CASE REPORT

INHERITED CONGENITAL PORPHYRIA IN CALVES

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Introduction

Inherited congenital porphyria is a rare disease among domestic animals. It is an inherited deficiency of an enzyme which is required for the normal synthesis of hemoglobin which results in an inadequate production of hemoglobin and an overproduction of porphyrins. The porphyrins are photodynamic which accounts for their fluorescent and photosensitization activities (2,3,4).

Due to the nature of the porphyrin molecule, energy in the form of light of short wavelengths, induces resonance in the porphyrin molecule. The energy emitted is of longer wavelength than visible light plus heat. The fluorescent property of porphyrins is thus accounted for by their excitation with ultraviolet light which has short wavelengths. The heat which is released in the skin is sufficient to cause cellular injury which is the sign of photosensitization (4).

In the bovine species, the disease is inherited as a single Mendelian recessive character; however porphyria does not occur with uniform severity (2,4). The bones, teeth and urine of affected cattle have a reddish-brown discoloration and these all fluoresce red under ultraviolet light. The reddish-brown discoloration of the teeth and bones has resulted in the respective terms "pink tooth" and "osteihaemochromatosis" being applied to animals affected with the disease (3,4,5). Photosensitivity of unpigmented areas of the skin is also another sign which occurs in affected animals exposed to sunlight. Because of inadequate hemoglobin synthesis as well as a decreased erythrocytic life span, affected animals are anemic in various degrees of severity (2,3,4).

History – Case I

A purebred female Ayrshire calf, born November 1973 was presented to the Vegreville Veterinary Clinic on June 24, 1974. The dam was a registered purebred Ayrshire which had been mated by artificial insemination to a registered purebred Ayrshire bull.

The calf had been stabled indoors since birth but had been turned outside for the two weeks previous to presentation to the veterinary clinic. The calf weighed about 250 pounds and was predominantly white in color. It was in poor bodily condition and it appeared small in body size for its age. The respiratory rate and the heart rate were approximately twice that of the normal and the calf appeared depressed. The ambient temperature on that particular day was approximately 80°F and the owner reported that the calf had been lying in the sun prior to the trip to the veterinary clinic.

When the calf was presented it had an abrased area on the back extending from the tips of the scapulae to the tuber ileum and extending distally from the spines of the vertebrae approximately two and one half inches. The calf was very uncomfortable in that it licked its back, alternating sides, so that it caused bleeding of the area. The muzzle was also abraded as far as the tongue would reach. The urine was amber colored and the owner reported that this had appeared about one week after the calf was turned outside. It had also started licking its back and muzzle at the same time.

History – Case II

Another female Ayrshire calf was presented to the Vegreville Veterinary Clinic on September 27, 1974. The calf had been born August 17, 1974 to a grade Ayrshire cow which had been mated to a registered Ayrshire sire by artificial insemination. The calf's general external appearance was normal, however it was passing amber colored urine. The owner reported that he had noticed this occurrence only several days previously. On examination of the calf's teeth, they were found to be reddish-brown in color and fluoresced pink under the ultraviolet lamp. This calf had access to a barn but was otherwise housed outside most of the day.

Laboratory Findings

From the calf described in Case I, an incisor tooth was extracted and submitted along with a urine specimen to the Provincial Veterinary Pathology Laboratory at Edmonton, Alberta. Sometime after the urine specimen had been collected, it was noted that the amber color

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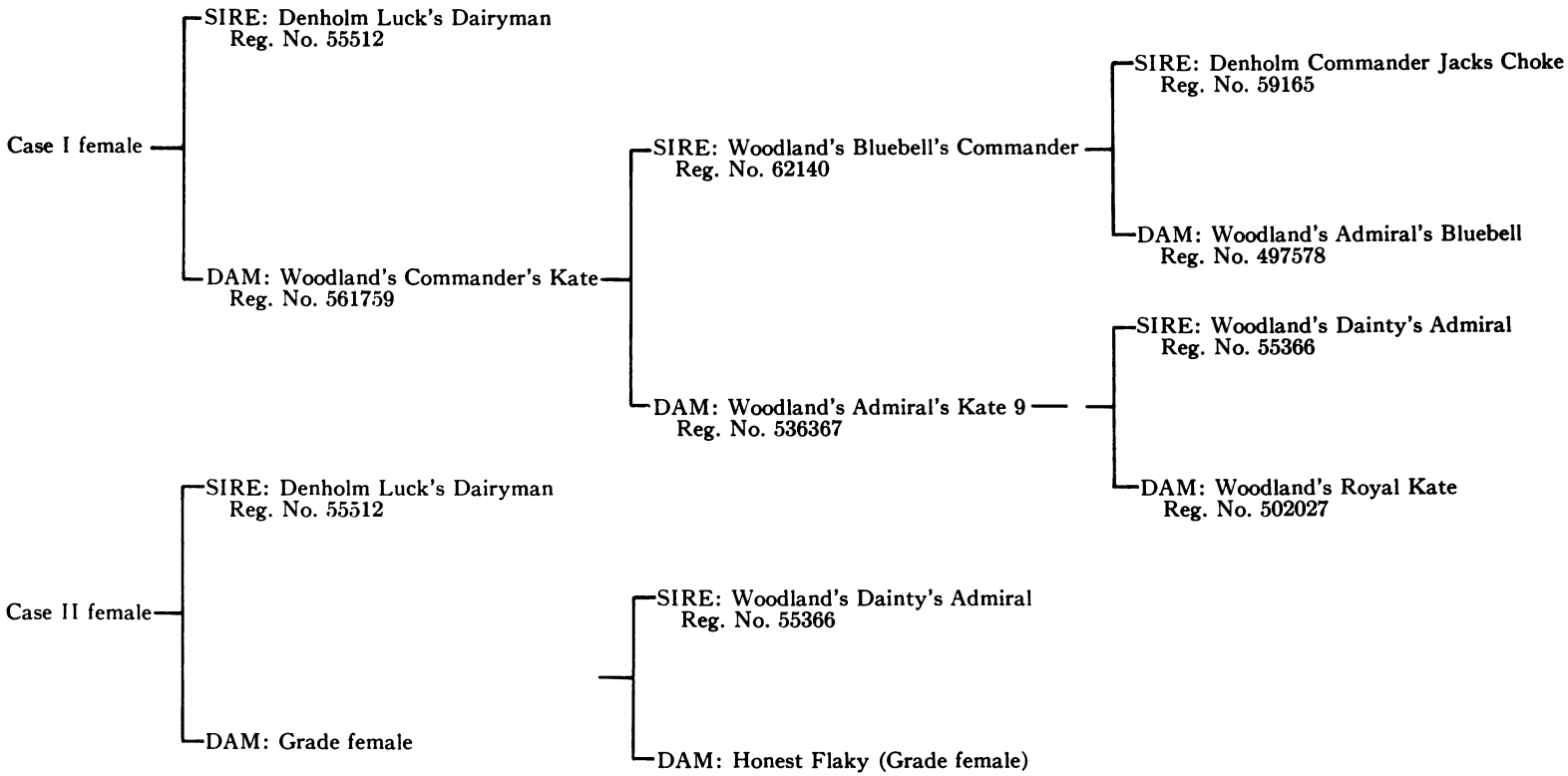

FIGURE 1. Pedigree of calves affected with inherited congenital porphyria.

had darkened considerably. The urine specimen of Case No. I contained 1196 ml coproporphyrine and 560 μg/ml uroporphyrins. The urine of normal cattle contains 1.84 μg/100 ml of coproporphyrins and no significant quantity of uroporphyrins. The urine of cattle affected with inherited congenital porphyria has contained 500–1600 μg/100 ml of uroporphyrins and 356–1530 μg/100 ml of coproporphyrins (1).

Inherited congenital porphyria must be differentiated from photosensitization due to other causes and also from symptomatic porphyrinuria caused by liver insufficiency. Discovery of the discolored teeth at birth would have been a positive diagnosis, however neither calf was examined at that time. In order to detect heterozygous normal carriers, breeding trials are necessary but the herd involved was too small and the owner planned to retain all females as herd replacements.

Both calves were sired by the same sire and the dams originated from the same herd in southern Alberta (Figure 1). The grade Ayrshire dam of Case No. II had given birth to a stillborn calf the previous year. It had been the result of an identical mating with the same sire. The owner had not felt the event was significant at the time and a necropsy had not been performed.

When the owner decided to keep the first calf he was advised to keep it indoors to prevent photosensitization, but not to use the animal as a female replacement in his herd. When examined three months later, it had grown but was still undersize for its age and the hair had grown back into the previously denuded areas affected by photosensitization. Probably because it had been housed indoors, the urine was not as dark as on the initial examination but the teeth, both deciduous and permanent, were the same reddish-brown color.

The Canadian Ayrshire Breeders Association was made aware of the occurrence of the two female Ayrshire calves born with inherited congenital porphyria. Since porphyria in the bovine species is thought to be due to a recessive factor, the heterozygotes are clinically normal as is the case with the dams of the two affected calves. There is no strict sex linkage known in the mode of inheritance but apparently the incidence is higher in females than in males (2). In the herd involved the numbers were too small and the same sire had been used to mate only the two dams.

Summary

Inherited congenital porphyria was diagnosed in two female Ayrshire calves sired by the same sire and born to dams which had originated from an Ayrshire herd in southern Alberta. The diagnosis was based on the reddish-brown discoloration of the teeth, high level of porphyrinuria, photosensitization and reduced growth rate.

Résumé

L'auteur a diagnostiqué de la porphyrie héréditaire chez deux génisses Ayrshire engendrées par le même taureau et nées de vaches provenant d'un troupeau Ayrshire du sud de l'Alberta. Son diagnostic reposait sur la décoloration brun-rougeâtre des dents, le taux élevé de porphyrinurie, la photosensibilisation et le retard de croissance.

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