International Journal of Contemporary Pediatrics
Majhi SC et al. Int J Contemp Pediatr. 2015 Feb;2(1):59-61
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: 10.5455/2349-3291.ijcp20150215

Case Report

Jeune syndrome - asphyxiating thoracic dystrophy and its prenatal diagnosis: a rare case report

Subash Chandra Majhi[1]*, Bobbity Deepthi[1], Sharmila Pradhan[2], Babita Ramani[2]

[1]Department of Pediatrics, VSS Medical College & Hospital, Burla, Sambalpur, Odisha, India
[2]Department of Obstetrics and Gynecology, VSS Medical College & Hospital, Burla, Sambalpur, Odisha, India

Received: 7 December 2014
Accepted: 22 December 2014

***Correspondence:**
Dr. Subash Chandra Majhi,
E-mail: majhi.subash660@gmail.com

ABSTRACT

Jeune syndrome is an extremely rare condition representing Asphyxiating Thoracic Dystrophy (ATD) and it is potentially lethal congenital dwarfism with estimated incidence of 1 per 100000-130000 live births.[1] A term male neonate, born by vaginal delivery to a primigravida with polyhydramnios, presented to our NICU with birth asphyxia and severe respiratory distress since birth along with multiple characteristic skeletal anomalies including small bell shaped thorax and short limbs leading to diagnosis of Jeune syndrome. Follow up of the mother in her second pregnancy with prenatal sonographic examination at 17 and 19 weeks revealed the same type of skeletal dystrophy in this pregnancy. Diagnosis was made on ultrasonographic findings and detected abnormalities were confirmed by X-ray following termination of her second pregnancy. This is a case of Jeune syndrome in siblings.

Keywords: Jeune syndrome, Asphyxiating thoracic dystrophy, Polyhydramnios, Skeletal anomalies

INTRODUCTION

Asphyxiating Thoracic Dystrophy (ATD) also known as Jeune syndrome is a rare autosomal recessive disorder with variable severity and multiple musculo-skeletal manifestations with an incidence estimated as 1:100000-130000 live births.[1] It is a rare short rib skeletal dysplasia characterized by short-limbed dwarfism, a small, narrow bell shaped thorax, micromelia, varying degrees of rhizomelic brachymelia, polydactyly of hands and feet, pelvic abnormalities, and renal anomalies, with a considerable neonatal mortality as a result of respiratory distress.[2] Renal, hepatic, pancreatic and ocular complications may occur later in life. The prenatal diagnosis of fetal skeletal dysplasia is a challenging task as there could be large number of possible diagnosis but skeletal dysplasia in particular includes lethal diseases, which makes prenatal diagnosis in such cases highly important, both medically and societally. Diagnostic imaging for prenatal diagnosis is accomplished by ultrasound, MRI and CT scan, and chromosomal and genetic diagnosis is also performed as needed.[3] We in our case focus on diagnosing this unique case by noninvasive ultrasound indicating possibility of an easy prenatal diagnosis.

CASE REPORT

A first order full term male baby vaginal delivered by face presentation born to a primigravida non consanguineous married couple presented to our NICU with history of birth asphyxia and severe respiratory distress soon after birth with the multiple skeletal anomalies. The antenatal ultrasonography had revealed polyhydramnios. Birth weight of the baby was 2.6 kg, length being 42 cm, head circumference being 33 cm. The neonate had a typical small narrow bell shaped thorax, bilateral short upper limbs (rhizomelic

shortening), small hands (short metacarpals) with ecchymosis and bruises of whole face. X-ray showed typical findings of horizontal short ribs, irregular enlarged costochondral junction, small and horizontal clavicles. Ultrasonography findings revealed bilateral hydronephrosis. The neonate required ventilatory support due to severe respiratory distress and died within 72 hours due to progressive respiratory failure.

The mother with the deceased child was followed 18 months later in her second pregnancy. Prenatal sonographic examinations were performed in 17 and 19 weeks. The length of the long bones like humeri, femora, and tibiae was short (below the mean) for gestational age, and the thorax was abnormally flat and narrow with short ribs. Thoracic circumference <3rd centile while abdominal circumference was within normal limits. We concluded that the fetus had Jeune syndrome following which pregnancy was terminated and detected abnormalities were confirmed by an X-ray of dead fetus. The characteristic skeletal changes of Jeune syndrome are distinct enough at 17 weeks of fetal age to permit sonographic diagnosis.

Figure 1: Neonate with Jeune.

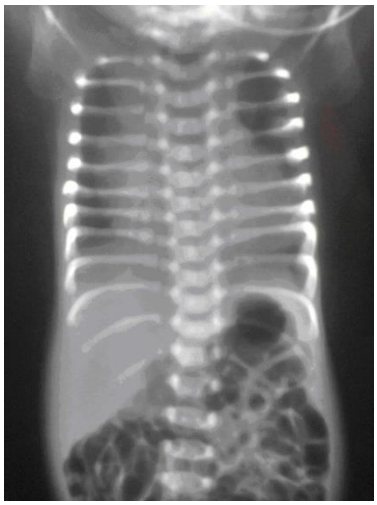

Figure 2: Radiographic findings of Jeune syndrome.

DISCUSSION

Jeune syndrome, also known as Asphyxiating Thoracic Dystrophy (ATD), is a rare autosomal recessive disorder. In 1955, Jeune et al., described familial asphyxiating thoracic dystrophy in a pair of siblings with severe narrowing of thoraxes. Its gene has not been mapped, but various studies have shown the locus to be on 12p and 15q13.[4] Jeune syndrome is the first chondrodysplasia to be linked to a defect in intraflagellar transport (IFT) or primary cilia function belonging to the family of skeletal ciliopathies. It is a disorder associated with dysfunction of primary cilia, classified as 1 of the 6 Short-Rib Polydactyly Syndrome (SRPS) disorders.[5] Classic manifestations in infancy include dwarfism with short ribs, short limbs, polydactyly of hands and feet and characteristic radiographic changes in the ribs (small bell shaped thorax) and pelvis (short iliac bones with acetabular spurs)[1] along with retinal degeneration.[5] Severity of clinical and radiological features is variable. Lung hypoplasia, presumably due to restricted thoracic cage, causes alveolar hypoventilation, and approximately 60-70% patients of Jeune syndrome die from respiratory failure in infancy. Chronic renal failure may ensue in survivors. Variability in clinical, radiological and pathological manifestations may be related to genetic heterogenicity. Other skeletal dysplasias which are close mimics are achondrogenesis, achondroplasia, osteogenesis imperfecta, thanatophoric dwarfism, hypophosphatasia.[7] Prenatal ultrasonography may show narrow thorax, short hypoplastic ribs and short tubular bones.[8,9] Important in diagnosis of spectrum of skeletal dysplasias are measuremnts like Femur Length (FL), Thoracic Circumference (TC), Rib Cage Diameter (RCD), Abdominal Circumference (AC), TC/AC ratio. Thus based on these measurements, we can diagnose abnormalities like mesomelic (shortening of radius and ulna in upper limb and tibia and fibula in lower limb), rhizomelic (shortening of humerus in upper limb and femur in lower limb) and micromelic (shortening of all tubular bones), chest circumference <5th percentile for gestational age, suggests lung hypoplasia, and femur length/abdominal circumference ratio <0.16 suggests lung hypoplasia. Other ultrasonographic findings include polyhydramnios and absent or feeble respiratory movements. Confirming diagnosis of such lethal skeletal dysplasia at an early stage should be treated with care and attempt should be to reduce the discrepancy in timing of prenatal diagnosis and correct diagnosis rate.[3]

CONCLUSION

Jeune syndrome, though a rarity, prenatal diagnosis of this fatal condition is possible due to its characteristic skeletal abnormalities on ultrasonography thus helping parents make decision of pregnancy termination.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Verma A, Gurudatta HS. Jeune syndrome. Indian Pediatr. 2004;41:954-5.
2. Murotsuki, Haruki Nishizawa, Yasuhiro Udagawa. Ultrasonic diagnosis of fetal bone and small parts. Donald School J Ultrasound Obstet Gynecol. 2011 Jan-Mar;5(1):45-55.
3. Morgan NV, Bacchelli C, Gissen P, Morton J, Ferrero GB, Silengo M, et al. A locus for asphyxiating thoracic dystrophy, ATD, maps to chromosome 15q13. J Med Genet. 2003;40:431-5.
4. Beales PL, Bland E, Tobin JL, Bacchelli C, Tüysüz B, Hill J, et al. IFT80, which encodes a conserved intraflagellar transport protein, is mutated in Jeune asphyxiating thoracic dystrophy. Nat Genet. 2007;39:727-9.
5. Casteels I, Demandt E, Leguis E. Visual loss as the presenting sign of Jeune syndrome. Europ J Pediatr Neurol. 2000;4:243-7.
6. Twining P, McHugo JM, Pilling DW. Textbook of foetal abnormalities. In: Twining P, McHugo JM, Pilling DW, eds. A Book. 1st ed. Edinburgh: Churchill Livingstone; 2000: 254-255.
7. Das Bibhuti B, Nagaraj A, Fayemi A, Rajegowda Benemanahalli K, Giampietro Philip F. Foetal thoracic measurements in prenatal ultrasonography of Jeune syndrome. Indian J Pediatr. 2002;69:101-3.
8. Callen PW. Prenatal ultrasonography. In: Callen PW, eds. Ultrasonography in Obstetrics and Gynaecology. 4th ed. Philadelphia: WB Saunders; 2000.
9. Chen CP, Lin SP, Liu FF, Jan SW, Lin SY, Lan CC. Prenatal diagnosis of asphyxiating thoracic dysplasia (Jeune syndrome). Am J Perinatol. 1996;13:495-8.

DOI: 10.5455/2349-3291.ijcp20150215
Cite this article as: Majhi SC, Deepthi B, Pradhan S, Ramani B. Jeune syndrome - asphyxiating thoracic dystrophy and its prenatal diagnosis: a rare case report. Int J Contemp Pediatr 2015;2:59-61.