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OTOCEPHALY WITH CYCLOPIA AND HYDROCEPHALUS IN A BERGAMASCA LAMB (OVIS ARIES)

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Abstract: *Otocephaly is a rare lethal malformation in veterinary and human medicine characterized by severe facial dysmorphism. Otocephalic phenotypes contain synotia, agnatia or mandibular hypoplasia, microstomia and aglossia or microglossia. A stillborn male lamb with syndromic evolution of otocephaly was evaluated clinically, by computed tomography and dissection examination. An Bergamasca ewe in her third lambing gave birth to the lamb through assisted vaginal birth in a herd of 70 animals. This was the only newborn affected. Clinically the lamb had incomplete microstomia due to agenesia of the palate bones, aglossia, synotia, cyclopia without eyeballs and hydrocephalus. In CT scan, one frontal orbital cavity was observed in the medial plane between the frontal, nasal and maxillary bones, ventriculomegaly with cerebral and cerebellar hypoplasia and mandibular semilunar remnants. There were no modifications to the skeleton. Dissection revealed abnormally large pharyngeal cavity, tracheal hypoplasia, well-developed esophagus and thoraco-abdominal hemorrhagic effusion. The organs in the abdominal cavity had a hemorrhagic appearance, the spleen was slightly enlarged in volume, the left kidney was hypoplastic, the right kidney was normal, the abomasum was normal in appearance, without content, and the rumen had liquid content, possibly amniotic fluid. The aim of the paper was to present a rare case of otocephaly with syndromic evolution, as well as to review the etiopathogenesis of this congenital malformation.*

Keywords: *rare disease, microstomia, synotia, ventriculomegaly, hemorrhagic effusion*

• Introduction

Congenital anomalies in sheep and goats encompass a wide range of structural and functional defects that can impact survival, growth, and reproductive performance [1]. The term otocephaly has been applied to cases of dysgnathia in which the upper and lower jaws, tongue, and frontonasal bones are absent or hypoplastic, and the pinnae are often fused in the midbasal part of the head [2].

• Material and method

The case study was conducted on a Bergamasca lamb (Fig.1), stillborn in April 2025, with changes incompatible with life. The lamb comes from a herd of 70 heads where mating was carried out naturally, with one ram per 23 females. The female was on her third lambing, the previous ones being normal. No other congenital malformations were reported in the herd. Due to the severity of the clinical form and its rarity, the lamb was sent to the Faculty of Veterinary Medicine Timisoara for further investigations, being teaching and research material.

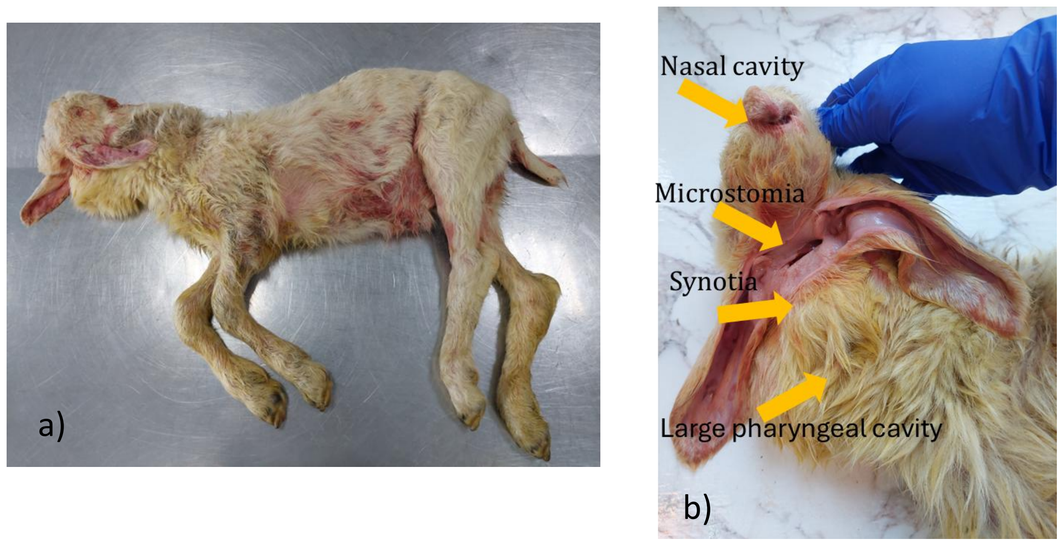

Fig 1. Lateral view of the lamb (a) and rostral part of the head (b)

References
1. Esmaeili H, Mahdi Joghataei S, Hamedi M, Lacasta D, Congenital anomalies in sheep and goats in Iran over ten years, Vet Journal, 315, 2026, 106533
2. Erlich MS, Cunningham ML, Hudgins L. Transmission of the dysgnathia complex from mother to daughter. Am J Med Genet. 2000;95(3):269–274.

• Results and discussions

In CT scan, one frontal orbital cavity was observed in the medial plane with no eye structure inside it (Fig.2). In ventral examination of the head, the vomer (*), the hyoid bones (yellow arrows) and remanences of the mandible (*) are seen (Fig.3.).

In Figure 4 it can be observed that the temporal bone is very extended (*), the parietal bone (*) is abnormal, and the frontal bone (*), is poorly developed. The two parietal bones fuse dorsally in the median plane, continuing caudally with the interparietal (*). Short nasal bone (*), frontal bone (*), zygomatic bone (*) and maxillary bone (*) are notice.

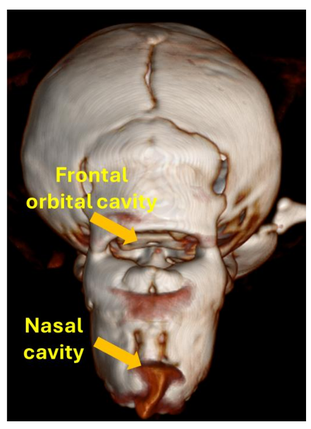

Fig.2 Rostral view of the head

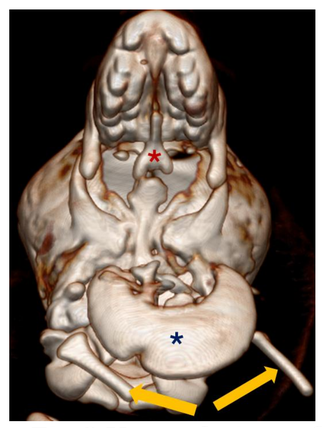

Fig.3 Ventral view of the head

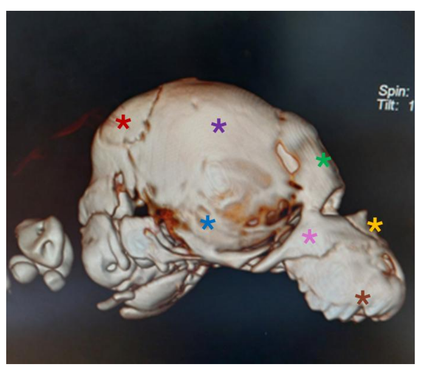

Fig.4 Lateral view of the head

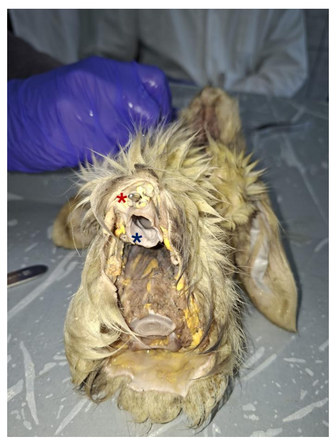

Fig.5 Trachea and esophagus

Dissection revealed abnormally large pharyngeal cavity, tracheal hypoplasia (*), well-developed esophagus (*)(Fig.5) and thoraco-abdominal hemorrhagic effusion (Fig.6).

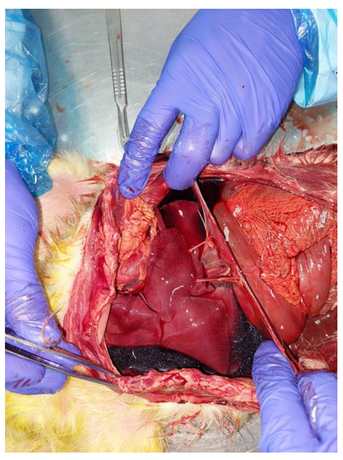

Fig.6 Hemorrhagic effusion in thoraco-abdominal cavities

• Conclusions

Due to the large genetic contributing factors for the production of otocephaly associated with cyclopia, anophtalmia and hydrocephalus, it is impossible to define the specific causes.