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CASE REPORT

Discordant monoamniotic twins with Pena–Shokeir phenotype

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Key Clinical Message

Pena–Shokeir phenotype is a rare disorder. However, its etiology is incompletely understood. It may be familial or may be due to anoxic–ischemic etiology. Although rare, it can affect one twin in a monoamniotic pregnancy, most likely due to early cord entanglement.

Keywords

Fetal akinesia deformation sequence, fetal cord entanglement, monoamniotic twins, Pena–Shokeir syndrome.

Introduction

Pena–Shokeir phenotype/fetal akinesia deformation sequence is characterized by the lack of fetal movement, fetal intrauterine growth restriction, craniofacial anomalies, arthrogryposis, polyhydramnios, and pulmonary hypoplasia. We report a case of Pena–Shokeir phenotype occurring in one fetus of a monoamniotic pair. We postulate that this resulted from hypoxia/anoxia due to early cord entanglement.

Case Report

A 43-year-old patient, para 2, gravida 3, with two prior uneventful pregnancies, presented to our unit at 27 weeks of gestation. We confirmed a monoamniotic twin pregnancy. Twin A had decreased fetal movements, a thickened nuchal fold, hypertelorism, micrognathia, bilateral clubbed feet and clenched hands, and bilateral moderate hydrothoraces (Fig. 1). Twin B had a ventricular septal defect but was otherwise structurally normal. An amniocentesis confirmed normal 46 XX karyotype. The patient was followed up weekly, but twin A developed ascites and the hydrothoraces worsened. Delivery was by Cesarean section at 32 weeks of gestation.

Twin A weighed 2450 g and had an Apgar score of 1/10 and 1/10 at 1 and 5 min, respectively. She was markedly hydropic, which accounted for the increased weight, with no palmar or plantar creases. She had a high-arched palate, hypertelorism, abnormal low-set ears, micrognathia, ankylosis of large joints with hyperextension of upper and lower

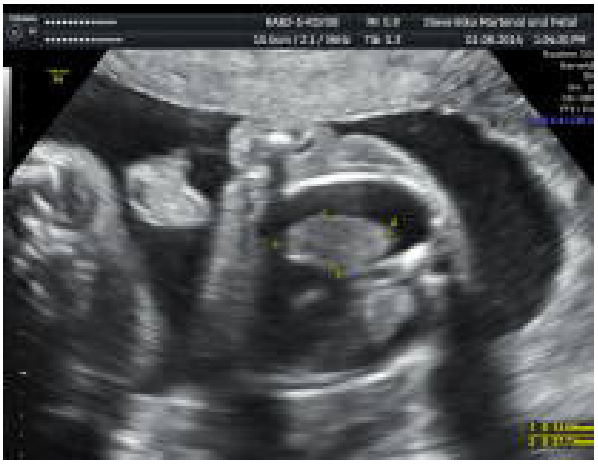

Figure 1. Ultrasound image of twin A at 30 weeks of gestation showing pleural effusions.

limbs, bilateral talipes equinovarus, and campylodactyly (Fig. 2). Resuscitation of twin A was difficult, and the baby demised in the first hour of life. Twin B was appropriately grown and weighed 1600 g and had an Apgar score of 7/10 and 9/10 at 1 and 5 min, respectively. She was admitted to neonatal ICU as she was premature but demised 2 weeks later due to neonatal sepsis.

The postmortem of twin A revealed that there were thin cerebral and cerebellar cortices, cerebral cortical dysplasia, and diffuse skeletal muscle atrophy with fibrous displacement (neurogenic atrophy). The lungs were severely hypoplastic with a lung/body weight ratio of 0.0016. There was absence of anterior motor horn cells in the spinal cord. The postmortem concluded that the baby had fetal akinesia deformation sequence (Pena–Shokeir Syndrome) most likely due to anoxic–ischemic etiology following cord entanglement, even though the cord entanglement was not identified at the time of delivery.

The pathogenesis of Pena–Shokeir phenotype is attributable to familial muscle dystrophy or anoxic–ischemic etiology. However, the details of how this develops are still unclear. As there was no family history of Pena–Shokeir phenotype in this patient and only one fetus was affected, the most likely etiology was thought to be anoxic–ischemic. The pathologist concluded that the absence of anterior motor horn cells in the spinal cord supported this. Cord entanglement was postulated to be the initiating event.

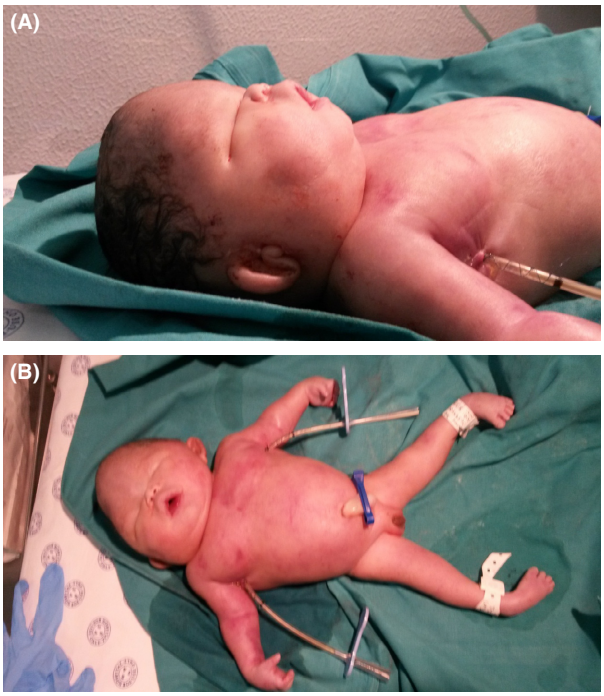

Figure 2. (A) Postdelivery image of twin A showing the facial profile (hypertelorism, abnormal low-set ears, high-arched palate). (B) Postdelivery image of twin A showing hyperextended upper and lower limbs, bilateral clubbed feet, and campylodactyly. Intercostal drains were inserted in an attempt to resuscitate the baby in accordance with the mother's wishes.

Discussion

A prospective observational study showed that cord entanglement is present in all monoamniotic twins when systematically evaluated by color Doppler [1]. In most cases, cord entanglement has no sequelae. In other cases, it can cause fetal death of one or both twins or discordant weights between twins. In our case, we attribute the Pena–Shokeir phenotype to anoxic–ischemic damage due to cord entanglement. We find a hereditary mechanism unlikely as twin B and the patient's previous two children were unaffected.

Pena–Shokeir syndrome or fetal akinesia deformation sequence refers to early lethal neurogenic arthrogryposis and pulmonary hypoplasia. The classic features include intrauterine growth restriction, craniofacial anomalies, limb contractures, pulmonary hypoplasia, short umbilical cord, short gut, and pregnancy complications such as polyhydramnios, abnormal uterine positioning, and decreased fetal movements. Craniofacial abnormalities related to decreased fetal movements include ocular hypertelorism, high bridge of the nose, underdeveloped tip of the nose, posteriorly angulated ears that appear low-set, short appearing neck with mild webbing, microretrognathia, high-arched palate, and cleft palate [2, 3]. Approximately 30% of affected children are stillborn. The remainder mostly die of pulmonary complications, as in our case.

Phenotypic abnormalities are mostly caused by decreased or absent movements in utero. Neuromuscular abnormality of the diaphragm and intercostal muscles causes pulmonary hypoplasia. Multiple ankylosis of elbows, knees, hips and ankles, rocker-bottom feet, talipes equinovarus, and campylodactyly are present. The absence of the flexor creases on fingers and palms is seen. There is usually spinal cord involvement with loss or reduction in anterior motor horn cells. Skeletal muscle shows diffuse and group atrophy consistent with neurogenic atrophy. Often thin cerebral and cerebellar cortices with polymicrogyria are noted [3]. Twin A in our study demonstrated most of these phenotypic findings, and a diagnosis of Pena–Shokeir syndrome was made. The pathogenesis of Pena–Shokeir phenotype may be attributed to familial muscle dystrophy or anoxic–ischemic etiology. However, the details of how this develops are uncertain.

Six cases of Pena–Shokeir phenotype have been reported in monochorionic twins in the literature [4–7]. Five cases occurred in monochorionic diamniotic twin pregnancies [4–6]. In two of these cases, both twins were

affected, and postmortem concluded that the etiology was myogenic as there were a normal number of anterior horn cells in the spinal cord [4]. The other four cases were attributed to intrauterine anoxic–ischemic damage which was confirmed on autopsy [4–6]. There is only one published case of a monoamniotic twin pregnancy with discordance in the Pena–Shokeir phenotype [7]. That case was similar to ours in that that fetus also had bilateral pleural effusions with a low lung/body weight ratio. Histology of that baby also showed a disuse atrophy pattern as well as loss of anterior horn cells.

This is a novel case as it is only the second reported in the literature of a discordant Pena–Shokeir phenotype in a monoamniotic twin pregnancy.

Conflict of Interest

None declared.

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