J. Comp. Path. 2014, Vol. 150, 245–252

Available online at www.sciencedirect.com

ScienceDirect

ELSEVIER

www.elsevier.com/locate/jcpa

SPONTANEOUSLY ARISING DISEASE

Skeletal Dysplasia with Craniofacial Deformity and Disproportionate Dwarfism in Hair Sheep of Northeastern Brazil

F. P. M. Dantas*, G. X. Medeiros*, A. P. M. Figueiredo*, K. Thompson† and F. Riet-Correa*

** Veterinary Hospital, Federal University of Campina Grande, Patos, Paraíba CEP 58700-000, Brazil and † Institute of Veterinary, Animal and Biomedical Sciences, Massey University, Private Bag 11222, Palmerston North, New Zealand*

Summary

This paper reports a newly described form of skeletal dysplasia affecting Brazilian hair sheep of the Cabugi breed. This breed is characterized by having a short head and in some cases the animals are smaller and more compact than sheep of similar breeds. Lambs born with craniofacial abnormalities and dwarfism that die at 2–6 months of age are frequent in this breed. In a flock of 68 ewes and three rams of the Cabugi breed, 134 lambs were born over a 4-year period. Of these, 14 (10.4%) had marked cranial abnormalities and dwarfism and died or were humanely destroyed, 43 (32%) had a normal face and 77 (57.5%) had the short face characteristic of the breed. Dwarf lambs were much smaller than normal, with short legs, a domed head with retruded muzzle and protruded mandible, sternal deformities and exophthalmic eyes situated more laterally in the face than normal. Microscopical examination of long bones of the limbs, bones of the base of the skull and vertebrae showed no lesions. Bones from four affected lambs and one control lamb were macerated for morphometric examination. Although the length of the spinal cord was similar, there was disproportionate shortening of the appendicular bones, particularly the distal segments. Thus the disease was defined as a skeletal dysplasia characterized by craniofacial deformity and disproportionate dwarfism. It is suggested that the disease is inherited as an incomplete dominant trait. The shortened face, which is a feature of the Cabugi breed, may represent the heterozygous state and the more severe, often lethal, dwarfism may occur in homozygotes.

Keywords: brachycephaly; disproportionate dwarfism; sheep; skeletal dysplasia

Introduction

In northeastern Brazil there is a type of hair sheep known as 'cara curta' (short face), recognized as the Cabugi breed and characterized by shortening of the face and to a lesser degree the limbs (dos Santos, 2003). In flocks of this breed, lambs are often born with more severe skeletal abnormalities and frequently die from a few weeks to 6 months of life.

A wide variety of skeletal dysplasias, including several forms of disproportionate dwarfism, have been described in man and domestic animals. The most common skeletal dysplasia of sheep is 'spider lamb syndrome' (SLS), an inherited chondrodysplasia reported in the Suffolk and Hampshire breeds and characterized by increased length of long bones (Vanek *et al.*, 1986, 1989; Rook *et al.*, 1988). A single base change in the tyrosine kinase II domain of gene encoding fibroblast growth factor receptor 3 (FGFR3) has been recognized as the underlying defect in this disease. Chondrodysplastic diseases characterized by disproportionate dwarfism have also been reported occasionally in sheep (Thompson *et al.*, 2008) and are well recognized in cattle (Thompson, 2007).

In this paper we report a newly described form of skeletal dysplasia in sheep of the Cabugi breed

Correspondence to: F. Riet-Correa (e-mail: franklin.riet@pq.cnpq.br).

0021-9975/$ - see front matter
http://dx.doi.org/10.1016/j.jcpa.2013.11.208

Table 1
Craniometric points according to von den Driesch (1976)

Craniometric point	*Definition*
Akrokranion	Most caudal point of the skull (the external occipital protuberance)
Basion	Point in the median plane of the rostral–ventral border of the foramen magnum
Bregma	Point median of the suture between the parietal and frontal bones
Euryon	Most lateral point of the neurocranium, caudal to the zygomatic process of the frontal bone
Gonion caudale	Most caudal point of the mandibular angle
Infradentale	Most rostral median point of the alveoli of the incisor teeth
Nasion	Most caudal point of the interfrontal suture
Otion	Most lateral point of the mastoid region, located dorsal and caudal to the external acoustic pore
Palatino-orale	Median point of the suture between the palatine and maxillary bones
Prosthion	Midpoint of the line joining the rostral extremities of the incisive bones
Rhinion	Midpoint of the line joining the rostral extremities of the nasal bones
Staphylion	Most caudal point of the palatine bone, at the median plane
Zygion	Most lateral point of the zygomatic arch

characterized by cranial abnormalities and disproportionate dwarfism.

Materials and Methods

This study was conducted in a flock of Cabugi sheep in the municipality of Taperoá, Paraíba State, in the semiarid region of northeastern Brazil. Between May 2008 and December 2011 the flock was observed for the presence of lambs with skeletal deformity, including short limbs and cranial abnormalities. Ten affected lambs, four males and six females, were examined clinically. Five affected lambs were subjected to necropsy examination. Four were humanely destroyed and examined between 15 and 45 days of life. Another died spontaneously after approximately 6 months of age and was also examined. A control 1-month-old normal lamb from the same Cabugi flock was also examined.

During the necropsy examination, samples from thoracic and abdominal viscera, skeletal muscles, pituitary, parathyroid, skull, long bones, vertebrae, ribs, sternum and central nervous system were collected and fixed in 10% neutral buffered formalin. Bones were decalcified in formic acid and sodium citrate. All tissues samples were embedded in paraffin wax and sections (4–6 μm) were stained with hematoxylin and eosin (HE).

In addition, another three 1-month-old dwarf lambs and one normal lamb from the same Cabugi flock and of the same age were used for morphometric studies of the bones. For this, the skeletons of these lambs were macerated in hot water then washed in running water and submerged in H_2O_2 for 12 h for bleaching (Tasbas and Tecirlioglu, 1996; Onar *et al.*, 1997). They were then dried for 2–3 days. The joints that were released during the maceration were fixed with glue and copper wire. Different osteometric parameters were recorded following the methods of Guintard and Fouché (2008), Parés *et al.* (2010) and Salami *et al.* (2011) with the help of Vernier calipers. The measurements were made in millimeters to the nearest 0.05 mm.

Osteometry of the Axial Skeleton

The skull biometric examination was performed using the craniometric points defined by von den Driesch (1976) and presented in Table 1. The following parameters were measured: total length of the skull (the distance between the akrokranion and prosthion); width of the skull (the distance between the zygion); total length of the cranial base (the distance between basion and prosthion); total width of the cranial base (the distance between the otion); neurocranium length (the distance between the akrokranion and nasion); neurocranium width (the distance between the euryon); neurocranium height (the distance between the bregma and midpoint between the foramen oval); viscerocranium length (the distance between the prosthion and nasion); viscerocranium width (the distance between the facial tuberosity); viscerocranium height (the distance between the staphylion and nasion); total length of the nasal region (the distance between the nasion and rhinion); nasal bone length (the distance between the rostral border of the nasal bone and frontonasal suture); nasal bone width (between the lateral and medial borders of the nasal bone); total length of the palatal region (the distance between the prosthion and staphylion); rostral palatal region length (the distance between the prosthion and palatino-orale); greatest width of the region palatal (maximum distance at the horizontal plate of the palatine bone behind the last molar tooth); mandible length (the distance between the infradentale and gonion caudale).

The total length of the vertebral column was taken between the cranial edge of the ventral arch of the atlas (C1) and the apex of the sacrum. The length of the sternum was taken between the manubrium and the xiphoid process. The width of the sternum was taken at the level of the 3rd sternebra.

Osteometry of the Appendicular Skeleton

The length of the following long bones was determined: scapula (the distance between the proximal border and