Fetal Thoracic Malformation at 13 weeks of Gestation Associated With Turner Syndrome: A Case Report

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In the present case report, we stated the intercourse between thoracic vascular malformations observed in the first trimester of pregnancy and Turner syndrome. Prenatal doppler findings of the vascular tumor and its relation with Turner syndrome is also evaluated.
Since the introduction of antenatal screening programs for Turner syndrome based on nuchal translucency thickness in the first trimester of pregnancy, increasing number of fetal structural abnormalities are being detected at the 11-14 week ultrasound scan. Furthermore, cardiovascular complications are the main cause of increased mortality in Turner syndrome.

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Key Words: Turner, Vascular malformations, Nuchal thickness

The Turner syndrome is associated with a chromosomal defect (monosomy X0 or structural abnormality of one of the X chromosomes) that affects crudely 1 in 2000 to 1 in 5000 live female births.[1] The most common features are short stature and gonadal dysgenesis, but the most serious clinical aspect of the syndrome is due to congenital cardiovascular anomalies that include most critically, aortic coarctation and dissection.[2-3]

An increased nuchal translucency measurement at 10-14 weeks of gestation is known to be associated with fetal chromosomal abnormalities[4] and other structural or genetic defects.[5]

During the last 30 years, extensive research has been aimed at developing a noninvasive method for prenatal diagnosis of chromosomal and other abnormalities through the isolation and examination of fetal cells that are found in the maternal circulation. However, there is no realistic prospect that, in the foreseeable future, noninvasive diagnosis will replace the need for invasive testing.

Case Report

The patient we have reported was 19 years old and the pregnancy was her first one. She referred to our perinatology unit at 13 weeks of gestation for the first trimester screening.The patient had a healthy baby from her first normal labor.

The fetus was scanned transabdominally in sagital planes such that it occupied three quarters of the image field. The maximal thickness of the sonolucent zone was measured between the inner aspect of the fetal skin and the outer aspect of the soft tissue overlying the cervical spin or the occipital bone. Three nuchal translucency thickness measurements were made for fetus, with the longest value chosen for analysis.

According to the findings in her first trimester ultrasound screening, there was a single vital fetus in intrauterine cavity and the measurement of the CRL is 67,4 mm. Nuchal translucency measurement was 3,2 mm. The use of color doppler allowed the serendipitious identification of a highly vascular structure in the posterolateral portion of the fetal thorax, in proximity to the costovertebral angle, at the level of a four-chamber view of the heart. The presence of the mass was confirmed at two different angles of insonation, at least 45 degrees apart. The lesion had a spheric shape and was 6 mm in diameter. Color doppler analysis stated the highly turbulent flow within the lesion which was confirmed by pulsed wave doppler ultrasonography (Figure 1).

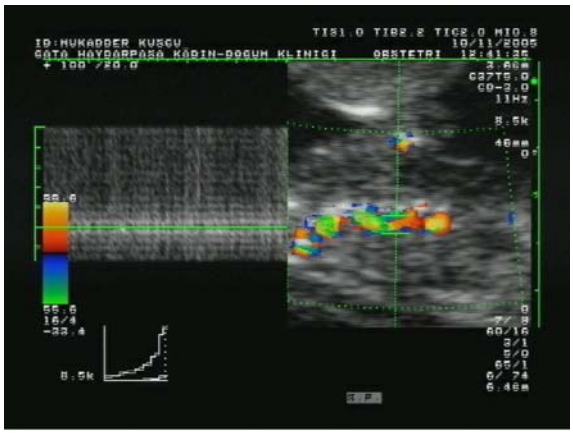

Figure 1: Color doppler analysis stated the highly turbulent flow within the lesion which was confirmed by pulsed wave doppler ultrasonography.

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The termination of pregnancy was proposed to the patient according to the accomplished lesions. After the parents decision, we took the informed consent from them. Before the medical termination of the pregnancy, CVS was performed in order to get karyotype of fetus. The pregnancy was terminated by two doses of vaginal misoprost

Histologic examination revealed a malformation composed mainly of blood capillaries and to a lesser extent, lymphatic capillaries (Figure 2). The determination of fetal karyotype, sampled from the nuchal zone of the abortus material was 45 X0 monosomy (Turner syndrome).

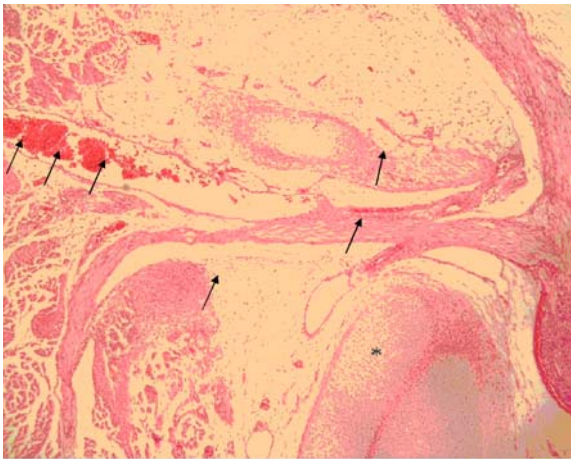

Figure 2 : Histologic picture demonstrating dilated vessels, a feature of vascular malformation. Hematoxylin and eosin. Original magnification ×100. Arrows: Vessels, Asteriks: ribs

Discussion

This study demonstrates the association between the prenatal doppler finding of a vascular tumor in the fetal chest and Turner syndrome. This association is varified by chromosomal and histological analysis.

One article reported the association between thoracic vascular malformations observed in the first trimester of pregnancy and Down syndrome.[6] In that study, lesions had a globular shape and were 4-6 mm in diameter, occupying almost one third of the hemithorax. Down syndrome was diagnosed in five out of the seven cases. This association between thorasic vascular malformation and Turner syndrome was not indicated in past case reports.

The occurence of vascular tumors is not well documented in Turner syndrome. Although it is possible to postulate that, a fetal hyperdynamic circulation as a consequence of blood shunting within the vascular tumor may contribute to the development of cardiac strain and increased NT.[7] During the last 30 years, extensive research has been aimed at developing a noninvasive method for prenatal diagnosis of chromosomal and other abnormalities through the isolation and examination of fetal cells that are found in the maternal circulation.

Increased fetal nuchal translucency thickness is a useful ultrasonographic marker for screening of fetal aneuploid conditions, particularly chromosomal trisomies,[8] Turner syndrome [9] and triploidies.[10]

Within a transluceny value set at 3,0 mm, the detection rate of unbalanced translocation pregnancies from a balanced translocation population was 71% and specificity was 96% in the case control study. Possible mechanisms include cardiac dysfunction, venous congestion in the head and neck, alteration in extracelluler matrix, lymphatic vessel hypoplasia, congenital infection, anemia and hypoproteinemia. There is increasing evidence that many neural crest - related cardiovascular defects may be caused by genetic imbalance.[11]

Nuchal cystic hygroma is the most frequent fetal neck pathology observed at antenatal sonograms.[12] The sonographic diagnosis is based on the proof of the cystic structures in constant position in the cervical region possibly with extension to the lateral, caudal, cranial and anterior aspects. By making careful and detailed prenatal diagnosis of it,observed associated sonographic findings are expected to identify the more normal cases.

If an extremity, craniofacial, renal or cardiac anomaly is observed beside the nuchal cystic hygroma it might be an indication for a trisomy on Turner syndrome.

The differential diagnosis of nuchal cystic hygroma includes nuchal oedema, meningocele, encephalocele, cervical teratoma, the pseudomembrane, hemangiomas, subchorial placental cyst and familial nuchal blebs. It is known that nuchal translucency may resolve spontaneously and if the karyotype is normal it has a relatively good fetal prognosis compared to the persistent nuchal translucency cases.[13] Pathophysiology basis of Turner syndrome is considered a consequence of haploinsufficiency of certain genes on X chromosome.[14] Most often, Turner syndrome is first diagnosed incidentally by amniocentesis or chorionic villous sampling performed for unrelated reasons. Nuchal translucency, cystic hygroma, coarctation of aorta, renal abnormalities, growth retardation and fetal hydrops are the most common prenatal ultrasound signs. Abnormal maternal serum alpha-fetoprotein, human choronic gonadotropin and unconjugated estriol are also suggestive of Turner syndrome.[15]

Regular ultrasound examinations have aided the detection of nuchal cystic hygroma and non-immun hydrops during pregnancy, which are typical but not patognomonic of Turner syndrome. As doppler artefacts can easily be obtained in proximity of skeletal structures, the presence of the mass had to be confirmed at different angles of insonation to exclude false positive findings.

Although it was stated as normal variation for vascular malformation in normal populations, the implication of an

association between readily identifiable fetal doppler masses in the first trimester of pregnancy and Turner syndrome is evident. As this finding is observed in this patient for fetal karyotyping, future studies should address the confirmation about this.

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