Trisomy 13 with anorectal malformation: an association or an incidental finding?


Keywords: Anorectal malformation; imperforate anus; Patau’s syndrome.

Case report

The patient was a female infant born at term to non-consanguineous parents by emergency caesarean section for non-reassuring foetal monitoring. Antenatal scan at 11 weeks’ gestation showed increased nuchal translucency of 4.2 mm. Repeat ultrasound scan at 19 weeks showed bilateral borderline ventriculomegaly (9–11 mm) and bilateral renal pelvic dilatation. These findings persisted and were also seen in the later scans done at 26, 32 and 35 weeks. The antenatal foetal echocardiogram was suggestive of double outlet right ventricle. The rest of the antenatal ultrasound examination was within normal limits. Genetic counselling was offered to the parents who refused any further genetic screening and decided to continue with the pregnancy.

The patient was born in good condition and did not require any active resuscitation at birth. Her birth weight was 2580 g, and the head circumference was 35.3 cm. On initial examination, she was noted to have overlapping fingers, widely spaced nipples and ulnar deviation of hands. Systemic examination revealed an imperforate anus. Looking at the clinical features, especially the overlapping digits and imperforate anus, our provisional diagnosis was trisomy 18. A genetics consult was requested, and a blood sample was sent for karyotype. She was then transferred to a surgical unit, and colostomy was performed under general anaesthesia. The echocardiogram done by a cardiologist excluded double outlet right ventricle but confirmed the presence of two atrial septal defects.

Eye examination by the ophthalmologists revealed microphthalmia, complete retinal detachment and congenital cataract on the right side. The left eye examination showed inferior iris and retinal coloboma. Cranial ultrasound scan revealed slightly bulky lateral ventricles with no other abnormality. Renal ultrasound showed pelvic calyceal dilatation of 17 mm on the right side.
Fluorescent in situ hybridisation and karyotype analysis confirmed trisomy 13. The infant had a genome of 47 XX+13 with no mosaicism or translocation. This infant is unique as she is possibly the only reported child with Patau syndrome 47 XX+13 (pure non-disjunction and not mosaic/translocation) to have an anorectal malformation. She was discharged home on day 21 of life, self-ventilating in air and requiring bottle feeding.

Discussion

The incidence of trisomy 13 is 1 in 10,000 newborns. It is caused by trisomy for all or a large part of chromosome 13 and is associated with severe intellectual disability multiple physical abnormalities and a limited life span. Extra chromosome 13 may be present in all cells (complete trisomy) or in some of the body cells (mosaicism). If there is a part of an extra chromosome in the cells, it is referred to as partial trisomy. The usual phenotypic features are small for gestational age, central nervous system anomalies, mid facial defects and uro-genital malformations. Other associated features are polydactyly, narrow hyper-convex finger nails and skin defects of the posterior scalp. The features and severity of presentation may vary with the degree of cells involved with trisomy.

Current medical literature describes [2] anorectal malformations as a feature of trisomy 18 and 21 but not of trisomy 13. Out of the cases of trisomy 13 described so far with imperforate anus, all have been either mosaics or translocations. This is the only pure trisomy 13 to have an imperforate anus as an associated finding. This report raises a question for the perinatal physicians whether imperforate anus is an unrecognized association of trisomy 13 or this was just a coincidental finding.

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References


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