Case Report

A NOVEL CASE REPORT ON APLASIA CUTIS CONGENITA CO-EXISTENCE WITH ABLEPHARON MACROSTOMIA SYNDROME (AMS) IN A NEW BORN BUFFALO CALF

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ABSTRACT

The present case describes the successful vaginal delivery of a buffalo calf with Aplasia Cutis Congenita co-existence with Ablepharon Macrostomia Syndrome (AMS) in a graded Murrah buffalo.

Keywords: Aplasia cutis congenita, ablepharon macrostomia syndrome, buffaloes.

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INTRODUCTION

Aplasia cutis congenita which is also known as congenital epitheliogenesis imperfecta, a recessive hereditary condition characterized by congenital discontinuity of the squamous epithelium of the skin and oral mucous membranes (Yager and Scott, 1993) where the severity is variable among domestic animals. The size and location of the lesions vary but usually consist of irregular patches of hair and discontinuity of squamous epithelium, more frequently occurring on the distal extremities of the limbs below the knees and hocks and on the muzzle, ears, tongue and oral mucosa due to defect in the single autosomal recessive gene causing complete local failure of tissue growth (Roberts, 1986). Affected calves may lack the epithelium in the oral mucosa and tongue and the defect is usually incompatible with life.

Ablepharon Macrostomia Syndrome (AMS) is a congenital condition seen in human which is mainly characterised by aberrant genitalia, redundant skin, non-existent or sparse eyelashes and eyebrows, ear anomalies and macrostomia. Documentation of this condition in veterinary practice is very rare; hence the present condition is compared

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with the findings of Mazzullo *et al.* (2012) in a crossbred calf.

CASE HISTORY AND CLINICAL OBSERVATIONS

A pluriparous graded Murrah buffalo was presented to the Veterinary Clinical Complex, during off hours, which was at full term gestation with a history of prolonged second-stage labour showing unproductive straining since 24 hrs with scanty whitish vaginal discharges. On clinical examination, all the vitals were within the normal range. Detailed obstetrical examination revealed that the cervix was fully dilated with intact foetal membranes.

OBSTETRICAL INTERVENTION AND TREATMENT

On administering a low plane of epidural anaesthesia with 4 ml of 2% Lignocaine and sufficient lubrication, the intact foetal membranes were ruptured manually to deliver the calf safely. After the rupture of foetal membranes, blood-tinged foetal fluids were expelled. The calf was in anterio-longitudinal presentation, dorso-sacral position and both the fore limbs and head were extended towards the birth canal. Obstetrical snares were applied above the fetlock joints and the foetus was delivered by gentle manual traction. The foetal membranes were expelled simultaneously with the live foetus. Clinical examination of the new born calf revealed, absence of skin on major portions of the body except for a few patches of skin present on the head up to shoulder region and some patches over the rump region (Figure 1).

The head of the calf was observed with some of the peculiar lesions similar to a rare condition recorded in humans called as Ablepharon macrostomia syndrome (Figure 2), which includes, brachygnathism, absence of eyelids (ablepharon), posteriorly fixed auricular cartilage and ear pinnae that are not formed fully. Initially, the calf was treated with 1 ml of Dexamethasone and Vitamin-A injections intramuscularly, but ultimately it succumbed to death within a few hours.

DISCUSSION

Epitheliogenesis Imperfecta was first reported in cattle in the year 1928. Etiological factors included, inbreeding (Leito *et al.,* 2002) caused by recessive hereditary gene (Yamini *et al.,* 2020). According to Venkataramanan *et al.* (2010) and Azizi *et al.* (2016), epitheliogenesis imperfecta is a rare congenital condition marked by an abrupt segmental lack of the epidermis and skin structures descended from epithelial cells, which is most noticeable on the trunk area. The present findings, which include the lack of epithelium across significant portions of the limbs, muzzle, nostrils, tongue, hard palate, and cheeks, are typical and comparable to those documented by Dalir-Naghadeh *et al.* (2004) and Yamini *et al.* (2020) in bovines, Bentinck-Smith (1951) in swine, Tontis and Hofstetter (1991) in lambs and Kumar *et al.* (2021) in Kanni doe.

Mazzullo *et al.* (2012) reported a case of ablepharon macrostomia syndrome in a calf

having characteristic lesions on the head and resembling with that observed in the present case such as brachygnathism, abnormal ear and nose, ablepharon condition and epithelial deformities of adnexa of skin. Hence, finding ablepharon macrostomia syndrome, along with epitheliogenesis imperfecta in a buffalo calf born alive, was exceptionally rare and was not reported previously in the literature for buffaloes.

Fig. 1. Live calf with aplasia cutis congenita condition

Fig. 2. Ablepharon Macrostomia syndrome showing wide mouth, brachygnathism, ablepharon and abnormal ear pinnae

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