

TRANSVERSE TERMINAL HUMERAL BILATERAL HEMIMELIA IN A DOG – CASE REPORT

[*Hemimelia transversa terminal umeral bilateral em um cão – relato de caso*]

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ABSTRACT – Hemimelia is a rare congenital anomaly, characterized by the absence of a part or the whole distal segment of a body member. It is usually unilateral and is classified according to the affected bone segments. In the transverse terminal hemimelia, there is the absence of the whole distal part of a body member, what is defined as congenital amputation. Several studies about cats and dogs concluded that radial hemimelia is the most usual. However, other types of this anomaly are rarely observed in dogs. Therefore, the aim of this study is to report a case of transverse terminal humeral bilateral hemimelia in a mixed-breed dog. In the physical exam was observed a bilateral malformation of the thoracic members. The radiographs revealed the presence of a well-developed scapula, humeral vestiges and the absence of other appendicular segments on the right and left antimeres, what made the hemimelia diagnosis possible. Consanguinity was the probable cause of the anomaly. Because of the scarcity of scientific reports about this pathology in dogs, further studies are needed to investigate the here observed.

Keywords: canids; congenital amputation; bones.

RESUMO – Hemimelia é uma rara anomalia congênita caracterizada pela ausência de parte ou todo o segmento distal de um membro, de ocorrência geralmente unilateral e classificada de acordo com os segmentos ósseos afetados. Na hemimelia transversa terminal toda a porção distal de um membro está ausente, sendo definida como amputação congênita. Diversos estudos em cães e gatos reportam a hemimelia radial, sendo esta a forma mais frequente. Não obstante, raras são as abordagens em cães acerca das demais apresentações da hemimelia. Assim, este estudo objetiva relatar um caso de hemimelia transversa terminal umeral bilateral em um cão mestiço. No exame físico, observou-se má formação dos membros torácicos. As imagens radiográficas revelaram nos antímeros direito e esquerdo a presença de escápula desenvolvida, resquícios umerais e ausência dos demais segmentos apendiculares, permitindo assim o diagnóstico de hemimelia. Como causa provável constou apenas o fator de consanguinidade. Devido escassez de abordagens científicas acerca deste tipo de patologia em cães, demais estudos são necessários para investigar o aqui observado.

Palavras-Chave: canídeos; amputação congênita; ossos.

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INTRODUCTION

Dysostosis are a group of bone deformities characterized by the abnormal development of a bone or a part of it (Towle; Breur, 2012). Hemimelia is a rare bone dysostosis of the appendicular skeleton (Ferreira et al., 2012; Campos et al., 2014). It is classified as terminal when part or the whole intermediary and distal bones are absent until a certain point (called congenital amputation); intercalary, when part or the whole intermediary bones are absent. However, in the last type, it is possible to find bone segments, proximal or distal to the change (Towle; Breur, 2012).

Both intercalary as terminal hemimelia can be categorized in transverse when occurs an absence of all the distal skeletal elements, and longitudinal, when the losses are extended parallelly along the axis of the body member. The diagnosis is made by radiographic testing (Kay et al., 1974; Towle; Breur, 2012; Pollard; Wisner, 2015).

The anomaly can be uni or bilateral. The unilateral is the most usual one. In dogs, radial hemimelia is the most common type, what causes deformity of the affected body member and as a result, it triggers severe functional limitations (Mendes; Rausch; Carapeto, 2004; Towle; Breur, 2012; Pollard; Wisner, 2015).

Since many dysostosis are not diagnosed, or yet diagnosed but not reported, it is hard to ascertain its exact incidence in cats and dogs (Towle; Breur, 2012). Nevertheless, it is estimated that 1 to 2% of puppies are born with some congenital malformation, a part of them being detected at birth and the others identified only several days or weeks after birth (Pollard; Wisner, 2015).

Among the cases of malformation, there are the use of teratogenic drugs, infectious, chemistry and physical agents and finally, consanguinity. In general, the fetal anomalies can be a result of an unbalanced diet during the pregnancy, drug administration or viral infections (Johnson, Kenneth and Watson, 2014; Riaz et al., 2016). However, the

etiology of hemimelia is still unknown. As it is a rare anomaly and there are few approaches about the subject, the knowledge about the disease is limited (Peres et al., 2013). In this way, the purpose of this study is to describe an unusual case of humeral bilateral hemimelia, diagnosed in a dog, by radiographic testing and classified as transverse terminal.

CASE REPORT

A three months old mixed-breed dog was lead to clinical care, weighing 3.2kg. The owner intended to receive medical veterinary guidance about dealing with the malformation of the animal body members, what has been present since the birth.

During the medical history, the owner reported that in the same litter (of an amount of two puppies), just this dog presented changes. He also informed that the puppies are sons of consanguineous parents (mother and son). In addition, he stated that it was not observed occurrence of malnutrition or diseases during the pregnancy, neither exposition to radiation, drugs or chemical agents throughout this period. There was no report of changes related to the animal general state of health. And the body condition score had a good evaluation.

In the physical exam was observed a bilateral malformation of the thoracic members (Figure 1 - A). There was no evidence of pain, crackles or vestiges of fractures in the thoracic region which corresponded to the deformity. The dog moved itself in a limited way: using the impulse of the hindlimbs and slide the thoracic region on the floor. A radiographic testing was performed on the dorsoventral and latero-lateral directions of the thoracic region, which made possible to observe the presence of a well-developed scapula, humeral bilateral vestiges and bilateral agenesis of the fore appendicular segments (Figure 1 - B, C). The thoracic members hemimelia was classified in transverse terminal bilateral, according to Towle; Breur (2012). A complete blood count (CBC) was also performed, which presented normal ranges. Still, an abdominal ultrasound was performed as well, although did not demonstrate changes.

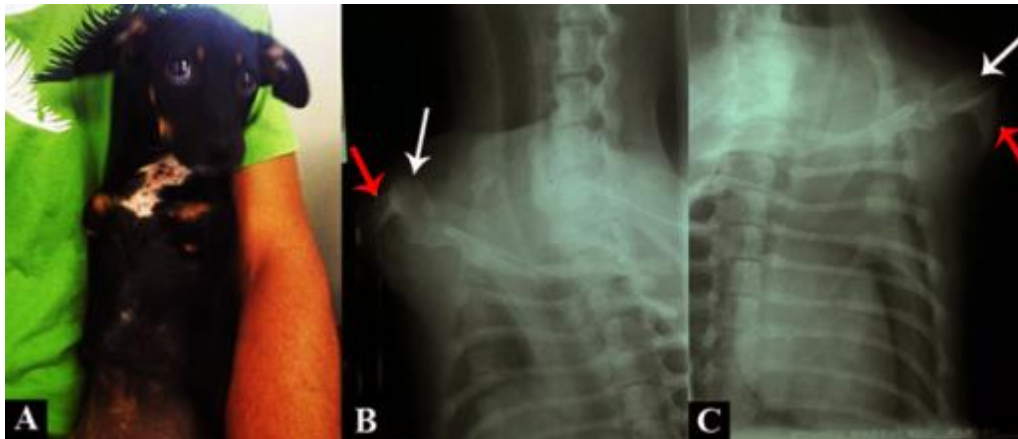


Figure 1. A) A three months, mixed-breed dog with transverse terminal humeral bilateral hemimelia. B) Radiographic image in the dorsoventral position, right antimerer, and left antimerer (C). Both images demonstrated the presence of a well-developed scapula (white arrows) and bilateral vestiges of the humerus proximal end (red arrows). Source: Author's collection.

After the clinical evaluation and an analysis of the tests, it was possible to observe that the animal had been undergoing conservative treatment, since it presented a good quality of life, despite the orthopedic change. The owner was oriented to acquire a wheelchair-like prosthesis due to the inability of the forelimbs and to castrate the animal in its adult phase, considering that the anomaly is related to heredity. Along a year, every three months, the dog was monitored. At six months old, in clinical evaluation, the owner informed that he had not acquired the prosthesis due to financial issues. Nevertheless, the animal was able to move itself even without the prosthesis, a sign of a considerable adaptation to the absence of the body

members. The moves observed since then were based in the underpinning of the body weight on the hindlimbs when moving to short distances (about 2 meters), with an intermittent maintenance of the balance. In the next evaluation, the owner reported that the dog's locomotion had improved. During this period, when the animal was six months old, it was castrated. When the animal was one year old, it was detected a better underpinning of the body weight on the hindlimbs during a larger period of time, what allowed it to move through bigger distances. New radiographies were performed so as to observe the skeletal development in view of the deformity. No bone change resulting from hemimelia has been noticed in the adult phase (Figure 2).



Figure 2. A) Macroscopic observation of transverse terminal humeral bilateral hemimelia after nine months. B) Radiographic observation of bone development in transverse terminal humeral bilateral hemimelia. In the dorsoventral position, it is possible to see humerus proximal ends on the left and right antimeres (circles). Source: Author's collection.

DISCUSSION

The denominations of malformations are rarely used in veterinary science, by the reason that there is not a uniform and accurate terminology for several species (Macrí et al., 2014). However, the congenital classification disorders in humans, described by Kay et al. (1974) has been used to classify animal dysostosis, as was done by Johnson, Kenneth, and Watson (2014). In this present case, according to the radiographic findings, the hemimelia was classified in transverse terminal humeral bilateral, confirming the literature descriptions to this pathology (Kay et al., 1974; Towle; Breur, 2012; Johnson, Kenneth, and Watson 2014). A similar case, containing the same classification, was reported by Rossi and Kruger (1975) in a child. Nonetheless, in dogs, there is no occurrence of a similar case to the one reported in this research.

Hemimelia is a rare dysostosis found in animals; the radius is the most affected bone in cats and dogs (Towle; Breur, 2012). Thus, radial hemimelia is the most reported form when talking about cats and dogs (Alam et al., 2006; Pisoni et al., 2012; Peres et al., 2013). As consequence, it is rare to find descriptions involving the other types of the pathology.

The patient's medical history was performed as described by Towle; Breur (2012). The owner was inquired about the condition of the animals in the same litter, incidence of malnutrition, maternal disease and potential fetal exposition to drugs or radiation during the first or second development phase. However, in this case, the questions above did not befall according her. Although there are various suspicion about the causes of hemimelia, some authors state that its etiology is unknown (Macrí et al., 2009; Pisoni et al., 2012; Peres et al., 2013; Macrí et al., 2014), including in humans (Riaz et al., 2016). In this case, the presence of consanguinity, as reported by the owner, can be considered as a possible cause of dysostosis, in accordance with Prats (2005). Despite the fact that there is little study concerning the consanguinity as a cause of these changes in dogs, some cases have already been observed in other species, as in caprines and equines (Corbera et al., 2002; Marchiori et al., 2014). Indeed, Marchiori et al. (2014) describes a bilateral amelia of the forelimbs in an Arabian foal, daughter of parents with 25% of consanguinity. However, in dogs are not found approaches.

Alam et al. (2006) explain the larger frequency of congenital malformations in thoracic members due to the fact that they are developed before the hindlimbs and that the bones are formed of the

proximal to the distal part. It can be related to what was verified in the present case.

The dysostosis diagnosis is simple and based on the physical and radiographic examination of the affected segment. Concomitant fractures may be present (Towle; Breur, 2012), however, not was observed in this case. The proximal terminal transverse hemimelia must be distinguished from true amelia by radiographic examination. The radiographics performed in this patient allowed to classify the hemimelia by observing the development of both scapulae and the humeral proximal ends. In human medicine, the most common way to diagnose the congenital deformities is through prenatal ultrasound, which recognizes the abnormality before birth. Therefore, the postnatal radiography is highly relevant to confirm the diagnosis and the ultrasound findings (Pallavee, 2016). In veterinary medicine, the use of prenatal ultrasound is rarely used when it comes to diagnose congenital deformities.

The main complications that may be observed in this type of dysostosis are the result of repetitive strain injury (RSI), due to the poor posture the animals have in order to be able to move themselves (Alam et al., 2006). Nevertheless, in the present case it was not observed any complication. The animal had a good adaptation to the skeletal pathology, even without the prosthesis. Thereby, Peres et al. (2013) and Campos et al. (2014) also reported a good adaptation to the bilateral radial hemimelia by cats and dogs.

For Mosbah et al. (2012), hemimelia was classified as a congenital anomaly that does not have to be surgically corrected, since the animal is able to survive with a discreet impediment. The observations related to this case confirm this ease adaptation addressed in the literature. In addition, the dog can get around, even without use of an orthopedic device.

It is recommended to castrate the animal in its adult phase, considering the hereditary disposition of the anomaly. The absence of body member's formation is associated with an autosomal dominant inheritance, but can also be recessive and caused by chromosomal abnormalities (Alam et al., 2006). This recommendation was followed by the owner, who castrated the animal when it was seven months old, since he could be able to copulate.

Although the congenital deformities on the dog's body members have been previously documented, the bilateral congenital absence of the humerus distal ends along with the agenesis of the remaining thoracic members represents an extreme variant of hemimelia. As it is already known by the authors,

the present case is the first report regarding this type of forelimb malformation in a dog, what depicts a significant addition to the field's literature.

CONCLUSION

Hemimelia has been rarely published in Veterinary Science. There were no reports of transverse terminal bilateral hemimelia affecting the humerus in dogs, as it was demonstrated in this case. Since it is a rare occurrence, these descriptions can contribute to future similar cases. The presence of consanguinity reported during the research highlighted a presumable cause of deformity in the animal. However, since there are few scientific approaches regarding this issue in the canine species, other research must be done to explore this bone deformity, which has not been completely elucidated yet, in a greater extent.

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