A case of amniotic band syndrome

Inan C, Sayin NC, Gurkan H, Erzincan SG, Uzun I, Sutcu H, Atli E, Varol FG
Trakya University, Faculty of Medicine, Departments of Obstetrics&Gynecology and Medical Genetics, Edirne, Turkey, Edirne, Turkey

Objective

Amniotic band syndrome (ABS) can cause a wide spectrum of congenital abnormalities, including orofacial and visceral defects. It is also associated with malformations in truncal, craniofacial regions and the limbs. Sometimes this phenomen may imitate some genetic disorders. In our abstract we present a case report of amniotic band syndrome.

Methods

This is a case report.

Results

A 28-year-old primigravid woman was referred at 21 weeks of gestation in our clinic. Ultrasonographic (USG) examination revealed bilateral broad cleft lip and palate, scoliosis, a right upper limb that appears in a form of retrograde flexion contracture, left anophthalmia and right microphthalmia. Atrial width of the lateral ventricle was measured as 18 mm. Left kidney was observed larger than the left one (Figure 1a,b,c,d). Cordocentesis was performed at 22 weeks gestational weeks. The parents decided to terminate the pregnancy. Postmortem clinical examination revealed a band structure starting from the middle line of the face, extending to the back of the head and terminating on the right upper extremity which makes the extremity turn back to the back of the fetus. Right upper limb was also turned to the back and seemed as a flexion contracture. Left anophtalmia and scoliosis were also confirmed (Figure 2a, b,c). Because of the possibility of a genetic abnormality, we perfomed array-CGH after the analysis of fetal karyotype and FISH, all of which revealed normal results.

Conclusion

The most common malformations in ABS are limb defects, including amputations, constrictions, brachydactyly and pseudosyndactyly; followed by acalvaria, acrania and orofacial clefts. In ABS phenomenon severe ventriculomegaly and asymetric fetal kidneys are not expected to be observed. In our case these abnormalities were also accompanying ABS. Although this case is considered most probably to be compatible with ABS, whole exome sequencing (WES) is suggested for such cases, involving atypical findings after karyotype, FISH and array-CGH analyzes.

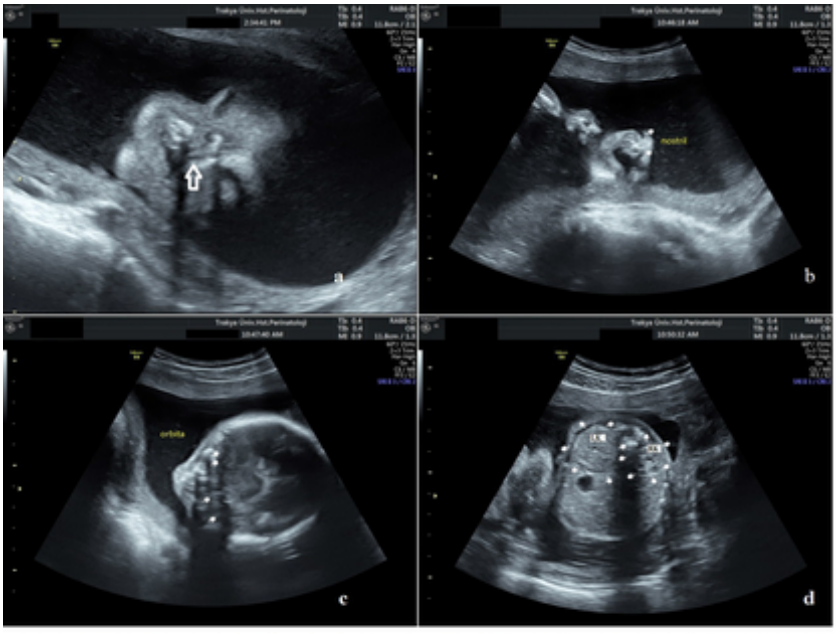

Figure 1. Ultrasonographic imagings of the fetus at 21 weeks of gestation. **a,b.** Bilateral broad cleft lip and palate (Arrow). **c.** Left anophthalmia and right microphthalmia. **d.**The different sizes between fetal kidneys. LK: Left kidney, RK: Right kidney

Figure 2. Postmortem views of the fetus. **a.** Bilateral broad cleft lip and palate (white arrow); anophptalmia (white asterix). **b,c.** A band structure starting middle line of the face and terminating on the right upper extremity (black arrows); right upper extremity which turned back (black asterix).