

Validity of Sonologic Soft Marker for Chromosome Abnormality

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Abstract- A study was conducted to evaluate the validity of sonologic soft marker for chromosome abnormality. There were 200 subjects referred from Obstetrics and Gynaecology OPD of SM CSI Medical College, Karakonam, Trivandrum for a period of 2 years. The study was done using Siemens sonolin 50 USS scanner. The different sonologic soft markers used for the evaluation were nuchal fold thickness, short long bones, mild pyelectasis, echogenic bowel, echogenic intra cardiac focus and choroid plexus cyst. The karyotype analysis was carried out in antenatal patients showing isolate and multiple sonologic soft markers. The study observed that there is no isolated soft marker is found to have higher risk for chromosome abnormality whereas cluster of markers seems to confer higher risk for aneuploidy. Thus sonologic soft markers can be used as a good tool along with cytogenetic markers to reduce the risk of chromosomal abnormalities during prenatal period.

Index Terms- Aneuploidy, Antenatal USS, Chromosome abnormality, High risk pregnancy, Karyotype, Sonologic soft marker

1 Introduction

Chromosomal abnormalities are one of the leading causes of pregnancy loss. Chromosome abnormalities occur in 0.1-0.2% of all live births [1]. Trisomy 21 is the most common karyotypic abnormality in live born infants [2]. Sonographic finding in fetuses with Down syndrome include both structural and non-structural abnormalities. Second trimester ultrasound detects two types of sonographic findings suggestive of aneuploidy. Detection of major fetal structural anomalies comprises the first group. The second group includes soft markers that are non specific often transient and can be detected during 2nd trimester ultrasound scan [3]. The most commonly studied soft markers of aneuploidy included nuchal fold thickness, rhizomelic limb shortening, mild pyelectasis, echogenic bowel, echogenic intra cardiac focus and choroid plexus cyst. Unfortunately the studies evaluating significance of soft markers for aneuploidies vary widely and show contradictory results. The present study reviews the validity of sonographic markers for chromosome abnormalities in study population to identify the best marker for detecting at risk pregnancy for chromosome abnormality.

2 Materials and Methods

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The study was conducted on 200 subjects referred from Obstetrics and Gynaecology OPD of SM CSI Medical College, Karakonam, Trivandrum for a period of 2 years.

Inclusion criteria:

1. Pregnant subjects below 35 years of age.
2. Subjects without any added risk for chromosome abnormality.

The study was done using Siemens sonolin 50 USS scanner. The following soft markers were evaluated during 18-20wks scan.

Nuchal fold thickness

The measurement is made in transverse plane of fetal head slightly off the biparietal diameter which includes cerebellum, occipital bone and cavum septum pellucidum. The Nuchal fold was measured with placement of calipers from outer edge of occipital bone to outer edge of skin [4]. 5mm was used as single cut off.

Echogenic bowel

Fetal echogenic bowel refers to presence of hypoechoic bowel as compared with echogenicity of adjacent iliac bone. Once echogenic bowel was suspected the gain of USS unit was lowered gradually until only bone or bowel was visible.

Short long bones

Shortened humerus or femur is identified by comparing actual measurement with expected measurement on the basis of biparietal diameter. The femur is considered shortened when measured to expected ratio that is < 0.89 [5].

Echogenic Intracardiac focus

Echogenic Intracardiac focus is seen as discrete areas of echogenicity compared to bone in the region of papillary muscle. The foci were visualized from different angles to make sure that one does not include specular reflections of papillary muscle.

Choroid plexus cyst

Looked for in axial plane of head within lateral ventricle.

Mild pyelectasis

Mild pyelectasis is diagnosed when renal pelvis measures > 4 mm and

< 10 mm in AP dimension in axial scan of abdomen without caliceal dilatation.

Karyotyping

Karyotyping was done in case of subjects with positive soft markers for chromosome abnormality using the blood collected by cordocentesis of foetus or by cord blood of baby. Karyotyping was done by lymphocyte culture method at Ittyavirah Scan and Genetic Research.

3 Observations and Results

Of the 200 subjects studied the commonest sonologic marker observed was pyelectasis ($n=6$) followed by choroid plexus cyst ($n=5$).

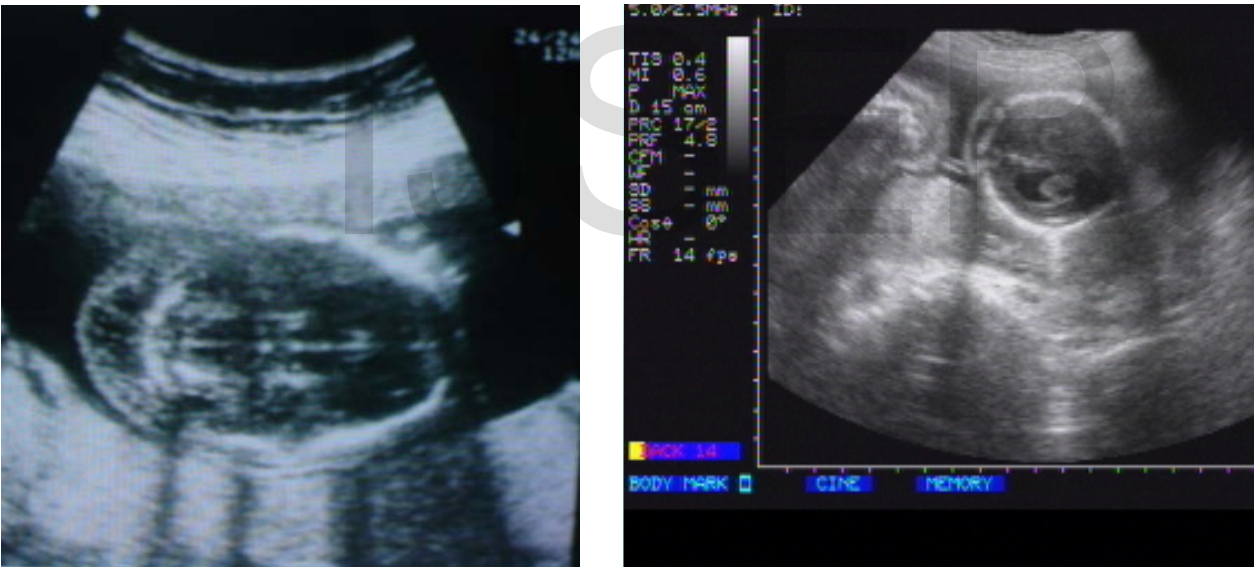




FIG. 3 (a &b)
Minor pyelectasis

Sl.	Soft marker	Number	Percentage
1	Nuchal thickness	2	1
2	Choroid plexus cyst	5	2.5
3	Echogenic bowel	1	0.5
4	Pyelectasis	6	3
5	Short long bone	1	0.5
6	Echogenic Intracardiac focus	4	2
	Total	19	9.5

TABLE 1:
Sonographic markers observed in 200 antenatal patients

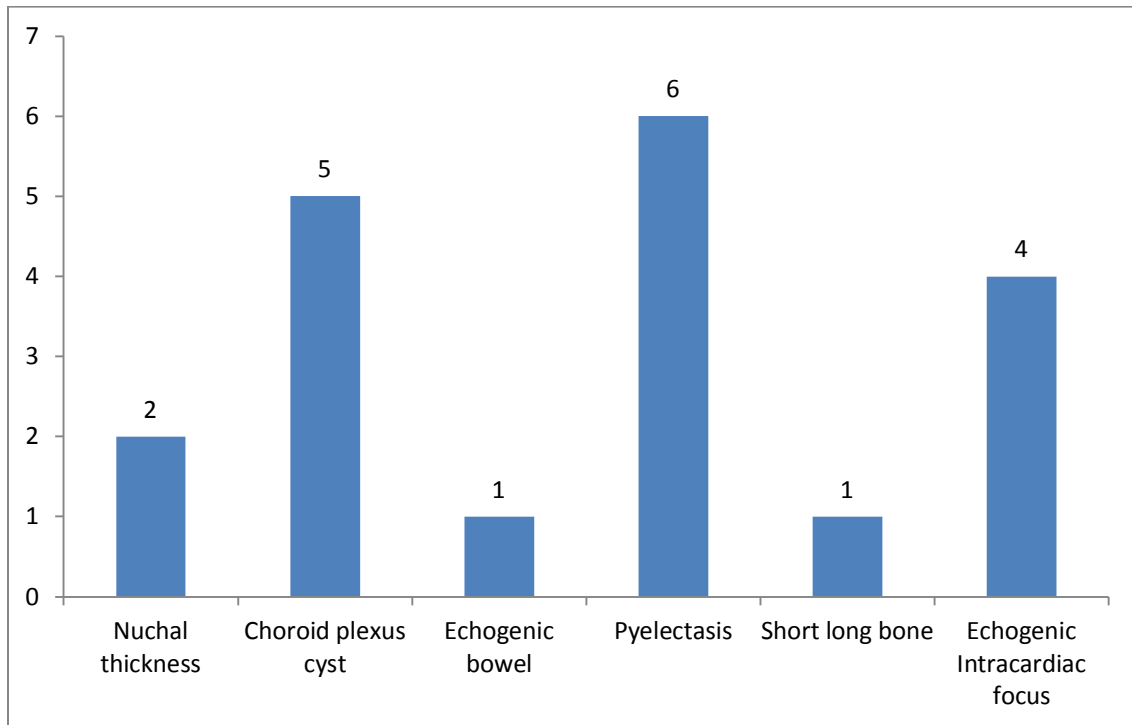


FIG. 4
 Sonographic markers observed in 200 antenatal patients

	Soft markers	Number	Abnormal karyotype
1	Choroid plexus cyst and Nuchal thickness	6	0
2	Echogenic bowel and short long bone	1	1
3	Choroid plexus cyst with echogenic bowel	2	1
4	Pyelectasis and NT	4	0
	Total	13	2

TABLE 2:

Multiple sonographic markers in 200 antenatal patients and their correlation with chromosome abnormality

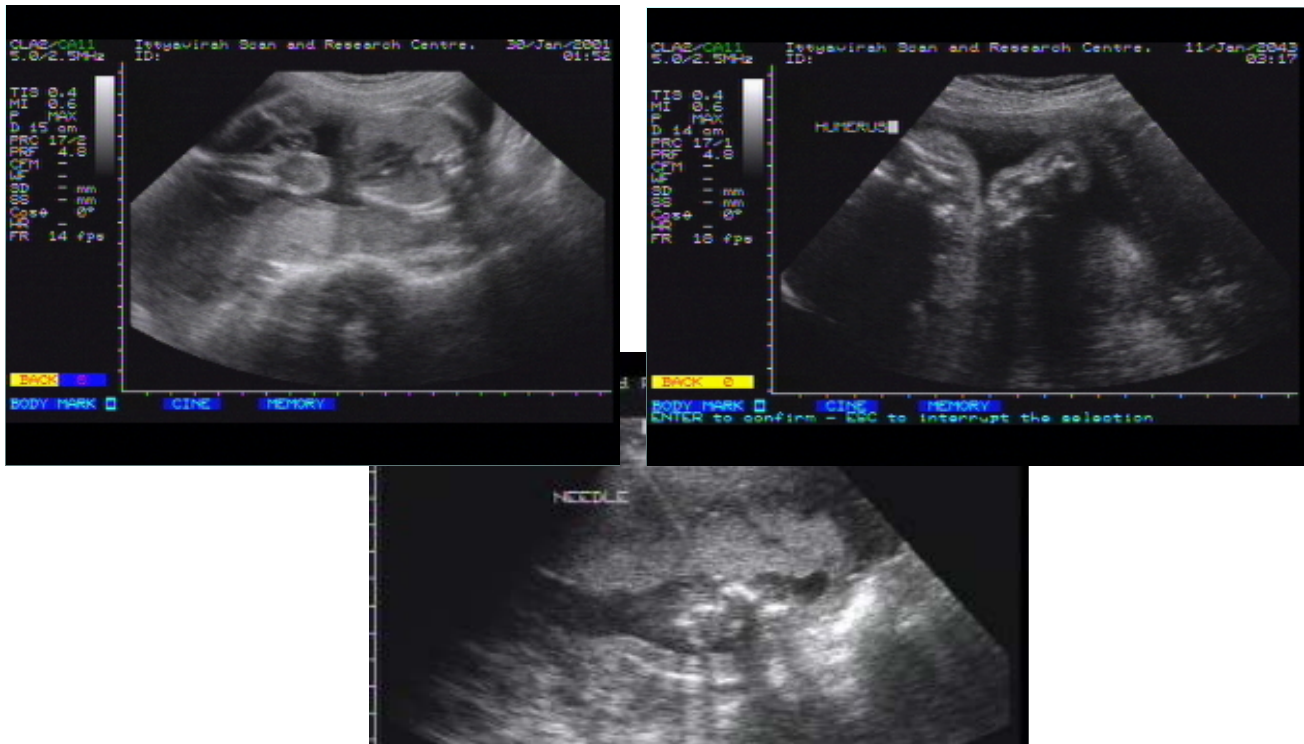


FIG. 7

Cordocentesis under ultrasound guidance

Out of the 200 subjects included in the present study 19 subjects had solitary sonographic soft marker for chromosome abnormality. The commonest marker observed was pyelectasis followed by choroid plexus cyst. None of the cases with solitary soft marker had chromosome abnormality detected by karyotyping. Among the 200 subjects 9 subjects had multiple soft markers; out of these, 2 fetuses had chromosome abnormality. One was Down syndrome and the other was Turner's syndrome.

5 Discussion

In the present study out of 200 subjects, 19 antenatal cases showed isolated soft marker for chromosome abnormality and none of these fetuses had chromosome abnormality on karyotyping. Nyberg DA et al reported that isolated soft marker was the only sonographic finding in 42 (22.6%) of 166 fetuses with trisomy. When compared with 11% of control population [6], in this study the percentage of isolated marker in subjects is 9.5%. According to Bomle B et al [7] echogenic intracardiac focus was the single most common isolated marker in both affected (7.1%) and control fetus (3.9%). In this study multiple marker was noted in 13 antenatal cases which included 6 cases of choroid plexus cyst with NT, 4 cases of

pyelectasis with NT, 2 cases of choroid plexus cyst with echogenic bowel and one case of echogenic bowel with short long bone. Out of the 9 cases with multiple markers, 2 fetuses had chromosome abnormality. Out of the 19 subjects those showed solitary marker none of them had chromosome abnormality which suggesting that cluster of markers seen to confer higher risk of aneuploidy than solitary marker. Similar conclusions were made by Sohi B et al [8]. According to Sameer Ramga et al, Nuchal fold thickening, short humerous, even as isolated findings, confirm high risk of aneuploidy [9].

6 Conclusion

Out of 200 subjects included in the study 19 had solitary soft marker for chromosome abnormality and none of them had chromosome abnormality. Out of the 9 subjects with multiple soft markers, 2 fetuses had chromosome abnormality. There is no isolated soft marker is found to have higher risk for chromosome abnormality whereas cluster of markers seems to confer higher risk for aneuploidy.

7 References

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