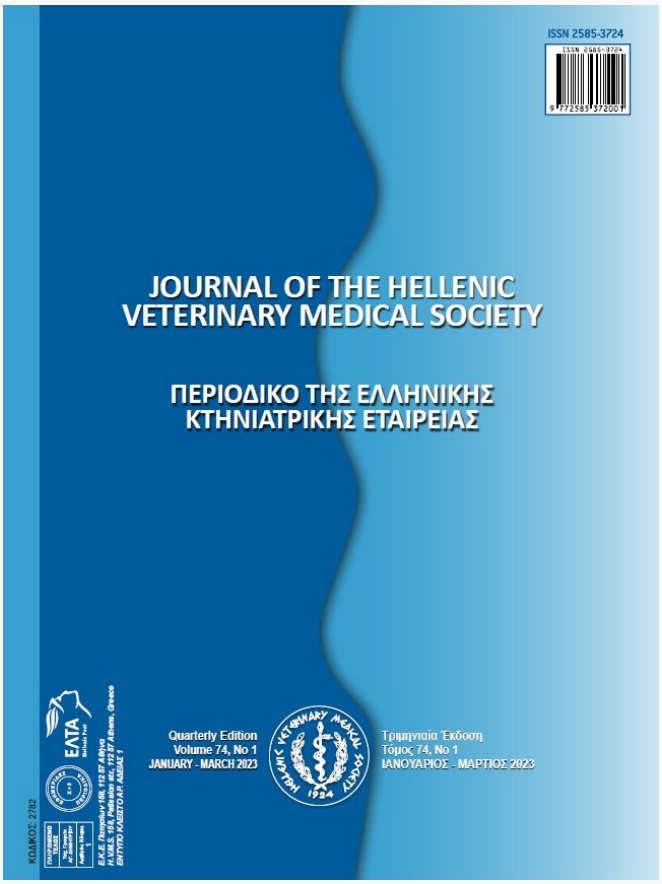
Journal of the Hellenic Veterinary Medical Society

Vol 74, No 1 (2023)



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AM Abu-Seida

doi: 10.12681/jhvms.28913

To cite this article:

Abu-Seida, A. (2023). Unilateral Partial Aphalangia and Hemimelia in an Adult Dog and Cat: Clinical Presentation and Radiography Findings. *Journal of the Hellenic Veterinary Medical Society*, *74*(1), 5463–5468. https://doi.org/10.12681/jhvms.28913 (Original work published April 12, 2023)

J HELLENIC VET MED SOC 2023, 74 (1): 5471-5476
ΠΕΚΕ 2023, 74 (1): 5471-5476

Case report
Κλινικό περιστατικό

Unilateral Partial Aphalangia and Hemimelia in an Adult Dog and Cat: Clinical Presentation and Radiography Findings

A.M. Abu-Seida

Department of Surgery, Anesthesiology and Radiology, Faculty of Veterinary Medicine, Cairo University, Giza, Egypt

ABSTRACT: Congenital digital deformities are uncommon anomalies in dogs and cats and many of them are still not recorded in veterinary literature. This report describes the clinical presentation and radiological findings in a 3-year-old male German shepherd dog with a right pelvic limb partial aphalangia and an 11-year-old female Persian cat with a left thoracic limb partial hemimelia. The dog showed an absence of the fourth digit of right pes with abnormal footpads and a wide interdigital space between the third and fifth digits. Radiography revealed presence of all metatarsals, absence of the middle and distal phalanges of the fourth digit, poorly developed proximal phalanx and foot pad of the fourth digit as well as fusion between the foot pads of the fourth and fifth digits. The cat had only one digit, two nails and one rudimentary foot pad in the left thoracic limb, which was shorter than the contralateral limb. Radiography revealed short ulna, presence of three carpal bones, absence of all metacarpals and presence of one digit with three phalanges. No other congenital disorders were noted in both animals. No treatment was offered because the dog and cat were coping well with their defects. In conclusion, dogs and cats with unilateral partial aphalangia and hemimelia can survive into old age with a reasonable quality of life, particularly indoor animals. Radiography plays a crucial role in complete description, classification and definite diagnosis of these defects in companion animals.

***Keywords*:** Aphalangia /Cat / Dog / Digital deformity /Dysostoses / Hemimelia

Corresponding Author:
Ashraf M. Abu-Seida, Faculty of Veterinary Medicine,
Cairo University, Department of Surgery, Anesthesiology & Radiology, Giza
Square PO: 12211 Giza, Egypt.
E-mail address: ashrafseida@cu.edu.eg , ashrafseida@yahoo.com

Date of initial submission: 11-12-2021
Date of acceptance: 04-04-2022

INTRODUCTION

Dysostoses are congenital skeletal malformations characterized by an abnormal development of individual bones or parts of bones (Carvallo et al., 2010). Numerous dysostoses have been previously recorded in domestic animals and birds (Carrig et al., 1981; Barrand, 2004; Abu-Seida 2014; Abu-Seida et al., 2015; Di Donaet al., 2016). Compared to human literature, description and classification of dysostoses are still lacking in veterinary literature (Temtamy and Aglan, 2008).

Dysostoses can occur in the axial or appendicular skeleton (Kerrigan and Robinson, 2016). Axial dysostoses include hemivertebra, block vertebra, butterfly vertebra, transitional vertebra, spina bifida, facet aplasia, and dens malformation (Kerwinet al., 2012; Salas et al., 2014). Several appendicular dysostoses have been recorded in the veterinary literature such as; amelia: absence of limbs, dimelia: complete duplication of a limb, brachydactyly: small-sized digit (Hoskins, 1995), aphalangia: lack of a digit or part of digit (Macriet al., 2012), adactyly: absence of all digits and their metacarpals or metatarsals (Barrand and Cornillei, 2008), polydactyly: high number of digits (Jezyk, 1985), oligodactyly (hypodactyly): low number of digits (Di Donaet al., 2016), hemimelia: complete or partial absence of one or more bones, syndactyly: fused digits (Towle and Breur, 2004) and ectrodactyly: digital cleft extending upward to various levels (Ferreiraet al., 2016; Di Pietroet al., 2021).

The etiology of dysostoses may be hereditary (malformation) or environmental (disturbed development) in origin (Towle and Breur, 2004). Genetic underpinnings of dysostoses are complex and at this time are poorly understood. There are several known mutations affecting inherited skeletal phenotypes in dogs and cats such aspolydactyly, syndactyly, spondylocostal dysostosis, disproportional dwarfism,osteogenesis imperfect, short tail and tail-less, brachycephaly and orofacial cleft (Haase et al., 2016).

Environmental factors include; drugs (tetracycline, griseofulvin, parabendazole, thalidomide or corticosteroids), maternal diseases, faulty maternal diet, modified-live vaccines, transplacental virus infections, radiation, and trauma to the mother, embryo, or placenta (Hoskins, 1995; Johnson et al., 1995;Towle and Breur, 2004). These environmental factors usually result in several failures during formation of mesenchymal bone model, transformation of anlagen into cartilage, or conversion of cartilage into bone between 23 and 35 day of gestation (Johnson et al., 1995; Towle and Breur, 2004).

Congenital digital deformities in dogs and cats have been rarely recorded (Macri et al., 2011; Macriet al., 2012; Di Donaet al., 2016). Information about the etiology, diagnosis and treatment of these defects is still scarce.

Aphalangia means lack of a digit or part of digit (Macri et al., 2012) whereas; adactyly is commonly referred to the total absence of digits and their metacarpals or metatarsals (Noden and Lahunta, 1985; Johnson et al., 1995; Macri et al., 2012). Moreover, partial aphalangia is the absence of one or more phalanges from one to four digits (Noden and Lahunta, 1985), whilst partial adactyly is the absence of one to four digits and their metacarpals or metatarsals.

Hemimelia is a congenital complete or partial absence of one or more bones. Hemimelia is called terminal if all or part of the middle and distal bones of a limb are absent (Towle and Breur, 2004). If all or parts of the middle bones of a limb are absent, with the proximal and distal parts being present, the hemimelia is called intercalary (Towle and Breur, 2004). Each of these two main groups can be subdivided into transverse and longitudinal hemimelia. Transverse hemimelia refers to complete absence of one or more bones across the limb's width while the longitudinal hemimelia means absence of one or more bones along the preaxial (medial)or postaxial (lateral) side of a limb (Towle and Breur, 2004).

In dogs and cats, most of the recorded cases of aphalangia and hemimelia had mild signs and the deformed animals appeared to cope well, particularly indoor animals (Towle and Breur, 2004; Barrand and Cornillie, 2008; Di Dona et al., 2016).

Many dysostoses can be diagnosed by physical examination, radiography, computed tomography and magnetic resonance imaging (Wrzosek et al., 2014).

The overall prognosis for appendicular dysostoses is generally good; however, treatment either medical or surgical may be necessary when the defect degrades the animal's quality of life (Kerrigan and Robinson, 2016).

Medical therapy includes physical rehabilitation to prevent pain, build muscle, and regain function as well as splinting when laxities or contractures are present. Palliative surgery includes total or partial

amputation of severely deformed limb or digit while reconstruction surgery includes realignment arthrodesis. Moreover, neutering is highly recommended in cases with known inheritance (Pratschke, 1996; Kerrigan and Robinson, 2016).

This report represents an addition to the scant literature on aphalangia in dogs and hemimelia in cats through describing the clinical presentation and radiological findings of unilateral partial aphalangia in a 3-year-old German shepherd dog and unilateral partial hemimelia in an 11-year-old Persian cat.To the author's knowledge, the clinical presentation and radiological findings of the congenital deformities described here are firstly recorded in an adult dog and cat.

CASES PRESENTATION

Case 1

A 3-year-old male German shepherd dog was admitted for vaccination to the Small Animal Clinic at Faculty of Veterinary Medicine, Cairo University, Egypt.A full case history was taken and a complete physical examination was performed.

Physical examination was within normal limits except that the dog had an absence of the forth digit of the right pes (Fig. 1). Digital deformity was incidentally detected during examination. Digital examination revealed a wide interdigital space between the third and fifth digits (Fig. 2a). The footpads were abnormal in their size and shape. The dog was able to walk reasonably normally without lameness. The dog's owner reported that the dog was the only puppy affected out of a litter of seven puppies.

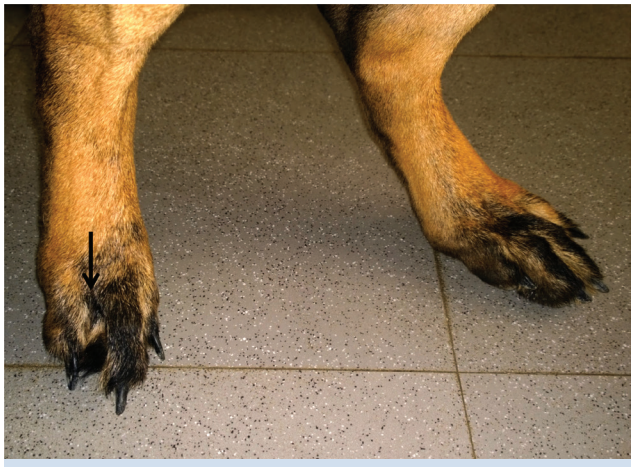

Fig. 1 Dorsal view of both pelvic limbs of a 3-year-old German shepherd dog with partial aphalangia showing absence of the fourth digit of the right pes with a wide interdigital space between the third and fifth digits (black arrow)

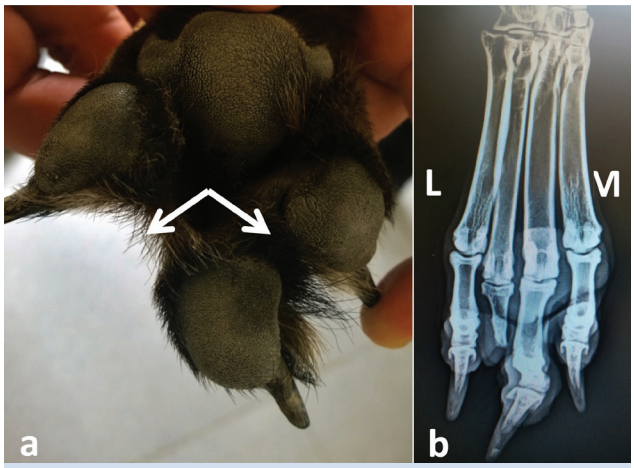

Fig. 2 (a) Plantar view of the right pes of a 3-year-old German shepherd dog with partial aphalangia showing absence of the fourth foot pad. Notice: the interdigital space between the second and third digits is narrower than that between the third and fifth digits (white arrows). **(b)** Dorsoplantar radiographic view of the right pes showing presence of all metatarsals, absence of the middle and distal phalangeal bones of the fourth digit, poorly developed proximal phalanx and foot pad of the same digit and fusion between the foot pads of the fourth and fifth digits. L: lateral side and M: medial side

Radiography was carried out on a stationary X-ray unit (Fischer Imaging, Stuttgart, Germany) and X-ray settings were 50 kV and 10 mAs at a film focus distance of 70 cm. Distal segments of both pelvic limbs were radiographed in mediolateral and dorsoplantar views.

Radiographs revealed normal distal extremity of the left pelvic limb. Dorsoplantar radiographic view of the right pes showed presence of all metatarsals, absence of the middle and distal phalangeal bones of the fourth digit, poorly developed proximal phalanx and foot pad of the fourth digit and fusion between the foot pads of fourth and fifth digits (Fig. 2b). No congenital disorders other than partial aphalangia were noted.

Clinical and radiological results confirmed a unilateral partial aphalangia in this dog. As the dog was coping well with the defect, no therapy was carried out.

Case 2

An 11-year-old indoor Persian cat was admitted for vaccination to the Small Animal Clinic at Faculty of Veterinary Medicine, Cairo University, Egypt. A complete case history was taken and a full physical examination was performed.

Physical examinations were within normal limits

and the cat was able to walk and jump reasonably normally. Digital deformity was incidentally found in the cat. On examination, the cat showed normal right thoracic limb while the left one was normal to the level of elbow joint.The left thoracic limb was shorter than the right one. The distal limb segment of left thoracic limb was clearly deformed, medially rotated and smaller than the normal contralateral limb. The left thoracic limb had only one digit, two nails and a small rudimentary digital pad (Fig. 3). The observed digit was present at the level of the third digit. One nail was found at level of the first digit and the other one was seen in the present digit.

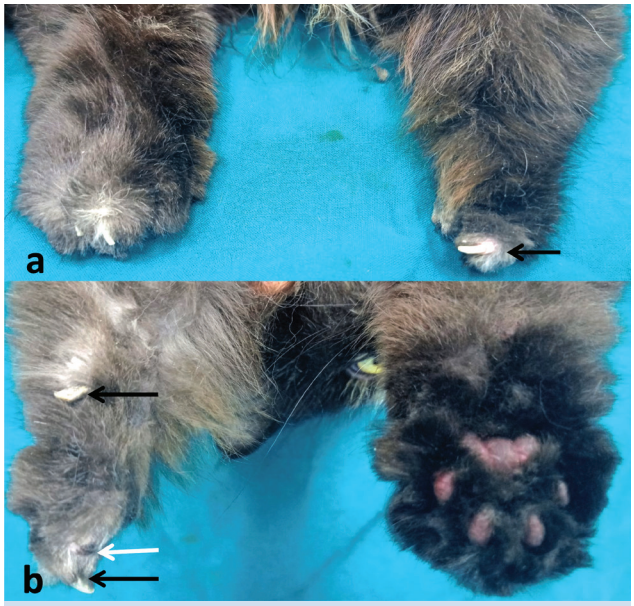

Fig. 3 (a) Dorsal view of both thoracic limbs of an 11-year-old Persian cat with a partial hemimelia showing presence of only one digit in the left thoracic limb (black arrow). Notice that the distal segment of the left thoracic limb is smaller than that of the normal contralateral one. **(b)** Plantar view of both thoracic limbs of the same cat showing normal foot pads in the right thoracic limb and presence of only one rudimentary foot pad (white arrow), two nails (black arrows) and one digit in the left deformed thoracic limb

Radiography was carried out with X-ray unit (Fischer Imaging, Stuttgart, Germany) and settings of 42 kV, 10 mAs and a film focus distance of 70 cm. Distal segments of both thoracic limbs were radiographed in mediolateral, oblique and dorsopalmar views. Radiographs of both thoracic limbs revealed a deformed left thoracic limb compared to the normal contralateral right one. The left thoracic limb showed shortened ulna, presence of only three carpal bones, absence of all metacarpal bones, absence of four digits and presence of only one digit with three phalanges (Fig. 4). No other associated defects were recorded in this cat.

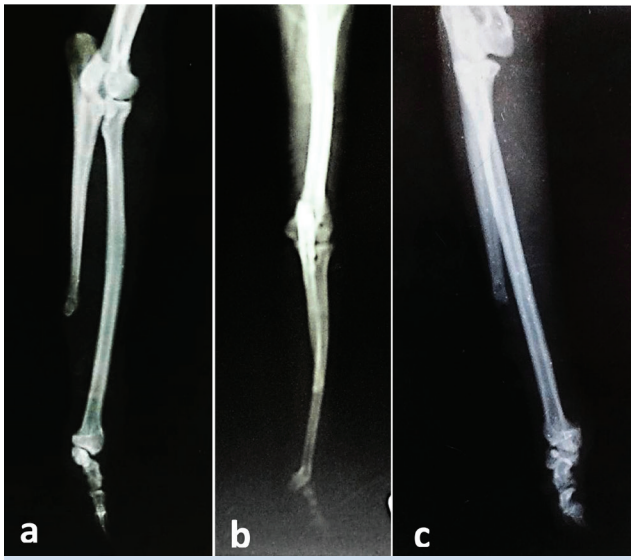

Fig. 4. (a) Mediolateral, dorsopalmar **(b)** and oblique **(c)** radiographs of the left thoracic limb of the same cat in figure 3 showing shortened ulna, presence of only three carpal bones, absence of all metacarpals and presence of only one digit with three phalangeal bones.

Clinical and radiological results confirmed a unilateral partial hemimelia in this cat. No treatment was performed because the cat was coping well with her defect.

Discussion and conclusions

Up to date, several dysostoses are not yet recorded in veterinary literature. Few reports have been described canine aphalangia and feline hemimelia in veterinary literature (Barrand and Cornillie, 2008; Macri et al., 2011; Macriet al., 2012; Di Dona et al., 2016). The present report describes unique cases of unilateral partial aphalangia and hemimelia in an adult dog and senior cat, respectively. These cases are unique due to their presenting clinical signs that differ from previously recorded cases and in that the deformed animals could be survived to old age without any complications. The present cases represent an addition to the veterinary literature on this topic.

Although *Nomina Embryologica Veterinaria* (2006) is the gold standard for identification and classification of congenital defects in veterinary medicine, there is a great shortage in a uniform and consistent nomenclature and classification of canine and feline distal limb anomalies (Cornillie et al., 2004; Di Dona et al., 2016). Therefore, a standard source of unequivocal and well-described nomenclature of these congenital defects is highly required.

In partial aphalangia one or more phalanges from

one to four digits are absent (Macri et al., 2012). Therefore, the term partial aphalangia used in the present dog is most appropriate due to absence of the middle and distal phalangeal bones of the fourth digit. As well, the term partial hemimelia used in the present cat is most suitable due to underdeveloped ulna, presence of three carpal bones only and absence of all metacarpals and four digits.

In case 1, a unilateral absence of the fourth digit of the right pes was clinically seen whereas, on radiography, an absence of two phalanges of the fourth digit of right pes and presence of the fourth metatarsal bone were viewed. Therefore, the defect could be identified clinically as a partial adactyly; however it was a partial aphalangia after radiography. Similar findings were recorded before in dogs (Di Donaet al., 2016). Thus, radiography plays a crucial role in a definite diagnosis of congenital digital defects in dogs. Moreover, radiography is essential for differential diagnosis of the congenital digital defects and any earlier amputation (Di Donaet al., 2016).

According to the available literature, there is a one case report of a unilateral thoracic limb partial aphalangia with mild lameness. It was recorded in a 2-month-old male kitten with no digits, one nail, two footpads, and presence of the first metacarpal as well as proximal and distal phalanges of the first digit, reduced metacarpal bones and incompletely ossified carpal bones (Macri et al., 2012). In contrast, the recorded partial aphalangia here was seen in the pelvic limb of an adult dog with one deformed digit.

The footpads on the deformed limb of the present dog were also abnormal in their number, shape and size. Footpad anomalies are mentioned briefly in few previous reports of congenital digital defects. In one case, the metacarpal and digital pads were smaller than usual and had an unusual shape (Pratschke, 1996). Another case had a carpal pad 1.5 times larger than usual (Montgomery and Tomlinson, 1985). The largest case study, of 14 dogs, described the radiographic findings only and did not record the anomalies defects of footpads (Carriget al., 1981). It seems probable that some previously recorded cases had pad defects but that they were not reported.

Although the etiology of aphalangia is unknown, inherited autosomal dominant manner in cats was suggested (Schneck, 1974; Jezyk, 1985). A possible non-hereditary basis in dogs, due to the low incidence of bilateral form and lack of breed predisposition is also suggested (Carriget al., 1981). In the present cases, the author was unable to study littermates, parents and environmental circumstances of the diseased animals. Genetic or environmental teratogens are not possible etiologies for the recorded defect here because these causes would have had an effect on all limbs or at least on both thoracic or both pelvic limbs (Macri et al., 2011; Di Donaet al., 2016). Therefore, the cause of the current congenital partial aphalangia remains unclear. Similar unknown etiology was mentioned before in a cat with partial thoracic limb aphalangia (Macri et al., 2012).

In case 2, a partial unilateral hemimelia was recorded in the left thoracic limb of an 11-year-old cat. In contrast, cases of bilateral hemimelia of both thoracic limbs were reported in kittens (Lockwood et al., 2009; Pisoni et al., 2012).

The left deformed thoracic limb was shorter than the normal right one in the recorded cat here. Similar finding was recorded before in a cat with unilateral partial hemimelia (Makino et al., 2016). This shortening could be attributed to absence of all metacarpal bones in the deformed thoracic limb.

Clinically the cat had one digit of the left thoracic limb whereas, on radiography, underdeveloped ulna, presence of only three carpal bones and absence of all metacarpals as well as four digits were observed. Therefore, the defect could be identified clinically as a partial adactyly; however it was a partial hemimelia after radiography.

Unlike the recorded kitten by Pisoni et al. (2012) that was unable to use the deformed limb well and was treated conservatively with splint bandage, the reported cat here was able to use the deformed limb well. On contrary, two kittens with brachymelia (ectromelia, hemimelia, meromelia and apodia/adactyly) in all limbs were euthanized (Cornillieet al., 2004). In this regard, cats with mild cases of hemimelia, do not necessarily have a bad quality of life necessitating euthanasia. Survival into adulthood with a favorable quality of life is possible (Barrand and Cornillie, 2008; Di Pietro et al., 2021).

No associated congenital defects were reported in the present cat. In contrast, polydactyly, cardiomegaly, five thoracic vertebrae, six lumbar vertebrae and fifteen ribs, with hypoplasia of the first pair were associated with bilateral hemimelia in cats (Lockwood et al., 2009; Pisoni et al., 2012).

Regarding etiology, it has been suggested that hemimeliamay be a hereditary trait in Siamese and Domestic short hair cats (Di Pietro et al., 2021). In contrast, the hemimelia recorded here was seen in a Persian cat.

Treatment of digital deformities depended upon the severity of clinical signs (Pratschke, 1996). Therefore, the therapy should be in compliance with every single case (Tchaprazov et al., 2007; Harasen, 2010). Conservative management with the use of protective braces can avoid the secondary lesions that can be associated to the underdevelopment of the toes and the digital pads in particular (Di Dona et al., 2016). The dog and cat in the present study did not need any treatment because of the good function of the affected limbs

Moreover, animals with appendicular dysostoses should be sterilized in order to avoid the potential hereditary nature of these disorders as mentioned before (Ferreira et al., 2016).

Further studies are recommended to determine the prevalence of dysostoses and clinical significance in dogs and cats and to put in record any defect that has not been reported previously.

In conclusion, dogs and cats with partial aphalangia or hemimelia do not necessarily have a bad quality of life necessitating treatment or euthanasia. Survival into old age with a reasonable quality of life is possible, particularly for indoor animals. Clinical presentation is not enough for definite diagnosis of aphalangia as well as hemimelia and radiography is essential for complete description, classification and definite diagnosis of these dysostoses in dogs and cats.

CONFLICT OF INTERST

The author declares no conflicts of interest.

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