



ETIOLOGY, CLASSIFICATION OF IMPORTANT SYNDROMES FOUND IN INFANCY

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Annotation:

Failure to thrive is the term used to describe the condition in which a young child fails to gain weight satisfactorily. Common reasons for such poor weight gain are parental neglect or lack of food. On the other hand, a large number of important gastrointestinal disorders may be responsible, including those associated with vomiting, such as food intolerance or obstruction of the upper bowel by pyloric stenosis; disorders of digestion and absorption, including celiac disease and cystic fibrosis; and bowel infections. 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.

Keywords: amniotic band syndrome, intrauterine amputation, congenital anomalies .

In developed countries, SIDS (also called crib death or cot death) accounts for 20 percent of deaths between the ages of one month and one year. SIDS is a categorization rather than an explanation, for the label is given when no reason for death can be found from the infant's medical history or even after autopsy.

Most crib deaths occur in the first five months of life and strike at home during the night. They are more common in the winter and in poor social circumstances. A preceding minor respiratory infection is common. This has prompted some investigators to suggest that the underlying defect is the presence of a virus in the bloodstream, leading to instability of cardiac and respiratory mechanisms. Many other hypotheses have been proposed to explain such deaths, however, and it is likely that several different causes may be involved.

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.

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, consanguinity or malformations in the family. Pregnancy was regularly controlled, had favourable course until premature labour, ultrasound examination did not show any fetal malformations.



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 karyotypisation, 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, funnellike 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.

ABS etiopathogenesis 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, peripheral nerve palsy (1,21). If bands compress the fetal head or face, different cranio-facial disturbances appear – asymmetric face clefts, orbital defects (anophthalmos, microphthalmos, enophthalmos), corneal abnormalities, central nervous system malformations (anencephaly, encephalocoele, asymmetric meningocele), 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 fourdimensional 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).

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