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The Occurence of Dermatosparaxis in a Herd of Holstein Dairy

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Objective

Dermatosparaxis (Cutanous asthenia) is a heritable collagen dysplasia causing hyperextensibility and extreme fragility of the skin. Joint laxity, ocular and capillary abnormalities have also been described. Dermatosparaxis has been reported in human, horses, sheep, dogs, cats and some breeds of cattle (Belgian Blue, Holstein, Charolais, Hereford, and crossbred ...). In most cattle, cutaneous asthenia is inherited as a lethal autosomal recessive trait. Both of Human Ehlers-Danlos syndrome (EDS) type VIIC and bovine dermatosparaxis are caused by mutations in the procollagen I N-Proteinase (pNPI) gene (or ADAMTS2 gene). The condition results from the absence of activity of an enzyme, resulting in accumulation of pre-cursor molecules (pro-collagen) that self-assemble into ribbon-like fibrils that fail to provide normal tensile strength to tissues. Various forms of EDS have been identified in affected animals. Mutations in other genes (COL5A1, PLOD1, EPYC, TNBX ...) are likely responsible for other types of dermatosparaxis. Within and between species there are clinical forms (expression of the phenotype), biochemical and ultrastructural variations.

In Holstein cattle onset is soon after birth, and the condition to be lethal. Typical clinical signs include subcutaneous oedema of the neck, thorax, limb and dewlap. Severe, extensive ulceration secondry to minor trauma. Large skin areas became necrotic and sloughed, that are prone to infection. The lesions responded poorly to treatment, heal slowly with thin, papyraceous scars, and new lesions developed time to time. Abnormal lowing and poor vision has been reported.

Case presentation

In a small dairy near Tehran, three heifers with the same age (one pregnant, 4 month) and eyes with dark blue appearance, were examined due to reduced vision. The heifers was not blind, but the vision moderately decreased. The cornea was uniformly dark blue so that pupil and iris were indistinguishable. The eyes had no other problem. Inerestingly, the heifers' skin had other abnormal symptoms, presence of thin and loose skin in excessive amount with folds all over the neck and in the lower parts was hanging. The skin can easily be pulled up to 20-30 cm with a hand and skin folds remained intact for a few minutes. Two non-pregnant heifers were removed and five months later, the male calf was born and soon after showed symtoms similar to those of her mother but more severe, which from three month onwards intensified rapidly with age. All three heifers were the offspring of a domestic sperm from an imported bull and male calf was the progeny of a sperm imported.

Clinical signs in calf included, eyes with dark blue corneas, very thin skin with severe elastisity and numerous folds on the neck, multiple skin ulcers of various sizes on the tighs, perineum, scrotal sac and abdomen with peeled skin, often without bleeding, and or a thin scab, seperation of the skin from the subcautaneous and accumulation of edema fluid under the skin of the neck and dewlap that during three month hangs in a cavity with a volume of several (>5) litres. The fluid of skin cavity was sampled three times and the result of bacterial culture was negative in all cases. The calf was autopsied at eight month of age after the skin cavity was torn suddenly and a large amount of pale serosanguinous, clear and odourless fluid was drained and then began to bleed. During the eight months the calf had no other disease.

At autopsy the most prominent sign was very thin and fragile skin with a diameter of 2-3 mm, which could be easily torn like a thick cloth with hand. Extensive detachment of the skin from the subcutaneous tissue, subcutaneous bleeding, and the presence of granular tissue were also signs of autopsy. Other body organs seemed normal.

Microscopic examination on cutaneouus tissue shows mild hyperkeratotic epidermis with marked reduction in the density and number of mature collagen fibers in superficial dermis. Remaning fibers were short, fine, pale, fibrillated and fragmented. Irregular fibrils arranged in loosely woven flat with lost transverse striation.collagen fibers of the deep dermis appeared relatively spared. Ulcerative neutrophilic dermatitis with granulation tissue formation in some areas were present.

Conclusions

Unfortunately, genetic disorder such as dermatosparaxis are usually not reported or diagnosed, animals suspected of having a genetic disease are usually removed from the herd quickly, because of a lack of resources and diagnostic tools, and/or low value of the animals.

Based on the clinical presentation, autopsy and light microscopic findings a diagnosis of dermatosparaxis was made. The recessive nature of inheritance in this disease, may suggest that the genetic anomaly is present in the papultionin in a heterozygus state and thus can be preserved. Therefore, the clinical form of the disease first occurred after inbreedings or use a large number of one type of sperm with a defective gene for a long time. This case highlights that the potential risk of the widespread and rapid dissemination of genetic disease associated with AI and espesially Emberyo transfer.

Due to the lethal nature of the phenotype observed in the offspring of carriers of deffective genes, it seems that further genetic investigation is necessary for the import of sperm or emberyos. Affected animals and their relatives should not be used for breeding. Recently, the method of whole-genome sequencing (WGS) has been used to make precision diagnoses for understanding rare genetic disorders in animals, with an available reference genome sequence for surveillance of cattle breeding papulations for harmful genetic disorders.

To the authors' knowledge this is the first report of dermatosparaxis in the Holstein cattle breed in Iran.

Keywords

Cattle, Collagen, Dermatsparaxis, Holstein, Heritable, Iran.

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