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CASE REPORT

Congenital kyphoscoliosis associated with thoracic hemivertebrae in a 9-month-old Racking filly

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Summary

Thoracic hemivertebra is a rare congenital vertebral malformations in horses resulting in vertebral column deformity with or without neurological signs. A 9-month-old Racking filly was presented with a distinct lump over the back region. This lump was apparent at birth and had been increasing in size. Clinical examination revealed painless kyphoscoliosis over the thoracic vertebrae with mild hindlimb ataxia and gait abnormality. Plain radiographs of the vertebral column showed 13th–16th thoracic hemivertebrae. Myelography was performed under general anaesthesia. The ventral and dorsal aspects of the contrast medium column were narrowed at the level of the 13th–14th thoracic hemivertebrae. The contrast medium was not seen caudally, suggesting spinal cord compression. Little information about hemivertebra in horses exists, and a better understanding of hemivertebra aetiology is required.

KEYWORDS
horse, congenital abnormality, hemivertebra, kyphoscoliosis, spinal cord compression

INTRODUCTION

Several developmental vertebral defects are described in horses, with cervical vertebral stenotic myelopathy (CVSM) and occipitoatlantoaxial malformation (OAAM) being the commonest (Crochik et al., 2009; Fürst, 2018; Unt & Piercy, 2009). However, other congenital malformations of the vertebral column including atlantoaxial instability, block vertebrae, butterfly vertebrae, hemivertebrae and spina bifida occur rarely in horses (Fürst, 2018). Developmental abnormalities of the vertebral column occur due to a failure of normal formation of vertebral formation, segmentation, differentiation or abnormal union of structures during embryologic development stages (Castriota-Scanderbeg & Dallapiccola, 2005; Fürst, 2018). Hemivertebra is a condition in which asymmetric vertebral body formation occurs, so a part of the vertebral body is deficient (Bertram et al., 2019; White & Goldberg, 2012). Depending on hemivertebra location and severity, it may be associated with moderate-to-severe angulation of the normal vertebral column alignment with or without neurologic gait deficits (Fürst, 2018; Haussler, 2018). This case report describes the clinical and radiographic findings of congenital thoracic hemivertebrae in a 9-month-old Racking filly.

CASE HISTORY AND CLINICAL FINDINGS

A 9-month-old 153 kg Racking filly with a distinct bump over the back region was referred to the Surgery Section of the Veterinary Teaching Hospital of the Ferdowsi University of Mashhad. According to the owner, this lump was present since birth and increased in size over time. There was no history of trauma. On physical examination, the filly was alert and her body condition was normal. The respiratory rate, heart rate and temperature were within normal limits. The physical examination revealed abnormal curvature of the vertebral column along the sagittal plane (kyphosis) with mild scoliosis (kyphoscoliosis) over the thoracic vertebrae (Figure 1). Examination of the filly's back revealed no pain in this region. No forelimb deficits or abnormalities of the cranial nerves were identified. The cervical

range of motion was normal. Very mild ataxia and abnormal gait in the hindlimbs were observed on examination (grade 1/5). No urinary or faecal incontinence was reported by her owner.

A blood sample was obtained and haematology and serum biochemistry profiles were evaluated. Haematological parameters were within the normal range. Biochemical analyses showed an increase in aspartate aminotransferase (AST: 393; reference range: 226–366 U/L) and creatine kinase (CK: 1083; reference range: 108–430 U/L). Plain and contrast radiography was carried out. A myelogram was performed under general anaesthesia. For this purpose, the filly was sedated with 1.1 mg/kg of xylazine (20 mg/mL; Alfasan) intravenously, followed by 0.1 mg/kg of diazepam (5 mg/mL; Zepadic®; Caspian Tamin Pharmaceutical Co.). Anaesthesia was induced by 2.2 mg/kg ketamine (100 mg/mL; Alfasan) intravenously and the filly was intubated orotracheally. Anaesthesia was maintained with isoflurane (AErrane®; Baxter Healthcare Corporation). Myelographic evaluation was performed with the cisterna magna technique (Butler et al., 2017). The area was aseptically prepared. The head was maintained in a flexed position, and an 18G spinal needle was placed in the atlantooccipital space. About 50 mL of cerebrospinal fluid (CSF) spontaneously was slowly withdrawn. A similar volume of iodixanol 270 mgI/mL (270 mgI/mL; VisipaqueTM; GE Healthcare Inc.) was then injected slowly over 3 minutes. Before imaging, the head was maintained in an elevated position. Multiple hemivertebra between the 13th and 16th thoracic vertebrae and several abnormal ribs were apparent on plain lateral radiographs. The proximal aspect of several ribs over ribs 13–18 was irregular in shape and diameter, and there was decreased space between ribs on the left side (Figures 2 and 3). No evidence of any significant abnormality in the contrast medium column was seen in lateral and flexed radiographs of the cervical region. In the thoracic region, the ventral and dorsal aspects of the contrast medium column were narrowed at T13–T14 and failed to flow beyond this location, indicating extradural compression of the spinal canal in these thoracic regions (Figure 4). Collected CSF was evaluated and the results were in the normal range. Various aspects of this disorder, including its progression, treatment options (conservative and surgical) based on human cases and veterinary literature, outcomes, prognosis and animal welfare, were fully discussed with the owner, but the owner refused clinical advice. Follow-up was obtained from the owner by phone 1 month after presentation, who reported that the filly's clinical signs had not changed. Unfortunately, the owner did not respond to subsequent follow-up requests.

FIGURE 1 Clinical appearance of thoracic kyphoscoliosis in the presented filly.

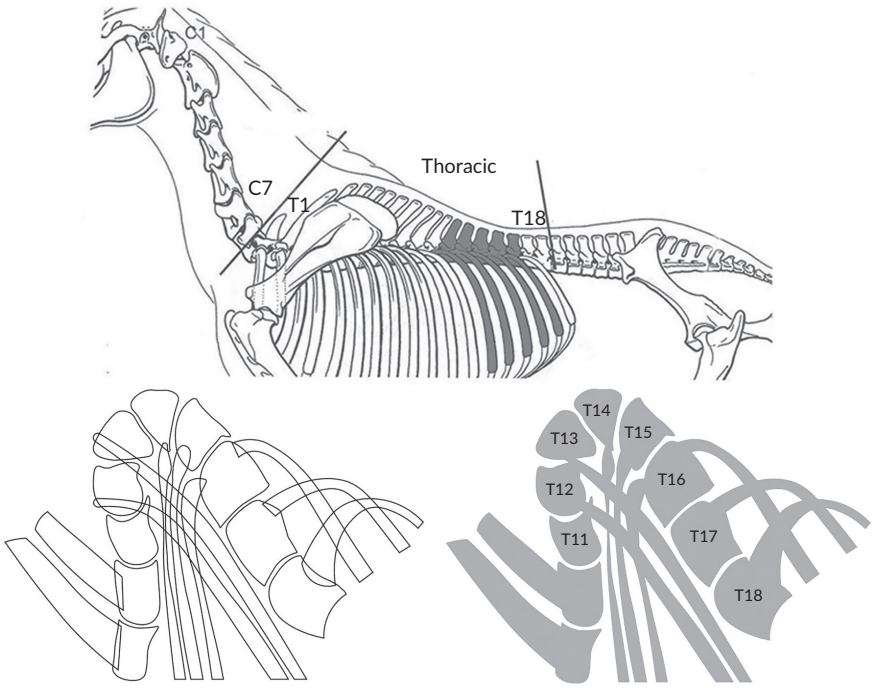

FIGURE 2 Schematic representation of the abnormality in vertebrae noted. Grey shadow areas show abnormal shape of the 12th–16th thoracic vertebrae and wedge shape in body of the 13th–16th thoracic vertebrae.

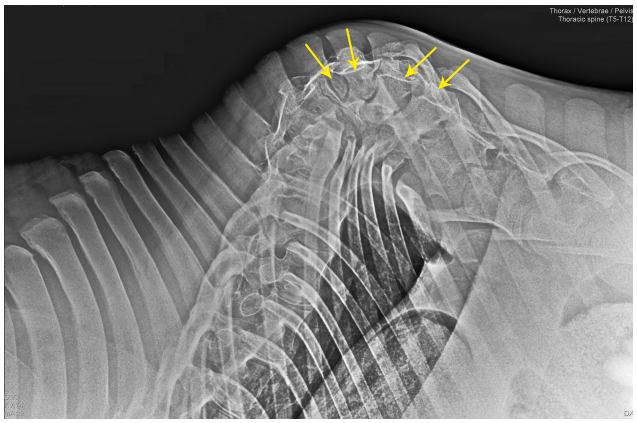

FIGURE 3 Laterolateral radiograph of the thoracic vertebrae in the mid to caudal-thoracic region. Kyphosis shows in the caudal-thoracic region with hemivertebrae in the 13th–16th thoracic vertebrae (arrows).

DISCUSSION

In horses, developmental vertebral disorders commonly cause malformations of the cervical vertebrae (de Heer & Nout, 2011; Dorner et al., 2022; Rendle et al., 2008; Unt & Piercy, 2009; Wong et al., 2005) while thoracic and lumbar vertebral malformations are infrequent (de Heer & Nout, 2011; Wong et al., 2005). Vertebral malformations can be congenital (present at birth) or acquired (later in life; Wong et al., 2005). Hemivertebra is one of the congenital vertebral malformations. Failure of chondrification centres during the formation or the segmentation in the somitogenesis stages and subsequent disruption in ossification during vertebral body formation has been proposed as the pathogenesis of hemivertebrae (Besalti et al., 2005; Goldstein et al., 2005; Johal et al., 2016; Powel et al., 2022; Varras & Akrivis, 2010). Improper formation of the intersegmental arteries of the vertebral column and, therefore, alternation in vascular supply at the developmental stage is postulated as another aetiology for hemivertebrae (Besalti et al., 2005; Goldstein et al., 2005; Johal et al., 2016; Powel et al., 2022; Varras & Akrivis, 2010). Hemivertebrae can result in deviations of the vertebral column in the coronal plane laterally (scoliosis), sagittal plane dorsally (kyphosis) or ventrally (lordosis; Denoix, 2005; Fürst, 2018).

In humans, hemivertebra is a rare congenital malformation, and its incidence is estimated at 0.05%–0.1% and is more common in females (Shah et al., 2020; Varras & Akrivis, 2010; Xu et al., 2020). In animals, hemivertebra is the most common congenital malformation in dogs (De Rycke & Saunders, 2017; Ryan et al., 2017; Schlensker & Distl, 2016; Wyatt et al., 2018). The breed-related incidence of hemivertebra in dogs has been investigated. Hemivertebra is reported to be common in screw-tailed brachycephalic breeds, with a prevalence range from 78% to 94% (Davitkov et al., 2020; Wyatt et al., 2018).

A study, which described vertebral lesions in 202/443 horses (38.6%), reported only 15 horses (2.9%) with vertebral malformations, including scoliosis, lordosis and kyphosis being identified (Jeffcott, 1980).

Only a few cases of hemivertebra in horses have been reported previously. According to these and the present case, the horses affected by hemivertebra are of various breeds, including the Quarter (Wong et al., 2005), Friesian (de Heer & Nout, 2011) and American saddle horses (Kirkberger & Gottschalk, 1989). To our knowledge, this is the first reported case of thoracic hemivertebra in the Racking horse. Previous case reports of hemivertebra in horses were restricted to males, including geldings and colts (de Heer & Nout, 2011; Kirkberger & Gottschalk, 1989; Rendle et al., 2008; Wong et al., 2005). The current case is unusual in being a female foal (a filly). Given the few published case reports, it is not possible to identify sex or breed predisposition or incidence of hemivertebra in the horse with any confidence.

In agreement with the case reported by Wong et al. (2005), rib abnormalities were observed in our case. This finding is often noted as an incidental finding on radiography or in post-mortem specimens and does not usually contribute to neurological signs (Denoix, 2005; Wong et al., 2005). Hemivertebrae have been found to be associated with other congenital musculoskeletal abnormalities, including those affecting ribs, limbs, neural tube and extra-musculoskeletal tissues. The latter include abnormalities of the heart, genitourinary system, central nervous system and gastrointestinal tract. These other lesions may arise due to anatomically unbalanced growth as a consequence of the hemivertebra and associated abnormalities in structural differentiation of tissues surrounding the vertebral column (Bohiltea et al., 2022; Chaturvedi et al., 2018; Moser, 2005; Varras & Akrivis, 2010; Yang et al., 2020). As the ribs develop closely with vertebrae during gestation, the same developmental failures that lead to hemivertebrae are attributed to rib abnormalities (Fischer & Degenhardt, 2008).

Structural abnormalities of the vertebral column may affect the spinal cord and a neurological examination should be conducted (Kaplan et al., 2005), although neurological deficits are variably present (Denoix, 2005; Wong et al., 2005). It has been noted that horses with deviations of the thoracolumbar vertebral column without neurological deficits can be used normally, although these horses are predisposed to intervertebral osteoarthritis (Denoix, 2005). Unlike the thoracolumbar region, hemivertebrae and deviations of the cervical vertebral column are usually accompanied by neurological deficits (Denoix, 2005). Kyphoscoliosis of the thoracolumbar vertebral column (T7-L4) due to malformation of T1–T15, including hemivertebra with no neurological deficits were reported in a 1-month-old American saddle foal (Kirkberger & Gottschalk, 1989). A 9-month-old Quarter gelding with severe hindlimb ataxia and paresis was reported to be associated with multiple thoracic malformations of T4–T8, including hemivertebrae, kyphosis and fused dorsal spinous processes (Wong et al., 2005). In another case report, a 5-day-old Friesian colt that presented for kyphosis at the level of T18–L3 was affected by neurological deficits, including pelvic limb paraparesis and severe ataxia (de Heer & Nout, 2011). Acquired kyphosis due to previous compression fractures of T14–T18 in association with mild hindlimb ataxia was described in a 20-month-old Tennessee Walking horse (Kothstein et al., 2000). Kyphosis of the

thoracolumbar vertebral column with a thoracic neurenteric cyst and butterfly vertebrae at T6 and T7 was reported in a 7-month-old mixed breed colt associated with severe neurologic deficits (Rendle et al., 2008). The filly reported in this paper showed a mild neurological deficit in its hindlimbs that may have been due to compression and narrowing of the spinal cord at T13. It has been described that several factors, such as breeds, the severity of kyphosis, hemivertebra subtype and the presence of other pathologic conditions, may be associated with the presence or absence of neurological signs in dogs (De Decker et al., 2019). However, the development of clinical signs related to hemivertebra in horses is not entirely documented.

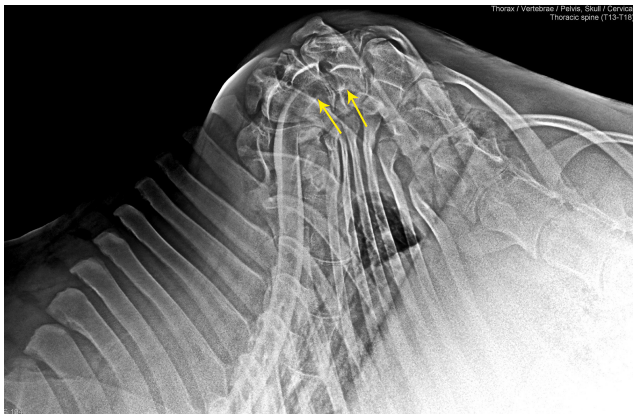

FIGURE 4 Laterolateral radiograph of the caudal-thoracic vertebrae showing the abnormal myelogram. Note the narrowing of the ventral contrast medium column at the level of the 12th thoracic vertebra. The ventral and dorsal contrast medium columns are narrower at the level of the 13th–14th thoracic vertebrae (arrows), and there is no sign of contrast medium distal to the 14th thoracic vertebra suggesting spinal cord compression.

Some of the thoracic vertebral malformations reported in the literature are summarised in Table 1.

Increased AST and CK concentrations were observed in the case presented here. Spinal misalignment can alter body biomechanics and muscle loading, and it has been shown that kyphosis may be associated with several muscle impairments (Briggs et al., 2007; Fasser et al., 2021). Although an elevation in serum CK along with AST can indicate muscle damage, other factors may affect these enzymes, such as transportation (Harris et al., 1990; Satué et al., 2022; Wessely-Szponder et al., 2015).

The aetiological factors causing vertebral malformations have not been fully explained in horses. Still, genetic and heredity factors, dietary imbalances, toxic factors, hormonal changes, environmental causes, trauma and in-utero insult (oxygen deficiency, increased temperature and carbon monoxide) may be responsible (Dorner et al., 2022; Fürst, 2018; Kirkberger & Gottschalk, 1989; Kothstein et al., 2000; Unt & Piercy, 2009; Witzmann et al., 2014; Wong et al., 2005). An interaction between genetic susceptibility of the developing embryo or fetus and these factors probably exists (Witzmann et al., 2014). It has been mentioned that alteration in gene expression, especially members of the Hox family, may contribute to malformations of the axial skeleton and vertebrae (Seki et al., 2008; Unt & Piercy, 2009). Hox genes play an essential role in the body plan along the anterior–posterior axis and specify the morphology and patterning of the vertebrae (Böhmer et al., 2015; Kappen, 2016; Mallo et al., 2010). Their function in patterning the vertebrate body plan is highly complex (Böhmer et al., 2015; Mallo et al., 2010). It has been suggested that defects in molecular and cellular signalling pathways related to Hox genes, including Wnt signalling pathways, retinoic acid and fibroblast growth factor, as well as variations in the spatial expression of Hox genes may result in malformations and abnormalities of the axial skeleton and

TABLE 1 Review of reported thoracic abnormalities in horse.

Signalment	Abnormality	Clinical sign	Outcome	Reference
One-month-old American saddle foal	Kyphoscoliosis, malformation of T7-L4, hemivertebra of T15	No neurological deficits	Euthanised	Kirkberger and Gottschalk (1989)
Thirteen-month-old Akhal-Teke colt	Severe malformation of T4	Neurological deficits	Euthanised	Johnson et al. (1997)
Twenty-eight-month-old American Saddlebred gelding	Severe malformation of T4	Neurological deficits	Euthanised	Johnson et al. (1997)
Three Haflinger newborn foals	Midthoracic lordosis, malformation of T7–T10 in one foal and malformation of T7–T11 in another foal	Not reported	Euthanised	Coates and McFee (1993)
Twelve-month-old Tennessee Walking horse	Acquired kyphosis, compression fractures of T14–T18	Neurological deficits	Euthanised	Kothstein et al. (2000)
Nine-month-old Quarter gelding	Kyphosis, malformation of T4–T8, hemivertebrae of T7	Neurological deficits	Euthanised	Wong et al. (2005)
Seven-month-old cross-breed colt	Kyphosis, butterfly vertebrae of T6 and T7, thoracic neurenteric cyst	Neurological deficits	Euthanised	Rendle et al. (2008)
Eleven-month-old Chilean Caballo Raza Chilena colt	Malformation of C5, C6, and T1, cervical spondylolisthesis	Neurological deficits	Euthanised	Dorner et al. (2022)

vertebrae during sclerotome differentiation, chondrification and ossification (Mallo et al., 2010; Unt & Piercy, 2009). However, the role of genetics, heredity and other factors of the vertebral column abnormalities and hemivertebra in horses are generally considered unknown, probably because of the condition's rarity. Although hemivertebra was present at birth in the present case here and genetics may explain the abnormality, the exact aetiology remains unclear as it is the case with previous hemivertebra case reports (de Heer & Nout, 2011; Wong et al., 2005). In the case presented here, the filly's parents had no vertebral column abnormalities, and the owner did not know about other previous generations. Also, this foal had no known exposure to toxic substances or trauma.

In humans, early diagnosis, conservative treatment and corrective surgical techniques including hemivertebra resection are recommended before developing neurological signs or severe deformity and disability in cases with scoliosis and kyphoscoliosis (Crostelli et al., 2022; Han et al., 2011; Oksanen et al., 2021; Ruf et al., 2006; Xia et al., 2022; Yang et al., 2020). Still, there is no treatment for this condition in horses (Fürst, 2018). In dogs, surgical management of cases with clinical signs associated with hemivertebra and vertebral column malangulation can be challenging. There are only a few reports of such cases in the veterinary literature (Charalambous et al., 2014).

Congenital thoracic hemivertebra in horses is a rare anomaly that may come along with a spinal deformity resulting in congenital scoliosis and kyphosis. This condition may also cause spinal cord compression and neurological signs. Rib anomalies may occur in association with hemivertebrae. The actual incidence and prevalence of thoracic hemivertebra in horses are unknown, and future reports are required to better understand the different aspects of such an abnormality.

AUTHOR CONTRIBUTIONS

The first three authors contributed to the clinical examinations and interpretation of the clinical data. All the authors contributed to the preparation of the manuscript, table and figures. All authors approved the final version of the manuscript.

CONFLICT OF INTEREST STATEMENT

No conflicts of interest have been declared.

FUNDING INFORMATION

No funding was received.

ETHICS STATEMENT

This case report details the management of a clinical case that was a part of the clinical caseload. High veterinary care has been performed with the consent of the animal owner. All identifying information has been removed.

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