## Radiological Findings in Three Cases of Paraxial Radial Hemimelia in Goats

Juan Alberto CORBERA<sup>1)</sup>, Mónica PULIDO<sup>1)</sup>, Manuel MORALES<sup>1)</sup>, María Candelaria JUSTE<sup>1)</sup> and Carlos GUTIERREZ<sup>1)</sup>

<sup>1)</sup>Department of Animal Medicine and Surgery, Veterinary Faculty, University of Las Palmas de Gran Canaria, Arucas 35416, Canary Islands, Spain

(Received 26 December 2001/Accepted 1 May 2002)

ABSTRACT. Hemimelia is a congenital abnormality characterized by the absence of a portion of the normal structures in a limb. Hemimelia is classified as transversal and paraxial and is related to genetical and environmental factors. This article shows the radiolo gical findings observed in three different cases of paraxial hemimelia occurred in goats (radial agenesia, absence of the portion of the distal epiphysis of the radius and anomalous radius with ulnar hypoplasia). Possible causes related to these abnormalities are discussed.

KEY WORDS: caprine, hemimelia, radiology.

- J. Vet. Med. Sci. 64(9): 843–845, 2002

Hemimelia is a congenital abnormality characterized by the absence of a portion of the normal structures in an extremity. Radial hemimelia occurs when a total or partial congenital deficiency affecting the radius is present [15].

In humans, this deformity is considered to be sporadic [13] or very rare, considering that human incidence is approximately 1 per 1 million of live births [7] and not considering the high percentage of the disease occurred in the past due to thalidomide embryopathic effects.

From an anatomical point of view, hemimelia is classified as *transversal hemimelia*, characterized by a complete absence of the distal portion of the limb, also called congenital amputation, and *paraxial hemimelia*, characterized by an aplasia of either the radius or ulna, or tibia and fibula [15]. Although, when an intercalary segment of the limb is absent, the term phocomelia is used. In contrast, amelia is the complete absence of one or more limbs. Otherwise, when the radius is completely absent, the lesion is described as radial agenesis [5].

The etiology of radial hemimelia is considered to be genetical, environmental or a combination of both factors. In humans, genetical defects that cause limb deficiencies are associated with autosomal dominant inheritance; however, other genetical causes include an autosomal recessive inheritance and chromosomal aberrations. Also, different teratogenic agents and drugs have been related to congenital defects [9, 18].

In the literature there are several reports of radial hemimelia in the cattle, sheep, dog, and cat. In goats, amelia and paraxial hemimelia [3, 6] and radial agenesia [4] have been reported. However, neither description could be related to genetical or environmental causes.

Given that clinical findings are similar in many cases, radiological examination is needed to determine the anatomical deformities presents in the newborn. This paper reports the radiological findings corresponding to three different cases of paraxial hemimelia occurred in Canarian goats.

In the last 10 years, among over thirty different congeni-

tal abnormalities observed in ruminants at the Large Animal Clinic, hemimelia in forelimbs was diagnosed in three newborn goats. These cases were from three different dairy goat farms located in Gran Canaria, Canary Islands.

All the animals, which presented deformities in the forelimbs, were assessed by physical and x-rays examinations. Of the three cases, necropsy could be performed in two animals, and no other gross lesions were evident in both animals. Karyotype was also carried out in two newborns and no apparent chromosomal aberrations were observed (58 XX, 58 XY). Serological studies on the dams using ELISAs for the detection of Toxoplasma, Akabane virus, and bovine viral diarrhea virus were also negative.

Case 1: A bilateral forelimb deformity was evident in a female goat kid. Clinically, the right forelimb showed a varus deviation. Radiologically, the presence of a curved cubitus without evident vestiges of the radius was observed (Fig. 1). A radial agenesia was diagnosed. This type of hemimelia was also observed in another newborn goat.

Case 2: The left forelimb of the same female kid showed a clinically strong varus deviation in the left forelimb. Radiologically, an absence of a portion of the distal epiphysis of the radius was evident (Fig. 2). This kind of hemimelia was classified as partial paraxial.

Case 3: A deformity presented in the right forelimb of a male kid and also a strong varus flexion was observed. A grosser and shorter radius as well as vestiges of cubitus (ulnar hypoplasia) was observed at radiological examination (Fig. 3).

An important bone deformity was evident at radiological examinations in cases 1 and 2. Bone deformity occurred during fetal development should be explained because the cubitus is not able to act as the sole weight-bearing bone between elbow and carpus. The extensor muscles pull the unsu and articulation with the carpus was not established, showing a permanent flexion.

From an etiopathogenic point of view, the factors that produce radial hemimelia remain unclear. During the thoracic limb development (around 4 week in the sheep) [14],



Fig. 1. Radial agenesia, characterized by the presence of a curved cubitus without evident vestiges of the right radius in a female newborn kid.



Fig. 2. Partial paraxial hemimelia, characterized by the absence of a portion of the distal epiphysis of the left radius in the same female newborn kid of Fig. 1.

somes have been described in the "tibial hemimelia syndrome" with a simple autosomal recessive mode of inheritance [12, 16]. This malformation has also been described in Shorthorn cattle, with similar characteristics and associated with a recessive allele in a environmental causes such as intrauterine compression, drugs, maternal nutritional deficiencies, inflammatory processes, irradiations, vaccines [17] and vascular defects [19] have been associated with radius deficiencies. On the other hand, genetic defects have been described in the "tibial hemimelia syndrome" with a simple autosomal recessive mode of inheritance [12, 16]. This malformation has also been described in Shorthorn cattle, with similar characteristics and associated with a recessive allele in a homozygous state [11].

In dogs, hemimelia has also been associated with an autosomal recessive inheritance [2].

In lambs and calves, hemimelia has been related to those dams that grazed *Lupinus consentinii* during mating or the early part of pregnancy, which contains significant amounts of alkaloids in its seeds, seed pods and leaves [1].

In the studied farms there were not any evidence of the use of retinoic acid, parbendazol, acetazolamide, adenine,



Fig. 3. Ulnar hypoplasia, characterized by the presence of a grosser and shorter radius as well as vestiges of cubitus observed in the right forelimb of a male newborn kid.

1,7-dimethylxanthine, aminophylline, acetoxy-methylmethylnitrosamine, aspirin or cadmium, all drugs associated with congenital abnormalities [9, 18]. The main infection agents, teratogenic plants, parasites and drugs were also ruled out as possible causes, because the farms were managed attentively and the presentations were spontaneous.

The relationships and the phenotypic similarity to the proven genetic disorders in Galloway cattle, Shorthorn cattle and dogs could suggest a simple autosomal recessive trait. The study of the three herds revealed to be composed by a small number of animals and that the introduction of new males was not an usual practice. This fact could have promoted an inbreeding environment, which suppose that recessive abnormalities could occur [8].

Further studies including a major number of cases are necessary in order to establish the possible genetical or environmental cause related to radius hemimelia in goats.

## **REFERENCES**

- Allen, J.G., Fenny, R.E., Buckman, P.G., Hunt, B.R. and Morcombe, P.W. 1983. Aust. Vet. J. 60: 283–284.
- Alonso, R.A., Hernandez, A., Diaz, P. and Cantu, J.M. 1982. Vet. Rec. 110: 128.
- Aslanbey, D., Tasbas, M. and Olcay, B. 1988. Veteriner Fakultesi Dergisi, Ankara University 35: 93–103.
- Baum, K.H., Hull, B.L. and Weisbrode, S.E. 1985. J. Am. Vet. Med. Assoc. 186: 170–171.
- 5. Bennett, D. and May, C. 1997. pp. 149. *In*: Tratado de Medicina Interna Veterinaria, Intermedica, Buenos Aires.
- El Hariri, M.N.E. and Shawki, H. 1980. J. Egyptian Vet. Med. Assoc. 40: 89–98.
- 7. Fernandez-Palazzi, F., Bendaham, J. and Rivas, S. 1998. *J. Pediatr. Orthop. (B)* 7: 298–302.
- 8. Huston, K. 1993. Vet. Clin. North Am-Food Anim. Pract. 9: 1-
- 9. Kochhar, D.M. 1985. Prog. Clin. Biol. Res. 171: 267–281.
- 10. Lanthier, C., Dupuis, J. and Pare, J. 1998. J. Zoo Wildlife Med.

- **29**: 65–67.
- 11. Lapointe, J.M., Lachance, S. and Steffen, D.J. 2000. *Vet. Pathol.* **37**: 508–511.
- 12. Leipold, H.W., Guffy, M.M. and Cook, J.E. 1977. Comp. Pathol. Bull. 9: 2.
- 13. Lenz, W. 1980. Clin. Orthop. 148: 9-17.
- Noden, D.M. and de Lahunta, A. 1985. pp. 196–210. *In*: The Embryology of Domestic Animals. Developmental mechanisms and malformations. Williams & Wilkins, Baltimore.
- Palmer, N. 1993. pp. 45–47. *In*: Pathology of Domestic Animals 4th ed., Academic Press, San Diego.
- Pollock, D.L., Fitzsimons, J., Deas, W.D. and Fraser, J.A. 1979. Vet. Rec. 104: 258–260.
- 17. Richardson, D.C. 1979. J. Vet. Orthop. 1: 39-42.
- Sanders, D.D. and Stephens, T.D. 1991. Teratology 44: 335– 354
- Van Allen, M.I., Hoyme, E.E. and Jones, K.L. 1982. *J. Pediatr.* 101: 832–838.