

Objective

To highlight the rare antenatal findings in a case of Trisomy 21

Case report

- ❑ 27-year-old primigravida referred to us at 14 weeks GA in view of biochemical screening showing high risk for Trisomy 21(1: 224)
- ❑ High bhcg -2.76 moms
- ❑ PAPP-A 0.74moms
- ❑ Combined risk showing low risk for Trisomy 21. (1: 1389).

- ❑ Screening anomaly showed normally growing fetus with no anomalies and aneuploidy markers.

- ❑ Non invasive prenatal testing and amniocentesis was discussed but the patient declined both. She was having regular growth scans in view of high hCG.

- ❑ At 30 weeks, scan showed hepatosplenomegaly, echogenic bowel, bilateral echogenic kidneys, cardiomegaly, mild pericardial effusion and Tricuspid regurgitation with Placentomegaly.

- ❑ Amniocentesis was offered again.

- ❑ Maternal TORCH Panel showed no evidence of acute infection.

- ❑ Corticosteroids given for fetal lung maturity

- ❑ At 32 weeks emergency caesarean section was done in view of abnormal middle cerebral artery doppler and delivered a female baby of weight 2.09 kg with Apgar score of 7 at 1’and 8 at 5’ having dysmorphic features.

- ❑ Baby was administered surfactant and was on mechanical ventilation till D4 of life.

- ❑ On evaluation baby had hemoglobin of 9.8g/dl with hepatosplenomegaly, flow cytometry suggestive of abnormal myelopoiesis and tandem mass spectrometry for inborn errors of metabolism was negative.

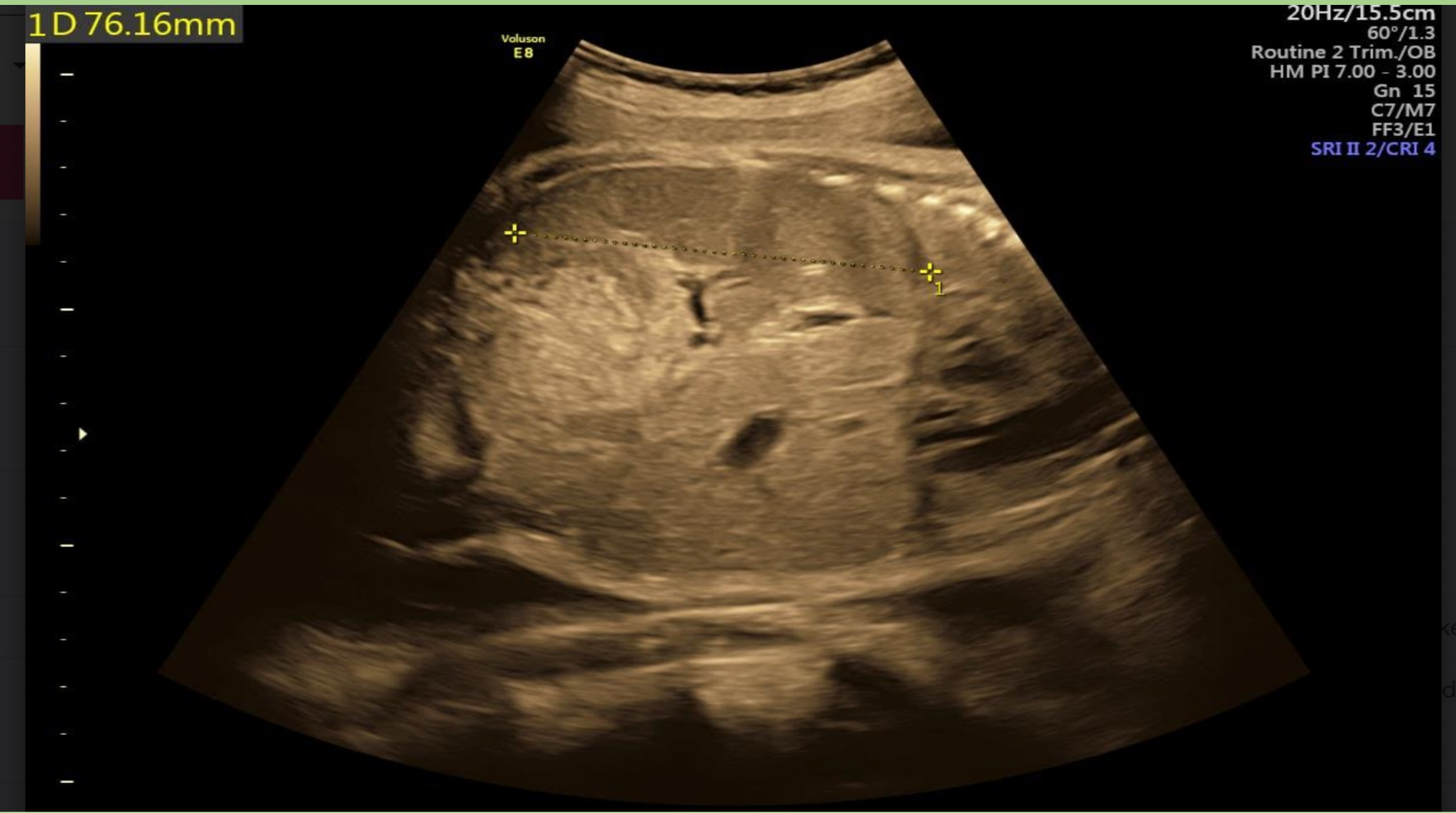
- ❑ Karyotype showed 47 XX, + 21 / 46 XX which was suggestive of Mosaic Down Syndrome.

- ❑ The baby was diagnosed with acute Myeloid leukaemia at 2 months of age.

- ❑ Baby was treated with chemotherapy and is on regular follow up

Discussion

- ❑ Fetal hepatosplenomegaly- HSM can occur in a number of pathologies like Trisomy 21, hydrops, infections, hypothyroidism, storage disorders and over growth syndromes and has to be taken into consideration as a possible marker.
- ❑ Fetal HSM could be an indicator of transient abnormal myelopoiesis(TAM) which occurs in 10% of newborns with Trisomy 21.
- ❑ Fetal hydrops and anemia may also be seen in such fetus.
 - Percutaneous umbilical cord blood sampling is needed to diagnose TAM prenatally- markedly increased leucocytes with blast cells.
- ❑ TAM usually resolves spontaneously within few months or may progress to myeloid leukemias.
- ❑ 13-33% of patients with TAM develop AML within the first 4 years of life.



Conclusion

Conclusions: Antenatal markers of Trisomy 21 are not limited to first and second trimesters alone and third trimester findings like HSM can be an isolated finding .

References

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