CASE OF THE MONTH (JUNE 21)

Theme: Downs detection still a challenge!

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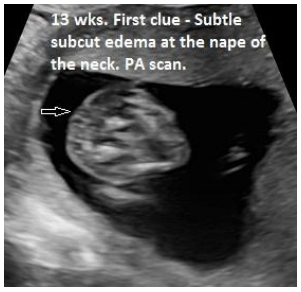
Background : IT'S an eye opener indeed and shows that there is a long way to go !

We try to understand, why the nasal bone measurement, borderline Nuchal Translucency (between 50 – 95 th % centile) and subtle variations in growth and biometry are so crucial in suspicion of underlying chromosomal anomaly in fetus.

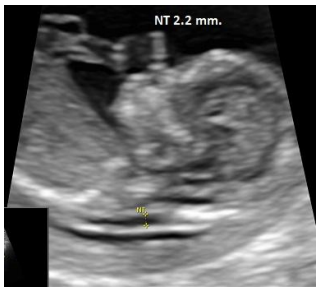
Case History – 36 yr lady, G2P2L1A0 , came for serial antenatal ultrasound examinations as described below. There was no relevant past medical and family history of any obvious illness. It was spontaneous conception. Her dating was accurate and GA assigned as per the LMP.

[Only few relevant representative images are included in document].

13 weeks [NT scan]: It was technically difficult scan due to persistently contracted uterus. Patient was examined with multiple attempts on two consecutive days , with per abdominal and trans vaginal route to get best possible images. Nuchal translucency was slightly prominent on eyeballing with NT measurement 2.2. There was a subtle nuchal edema noted (a) & (b). Nasal bone was short mes. 2 mm which was 1.2 % centile for the gestational age (c). Choroid plexus showed possible tiny cysts (d). There was a trivial tricuspid regurgitation (TR) [(f) , open arrow] , however Ductus venosus (DV) flow was acceptable (e).



(a)



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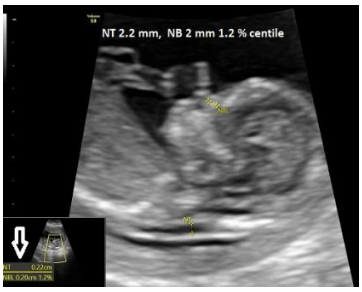
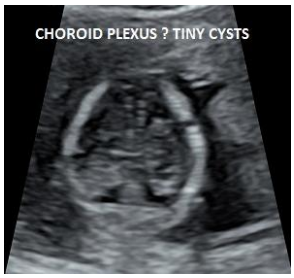
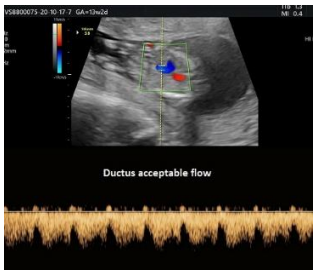


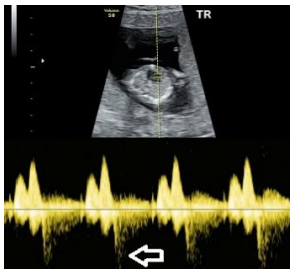
(c)



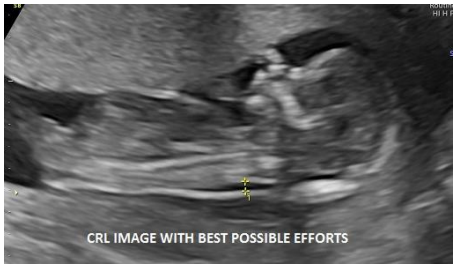
(d)

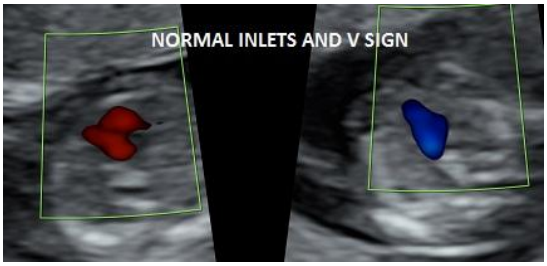


(e)



(f)





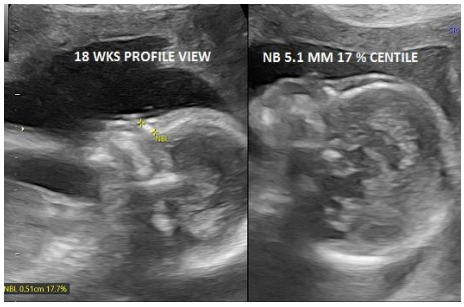
(g) (h)

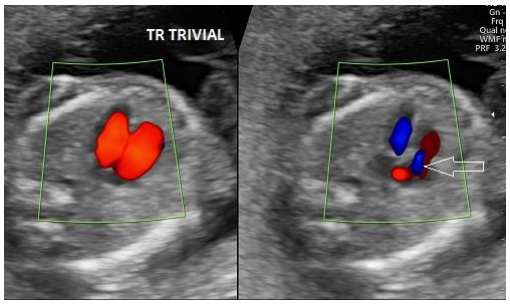
Fig. 1 , NT scan (a - h)

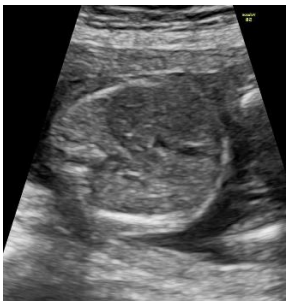
CRL (g) and cardiac evaluation (h) was normal for the gestational age.

In view of subtle findings detailed screening and an early anomaly scan was advised to patient. Due to Some reasons they missed combined screening and second trimester screening test was done which came low risk. Patient came for anomaly scan at around 18 + weeks.

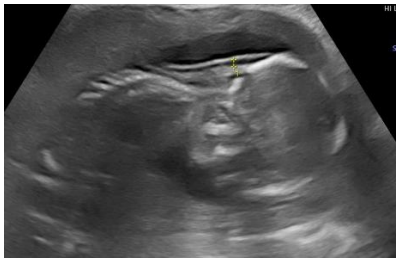
18-19 wks [Anomaly scan] : Growth was corresponding without any major anomaly in fetus. Profile view and nasal bone was relatively normal (a). There was trivial TR but not consistently demonstrated (b). Nuchal fold was relatively normal (d). Stomach bubble was small persistently (c) but showed acceptable distension on second day follow up (f). Head shape showed mild suspicion of brachycephaly (e) but not very convincing.
As there was no major anomaly and second trimester screening came low risk , expectant management and follow up considered .

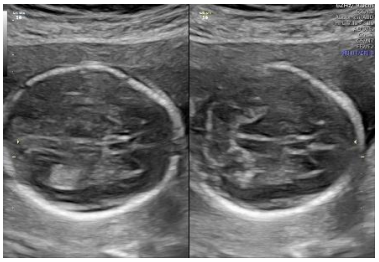


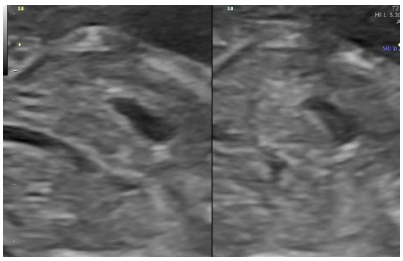




(a) (b) (c)



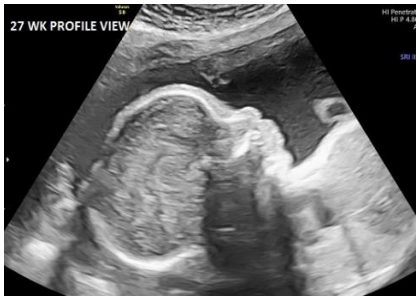


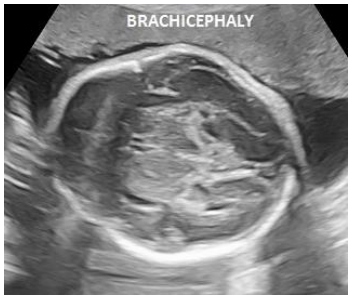


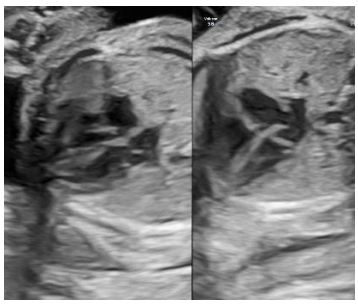
(d) (e) (f)

Fig.2 , Anomaly Scan. (a - f)

27 wks [Growth and cardiac scan] : Average growth was corresponding with expected gestational age ,however head parameters were around 29 weeks (67 % centile) and others parameters around 27 weeks. Long bones , especially proximal bones showed mild shortening (FL 17 % centile). Profile view almost looked normal (a). Cardiac evaluation was normal (c). Head shape revealed slight brachycephaly (b). AFI was 18. MCA PSV showed mild fetal anemia (Barcelona calculators).These subtle findings discussed with patient and treating physician. Detailed counselling about possible outcomes were intimated (either constitutional variation or rarely some underlying fetal disorder).



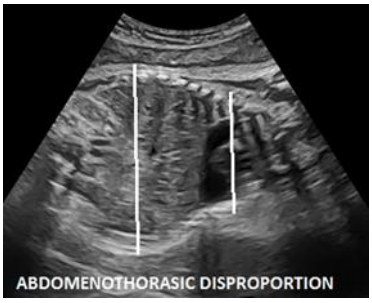


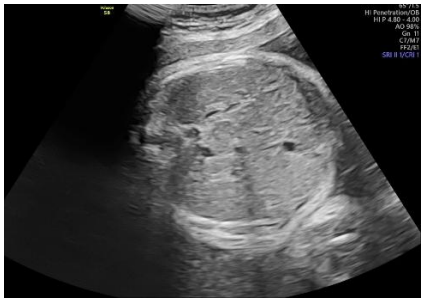


(a) (b) (c)

Fig.3 , Growth scan 27 Wks. (a,b and c).

Growth scan with doppler (34 weeks) : Interval growth showed growth lag, especially with proximal long bones shortening. Humerus less than 5 % centiles and Femur around 6-7 % centiles. There was mild hydroamnios. Doppler parameters were within normal limits. Fetal abdomen was protuberant probably due to hepatomegaly with narrow thorax on coronal view. In view of IUGR with polyhydroamnios and long bones shortening, detailed counselling about delivery management and possible outcomes were discussed with physician.





(a) (b)

Fig.4 , Growth scan 34 wks (a & b)

36 and 38 weeks scans were done outside in tertiary care center : There was a IUGR with abnormal Doppler parameters showing signs of placentofetal insufficiency. LSCS advised but patient insisted for vaginal delivery in spite of risk involved. With due consent and informing possible outcomes vacuum assisted vaginal delivery was conducted at 38 weeks.

Baby delivered with syndromic facies. Metabolic disease profile came negative. Genetic testing was done which came positive for the Downs Syndrome. Needed NICU stay for about 2 weeks.

Discussion & Conclusion : The only convincing finding before 20 th week in this case was a short nasal bone which was out of range i.e. below 5 th % centile. Double marker test was missed due to some reason and delay in doing NT scan. As the second trimester screening test came low risk and no obvious anomaly was detected at 18 weeks option of invasive testing was not considered. IUGR evolved after 28 weeks with proximal long bones shortening, brachycephaly and polyhydroamnios were clues for some underlying disease and discussed with patient and physician too. Possibility of minor constitutional variations and expectant management was also considered in this case. Only strong supporting factor in this case was maternal age which was 36 yrs.

We feel that structurally normal fetus with downs syndrome is still a challenge. However picking up the subtle clues on Ultrasound examination is crucial and consciously considered in counselling. Late onset IUGR , especially with normal liquor and long bones shortening is most of the time is constitutional however in presence of first trimester soft markers this can be an important clue for the suspicion of downs or any other syndromic condition in fetus. By following systematic screening protocols and detailed ultrasound examination in all trimesters we can improve the detection rates of genetic diseases and syndromic conditions in fetal life. Along with the systematic first trimester screening , NIPT and definitive tests like amniocentesis for karyotyping , FISH and microarray should be seriously offered to the women with high background risk especially in presence of ultrasound soft markers.

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