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CASE REPORTS

Importance of Anomaly Scan in 1st Trimester: Two Case Reports

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Abstract:
Fetal anomaly is scanned at the time of nuchal translucency(NT) assessment at 11 weeks to 13 weeks 6 days. Increased nuchal translucency is the strongest single marker for chromosomal abnormality. Here, we report two cases observed in 1st trimester, scanned by both TAS and TVS techniques. 1st case is about aneuploidy screening while the 2nd one is multiple congenital anomalies in hypothyroid mother. Early detection of anomaly improves the outcome of pregnancy which is beneficial for the parents.

Key words: Fetal anomaly scan, Nuchal translucency, chromosomal abnormality.

Introduction:

Nuchal translucency is sonographic appearance of fluid collection under skin behind fetal neck in 1st trimester of pregnancy. The term translucency is used irrespective of whether it is septated or not and whether it is confined to the neck or envelopes the whole fetus. Fetus with chromosomal abnormalities, cardiac defects and many genetic syndromes the NT is increased.[1] Fluid collects behind the fetal neck, much like it does in dependent ankle edema. This occurs partly because of the fetus's tendency to lie on its back and partly because of the laxity of the skin of the neck. The more fluid that has accumulated, the greater the risk of abnormality being present.[2] Screening NT can detect about 80% of fetuses with trisomy 21 and other major aneuploidies for a false positive rate of 5%. Nicolaides et al. shows that the combination of NT and maternal serum free â-hCG and PAPP-A improves the detection rate to 90%.[3] There is now evidence that the detection rate can increase to about 95% and the false positive rate can be reduced to 3% by also examining the nasal bone, ductus venosus flow and tricuspid flow.[1]

Other anomalies detected in 1st trimester include Major CNS anomalies (acrania - exencephaly-anencephaly), holoprosen-cephaly, cystic hygroma, hydrops, abdominal wall defect, distended bladder, posterior urethral valve in male fetus, urethral atresia in female fetus, abnormal ductus venosus blood flow, tricuspid regurgitation, skeletal defects and limb defects.[4]

Case Report I:

A 34 years old woman (gravida 4, para 3), age of last child 6 years, came for ultrasound with history of 3-month amenorrhea on 9 September.

Her USG scanning shows CRL measurement = 3.6 cm at 10W 4D, NT measurement 3.4 mm (Fig-1). She was advised for a follow-up scan which was done on 12 September (Table I). She was positive for Trisomy 21 screening in biochemical marker (Table II). The next follow up date was on 26 September.

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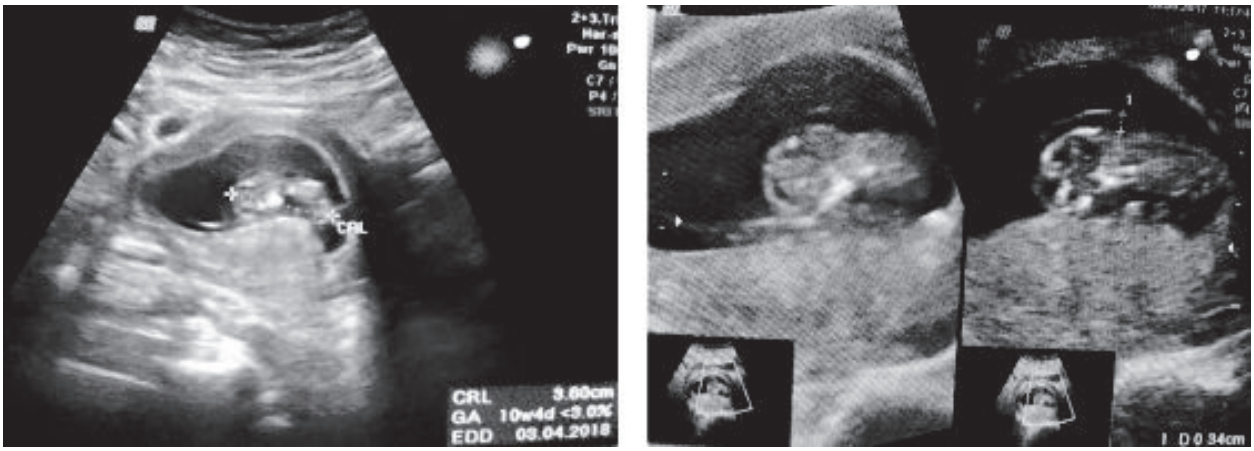

Fig 1: *USG on 1st visit (9 Sep)*

Table-I
USG data and findings

Date	Fetal parameters	NT (In mm)	GA
9 Sep	CRL 3.6 cm	3.4	10W+
12 Sep	CRL 4.2 cm	3.6	11W
26 Sep	BPD 1.75 cm	Subcutaneous translucency envelopes whole fetus.	12W+

Subcutaneous translucency is increased and extending from head to feet, enveloping whole fetus. BPD was 1.75 cm at GA 12 W 5 D. Eventually abdominal wall defect along with hernia was found (Fig- 2).

Table-II
Biochemical report

Test	Value Observed	Normal Range (10-12 Weeks)	Level
Free β-hCG	17.9 ng/mL	Up to 12.7 ng/mL	Raised
PAPP-A	0.48 Corr MoM	≥ 0.5 Corr MoM	Lowered
NT	3.4-3.6 mm	< 3 mm	Raised

Value of these markers in Table-II along with gestational age estimate the likelihood of pregnancy with Down's syndrome.

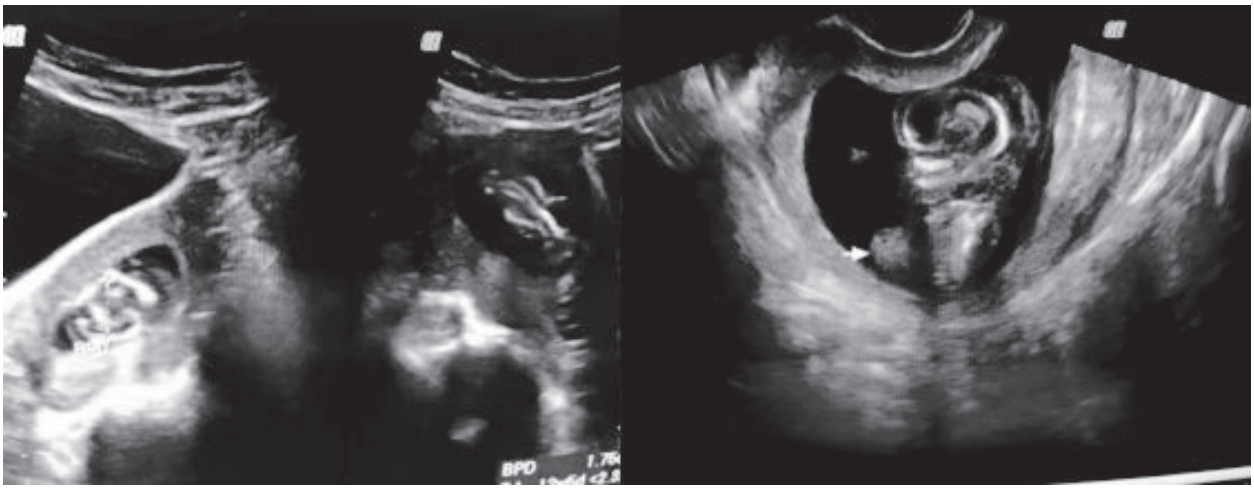

Fig 2 : *USG on 2nd Visit (29 Sep) (Case Report 1)*

Case report 2: A 43-year-old women (gravida 2, para 1) hypothyroid patient who was not taking her medications regularly for last 6 months came to BIRDEM OPD to do lower abdominal ultrasound due to irregular menstrual history, she was unaware of her pregnancy. She and her husband were non-consanguineous and there was no family history of congenital malformations. The woman did not have diabetes mellitus.

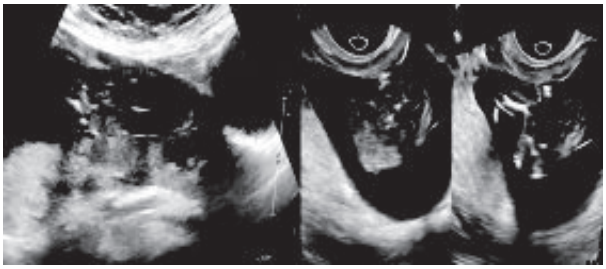

Fig.-3 : *(Left) TAS, (Right) TVS (Case Report 2)*

On lower abdominal ultrasound an early pregnancy was seen with FL about 8.2 mm corresponding to 12 weeks 04 days of gestation, FL was well outlined which indicated that the foetus was more than 12 weeks but cranial vault could not be well outlined, so TVS was done in the same setting for further evaluation and multiple structural congenital anomaly was seen. On TVS, fetal skull vault was absent indicating anencephaly (Fig-3). Defect was seen in lower chest and upper abdomen of foetus leading to external position of heart (ectopia cordis) and liver (Fig-4).

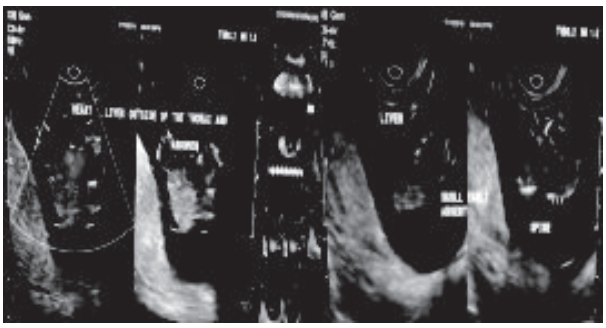

Fig.-4: *Ectopia cordis and herniated liver(Left) & Spinal defect revealed in TVS (Right).*

Gastric bubble was absent indicating tracheo-oesophageal fistula, single lower limb was seen which has significant association with maternal hypothyroidism (Fig-4). The parents elected to terminate the pregnancy and a malformed fetus was delivered with anencephaly, ectopia cordis, defective upper anterior anterior abdominal wall through which liver is herniationg outside, absent left lower limb, and an imperforate anus (Fig-5).

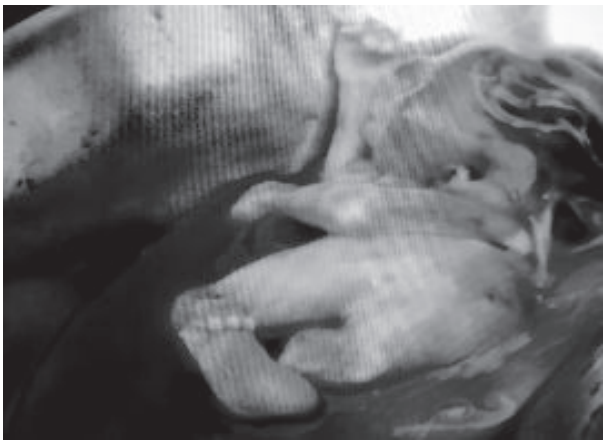

Fig 5: *Malformed fetus*

Discussion:

Combination of NT and maternal serum free β-hCG and PAPP-A improve detection of trisomy 21 and other aneuploidy up to 90%. Usually, maternal serum PAPP-A increases and β-hCG decreases with gestation. In 1st case report, initial ultrasound findings show nuchal translucency is increased (>3mm). Follow up scan shows further increased (3.6mm from 3.4mm). At that time biochemical markers PAPP-A is low and & free β-hCG is raised. This indicates fetus is positive for trisomy 21 screening. In follow up scan, subcutaneous translucency is increased and extending from head to feet and envelopes whole fetus. Obstetrician counselled the patient and terminated pregnancy. Several studies from Markov et al., Abu-Rustum et al. show maternal age has relation with increased NT.[5,6] Brizot et al[9] found that an improved estimate of risk for fetal trisomies at 10-13 weeks' gestation can be derived by combining data on maternal age, maternal serum or free β-HCG and fetal nuchal translucency thickness.[7]

In 2nd case report we found multiple fetal anomalies in early pregnancy scanning at 11-14 weeks. Detection rates are better after 13 weeks and improved with the use of transvaginal scanning. However, caution is required and we should be aware of the potential pitfalls which are related to embryological development. The

detection rate is better after 13 weeks and improved with use of TVS. Skull ossification begins at 10 weeks and is complete by 12 weeks, The lateral ventricles occupy most of the cerebral hemisphere and within each the choroid plexus is very prominent. This is not hydrocephalus. Before this, at 7-9 weeks, the forebrain appears as a single ventricle. Midgut herniation into the umbilical cord is physiological between 8-12 weeks. This should not be mistaken for exomphalos which is more likely when the mean diameter of the herniated sac exceeds 7 mm and the CRL is more than 68 mm.

Kashyap et al found in their study almost half of malformations were amenable to be diagnosed in 1st trimester.[8] Their study revealed fetuses having malformations like neural tube defect, anencephaly, holoprosencephaly, gastroschisis. Castro-Aragon and Levine reported that 60-70% of malformations could have been diagnosed prior to 12 weeks. Fong et al[10] in their study scanned 8,537 women between 11 and 14 weeks' gestation (CRL 45-84 mm); there were 175 fetuses with an increased NT. Besides nuchal abnormalities, a wide range of other congenital anomalies can be diagnosed with US at 11–14 weeks of gestation, including defects of the central nervous system, heart, anterior abdominal wall, urinary tract, and skeleton.

Oztekin et al. analyzed 1085 pregnancies; 21 (1.93%) fetuses had at least one major structural defect considered detectable by routine ultrasound screening.[11] 14 (1.29%) were identified at early (first trimester) screening and an additional 5 (0.47%) were identified at late (second trimester) USG. They found that majority of fetal structural abnormalities can be detected by sonographic screening at 11–14 weeks, but detailed fetal anatomic survey performed at 18–22 weeks should not be abandoned.

Rossi and Prefumo also laid stress that first trimester ultrasound can detect half of fetal malformations. They included nineteen studies on 78,002 fetuses, with 996 with malformations that were confirmed by postnatal or postmortem examinations. USG at 11 to 14 weeks detected malformation in 472 of the malformed fetuses (51%). Detection rate was highest for neck anomalies (92%) and lowest for limbs, face, and genitourinary anomalies (34% for each).[12]

Early ultrasound might be more accurate than second trimester ultrasonography for detection of malformations associated with oligohydramnios and anhydramnios which lead to poor visualization at later gestation necessitating amnioinfusion. However, accurate detection requires awareness and expertise along with proper instruments.

Conclusion:

These two case report emphasizes the importance of the first trimester scan in early detection of aneuploidy and structural fetal anomaly. TVS should be incorporated as routine first trimester ultrasound scan as it helps to detect some defects which can easily be missed in per abdominal ultrasound. When diagnosis before viability is possible, management options can be discussed with the couple; including medical termination of pregnancy. Early detection allows a safer termination of pregnancy, if this option is considered. Even when the decision to continue the pregnancy is taken, prenatal diagnosis is very important to improve outcome.

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