Ensaios e Ciência
ISSN 1415-6938 e ISSN 2178-695X

DOI: https://doi.org/10.17921/1415-6938.2025v29n1p151-160

Genitourinary and Skeletal Congenital Malformations in Lamb: Case Report in Western Region of São Paulo

Malformações Congênitas Geniturinárias e Esqueléticas em Cordeiro: Relato de Caso no Oeste Paulista

Received on: 27/08/2024
Accepted on: 08/10/2024

Gabriella Capitane Sena: Universidade do Oeste Paulista, Programa de Pós-Graduação em Ciência Animal. SP, Brasil.

Isabella Guartieri da Silva: Universidade do Oeste Paulista, Programa de Pós-Graduação em Ciência Animal. SP, Brasil.

Felipe Rydygier de Ruediger: Universidade do Oeste Paulista, Faculdade de Ciências Agrárias. SP, Brasil.

Rejane Batista Brinholi: Universidade do Oeste Paulista, Faculdade de Ciências Agrárias. SP, Brasil.

Leticia Jalloul Guimarães: UniFatecie Centro Universitário. PR, Brasil.

Maria Fernanda Gibim: Universidade do Oeste Paulista, Programa de Pós-Graduação em Ciência Animal. SP, Brasil.

Isadora de Souza e Silva: Universidade do Oeste Paulista, Programa de Pós-Graduação em Ciência Animal. SP, Brasil.

Rayane Cardoso Melozo: Universidade Anhanguera Unopar, Programa de Pós-Graduação em Saúde e Produção Animal. PR, Brasil.

Fabíola Cristine de Almeida Rêgo: Universidade Anhanguera Unopar, Programa de Pós-Graduação em Saúde e Produção Animal. PR, Brasil.

Marilice Zundt: Universidade do Oeste Paulista, Programa de Pós-Graduação em Ciência Animal. SP, Brasil.

Abstract
Congenital malformations are structural defects that occur during intrauterine development and can result from genetic, environmental, or infectious factors. Susceptibility to these malformations varies according to the embryonic development stage. In Brazil, the ingestion of toxic plants, is a significant cause of deformities in ruminants, especially sheep, leading to conditions like arthrogryposis and craniofacial anomalies. This report describes a rare case of genitourinary and skeletal malformations in a lamb born in Pirapozinho, SP, in September 2022. The animal presented changes incompatible with life, leading to euthanasia. The necropsy revealed several anomalies, including vertebral deformities, pulmonary edema, cardiac hypertrophy, and anal atresia. The presence of the toxic plant juá bravo (*Solanum viarum*) on the property suggests its possible contribution. The observed malformations indicate a multifactorial etiology, involving potential nutritional deficiencies, plant toxins, and maternal infections. This case highlights the complexity of the causes of congenital malformations in sheep and underscores the need for comprehensive preventive approaches, such as nutritional monitoring, toxin control, and infection surveillance, to minimize these risks in sheep.

Keywords: Autopsy. Birth Anomaly. Ovine. Veterinary Pathology.

Resumo
Malformações congênitas são defeitos estruturais que ocorrem durante o desenvolvimento intrauterino e podem resultar de fatores genéticos, ambientais ou infecciosos. A susceptibilidade a essas malformações varia de acordo com o estágio de desenvolvimento embrionário. No Brasil, a ingestão de plantas tóxicas, é uma causa significativa de deformidades em ruminantes, especialmente ovinos, resultando em condições como artrogripose e anomalias craniofaciais. Este relato descreve um raro caso de malformações geniturinárias e esqueléticas em um cordeiro nascido em Pirapozinho, SP, em setembro de 2022. O animal apresentava alterações incompatíveis com a vida, levando à eutanásia. A necropsia revelou diversas anomalias, incluindo deformidades vertebrais, edema pulmonar, hipertrofia cardíaca e atresia anal. A presença da planta tóxica juá bravo (*Solanum viarum*) na propriedade sugere sua possível contribuição. As malformações observadas indicam uma etiologia multifatorial, envolvendo possíveis deficiências nutricionais, toxinas de plantas e infecções maternas. Este caso reforça a complexidade das causas das malformações congênitas em ovinos e destaca a necessidade de abordagens preventivas abrangentes, como monitoramento nutricional, controle de toxinas e vigilância de infecções, para minimizar esses riscos em rebanhos ovinos.

Palavras-chave: Anomalia de Nascimento. Necropsia. Ovinos. Patologia Veterinaria.

1 Introduction

Congenital malformations, also known as birth anomalies, are structural defects that develop during intrauterine life, affecting the formation of organs, bones, or entire systems. These malformations may result from genetic factors, environmental influences, infectious diseases, or a combination of these factors (WHO, 2022).

Susceptibility to congenital defects varies according to the stage of embryonic development at the time of exposure to teratogenic agents. In the early days after fertilization, the embryo is more resistant to environmental influences but highly vulnerable to genetic mutations and chromosomal aberrations (Paniagua; Jacob, 2023). On the other hand, between the 10th and 40th day of gestation in sheep, the embryo becomes more susceptible to environmental teratogenic influences, such as intoxications,

nutritional deficiencies, infectious diseases, and radiation (Santa Rosa, 1990; Succu *et al*., 2023).

A study on congenital anomalies in sheep, conducted in Rio Grande do Sul, identified isolated cases of abnormalities, with defects affecting various systems, such as the skeletal, cardiovascular, respiratory, digestive, genitourinary, and nervous systems, including anomalous twins and total or partial absence of the head (Marcolongo-Pereira *et al*., 2010).

In Brazil, the ingestion of toxic plants is one of the main causes of abortions and malformations in ruminants (Bezerra; Pinheiro; Lucena, 2021; Riet-Correa; Medeiros; Lima Bezerra; Lucena, 2024; Schild, 2012; Withoeft *et al*., 2024). Among the toxic plants with teratogenic or abortifacient effects, *Mimosa tenuiflora* (black jurema) stands out. Its intoxications cause various malformations, especially in the head and limbs. These deformities frequently occur in sheep and goats (Nogueira *et al*., 2022).

The objective was to report a rare case of genitourinary and skeletal congenital malformations in a lamb born on a sheep farming property in the interior of São Paulo state, describing the clinical and pathological findings observed and discussing possible etiologies and associated risk factors.

2 Case Report

This case report describes a rare occurrence of multiple malformations in a lamb (Figure 1) born on a rural property in Pirapozinho, SP, during the birth season in September 2022. The lamb was born to a three-year-old Santa Inês ewe, which was fed commercial concentrate and grazed predominantly on *Panicum maximum* 'Aruana'. The birth was twin, with the other lamb being male and showing no anomalies. According to the owner, the ewe had no history of complications in two previous births. Both the ewe and the surviving lamb underwent clinical examination and were found to be healthy.

Figure 1 - Neonatal lamb presenting skeletal deformities of the spine and forelimbs

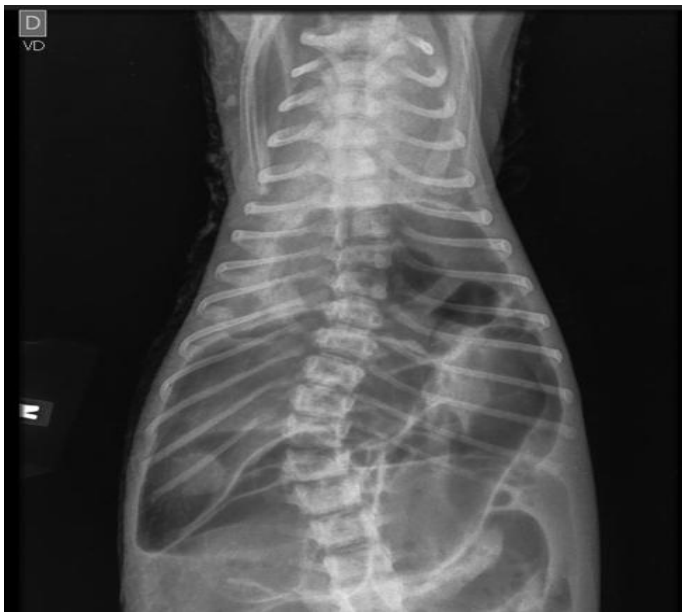
Source: the authors.

The property used the anthelmintic Nitroxinil 34%, but there were no records of application during the gestational period. The presence of the toxic plant *Solanum viarum* (juá bravo) was also observed on the property, suggesting a possible contributing factor to the malformations.

The malformed lamb was born alive but exhibited difficulty in locomotion, pronounced vocalization, agenesis of the reproductive system, and anal atresia. Due to incompatibility with life, euthanasia was necessary a few hours after birth.

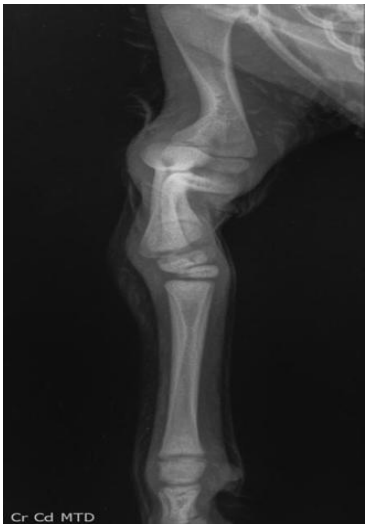
After euthanasia, the lamb was sent to the Veterinary Hospital of the University of Western São Paulo, Campus 2, in Presidente Prudente – SP, for detailed examinations. Radiographs were taken in various projections to assess the skeletal system. The ventrodorsal radiograph (Figure 2) revealed the presence of 14 pairs of ribs and 14 thoracic vertebrae, while another radiographic image showed 5 lumbar vertebrae. Additionally, deformities in the radius and ulna of the right forelimb were observed (Figure 3). A necropsy was conducted at the Animal Pathology Laboratory to examine the affected systems.

Figure 2 - Ventrodorsal radiograph allowing the analysis of 14 thoracic vertebrae and 14 pairs of ribs in a lamb with skeletal malformations



Source: the authors.

Figure 3 - Radiograph of the right forelimb showing deformities in the radius and ulna

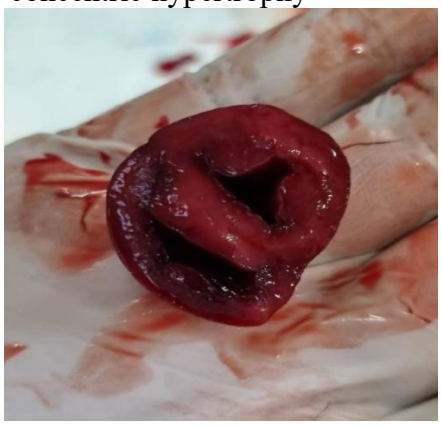


Source: the authors.

The lamb's necropsy revealed a series of significant anomalies affecting multiple organ systems. In the respiratory system, marked pulmonary edema and diffuse congestion were observed, indicating excessive fluid accumulation in the lungs and possible circulatory complications. These changes suggest respiratory failure or blood circulation issues that could be related to structural abnormalities in the respiratory or cardiovascular system.

In the cardiovascular system, bilateral concentric hypertrophy of the heart was identified (Figure 4), characterized by thickening of the ventricular walls. This alteration is often an adaptive response to pressure or volume overload and may indicate the presence of congenital heart anomalies or heart failure secondary to other malformations.

Figure 4 - Interior of the heart (cross-section through the ventricles) showing bilateral concentric hypertrophy

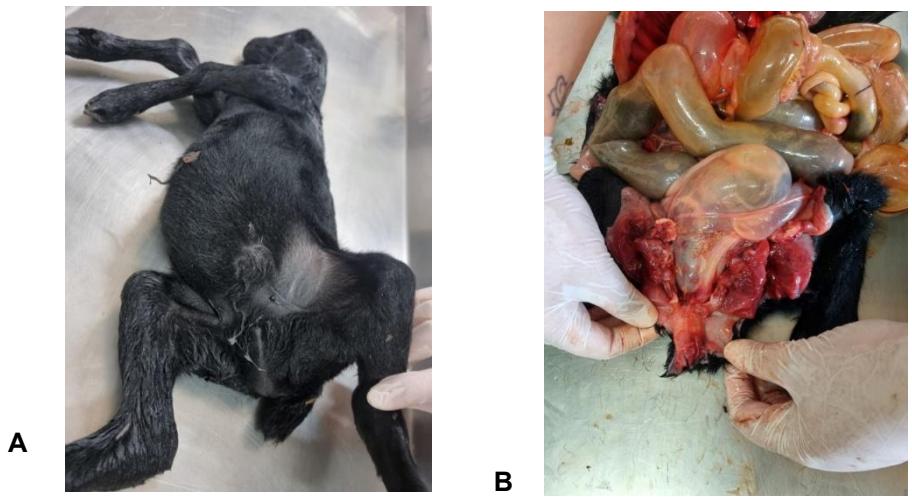


Source: the authors.

Regarding the digestive system, the necropsy revealed moderate diffuse hepatic congestion, possibly associated with heart failure, resulting in poor hepatic venous drainage. Additionally, rectal and anal atresia was identified, a complete obstruction of the lower digestive tract that prevents normal excretion. This condition can lead to abdominal distension, accumulation of fecal content, and metabolic complications.

The lamb exhibited agenesis of the reproductive system, showing the absence of the formation of internal and external genital organs (Figure 5).

Figure 5 - Lamb presenting anal atresia and reproductive system agenesis. A) Before necropsy, showing the absence of external reproductive organs and anus; B) During necropsy, confirming the absence of internal reproductive organs and complete obstruction of the final portion of the digestive tract



Source: the authors.

Bilateral renal congestion and patent urachus were observed in the urinary system, a condition in which the urinary duct connecting the bladder to the navel does not close properly after birth, potentially leading to urinary infections and other complications.

In the skeletal system, notable anomalies were detected, such as an additional pair of ribs and thoracic vertebrae, which differs from the normal anatomical pattern of sheep, which have 13 pairs of ribs and 13 thoracic vertebrae. Furthermore, the absence of a cervical vertebra was observed, a deformity that may be associated with the clinically observed neurological abnormalities, including ataxia and dysmetria.

These findings indicate a complex interaction of multiple congenital factors that resulted in the various observed anomalies. The combination of structural defects in the respiratory, cardiovascular, digestive, genitourinary, and skeletal systems suggests a multifactorial etiology, possibly involving genetic and environmental factors, such as exposure to toxic plants or nutritional deficiencies during gestation.

The analysis of the different systems of the malformed lamb revealed significant congenital anomalies, suggesting a multifactorial etiology. Congenital malformations in sheep, such as those observed in this study, may occur due to a range of factors, including genetic, environmental, and infectious influences.

In accordance with the data from Kumar *et al*. (2010) and Frey *et al*. (2018), anomalies or congestion in various systems can be attributed to impaired venous blood flow, resulting in inadequate venous drainage and distension of veins, venules, and distal capillaries. This congestion pattern observed in the lamb's respiratory and digestive systems is consistent with previous reports, suggesting a common pathophysiological response to a range of teratogenic insults.

Regarding the skeletal system, the presence of 14 pairs of ribs and 14 thoracic vertebrae differs from the typical anatomy of sheep, which have 13 pairs of ribs and 13 thoracic vertebrae. This anatomical variation is indicative of a significant skeletal malformation that may have multiple etiologies.

Previous studies, such as those by Santa Rosa (1990), Capone and Sentongo (2019), and Kiani *et al*. (2022), indicate that nutritional deficiencies, particularly of vitamins A and E, selenium, copper, iodine, zinc, and manganese, are predisposing factors for the development of skeletal deformities. In the present case, the bone deformity may be correlated with a possible nutritional deficiency in the ewe during gestation, although no laboratory confirmation of this hypothesis was obtained.

The literature also highlights the importance of environmental factors, particularly the ingestion of toxic plants, in the pathogenesis of congenital malformations in ruminants (Bezerra; Lucena, 2024; Brülisauer *et al*., 2017; Johnsen *et al*., 2018; Leong *et al*., 2017). In the specific case of Juá-bravo (*Solanum viarum*), the plant contains steroidal alkaloids known for their toxicity to animals.

Although it is not widely reported as a plant directly associated with congenital malformations, plants with similar toxins, such as *Aristolochia* spp., *Veratrum* spp., and *Lupinus* spp. (Dirks *et al*., 2021; Khan *et al*., 2024; Seale; McDougal, 2022; Schneidewind *et al*., 2024), are documented as causes of congenital defects in ruminants. If the lamb’s mother ingested this plant during gestation without it being noticed, it is possible that the exposure led to fetal developmental alterations, resulting in malformation.

The toxicity of these plants varies according to the stage of gestation, the type, and the quantity ingested, with teratogenic plants tending to be more harmful when consumed early in pregnancy, during the critical periods of embryonic development (Panter *et al*., 1998).

The findings in the reproductive system of this lamb represent a rare discovery, as they contradict the literature, where the reported alterations are usually partial, and it is possible to identify the animal's sex. In this case, the lamb had no internal or external reproductive organs (Salvador, 2021; Marcolongo-Pereira *et al*., 2010; Ladds, 1993).

Additionally, the persistence of the urachus was found, which is a tubular structure that connects

the fetal urinary bladder to the allantois during fetal development, allowing the excretion of urine-like aqueous fluid. After birth, this structure normally closes and transforms into the median umbilical ligament (Bassert; Colville, 2010; Noden; De Lahunta, 1985).

Several authors report that this is a congenital or acquired anomaly that interferes with the normal development of the urinary tract and can lead to additional complications, as the umbilical stump may remain moist or continuously drip urine through the navel, which can lead to infections (Tatekawa, 2019; Dos Santos Sousa *et al*., 2017).

Dittmer and Thompson (2015) and Macêdo *et al*. (2011), in their investigation of abnormalities, report that when an abnormality is restricted to a single animal, the cause is usually impossible to fully diagnose, except in cases where the anomalies are preexisting in specific breeds or if the gene can be detected.

This confirms the observations of Dennis and Leipold (1986) that congenital defects, such as rectal atresia, ankylosis, and reproductive system agenesis, may sporadically arise in herds without a well-defined cause, as observed in this study, where multiple defects were identified without a clear etiological cause.

In summary, the analysis of this case suggests a complex combination of genetic, environmental, and possibly infectious factors that resulted in the multiple observed anomalies. This multiplicity of etiological factors reflects the complexity of the pathogenesis of congenital malformations in sheep, as described in the literature. The integration of this study's findings with other reports underscores the need for a comprehensive approach in managing sheep pregnancies, including rigorous nutritional monitoring, control of exposure to toxic plants, and continuous surveillance for infectious agents.

4 Conclusion

This study documented a rare case of multiple congenital malformations in a lamb, reinforcing the multifactorial etiology, which may involve genetic, environmental, and possibly infectious factors. The identified anomalies highlight the importance of stricter monitoring during gestation, particularly concerning nutrition and toxin exposure, as well as continuous surveillance for potential infections. The absence of a clear etiological diagnosis underscores the need for further investigations to better understand the associated risk factors and prevent similar cases in sheep flocks. Future studies may contribute to improving management strategies, helping breeders minimize the incidence of malformations and promote animal's health.

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