A case of pseudoaminopterin syndrome

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Objective

Pseudoaminopterin syndrome or aminopterin syndrome-like (ASSA syndrome--OMIM 600325] is a rare multiple congenital anomaly disorder characterized by ossification defects of the skull, dysmorphic facial features, delayed development, and variable limb defects. The clinical features resemble the embryopathy caused by maternal treatment with the folic acid antagonist, Aminopterin, or its methyl derivative, methotrexate, in early pregnancy and include very unusual facies, skull anomalies, and skeletal defects.

Methods

This is a case report.

Results

37 year old, G2 P0+1 (6 weeks) attended FMU at 30 weeks with a fetus with abnormal facial features. She had a cff DNA test, which suggested low risk for trisomies. No history of consanguinity. Several family members on paternal side had syndactyly on either the foot or the hand. The patient had myomectomy in 2011 and was advised to have a CS. The ultrasound findings in FMU confirmed talipes of the left foot, syndactaly affecting the left hand with a fusion of the third and fourth finger, abnormal facial features with nasal bone hypoplasia, depression of the nasal bridge and prominent frontal bossing and severe IUGR with normal liquor and Dopplers. Rare genetic syndromes and chromosomal disorders in association with this mid-facial anomaly could not be excluded. Invasive testing was offered but the parents declined it. The couple was informed that the prognosis for this baby depends on the underlying cause which will be established after birth. The patient delivered at 37 weeks a female baby weighting 1700 g. The baby was dysmorphic with hypomineralization of the skull bones and very wide anterior and posterior fontanelle and sutures, prominent forehead and flat mid-face with severe depression of the nasal bridge, the face was triangular. There was a complete syndactyly with the fusion of the middle and the ring finger on both hands, with ulnar deviation of fingers, valgus deformity of the feet and overlapping toes. CT of the head suggested the sagittal suture was quite wide/opened with a gap of about 3 cm in-between. The mean bone density of the bones was normal. The left nasal cavity was narrowed and almost obliterated as a result of the deviated nasal septum. There appeared to be a central hard palate defect, most likely representing a cleft palate lined by a membrane.

Conclusion

The syndrome is characterized by hypomineralization of the skull bones associated with various limb defects. The inheritance is likely autosomal recessive. It is a multiple congenital anomaly syndrome that resembles the aminopterin embryopathy with no history of fetal exposure to aminopterin. It is associated with mild to moderate intellectual disability and short stature. The prognosis is variable and depends on the individual case. This is a rare condition, therefore it is difficult to make the diagnosis prenatally. However, the multiple anomalies can raise the suspicion.