A rare case of Jeune syndrome (Asphyxiating thoracic dystrophy)

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Abstract

A rare case of Jeune syndrome (Asphyxiating thoracic dystrophy) found in ultrasound examination of fetus describing short thorax, short all long bones with polydactyly.

Keywords: Jeune Syndrome, USG, Asphyxiating thoracic dystrophy

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Introduction

Jeune syndrome also known as Asphyxiating Thoracic Dystrophy is a rare genetic congenital disorder that affects a fetus's cartilage and bone development. It affects the pelvis, ribcage, arms and legs.[1]

Case Report

A 27 years old Hindu primi female with BMI 17 had 23 weeks of pregnancy and came to Mahavir Hospital (Scientific Research Institute) for the first time. Routine check-up was done.

On ultrasound examination[2]

Biometry:
BPD: 57.4mm
HC: 205.6mm
AC: 169.4mm
FL: 25.4 mm
HUMERUS: 23.1 mm
Efw: 350gm
Presentation: cephalic
Fetal heart activity visualized and fetal movement was normal
Placental site: anterior grade 0
Nuchal and Skin: Nuchal oedema
Thorax: chest wall was long and narrow (BELL SHAPED)
Heart: Normal 4 chambers, 3 vessels outflow tract
GIT and Urinary tract normal

Extremities
Left foot: 39.9mm
Right foot: 40.2mm
Left Humerus: 23.0mm
Right Humerus: 22.3mm
Left Femur: 27.6mm
Right Femur: 27.0mm
Left Radius: 23.0mm
Right Radius: 21.2mm
Left Ulna: 20.3mm
Right Ulna: 20.1mm
Left Tibia: 20.1mm
Right Tibia: 22.6mm
Left Fibula: 21.3mm
Right Fibula: 23.3mm
Left Femur/foot: 0.69
Right Femur/foot: 0.67

Hands

1. overlapping fingers left side
2. right side polydactyly

Feet

1. Humerus: fractured and short bilateral[3]
2. Femur: bowed and short bilateral
3. Tibia: short bilateral
4. Fibula: short bilateral
5. Ulna: short bilateral
6. Radius: short bilateral

Discussion

Jeune syndrome was first described by M Jeune in 1955[1,2,14]: Jeune syndrome is an autosomal recessive disorder[3] that affects the bone development in fetus. It affects about 1 in every 100,000 to 130,000.

If mother and father both carry this gene, every child they conceive has a one-in-four chance of having this syndrome.

Jeune syndrome cause abnormality in child's thoracic cage,[5] pelvis, both upper limbs and lower limbs thoracic cage are smaller and narrower than usual and keep fetus lungs under develop so fetus is not able to breath properly ex utero and can have URTI or LRTI or respiratory failure.

Usually children born with jeune syndrome are not able to pass infancy but those have early childhood have to suffer from renal and cardiac problems which are severe in nature[3]

In utero fetus have both limbs are underdeveloped and small in size and fragile and get easy fracture in side uterus also

Fetus have renal abnormality like renal failure form cystic renal disease.

Liver failure because of hepatic cirrhosis and retinal problem.[3,4,5]

X- Ray features[6,7]:

1. Short and narrow elongated thoracic cage (Bell shaped)
2. Handle bar (High Riding) clavicle
3. Costochondral junction are irregular
4. Short and broad phallanges
5. Short distal upper and lower limbs
6. Polydactyly
7. Epiphysis are coned shaped
8. Acetabulam may be trident or dyspalstic with flat roof
9. Iliac bones –short and flared
10. Femoral epiphysis may have premature closure

USG finding

1. cirrhosis of liver[8]
2. renal disease (cystic kidney)[9]
3. cystic disease of pancreas

Genetic Make Up

Different 11 genes are found to have mutation for Asphyxiating Thoracic dystrophy most common change in gene IFT80, DYNC2CHI has also found in 50% of cases.[10]

The genes are involving in making of protein that found on tip of cilia and are involved in a process called IFT- intra flageller transport.

Sonic Hedgehog pathways is essential for growth and proliferation and maturation of cells to live cartilage and bone and this is deficient in Jeune syndrome.

Abnormality of cilia in tissue of kidney, liver, retina can cause symptoms in respective organs.

Asphyxiating thoracic dystrophy is one part of group disorders like skeletal ciliopaphy or ciliary chondrodysplasias.[11,13]

SRPSs(short rib-polydactyly Syndrome) are also seen in jenue syndrome.

This is an inherited autosomal resseive pattern means both copies of the gene in each cell have mutations

Treatment

Prognosis is very poor and variable ad there is marked phenotypic variation.

Ex utero those survivors have lung complication because of thoracic cage abnormalities and may go for mortality.

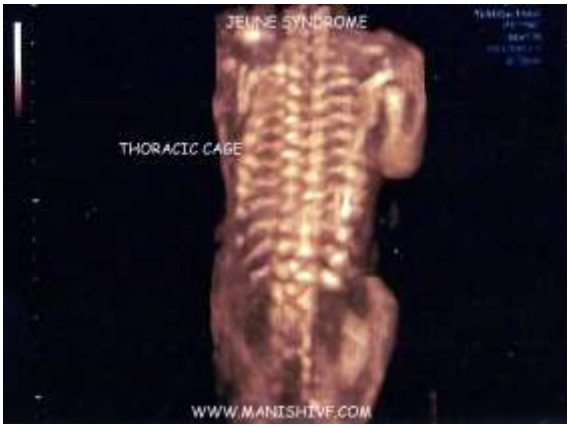


Fig. 1 a: Thoracic cage 3D

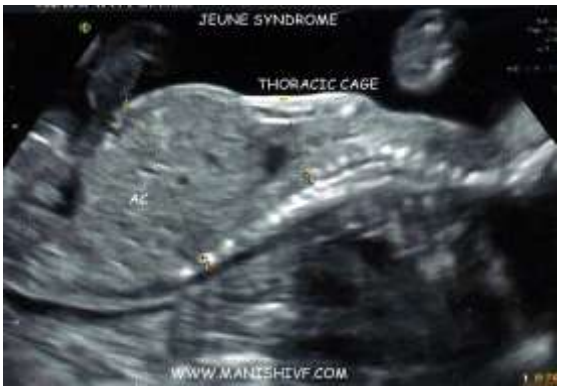


Fig. 2 b: Thoracic cage 2D

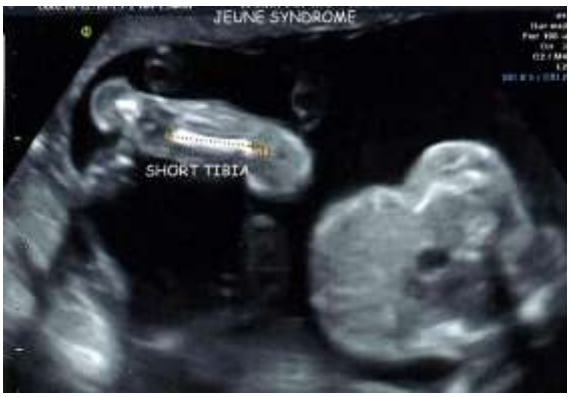
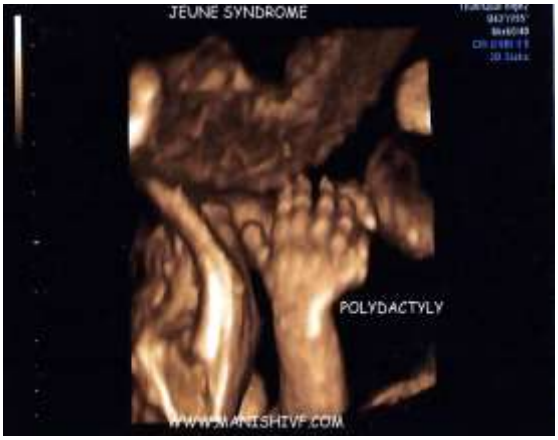


Fig. 2: Short tibia



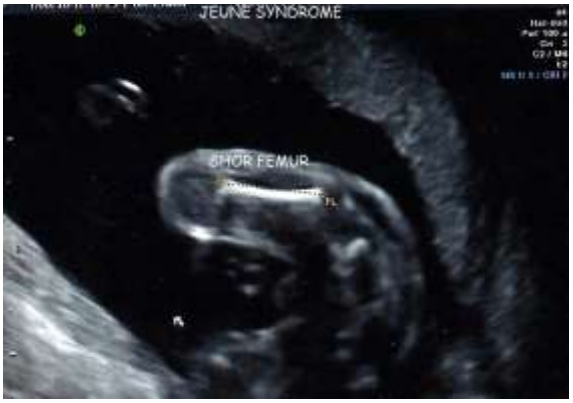


Fig. 5: Short Femur

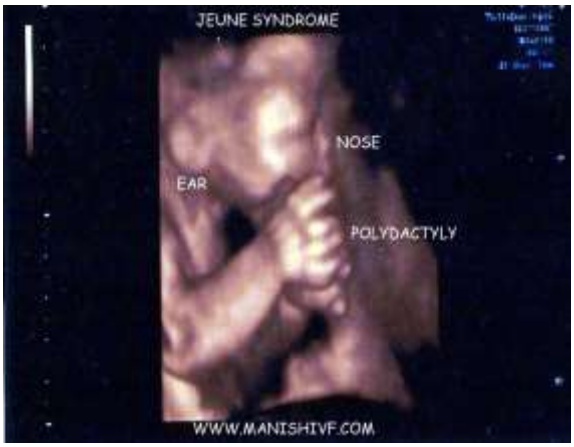


Fig. 3: Polydactyly 3D

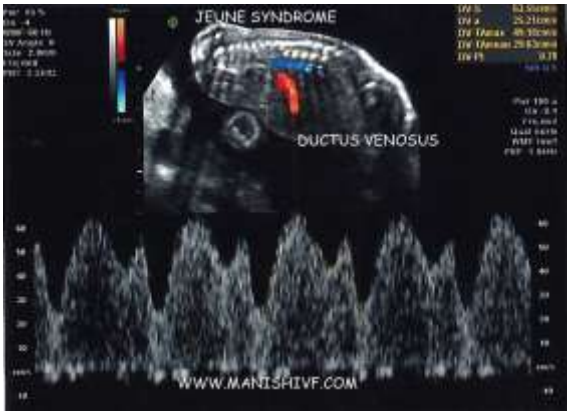


Fig. 6: Normal Ductus Venosus

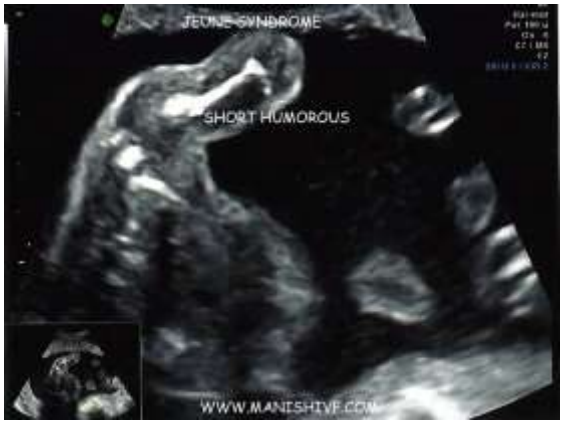


Fig. 4: Short Humerus

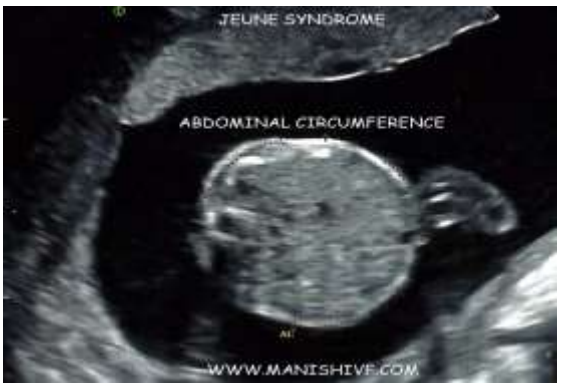
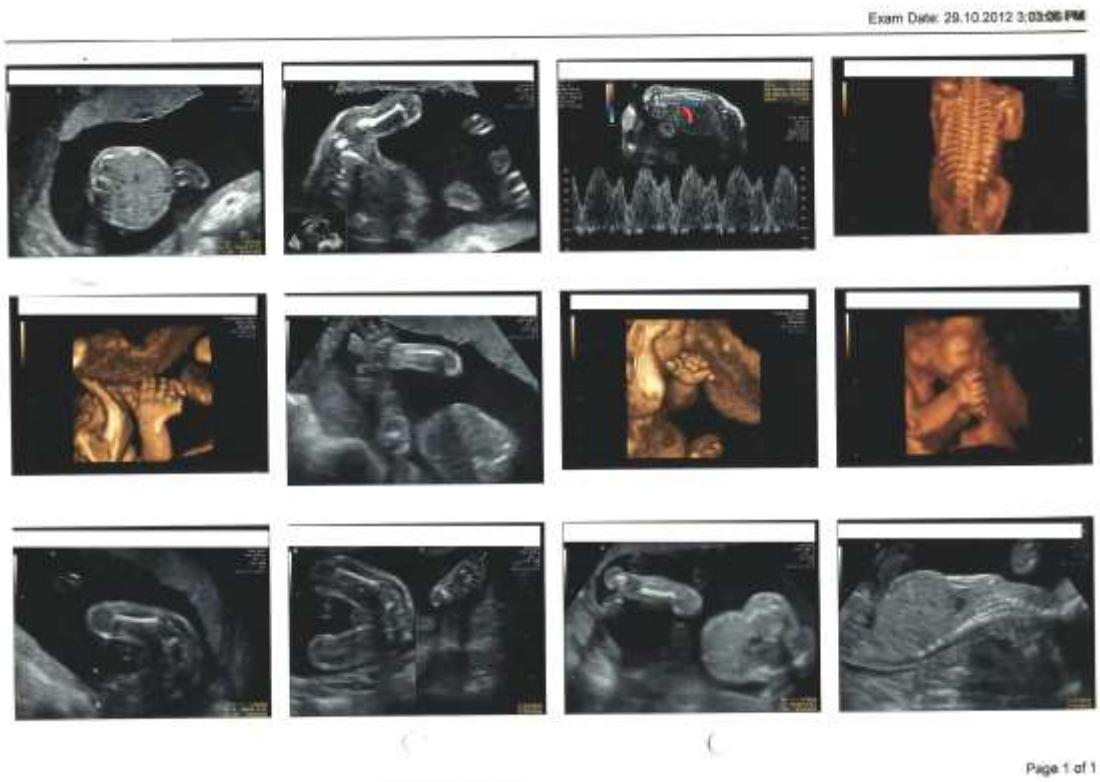


Fig. 7: Abdominal Circumference

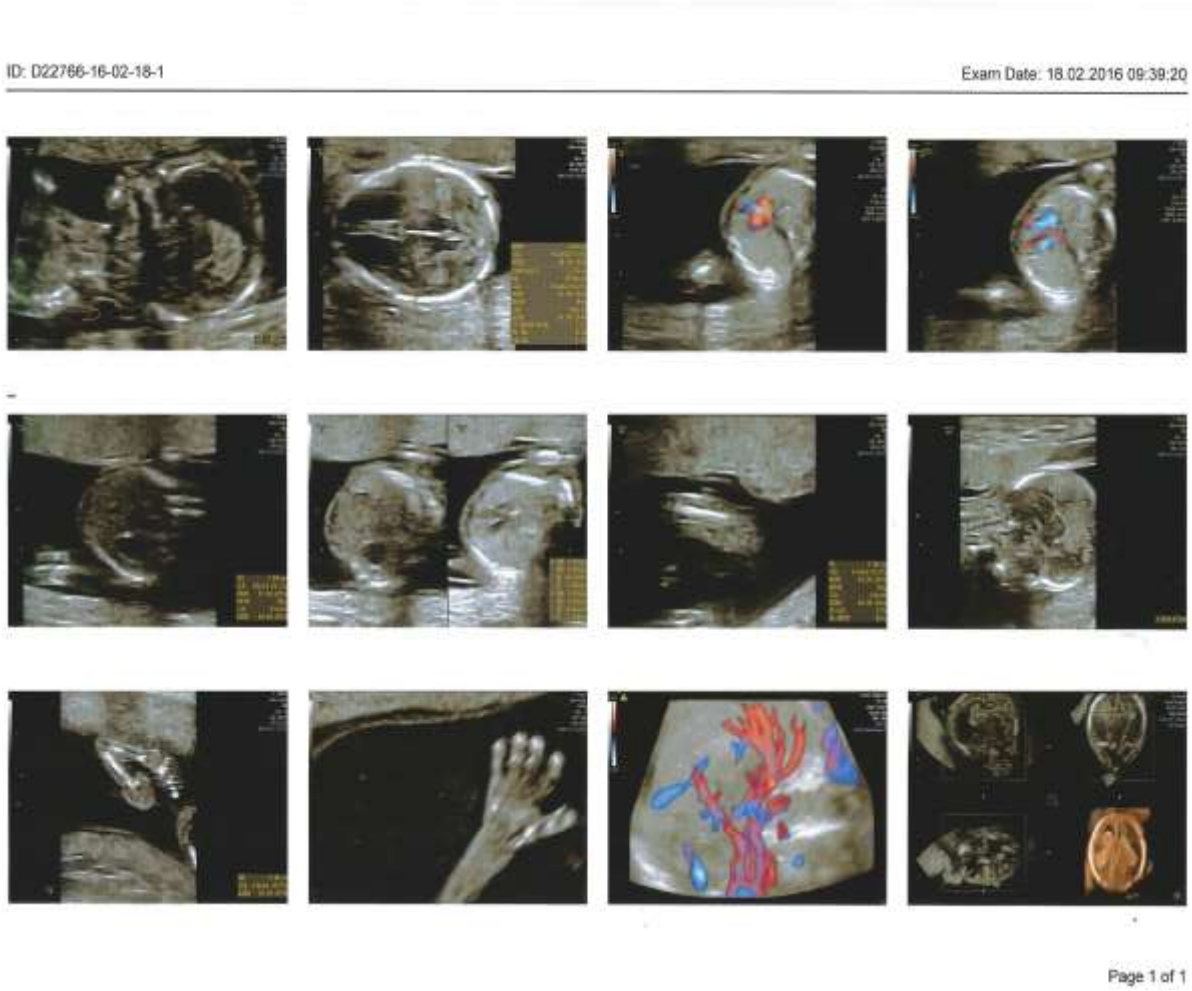


A case of Jeanu syndrome

Follow up

- She had terminated previous pregnancy somewhere else so autopsy findings were not found.
- She again came with pregnancy after 4 years.
- Her LMP IS 9/11/2015.
- Present ultrasound revealed no obvious fetal anomalies.
- Fetal biometry suggestive of NORMAL fetal growth as per gestation.
- Fetal Maturity is about 14week 3 Days at the time of scan.
- Normal and adequate quantity of liquor.
- Primary evaluation of fetal heart by 4 chamber heart, 3 vv and STIC shows no abnormality on present examination.
- There is no polydactyly.
- Her EDD is 15/8/2016. And she delivered normal male child of 2.9 kg on 04/08/2016

Normal usg in this pregnancy and normal healthy baby…….



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