Pena-Shokeir Syndrome's first case report from Syria

Mohammad Almoshantaf[1], Hidar Alibrahim[2], Haidara Bohsas[2], Shorouk Alabdo[2], Abdulqadir Nashwan[3], and Sarya Swed[2]

[1]Ibn Al-Nafees Hospital
[2]University of Aleppo Faculty of Medicine
[3]Hamad Medical Corporation

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Abstract

Pena-Shokeir Syndrome is considered to be a fatal congenital condition that is rarely diagnosed in neonates. We present the first ever reported case of Pena-Shokeir Syndrome from Syria. Both clinical assessment and early prenatal diagnosis are necessary to give more realistic options for the mother and the baby.

Running head: Pena-Shokeir Syndrome

Pena-Shokeir Syndrome's first case report from Syria

Mohammad Badr Almoshantaf[1], Hidar Alibrahim[2], Haidara Bohsas[2], Shorouk Alabdo[3], Abdulqadir J. Nashwan[4*], Sarya Swed[2]

1- Department of Neurosurgery, Ibn Al-Nafess Hospital, Damascus, Syria. (baderalmoushantaf1995@gmail.com)

2-Faculty of Medicine, Aleppo University, Aleppo, Syria. (saryaswed1@gmail.com, haideralibrahem1999@gmail.com, haidara.bohsas@gmail.com)

3-Department of Pediatrics, University Aleppo Hospital, Aleppo, Syria. (syshorouk@gmail.com)

4- Nursing Department, Hamad Medical Corporation, Doha, Qatar. (anashwan@hamad.qa)

***Corresponding author** : Abdulqadir J. Nashwan, Hamad Medical Corporation, Doha, P.O. Box 3050, Qatar. Email: anashwan@hamad.qa

Informed Consent

A written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

Abstract

Pena-Shokeir Syndrome is considered to be a fatal congenital condition that is rarely diagnosed in neonates. We present the first ever reported case of Pena-Shokeir Syndrome from Syria. Both clinical assessment and early prenatal diagnosis are necessary to give more realistic options for the mother and the baby.

Keywords: *Pena–Shokeir syndrome (PSS); Atrial septal defect (ASD); Congenital Anomalies*

Key Clinical Message

There is no current treatment for babies with Pena-Shokeir Syndrome, so continuous monitoring and palliative care are the keys to management.

Introduction

Pena-Shokeir Syndrome (PSS) is a deadly condition of numerous congenital contractures [1]. It was first identified in 1974, and the estimated frequency is 1 in 12,000 births [2]. Type 1 has been classified into 2 types; type 2 is described with a fetus's hypokinesia/akinesia marked by joint contractures, malformations of the face, and undeveloped lungs. PSS Type 2 is a severely degenerative neurological condition that leads to brain atrophy and is characterized by cataracts, intracerebral calcifications, micro-corners, optic atrophy, growth failure, and progressive joint contracture [3]. This syndrome may be difficult to diagnose since it has comparable ultrasonographic characteristics with other disorders [4]. However, as early as 12 weeks of gestation, ultrasonography may detect PSS by analyzing patterns such as hypoplasia of the lung tissue, reduced intrauterine movement, fetal edema, and locked limb posture. Extremities might have been stretched or contracted; the knees are often extended, the elbows are flexed, and the feet may have substantial rocker-bottom or equinovarus deformity. Hypertelorism, a sunken nose tip, micrognathia, and low-set ears are among the facial traits. The most common facial traits are hypertelorism, low-set ears, a depressed tip of the nose, micrognathia, and the head is often regarded as abnormally big compared to the body [3, 5]. Although cesarean delivery is normally reserved for obstetric reasons, the obstetric treatment will usually continue to be supportive, and a neonatologist and geneticist will assess the newborn after they are born. The patient should be informed about the options for palliative care after a second postnatal examination [3]. Although the eventual prognosis of PSS depends on the etiology, this syndrome has been characterized as nearly consistently fatal. Thirty percent of the fetuses affected by PSS are stillborn, and live-born infants often die after about a month of their lives. A congenital cerebral anomaly or severe respiratory failure related to pulmonary hypoplasia are the most common causes of mortality in infants born prematurely [3, 6, 7]. Patients who have previously given birth to a child affected by PSS should have strict fetal monitoring during future pregnancies. This will enable the early detection of any defects that may arise, and the recurrence risk may range from 0% to 25% [2]. We reported the first case of Pena-Shokeir syndrome from Syria; thus, we recommend that clinical physicians be aware of any abnormalities in ultrasonography during pregnancy, which could assist the pregnant woman and the doctor in recognizing the recurrence risk of this syndrome and making the necessary decisions.

Case presentation

We report a case of 33-year-old G3A1D1 married woman. In her last pregnancy, she had a history of blood transfusions in the first trimester, hydramnios, and was absent of fetal movement. Cesarean delivery was due to fetal-pelvic maladjustment. At birth, the female neonate was cyanosed and suffered from delayed screaming. Thus, the resuscitation protocol was initiated and successfully delivered. On the second day of birth, the neonate had suffered from severe dyspnea with a Spo2 of 80%, a heart rate of 180, and a respiratory rate of 76. On clinical examination, the baby had multiple skeletal malformations (rocker bottom foot (Figure 1), very short neck), microcephaly (head perimeter of 32 cm), large ears (Figure 2), upper teeth, micro mouth, cleft palate, hypotonia, absent reflexes, malformation in genital organs, and a hairy back (Figure 3). No cardiac murmurs were reported, but cardiac echography findings were atrial septal defect (ASD), tricuspid valve failure, and high pulmonary tension. The chest X-ray was unremarkable. Depending on the clinical and morphological features observed, we diagnosed the neonate with PSS. Due to the overall bad respiratory condition, the neonate was admitted to the incubator unit with an oxygen mask and prophylactic antibiotics. No signs of recovery were noted, and eventually, the baby died due to cardiopulmonary failure as resuscitation was ineffective.

Discussion

Fetal development akinesia deformity sequence, known as the Pena–Shokeir Syndrome (PSS), is characterized by early-onset neurogenic arthrogryposis and hypoplastic lungs [8]. Several studies have suggested that this syndrome is caused by mutations in the RAPSN and DOK7 genes [9]. Many risk factors contribute

to developing this disorder, including positive family history, environmental conditions like trauma, and hypotension [10]. Despite various possible diagnoses, PSS is related to chromosome 18 trisomies such as arthrogryposis and micrognathia [11]. It is possible to diagnose PSS in the case of a normal chromosomal study, micrognathia, and arthrogryposis in the child [10]. The inclusion of multi ultrasound in rendering mode enables a more thorough assessment of each suspected embryonic anomaly, completing the two-dimensional approach [12]. Current molecular genetic studies have increased our understanding of the hereditary reasons for this syndrome, indicating that many patients are at the extreme end of other identified neuromuscular diseases [13]. Despite pulmonary hypoplasia being crucial for conclusively defining PSS, this finding is often seen in the latter stage of fetal akinesia [14]. Magnetic resonance imaging (MRI) is another imaging technique for determining the presence of PSS. MRI should be requested even if it is not necessary to diagnose PSS when there is a suspicion of prenatal central nervous system defects. By comparing our case to other reported examples in the literature, we found Sumaiya Adam et al., [15], A young pregnant woman who had a standard ultrasound scan at 24 weeks of pregnancy. The routine second-trimester ultrasonography was performed and showed fetal micrognathia, a missing septum pellucidum, significant hyper-lordosis, and decreased fetal movements. PSS was diagnosed based on prenatal ultrasound, MRI results, and normal fetal chromosomes. The presence of pulmonary hypoplasia revealed in postnatal ultrasonography with a lower ratio of the fetal lung to head circumference (LHR) = 0.62 corroborated the final diagnosis of PSS. According to Eduardo Santana et al. [11], Another young nulliparous woman in her second pregnancy was hospitalized in the 28th week due to possible fetal arthrogryposis. The 2D ultrasonography revealed chronic spine hyperextension with head extension, persistent arm and leg flexion, hands and feet twisting, evidence of pulmonary hypoplasia, and retrognathia. The fetal position included continuous bending of the upper and lower limbs, hands and feet twisting, and more details of the micrognathia. A peri-membranous interventricular septal defect associated with intermittent fetal bradycardia was observed during the fetal echocardiography examination.

The cesarean birth went off without a hitch. The female infant weighed 1050 g and got APGAR scores of 0/0 in the 1^{st} and 5^{th} minutes. Unfortunately, the infant died after extensive resuscitation efforts. Because of the poor prognosis of PSS, in some countries where it is legal or socially-religiously unaccepted, late pregnancy termination may be offered as a therapy option. Instead, the parents might choose complete resuscitative action post-delivery or comfort treatment after birth. Unfortunately, not all patients will have a prenatal diagnosis and making decisions about lifesaving postpartum measures may be challenging. This article showed a rare case that causes severe abnormalities and requires constant follow-up and monitoring to reach the best management.

Conclusion

In this case, PSS has never before been reported in Syria. For better treatment choices for the mother and the child, a professional understanding of this disease and early prenatal diagnosis are needed. Continuous monitoring and palliative care are the main therapeutic strategies for newborns with PSS since there is currently no cure.

Declarations

Acknowledgment

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Conflicts of interest

All authors declared no conflict of interest.

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Ethical approval

This case report was reviewed and approved by the ethics committee, Aleppo University Hospital, Aleppo university, Syria.

Registration of research studies

Not applicable.

Author contribution

All authors have contributed to writing and reviewing the manuscript.

Guarantor

D. Mohammad B. Almoshantaf

US Sanctions

I would like to declare that the authors have prepared this submission in their personal capacity and not as an official representative or otherwise on behalf of a sanctioned government.

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Figure 2: Large Ear

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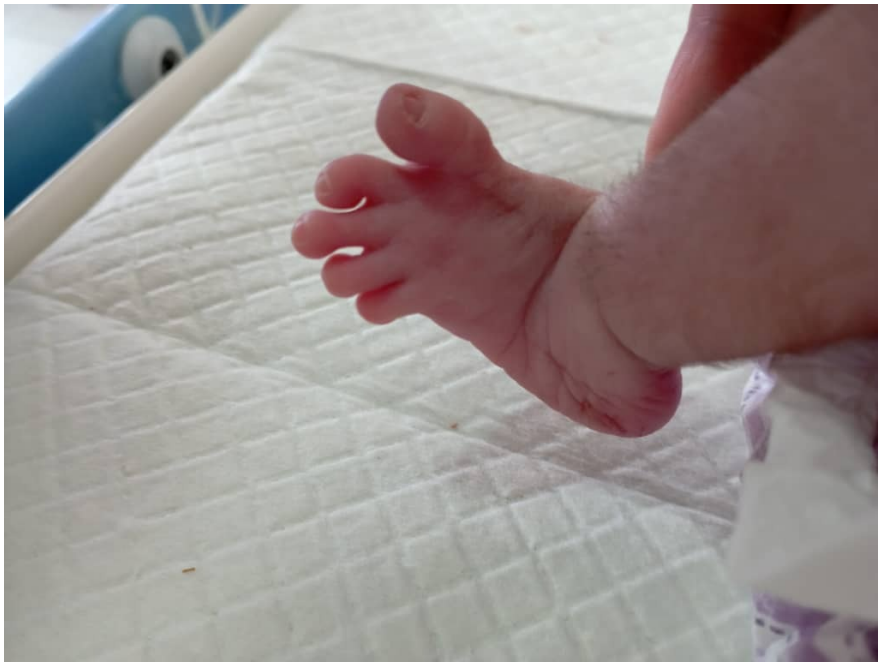

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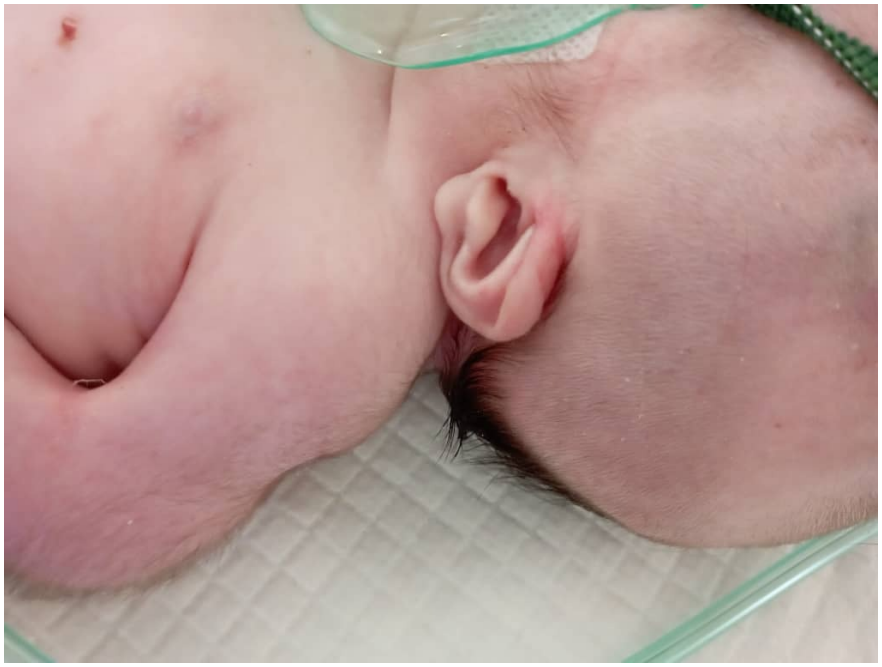

Figure 2: Large Ear

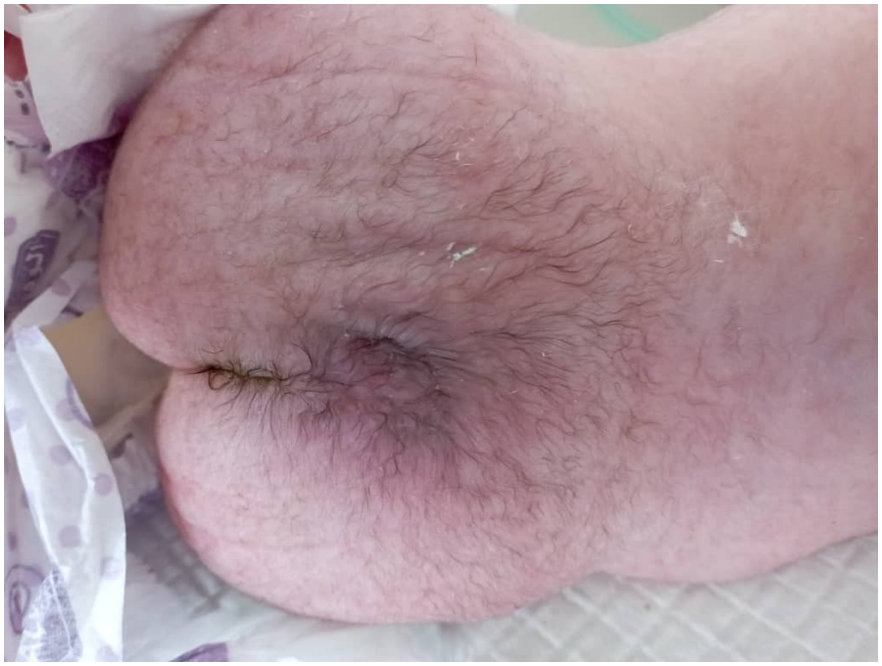

Figure 3: Hairy Back