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Case Report

# Discussion on Cystic Hygroma Based on Its Natural History Through Two Cases Diagnosed by Ultrasonography

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## ABSTRACT

Cystic hygroma has an incidence rate of 1 case per 1000 birth. Half of cystic hygroma may have chromosomal abnormalities, and demonstrate poor prognosis. We experienced two cases of cystic hygroma and report with a literature review. Case 1 is a 37-year-old woman. Transabdominal ultrasonography at 12 weeks of gestation demonstrated bilateral cysts at neck, and diagnosed as cystic hygroma. No abnormal findings are detected by amniocentesis. With a request for artificial abortion, vaginal delivery was performed. Case 2 is a 28-year-old woman. Thick nuchal translucency was detected at 12 weeks of gestation. There were bilateral cysts at neck, and diagnosed as cystic hygroma. Amniocentesis revealed the inversion of chromosome 13. With a request for continuing pregnancy, regular prenatal visits have been performed. Half of cystic hygroma may have chromosomal abnormalities. Based on its natural history with decision of performing amniocentesis, providing appropriate information through ultrasonography can be extremely useful.

**Keywords:** Cystic Hygroma; Chromosomal Abnormalities; Nuchal Translucency; Ultrasonography

## INTRODUCTION

A cystic hygroma is a singular or multi-loculated fluid collection, typically along the poserior fetal neck and back. It is often pointed out that cystic hygroma has an estimated incidence rate of 1 case per 1000 birth. About half of cystic hygroma seen before 30 weeks of gestation may have chromosomal abnormalities or nonimmune fetal hydrops, and demonstrate poor prognosis [1]. Cystic hygroma is associated with an increased risk of not only structural malformations but also autosomal trisomy and copy number variants compared with fetuses without an enlarged nuchal translucency [2,3]. A previous study reported that 18% had an adverse pregnancy outcome (termination for fetal abnormality, intrauterine fetal death, or miscarriage) in fetuses with an enlarged nuchal translucency versus for those with a normal measurement [4]. Although it is not a simple process for parents to make a decision of performing amniocentesis since sometimes these diagnostic testing may be done only at higher medical facilities, careful ultrasound examination and explanation can be helpful in decision making. We experienced two cases of cystic hygroma and report with a literature review.

## CASE REPORT

Case 1 is a 37-year-old primigravid woman. Transabdominal ultrasonography of her fetus at 12 weeks of gestation demonstrated echo free spaces at neck. There were bilateral cysts at neck with diameters of 8 mm and 10 mm respectively and diagnosed as cystic hygroma by ultrasound (Figure 1). No abnormal findings are detected by amniocentesis. With a request for artificial abortion, vaginal delivery was performed at 19 weeks 3/7 days of gestation.

Case 2 is a 28-year-old woman, gravida 3, para 2. Thick nuchal translucency of 6 mm was detected at 12 weeks of gestation (Figure 2). There were bilateral cysts at neck with major axes of 15 mm and 10 mm respectively, and diagnosed as cystic hygroma (Figure 3). Amniocentesis revealed the sporadic inversion of chromosome 13, with no chromosomal abnormalities of parents. With a request for continuing pregnancy, regular prenatal visits have been performed.

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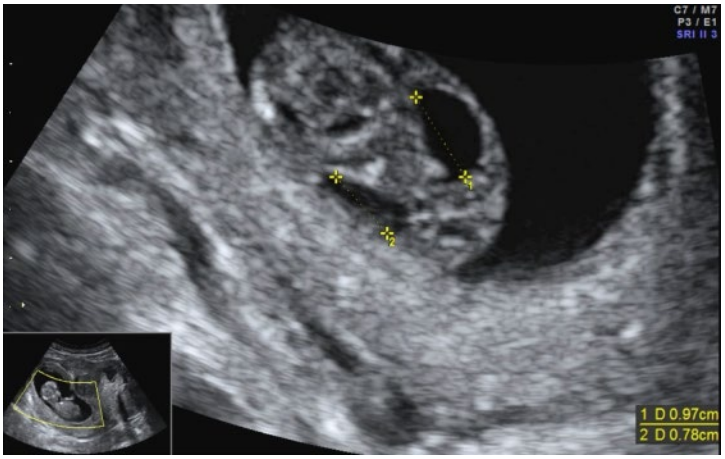


**Figure 1:** Transabdominal ultrasonography finding of case 1. There were bilateral cysts at neck with diameters of 8 mm and 10 mm respectively.

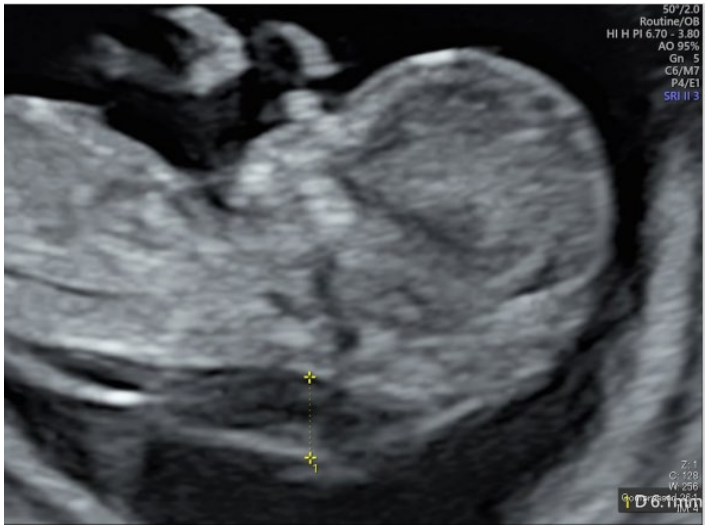

**Figure 2:** Transabdominal ultrasonography finding of case 2. Thick nuchal translucency of 6 mm was detected at 12 weeks of gestation.

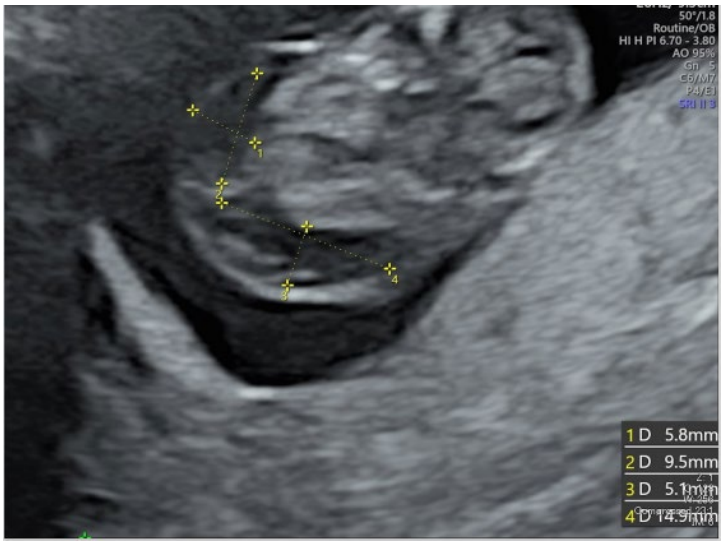


**Figure 3:** Transabdominal ultrasonography finding of case 2. There were bilateral cysts at neck with major axes of 15 mm and 10 mm respectively**.**

## DISCUSSION AND CONCLUSION

We experienced two cases of cystic hygroma and herein showed two important clinical observations. First, the conventional finding seems to be correct that about half of cystic hygroma may have chromosomal abnormalities, while the rest may have normal karyotype. Second, providing appropriate information through ultrasonography can be extremely useful with decision of performing amniocentesis or not.

The first finding is important for evaluating cases with cystic hygroma both in embryological aspect and in obstetric practice. About half of cystic hygroma seen before 30 weeks of gestation may have chromosomal abnormalities, while the rest may have normal karyotype [1,2]. The half of the cystic hygroma with normal karyotype might have congenital heart disease and skeletal abnormalities [5,6]. Possibility of normal child with all of the cases of cystic hygroma is reported as 20% [7]. We experienced two cases of cystic hygroma. Case 1 showed no abnormal findings by amniocentesis. Case 2 showed the sporadic inversion of chromosome 13, with no chromosomal abnormalities of parents. Not all of the cases with fetal cystic hygroma may demonstrate chromosomal abnormalities as per previous reports. It is imperative to recognize that point since all cases should not have fatal anomalies, although case 1 had requested for artificial abortion.

The second point is extremely important as providing appropriate information through ultrasonography can be helpful for parents to make decisions. Lithner et al. reported that the overall risk for an adverse pregnancy outcome with a nuchal translucency more than 3.5 mm and normal karyotype was 25.9%, demonstrating an overall chance of a favorable outcome of 74.1% [8]. If a cystic hygroma is detected on ultrasound examination, genetic counseling should be given to discuss its clinical significance and options for further evaluation [9]. Diagnostic genetic testing and performance of a detailed ultrasound examination to assess for structural abnormalities are recommended. We consider that careful ultrasound examination and polite explanation led to the choice of amniocentesis in both case 1 and case 2. It is often pointed out that whether patients are convinced is very important, and we believe that polite explanation as well as detecting cystic hygroma by ultrasound should be helpful guidance to parents.

There are some sonographic findings associated with, but not diagnostic of, major aneuploidies such as cystic hygroma, enlarged nuchal translucency, absent nasal bone, fetal growth restriction, and structural anomalies. Numerous papers have reported that their use as a component of the combined test with maternal age, serum total or free beta human chorionic gonadotropin, serum pregnancy-associated plasma protein A, and so on. Before giving pregnant people diagnostic genetic testing, careful ultrasound examination and polite explanation are imperative since sometimes these diagnostic testing may be done only at higher medical facilities.

In conclusion, we experienced two cases of cystic hygroma. Based on its natural history with decision of performing amniocentesis or not, providing appropriate information through ultrasonography can be extremely useful.

## DISCLOSURES

Consent was obtained or waived by all participants in this report.

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