

ISSN: 2456-298X

Tetra – Amelia with Brachygnathia inferior in a Holstein- Friesian

cross bred calf

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Abstract

A case of live male calf with the congenital tetra – Amelia condition is reported in this paper.

External examination of the calf revealed total absence of all the four limbs. On physical

examination there is brachygnathia inferior, no other phenotypic anomalies were found, while

post-mortem examination revealed the fully developed visceral organs.

Key Words: Tetra-Amelia, Brachygnathia inferior, Calf, Congenital disorder

Introduction

Malformations of the extremities varied in their manifestations, ranging from absence of a single

structure to partial or complete absence of the limbs (Lallo et al., 2001). Amelia means total

absence of one or more limbs, is a rare congenital malformation diagnosed in newborn domestic

animal. In cattle, cases of hemimelia (absence of a portion of a limb) and deformities of hind

limbs have been reported (Corbera et al., 2002). Mosbah et al (2012) reported a case of

acroteriasis congenita in male calf borned after dystocia attributed to hydrocephalus. The calf was

missing all four limbs below the elbow and stifle. There were also defects in the jaw and facial

bones in addition to hydrocephalus. Tetra-amelia is a very rarest congenital disorder diagnosed in

domestic animals with total absence of all four limbs. Previously a case of still born male calf

with the congenital tetra – Amelia condition is reported by Kokila et al. (2014). These defects

tetra- Amelia with brachygnathia inferior have not been reported in a live calf in the literature.



ISSN: 2456-298X

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Case history and observation

A three year old crossbred Holstein- Friesian heifer calved a live male calf with complete absence of all the four limbs. Physical examination of the animal total absence of both the thoracic and pelvic limbs (Fig.1 & 2). Also brachygnathia inferior was noticed. Neck, thorax and trunk were normal. At necropsy findings, development of other visceral organs was absolutely normal. Mandible was 2 inches smaller than maxilla. Calf has normal suckling reflex, defectaion and urinating normally for five days. Calf was euthanized on the request of farmer.

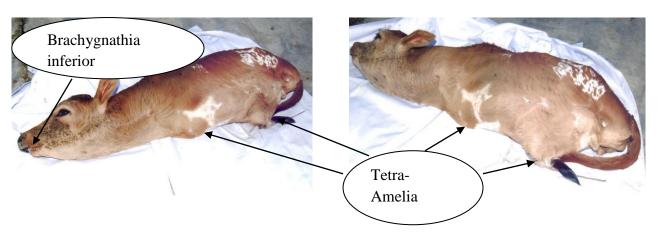


Fig.1 Fig.2

Discussion

The etiology of limb malformation includes hereditary factors, environmental factors, or a combination of both. The complete absence of a limb in amelia occurs as a result of the limb formation process being either prevented or interrupted very early in the developmental stages of embryo. Amelia may be present as an isolated defect, but it is often associated with major malformations in other organ systems. In humans this syndrome can cause severe malformations of other parts of the body, including the face, head, heart, nervous system, skeleton, and



genitalia. The lungs are underdeveloped in many cases, which make breathing difficult or

impossible pattern might be the cause for tetra Amelia condition. In autosomal recessive

inheritance both copies of the gene in each cell attain mutations. Each parents of affected

individual with tetra-amelia syndrome carry one copy of the mutated gene, but do not show signs

and symptoms of the condition. Researchers believe that unidentified mutations in WNT3 or in

other genes that involve in limb development are probably responsible for the disorder in these

cases.

In human beings, a mutation in the WNT3 gene prevents and leads to the other serious birth

defects associated with tetra-amelia syndrome (Niemann et. al. 2004). Chromosome instability

was also diagnosed in a calf affected by congenital malformation namely lack of the distal left

anterior leg and right anterior leg ended with a hook-shaped, nail like structure, high rates of

structural chromosome aberrations and increased yields of sister chromatid exchanges (Di

Berardino et al., 1983). Mutagen-induced chromosome instability was analyzed in cattle, and the

most expressive fragile sites in cows were observed in chromosomes 1 and X. (Danielak and

Slota 2004). Under-developed mandible is due to a recessive gene (Grant 1956). Etiology of

present case of tetra- Amelia with brachygnathia inferior may be due to hereditary and

environmental factors. Subsequent calving cow delivered normal female calf.

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ISSN: 2456-298X



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ISSN: 2456-298X