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Case of Chondrodysplasia in a Holstein Calf

Holstein Irkı Bir Buzağıda Kondrodisplazi Olgusu

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ABSTRACT

The aim of this study was to describe pathomorphologically a case of chondrodysplasia observed in a newborn Holstein calf. According to the anamnesis, the first remarkable finding in the calf, which was delivered in the Department of Obstetrics and Gynecology of our faculty, was that the joints were easily broken during birth. Multiple anomaly findings were observed in the calf that was born. Macroscopic examination revealed that the calf had a disproportionately short body and an enlarged skull. It was also observed that the forelimbs and hind limbs were short and arthrogryposis, scoliosis of the vertebrae and herniation in the umbilical region. When the chest cavity was opened, it was noted that the costae and sternum were not fully developed and the vertebrae were curved. It was noticed that the trachea was smaller and curved than normal and the bronchial structures of the lung were not formed. When the skull was opened, hydrocephalus was noted. The most striking finding was the softness and fragility of all the body bone and cartilage tissues. For histopathological examination, tissue samples were fixed in a 10% buffered formalin solution for 48 hours and decalcified. Following this, routine tissue processing procedures, 4-5 µm thick sections were taken with a microtome, stained with haematoxylin-eosin (H&E), and examined under a light microscope. Histopathological examination revealed degenerative and hypertrophic chondrocytes, as well as incomplete vascularization in the affected area.

Keywords: Calf, Chondrodysplasia, Histopathology

ÖZ

Bu çalışmada Holstein ırkı, yeni doğmuş bir buzağıda gözlenen kondrodisplazi olgusunun patomorfolojik olarak tanımlanması amaçlandı. Alınan anamnez bilgilerine göre, fakültemiz Doğum ve Jinekoloji Anabilim dalında doğumu gerçekleştirilen buzağıda ilk dikkat çekici bulgu doğum esnasında eklemlerinin kolayca kırılması durumuydu. Doğumu gerçekleştirilen buzağıda birden fazla anomali bulguları gözlendi. Yapılan makroskobik incelemede buzağının gövdesinin kısa, kafatasının ise oldukça büyük şekillendiği görüldü. Ayrıca ön ve arka bacakların kısa ve artrogripozisli olduğu, omurlarda skolyoz ve ayrıca umblikal bölgede fıtıklaşmanın şekillendiği gözlendi. Göğüs boşluğu açıldığında ise kostalar ve sternumun tam gelişmediği, vertebraların eğri olduğu dikkat çekti. Trakeanın normalden küçük ve eğri şekillendiği, akciğer bronş yapılarının şekillenmediği fark edildi. Kafatası açıldığında ise hidrosefalus dikkati çekti. En dikkat çekici bulgu ise vücudun tüm kemik ve kıkırdak dokusunun yumuşak ve kırılgan olmasıydı. Histopatolojik inceleme amacıyla doku örnekleri %10'luk tamponlu formalin solüsyonunda 48 saat süreyle fikse edilmiş ve dekalsifikasyon işlemi uygulanmıştır. Bunu takiben, rutin doku işleme prosedürleri gerçekleştirilmiş, mikrotom ile 4-5 µm kalınlığında kesitler alınmış, hematoksilen-eozin (H&E) ile boyanmış ve ışık mikroskobu altında incelenmiştir. Histopatolojik incelemelerde; dejeneratif ve hipertrofik kondrositlerin yanısıra bu bölgede damarlaşmanın da tam şekillenemediği gözlendi.

Anahtar Kelimeler: Buzağı, Kondrodisplazi, Histopatoloji

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INTRODUCTION

Congenital anomalies are pathological conditions that can occur during embryonic or fetal development in all animal species including structural or functional defects in tissues, organs, or systems.[1] These malformations are common worldwide and can lead to fetal losses, neonatal deaths, or pregnancy termination. In cattle, in particular, such anomalies result in significant reproductive losses and cause economic losses in the livestock industry.[2]

Although the etiology of congenital malformations has not been fully elucidated, both hereditary and non-hereditary factors have been reported to play a role in the development of these anomalies.[3]

The etiological factors of non-hereditary congenital anomalies in animals include genetic predisposition,3 faulty breeding selection, breed characteristics, consanguinity,[4] parental age, stress factors, nutritional disorders,[5] vitamin deficiencies, exposure to teratogens, and endocrine disorders.[6]

In addition, factors such as fetal position in the uterus, exposure to carcinogenic substances, and pregnancy diagnosis by rectal palpation before day 42 or by rectal ultrasonography between days 28 and 90 of gestation may also contribute to the development of congenital malformations.[2] Mutant genes or chromosomal abnormalities have been shown to play a role in the occurrence of inherited malformations.[3] Chromosomal changes typically manifest as a broad spectrum of malformations rather than as isolated deformities.[1]

Chondrodysplasia, a genetic disorder, is defined as an extremely rare abnormality in cartilage growth or development, which can affect genes or chromosomes and result in varying degrees of disproportionate dwarfism.[2] It typically disrupts transverse or longitudinal cartilage growth, negatively impacting the entire skeletal system. Chondrodysplasia is characterized pathologically by the destruction of endochondral osteogenesis.[6] The probability of birth defects in calves has been reported to be between 0.2% and 0.3%, with 26.6% of these cases involving skeletal and muscular anomalies.[7]

In addition to cattle, chondrodysplasia has also been reported in dogs, pigs, rabbits, and sheep as well as in humans and mice. The aim of this study was to describe pathomorphologically a case of chondrodysplasia observed in a newborn Holstein calf.[5]

CASE PRESENTATION

The subject of this case report was a newborn Holstein calf that was delivered at Atatürk University, Faculty of Veterinary Medicine, Department of Obstetrics and Gynaecology, and died a few minutes after birth. Bone samples obtained from the systemic necropsy were fixed in 10% buffered formalin solution for 48 hours and decalcified in 36.8% formic acid and 6.8% sodium formate for histopathological examinations. They were then embedded in paraffin blocks following a routine tissue processing procedure. Sections of 4–5 µm thickness were taken using a microtome, stained with haematoxylin-eosin (H&E), and examined and imaged with an Olympus BX52 light microscope (Japan) equipped with a DP72 camera system.[8]

Macroscopic examination revealed that all bones of the calf were extremely soft and fragile at birth (Figure 1A). The body of the calf was short, and the skull was disproportionately large. The forelimbs and hind limbs were shortened, and arthrogryposis, scoliosis of the vertebrae, and herniation in the umbilical region were observed (Figure 1B). Upon opening the skull, hydrocephalus was detected (Figures 1C–D). Examination of the thoracic cavity revealed that the ribs and sternum were underdeveloped, and the vertebrae were curved (Figure 1E). Additionally, the trachea was smaller and more curved than normal, and the bronchial structures of the lungs were absent (Figure 1F).

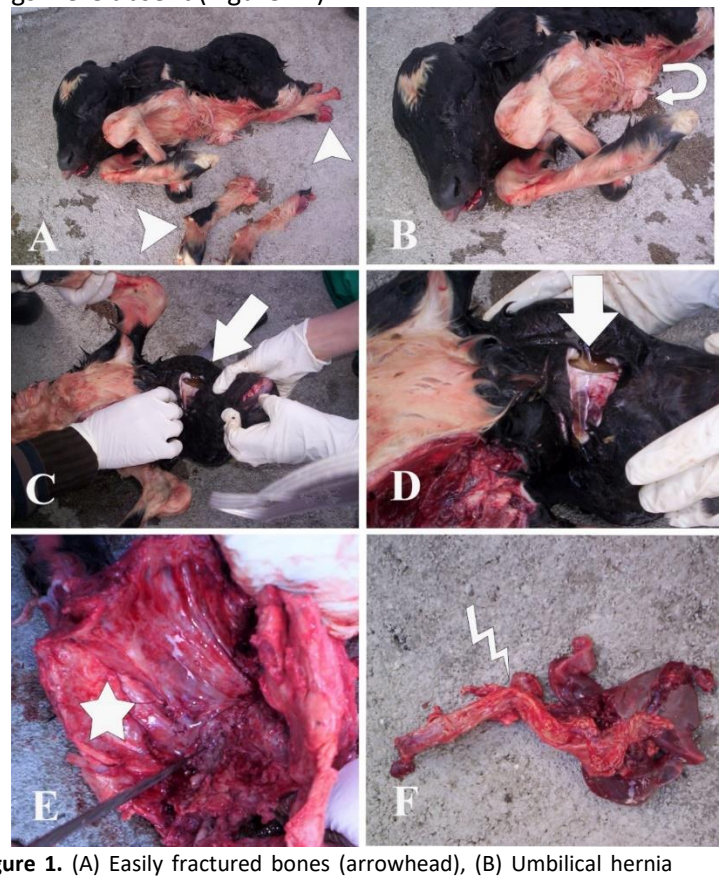

Figure 1. (A) Easily fractured bones (arrowhead), (B) Umbilical hernia (curved arrow), (C–D) Hydrocephalus (arrow), (E) Underdeveloped ribs and sternum (star), (F) Smaller than normal and curved trachea (lightning).

Microscopic examination revealed an increased number of immature chondrocytes, along with decreased or

incomplete vascularization and calcification (Figure 2A–B). Degenerative changes were observed in mature chondrocytes (Figure 2C–D).

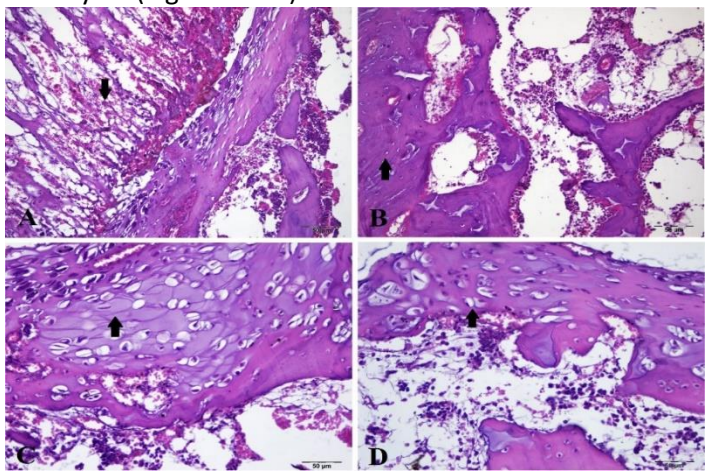

Figure 2. (A–B) Incomplete calcification (arrow), (C–D) Mature chondrocytes and degenerating chondrocytes (arrow), (H&E, 50 µm).

DISCUSSION

Although congenital anomalies in cattle are rare in cattle breeding, they represent a serious problem that can lead to significant economic losses. Various studies on the etiology of these anomalies have shown that multiple factors, including genetic predisposition,[3] environmental influences, and nutritional deficiencies,[5] may play a role.

Genetic factors have been identified as a significant contributor to the development of chondrodysplasia in cattle, and this condition has been shown to be fatal in affected individuals.[3] Chondrodysplasia is a skeletal malformation characterized by defects in endochondral osteogenesis. Macroscopic findings vary widely; however, the common feature in all cases is a disruption in transverse and longitudinal bone development.[6]

The term 'bovine prenatal fatal chondrodysplasia' refers to a genetically heterogeneous group of chondrodysplasias and is characterized by severe micromelic dwarfism.[9] Hereditary chondrodysplasia has been reported in humans, cattle, sheep, dogs, pigs, rabbits, and mice. According to the anamnesis, an aborted fetus with similar findings had previously been obtained from the same bull on this farm, leading to the conclusion that this condition may be hereditary.

It has been reported that in cases of chondrodysplasia, intrauterine death typically occurs between the 6th and 8th months of gestation; however, some affected calves may be born alive, with the majority of these cases classified as bulldog calves.[6] In the present case, chondrodysplasia was observed in a Holstein calf, which died a few minutes after birth. The results of our study aligned with those reported in the existing literature.

Generalized chondrodysplasia is primarily characterized by cartilage defects[10] and is associated with various skeletal and soft tissue abnormalities, including disproportionate dwarfism,[11] a large and domed skull,[10] short limbs,[11] a protruding tongue,[11] cleft palate,[11] tracheal malformations, underdeveloped lung lobes,[12] and ventral abdominal hernia.[13]

Various forms of chondrodysplasia have been identified in cattle, including snorter (brachycephalic), Dexter, and Telemark chondrodysplasias. Generalized chondrodysplasias have been observed in Dexter Holstein,[11,12] Jersey,[14] Highland,[15] Belted Galloway,[6] Nellore,[7] and Miniature Zebu,[16] cattle, and these conditions are collectively referred to as bulldog-type dwarfism.

The diagnosis of the bulldog fetus was based on its physical characteristics, including deviations and deformities from normal development, and was confirmed by previously reported cases in different cattle breeds.[12,15]

In our study, chondrodysplasia was detected in a Holstein calf, and the macroscopic findings were similar to those reported in cases of chondrodysplasia anomaly described by Jacinto et al.[11] These changes, which align with previously described findings in different cattle breeds, including Holstein, consist of structural anomalies thought to be associated with genetic mutations.[11,12]

Bulldog-type chondrodysplasia is characteristically defined by short limbs, a bulging abdomen, a flattened head, and micromelia. Such anomalies have been reported in various cattle breeds, including Jersey,[14] Highland,[15] Belted Galloway,[6] Nellore[7], and Miniature Zebu.[16] Similar macroscopic findings were observed in our case and were consistent with the literature.

In previously reported cases, histopathological examination of bones such as the humerus, radius, femur, and tibia revealed that intramembranous ossification occurred beneath the periosteum, whereas endochondral ossification was severely limited.[17] In addition to abnormal chondrocytes, a large number of incompletely matured chondroblasts were observed, and vascularization was found to be insufficient in these areas.[12] The histopathological findings observed in our case were consistent with those reported in the literature.

In conclusion, chondrodysplasia in calves leads to severe skeletal deformities, reduces the animal's quality of life, and causes economic losses. Early diagnosis is crucial to preventing the spread of this genetic disorder within herds. To achieve this, genetic isolation strategies should be implemented, and selective breeding programs should be established by removing individuals carrying the chondrodysplasia mutation from the herd.

Additionally, the widespread adoption of genetic

testing, raising awareness among breeders, and ensuring regular veterinary inspections of calves are critical measures to control the spread of the disease. These actions will not only improve animal welfare but also minimize potential negative impacts on the national economy. Therefore, this case was considered noteworthy and was reported to our department.

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