

# Morphopathological Examination of Anencephaly and Concomitant Congenital Malformations in a Lamb

## Kuzuda Anensefali ve Eşlik Eden Konjenital Malformasyonların Morfopatolojik İncelenmesi

Muhammet Bahaeddin DÖRTBUDAK<sup>1\*</sup>



Gamze ÇAKMAK<sup>2</sup>



Serkan YILDIRIM<sup>3</sup>



Funda EKŞİ<sup>4</sup>



Fatma İLHAN<sup>5</sup>



Nebi ÇETİN<sup>2</sup>



Sait ŞENDAG<sup>2</sup>



<sup>1</sup>Harran University, Faculty of Veterinary Medicine, Department of Pathology, Şanlıurfa, Türkiye

<sup>2</sup>Yüzüncü Yıl University, Faculty of Veterinary Medicine, Department of Anatomy, Van, Türkiye

<sup>3</sup>Atatürk University, Faculty of Veterinary Medicine, Department of Pathology, Erzurum, Türkiye

<sup>4</sup>Çukurova University, Faculty of Veterinary Medicine, Department of Obstetrics and Gynecology, Adana, Türkiye

<sup>5</sup>Balıkesir University, Faculty of Veterinary Medicine, Department of Pathology, Balıkesir, Türkiye

*This study was presented in a poster session at the "48<sup>th</sup> Annual Conference of Physiology and Pathology of Reproduction and 40<sup>th</sup> Joint Congress of Veterinary and Human Medicine" on 11<sup>th</sup>-13<sup>th</sup> February in Switzerland.*

**Geliş Tarihi/Received** 23.05.2024  
**Revizyon/Revision** 11.06.2024  
**Kabul Tarihi/Accepted** 26.06.2024  
**Yayın Tarihi/Publication Date** 30.06.2024

**Sorumlu Yazar/Corresponding author:**

Muhammet Bahaeddin DÖRTBUDAK

E-mail: mbdortbudak@gmail.com

**Cite this article:** Dörtbudak MB, Çakmak G, Yıldırım S et al. First Case Report: Morphopathological Examination of Anencephaly and Concomitant Congenital Malformations in a Lamb. *J Vet Case Rep.* 2024;4(1):19-22.



Content of this journal is licensed under a Creative Commons Attribution-Noncommercial 4.0 International License.

### ABSTRACT

The macro-pathology changes of anencephaly and other accompanying congenital anomalies in a lamb are described in this report. Anencephaly is a condition in which a large part of the brain does not develop or is not formed at all as a result of incorrect closure of the neural sulcus during the development of the offspring in the intrauterine period. Anencephaly is generally not manifested alone and includes various anomalies. Although congenital malformations are very common in domestic ruminants, there is no report in the literature reporting an anencephaly anomaly in lambs. In this study, for the first time in the world, anencephaly in a lamb and some concomitant anomalies were presented. In this case in which morphological changes were examined, anencephaly anomalies, as well as anomalies of acrania, arhinia, anotia, anophthalmia, agnathia and microglossia were also encountered. Thanks to this case, the macroscopic findings of congenital head anomalies in a lamb that had not been previously reported were presented and evaluations were made on their possible etiological causes.

**Keywords:** Anencephaly, Congenital anomaly, Lamb, Morphopathology.

### Öz

Bu olgu sunumunda bir kuzuda anensefali ve eşlik eden diğer konjenital anomalilerin makropatoloji değişiklikleri anlatılmaktadır. Anensefali, intrauterin dönemde yavrunun gelişimi sırasında nöral sulkusun yanlış kapanması sonucu beynin büyük bir bölümünün gelişmemesi veya hiç oluşmaması durumudur. Anensefali genellikle tek başına ortaya çıkmaz ve çeşitli anomalileri içerir. Evcil ruminantlarda konjenital malformasyonlar oldukça yaygın olmasına rağmen literatürde kuzularda anensefali anomalisini bildiren bir yayın bulunmamaktadır. Bu çalışmada dünyada ilk kez bir kuzuda anensefali ve buna eşlik eden bazı anomaliler sunulmuştur. Morfolojik değişikliklerin incelendiği bu olguda anensefali anomalilerinin yanı sıra akrania, arhinia, anotia, anoftalmi, agnati ve mikroglossia anomalilerine de rastlandı. Bu vaka sayesinde bir kuzuda daha önce bildirilmemiş konjenital kafa anomalilerinin makroskobik bulguları sunulmuş ve bunların olası etiyolojik nedenleri üzerine değerlendirmeler yapılmıştır.

**Anahtar Kelimeler:** Anensefali, Konjenital anomali, Kuzu, Morfopatoloji

## INTRODUCTION

Congenital malformations may manifest as any organ or organs in the body not forming at all during or after the intrauterine period, their inadequate development, and various developmental deformities.<sup>1,2</sup> Congenital anomalies of the central nervous system (CNS) are more common than other system or organ anomalies in domestic animals.<sup>3,4</sup> Anencephaly, which is one of the most important CNS anomalies, is the case where both hemispheres that form the brain are not shaped. Closure errors in the cranial part of the neural tube are involved in the pathogenesis of anencephaly. In anencephaly anomalies the offspring are usually born dead. Even if live-borns usually don't have a chance to survive. Anencephalic cases generally do not have skin, and hemispheriums are replaced by the area cerebrovasculosa an irregular mass consisting of a mixture of nerve tissue and blood vessels.<sup>5,6</sup> The phenomenon of anencephaly often does not occur as a single anomaly. Anencephalic anomalies occur along with developmental disorders such as acrania, agnathia, anophthalmia, arhinia, anotia, aglossia, craniosynostosis, myeloschisis, cheiloschisis, and palatoschisis. Acrania is the absence of the calvarial bone that preserves the brain; arrhinia is the absence of the nose; anotia is the absence of the ear; aglossia is the absence of the tongue. Craniosynostosis is the bifidity of the calvarian bone; myeloschisis is the bifidity of the cheek; cheiloschisis is of the lip; and palatoschisis is of the palate. Although the etiology of the congenital malformations of the CNS has not been definitively explained, toxic-infectious diseases, hereditary and environmental factors have been suggested to play a role.<sup>1,7,8</sup>

The case being presented here might be of interest because it reports anencephaly and other head anomalies that have not been previously reported in lambs. In addition, some evaluations were made on the etiology of congenital anomalies based on this case.

## CASE PRESENTATION

The case material consisted of the offspring of a five-year-old Akkaraman breed sheep brought to Yüzüncü Yıl University, Faculty of Veterinary Medicine, Department of Obstetrics and Gynecology with the complaint of difficult birth. Examination of the birth canal revealed that the fetus was swollen and therefore could not leave the canal. When it was understood that there could be no normal delivery, a cesarean section was decided. The lamb, which was removed from the birth canal after the operation was performed by obtaining the consent form from the patient owner, was found to have no vital activity. In addition, the

examination showed that the fetus died in the womb. No significant clinical symptoms were observed in the mother, who was kept under surveillance for 24 hours. The offspring with congenital malformations were sent to the Department of Pathology to determine the existing anomalies and undergo a macroscopic examination.

## Macroscopic Findings

Examination revealed that the offspring died approximately three days earlier. It was observed that the placenta carried with the offspring was dark. It was determined that the congenital malformations in the offspring were mostly located in the head and neck region (Fig. 1A). It was the first remarkable finding that the calvarial bone was not shaped in the offspring, i.e., acrania. The presence of a cystic pouch with a fluctuant content was detected instead of the head, which was not protected by the skin.

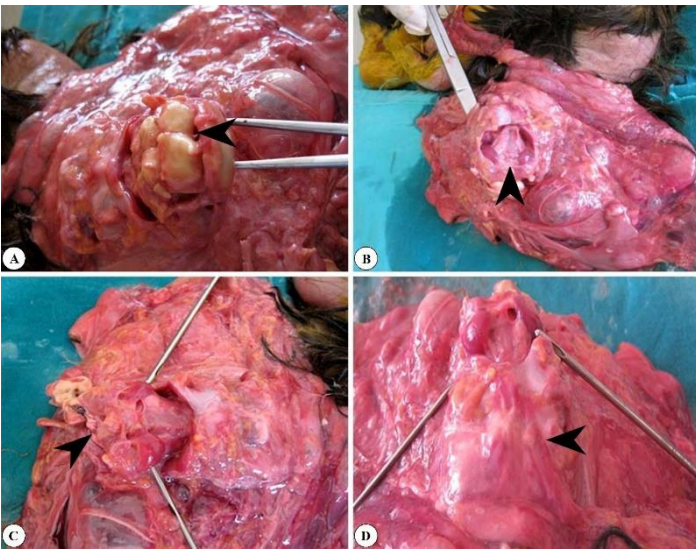


**Figure 1.** A) Cystic structure under the undeveloped scalp (arrowhead). B) The mouth, nose, ears and eyes are not formed (arrowhead). C) Air sacs containing otolytic fluid in the cystic structure (arrowhead). D) Fully formed hyoid bone, larynx, trachea, and esophagus under the air sacs (arrowhead).

A rudimentary residue consisting of immature nerve and vein formations was detected in and around the connective area of this cystic formation. Since the facial bones did not form to a large extent, it was observed that no facial organs such as the jaw (agnathia), nose (arhinia), ear (anotia), and eye (anophthalmia) were formed (Fig. 1B). With the opening of the cystic formation in the head, two small air sacs in the form of gas bubbles and autolytic tissue residues with some amount of smelly brownish fluid were detected in the sac (Fig. 1C). Inside this cystic formation, a small unformed tongue and hyoid bone were found. It was

found that the larynx and the following esophagus and trachea were formed when advanced to the throat region (Fig. 1D).

The spinal cord was very regular and reached exactly the os occipitale, and there was a blocked spinal canal just after the atlanto-occipital joint, just like a plug (Fig. 2A). With this plug-like behavior, it was seen that the nervous system pathway was blocked and the nervous system ended in this region (Fig. 2B). It was found that hemispherium, cerebellum, mesencephalon, and pons were not shaped at all in this anencephalic case (Fig. 2C). Medulla oblongata was found to be present as a rudimentary remnant. It was determined that labor could not be started because the hypothalamus did not form, and therefore, the tissues were severely autolytic (Fig. 2D). Autolytic changes were quite common in the rest of the body.



**Figure 2.** A) The spinal cord forms a plug at the atlanto-occipital joint (arrowhead). B) Termination of the nervous system with the plug formed by the spinal cord (arrowhead). C) The hemispheres, cerebellum, mesencephalon and pons are not formed at all (arrowhead). D) Excessive autolysis and adhesions in other internal organs (arrowhead).

## DISCUSSION

Although studies on mammalian anomalies caused by inadequate development in the intrauterine period are quite insufficient, the incidence of congenital malformations in sheep has been reported to be between 0.5-1%.<sup>1,9</sup> This prevalence cannot be underestimated, taking into account the cases that may not have been reported. In order to address the importance of congenital anomalies and draw attention to their etiologies, such cases should be reported, and their etiology should be scrutinized. CNS anomalies are among the most common congenital malformations in domestic animals. CNS

anomalies are more common than other system and organ anomalies. This is due to the fact that the embryonic development of the CNS is earlier than other systems and organs. It has also been suggested that complications in the embryonic development of the CNS may be effective in the development of abnormal encephalon.<sup>5,6</sup> Although anomalies of the CNS, which has a complex embryogenesis and a long intruterine process since the early stages of pregnancy, are more common, few reports have been made regarding this system.<sup>2,4</sup> In Turkey, Hazıroğlu et al., for the first time, reported a calf case with anencephaly and various accompanying congenital malformations.<sup>10</sup> Dogan et al., in their examination of 6 calves with congenital anomalies, they reported that they found findings related to head anomalies in 3 of them.<sup>11</sup> Due to the fact that the head region consists of important organs, anomalies here have vital consequences. Especially the anomalies in the CNS, which is the root of the organism, are the causes of functional disorders in other organs and systems of the body that appear morphologically normal.

Cranial malformations do not usually occur alone and more than one anomaly is observed together.<sup>3,12</sup> Many cranial anomalies were also present in this case. Lakkawar et al. who reported a calf case with anencephaly and associated malformations, reported that they observed erroneous and inadequate development of the tongue and mandibula, acrania, anophthalmia, anotia, and arhinia.<sup>13</sup> In addition, Lakkawar et al., stated that there was no skin in the skull area and that there were three cystic formations present here.<sup>13</sup> Hazıroğlu et al., in their study describing anencephaly and accompanying malformations in a calf fetus reported that the upper jaw was short (brachiganti superior), the upper lip had a cleft (cheiloschisis), the palate had a cleft (palatoschisis), the ear was rudimentary (microtia), the eye was small (microphthalmia), and there were no hemispheriums (anencephaly).<sup>10</sup> Hazıroğlu et al. reported in their cases that although hemispheriums were not shaped, calvarial bone was formed, the cerebellum was larger than usual and pituitary was existent.<sup>10</sup> In this presented case, it was observed that more severe malformations were present compared to the case reported by Hazıroğlu et al. However, similar to the case reported by Lakkawar et al. anophthalmia, anotia, arhinia, acrania and anencephilia were detected.

Various factors, such as genetic disorders, infectious and toxic diseases, fetal hypoxia, nutritional deficiency, trauma, and irradiation play a role in the etiology of CNS anomalies.<sup>14,15</sup> According to the anamnesis obtained from this case, it was stated that such anomalies were not previously seen in the herd, one-way nutrition was performed, there was no epidemic infection in the herd, and there was no condition that would lead to poisoning.



In light of the anamnesis and literature information obtained, it was thought that the anomaly in this case may have been caused by nutritional deficiency.

As a result, the findings of a case with anencephaly and related malformations such as anophthalmia, arhinia, anotia, and agnathia in a lamb were reported for the first time in the world with this study. In addition to these morphological changes determined in the case of congenital anomalies, an evaluation was made on possible etiological causes, and it was thought that malnutrition may have a role in the pathogenesis of such anomalies.

**Hakem Değerlendirmesi:** Dış bağımsız.

**Yazar Katkıları:** [Muhammet Bahaeddin DÖRTBUDAK (MBD), Gamze ÇAKMAK (GÇ), Serkan YILDIRIM (SY), Funda EKŞİ (FE), Fatma İLHAN (Fİ), Nebi ÇETİN (NÇ), Sait ŞENDAĞ (SŞ): Fikir-GÇ; Tasarım-SŞ; Denetleme-NÇ; Kaynaklar-SY; Veri Toplanması ve/veya İşlemesi-MBD; Analiz ve/veya Yorum-Fİ; Literatür Taraması-MBD; Yazıyı Yazan-SY; Eleştirel İnceleme-Fİ]

**Çıkar Çatışması:** Yazarlar, çıkar çatışması olmadığını beyan etmiştir.

**Finansal Destek:** Yazarlar, bu çalışma için finansal destek almadığını beyan etmiştir.

**Peer-review:** Externally peer-reviewed.

**Author Contributions:** [Muhammet Bahaeddin DÖRTBUDAK (MBD), Gamze ÇAKMAK (GÇ), Serkan YILDIRIM (SY), Funda EKŞİ (FE), Fatma İLHAN (Fİ), Nebi ÇETİN (NÇ), Sait ŞENDAĞ (SŞ): Concept -GÇ; Design-SŞ; Supervision-NÇ; Resources-SY; Data Collection and/or Processing-MBD; Analysis and/or Interpretation-Fİ; Literature Search-MBD; Writing Manuscript-SY; Critical Review-Fİ]

**Conflict of Interest:** The authors have no conflicts of interest to declare.

**Financial Disclosure:** The authors declared that this study has received no financial support.

## REFERENCES

- Jubb KVF, Kennedy PC, Palmer N. Pathology of domestic animals. Fourth Edition. Academic Press. United States. Vol. 1, 2013, pp. 270-286.
- Singh M, Parrah JUD, Moulvi BA, Athar H, Kalim MO, Dedmari, FH. A review on pectus excavatum in canines: A congenital anomaly. *Iran J Vet Surg.* 2013;8(1):59-64.
- Dos Anjos Nonato I, Teixeira MR, De Miranda JL, Braz HMB, Machado JP. Cranioschisis and anencephaly in a dog-challenging etiology. *Acta Sci Vet.* 2019;47(1):449.
- İşler CT, Altuğ M, Gönenci R, Aytekin İ. Evaluation of anomaly cases detected in calves in Bolu region between 2005-2009. *Harran Üniv Vet Fak Derg.* 2016;5(2):100-104.
- Di Muro G, Cagnotti G, Bellino C, Capucchio MT, Colombino E, D'Angelo A. Multiple Cephalic Malformations in a Calf. *Animals.* 2020;10(9):1532.
- Silva LF, Rabelo RE, Santos GP, et al. Multiple congenital encephalic malformations in a calf. *Braz J Vet Pathol.* 2016;(9):103-107.
- Islam MR, Roh YS, Cho A, et al. Multiple congenital anomalies in a Korean native calf (*Bos taurus coreanae*). *Korean J Vet Res.* 2011;51(1):63-67.
- Ortega-Pacheco A, Lezama-García MA, Colín-Flores R. Presence of congenital anomalies in three dog litters. *Reprod Domest Anim.* 2020;55(5):652-655.
- Raofi A, Dehghan MM, Mardjanmehr SH, et al. Cranium bifidum with meningocele in a lamb. *Small Rumin Res.* 2004;55(1-3):253-256.
- Haziroğlu RM, Atasever A, Yildiz D. Anencephaly and other congenital malformations in a calf [Turkish]. *Ankara Univ Vet Fak Derg.* 1996;(43):303-305.
- Doğan GK, Karakurt E, Kuru M, Nuhoglu, H. Anatomical and pathological evaluation of congenital anomalies in calves: 6 cases. *Pakistan J Zool.* 2022;54(3):1345-1354.
- Yurdakul İ, Yalçın M, Karataş Ö. A Case of Multiple Congenital Anomaly in a Calf. *Erciyes Üniv Vet Fak Derg.* 2020;17(3):342-345.
- Lakkawar AW, Kumar R, Nair MG, Varshney KC. Anencephaly and Associated Malformations in a Ovine Foetus. *Folia Vet.* 2002;46(1):39-41.
- Rousseaux CG, Ribble CS. Developmental anomalies in farm animals: II. Defining etiology. *Can Vet J.* 1988;29(1):30-40.
- Samuel M, Wanmi N, Usende L, Ozegebe PC. Review of Some Developmental Anomalies in Domestic Species in North Central Region of Nigeria-Contributions of Normadism and Climate Changes in Embryo-toxicity. *Anim Mol Breed.* 2016;6(1):1-7.