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CASE REPORT

# Prenatal diagnosis of mediastinal neurenteric cyst: a case report and review of the literature

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## Abstract

Neurenteric cyst is a very rare developmental anomaly. Prenatal diagnosis of mediastinal neurenteric cysts has been reported rarely. We present a case of neurenteric cyst associated with vertebral anomalies diagnosed by prenatal ultrasonography at 31 weeks of gestation, which was treated successfully in the early neonatal period. In addition, we searched the English literature for all cases of mediastinal neurenteric cyst diagnosed in the prenatal period reported to date. We found that only 17 cases were reported previously. We reviewed the reports of these 17 patients along with our case, and we investigated the prenatal and postnatal diagnosis and treatment approaches and the factors influencing the prognosis. Fetuses with mediastinal neurenteric cysts should be monitored regularly by ultrasonography. Fetuses with no signs of hydrops are more likely to survive with proper neonatal center transfer, regular follow-up, and appropriate postnatal approach. Fetuses with hydrops findings have a high risk of fetal and neonatal death.

**Keywords** Mediastinal neurenteric cyst · Prenatal diagnosis · Thoraco-amniotic shunt

## Introduction

According to our systematic review in the English literature, all articles about neurenteric cysts were reviewed. There were just 17 of these articles in the literature [1–17] (Table 1).

Neurenteric cysts are posterior enteric residues that form a cystic mass in the posterior mediastinum. [4, 6, 9, 10, 12, 16]. These cysts can cause fetal hydrops, and if left untreated, hydrops causes fetal death in almost all cases [6, 8, 10, 12, 13, 16]. In addition, these cysts can cause severe respiratory problems in the postnatal period and may require respiratory support and emergency surgery [1, 8, 10, 18, 19]. If the diagnosis is determined at an early stage, deterioration of the surrounding structures may be avoided [5, 7, 8, 13]. Diagnosis of neurenteric cysts in the prenatal period allows time to inform the family, transport the patient to an appropriate neonatal surgical center, and increase the patient's survival by pre- and postpartum intervention [3, 5, 7, 8, 13]. In this presentation, we have reviewed all cases of mediastinal neurenteric cyst detected in the prenatal period reported in the literature along with our own case, and we tried to reveal prenatal diagnostic features, prenatal and postnatal approaches, and prognostic factors in these cases.

## Case report

A 20-year-old primigravida woman was referred to our hospital with the diagnosis of right-sided fetal cystic mass detected by fetal ultrasonographic examination at 31 weeks of gestation. Ultrasonography revealed a 25.7 × 27 × 32-mm hypoechoic, avascular, unilocular cystic mass in the right posterior fetal chest (Fig. 1). The cyst displaced the mediastinal structures anteriorly and to the left (Fig. 2). There were no signs of hydrops or lung hypoplasia. Although the

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**Table 1** Prenatally detected cases of mediastinal neurenteric cysts (in chronological order)

| Author | GA at diagnosis (weeks) | Size of cyst | Vertebral anomalies | Associated findings | Location | İntrautrine intervention | Treatment | Connection | Out come |
|---|---|---|---|---|---|---|---|---|---|
| Newnham [1] | 34 | 6.3 × 3 × 3.4 cm | – | The heart displaces to the left_ | Right HT PM | | PLT and resection of the cyst 5th day | NO | Good |
| Fernandes [2] | 22 | Large | Meningocele, tethered cord | Respiratory distress at 1st day | Right HT PM | | Resection of the cyst at 1st day | Jejenum | Good |
| Gulrajani [3] | 38 | 3 × 2 cm | C7-T1 vertebral defect kyphosis | No respiratory distress | Anterior Para spinal | | Resection of the cyst at 8th months | Spinal Canal | Good |
| Rizalar [4] | 32 | 4 × 5 × 5 cm | Cervical scoliosis, hemivertebra anterior spina bifida | Respiratory distress at 1st day | Right HT PM | | Resection of the cyst at 1st day | NO | Good |
| Daher [5] | 32 | 2 cm | C6, C7 fusion hemivertebra syringomyelia | Respiratory distress at 1st day | Right HT PM | | Resection of the cyst at 1st day | NO | Good |
| Macaulay [6] | 23 | 6.1 × 4.6 × 2.7 cm | C5-C6 block vertebrae T1-T2 cleft syringomyelia | Fetal ascites, hydrops | Right HT PM | Thoraco-amniotic drainage (25th week) + Shunt | Resection of the cyst at 3rd week | NO | Good |
| Perera [7] | 34 | 4.6 × 2.4 × cm | T3–4 hemivertebra | Increase in cyst size respiratory distress | Right HT PM | | Resection of the cyst at 2nd day | NO | Good |
| Wilkinson [8] | 28 | 5 cm | T1–3 hemivertebra | Fetal ascites hydrops Tracheomalasia right Bronchomalasia respiratory distress | Right HT PM | Thoraco-amniotic drainage (28–31st week) | Resection of the cyst at 4th day | Spinal Canal with 2 cm stalk | Good |
| Olavarria [9] | 35 | Large | Hemivertebra | Pulmonary hypoplasia, macrocephalus | Right HT PM | | – | Spinal Canal | Exitus |
| Uludağ [10] | 34 | 3.14 × 4.39 × cm | Scoliosis, hemivertebra | Bilateral caliectasia | Right HT PM | | Resection of the cyst at 4th day | NO | Good |
| Reisli [11] | 32 | 6 × 12 cm | – | Ductus thoracicus atrophy Respiratory distress at 6th month | Right HT PM | Prenatal aspiration (Thoracentesis) | Resection of the cyst at 6th month | NO | Good |

**Table 1** (continued)

| Author | GA at diagnosis (weeks) | Size of cyst | Vertebral anomalies | Associated findings | Location | İntrautrine intervention | Treatment | Connection | Out come |
|---|---|---|---|---|---|---|---|---|---|
| Aslan [12] | 32 | 2 × 3.6 × cm | Thoracal scoliosis | Increase in cyst size | Right HT PM | | Resection of the cyst at 14st day | NO | Good |
| Şahinoğlu [13] | 22 | 5 × 2.7 cm | – | – | Right HT PM | | Resection of the cyst at 1st day | NO | Good |
| Bernasconi [14] | 38 | 4 cm | Thoracal vertebral defect | – | Right HT PM | | Resection of the cyst at 3rd week | Spinal Canal | Good |
| Kimya [15] | 21 | 2.5 × 1.1 cm | T5–7 vertebral defect, split vertebra | – | Right HT PM | | – | Epidural Space | Abortion at 24th week |
| Gadodia [16] | 28 | 4 × 5 cm | Vertebral segmentation | – | Right HT PM | | Resection of the cyst at 2nd week | Spinal Canal | Good |
| Petrovic [17] | 13 | Two cyst 3–3.4 cm | – | Increase in cyst size | Left HT | | | Between the cyst | Abortion at 14th week |
| Çay | 31 | 2.7 × 3.2 × 2.7 cm | Hemivertebra butterfly vertebrae cervico-thoracal rotoscoliosis | Increase in cyst size respiratory distress | Right HT PM | | Resection of the cyst at 7th day | Closed adherent to the spinal column | Good |

*HT* hemithorax, *PM* posterior mediastinum, *GA* gestational age

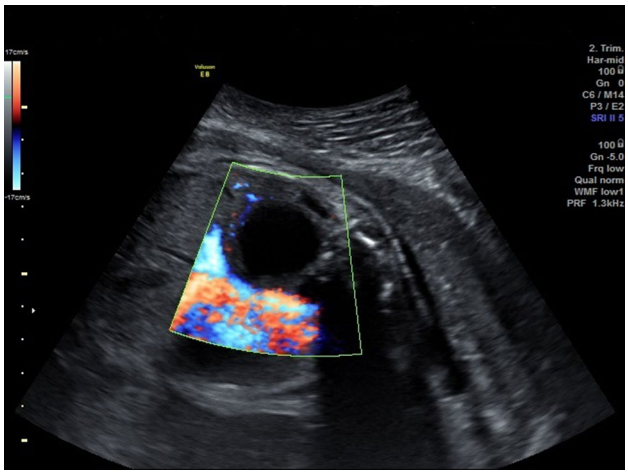

**Fig. 1** Transverse color Doppler image shows an anechoic cystic lesion (4 × 3 cm) located in the right posterior mediastinum, anterolateral to the dorsal spinal column and posteromedial to the heart. On color Doppler, no vascularity is present in the lesion

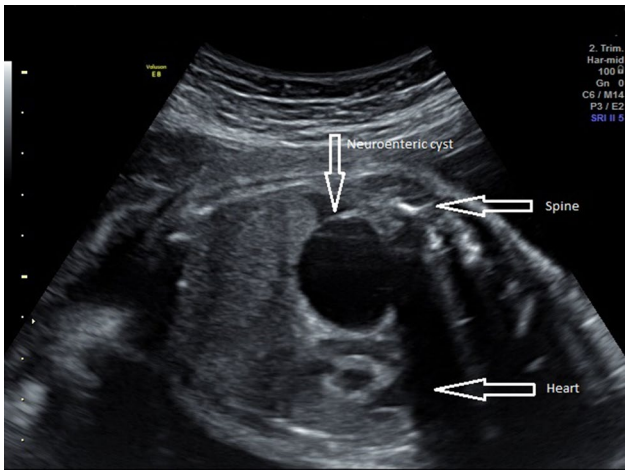


**Fig. 2** Prenatal Ultrasonography shows an anechoic cystic lesion located in the right posterior mediastinum, anterolateral to the dorsal spinal column and posteromedial to the heart

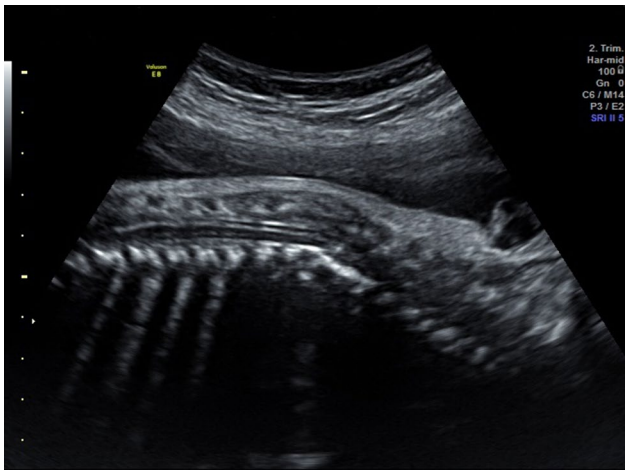

**Fig. 3** Sagittal spine sonogram demonstrates increased thoracic kyphosis and widened spinal canal

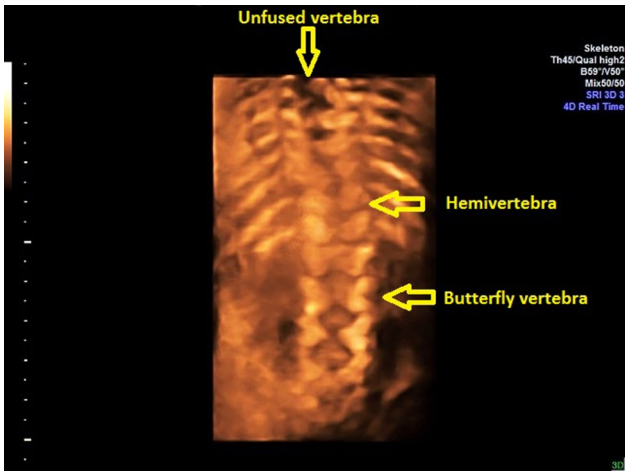


**Fig. 4** 3D ultrasound scan of fetal spine reveals vertebral segmental anomalies including hemivertebra, butterfly vertebra, and unfused vertebra at the thoracic region

differential diagnoses are fetal cystic masses, we strongly suspected a neurenteric cyst because there were numerous vertebral anomalies associated with the fetal mediastinal cyst (Figs. 3, 4). The patient was followed at 1–2 week intervals. A small increase in the size of the cyst was noted at the 36th week, and minimal pleural effusion was detected on ultrasonography at 37 weeks. It was decided that the baby should be delivered normally based on these developments, but cesarean delivery was performed because of respiratory distress during normal birth. The patient was followed up in the neonatal intensive care unit due to postpartum respiratory distress and respiratory support was applied.

When the condition was stable, thorax CT was performed, and the diagnosis was confirmed. There were many associated vertebral anomalies including rotoscoliosis, hemivertebra, and butterfly vertebra detected at the thoracocervical level (Figs. 5, 6). The patient was then taken for surgery for excision of the mass by posterolateral thoracotomy. The cyst, diaphragm, and mediastinum were in close relationship but easily separated. Despite the fact that there was a close proximity with the cyst to the spinal canal, no relation was found. Histopathology examination was compatible with neurenteric cyst (Fig. 7), and the patient was discharged without any problems on the 7th day post op. One-year follow-up was normal.

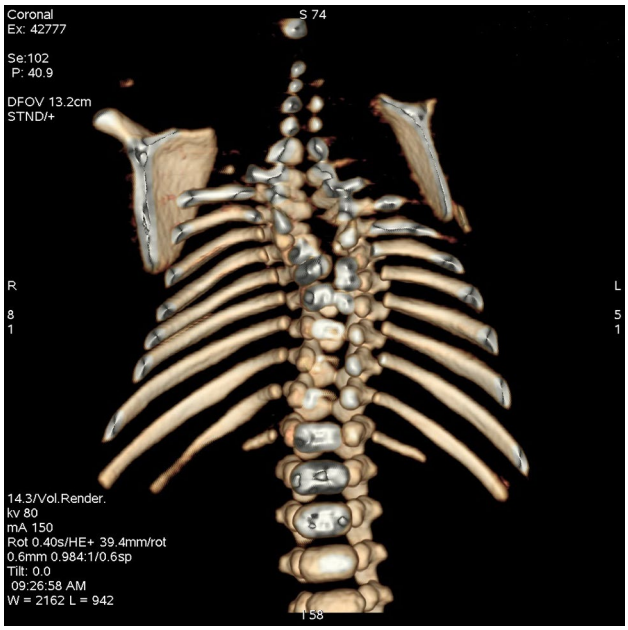


**Fig. 5** Coronal VR spinal image shows rotoscoliosis, hemivertebra, unfused vertebra and butterfly vertebra

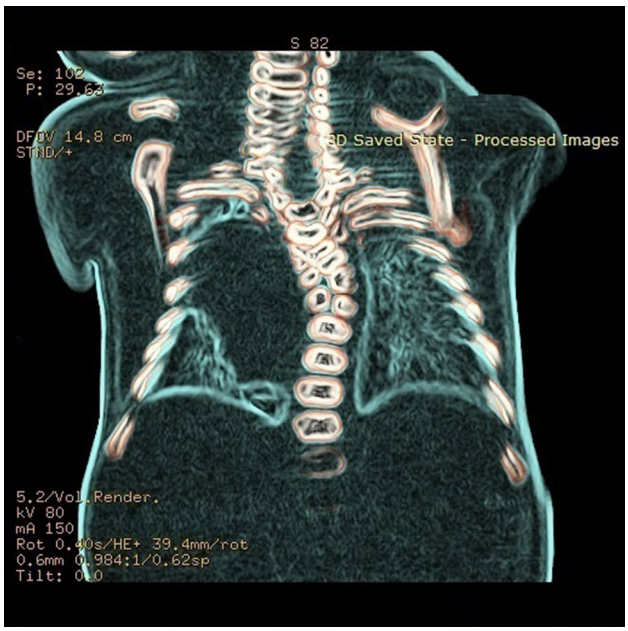


**Fig. 6** Thoracic CT demonstrates a thoracic cystic mass and rotoscoliosis, hemivertebra, unfused vertebra, and butterfly vertebra

## Discussion

Neurenteric cysts are uncommon posterior enteric remnants that result from incomplete separation of the notochord from the foregut in the third week of embryogenesis [4, 6, 9, 10, 12, 16]. The notochord begins forming

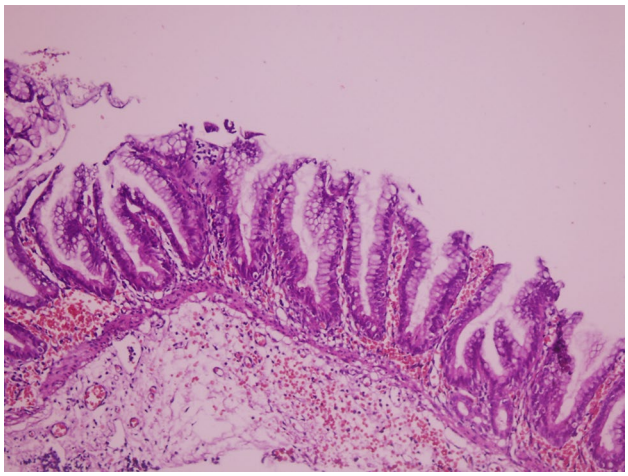

**Fig. 7** Cystic wall where columnar epithelium is observed with villous structures. (HE, ×20)

with the migration of ectodermal cells when the embryo is 18 days old [18]. As the notochord forms, it fuses with endoderm [10, 19, 20]. The vertebral bodies form around the notochord. If some endoderm becomes entrapped in the ectoderm, a split in the notochord develops, known as split notochord syndrome. This malformation is thought to be the cause of neurenteric cysts. This helps to explain the common association of neurenteric cysts with vertebral body anomalies [20]. The association of such cysts with abnormalities of the spinal column is well documented and can range along a spectrum from mediastinal masses with minimal spinal abnormality to lesions completely within the dura with no extraspinal components [2, 7, 19]. Most of the cases examined here had vertebral anomalies (77.7%). There was also a relationship with the spinal canal in seven cases. Successful surgery was completed for these, but cerebrospinal fluid fistula in one patient caused hydrothorax, but it improved with 3 weeks of drainage.

Neurenteric cysts are most commonly located in the mediastinum 90% posteriorly and 66% on the right side [4, 6, 8–12, 14], as in the 16 cases presented here.

The differential diagnosis of a cystic mass in the fetal chest includes macrocystic adenomatoid malformations, pulmonary sequestration, bronchogenic cysts, duplication cysts, diaphragmatic hernia, teratoma, and neurenteric cysts [6, 8, 14, 15]. Neurenteric cysts can often be distinguished by the virtually pathognomonic association with vertebral anomalies. These include hemivertebra, butterfly vertebrae, cleft vertebral bodies, spina bifida, absent and fused vertebrae, vertebral segmentation, and scoliosis [4, 6, 7, 9, 10, 12, 14].

Ultrasonography has been the cornerstone of prenatal diagnosis for this lesion. If a fetal thoracic mass is detected, serial ultrasonography is required to assess for cyst growth, mediastinal shift, and the development of hydrops [8].

Prenatal intervention should be performed in cases where hydrops develops. If hydrops does not develop, prolonging pregnancy to term is acceptable [8, 18].

The problems that may arise in the prenatal period due to mediastinal neurenteric cysts depend on the size of the cyst and its secondary physiological deterioration [4, 8, 10–13, 18, 19]. A large cyst may cause mediastinal shift, hypoplasia of normal lung tissue, polyhydramnios, fetal hydrops, and cardiovascular complications leading to death [4, 8, 10–13, 18, 19]. Hydrops manifests itself in the prenatal period as fetal ascites, pleural and pericardial effusions, and skin and scalp edema [4, 8, 18].

Adzick et al. [18] have suggested that there are four treatment options for hydrops-causing cysts: (1) thoracentesis (2) thoraco-amniotic shunt, (3) fetal delivery (term or preterm), and (4) in utero resection. Thoracentesis and thoraco-amniotic shunt placement for hydrops are performed with the intention of prolonging pregnancy to term and resecting the lesion postpartum. Thoracentesis is only transiently effective [8, 11, 18]. The general experience with fetal cystic lesions is that the intracystic fluid rapidly reaccumulates [4, 8, 11, 18]. In three patients, fluid was collected again after intervention. Although migration and occlusion can occur, a hydropic fetus at 30 weeks of gestation should be delivered and the mass resected ex utero. Theoretically in utero resection would be reserved for hydropic fetuses younger than 30 weeks of gestation [18].

Complications related to cyst growth in the intrauterine period occurred in six of 18 patients examined here. Three of these six patients were prevented from developing hydrops by intrauterine interventions such as thoracentesis and thoraco-amniotic shunt on the appearance of hydropic changes during prenatal follow-up, and postnatal treatments were performed. In the other three patients, no action was taken in the prenatal period and these patients died. Two patients were aborted based on a decision by the ethics committee and one patient died due to lung hypoplasia during the early postnatal period. In three patients who developed hydrops during the intrauterine period, it was possible to give birth by preventing cyst growth with thoracentesis and thoraco-amniotic shunt. Intrauterine aspiration has a critical prognosis in terms of prevention of hydrops development of cyst growth. Intrauterine cyst aspiration was not performed in the three patients who died. If the cyst was minimized by intrauterine intervention, maybe it would have been possible for these cases to survive.

Postnatal symptoms due to a mediastinal cystic mass are a reflection of its size and proximity to other structures [8, 18, 19]. Many infants become symptomatic shortly after birth as the lesion fills with fluid, expands, and compresses vital surrounding structures [8, 10, 18, 19]. Signs and symptoms of neurenteric cysts are related most frequently to the compressive effect of the mass on the airway [8, 10, 12, 16, 18, 19]. Dyspnea, cough, stridor, and respiratory distress are noted frequently. Respiratory symptoms, presence of a mediastinal mass, and a vertebral anomaly form a triad present in 70% of patients. Occasionally, lesions are asymptomatic and are incidentally found on chest radiography [8].

Nine of the sixteen living patients had respiratory distress symptoms, one of whom was lost due to respiratory distress as a result of lung hypoplasia. Urgent intervention was required, and cyst excision was performed in the other eight patients, six of whom underwent surgery in the first few days of life. Two patients, who had received intervention in the intrauterine period, had no early postnatal respiratory distress, and these cases were discharged, but they returned with severe respiratory distress due to cyst regrowth on the 20th day and at 6 months, respectively, and had to undergo urgent surgical intervention. In this way, respiratory distress, often only after a few days of life, is a common presentation of thoracic neurenteric cysts. According to us, patients who develop hydrops in the intrauterine period and need intervention should not be discharged in the postnatal period, and cyst excision should be performed as early as possible.

For the rest of the patients, cyst resection was performed in 1–21 days (average 9.8 days) in six cases. In one case, cyst resection was performed in the 8th month.

If a mediastinal cystic mass is detected on prenatal ultrasonography and accompanied by vertebral anomalies, it is most likely to be a neurenteric cyst. Mediastinal neurenteric cysts should be carefully monitored for cyst growth, hydrops, and pulmonary hypoplasia development. The follow-up interval may be 1–2 weeks. This follow-up should be done more frequently in patients with hydrops findings and intrauterine intervention. Hydrops findings such as scalp and skin edema, ascites, and pleural effusion in the prenatal period, thoracentesis and thoraco-amniotic shunt should be tried to prevent hydrops development. Cyst excision should be performed in the early postnatal period in patients with respiratory distress in the postpartum period, especially in patients requiring intrauterine intervention.

## Compliance with ethical standards

**Ethical statements** This article does not contain any studies with human or animal subjects performed by any of the authors. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1964 and later versions. Informed consent was obtained from all patients for being included in the study.

**Conflict of interest** Ali Çay, Semih L Mirapoğlu, İbrahim Aydoğdu, and Hüseyin Toprak declare that there are no financial or other relations that could lead to a conflict of interest.

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