Case report

AMNIOTIC BAND SYNDROME

Aleksandra Matic and Jovan Komazec

Amniotic band syndrome (ABS) is a set of congenital malformations attributed to amniotic bands that entangle fetal parts during intrauterine life, which results in a broad spectrum of anatomic disturbances - ranging from minor constriction rings and lymphedema of the digits to complex, bizarre multiple congenital anomalies incompatible with life. ABS is not very often, but should be considered in every newborn with congenital anomalies, especially defects of extremities and/or body walls. ABS can be diagnosed prenatally by ultrasound; otherwise, the defects are seen after birth. Child's karyotyping is of great importance, in order to avoid misdiagnosis and incorrect information of recurrence risk. A team of specialists should be included in the treatment and follow-up of children with ABS, according to individual needs of every single patient.

The aim of this paper is to point out diagnostic and therapeutic approaches in newborns with ABS trough the report of two cases. *Acta Medica Medianae 2009;48(2):44-48.*

***Key words**: amniotic band syndrome, intrauterine amputation, ring constrictions*

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Introduction

Amniotic band syndrome (ABS) is a set of congenital malformations attributed to amniotic bands that entangle fetal parts during intrauterine life, which results in a broad spectrum of anatomic disturbances - ranging from minor constriction rings and lymphedema of the digits to complex, bizarre multiple congenital anomalies incompatible with life (1,2).

Incidence of ABC is estimated in wide range of 1 : 1200 – 1 : 15000 live births (1,3,4-6) in 1 : 70 in stillborns (7) and among abortuses as high as 178 : 10000 (2). Among total of 3% major malformations in general population (8), ABS is responsible for 1-2% (9).

The aim of this paper was to point out diagnostic and therapeutic approach in newborns with ABS through the report of two cases.

Case report No 1

Patient: female preterm newborn, second child in the fourth pregnancy (1 healthy child, 2 artificial abortions) in a healthy 28-year old mother. There were no trauma or drug taking during pregnancy, no prenatal chromosomopathy diagnostics, consangvinity or malformations in the family. Pregnancy was regularly controlled, had favourable course until premature labour, ultrasound examination did not show any fetal malformations. Delivery occurred in 35 1/7 gestational week. Birth weight was 2880 g (<90 percentils), birth length 45 cm (>10 percentils), head circumference 32 cm (<50 percentils), Apgar score 9/10. The first examination showed the following malformations: missing second finger on the left hand, missing third and forth fingers on the right hand and replaced with short edematous rudiment. On the right leg, above the ankle there were circulatory ring, the right foot slightly smaller than the left. Besides, there were few minor malformations present: hypertelorism, flat nasal bridge, thin lips, malposition of the ears. Karyotyp was normal female (46, XX). All performed examinations (ultrasound of central nervous system and abdominal organs, echocardiografy, oftalmoscopic examination) showed normal anatomy of examined organs. Plastic surgeon was consulted, and edematous rudimentary part replacing third and fourth finger on the right hand were banded.

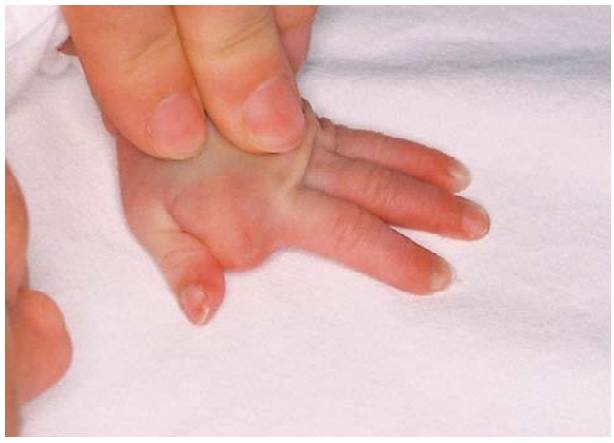

Figure 1a. Left hand with a digit amputated

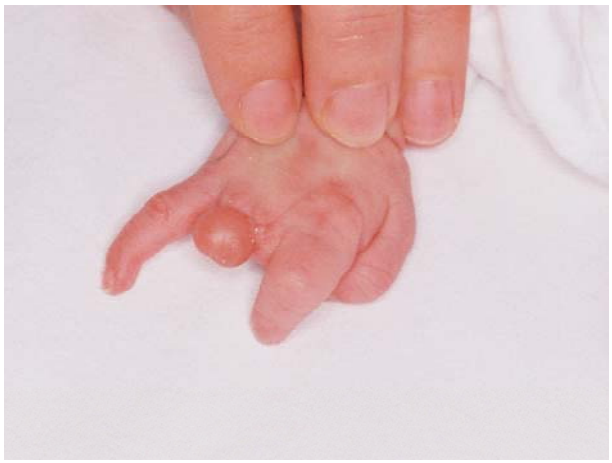

Figure 1b. Right hand with two digits amputated

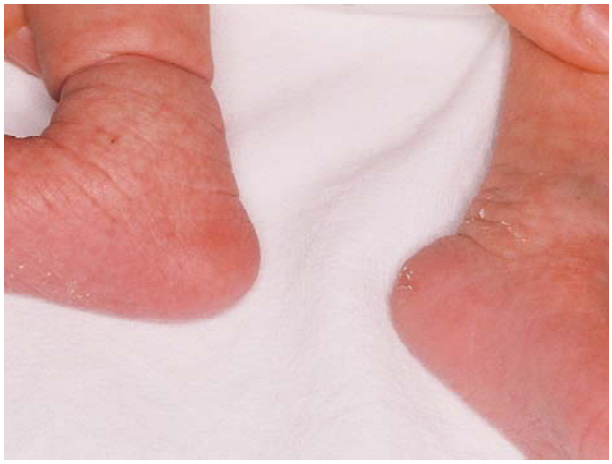

Figure 2. Constriction ring above the ancle of the right leg

Case report No 2

Patient: male preterm newborn, born after 34 gestational weeks as a first child from a first, regularly controlled pregnancy to a 27-year old healthy mother. Pregnancy had favorable course until premature labor. Trauma during pregnancy, prenatal fetal karotypisation, cerclage, drug taking, consanguinity or malformations in family were denied. Routine ultrasound examination did not show any fetal malformations. Birth weight was 2250 grams (50 percentils), birth length 46 cm (50 percentils), head circumference 29,5cm (10 percentils), Apgar score 8/9. Immediately after birth, the following malformations were observed: cleft lip and palate, left-sided uvula, absent lower wall of the left nostril. On the right hand, third finger was shortened with the presence of two constriction rings; bellow lower ring the presence of edema. On the upper part of the right foot there were amniotic band mark, syndactylia of II and III, and IV and V right tows; right tows were shortened with hypoplastic nails. Besides, there were some more minor malformations present: asymmetric and malformed ears, funnel-like bended thoracic cage, bilateral klyndactily of the fifth finger, sandal creases on the feet. Karyotyp was normal male (46, XY). All performed examinations (ultrasound of central nervous system and abdominal organs, echocardiografy, oftalmoscopic examination) showed normal anatomy of examined organs. Plastic surgeon and orthodontist were consulted.

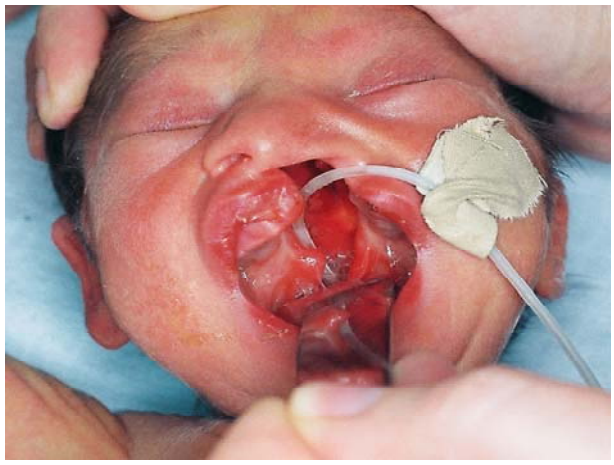

Figure 3. Cleft lip and palate

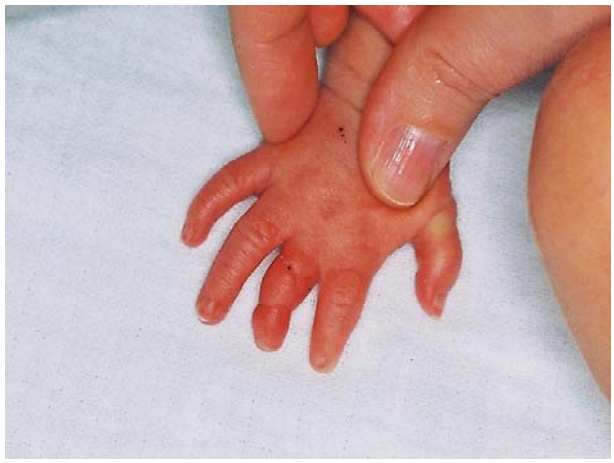

Figure 4a. Deformities of the right hand middle finger

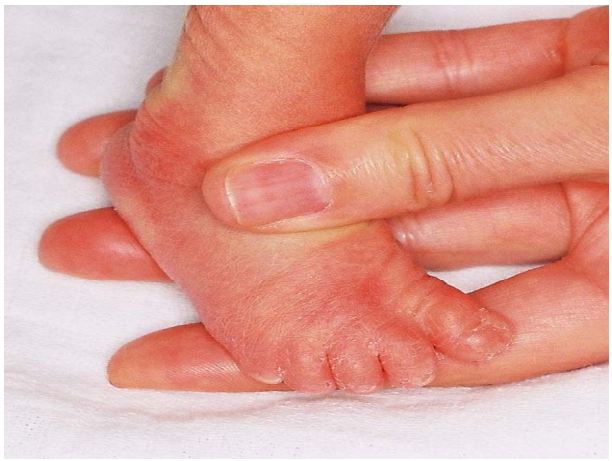

Figure 4b. Right foot deformities

Discussion

ABS ethiopathogenesis is still unknown, but there are two main theories (10-13). Widely accepted "extrinsic model", proposed by Torpin and Faulkner in 1966 explains defects genesis by rupture of the amnion in early pregnancy, with forming of amniotic bands and amniotic liquid

loss, followed by extrusion of all or parts of the fetus into the chorionic cavity. Bands entrap the parts of the growing fetus, and fetus' limbs and other body parts become entangled and subjected to compression, which compromises fetal circulation and also his growth and development with consecutive disturbances of functions and anatomy. The intrinsic model was proposed by Streeter in 1930 and suggests that the anomalies and the fibrous bands have a common origin, caused by a perturbation of developing germinal disc of the early embryo.

Beside ABS etiopathogenesis, risk factors which start such sequence of events are also poorly known. Most cases of ABS are not of genetic origin, and there is no recurrence in siblings or children of affected adults. However, there are some reports of amniotic band syndrome among families with collagen disorders, more specifically Ehler-Danlos syndrome (2,6), in other diseases that involve connective tissue, e.g. in osteogenesis imperfecta , and in one case of epidermolysis bullosa congenital (14,15). Some other possible etiologic factors besides inheritance were explored in several studies. Some studies found connection between ABS and mother's age (especially primiparas under the age of 25 (12,15), prematurity (16), abdominal trauma (2,15), unsuccessful abortion (17), intrauterine contraception (2), cerclage (18), chorionic villus sampling (17-20), amniocentesis (2,17-20), malformations of the uterus (15), some drugs taking, like ergotamine (21), acetaminophen (12), misoprostol (21). However, there is no firm evidence of definite causality for any of these factors, and therefore a great number of authors considers ABS as defects with sporadic occurrence, with no gender prevalence, and no strong risk factors (1). Several studies confirm this opinion, with evidence that most of the cases of ABS have no risk factors in prenatal anamnesis. Both our patients have only prematurity in their family and prenatal anamnesis.

Amniotic band syndrome has very polymorphic clinical findings, because type of deformities depends on the time of amniotic rupture during pregnancy and part of the fetal body which is entangled in amniotic bands. Early amniotic rupture, during first 45 days, leads to the most severe cranio-facial and visceral malformations (5). Every part of the fetal body can be damaged, but most often extremities, especially upper extremities. Most often there are minor defects, such as constriction rings or digit amputations; but, even minor defects are multiple in 77% of cases (4). Abnormalities of the extremities can be expressed in several ways: constriction rings of the soft tissue accompanied by distal edema, shortenage of the limb or intrauterine limb amputation, amputation of the digits (most often II, III and IV finger) and toes, syndactyly, hypoplasia of the digits, foot deformities, pseudoarthrosis, periferal nerve palsy (1,21). If bands compress the fetal head or face, different cranio-facial disturbances appear – asymmetric face clefts, orbital defects (anophtalmos, microphtalmos, enophtalmos), corneal abnormalities, central nervous system malformations (anencephaly, encephalocoele, asymmetric meningocoele), calvaria defect. Amniotic bands can also cause abdominal wall defect and abdominal organs extrophy (1), chest wall defect with heart extrophy (6), umbilical cord strangulation with often lethal outcome (1). Amniotic rupture and consecutive olygoamnion can, by mechanical pressure on the fetus, cause deformities such as metatarsovarus, scoliosis (5) or hip dislocation (1). Because of such a wide spectrum of possible anomalies and many combinations of their simultaneous appearance, there are no two identical cases of ABS (1). Beside all previously mentioned malformations caused by amniotic bands itself, a subset of cases manifest additional findings that are not consistent with that mechanism, such as congenital heart defects, renal anomalies, hemangiomas, imperforate anus, polydactyly, septo-optic dysplasia, typical cleft lip and palate (10).

ABS can be diagnosed prenatally by ultrasound, which can sometimes show amniotic bands, but more often malformations consistent with ABS, as well as olygoamnion and reduction of foetal movements (22). ABS can be diagnosed as early as 12 gestational weeks (23); in the second trimester of gestation most of ABS defects could be seen during routine ultrasound examinations (22). The most important ultrasound diagnostic criteria are visible amniotic bands, constriction rings on extremities and irregular amputations of fingers and/or toes with terminal syndactily. Mild defects, however, are less likely to be diagnosed prenatally, in which case defects are seen after birth (23). Latest ultrasound techniques – three-dimensional and four-dimensional ultrasound contribute to more sensitive prenatal diagnostics of ABS, and in complicated cases foetal magnetic resonance can be helpful (3).

Placenta and amnion examination after the delivery should be obligatory part of the newborns health evaluation because it can show presence of amniotic bands, among other things (3,17,19).

Physical examination is the main way of postnatal diagnostic of ABS, with usage of addition searches in order to establish potential malformations of different organs and body parts: ultrasound, echocardiography, X-ray.

ABS must be considered in differential diagnosis of all complex or asymmetric malformations, especially those on extremities, face and body walls. ABS should be differentiated from the whole spectra of symmetric fusion defects of middle body line (19). In differential diagnosis some rare findings, such as amniotic folds, complex extremity-body wall, extraamniotic

pregnancy should be taken into consideration (2). Exclusion of chromosomopathies in ABS cases is of great importance, because of informing the parents of future recurrence risk, which is very low for ABS (19).

Therapy of ABS is mostly surgical, with an individual approach to every single case. Interdisciplinary consulting and work is very often needed (plastic surgeon, orthopedic surgeon, orthodontist, ophthalmologist, neurosurgeon…) (1). Lately, there have been some attempts of prenatal ABS treatment - foetoscopic laser cutting of amniotic bands, before their compression on the fetus makes malformations (25). In cases when foetal anomalies incompatible with life are prenataly seen, pregnancy termination is advised (2).

Conclusion

ABS is not very often, but should be considered in every newborn with congenital anomalies, especially defect of extremities and/or body walls.

The basis for postnatal diagnosis is physical examination of the newborn, with additional examinations after potential internal organs malformations. Child's chromosomal karyotype analyses in order to exclude wrong diagnosis and consecutive incorrect information about recurrence risk for the parents is of outmost importance.

Because of ABS complexity, the treatment and follow-up of these children requires a team of specialist, according to special needs of every single patient.

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SINDROM AMNIONSKIH BRIDA

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Sindrom amnionskih brida (ABS) je grupa kongenitalnih anomalija nastalih obuhvatanjem delova fetusa od strane fibroznih amnionskih traka tokom intrauterinog života, što dovodi do širokog spektra poremećaja u njegovoj anatomiji - od minornih konstrikcijskih prstenova i limfedema prstiju, do multiplih, kompleksnih, bizarnih malformacija inkompatibilnih sa životom. ABS nije čest, ali kod svakog deteta sa kongenitalnim anomalijama, pre svega defektima ekstremiteta i telesnih zidova, na njega treba misliti. Dijagnozu je nekad moguće postaviti već prenatalno, putem ultrasonografskog pregleda ili postnatalnim pregledima deteta. Veoma je važan kariotip deteta da bi se izbegla pogrešna dijagnoza i netačna informacija o rekurentnom riziku za dalje potomstvo. S obzirom na kompleksnost ABS, u tretman i praćenje dece sa ovim anomalijama trebalo bi da bude uključen tim lekara, prema specifičnim problemima svakog pojedinog bolesnika.

Cilj ovog rada bio je da kroz prikaz dva slučaja sindroma amnionskih brida ukažemo na dijagnostički i terapijski pristup kod novorođenčadi sa ovim problemom. *Acta Medica Medianae 2009;48(2):44-48.*

Ključne reči: *sindrom amnionskih brida, intrauterina amputacija, konstrikcijski prsten*