Bulletin of Environment, Pharmacology and Life Sciences
Bull. Env. Pharmacol. Life Sci., Spl Issue [4] November 2022 : 202-204

Online ISSN 2277-1808
Journal's URL:http://www.bepls.com
CODEN: BEPLAD

ORIGINAL ARTICLE **OPEN ACCESS**

Hydrops Fetalis with Cystic Hygroma: A Case Report and Its Embryological Basis

Garg S*[1], Agarwal S*[2], Saha S*[3]
*[1] Associate Professor, Dept. of Anatomy, SGT Medical College, Gurugram.
*[2] Associate Professor, Dept. of Anatomy, Al Falah Medical College, Faridabad
*[3] Professor, Dept. of Anatomy, SGT Medical College, Gurugram
* **Corresponding author**: Dr. Shilpi Garg, Associate Prof, Dept. of Anatomy, SGT Medical College, Gurugram. Email: mdshilpi@gmail.com

ABSTRACT
Cystic hygromas are congenital malformations of the lymphatic system that appears commonly in the head and neck area. They are usually the result of juglar lymphatic obstruction in which the normal communication between the juglar veins and the juglar lymphatic sacs fails to develop. Various genetic factors like SHH (sonic hedgehog) and VEGF (vasculo endothelial growth factor) are involved in regulation of vascular system including lymphatics. An 18weeks old male aborted fetus was examined for swelling in the neck region present in museum, Department of Anatomy, WCMSR&H, Jhjjar. There was generalized edema along with a cystic swelling of well-defined margins around 2×1.5 cm in the posterolateral compartment of the neck. This case report highlighted the cystic hygroma condition with hydrops fetalis. The diagnosis can be made early by routine antenatal USG screening method and genetic evaluation should be considered.
KEY WORDS: *Cystic hygroma, lymphatic obstruction, Hydrops fetalis, USG screening*

Received 12.11.2022 Revised 23.11.2022 Accepted 01.12.2022

INTRODUCTION

The Cystic hygromas are defined as congenital malformations of the lymphatic system. They appear as epithelium lined fluid filled membranous cyst. They present mostly in thehead and neck area with respect to long axis of fetus [1].In most cases of cystic hygroma, 70-80% of it appears in the neck and lower parts of face but can be present on other sites likeaxilla, superiormediastinum, retroperitoneummesentery, pelvis and peritoneum [2]. They areusually caused by obstruction in the juglar lymphatic system, failure of communication between the juglar veins and the juglar lymphatic sacs leads to the development of progressive lymphedema. Due to massive lymphedema hydrops fetalis condition can occur leading to intrauterine death of fetus. The incidence of cystic hygroma is 1 case per 6000-16000 live births with a survival rate of 2-6 % only [3].When cystic hygromas appears before 30th weeks of gestation it is associated with various chromosomal anomalies like Turner syndrome, Noonansyndrome, trisomies, fetal hydropsand various cardiac anomalies like coarctation of aorta and a left hypo plastic heart. Exposure to alcohol and various drugs including aminopterin and trimethadione duringintrauterine life are alsosaid to be associated with cystic hygroma. Cystic hygroma can be detected by abdominal USG as early as 10 weeks of gestation however transvaginal ultrasonography can provide better diagnosis over trans abdominal USG. [4, 5].

CASE REPORT

An 18weeks old male aborted fetus was examined for swelling in the neck region present in museum, Department of Anatomy, WCMSR&H, Jhjjar. Generalized edema along with a cystic swelling of well-defined margins around 2×1.5 cm in the posterolateral compartment of the neck was seen. Gestational age was determined by taking various parameters like C.R.L =16.2 cm, B.P.D =8.5 cm, F. L=2.8 cm. All the external features were examined for any gross deformity. All the facial features including eyes, nose, ear, lips were formed. Umbilical cord was normally present. No limb deformity was seen.

PATHOPHYSIOLOGY AND EMBRYOLOGY

Development of lymphatic vessels takes place after the formation of main vascular system including arteries and veins. Angiogenesis from the cardinal veins and subsequently proliferation of lymph angioblast are important developmental steps in formation of lymphatic system. The juglar lymph sac is first one to develop at the junction of subclavian vein and pericardinal vein. Any malformation of juglar lymphatics and their remodeling can result in increased accumulation of tissue fluid or lymphedema posterior to the cervical spine in neck regionleading to clinical condition called cystic hygroma [6].Various molecular factors are involved in the specification of arteries, veins and the lymphatic system after angioblast induction. SHH (Sonic hedgehog) molecular factor secreted by the notochord causes induction of surrounding mesenchyme to express VEGF(Vascular Endothelial Growth Factor).VEGF further induces PROX1 transcription factor for lymphatic vessel differentiation. Any aberrations in these factors can lead to the cascade of events leading to various vascular anomalies like cystic hygroma [7].

DISCUSSION

Cystic hygromas are congenital malformations and can be detected by USG in the first trimester of pregnancy.They are presented by single or multiple congenital cysts of lymphatic system and are usually associated with the Hydrops fetalis condition. Hydrops fetalis condition is characterized by accumulation of fluid in the serous cavities and oedema of soft tissues.Antenatal mortality rate is higher in fetuses having cystic hygroma and hydops fetalis condition [8]. 80% of cystic hygromas are present in the neck region [9].Cystic hygromas can have rare presentation on other unlikely sites likeaxilla and mediastinum [10].

In 60 – 70 % of the cases, cystic hygromas are associated with chromosomal anomalies like Turner's syndrome, Down's syndrome and various other trisomies. Identification of the nuchal ligament within the cyst is the characteristic sign in diagonosis of cystic hygroma via ultrasonography. Other diagnostic tool includesFISH (Fluorescent In Situ Hybridization) technique which helps in prenatal chromosomal analysis during intrauterine life [11].During pregnancy elevated alpha protein levels in amniotic fluid are also associated with Cystic hygroma [12]. Cystic hygroma with normal chromosome and without other associated congenital anomaly have a better prognosis and can have a spontaneous resolution before 20 weeks of gestation. The mortality rate is higher in fetuses having cystic hygromas associated with hydrops fetalis [13]. Cystic hygroma should be differentiated from other congenital anomalies like Encephalocoele, ocipito cervical meningomyelocoele and rarely posterior teratoma.

CONCLUSION

This case report has highlighted the pathophysiology of clinical condition cystic hygroma with hydrops fetalis. There is a need for better antenatal diagnostic tools along with genetic evaluation of fetus to prevent such adverse outcomes during pregnancy.

CONFLICT OF INTEREST

The authors declare that there are no conflicts of interest. The research received no specific grant from any funding agency in the public, community, or non-for profit sectors.

REFERENCES

1. Melissa S, Mancuso, Joseph Biggio. Foetal tumors. In: James, Steer, Weiner, Gonick, eds. (2012). High Risk Pregnancy Management Options. 4th ed. New Delhi: Elsevier; 387.
2. Pandit SK, Rattan KN, Budhiraja S, Solanki RS (2000) Cystic lymphangioma with special reference to rare sites. Indian J Pediatr 67: 339-341.
3. F. Cunningham, Kenneth Leveno, Steven Bloom, John Hauth, Dwight Rouse, Catherine Spong. Cystic hygroma. In: F. Cunningham, Kenneth Leveno, Steven Bloom, John Hauth, Dwight Rouse, Catherine Spong, eds. William's Obstetrics. 23rd ed. New York: McGraw Hill Professional; 2009: 356-357.
4. Papp C, Beke A, Mezei G, Szigeti Z, Bán Z, Papp Z. Prenatal diagnosis of Turner syndrome: report on 69 cases. J Ultrasound Med. 2006 Jun; 25(6):711-7; quiz 718-20.
5. Entezami M, Albig M, Knoll U, Gasiorek-Wiens A. Chromosomal disorders. In: Entezami M, Albig M, Knoll U, Gasiorek-Wiens A, eds. Ultrasound Diagnosis of Fetal Anomalies. 1st ed. New York: Thieme; 2003: 178-231.
6. Standring S. Gray's Anatomy: The Anatomical Basis of Clinical Practice, 41st Edition. Elsevier Churchill Living tone: New York, 2016: 205-209.
7. 7.Sadler. T.W: Langman's Medical Embryology, 12th edition. Lippincott Williams &Wilkins: Philadelphia; 2012:75-77.
8. Kiyota A, Tsukimori K, Yumoto Y, Hojo S et.al. Spontaneous Resolution of Cystic Hygroma and Hydrops in Fetus with Noonan's Syndrome. Fetal Diagn Ther. 2009:24(4): 499- 502
9. Merz E, Bahlmann F. Cystic hygroma. In: Merz E, Bahlmann F, eds. Ultrasound in Obstetrics and Gynecology. 2nd Ed. New York, NY: Thieme Medical Publishers; 2005: 23-29.

10. Brown LR, Reiman HM, Rosenow EC 3rd, Gloviczki PM, Divertie MB. Intrathoracic lymphangioma. Mayo Clin Proc. 1986; 61(11):882-92.
11. Cohen MM, Schwartz S, Schwartz MF, Blitzer MG, Raffel LJ, Mullins-Keene CL, et al. (1989). Antenatal diagnosis of cystic hygroma. Obstet Gynaecol Surv. 44(6):481-90.
12. Sorokin Y, Johnson MP, Drugan A, Koppitch FC 3rd, Evans MI. (1989). Amniotic fluid alpha-fetoprotein levels in the differential diagnosis of cystic hygroma. Fetal Ther; 4(4):178-84.
13. 13.Tanriverdi HA, Hendrik HJ, Ertan AK, Axt R, Schmidt W. (2001). Hygroma colli cysticum: prenatal diagnosis and prognosis. Am J Perinatol. 18:415-20.

CITATION OF THIS ARTICLE

S.Garg, S.Agarwal. S. Saha, Hydrops Fetalis with Cystic Hygroma: A Case Report and Its Embryological Basis. Bull. Env.Pharmacol. Life Sci., Spl Issue [4]: 2022: 202-204