

Case of the month : July 21.

Case Courtesy : Dr. Sanyogeeta Benare.

CASE TITLE – SKELETAL DYSPLASIA (OSTEOCHONDRODYSPLASIA)

Patient profile - 30 Yrs primi came for Anomaly scan. Her gestational age was 17 wks 0 days, assigned as per the LMP i.e. 10/4/2021. Dating scan was done elsewhere at 10 weeks . NT scan was offered but not done.

Clinical history - There was no specific family history of any disorder or skeletal dysplasia. No H/O consanguinity. Parenteral height was average.

Ultrasound findings – Single live Intrauterine fetus with normal amniotic fluid.

Fetal Growth Parameters - BPD ,HC and AC were corresponding with 17 to 18 weeks of gestational age. There was a significant shortening of all long bones with parameters corresponding to 13 to 14 weeks of gestational age i.e. less than 1 % centile. Detailed fetal anatomical survey was done with findings described as below -

Skeletal system - Long bones of all four limbs were short (less than 1 % centile) i.e. MICROMELIA .Limb movements were seen but flicker. No flexion – extension movements seen through out the scan.Bone mineralisation was normal. No evidence of fracture . No digital abnormality found. Right femur was short and bowed “ TELEPHONE RECEIVER FEMUR ”. Femur to foot ratio less than 1.

Fig. 1. [Page 2]

Thoracoabdominal Sagittal view showed disproportionately smaller thorax with Protruberant abdomen with dip at thoracoabdominal interphase. Axial view showed Hypoplastic ribs. Thoraco-abdomemal ratio was abnormal. (less than 0.6). Trunk length was short.

Face & CNS – There was a flat nasal bridge / midface hypoplasia with frontal bossing and macrocranium. Fetal calvarium & brain was normal. Spine was visualised in transverse and longitudinal view which appeared normal. Orbits & lips were normal.

There was no other significant anomaly found in fetus. Fetal heart , abdominal organs and KUB was normal. Uterine artery Doppler indices were within normal limits. **Fig.2.[Page 3]**

CONCLUSION – Considering the significantly abnormal findings like micromelia , narrow thorax , short trunk length , macrocranium & flat facial profile , diagnosis of LETHAL TYPE SKELETAL DYSPLASIA was made. Most common differentials considered in this case were THANATOPHORIC DYSPLASIA & ACHONDROGENESIS. After counselling about the findings and prognosis parents opted for the termination of pregnancy. External features were confirmed after the termination. Post abortus radiograph and genetic work up advised but not done. **Fig.3. [Page 4]**.

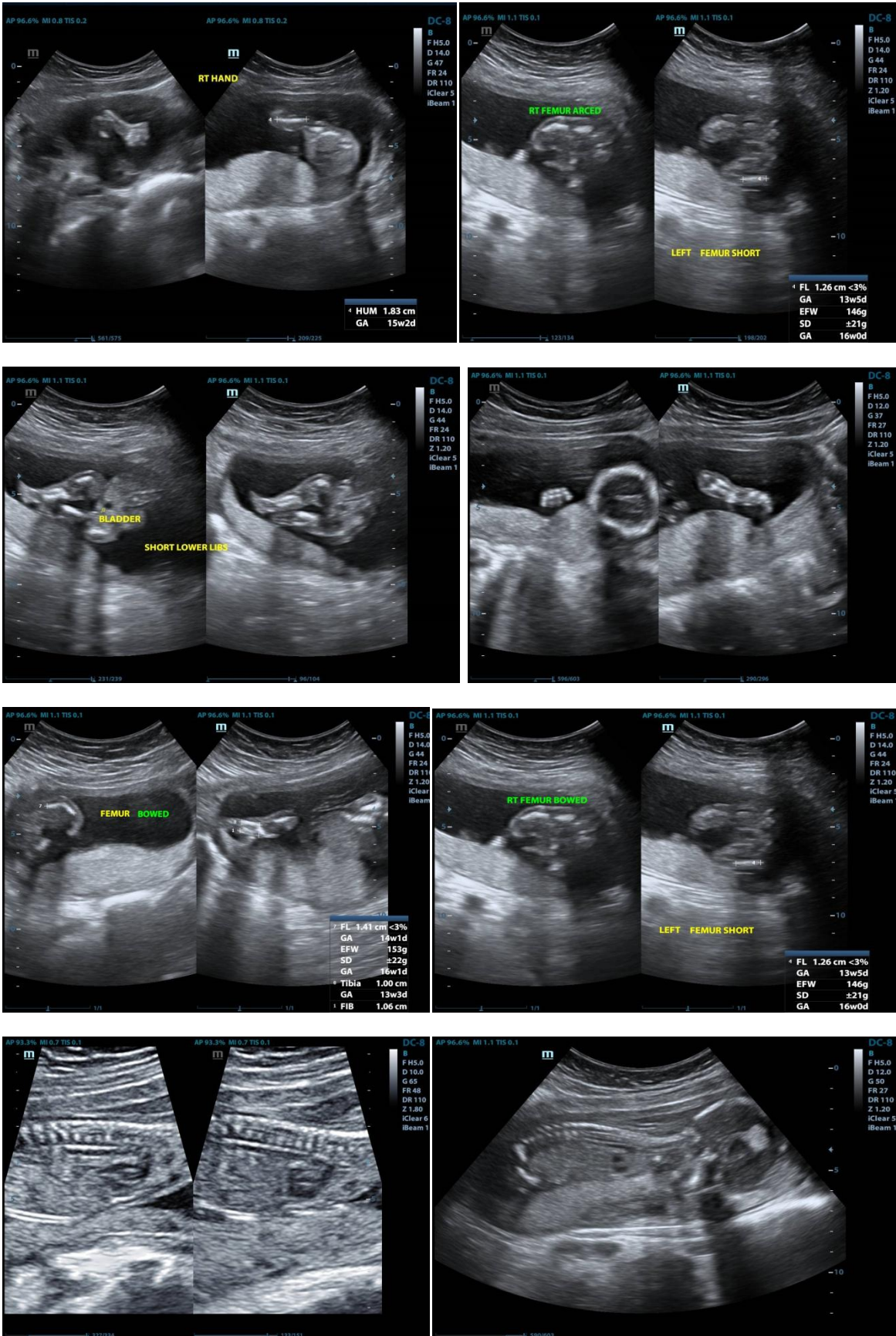
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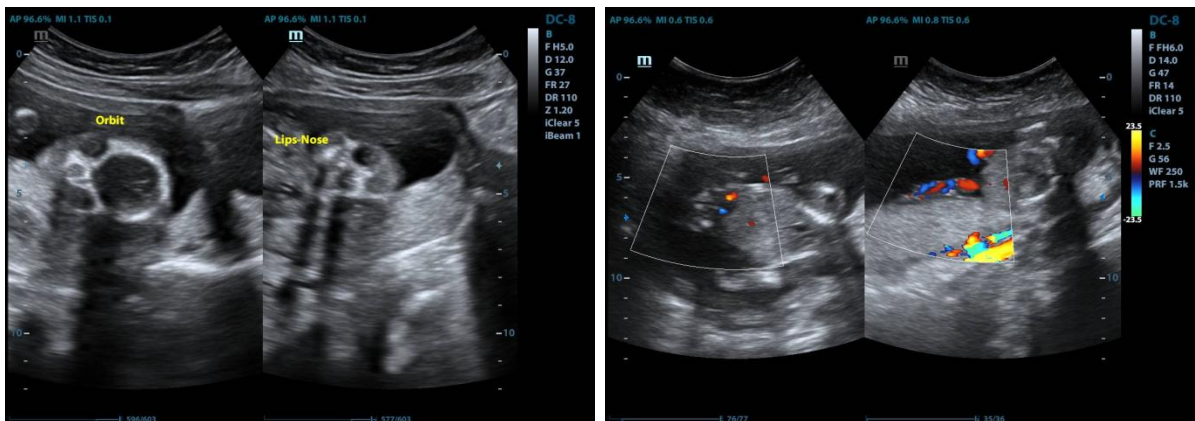
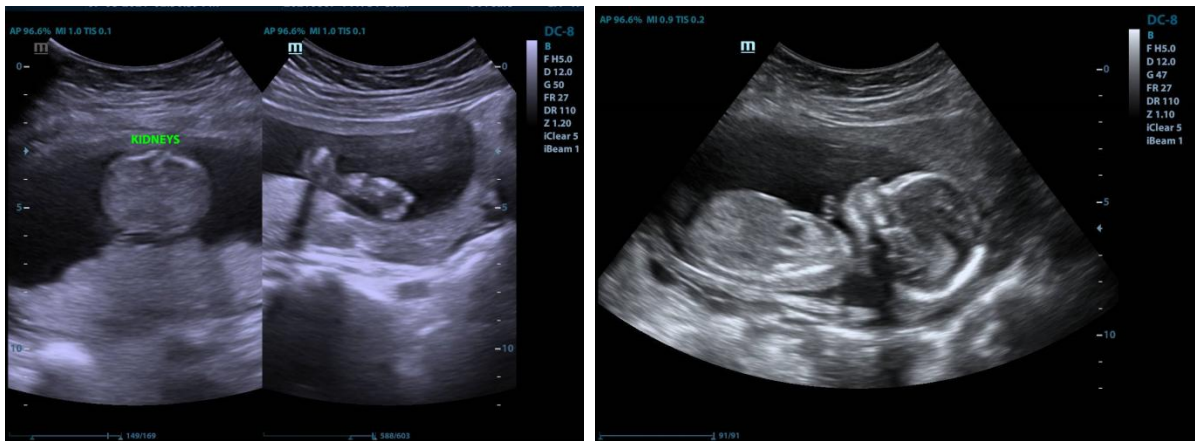
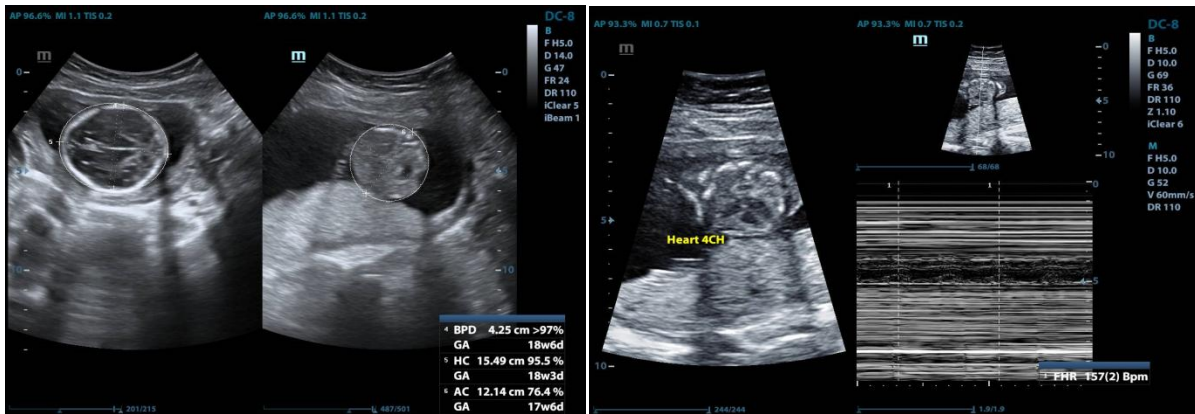
Fig 1. Short long bones in all limbs and normal spine.



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Fig 2. Cranium , face and thoracoabdominal contour.



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Fig.3. Post abortal images. [Pic Curtery : Dr.Sanat Pimpalkhare. MBBS.MD.OBGY]



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