NOTE

Anatomy

Radiological Findings in Three Cases of Paraxial Radial Hemimelia in Goats

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(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 genetic and environmental factors. This article shows the radiological 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.

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 agenesis [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 congenital 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 agenesis 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],...
somes have been described in the “tibial hemimelia syn-
drome” with a simple autosomal recessive mode of inherit-
ance [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 au-
tosomal 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,
1,7-dimethylxanthine, aminophylline, acetoxymethyl-
methylnitrosamine, 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 man-
aged attentively and the presentations were spontaneous.

The relationships and the phenotypic similarity to the
proven genetic disorders in Galloway cattle, Shorthorn cat-
tle 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 envi-
ronmental cause related to radius hemimelia in goats.

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