic villus sampling. The diagnosis can be confirmed after birth by chromosome analysis of tissue (skin) obtained from the affected infant. Treatment of triploid syndrome is symptomatic and supportive.

Biography

Karan Soni has graduated from Christian Medical College, Ludhiana, India in 2017 achieving numerous accolades during his studies. He is undergoing Medical Observership at Ahalia Hospital Musaffah, Abu Dhabi, UAE. He has acquired the ECFMG certificate in 2018. He is applying for Residency in USA for 2019. Ahalia Hospital Mussafah, Abu Dhabi, U.A.E.

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