

EPITHELIOGENESIS IMPERFECTA IN A GRADED FRIESIAN CALF

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ABSTRACT

Epitheliogenesis imperfecta, a congenital abnormality involving the skin has been reported in several domestic and wild animal species. In cattle, affected calves die shortly after birth due to septicaemia. A case of this disease in a graded Friesian calf, in which the animal has survived, is reported. Histopathology and type of lesions confirmed the disease. Karyotype revealed normal diploid chromosome complement of 60,XX. The development of epidermis was partial and the animal responded well to treatment with antibiotic, antihistamine and anti-inflammatory therapy. Scarring of affected regions commenced and hair started growing gradually. Skin lesions were accompanied by rolling of ears, and surfaces brought in contact grew together. The ear defect was corrected surgically and a pressure bandage similar to the one used in ear hematoma was applied to prevent reunion of adhered surfaces. Almost complete resolution was noticed in 3 months time and the calf survived.

Key words: *Epitheliogenesis imperfecta*, skin, congenital, Friesian calf, Karyotype.

INTRODUCTION

Epitheliogenesis imperfecta is a congenital abnormality in which discrete areas of the body are devoid of skin at the time of birth. Several domestic and wild animal species have been reported with this condition. In cattle, the affected calves die shortly after birth due to septicaemia (Hadley, 1927; Naghadeh *et al.*, 2004). A case of *epitheliogenesis imperfecta* in a graded Friesian calf, in which the animal has survived, is reported. Karyotyping was also done to detect for any chromosomal abnormalities.

MATERIALS AND METHODS

A graded Friesian calf with skin defects, belonging to a farmer's dairy unit located in the Nilgiris district of Tamil Nadu was brought for treatment. It was observed that the calf was unthrifty

and suffered from wound throughout the body. History revealed that it was the thirteenth calf of the dam and sired by an unknown bull through natural service. The dam had died of milk fever and calf was bottle fed with milk. On examination, the lesions were bright red in colour, glistening and depressed. The distribution of lesions was bilateral, symmetrical and diffuse. Affected areas were the flank and regions surrounding elbow and stifle joints. The lesions around the joints were extensive both medially and laterally (Fig 1). The left ear was deformed due to rolling of margins and growing together of surfaces brought in contact. Itching was noticed, and the calf injured itself in the process.

Treatment included antibiotic, antihistamine and anti-inflammatory therapy. Parenterally, injections sulphadiazine with trimethoprim (Sulprim™, Vetcare), pheneramine maleate (Avil™, Intervet), and Vitamin AD₃E (VetAD3E™,

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Sarabhai Zydus) were given. Topicure™ gel (Herbal preparation, Natural Remedies) and zinc oxide cream were applied topically. The rolled ear was surgically corrected by separating the adhered surfaces. A pressure bandage similar to the one used in cases of ear hematoma was applied to prevent reunion of surfaces.

For karyological studies, blood samples were collected in vacutainer systems containing sodium heparin from. The cultures were set up using RPMI 1640 culture medium and buffy coat and autologous plasma from the blood samples. Mitosis was induced by the incorporation of pokeweed mitogen (10 µg/ml) and the cultures were incubated at 37.5° C for 72 h. The cultures were harvested with colchicine followed by hypotonic treatment (0.075 M KCl) and fixed in methanol: acetic acid (3:1). Air-dried slides were prepared and stained in 2 per cent Giemsa (Halnan, 1977). A total of 50 metaphase spreads were screened for chromosome complement. Non-overlapping chromosome spreads with clear staining were photographed (x 1000) using Leica DM2500 trinocular research microscope with DFC280 camera.

RESULTS AND DISCUSSION

Scarring of the affected region commenced and borders closed in progressively. Signs of itching disappeared and hair started growing in the region. Almost complete resolution was noticed in three months time, except that the affected region became crusted with scar tissue (Fig 2). The calf survived and was inseminated at the age of 15 months and gave birth to a normal female calf.

Histopathological examination revealed partial development of epidermis in the affected areas (Fig 3a). Stratum basale with single layer

of columnar cells were noticed. Thickness of the stratum spinosum was reduced, stratum granulosum was absent and stratum corneum was partially developed. Hair follicle and secretory acini of tubuloacinar serous glands were seen in the dermis. Histological examination of normal skin from the same animal showed intact layers (Fig 3b).

The normal diploid chromosome complement of 60, XX was observed in all the metaphase spreads observed. No numerical chromosome abnormality was observed (Fig 4).

In the present case, the observations of type and distribution of lesions on the calf were indicative of *epitheliogenesis imperfecta*. The calf was a graded Holstein Friesian. Hadley (1927) noticed the condition in purebred or graded Holstein animals. *Epitheliogenesis imperfecta* was diagnosed for the first time in Hereford cattle by Agerholm *et al.* (1993). In Jerseys and Ayrshires, the epithelial defects could be more extreme and affected calves may be aborted (Jubb and Kennedy, 1970).

Histopathological examination confirmed the same and the development of epidermis was partial. Generation of epidermis cannot occur when the basal cells are incomplete and the lesions were patched up in time by scar tissue. The hair follicles were well developed in the dermis and the affected areas were covered gradually with hair. Biancamano *et al.* (2006) made an extensive study on 70 piglets with *epitheliogenesis imperfecta*, and observed most of the lesions with absence of epidermis superficial to mid-dermis and adnexae.

In several studies on *epitheliogenesis imperfecta* in cattle, lesions have been found to be located in skin of digital extremities, muzzle, oral mucosa and tongue (Hadley, 1927; Jubb and Kennedy, 1970; Naghadeh *et al.*, 2004). The calf under study had most of its lesions concentrated

over the flank and region surrounding elbow and stifle joints. However, Hadley (1927) and Jubb and Kennedy (1970) described the lesions to be bilateral and symmetrical and a similar distribution was noticed in this case also.

As observed by Hadley (1927) and Jubb and Kennedy (1970), skin lesions were accompanied by ear abnormalities. The growing together of surfaces after rolling are indicative of the fact that the lesions antedated birth by few weeks. The application of a pressure bandage after correction of the abnormality helped to prevent the reunion of adhered surfaces.

Based on the history of 13 affected Friesian calves Hadley (1927) observed that the defect was due to inheritance of a recessive allele. Other authors also have described the cause to be hereditary, due to a single recessive autosomal factor (Jubb and Kennedy, 1970; Naghadeh *et al.*, 2004; Gruys, 2007). Tontis and Hoffstetter (1991) observed congenital *epitheliogenesis imperfecta* in five lambs born to the same sire, and the abnormality disappeared after removal of the sire. Lieto and Cothran (2003) have studied *epitheliogenesis imperfecta* in American Saddlebred horses and suggested that the disease locus was located in equine chromosome 8q. Biancamano *et al.* (2006) observed a high ratio of affected males to female animals in an extensive study on 70 piglets with *epitheliogenesis imperfecta*.

In the present case the calf was the thirteenth of the dam, and this was the first incidence in all. The chromosomal source of the defect was presumed to be from the sire. However, the sire was a local bull and history of other services by this bull was not available.

In general, *epitheliogenesis imperfecta* has been found to be incompatible with life (Hadley,

1927; Naghadeh *et al.*, 2004). The lesions bleed and become infected readily and septicaemia is usually the cause of death. However, some of the affected animals survive under proper care and treatment (Jubb and Kennedy, 1970; Biancamano *et al.*, 2006). In the present case, the calf had survived, and calved for the first time to produce a normal female calf.

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Figure 1.
Lesion at birth



Figure 2
Lesion after scarring



Figure 3.
Histology: Normal skin (a) and skin from affected area (b)

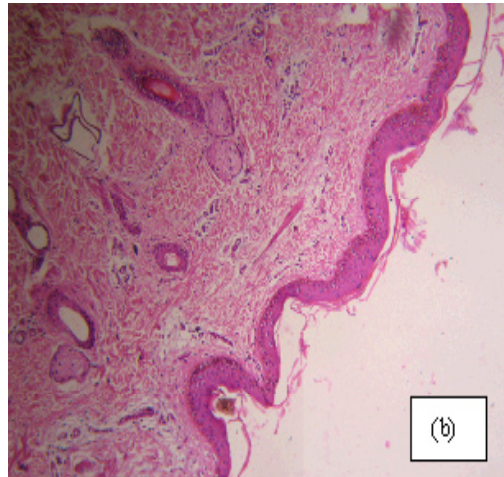
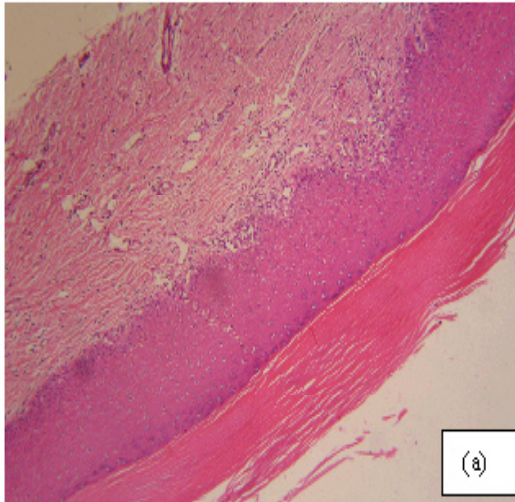
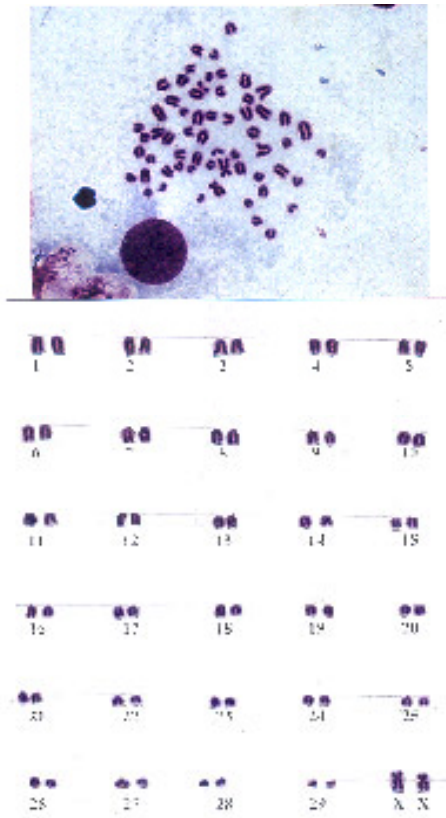


Figure 4

Metaphase spread and karyotype from affected animal



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