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FIRST-TRIMESTER DIAGNOSIS OF CYSTIC HYGROMA WITH SPINAL DYSRAPHISM: A RARE AND COMPLEX CO-OCCURRENCE.

Dr Nitika Sharma MS OBG

Dr Vivek Sharma MS Surgery

ABSTRACT

Background: Cystic hygromas and spinal dysraphism are rare congenital anomalies with significant perinatal implications. Their co-occurrence in a single fetus is extremely rare and, to the best of our knowledge, has not been previously documented. **Case Presentation:** A 26-year-old multigravida with 13+1 weeks' gestation presented for a routine antenatal checkup. Patient was advised ultrasound. The scan revealed a single live fetus with a large cystic hygroma surrounding the fetal head and trunk, as well as spinal dysraphism with kyphotic deformity and pleural effusion. No folic acid supplementation had been taken during early pregnancy. Multidisciplinary counselling was offered, with options for invasive testing and pregnancy management discussed. **Conclusion:** Early prenatal diagnosis of severe anomalies enables informed decision-making and counselling. Folic acid supplementation and routine anomaly scans remain pivotal in preventing and detecting such complex congenital malformations.

BACKGROUND

Among the lymphangiomas that occur most frequently are cystic hygromas.[1] Cystic hygromas are benign cysts that develop because of lymphatic system abnormalities. Although their precise embryonic origin is unknown, developmental abnormalities or cystic malformation of dilated lymphatic channels are thought to be the cause.[2] It is known that they develop from the remaining lymphatic tissue from embryos that has the capacity to divide.[3] They have been linked to chromosome aneuploidies, intrauterine mortality, and hydrops fetalis, among other disorders.[4] Meninges or neural components may herniate through a faulty neural arch caused by a range of congenital abnormalities in spinal dysraphism, resulting in a variety of clinical symptoms. They are separated into occulta (without an external lesion) and aperta (obvious lesion).[5,6] Depending on the pathological features, the following names are typically used: meningocele, myelomeningocele, lipomeningomyelocele, myeloschisis, and rachischisis. In rare instances, cystic hygroma may coexist with other structural anomalies such as spinal dysraphism, resulting in a complex clinical picture and potentially poor pregnancy outcome. Multidisciplinary management including detailed fetal ultrasonography, genetic counselling, and, when indicated, invasive diagnostic procedures is vital for optimal counselling and shared decision-making.

Case Presentation

A 26-year-old multigravida at 13 weeks and 1 day of gestation, presented for .antenatal checkup. Patient was advised routine obstetric ultrasound scan. Her last menstrual period was dated 05.01.2025, and the pregnancy was spontaneously conceived. She had not taken periconceptional folic acid supplementation during the first trimester. The patient's obstetric history revealed that this was her third pregnancy; however, further details of her obstetric past were not significant, and she had no history of previous congenital anomalies or miscarriage. The patient had been otherwise healthy without any significant medical or surgical history. No history of teratogenic drug use or infections during pregnancy was elicited. Routine antenatal tests including hemogram, blood grouping, infections screening, and fasting plasma glucose were within normal limits. On examination, her vitals were stable, and abdominal examination was unremarkable for her gestational age. The sonographic evaluation at 13+1 weeks revealed a single live intrauterine pregnancy with an estimated fetal weight of 70 grams. The fetal biometry — biparietal diameter (BPD) of 21.1 mm, femur length (FL) of 9.5 mm, abdominal circumference (AC) of 69.0 mm, and crown-rump length (CRL) of 57.6 mm — were consistent with the stated gestational age. Cardiac pulsations were present at 165 beats per minute, with regular fetal movements noted. The placenta was posterior and appeared Grade I in maturity. Amniotic fluid volume was adequate for the period of gestation. The internal os was closed. Ultrasound findings revealed significant congenital anomalies. A large, cystic, heterogeneously fluid-filled structure was seen in the subcutaneous tissue surrounding the fetal head, neck, and trunk. The findings were highly suggestive of a cystic hygroma. In addition, there was evidence of spinal dysraphism, characterized by a defect along the fetal spine with herniation of meninges suggestive of a meningocele or myelomeningocele. Kyphotic deformity of the spine was also noted at the thoracolumbar level. The fetal skull appeared well ossified; however, the nasal bone was not clearly visualized at this stage. There was also a mild right-sided pleural effusion in the fetus. No other obvious structural anomalies were noted at this scan, and the fetal heart appeared structurally normal for the gestational age. Given these findings — cystic hygroma, spinal dysraphism, and pleural effusion — the case was consistent with a severe congenital anomaly complex that is often associated with chromosomal and structural anomalies. Following a multidisciplinary counselling session, the couple was informed about the prognosis, further diagnostic options including fetal karyotyping, and potential pregnancy outcomes. Options for continuation versus termination of pregnancy were discussed in detail. The couple was advised genetic counselling and further targeted anomaly scans as per protocol. This case highlights the critical role of early first-trimester ultrasound in detecting congenital anomalies and underscores the importance of folic acid supplementation in the periconceptional period as a primary prevention strategy.

DISCUSSION

This case describes a rare and complex constellation of congenital anomalies — cystic hygroma and spinal dysraphism — detected at 13 weeks of gestation. To the best of our knowledge, there are no previously published reports describing the co-occurrence of these two anomalies in a single fetus, making this case particularly unique. The incidence of Spinal dysraphism is not clearly estimated. Due to advancements in antenatal care, folic acid supplements, high-resolution ultrasonography for prenatal screening, and biochemical indicators, the prevalence of spinal dysraphism has decreased globally in recent decades.[7] Cystic hygromas, arising from disrupted lymphatic development, are most seen in isolation or associated with chromosomal aneuploidies. They often herald a poor fetal prognosis, with an increased risk of miscarriage, hydrops fetalis, and adverse pregnancy outcomes.[8] The simultaneous presentation of these defects may reflect a multifactorial pathogenesis, potentially involving a shared disruption in early embryonic

morphogenesis. Despite their independent etiologies, the presence of both anomalies considerably worsens the prognosis. Early prenatal diagnosis is crucial for timely counselling and decision-making, especially in a pregnancy that is already at risk for chromosomal or structural anomalies.

CONCLUSION

The rare combination of cystic hygroma and spinal dysraphism presents a profound diagnostic and counselling challenge. Early ultrasonographic detection is invaluable for prognosis, parental guidance, and care planning. This case reinforces the need for folic acid supplementation in the periconceptional period to reduce the risk of neural tube defects and highlights the vital role of routine first-trimester ultrasound in detecting complex congenital anomalies at a stage where reproductive options can be offered to the parents.

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