Fetal Soft Markers in Obstetric Ultrasound

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Abstract

Background: The use of ultrasound in pregnancy has significant health and economic outcomes for families and the health care system. With high resolution ultrasound, it is possible to examine fetal anatomy in great detail. The quality of images obtained today is such that minor deviations from normal can be clearly identified.

Aim of Study: The purpose of this prospective study is the observation after detection of ultrasound soft markers, and to evaluate the usefulness of each ultrasound soft marker, and assess whether a specific soft marker should be looked for routinely on screening ultrasound.

Patients and Methods: This was a prospective studyin 270 pregnant women at 16-24 weeks of gestation. All women were examined twice with ultrasonography for detection of any tissue abnormalities. First at 16-24 weeks of pregnancy and repeated for follow-up of the soft marker once detected at 32-36 weeks of pregnancy.

Results: This study has demonstrated that when a soft marker is identified, there must be a careful search for other markers. The study showed a total of 27 (10%) of the studied women had tissue anomalies; 25 (9.3%) women had isolated soft tissue anomalies while 2 (0.7%) women had mixed anomalies. Three markers were not found in any woman, namely, increased nuchal fold thickness, absent nasal bone and ventriculomegaly. While the most frequent tissue anomalies were pyelectasis (3.7%) and choroid plexus cyst (2.2%). Another finiding in our study was echogenic bowel (EB) by 1.5%, EIF and shortened long bone by 1.10% and 0.75%, respectively; and the outcome of the new born are normal. There are some ultrasound findings commonly seen on second trimester routine scan that are associated with possible progression (e.g. renal pelvis dilatation). These findings should be reported as anatomical variants and appropriate follow-up arranged. For renal pelvis dilatation, then it is common practice to re-scan in the third trimester to see if there is progression.

Conclusion: Soft markers in second-trimester ultrasonography have limited use in screening for fetal aneuploidy. However, these markers can be used as a screening tool for adverse outcomes other than chromosomal abnormality.

Key Words: Fetal soft markers – Obstetric ultrasound.

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Introduction

ULTRASONOGRAPHY is a common component of prenatal care worldwide and is often used in early pregnancy to determine gestational age, number of fetuses, fetal cardiac activity, and placental location. Ultrasound screening is typically scheduled in the second trimester to visualize fetal anatomy and confirm gestational age. Most ultrasound examinations are reassuring, but some incidentally identify structural anomalies and soft markers for aneuploidy, making it necessary for health care providers to correctly interpret these findings [1].

When ultrasonographic soft markers (USMs) are detected in second trimester, the finding can raise questions about prenatal diagnostic testing procedures. As technologies evolve, various methods are currently implemented in prenatal genetic testing, including non-invasive prenatal testing (NIPT) and invasive testing using karyotyping or chromosomal microarray (CMA) [2].

As compared to invasive testing, NIPT using cell-free fetal DNA (cffDNA) from maternal plasma is non-invasive and more acceptable by women. In contrast, invasive prenatal testing is performed by chorionic villi sampling, amniocentesis, or cord blood sampling, which is associated with an increased risk of miscarriage and other side-effects [2].

Fetal Soft markers are incidental ultrasound findings of uncertain clinical significance. Soft markers found during second-trimester ultrasound imaging can include nuchal thickening, echogenic cardiac focus or foci, echogenic bowel, pyelectasis, choroid plexus cysts, shortened femur or humerus, absent nasal bone, and single umbilical artery [1]. They are usually normal variants, have no clinical sequelae, and are transient, resolving with advancing gestation or after birth [3].

These markers may, however, be associated with an increased risk for fetal aneuploidy, and individual risk assessment needs to be done. The risk for aneuploidy is higher whenever more than one marker is found or if markers are associated with single or multiple structural findings [4]. The actual magnitude of the increase is dependent upon the specific markers found. For example, echogenic bowel has been associated with blood in the bowel lumen, cystic fibrosis, growth restriction, infection, and gastrointestinal obstruction [5].

Therefore, researchers have long investigated the impact of isolated and multiple soft markers on the risk assessment of the aneuploidies and the invasive procedures [6].

The purpose of this prospective study is the observation after detection of ultrasound soft markers, and to evaluate the usefulness of each ultrasound soft marker, and assess whether a specific soft marker should be looked for routinely on screening ultrasound.

Patients and Methods

Study design: This is a cross sectional observational prospective study that was performed in 298 pregnant women in the period from October 2020 to December 2021.

Study approval: This study was approved by the Ethical Committee at Faculty of Medicine, Al-Azhar University. Privacy and confidentiality of all data were assured. 298 Pregnant women were enrolled in the study, the nature of the study was explained to each participant.

Setting: The cases were selected from Outpatient Clinic of Obstetrics and Gynecology Department, Assuit Police Hospital. Ultrasound examination was performed at period form 16-24 weeks of gestation. All soft markers which were detected in the mid trimesteric scan were confirmed by second ultrasound at 32-36 weeks and documented after delivery.

Participants: 298 pregnant women included in the study have the following selection criteria.

Inclusion criteria: Any pregnant patient between (16-24) weeks for the mid-trimestric scan, and singleton pregnancy.

Exclusion criteria: Patient with known or discovered fetal congenital anomalies during scanning, and multiple pregnancies.

Study plan:

Each pregnant woman was scheduled for two ultrasound examinations:

1- Each pregnant women was subjected to: History taking. (Risk factors) including: Age, consanguinity, previous history of congenital anomalies.

2- First trans-abdominal ultrasound was done at the time (16-24) weeks.

3- A second examination was scheduled at 32-36 weeks for confirmation of the presence or absence of the soft marker.

4- Fetal anomaly scan was done.

5- Patients were followed-up till delivery and the new born was examined by a neonatologist carefully to confirm the final diagnosis.

Procedure:

270 cases completed the study for the mid trimetric scan and for the follow-up of the soft marker with second ultrasound at 32-36 weeks as well the outcome of pregnancy were evaluated.

Mid trimestric scan examination consisted of:

(1) Estimation of the gestational age by femur length, biparietal diameter, abdominal circumference. (2) Fetal anatomy scans against a check list. This also included the degree of visibility of the target organ it is considered complete or incomplete according to the ability to visualize the following criteria.

For fetal normality, the following are assessed:

(1) Head shape and internal structures. (2) Cavum pellucidum, cerebellum, ventricular size at the atrium (<10mm). (3) Spine: Longitudinal and transverse. (4) Abdominal shape and content at the level of the stomach. (5) Abdominal shape and content at the level of the kidneys and umbilicus. (6) Renal pelvis (<5mm anteroposterior measurement). (7) Longitudinal axis abdominothoracic appearance (diaphragm/bladder). (8) Thorax at the level of the four-chamber cardiac view. (9) Arms, three bones and the hand (not counting the fingers). (10) Legs, three bones and the foot (not counting the toes).

Fetal soft markers includes:

(1) Choroid plexus cyst. (2) Mild ventriculomegaly. (3) Enlarged cistern magna. (4) Nasal bone hypoplasia. (5) Increased nuchal fold. (6) Echogenic cardiac focus. (7) Single umbilical artery. (8) Pyelectasis. (9) Echogenic bowel. (10) Short femur. (11) Short humerus.

Ultrasound criteria of each:

Choroid plexus cyst:

As the choroid plexus develops, the covering epithelium changes and choroidal villi are formed. The spaces between these projections (villi) may be enlarged by fluid or debris to create choroid plexus cysts. Seen sonographically as discrete fluid filled small cysts (≥3mm) in the choroid plexus within the lateral cerebral ventricles.

Mild ventriculomegaly:

Mild ventriculomegaly is defined as an axial diameter of greater than 9.9mm, measured across the atrium of the posterior or anterior horn of the lateral ventricles at any gestation. Sometimes, it is described as a separation of more than 3mm of the choroid plexus from the medial wall of the lateral ventricle. Ventriculomegaly is distinguished from hydrocephalus where there is an atrial diameter of greater than 15mm. Hydrocephalus is not considered here.

Enlarged cistern magna:

The cisterna magna is a fluid collection posterior to the cerebellum. It is seen as an echo-free triangle with the point oriented towards the cerebellar vermis. Prenatally, the anterior/posterior diameter should be <10mm, with a normal appearing vermis, and without hydrocephalus. The cisterna magna is measured on a transaxial view of the fetal head angled 15 degrees caudal to the canthomeatal line. The anterior/posterior diameter is taken between the inferior/posterior surface of the vemis of the cerebellum to the inner surface of the cranium. An enlarged cisternal magna is defined by an anterior/posterior diameter ≥10mm.

Nasal bone:

The fetal nasal bone can be visualized by sonography throughout pregnancy. This examination requires that the image be magnified so that only the head and the upper thorax are included in the screen. A mid-sagittal view of the fetal profile is obtained with the ultrasound transducer held parallel to the longitudinal axis of the nasal bone. In the correct view, there are 3 distinct lines. The first 2 lines, which are proximal to the forehead, are horizontal and parallel to each other, resembling an equal sign (=). The top line represents the skin and the bottom line, which is thicker and more echogenic than the overlying skin, represents the nasal bone. A third line, which is almost in continuity with the skin, but at a higher level, represents the tip of the nose.

Nuchal fold thickness (6mm or over):

It is the skin thickness in the posterior aspect of the fetal neck. It should be measured between 15-20 weeks of gestation. The magnification should be as large as possible, and only the fetal head and upper thorax should be included in the image. A good sagittal section of the fetus in the neutral position should be obtained, and the maximum thickness of the subcutaneous translucency between the skin and the soft tissue overlying the cervical spine should be measured. During the scan, more than one measurement must be taken, and the maximum one should be used for the risk assessment. Good measurement should not be averaged with two bad ones.

Echogenic cardiac focus:

Intracardiac echogenic foci (ICEF) are small areas of increased echogenicity located in the vicinity of the papillary muscles or chordae tendinae inside the fetal ventricles and moving synchronously with the cardiac valves.

Single umbilical artery:

Single umbilical artery (SUA) is the absence of one of the arteries surrounding the fetal bladder and in the fetal umbilical cord. Assessment of the umbilical arteries can be made from the cord itself in either transverse or longitudinal sections. The umbilical arteries can also be assessed at the cord insertion site into the fetal abdomen and on either side of the fetal bladder as the vessels originate from the iliac arteries. If needed, the assessment can be enhanced with colour flow Doppler.

Pyelectasis:

Pyelectasis which is a dilatation of the renal pelvis anteroposterior diameter greater or equal to 4mm in fetuses between 15 and 20 weeks; an anteroposterior renal pelvis diameter greater than or equal to 5mm in fetuses between 20 and 30 weeks; and a renal pelvis anteroposterior diameter greater or equal to 7mm in fetuses between 30 and 40 weeks. Other authors used a cutoff of 4mm to define pyelectasis.

Echogenic bowel:

Echogenic bowel is defined as fetal bowel with homogenous areas of echogenicity that are equal to or greater than that of surrounding bone.

Short femur:

A short femur length is defined as either a measurement below the 2.5th percentile for gestational age or a measurement that is less than

0.9 of that predicted by the measured biparietal diameter.

Short humerus:

A short humerus length is defined as either a measurement below the 2.5th percentile for gestational age or a measurement that is less than 0.9 multiples of that predicted by the measured BPD. The relationship between bone length and head size may differ across racial groups.

Statistical analysis:

Data was collected and analyzed by using SPSS (Statistical Package for the Social Science, version 20, IBM, and Armonk, New York). Quantitative data with were expressed as mean and standard deviation and were compared by Student *t*-test. Nominal data expressed frequency (percentage) and Chi² test was implemented for such data. Level of confidence was kept at 95% and hence, *p*-value was considered significant if <0.05.

Results

Baseline data among studied group (n=270): Mean age of studied women was 31.86±5.05 years with range between 21 and 39 years. Majority (57%) of studied women was between 25-35 years old while 42 (15.6%) and 74 (27.4%) women were <25 and >35 years old, respectively. Out of the studied group; 199 (73.7%) women came from rural areas and 71 (26.3%) women came from urban areas. Gravidity ranged between twice and 10 times while parity ranged between once and 9 times. It was found that 63 (23.3%) women had positive history of consanguinity while history of previous fetal anomalies was present in 18 (6.7%) women (10 cases had history of cleft lip, five cases had history of atrial septal defect and three cases had history of inguinal hernia). All women were subjected to ultrasound examination for the first time between 16 and 24 weeks for detection of soft marker. Three markers were not found in any woman, namely, increased nuchal fold thickness, absent nasal bone and ventriculomegaly (Table 1).

A total of 27 (10%) of the studied women had tissue anomalies; 25 (9.3%) women had isolated soft tissue anomalies while 2 (0.7%) women had mixed anomalies. The most frequent tissue anomalies were pyelectasis (3.7%) and choroid plexus cyst (2.2%). Three (1.10%) women had echogenic foci in the heart. It was found that echogenic bowel and shortened lone bone were present in 4 (1.5%) and 2 (0.75%) women, respectively (Table 2).

Table (1): Baseline data among studied group.

	N=270
Age (years):	31.86±5.05
Range	21-39
Age groups:	
<25 year	42 (15.6%)
25-35 year	154 (57%)
>35 year	74 (27.4%)
Residence:	
Rural	199 (73.7%)
Urban	71 (26.3%)
Gravidity	2-10
Parity	1-9
Consanguinity	63 (23.3%)
History of congenital anomalies	18 (6.7%)

Data expressed as frequency (percentage), mean (SD), range. N: Number.

Table (2): Frequency of tissue anomalies in the studied group during the first scan.

	N=270
Pyelectasis	10 (3.7%)
Chorioid plexus cyst	6 (2.2%)
Echogenic bowel	4 (1.5%)
Echogenic foci in heart	3 (1.1%)
Shortened long bone	2 (0.75%)
Mixed anomalies	2 (0.75%)

Data expressed as frequency (percentage). N: Number.

Tissue anomalies in the studied group during the first scan based on the maternal age: The presence or absence of each soft tissue anomalies had no relation to the maternal age ($p>0.05$) (Table 3).

Table (3): Tissue anomalies during the first scan based on the maternal age.

	Anomaly		*p*-value
	Present	Absent	
Pyelectasis	31.87±3.54	32.10±5.13	0.16
Chorioid plexus cyst	32.13±5.09	31.31±3.45	0.19
Echogenic bowel	31.88±3.95	32.32±2.14	0.29
Echogenic foci in heart	32.01±2.22	31.90±5.30	0.29
Shortened long bone	31.90±3.48	31.89±3.03	0.45
Mixed anomalies	31.78±2.67	32.01±3.11	0.33

Data expressed as mean (SD). *p*-value was significant if <0.05.

It was found that frequency of different soft tissue anomalies was significantly increased with positive history of consanguinity ($p<0.001$) (Table 4).

Table (4): Tissue anomalies during the first scan based on consanguinity.

| | Consanguinity | | p-value |
	Negative (n=207)	Positive (n=63)	
Chorioid plexus cyst	14 (6.8%)	63 (100%)	<0.001
Pyelectasis	7 (3.4%)	58 (92.1%)	<0.001
Echogenic foci in heart	0	28 (44.4%)	<0.001
Shortened long bone	0	23 (100%)	<0.001
Echogenic bowel	0	8 (100%)	<0.001

Data expressed as frequency (percentage).
p-value was significant if <0.05.

Both groups of studied women based on previous fetal anomalies showed no significant differences as regard frequency of different tissues anomalies (*p*>0.05). All 270 women were then subjected to another ultrasound examination between 32 and 36 weeks for assessment of soft tissue anomalies (Table 5).

Table (5): Tissue anomalies during the first scan based on previous fetal anomalies.

| | Previous fetal anomalies | | p-value |
	Negative (n=252)	Positive (n=18)	
Pyelectasis	8 (3.2%)	2 (11.1%)	0.13
Chorioid plexus cyst	5 (2%)	1 (5.6%)	0.34
Echogenic bowel	3 (1.2%)	1 (5.6%)	0.24
Echogenic foci in heart	2 (0.80%)	1 (5.6%)	0.18
Shortened long bone	2 (0.80%)	0	0.87
Mixed anomalies	2 (0.80%)	0	0.87

Data expressed as frequency (percentage).
p-value was significant if <0.05.

Frequency of tissue anomalies in the studied group during the 2nd scan (n=270): All frequency of the tissue anomalies decreased during the second scan. Only six (2.2%) women had soft tissue anomalies in form of pyelectasis (1.10%), chorioid plexus cyst (0.80%) and echogenic bowel (0.40%) (Table 6).

Table (6): Frequency of tissue anomalies in the studied group during the second scan.

	N=270
Pyelectasis	3 (1.10%)
Chorioid plexus cyst	2 (0.80%)
Echogenic bowel	1 (0.40%)
Echogenic foci in heart	0
Shortened long bone	0
Mixed anomalies	0

Data expressed as frequency (percentage). N: Number.

Neonatal outcome among the studied group (n=270): Mean Apgar score was 8.10±0.67 with mean birth weight was 3315.34±322.41 (gm). Twenty neonates required admission to neonatal intensive care unit. All fetuses were free of anomalies (Table 7).

Table (6): Frequency of tissue anomalies in the studied group during the second scan.

	N=270
Pyelectasis	3 (1.10%)
Chorioid plexus cyst	2 (0.80%)
Echogenic bowel	1 (0.40%)
Echogenic foci in heart	0
Shortened long bone	0
Mixed anomalies	0

Data expressed as frequency (percentage). N: Number.

Discussion

This study has demonstrated that when a soft marker is identified, there must be a careful search for other markers, which may include fetal growth restriction. Whilst the exact relationship between soft markers and abnormal chromosomes remains unclear.

In our study, a total of 27 (10%) of the studied women had tissue anomalies; 25 (9.3%) women had isolated soft tissue anomalies while 2 (0.7%) women had mixed anomalies. Three markers were not found in any woman, namely, increased nuchal fold thickness, absent nasal bone and ventriculomegaly.

While the most frequent tissue anomalies were pyelectasis (3.7%) and choroid plexus cyst (2.2%). In contrast, Ahman, et al. [6] showed that the most common marker was echogenic foci in the heart (EIF) by 2.5%, also, in another study the most common marker identified was EIF which accounted for 46.2% of all markers, followed by CPC (32.6%).

Another finding in our study was echogenic bowel (EB) by 1.5%, EIF and shortened long bone by 1.10% and 0.75%, respectively.

Furthermore, we found that the presence or absence of each soft tissue anomalies had no relation to the maternal age, however, it was found that frequency of different soft tissue anomalies was significantly increased with positive history of consanguinity. Also, both groups of studied women based on previous fetal anomalies showed no significant differences as regard frequency of different tissues anomalies.

In accordance with other studies that showed different percentages of tissue anomalies as EIF is detected in 3.0% 0.5-20%, Fetal CPCs are found in 0.18-3.6%, 2.2% and 1.6% of prenatal ultrasonographic examinations, isolated fetal pyelectasis was found in 1-3%, 3.8%, 1.1% and 0.8% of fetuses during second trimester ultrasonography, The incidence of EB reported in the literature ranges from 0.2 to 1.8% and 0.08%. While the incidence of thickened nuchal fold (ThNF) was 0.1% and 0.05% [7-10].

Regarding EIF, it occurs in 0.5-20% of the genetic sonogram [11,12], by about 11% to 18% of fetuses with DS [12,13], and in 4-5% of chromosomally normal fetuses [13]. In the low-risk population, the incidence of DS ranges from 0.1% to 0.4% [14]. The documented chromosomal abnormality rate is 3.3-4.4% in a low-risk population in the presence of EIF [15].

Despite various research endeavors, the relationship of EIF with congenital malformations and chromosomal abnormalities is unclear [15]. Carriço et al. [16] detected 8.1% cardiac defects rate in fetuses with EIF without aneuploidy in fetal echocardiography and concluded that their presence should be interpreted as a possible risk factor for congenital heart defects. However, other authors found that fetal EIF was not associated with heart disease, structural heart defects, or extracardiac anomalies [17].

The low prevalence of EIF in our study population may indicate that EIF was underreported or not revealed in low-risk patients. It is reportedly more common in Asian than non-Asian individuals. It is currently believed that isolated EIF does not increase the risk of aneuploidy in a population previously evaluated by first-trimester combined screening [18]. Therefore, when isolated EIF is found in a low-risk patient for whom aneuploidy screening has been performed, no further risk assessment is required. Furthermore, if there is no evidence of altered cardiac function, a detailed echocardiogram is not recommended as long as the second trimester scan is normal [19].

Fetal CPCs are found in 0.18-3.6% of prenatal ultrasonographic examinations. CPCs typically undergo involution and are no longer detectable by the second trimester in serial ultrasound studies. CPC is anechoic and usually simple in appearance, although it can be complex, and it may be unilateral or bilateral. However, the appearance and laterality have no clinical relevance. Most researchers have indicated that isolated CPC is not associated with a higher risk of aneuploidy [20].

In addition, it has been reported that the presence of CPCs does not affect the neurological outcome during childhood. Recent guidelines have suggested that the presence of isolated CPCs does not require ultrasonographic follow-up [21].

In a meta-analysis, isolated fetal pyelectasis was found in 1-3% of fetuses during second trimester ultrasonography. In accordance to our study, the prevalence of isolated fetal pyelectasis was 3.7%. A recent meta-analysis showed that the presence of pyelectasis increases the likelihood of aneuploidy [8].

However, there is a consensus that isolated fetal pyelectasis is not a justification for karyotyping in low-risk patients [22]. Fetal pyelectasis is often associated with congenital hydronephrosis. Thus, when pyelectasis is observed in mid-trimester ultrasonography, follow-up examination is required. In a study of 8,873 pregnant Korean women who underwent routine mid-trimester screening ultrasonography, 249 (2.8%) cases were identified as isolated pyelectasis. Among them, 18.2% were persistent or progressive pyelectasis based on the third trimester ultrasound, and 3.2% were diagnosed with significant neonatal hydronephrosis after delivery [23].

The definition of a short FL varies. A short femur was defined when the femur length measurement compared to the expected femur length measurement for gestational age was ≤0.91 [24]. In contrast, in a national study of 147,776 fetuses in Denmark, 16.8% of the fetuses with Down syndrome had a short FL below the fifth percentile [25]. Cho et al. [26] concluded that short FL is a poor marker of Down syndrome in the second trimester.

A short humerus was defined by a measured to expected HL ratio ≤0.89 [24]. Gray et al. [27] concluded that an HL below the fifth percentile is the most efficient parameter for the detection of down syndrome. However, a recent meta-analysis showed that an isolated short humerus or femur does not increase the risk of aneuploidy.

When marked shortening of long bones is identified in second trimester ultrasonography, severe skeletal dysplasia should be suspected. Besides, associations have been observed between the presence of a short femur in midtrimester scans and the subsequent development of preterm birth, preeclampsia or SGA [28].

Therefore, when a short femur is suspected at the time of the second trimester screening, follow-

up sonography for fetal growth and heightened awareness of preterm birth or preeclampsia are recommended. De Carvalho et al. [29] demonstrated that fetuses with short HL, as well as FL, measured at mid-trimester ultrasonography were significantly associated with fetal growth restriction. However, contrary to short FL, there are few studies regarding the association between short HL and adverse pregnancy outcomes, such as preterm delivery or preeclampsia.

It has been argued that the presence of soft markers has no additional value in improving the detection of down syndrome in patients deemed to be at low risk at the first-trimester screening.

Few studies have investigated the performance of soft markers in a previously screened population. In a study by Kaijomaa et al. [30], only two fetuses had significant aneuploidy among 228 pregnancies that were found to be normal at the first-trimester screening and presented two or more soft markers at the mid-trimester ultrasonographic examination.

In our study, all frequency of the tissue anomalies decreased during the second scan. Only six (2.2%) women had soft tissue anomalies in form of pyelectasis (1.10%), chorioid plexus cyst (0.80%) and echogenic bowel (0.40%). It was found that shortened long bone, EIF and mixed anomalies were absent in all women.

So the study demonstrate that the Anomalies disappeared in 21 women (78%). This is supported by Loughna [31] who found that many soft markers will disappear or regress as gestation proceeds.

In our analysis, all fetuses with one or more soft markers had no significant findings after delivery. Accordingly, obstetricians should take our results into account when recommending invasive procedures. We did not evaluate the data of the non-invasive prenatal test (NIPT) for aneuploidy. However, the NIPT is a feasible option for women who present with one or more fetal soft markers.

Conclusion:

Soft markers in second-trimester ultrasonography have limited use in screening for fetal aneuploidy. However, these markers can be used as a screening tool for adverse outcomes other than chromosomal abnormality.

Sonography cannot be used to diagnose or exclude aneuploidy. It provides a noninvasive method by which to screen the risk of aneuploidy on the basis of a variety of sonographic features.

Although the management of each of the soft markers is different, a few generalizations can be made. First, the detection of any abnormal finding on ultrasound should prompt an immediate detailed ultrasound evaluation of the fetus by an experienced sonographer. If there is >1 abnormal finding on ultrasound, if the patient is older than 35 years of age, or if the multiple marker screen is abnormal, amniocentesis should be recommended to rule out aneuploidy.

Recommendations:

The screening ultrasound at 16 to 20 weeks should evaluate 8 markers, 5 of which (thickened nuchal fold, echogenic bowel, mild ventriculomegaly, echogenic focus in the heart, and choroid plexus cyst) are associated with an increased risk of fetal aneuploidy, and in some cases with non-chromosomal problems, while 3 (single umbilical artery, enlarged cisterna magna, and pyelectasis) are only associated with an increased risk of non-chromosomal abnormalities when seen in isolation.

Identification of soft markers for fetal aneuploidy requires correlation with other risk factors, including history, maternal age, and maternal serum testing results.

Soft markers identify a significant increase in fetal risk for genetic disease. Timely referral for confirmation, counselling, and investigation is required to maximize management options.

Further researches are needed on larger numbers of cases to give more accurate picture to evaluate the clinical significance of soft markers for aneuploidy screening.

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أهميـة العلامات الغيـر مؤكدة بالجنيـن
فى التنبؤ باختلال الصبغة الصبغيـة

خلفية الدراسة : إن استخدام الموجات فوق الصوتية فى الحمل له تأثير إيجابى هام على صحة الأم والجنين ونظام الرعاية الصحية والإقتصادية للأسرة بإستخدام الموجات فوق الصوتية عالية الدقة، أصبح من الممكن دراسة علم تشريح الجنين بقدر كبير من التفصيل، ولجودة نوعية الصور التى تم الحصول عليها اليوم أصبح من الممكن التعرف بوضوح على الانحرافات الطفيفة فى التكوين الطبيعى للجنين.

الهدف من الدراسة : إن الغرض من هذه الدراسة الاستطلاعية هو مراقبة العلامات اللينه بعد الكشف عنها فى الموجات فوق الصوتية، وتقدير مدى جدوى كل علامة لينة، وتقييم ما إذا كان ينبغى أن ينظر لعلامة محددة عن غيرها وذلك بالفحص الروتينى بالموجات فوق الصوتية. ومتابعة التوصيات للتعامل مع العلامات اللينة وتقديم تقييم لنوعية المعلومات المتعلقة بكل علامة.

المريضات وطرق الدراسة : وهى دراسة استطلاعية على ٢٧٠ من النساء الحوامل فى ما بين ١٦-٢٤ أسبوعاً من الحمل. تم فحص جميع النساء مرتين بالموجات فوق الصوتية للكشف عن تشوهات الأنسجة الرخوة. أولاً فى ما بين الأسبوع السادس عشر والأسبوع الرابع والعشرون والثانى للمتابعة فى حالة وجود أحدى العلامات اللينة فى ما بين الأسبوع الإثنين وثلاثين والاسبوع السادس والثلاثين.

نتائج الدراسة : أوضحت هذه الدراسة أن فى حالة وجود أحدى العلامات اللينة يجب التدقيق لإكتشاف وجود أى علامات أخرى، أوضحت النتائج أن ٢٧ (١٠٪) من النساء الخاضعات للدراسة كان لديهن تشوهات فى الأنسجة. كان لدى ٢٥ (٩.٣٪) من النساء تشوهات الأنسجة الرخوة المعزولة بينما كان لدى ٢ (٠.٧٪) من النساء تشوهات مختلطة. كانت التشوهات النسيجية الأكثر شيوعاً هى اتساع حوض الكلى (٣.٧٪) وكيس الضفيرة المشيمية (٢.٢٪). ثلاث نساء (١.١٠٪) لديهن بؤر مولدة للصدى فى القلب. وجد أن الأمعاء لمولدة للصدى والعظم الوحيد المقصر كانا موجودين فى ٤ (١.٥٪) و ٢ (٠.٧٥٪) من النساء على التوالى. ويستنتج من هذه الدراسة أن العديد من العلامات اللينة سوف تختفى أو تتراجع مع تقدم الحمل. إن استمرار أو أختفاء علامة لا يغير من خطر عدم توازن الصبغيات.

الاستنتاج : وجدت دراستنا أن الارتباطات بين اختلال الصبغة الصبغية والعلامات اللينة المكتشفة فى الثلث الثانى من الحمل كانت أقل أهمية مما تم الإبلاغ عنه سابقاً. ومع ذلك، لا تزال هناك حاجة إلى تقديم المشورة التفصيلية لكل علامة لينة، خاصة إذا لم تخضع المرأة الحامل لفحص اختلا الصبغة الصبغية، أو فى الحالات التى يتم فيها تحديد العديد من العلامات اللينة.