

Pena-Shokier Syndrome: A Case Report

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Abstract: *Pena and Shokier Syndrome (PSS) is an autosomal recessive non aneuploidic condition, Identified as a lethal disorder. It was initially described by S. D. J. Pena and M. H. K. Shokeir in 1974. The frequency is 1 in 12000 live births with autosomal recessive mode of inheritance. It is a rare congenital syndrome involving degeneration of the brain and spinal cord and characterized by facial, head, skeletal and muscular abnormalities. It involves multiple joint contractures, facial anomalies and pulmonary hypoplasia with an autosomal recessive mode of inheritance. Hall suggested clinical phenotype is secondary to decreased in fetal movements in utero.*

Keywords: Pena-Shokier Syndrome, Fetalakinesia, Ultrasound, early diagnosis.

1. Case Report

A two days old baby, born to non consanguineous parents 1st born female baby was brought with the complaints of respiratory distress. There was no significant family history of skeletal, genetic or congenital anomalies. Antenatal ultrasound was not performed (Un booked case). Baby was delivered by caesarean section. APGAR at one minute was 4 and at 5 minutes was 7 Baby progressively deteriorated and was referred to our institution.

Infantogram: arthrogryposis multiplexcongenita

Ultrasound abdomen was normal.

Nasogastric Tube aspiration done: bilious aspirate

Physical findings:

Face:

Rigid expressionless face, prominent eyes, hypertelorism, telecanthus, epicanthalfolds, poorly folded, small and posteriorly angulated ears, depressed nasal tip, small mouth, high arched palate, micrognathia

Limbs:

Multipleankylosis, ulnar deviation of hands, rocker-bottom feet, talipusequinovarus, camptodactyly, absent or sparse dermal ridges with frequent absence of flexion creases on the fingers and palms, clenched hands.

Genitalia. cryptorchidism.

Others:

Apparent short neck, polyhydramnios, short gut syndrome with malabsorption, small or abnormal placenta, short umbilical cord.

These features suggestive of Pena Shokier syndrome.

Fatal due to pulmonary hypoplasia

Pena and Shokier Syndrome type I is caused by mutations in the RAPSN or DOK7 genes.

Future scope of study:

Gene mapping and karyo typing should be done. Prenatal diagnosis by chorionic villus sampling and early termination of pregnancy should be advised.

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