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Case Report

Pena-Shokeir Syndrome: A Case Report

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Abstract

Introduction: Pena-Shokeir syndrome (fetal akinesia deformation sequence, FADS) is an autosomal recessive lethal disorder characterized by combination of abnormal limb position, facial anomalies (micrognathia), camptodactyly, restrictive fetal movement with reduced or absent response to acoustic stimulation, growth restriction, polyhydramnios, and pulmonary hypoplasia. Limb defects like rocker bottom foot and clubfoot are other prominents of the syndrome.

Case report: Obstetric ultrasonographic examination of a 24-year-old pregnant woman, consanguineous with her husband, revealed a single female fetus with contractures of the lower limbs, persistent flexion of the bilateral wrist, elbow joints and the knee joints consistent with Pena Shokeir syndrome phenotype. The parents were informed about the diagnosis and its poor prognosis. Fetus had no viability, therefore the termination of the pregnancy was offered to the parents and they accepted. We report the prenatal ultrasonographic and postnatal clinical findings suggestive of Pena-Shokeir syndrome case.

Conclusion: Pena–Shokeir syndrome is a potentially lethal condition and most cases are diagnosed prenatally by ultrasound.

Keywords: Pena–Shokeir syndrome, Fetal akinesia, Ultrasound.

Introduction

Pena and Shokier identified an early lethal disorder involving multiple joint contractures, facial anomalies, and pulmonary hypoplasia with autosomal recessive mode of inheritance with a frequency of 1 in 12,000 live births. Hall recently suggested that this clinical phenotype is secondary to decreased in-utero movement. Sonographic diagnosis is based on absent fetal movement and abnormal limb position. As such it is etiologically heterogeneous and is similar to fetal akinesia deformation sequence (FADS), a pattern of structural defects described by Moessinger.

Abnormalities [1]:

Growth: Prenatal onset growth deficiency. Head circumference is frequently spared.

Cranio facial: Rigid expressionless face, prominent eyes, hypertelorism, telecanthus, epicanthal folds, poorly folded small and posteriorly angulated ears, depressed nasal tip, small mouth, high arched palate, micrognathia.

Limbs: Multiple ankylosis (elbows, knees, hips and ankles), ulnar deviation of hands, rocker bottom feet, talipes equino varus, camptodactyly. Absent or sparse dermal ridges with frequent absence of the flexion creases on fingers and palms.

Lungs: pulmonary hypoplasia.

Genitalia: Cryptorchidism.

Others: Apparent short neck, polyhydramnios, short gut syndrome with malabsorption with small or abnormal placenta, relatively short umbilical cord, cleft palate, cardiac defects.

Some of these babies are born prematurely. Those born at term are invariably small for date. Approximately 30% are still born. Those live born die of complications of pulmonary hypoplasia within 1st month of life.

Case history:

A 24-year-old woman primigravida with II degree consanguineous marriage presented to our institution at 24 weeks gestation for pain abdomen and leaking per vagina. There was no significant family history of skeletal, genetic or congenital anomalies. There was no history of exposure to alcohol, teratogenic drugs. Antenatal ultrasound (US) performed at 24 weeks revealed right talipes equinovarus with bilateral clenched fists and micrognathia, persistent flexion of the bilateral wrist, elbow joints and knee joints. Ultrasound showed the biparietal diameter (53.8 mm) and femur length (40.1 mm) were consistent with the gestational date (24 weeks), while the abdominal circumference (185 mm) revealed growth restriction (22 weeks). Fetal posture was fixed and there was no movement during the ultrasound. Ultrasound images demonstrated scalp oedema. No neurological abnormalities were detected. Diagnosis of Pena-Shokeir syndrome was made. Parents were informed of the poor prognosis. A still born female child was born at 24 weeks of gestation with weight of 600 gm. Gross pathological examination revealed flexion deformity at wrist, hip and knee joints, micrognathia, camptodactyly, talipes equino varus, rocker bottom foot, low set ears, short neck, hypertelorism. Diagnosis of fetal dyskinesia syndrome, consistent with Pena-Shokeir syndrome was made.





Discussion:

In 1974, Pena and Shokeir first described this lethal autosomal recessive syndrome characterized by arthrogryposis, camptodactyly, facial anomalies and pulmonary hypoplasia in two siblings.[2] In 1985, MacMillan et al. diagnosed Pena–Shokeir syndrome type I prenatally using sonography.[3] Since, then several cases of prenatal sonography findings have been reported [4-6]. Sonographic diagnosis is based on absent fetal movement and abnormal limb position. We report the prenatal sonographic, post natal clinical findings of Pena–Shokeir phenotype with pathological correlation. It is a rare syndrome with an incidence of approximately 1 in 12,000 births. Such babies have a poor prognosis with approximately 30% of affected infant stillborn and most live births die in early neonatal period due to complications of pulmonary hypoplasia. In utero continuous fetal movement is

essential for development of normal respiratory and limb function. If movements stop, the joints become stiff and muscle mass decreases. The polyhydramnios and pulmonary hypoplasia are related to depressed swallowing and absence of normal fetal breathing[7].

Pena–Shokeir syndrome is characterized by the arthrogryposis, facial anomalies (micrognathia), pulmonary hypoplasia, and dysmorphic features resulting from fetal akinesia. Pena–Shokeir syndrome affects the muscles of the back, upper and lower limbs and the deformities are classically bilateral and symmetrical. The differential diagnosis includes Freeman– Sheldon syndrome, multiple pterygium syndrome, trisomy 18, trisomy 13, Potter syndrome, Neu–Laxova syndrome, restrictive dermopathy, Larsen syndrome, and cerebroocular- facial-skeletal syndrome.

In our case, diagnosis was established on basis of sonography, persistent flexion deformity of knees and wrist, absent fetal movement, micrognathia, camptodactyly, talipes equinus varus, rocker bottom foot, hypertelorism. Since Pena–Shokeir syndrome is a lethal anomaly with poor prognosis, search for associated malformation is of more of academic rather than therapeutic interest.

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