# Cystic Hygroma with Edward Syndrome: A Case Report

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## Abstract

Cystic hygromas are fluid filled sacs that result from blockage in lymphatic system. In developing foetus, it can progress to hydrops and eventually fetal death. Its occurrence is 1% in foetuses between 9–16 weeks of pregnancy. The risk for chromosomal abnormalities approaches 50% when cystic hygroma is found prenatally. In current case report, we have discussed a case of 20-year-old female diagnosed with cystic hygroma with Edward Syndrome of 15-weeks pregnancy. It could be managed with second trimester termination of pregnancy. Karyotyping of foetus diagnosed of Edward syndrome.

**Keywords:** Cystic hygroma, Edward syndrome, Lymphatic system.

## Introduction

Cystic hygroma was first described by Redenbacker in 1828 as abnormal accumulation of fluid in the region of the fetal neck(1). It results from early sequestration of embryonic lymphatic channels as sequestered tissue fail to communicate with remainder lymphatic(2). It is one of the major anomalies associated with aneuploidy. Cystic hygroma is often found with Edward syndrome i.e. trisomy 18 which is serious genetic error in cell division. Incidence of cystic hygroma is 1:5000 in babies born of Edward syndrome and they do not live beyond 1 year. Its inheritance is not known. Women with age more than 30 years with female foetus are predisposed to Edward syndrome.

## Case History

A 20-year-old primigravida with 15 weeks gestation came with ultrasonography report which showed foetal cystic hygroma and generalised anasarca. There were no complaints of pain in abdomen, bleeding per vagina. Menstrual and Obstetric history was normal. Patient was stable; she was of thin built with blood pressure of 120/80 mm of Hg on time of admission. No other significant medical history was noted. On abdominal examination, abdomen was soft, well relaxed and per speculum examination showed healthy cervix and vagina, and uterus was of 14 - weeks size. Routine investigations were done. Ultrasonography showed single live intrauterine pregnancy of 13 weeks 6 days with large cystic hygroma in foetal cervical region with oedematous foetal scalp (Fig. 1).

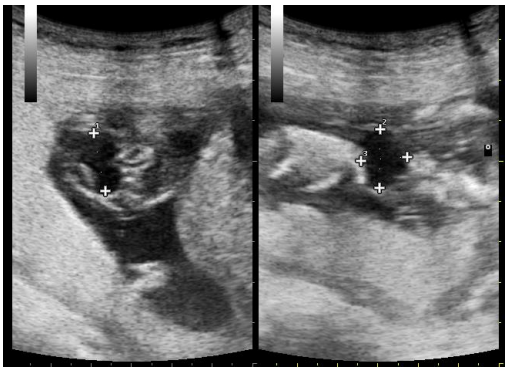

**Fig. 1: Ultrasonography showing cystic hygroma**

Thus prenatal cervical cystic hygroma was diagnosed. Counselling of the couple was done and option for medical termination of pregnancy was suggested. Termination of pregnancy was done by giving Tab. Mifepristone 400mg followed by Tab. Misoprostol 200mg per vagina after 48 hrs of Mifepristone. Placental tissue was sent for karyotyping. Cytogenetic profile of product of conception indicated trisomy 18, Edward syndrome Fig. 2(a) and Fig. 2(b).

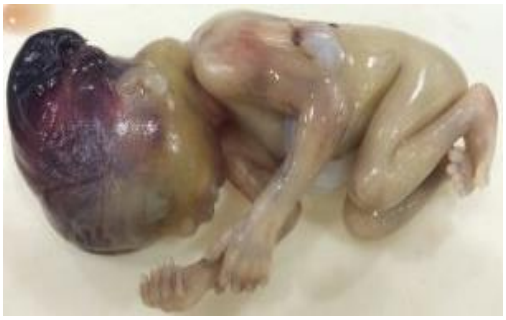

**Fig. 2 (a): Freshly aborted foetus with cystic hygroma (Anterior view)**

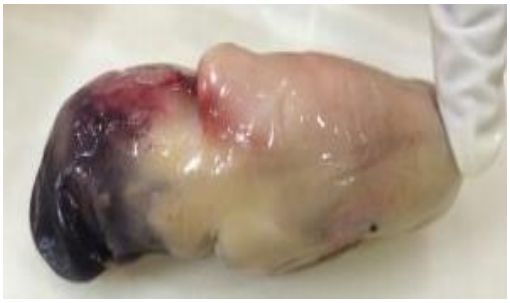

**Fig. 2 (b): Freshly aborted foetus with cystic hygroma (Posterior view)**

## Discussion

Cystic hygroma first described in 1828 by Redenbacher is an anomaly of lymphatic system that is characterised by single or multiple cysts within soft tissue up to size of 80 mm[3]. Cystic hygroma goes with aneuloidy. About 50% foetuses are associated with chromosomal abnormality like trisomy 18, Klinefelter[4]. If it is diagnosed early i.e. 9-16 weeks, it is not associated with chromosomal abnormality. Recurrent cystic hygromas are reported to have possible autosomal recessive pattern. Small lesions can be resolved and large ones persist. Prenatal USG diagnosis and invasive procedures for conformation are gold standard[5]. Patient should be counselled for continuation or termination of pregnancy on the basis of Karyotyping.

If there is euploid foetus (foetus in normal), they could survive until birth. 90% foetuses with septated cystic hygroma had completely normal postnatal neurologic follow up. Suitable optimum treatment is surgery, sclerotherapy, laser, and radiofrequency is recent modalities of treatment. If foetus is aneuploid, outcome is poor. For that, counselling for termination of pregnancy should be done. In case of foetal demise, post-mortem and Karyotyping is advised.

## Conclusion

Early diagnosis and counselling of couple should be the first line of management. In first trimester, increased Nuchal translucency (NT) at 11-13 weeks, we should go for chorionic villus sampling or amniocentesis for chromosomal analysis, triple marker test should be considered. In our study the diagnosis of cystic hygroma at 15-18 weeks has a strong association with chromosomal abnormalities. In such cases for early diagnosis, couple should be given option for triple marker test and 11-week NT scan should be mandatory. It will help in early detection of chromosomal abnormality.

**Conflict of Interest: None**
**Source of Support: Nil**

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