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Ultrasonographic Findings in pregnants with Down Syndrome

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Abtract

Objective: Review the ultrasonographic features of down syndrome cases during pregnancy.

Methods: Ultrasonographic features of Down syndrome cases diagnosed with karyotype investigations due to advanced maternal age, increased risk in biochemistry tests, and structural anomalies diagnosed during ultrasonography.

Results: Ultrasonographic features of 19 cases with Down syndrome were reviewed in a group of 1204 karyotipe investigation. In 7 cases ultrasonographic features were normal (36.8%). In 7 cases nuchal folds were increased. In 4 cases (21.1%) pelvicaliectasis were detected. In 2 other cases there were omphalocel and cardiac anomalies, and cystic hygroma and cardiac echogenic focus was detected in one cases.

Conclusion: During ultrasonographic investigations structural anomalies and soft markers should carefully searched and than decision for invasive procedures were should be considered.

Keywords: Down syndrome, ultrasonography, prenatal diagnosis .

Down sendromlu olgularda ultrasonografik bulgular

Amaç: Gebeliklerinde Down sendromu tanısı alan olguların ultrasonografik bulguları gözden geçirildi.

Yöntem: Gebeliği sırasında ileri maternal yaş, tarama testlerinde yüksek risk, ultrasonografide saptanan yapısal anomaliler nedeniyle amniosentez sonrası karyotip incelemesi ile Down sendromu tanısı alan olguların ultrasonografik bulguları gözden geçirildi.

Bulgular: Karyotip incelemesi yapılan 1204 olgudan kesin Down sendromu tanısı alan 19 olgunun ultrasonografik bulguları incelendi. 7 olgu normal değerlendirildi (%36.8). 7 olguda ense kalınlığı yüksekti. 4 olguda (%21.1) pelvikaliektazi saptandı. İkişer olguda omfolosel ve kardiak anomali tespit edildi. Birer olguda ise kistik higroma, kardiak ekojenik odak, koroid pleksus kisti ve pes ekinovarus saptandı.

Sonuç: Rutin ultrasonografik incelemeler ile down sendromlu olgularda saptanan soft markerlar üzerinde dikkatle durulmalı ve yaklaşım tekrar gözden geçirilmelidir.

Anahtar Sözcükler: Down sendromu, ultrasonografi, prenatal tanı .

Introduction

During ultrasonographic investigations some detected soft markers are related with Down syndrome and other aneuploidies such as increased nuchal fold thickness, echogenic bowels, short femur length, pyelectasis, hypoplastic nasal bone, choroid plexus cyst, echogenic intracardiac focus.[1] Prenatal biochemical tests are more valid than second trimester ultrasonographic investigations for diagnosis of Down Syndrome. Structural anom-

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alies are friquently related with chromosomal anomalies. Soft markers increases amniocentesis and this may lead to a little increase in abortion rates. In this study, ultrasonographic soft markers of fetuses diagnosed of Down Syndrome are retrospectively reviewed.

Methods

Our study group consisted of 19 cases with Down Syndrome . Amniocentesis was indicated in 1204 women due to family history of aneuploidies, maternal age, increased risk in biochemical tests, ultrasonographic soft markers and structural anomalies in these fetuses during the years 2006 and 2007. Ultrasonographic findings of these cases detected during ultrasonographic investigations which were performed in 11-22 weeks gestations were reviewed. The fetuses without karyotype analysis or diagnosed postnatally were not taken in to the study group since their records can not be reached. Early fetal deaths were also not included. Ultrasonographic investigations were made by using a Medison 3.5 MHz probe by our hospital physicians. Amniocentesis were performed by our physians under ultrasonography by using 22 g spinal needles after two times of povidone iodine aplication to lower abdomen. Our soft markers are nuchal translucensy more than 3 mm, nuchal fold thickness more than 6 mm,[1,2] choroid plexus cysts independent of number and size, intracardiac echogenic focus, echogenic bowels, renal pyelectasis (when anteroposterior length of renal pelvis is more then 4 mm), short femur or humerus length when actual length is shorter than 85% of expected length. Since ultrasonografic investigations were made by different physicians, we couldn't reach nasal bone findings in all hospital recordings. For this reason nasal bone findings are not included in our evaluation. All structural anomalies and soft markers were recorded.

Results

Ultrasonographic findings of 19 cases with Down syndrome diagnosed with karyotype investigations were included as the study group. Between 2006-2007 years we find out 1204 karyotypic analysis performed for advanced maternal age, increased risk in biochemical tests, structural anomalies and soft markers detected during ultrasonography and history of chromosomal anomalies in the family., Nineteen of cases were diagnosed as trisomy 21. Six patients were older than 35 years in this group. Mean maternal age of 19 cases was 30.9, average parity was 1.1, and mean gestational age was 18 weeks at the time of amniocentesis. Ultrasonographic findings of 19 cases with Down Syndrome are listed Table I. In 9 cases there were more than one ultrasonographic finding. All cases with Down Syndrome were discussed at our perinatology council. After situation was explained to the parents 16 cases were inducted by misoprostol. Three

Table 1. Ultrasound findings in Down syndrome cases.

Findings	Cases	%
Normal anatomy	7	36.84
NT>3 mm (5) NF>6 mm(2)	7	36.84
Renal pelvicaliectasia	4	21.05
Nasal bone hypoplasia	2	10.52
Cardiac anomaly	2	10.52
Omphalocele	2	10.52
Cystic hygroma	1	5.26
Intracardiac echogenic foci	1	5.26
Plexus choroideus cyst	1	5.26
Pes equinovarus	1	5.26

patients refused the council decision, 2 of those had gone cesarean section due to ablatio plasenta and fetal distress, one case was born spontaneously.

Discussion

Down Syndrome is seen in 1.41 of 1000 live births and ıt's the one of the most important chromosomal anomalies that is investigated by perinatologists.[3] Ultrasonographic soft markers are, increased nuchal fold thickness, echogenic bowels, short femur-humerus length, pyelectasis, echogenic intracardiac focus, and choroid plexus cyst. Nasal bone aplasia and hypoplasia were added to the list afterwards. All these markers are useful clues for diagnosis of Down Syndrome.[4] Cardiac anomalies are seen at about 50 % of Down Syndrome patients.[5] In spite of different study results structural anomalies can be detected at about 30 % of Down Syndrome cases by second trimester ultrasonographic investigations.[6-8] Yıldırım reported that 12.9 % of the cases had chromosomal anomalies and 4.6 % of these cases had Down syndrome in a group of cases with fetal anomalies detected during ultrasonography. They concluded that detection of chromosomal anomalies by amniocentesis was higher in cases with fetal anomalies rather than advanced maternal age or risk in triple test.[9] Sener reported that they foud pathologic ultrasonographic features in 30 % of cases with Down syndrome.[10] Soft markers are usually found with structural anomalies.[11] Furthermore it's said that isolated soft markers such as choroid plexus cyst, echogenic bowel, short femur, short humerus are not related with Down Syndrome. In our series there was no ultrasonographic marker among 7 cases. If it would be possible; could be find any marker in these pregnants?. The main question is what shall we do when find a soft marker. These markers can be found at about 14-15 % of normal second trimester pregnancies. False positivity and fetal deaths due to invasive diagnostic procedures will increase if we prefer karyotype analysis for every soft marker detected. According to Bindman et al. İsolated soft markers are seen at 13.9 % of Down Syndrome cases and 9.3% of normal fetuses.[11] The most common anomalies are congenital cardiac defects, cerebral ventriculomegalies, cystic hygroma, hydrops, hydrotorax, omphalocel, duodenal atresia and abnormal extremities.[8,12] Boyd et al reported that soft markers, increased the detection of malformation diagnosis rate by 4 % but on the other hand false positivity increased by 12 times.[13] Because of these increased likelihood ratio was proposed to be 2.0 when evaluating the soft markers.[14] And in the abcense of soft markers the case should be removed from the high risk group.[15] Nasal bone aplasia was detected in 0.5 % of normal fetuses and 43 % of trisomy 21 cases at Bromley's study[16] and they concluded that this soft marker increased the likelihood ratio 83 times. In our study we detected one nasal bone aplasia and one nasal bone hypoplasia (10.52%). This low percentage in our study may be due to evaluations done by different physicians. Also those patients may not be evaluated with same interest and care. In our opinion this ratio will increase when evaluations were done by well educated and carefull physicians. Bromley and et al reported that nuchal fold thickness 6 mm and/or more at 15-22 weeks of gestation increases trisomy 21 risk by 17 times but noted that this is not a common finding.[13,17] In our study, in 5 cases nuchal translucency and in 2 cases nuchal fold thickness was increased. Isolated increased nuchal

fold thickness was seen at 4% of Down Syndrome cases and together with 26 % of other anomalies.[17] It's accepted that this soft marker, even in isolated form, increases Down Syndrome risk.[17-18] Hyperechogenic bowel is encountered at 15 % of trisomy 21 patients and 6.6 % of normal fetuses.[19] We have no case with this finding in our study group. It should not be forgotten that hyperechogenic bowel can be related with swallowed blood, cystic fibrosis disease and fetal infections. It's known that short femur and humerus length increases chromosomal anomaly risk.[20-21] 2.5 times increase at standard percentiles is accepted as diagnotic criteria[22] and short humerus length is more diagnostic than femur length.[16,23] So it's advised to measure humerus length during second trimester ultrasonographic investigation.[14] Choroid plexus cysts are encountered in 25-30% of cases with trisomi 18 and 1 % of normal fetuses. They vary 3-16 mm in size can be detected at 14-16 gestational weeks and disappear at 22. gestational week. They are not accepted as soft markers for trisomy 21 but their importance is; they direct to reexamine fetal structures that may be related with trisomy 18. Renal pyelectasis is seen at 17% of Down Syndrome cases. We detected this finding in 4 of our 19 patients (21%). Aneuploidy ratio is 1/300 at isolated pyelectasis and this situation may also be related with hydronephrosis or postnatal urinary reflu disease.[24-25] Isolated pyelectasis is not accepted as an increased risk factor for trisomy 21 risk.[7] Middle phalanx hypoplasia of 5th finger, sandal gap, fetal ear size, brachycephaly are the other ultrasonographic findings, but these usually take place in case reports and not accepted as soft markers.[14] Single umblical artery may also seen in aneuploidic fetuses but it is usually together with other findings.[24-26] Isolated single umblical artery is not related with increased aneuploidy risk.[14]

Conclusion

As a conclusion when a soft marker is detected its risk accelaration should be considered than decision for invasive procedures should be evaluated Fetal structures should be reinvestigated carefully and than invasive procedures should be considered.

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