

A rare case of partial paraxial radial hemimelia in a puppy: a case report

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ABSTRACT: This article reports a case of a congenital unilateral forelimb defect in a two-month-old male puppy with clinical and radiographic findings. Congenital deformities are structural or functional congenital defects arising from errors during development, and they can affect an isolated portion of the body, the complete body or parts of several systems of the body. The congenital absence of a portion of the proximal epiphysis of the radius is an extremely rare condition in human and animal species. Although similar congenital limb deformities have been infrequently reported in dogs, the present case is, to the best of the authors' knowledge, the first report of this type of partial forelimb paraxial radial hemimelia in a puppy, and it represents an addition to the scant literature on this topic.

Keywords: radial hemimelia; radiography; congenital defect; dog

Errors during foetal development may be followed by structural or functional congenital malformations that are present at birth as defects (Noden and De Lahunta 1985). Congenital malformations can affect an isolated portion of the body, the entire body or parts of several systems of the body (Dennis and Leipold 1979).

Congenital limb malformations or partial malformations of limbs are varied in their manifestations, ranging from the absence of a single structure to partial or complete absence of the limbs (Lallo et al. 2001).

The lack of a uniform and precise nomenclature for limb malformations often complicates their description. This clinical case was of a type of hemimelia, a congenital abnormality with the complete or partial absence of one or more bones

(Towle and Breur 2004). Longitudinal hemimelia is the congenital absence of one or more bones along the preaxial (medial) or postaxial (lateral) side of a limb (Mo and Manske 2004; Towle and Breur 2004). Hemimelia can also be intercalary, in which case all or part of the middle bones of a limb are absent, with the proximal and distal portions being present.

Also, hemimelia can be classified as transverse hemimelia, with a complete absence of the distal portion of the limb, and paraxial hemimelia characterized by aplasia of either the radius or ulna, or tibia and fibula (Palmer 1993).

The objective of our study was to describe the macroscopic and radiographic findings of a rare case of a unilateral forelimb defect observed in a puppy.

Case description

A two-month-old male Chihuahua was presented to the Veterinary Teaching Hospital, Faculty of Veterinary Medicine, University of Messina, for an orthopaedic examination of a deformity of the right forelimb. The malformation had been present since birth and on physical examination a non-functional right forelimb, markedly reduced in size, with 90° varus deviation of the elbow, was observed (Figure 1). There was no pain, crepitation or evidence of fractures during physical examination of the deformed limbs. The paw was normal.

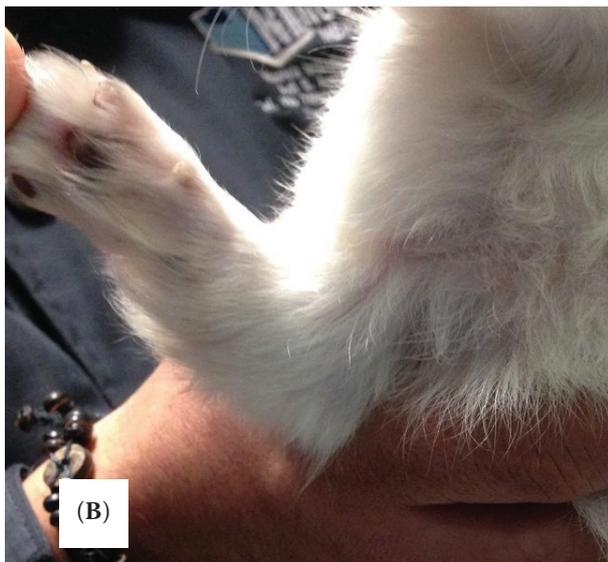


Figure 1. Dorsal (A) and ventral (B) views. Deficient right forelimb characterized by the complete ankylosis of the elbow and short leg

Radiographic views of the right forelimb were taken using an analogic Radiographic/Fluoroscopic Table System (Dedalus Mb 90/20 IMX-2A, Imago Radiology S.r.l., Abbiategrasso (MI), 20081, Italy) with a digital radiography system (Fujifilm Medical Systems, Italy); X-ray settings of 55 kV, 10 mAs at a film focus distance of 100 cm, without grid, were used. The site of deformation was radiographed in mediolateral and oblique views.

Radiographs showed a severe hypoplasia of the radius, pronounced curvature of the ulna and poor congruency of the humeroulnar joint (Figure 2). No surgical treatment was performed.



Figure 2. Lateral (A) and oblique (B) views. The X-ray of the right forelimb shows severe hypoplasia of the radius, pronounced curvature of the ulna and poor congruency of the humeroulnar joint

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DISCUSSION AND CONCLUSIONS

Different patterns of congenital lower extremity shortening have been reported in dogs and cats: brachymelia (Cornillie et al. 2004), hemimelia (Pedersen 1968; Alonso et al. 1982; Schultz and Watson 1995; Ahalt and Bilbrey 1997; Lallo et al. 2001; Rahal et al. 2005; Alam et al. 2006; Pisoni et al. 2012), ectromelia (De Lima 1915; Macri et al. 2009), adactyly (Barrand and Cornillie 2008; Macri et al. 2011), aphalangia (Macri et al. 2013a), radial agenesis (Swalley and Swalley 1978; Richardson 1979; Betts 1981; Winterbotham et al. 1985; O'Brien et al. 2002; Gemmill et al. 2004; Rahal et al. 2005; Hildreth and Johnson 2007; McKee and Reynolds 2007), syndactyly (Macri et al. 2013b) and preaxial terminal longitudinal hemimelia (Macri et al. 2014).

In our patient, the clinical signs were similar to those described in previous reports of hemimelia in dogs (Lallo et al. 2001; Rahal et al. 2005). Radial hemimelia can be caused by environmental or genetic factors, or by a combination of both.

Possible causes of hemimelia include administration of chemotherapeutics (tetracycline, griseofulvin, parabendazole, etc.), malnutrition (lack of riboflavin), intake of drugs such as thalidomide or corticosteroids (in chick embryos), trans-placental virus infections and X-rays, dietary mineral deficiency (e.g., zinc, manganese, copper) and vaccines (Johnson 1965; Karnofsky 1965; Warkany 1965; Riddle and Leighton 1970). Other causes of distal limb absence in young animals include strangulation by restrictive bands, *in utero* accidents and postnatal traumas (Johnson et al. 1995).

The heritability of radial hemimelia has been suggested: Alonso et al. (1982) described an autosomal recessive form of hemimelia in Chihuahuas, while Hoskins (1995) suggested that hemimelia in Siamese and domestic shorthair cats may be a hereditary trait.

Abnormalities in the function of the molecules responsible for embryonic limb development along the three main axes are responsible for developmental malformations in embryo limbs. Especially in human, chick and mouse embryos, mutation of *Wnt7a*, *En-1*, *FGF-2*, *Shh* and *Lmx-1* genes have been linked to the development of hemimelia (Chiang et al. 2001; Towle and Breur 2004; Woods et al. 2006). Also, hemimelia results from a lack of AER mesodermal interaction during limb outgrowth (Rantanen and Hegreberg 1982; Ogden and Grogan 1987; Towle and Breur 2004).

The critical period for the development of the entire limb in the canine embryo is between the 3rd and 4th weeks of gestation, when tissues are more susceptible to external influences (Noden and De Lahunta 1985).

The exact cause of the described malformations could not be ascertained. Preaxial longitudinal intercalary radial hemimelia is the most common type of hemimelia in dogs and cats (Alam et al. 2006), while congenital partial paraxial radial hemimelia represents an extreme variant of hemimelia.

Indeed, the present case is, to the authors' knowledge, the first report of this type of forelimb malformation in a dog and represents an important addition to the literature on this topic.

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