Case Report

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Spinal Hamartoma with Severe Kyphoscoliosis – A Rare Case of Spinal Mass in a Fetus

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Key Words
Spinal mass · Spinal hamartoma · Neural tube defect · Spinal dysraphisms · Teratomas · Kyphoscoliosis · Prenatal diagnosis · α-Fetoprotein levels

Abstract
Spinal hamartomas are rare lesions consisting of disorganized ecto- and mesodermal tissues of the spinal region. While postnatal identification of spinal hamartomas has been reported, a literature search did not reveal any published reports of prenatal identification of spinal hamartomas. Here we report a 46,XX fetus who presented at 20 weeks' gestation with a lower thoracic and lumbar kyphoscoliosis, suspected spina bifida, and amniotic fluid α-fetoprotein (AFP) levels within the normal range. Interestingly, autopsy at 22 weeks revealed a lumbosacral spinal hamartoma with kyphoscoliosis. We discuss the differential diagnosis for such spinal masses which includes congenital tumors and spinal dysraphism. This case illustrates that spinal hamartomas should be considered as part of the prenatal differential diagnosis of spinal dysraphisms, especially in the presence of normal AFP levels.

Introduction

Hamartomas are benign tumor-like lesions composed of mature, well-differentiated tissues arranged in a disorganized fashion, which can arise in almost any organ [1]. Spinal hamartomas are rare, and to our knowledge, prenatal diagnosis of a spinal hamartoma has not been previously described [1]. Here we report a fetus with a lumbosacral spinal hamartoma with kyphoscoliosis.

Case Report

A 29-year-old woman (gravida 1, para 0) presented with an abnormal fetal ultrasound at 20 weeks + 2 days which showed a severe scoliosis in the lower thoracic spine with a lesion suspected to be an open neural tube defect. As this was the first fetal ultrasound for this pregnancy, no previous scans were available for comparison. Spina bifida could not be ruled out with certainty by ultrasound given the limited visualization of the fetus due to severe kyphoscoliosis. A repeat ultrasound at 20 weeks + 4 days showed a kyphotic low thoracic and lumbar spina bifida (fig. 1a) as well as pericardial effusion and a small aorta. The fetal echocardiogram was normal. Amniocentesis was performed for analysis of fetal karyotype (46,XX) as well as amniotic fluid α-fetoprotein levels (AF-AFP) which were within the normal range [6.8

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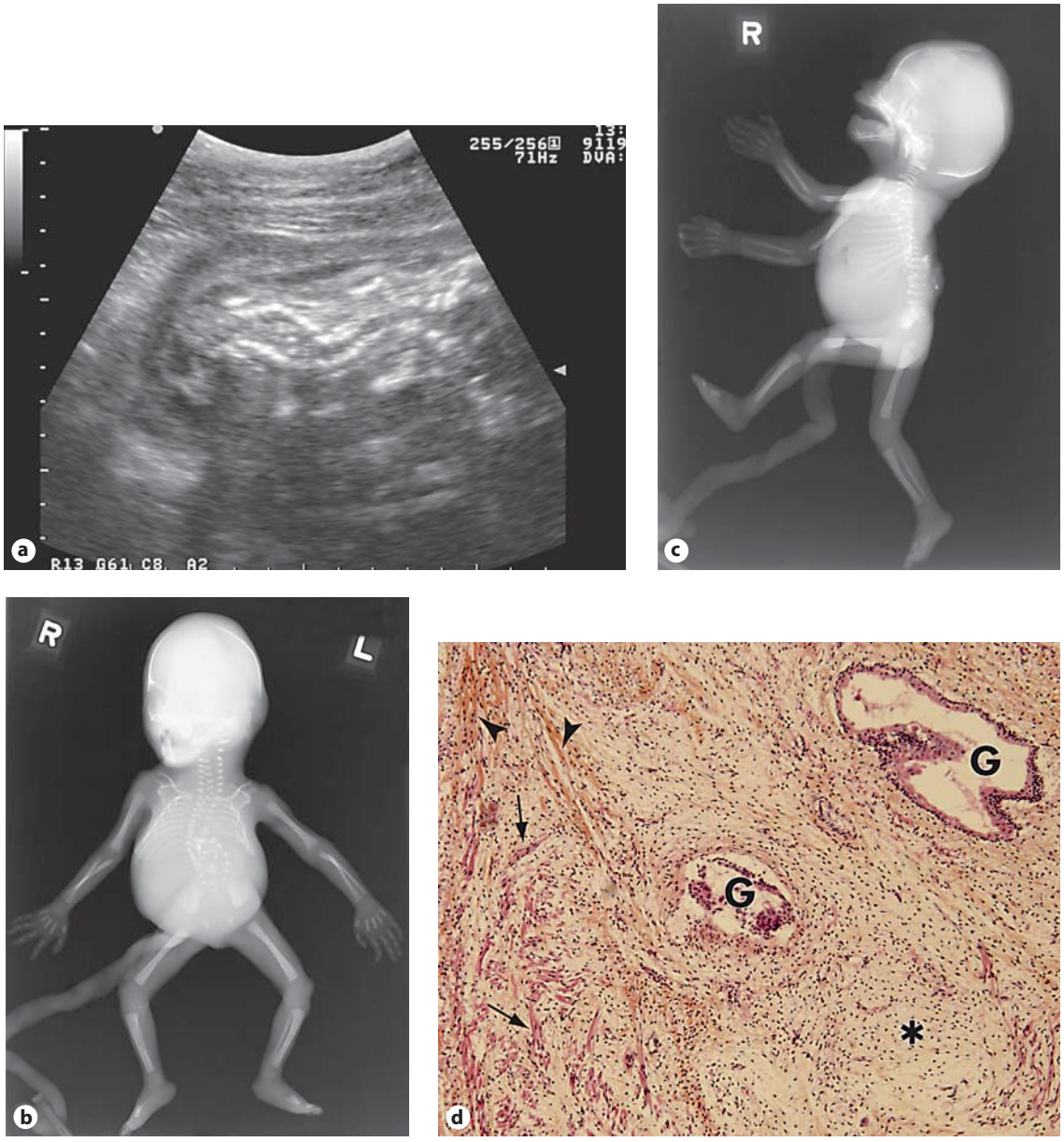

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Fig. 1. a Ultrasound at 20 weeks' gestational age. Longitudinal view with head on the right. Abnormal spine with severe mid-lumbar kyphosis and splaying of the vertebral bodies. Cerebral structures were normal (not shown). **b** The skull and extremities appear normal for gestational age. Abnormalities of the spine are present starting from the cervicothoracic junction and extending down to the mid-lumbar level. The abnormalities consist of hemivertebrae, as well as incomplete posterior elements at the thoracolumbar junction, as well as partially fused vertebrae and widening of the interpediculate distances from mid-thoracic spine down to the lumbar area. These are in keeping with presence of a neural tube defect. There are also abnormalities of the left side of the ribcage, where at least partial fusion of mid-thoracic ribs is suspected. This type of ribcage deformity can be encountered in cases of congenital scoliosis. **c** The lateral x-ray shows subtle kyphosis at the thoracolumbar junction with prominence and irregularity of the soft tissues posteriorly, as well as calcifications within these posterior soft tissues, which could dystrophically be associated with the spinal dysraphism and/or due to presence of calcific elements in the hamartoma at the level of the defect. Given the size of the fetus, the spine is also slightly shorter than expected. **d** Intermediate power photomicrograph (orig. magnif. 200×) of representative area of the hamartoma stained with hematoxylin-phloxine-saffronin. In a background of non-specific mesenchyme (asterisk), there are two glands (G). There are also bundles of skeletal muscle fibers (arrows). A few bundles of collagen fibers (arrowheads) are also present.

mg/l (0.96 MoM) at 20 weeks + 5 days: normal up to 3 MoM]. Based on the AF-AFP levels and the ultrasound findings, a closed spina bifida was suspected and, given the severity of the anomaly, the prognosis was likely to be poor. An MRI had been arranged to attempt to better visualize the spine, however, the couple decided to terminate the pregnancy prior to the MRI and the termination was performed at 22 weeks.

At autopsy several anomalies were identified: external findings included nuchal edema, lumbosacral kyphoscoliosis with normal overlying skin and a relatively shortened trunk (crown-rump length: 16.5 cm, normal ± range at 22 weeks: 19.8 cm ± 9.6 mm: crown-heel length: 26.5 cm, expected length at 22 weeks: 27.4 ± 2.5 cm). Internal findings included lumbosacral spinal hamartoma with kyphoscoliosis and an absent left kidney (subsequent parental kidney scans were normal). Radiological examination showed abnormalities of the spine and thoracic cage (fig. 1b, c) including areas of calcification which were confirmed within the hamartoma at autopsy. Neuropathological examination demonstrated an intraspinal lesion at the level of the kyphoscoliosis composed of a disorganized mixture of normal spinal and paraspinal tissues, including portions of spinal cord, notochordal islands, skeletal muscle, connective tissue, and glandular epithelium (fig. 1d), features which are typical of a spinal hamartoma. There was no evidence of spina bifida or of a meningo-(myelo)cele. The hamartoma filled the entire spinal canal, which was widened as a result with the greatest cross section being approximately 7 × 4 mm which significantly enlarged compared to the normal diameter of 2–3 mm. The size and extent of the hamartoma made it very likely that this was the causal factor for the vertebral anomalies as opposed to an incidental lesion.

Discussion

Spinal hamartomas are rare lesions consisting of mature and well-differentiated spinal and paraspinal tissues arranged in a disorganized fashion. Histological findings in spinal hamartomas can include nerves, fat, muscle, cartilage, bone, abnormal vessels, glands, synovial membranes, lymphoid tissue, and urinary tract tissue [2]. Spinal hamartomas may occur in the thoracic, lumbar, or sacral regions, and can present with a skin dimple, cutaneous angioma, subcutaneous mass, or normal overlying skin [2]. Formation of spinal hamartomas probably occurs from incomplete and premature disjunction of the neural tube and ectoderm and they are mostly sporadic – in this case the left renal agenesis is likely to be an incidental finding [3]. The renal agenesis was not detected on the ultrasound, and it is well established that renal agenesis may not be seen prior to 20 weeks due the expansion of the adrenal gland into the empty space. The nuchal fold measurement at 20 weeks + 6 days was 4.5 mm so either the nuchal edema was not visualized at the time of the scan or this was an artifact at autopsy. Hamartomas have been identified in patients with neurofibromatosis type 1, however these lesions are distinct from congenital lesions as they are composed of glial cells, ganglion cells, disoriented axons and vessels [4, 5]. Hamartomas have also been reported in association with spinal dysraphism [6, 7]. Furthermore, scoliosis has been associated with spinal dysraphism but not with spinal hamartomas [8, 9].

The postnatal differential diagnosis for spinal masses includes congenital tumors and spinal dysraphism. The most common congenital tumor in the differential diagnosis is the teratoma and these are usually located in the sacrococcygeal region, although spinal canal teratomas have been described [8, 10]. Teratomas arise from displaced germ cells, contain tissues from all three germ layers (ecto-, meso- and endoderm), and may be benign or malignant [10, 11]. Conversely, spinal hamartomas are composed of only ecto- and mesodermal tissue, and do not have malignant potential [2, 5]. As the histological findings of the spinal lesion in this case included glandular epithelium, a component of the endoderm, the distinction with teratoma is important. The lack of destruction of surrounding tissues and relatively organized nature of a limited spectrum of tissue components of this lesion are more consistent with the conventional description of a spinal hamartoma, and thus it is appropriate to describe it as such. Moreover, in their series of midline hamartomas, Tibbs et al. [2] identified glandular epithelium in at least 1 of the cases. Other congenital tumors in the differential diagnosis include dermoids and epidermoids, which contain only ectodermal tissues [5, 10]. Unlike spinal hamartomas, spinal dysraphisms arise from incomplete fusion of the posterior neuropore and they include myelomeningocele [12, 13], lipomyelomeningocele [14], myelocystocele [15], and meningocele [16]. Spinal dysraphisms may be identified prenatally by ultrasound combined with the measurement of AFP levels. AFP levels >3.0 MoM suggest the presence of spinal dysraphism, with a sensitivity of 85%. The normal AFP levels in this patient reduced the likelihood of an open spinal dysraphism, although a closed spinal dysraphism could not be ruled out.

The prognosis for spinal hamartoma is variable; surgical excision of the hamartoma is usually performed to prevent neurological damage and to correct the lesion. Following surgical excision, neurological function may range from normal to significantly impaired and partial paralysis of lower extremities, bowel and bladder dysfunction may occur. Midline hamartomas are not associated with hydrocephalus or Chiari malformations [8, 16]. Based upon the information available prenatally, it was

believed that the prognosis for our case was likely to be poor. A neurosurgical opinion was sought and they agreed that the chance of an adverse neurological sequence was believed to be high; the severe kyphoscoliosis would require major surgery and it was very likely that lower motor function and bladder and/or bowel function would be impaired. Had the couple chosen to continue the pregnancy, an MRI would have been performed to provide more information and assist in predicting the postnatal prognosis. The couple would have also been counseled regarding early neurological surgical interventions for their neonate.

Conclusion

While postnatal diagnosis of spinal hamartoma has been described, based on our literature review, no cases of prenatally diagnosed spinal hamartomas have previously been reported. The findings of this case suggest that spinal hamartomas, although rare, should be considered as part of the differential diagnosis of spinal dysraphisms, especially in the presence of normal AFP levels and cerebral structures.

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