

Cystic hygroma

Abstract

Cystic hygroma, also known as lymphatic malformation, is a congenital abnormality of the lymphatic system. It is characterized by the formation of fluid-filled sacs or cysts in the neck, axilla, or groin regions. These cysts are caused by the malformation or dysfunction of the lymphatic vessels and can lead to swelling and discomfort. Cystic hygroma can also be associated with chromosomal abnormalities, such as Turner syndrome or Down syndrome, and can have serious effects on an affected individual's health and quality of life. Treatment options for cystic hygroma include surgical excision, sclerotherapy, and laser therapy. In some cases, observation and close monitoring may be recommended if the cyst is small and asymptomatic. The prognosis for individuals with cystic hygroma varies depending on the size and location of the cyst, as well as the presence of associated anomalies.

This retrospective descriptive study aims to evaluate the incidence, presence of chromosomal anomalies and outcome of fetuses diagnosed with *cystic hygroma colli*.

Keywords: hygroma cyst • fetal • first trimester • prenatal ultrasound

Introduction

Cystic hygroma, or hygroma colli as it is commonly known is anatomically defined as a polycystic retrocervical and lateral mass ranging from a simple thickening of the posterior neck region to one or two large polycystic masses extending toward the scalp, back, and lateral portions of the fetal trunk[1].

From a physio-pathological point of view, the formation of hygroma colli is related to the sequestration of lymphatic fluid in the connective tissue of the fetal neck. It results on an abnormal fluid accumulation mainly detected by the ultrasound in the first trimester of pregnancy[2].

Karyotype abnormalities such as monosomy X, trisomy 21, trisomy 18 or 13 or others, as well as other morphological abnormalities like cardiac, diaphragmatic, skeletal or a set of syndrome are often associated with cistic hygroma.

Case Presentation

This is a retrospective observational study of records of patients hospitalized in the maternity departement of Menzel Temim for the management of cystic hygroma over a period from January 2020 to December 2022.

During this period, we reviewed 14 cases of cystic hygroma colli which had been prenatally diagnosed by fetal ultrasound. Maternal age at diagnosis, obstetric history, aneuploidy history as well as prenatal assessment was mentioned in the medical record. All patients underwent transabdominal and transvaginal ultrasonography.

Gestational age was determined from the last menstrual period and confirmed by measurement of the fetal crown-rump length (LCC) on a first trimester ultrasound. Between 11 weeks and 13 weeks and 6 days of gestation Nuchal Translucency (NT) measurements were performed.

The diagnosis of HK was made in presence of a retro-cervical anechogenic space bilateral (jugular sac) with one or more septamidline and paramedian sagittales forming lodges, HK thickness was measured in sagittal section (FIGURES 1 & 2).

Echographic abnormalities associated with cystic hygroma such as fetoplacental anasarca, organ malformations like cardiac, diaphragmatic, skeletal or a set of syndrome which are often associated with cistic hygroma were systematically investigated.

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All women were counseled about the association with chromosomal disorders and were offered fetal karyotyping by chorionic villous sampling at 11 weeks-14 weeks or amniocentesis from 15 weeks of gestation onwards. Alpha-fetoprotein was not routinely performed in our series.

Results

- Fourteen cystic Hygroma cases diagnosed by foetal ultrasonography fetal between January 2020 and December 2022.
- The average age of the patients was 36.45 (extremes: 29-45 years old).
- 14.3% were nulliparous or primiparous, 85.7% were more or equal to second parity.
- Consanguineous marriage represented 21.46% against 78.54% non-consanguineous
- A history of spontaneous abortion was found only in 21.42% of cases.
- Ultrasound discovery of Cystic hygroma occurs on average at 12 SA + 4j (extremes : 12SA+5j - 15SA)
- The thickness of the Hygroma was on average 7.1 mm (extremes: 5mm-8mm)
- Echographic abnormalities associated with cystic hygroma were systematically investigated for all patients in our study, 35.7% of fetuses had a fetoplacental anasarca (5 cases) and 7.14% had a pericardial effusion (One case) and the rest with no abnormality detected in the obstetric ultrasound.
- Karyotype testing was indicated for all patients included in our study and it was performed through transabdominal chorionic villus sampling or amniocentesis according to the term of pregnancy.
- An abnormal karyotype was detected in 85.68% of pregnancies; comparable rate to literature.
- Chromosomal abnormalities in order of frequency were:

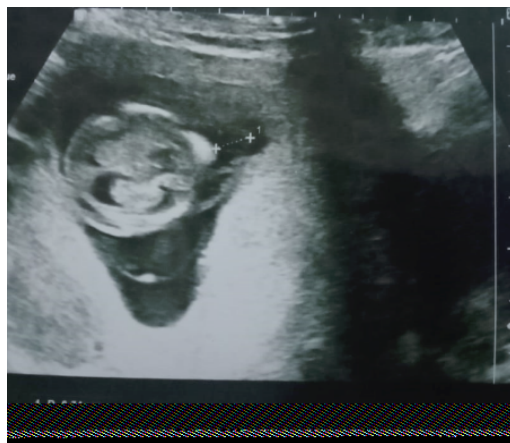


FIGURE 1: Sagittal ultrasound section showing a cystic hygroma.



FIGURE 2: Frontal ultrasound section showing a cystic hygroma.

- a. 49.98% Trisomy 21 (47XX+21) (7 cases)
 - b. 28.59% Turner's syndrome (45X) (4 cases)
 - c. 14.29% Trisomy 18 (47XX+18) (2 cases)
 - d. 7.14% Trisomy 13 (47XX+13)
- No fetuses with normal karyotype were found in our study.
 - Termination of pregnancy was performed at all patients in our study by medical method in 11 cases (78.54 %) and with aspiration for the rest.
 - Post-mortem findings confirmed the chromosomal abnormalities.

Discussion

Cystic hygroma is a congenital abnormality of the lymphatic system, characterized by the formation of fluid-filled cysts in various regions of the body [3]. This condition is a result of malformed or dysfunctional lymphatic vessels, which can lead to significant swelling and discomfort. In this literature review, the current understanding of cystic hygroma will be discussed, including its epidemiology, pathogenesis, clinical presentation, diagnosis, treatment options, and prognosis.

Cystic hygroma is a relatively rare condition, with an estimated incidence of 1 in 20,000 live births. It can occur in any race or gender, although there is a slightly higher incidence in females and in individuals with chromosomal abnormalities such as Turner syndrome or Down syndrome [4]. Cystic hygroma can affect different regions of the body, including the neck, axilla, or groin, with neck involvement being the most common presentation.

The exact cause of cystic hygroma is still not fully understood. However, it is believed to be related to a malfunction of the lymphatic vessels, which are responsible for the transport of fluid and waste products from tissues to the bloodstream [5]. In individuals with cystic hygroma, the lymphatic vessels are malformed, dysfunctional, or absent, leading to the accumulation of fluid in the affected area. This fluid accumulation results in the formation of a cyst.

The clinical presentation of cystic hygroma depends on the size and location of the cyst. Individuals with small, asymptomatic cysts may have no symptoms. However, larger cysts can cause significant swelling, discomfort, and pressure on surrounding structures. In some

cases, cystic hygroma can also cause respiratory distress, especially if the cyst is located in the neck region and compresses the airways [6]. Additionally, some individuals with cystic hygroma may have associated anomalies, such as chromosomal abnormalities or cardiac defects, which can further impact their health and quality of life.

The diagnosis of cystic hygroma is typically made through imaging studies, such as ultrasound or Computed Tomography (CT) scans. Ultrasound is the preferred initial imaging modality for cystic hygroma, as it is non-invasive, cost-effective, and can provide detailed images of the cyst and surrounding structures [7]. CT scans may be used to confirm the diagnosis and to evaluate the extent of the cyst and its relationship to surrounding structures.

Treatment for cystic hygroma depends on the size and location of the cyst, as well as the presence of associated anomalies. In some cases, observation and close monitoring may be recommended if the cyst is small and asymptomatic. However, larger or symptomatic cysts typically require intervention [2]. Treatment options for cystic hygroma include surgical excision, sclerotherapy, and laser therapy. Surgical excision involves the removal of the cyst and the surrounding tissue. Sclerotherapy involves the injection of a sclerosing agent into the cyst, which causes it to shrink. Laser therapy involves the use of a laser to destroy the cells that make up the cyst [8].

The prognosis for individuals with cystic hygroma depends on several factors, including the size and location of the cyst, the presence of associated anomalies, and the effectiveness of the treatment [5]. In general, individuals with small, asymptomatic cysts have a good prognosis, while those with larger or symptomatic cysts may have a more complicated course. Additionally, the presence of associated anomalies, such as chromosomal abnormalities or cardiac defects [9].

Conclusion

Cystic hygroma in fetuses is a congenital condition that can result in significant swelling and discomfort due to the accumulation of fluid in various regions of the body. It is caused by malformed or dysfunctional lymphatic vessels and can be diagnosed through imaging studies, such as ultrasound or computed tomography scans. The management of cystic hygroma in fetuses requires a multidisciplinary approach and may involve close monitoring, surgical intervention, or a combination of both. The prognosis for fetuses with cystic hygroma

depends on several factors, including the size and location of the cyst, the presence of associated anomalies, and the effectiveness of the treatment. Early detection and appropriate management can greatly improve the outcome for fetuses with cystic hygroma.

Competing Interests

No conflicts of interest

Authors' contributions

- Mhelheli Riadh, Hafsi Montacer: Data acquisition and data interpretation, article writing
- Ben Moumen Olfa, Gomri Emna, Hachicha Sarra: Article Writing
- Ragmoun Housseem: Supervision, correction and critical review of the article.

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