Anterior Cystic Hygroma with Normal Karyotype

Anterior Lokalizasyonlu Kistik Higroma

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ÖZET

Kistik higroma fetal dönemde %80 posterior servikal bölgede görülen kromozomal anomalilerle birlikteliği olan yapısal konjenital anomalidir. Bu yazıda anterior lokalizasyon gösteren normal karyotipli kistik higroma olgusu sunulmaktadır.

32 yaşında multigravid hastanın ikinci gebeliğinin 17. haftasında anterior çene kitlesi olan erkek fetus tespit edildi. Normal karyotipi olan fetus postpartum solonumu tıkayan kistik higroma nedeniyle başka merkezde sklerozan terapi aldı.. Şu anda 15 aylık olan fetusun tek gözde körlük dışında nörolojik gelişimi 3 ay geriden gelmektedir.

Kistik higroması olan fetuslar normal karyotipte olsa bile olumsuz perinatl sonuçları olan bir durumdur. Bu yüzden antenatal boyun kitlesi tanısı alan fetuslarda karyotipleme normal olsa bile kistik higroma ve olumsuz perinatal sonuçlar akılda tutulmalıdır.

Anahtar kelimeler: Kistik higroma, antenatal tanı

ABSTRACT

Cystic hygroma is a rare congenital cystic mass associated mostly with chromosal abnormalities and located 80% at the posterior cervical portion of the neck. This case is presented due to the atypical anterior location of the cystic hygroma and normal karyotype of the fetus.

A 32 year old, multigravid womans' male infant with anterior cervical cystic mass under chin was diagnosed at 17th week of gestation After birth sclerosan injection therapy applied at another pediatric clinic. After several times of injections the mass decreased in size that it does not obstruct respiration anymore. Baby is now 15 months old. His neurologic development is 3 months retarded than normal age and one side blindness observed.

Fetuses with cystic hygroma are at high risk for adverse outcomes and detailed prenatal diagnosis including karyotyping should be offered. Cystic hygroma should be kept in mind at all neck masses even the karyotype is normal or location of mass is atypic for cystic hygroma.

Key Words: cystic hygroma, antenatal diagnosis

INTRODUCTION

Cystic hygroma is a rare congenital cystic mass that generally arises from the failure of the lymphatic system to communicate with the venous system in the neck. A cystic hygroma also may arise from a failure of the juguloaxillary lymphatic sac to drain into the internal jugular vein. It seems like single or multiloculated fluid filled cavities. (1) The prevalence of isolated cystic hygromas at anterior part of the neck is very rare. 80% of all cystic hygromas involve the posterior portion of the neck and the lower part of the face (axilla 15%, retroperitoneum, abdominal viscera 2%, cervico-mediastinal 3%). In children the most common location is the posterior cervical space, followed by the oral cavity. They are usually very infiltrative in nature.

There is an association between fetal cystic hygroma and chromosomal abnormalities especially Turner syndrome, trisomy 18, 19 and 21 (2).

This case is presented due to the atypical anterior location of the cystic hygroma diagnosed at antenatal period (17. week) and normal karyotype of the fetus. Thus when a mass located in the anterior neck extending to the chin is detected; cystic hygroma should always be kept in mind.

CASE REPORT

A 32 year old, multigravid woman admitted to our clinical for following up of her present pregnancy. The follow up of the patient and the baby was normal until 17th gestation week. The nuchal translucency of the fetus was normal at the 11th-14th weeks of gestation and the combined trisomy 21 risk was detected 1/715. Male infant with anterior cervical cystic mass under chin was diagnosed at 17th week of gestation. After the diagnosis second trimester scanning done by a specialist and anterior multilocular predominantly cystic mass with septa of variable thickness observed. At a tertiary centre perinatology meeting the mass is decided to be a teratoma of the chin. The genetic analysis of the fetus was normal determined via percutenous umbilical blood sampling. Until 39 weeks of gestation fetal growing and maternal health was normal. Caesarean section performed because of prior surgery at 39 th gestational week. After birth the neck mass under chin was diagnosed as cystic hygroma. The mass was large and located in the anterior neck extending to the chin. Genetic analysis of the fetus was normal. There were no additional structural anomalies detected after birth. Baby was admitted to the neonatal intensive care unit. The apgar scores of the first and fifth minute were 9-10 respectively. Beside observation under free oxygen, antibiotherapy was applied. At the first day, both abdominal and cranial ultrasonographies reported as normal except fluid filled mass under chin. At the second day of birth subcostal retractions observed and at the third day the baby was intubated for respiratory obstruction of the mass. At the fourth day phototherapy started and after that sclerosing therapy by injections began at another pediatric clinic. After several times of injections the mass decreased in size that it does not obstruct respiration anymore. Baby is 22 months old now. His neurological development is 3 months retarded than normal age and one side blindness observed.

CONCLUSIONS

Fetuses with cystic hygroma are at high risk for adverse outcomes and detailed prenatal diagnosis including invasive procedures like amniocentesis and cordocentesis for genetic analysis should be offered. (3) Exclusion of fetal heart defects is very important. Cystic hygromas can be either with septation or without septation but the ones with septation like the big sized ones are related with bad prognosis and together with chromosomal anomalies. (4) In our case despite the septation of the mass no chromosomal anomaly was detected. Even though the normal genetic analysis is an indication of good prognosis as in our case, a tertiary centre needed for stabilization of baby until mass decreases or removal of respiratory obstruction. Prenatal drainage is useless (2), termination should be considered in severe cases especially to the ones with abnormal karyotype. In milder cases surgical removal may be postponed after birth. Instead of surgery, radiotherapy, aspiration, bleomycin injection can be used in suitable cases. (4) In that reported case, sclerosan therapy resulted in both functional and aesthetic good results. Hamartoma of the mandible, cervical thymus cyst, brachial cleft cyst, thyroglossal duct cyst must be considered for differential diagnosis of cystic hygroma of the anterior neck. This baby was considered as hamartoma of the chin at antenatal examinations. The atypical location and normal karyotype were the reasons of misdiagnosis. The delivery of the patients with a neck mass baby whatever the antenatal diagnosis was, should be in a centre with a genetic specialist, expert neonatal and pediatric surgical care team is available. Cystic hygroma should be kept in mind at all neck masses even the karyotype is normal or location of mass is atypic for cystic hygroma.

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