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Fellow Column: Identification of a Solitary Posterior Cervical Cystic Hygroma: A Case-report

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Abstract

Purpose: To report a case of a post-term neonate with an isolated cystic hygroma upon birth.
Methods: This is a retrospective case report followed by clinical observation, genetic testing, and surgical intervention.
Results: A post-term neonate was found with an isolated left parapharyngeal cystic hygroma with no incidence of concurrent genetic syndromes upon karyotyping and underwent surgical removal.
Discussion: Cystic hygromas are very commonly associated with genetic syndromes or maternal risk factors. If none exist, specific subtypes of cystic hygromas can also be inherited in an autosomal recessive fashion, a possibility that may need to be explored comprehensively.

"Cystic hygroma, also referred to as cystic or nuchal lymphangioma, is a congenital malformation of the lymphatic system resulting in a benign lesion composed of single or multiple cysts that can manifest anywhere in the body. (1)"

Introduction

Cystic hygroma, also referred to as cystic or nuchal lymphangioma, is a congenital malformation of the lymphatic system resulting in a benign lesion composed of single or multiple cysts that can manifest anywhere in the body. (1) These lymphatic malformations are most commonly found in the cervicofacial regions, particularly in the posterior cervical triangle. Approximately 20% occur in the axilla, and rarely the mediastinum, groin, and retroperitoneum. (1)

Cystic hygromas are thought to arise from maldevelopment or complete failure of the lymphatic system to communicate with the remainder of the lymphatic or venous circulation. (1) These isolated lymphatic sacs dilate from fluid retention and develop a cystic morphology due to the lack of venous outflow. They usually occur in the fetal population, with most lesions presenting by two years of age. The incidence is estimated to be 1 in 6,000 to 16,000 live births, with 50-65% apparent at birth, others manifesting later. (1)

The majority of prenatally diagnosed cystic hygromas are associated with aneuploidies, including Turner Syndrome, Noonan syndrome, trisomy 21, 18, and 13. (1) Isolated cystic hygroma can also be inherited as an autosomal recessive disorder. In addition to congenital development, these lymphatic malformations can arise as an isolated event or because of environmental etiologies, including substance abuse during pregnancy, vertical transmission of maternal viral infection, trauma, inflammation, or obstruction of lymphatic outflow. (1)

If sufficient in size, cystic hygromas can be visualized by abdominal ultrasound imaging at 8-10 weeks gestation. Detection in utero may indicate the need for further investigation, including amniocentesis to assess for genetic abnormalities and Fast Spin MRI to determine the extent of invasion into the surrounding fetal structures. (1) It is not uncommon for these lymphatic malformations to be discovered postnatally with negative intrapartum imaging. Management of cystic hygromas after birth include cytogenetic studies to rule out suspected chromosomal and genetic disorders. Imaging such as MRI or ultrasound can be advantageous in confirming the diagnosis as well as determining the size and infiltration of the lesion into surrounding neurovascular structures, which can fundamentally guide future management. (1) The standard care of treatment is surgical excision, attempting to remove the malformation in its entirety, sparing the surrounding structures. The cystic hygroma composition of microcystic lesions makes it challenging to remove due to its association with nearby tissues, with surgical excision rates up to 53%. (1) The exceptions to excision at the time of diagnosis include premature infants that are too small to completely identify crucial nerves, including the facial nerve encompassed by the lesion. (1) If no airway or vascular obstruction is present, surgical intervention can be delayed until 2 years of age especially if the lesion is located around the parotid gland or facial nerve. (1)

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Case Summary

The neonate is a post-term infant born at 40 weeks and 2 days gestation to a 33-year-old G4P3 Hispanic female with a blood type of O positive. Birth weight was 3250 grams (11-25th percentile), head circumference 35 cm (26-50th percentile), and length 48 cm (4-10th percentile). Except for being GBS positive, the mother of the baby had an uneventful pregnancy and a normal prenatal ultrasound exam. She received prenatal care and took prenatal vitamins throughout the pregnancy. At the time of delivery, the neonate presented in the transverse lie, indicating the need for a Cesarean section. APGAR score was 9 at both 1 and 5 minutes. The infant was active and crying upon delivery, with a large fluctuant neck mass on the left that warranted admission to the Neonatal Intensive Care Unit (NICU) for observation and evaluation.

On admission, the neonate's temperature was 36.1 C, heart rate of 164 beats per minute, respiratory rate of 49 breaths per minute,

Figure 1. Large fluctuant cystic hygroma located in posterior cervical triangle.

and blood pressure 63/27. Physical exam revealed a neck mass extending from below the left jaw to just inferior and slightly posterior to the left ear. No respiratory difficulties, murmurs, extremity deformations, or other abnormalities were found. The infant received ad lib feeds of expressed breast milk or Enfamil formula by mouth. Sepsis work-up included CBC, CRP, and blood cultures. CBC revealed WBC of 13.3, hemoglobin 18.1, hematocrit 52.8, and platelets 323. CRP was less than 0.4, and blood cultures were negative. Genetic testing was ordered to rule out chromosomal abnormalities and showed normal karyotype findings.

"Genetic testing was ordered to rule out chromosomal abnormalities and showed normal karyotype findings."

The neck mass was evaluated by ultrasound the day after admission. The imaging revealed a complex cystic structure measuring 4.8 x 4.2 x 4.6 cm with low-level echoes and internal septations. Two days later, an MRI of the neck was performed and revealed a 4.9 x 7.9 x 5.2 cm trans-spatial cystic mass with a medially protruding component into the left parapharyngeal space. Internal septations and internal fluid-fluid levels suggested a large lymphatic malformation. A normal left parotid gland was not visualized. Due to the mass effect from the cystic mass, the left submandibular gland was displaced anteromedially. The infant underwent successful surgical resection of the neck mass one week later. The researchers acquired written parental informed consent before publishing this case study.

Discussion:

While many causes of cystic hygromas exist, they most commonly (about 62% percent of the time) manifest as part of a syndrome, including but not limited to Down, Turner's, and Noonan syndromes, especially if the hygromas form in utero early in the pregnancy. (1) However, this case proved to be different as the karyotype showed no associated anomalies in the neonate, as well as no early detection of the hygroma in utero, despite its presence and large size upon birth. As there were no maternal risk factors present during the pregnancy, such as alcohol abuse or viral infection, the case includes the possibility of a simple malformation of the lymphatics or an autosomal recessive inheritance of the hygroma.

Studies have shown that there may exist an autosomal recessive

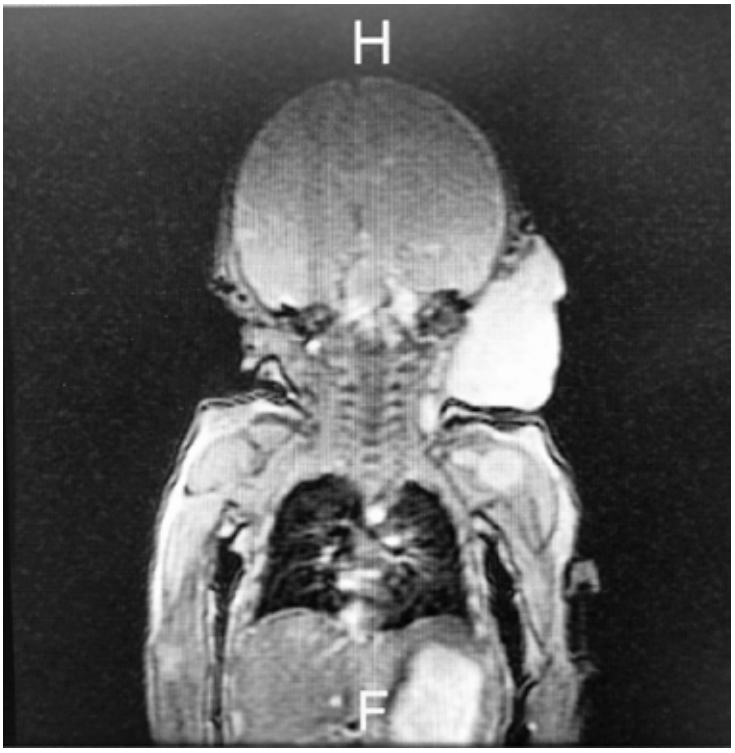

Figure 2. Upper body coronal MRI with contrast showing with cystic hygroma located in the posterior cervical triangle.

inheritance pattern of a familial nuchal subtype of cystic hygromas, which manifest less commonly without any associated fetal defects in 20-40% of the cases. (2) Similarly, it may also be possible that cervicofacial cystic hygromas, the most common manifestation of hygromas, may also be familial in cases without any other associated defects. Thus, it may become necessary to study further the cases of isolated cervicofacial cystic hygromas in families.

Once a cystic hygroma presents, they are often monitored for any signs of complications, such as hemorrhage, respiratory distress, infection, dysphagia. During infancy, a challenge of cystic hygromas is that the course is often unpredictable, as it may grow and cause the aforementioned complications or even spontaneously regress.(1) If they are asymptomatic, no treatment is usually needed or performed. (1) However, if they continue to enlarge, they may lead to airway obstruction and necessitate intervention. (3) In other cases, parents may also want to remove the hygroma due to aesthetics or disfigurement.

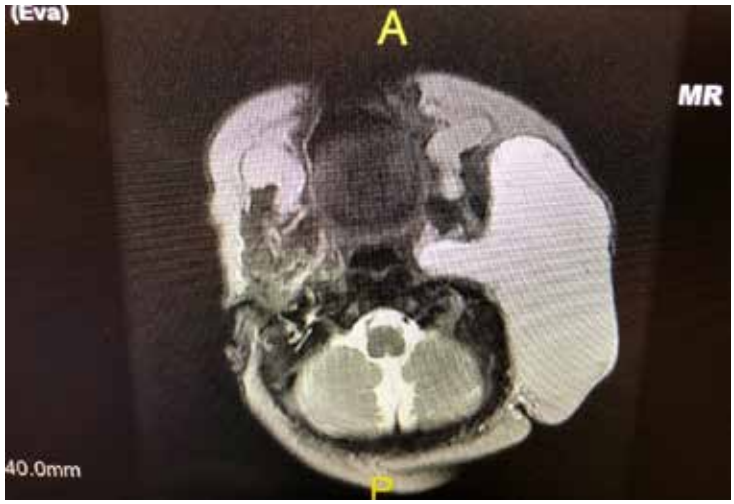

Figure 3. MRI with contrast showing lack of hygroma invasion into surrounding vasculature

Treatments currently used include surgical removal, drainage, sclerotherapy, or cauterization. (1) Aspiration is yet another form of treatment, but one that comes with a potential complication of necessary repetitive treatments or subsequent infection. (4) Sclerotherapy, a treatment that aims to shrink blood or lymphatic vessels via medications such as bleomycin, has become a popular choice for cystic hygromas due to its efficacy in eliminating the hygroma (5) but can potentially lead to complications such as discoloration of the skin, cellulitis, or rarely, an increase in the hygroma or a hard residual after shrinkage of the cysts, (1) In most cases, optimal treatment can be achieved by combining sclerotherapy with surgery or carrying out surgery alone. (1) Although surgery can also come with its own set of complications, such as facial nerve palsy or recurrence, many patients can be treated conservatively for complications and end with a good recovery. (6) However, in some instances, surgery may need to be delayed in instances of prior complications such as abscess formation, for which antibiotics would be administered and surgery delayed until 3 months. (1)

In this neonate, no complications such as respiratory distress or infection were perceived, and the decision to proceed to surgery was made. The neonate recovered well and was discharged without any further complications, with surgery proving to be a stable choice for treatment. However, as mentioned before, further evaluation and follow up would potentially be ideal for exploring the possibility of an inherited solitary cervicofacial cystic hygroma. The discovery of this inheritance pattern can lead to the development of further screening tools in utero, in addition to ones already established for cystic hygromas associated with genetic syndromes.

Conclusion:

This case presentation of a term neonate adds to the evidence for adequate surgical excision as a treatment for a cystic hygroma that was first identified after cesarean section, originally indicated for the transverse presentation. Though this case presented with a large cystic hygroma, the fact that it was not found prenatally is not particularly unusual. This case is an example of a good prognosis for cystic hygroma diagnosed after birth, as those found prenatally are more frequently associated with various malformation syndromes, karyotypic abnormalities, and various teratogenic agents. Furthermore, there may be a genetic basis for solitary cystic hygromas, potentiating the need for genetic testing for further exploration.

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