The prenatal diagnosis of iniencephaly accompanying with cystic hygroma: A rare case report

Comert Eh Sal H, Guvendag Guven ES, Guven S
Karadeniz Technical University, School of Medicine, Department of Obstetrics and Gynecology, Trabzon, Turkey.

Objective
Iniencephaly is a rare birth defect characterized by hyper-retroflexion of the head with absence of neck due to spinal deformity. The etiology is unknown and is seen 0.1 -0.01 of 10,000 deliveries. Cystic hygroma (CH) is a congenital anomaly of the lymphatic system and is a rare soft tissue tumor. CH can be seen isolated or may be associated with some chromosomal anomalies. In this case, we want to present case of the prenatal diagnosis of iniencephaly associated with cystic hygroma.

Methods
A 24-years-old woman was referred to our clinic for an antenatal anomaly scan at 13 week of pregnancy. CH was observed in a performed ultrasonographic examination of 1st trimester screening of the 13 week of pregnancy at the external center but the patient was applied to our clinic at 17 week of gestation. In the detailed ultrasonographic examination of the patient, shortening of spinal cord, hyperextension of fetal head and meningocoele sac in the occipital region were observed and also 45x35mm cystic hygroma with septation was observed in the neck and head. The iniencephaly associated with cystic hygroma was diagnosed after the prenatal examination. The prognosis was explained to the family. Due to the poor prognosis and the additional anomalies associated with it, the termination was recommended and accepted by the family. Hyperextension of head and wide cystic hygroma around the neck and head was observed in the abortion material.

Results
The etiology and pathogenesis of iniencephaly are not known exactly, both genetic and environmental reasons have been blamed. It is associated with chromosome anomalies including trisomy 13, trisomy 18, and monosomy X. Low socioeconomic level, folic acid deficiency, low parity, maternal obesity, tetracycline, sulphanamide, antihistamine, and antitumoral drugs may increase the risk of iniencephaly. CH is a congenital anomaly of the lymphatic system and is a rare soft tissue tumor. CH often seen in the neck region (%75-90), %20 in axillary, %5 in mediastinal, retroperitoneal area and rarely in chest wall. Cystic hygroma can be seen isolated or may be associated with some chromosomal anomalies such as turner syndrome (most common %40-80). Except this, related karyotype anomalies; down syndrome, trisomy 21, klinefelter syndrome, partial trisomies, partial monosomes, translocations, and mozaisms.

Conclusion
The prognosis of iniencephaly is inconsistent with life and is highly mortal. First mark for diagnosis of iniencephaly is fetal posture in obstetric ultrasonography. Repeated examination of the fetus posture in the 1st trimester when an abnormal finding is detected, will result in early diagnosis of fetal anomalies with posture changes. Concomitant cystic hygroma, which we present, was caused to delay the evaluation of the occipital region and bone structure in early prenatal assessment. In this cases, the prognosis of the pregnancy should be informed and termination should be offered to the family. Early diagnosis and termination are important for reducing maternal morbidity and mortality.