A case of amniotic band syndrome

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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 phenomenon 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 anophthalmia and scoliosis were also confirmed (Figure 2a, b,c). Because of the possibility of a genetic abnormality, we performed 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 asymmetric 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 2. Postmortem views of the fetus. a. Bilateral broad cleft lip and palate (white arrow); anophthalmia (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).