

From this trial, the use of different doses intravaginal progestagen in synchronization in different seasons of the year seems unnecessary.

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## Incidence of UMP synthase deficiency in South African Holstein cattle

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Deficiency of uridine monophosphate synthase (DUMPS) is an inherited, recessive metabolic defect identified in Holstein cattle. Since heterozygous carriers transmit the defective gene 50% of the time, one fourth of the offspring from matings between two carriers are expected to be homozygous for DUMPS. This is a lethal condition where embryos die early in gestation. Offspring produced from the imported semen of an American carrier bull showed a carrier frequency of 54%. Among 277 South African AI bulls active during 1991 and 1992, no incidence was observed.

Defek vir uridienmonofosfaatsintase (DUMPS) is 'n oorerflike, resessiewe metaboliese afwyking wat in Holsteinbeeste geïdentifiseer is. Omdat heterosigotiese draers die defekte geen in 50% van alle parings oordra, kan verwag word dat een kwart van die nageslag van parings tussen twee draers homosigoties vir DUMPS sal wees. Dit is 'n letale toestand waar die embryo vroeg afsterf. Nageslag afkomstig van ingevoerde semen van 'n Amerikaanse draerbul het 'n draerfrekwensie van 54% getoon. Van die 277 Suid-Afrikaanse KI-bulle wat gedurende 1991 en 1992 gebruik en vir die voorkoms van DUMPS getoets is, is geen draers geïdentifiseer nie.

**Keywords:** Holstein cattle, UMP synthase deficiency.

A deficiency of UMP (uridine-5'-monophosphate) synthase in cattle has been rediscovered in the late 1970s. It is suspected that DUMPS (deficiency of uridine monophosphate synthase), an inherited condition, has been present for 40 years and that the first known carrier, which was used extensively for artificial insemination (AI) purposes, was born in 1957 in America. Cattle have numerous different enzymes that are involved with metabolism, growth, movement, reproduction, lactation and other biological processes. Defects in any of these enzymes may cause moderate to severe physiological problems. In cattle, seven inherited enzyme deficiencies are known, with DUMPS being one of the most important (Shanks & Robinson, 1987).

UMP synthase, a bifunctional enzyme, is present in all body cells and is transmitted as an autosomal recessive trait (Robinson *et al.*, 1984), like pink tooth (Nicholas, 1987) and other enzyme disorders known in cattle. Therefore, matings between a normal individual and a carrier would be expected to produce one half normal and one half carrier offspring, regardless of sex. In matings between two carriers, one fourth of the offspring are expected to be normal, one half are expected to be carriers, and one fourth are expected to be

homozygous recessive, which implies that 25% of the offspring would lack the enzyme complex. This is a lethal condition because UMP synthase catalyses the last steps in the *de novo* synthesis of UMP, the precursor of all other pyrimidine nucleotides essential for DNA and RNA synthesis (Shanks *et al.*, 1984). In the homozygous recessive state, the embryos die early in gestation and appear to be lost or resorbed during the first two months of gestation. This defect may thus be responsible for many unexplained cases of embryo resorption. The defect also leads to more services per calving and longer than normal calving intervals for carrier-carrier matings (Shanks & Robinson, 1987).

Therefore, since UMP synthase is an enzyme that may directly affect calf survival and indirectly reproductive fitness (Robinson *et al.*, 1983), a breeding programme should be implemented to completely eliminate the deficiency. The purpose of this study was to investigate the incidence of DUMPS in Holstein cattle in South Africa and to identify possible carriers of the gene in bulls used for AI to limit the spread of the defective gene through the country.

UMP synthase activity of haemolysed erythrocytes was determined by the method of Jones *et al.* (1986). The assay consists of the conversion of orotate to UMP, carbon dioxide and inorganic phosphate (PP<sub>i</sub>) by a two-stage mechanism catalysed by phosphoribosyl pyrophosphate. An animal is designated a carrier for DUMPS if its activity for UMP synthase is less than 67% of the mean for normal animals of the same age and sex (J.L. Robinson, 1988, personal communication).

This genetic defect has been identified in two American bulls from which semen was imported for AI. Samples (13) received during one year for analysis from the offspring produced from one of these bulls, Happy Herd Beautician, showed a frequency of 54% for the DUMPS allele. The gene

did not occur in 277 South African Holstein bulls used by an AI company from 1991 to 1992 and which were tested in this investigation. This is very promising if compared to America where the carrier frequency was 1.4% among 297 Holstein bulls used by AI companies in 1985. It is estimated that in the United States 1–2% of all Holstein cattle are carriers for this condition (Shanks & Robinson, 1987). In a study of 880 Holstein cows in the state of Illinois in 1982, the minimal incidence was 1.7% (Robinson *et al.*, 1984).

Work is at present in progress to further study the incidence of DUMPS as a result of using imported semen and analyses will continue on all AI bulls in an effort to eradicate DUMPS in South Africa.

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