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Deficiency of uridine monophosphate synthase (DUMPS) and X-chromosome deletion in fetal mummification in cattle

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Abstract

Ten mummified fetuses were tested for the deficiency of uridine monophosphate synthase (DUMPS), which is known to contribute to the embryonic and fetal mortality in cattle. Genomic DNAs of the mummified fetuses were extracted from tissue samples collected from the mummies and were amplified by GenomiPhi™ DNA amplification kit. UMPS gene of the mummies was amplified by polymerase chain reaction (PCR) with DUMPS primers. Out of ten mummies examined, two fetuses were heterozygous (carriers) for DUMPS as indicated by the presences of three bands of 89, 53 and 36 bp. Estimated stage of gestation when the death occurred in the two mummies was 3.5 and 2.5 months, respectively. The other fetuses exhibited only two bands of 53 and 36 bp on the polyacrylamide gel indicated that they were normal. On the other hand, all the mummies were sexed using AMX/Y primers. Specific regions of Y and X chromosomes were amplified by PCR using AMX/Y. The expected 280 bp fragment in the female sample and the 280 and 217 bp in the male sample were observed. Nine mummies had a normal X and Y chromosome bands; however, the other mummified fetus exhibited only Y chromosome band, while the constitutive X chromosome fragment was missing. The estimated stage of gestation when the death occurred in this mummified fetus was

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100 days. This might be the first report of DUMPS and X-chromosome deletion at the amelogenin gene in bovine-mummified fetuses in Japan.

Keywords: DUMPS; X-chromosome; Fetal mummification; Cattle

1. Introduction

Although the causes of fetal mummification in cattle are often difficult to identify, some genetic factors due to autosomal recessive gene have been reported to be involved in bovine fetal mummification (Roberts, 1962; Stevens and King, 1968).

Recessive lethal genes contribute to embryonic and fetal mortality. About 2% of the Holstein in USA posses an autosomal recessive form of the gene for deficiency of uridine monophosphate synthase (DUMPS) which, if present in the homozygous condition, may lead to embryonic mortality before day 40 of gestation (Shanks et al., 1984). Uridine monophosphate synthase (UMPS) catalyzes the last two steps of de novo pyrimidine synthesis converting orotic acid to uridine 5′-monophosphate (Jones, 1980). Schoeber et al. (1992) isolated and sequenced the wild type bovine UMPS cDNA. One year later, Schwenger et al. (1993), identified the mutation responsible for DUMPS. Subsequent sequencing of the polymerase chain reaction (PCR) products revealed a mutation (C to T) with the loss of an *Ava*I site at codon 405, resulting in a premature stop codon with a truncated C-terminal catalytic subunit of the protein. A direct DNA test based on PCR was developed for the diagnosis of the problem, in which the PCR product is digested with the enzyme *Ava*I and the mutation can be identified (Schwenger et al., 1993).

A variety of methods have been used to sex bovine embryos, including immunological, cytogenetic and DNA-based techniques (van Vliet et al., 1989). Cytogenetic karyotyping is the classical and most reliable method for fetal sexing but has the disadvantage of being time consuming. Therefore, molecular genetic sexing by PCR is usually performed in preference to prenatal molecular tests for X-linked conditions (Jakubiczka et al., 2000). A number of DNA-based tests have been developed, most of which detect sequences present only on the Y chromosome; amplification of male samples gives a PCR product, whereas female samples give no product (Ennis and Gallagher, 1994). Both minisatellite (Schroder et al., 1990) and single-copy sequences (Aasen and Medrano, 1990) have been used for sex determination in cattle. Such assays need an internal control, usually amplification of a second autosomal locus, to verify that the absence of a signal is not the result of PCR failure. In cattle there are two different amelogenin transcripts (class I and class II), which are the products of genes located on the bovine X- and Y-chromosomes, respectively (Gibson et al., 1991, 1992). There is a 63-bp deletion in the fifth exon of the class II (Y chromosome) amelogenin gene (Gibson et al., 1991). This difference between the X and Y chromosomes at the amelogenin gene has been used for sex determination in cattle (Ennis and Gallagher, 1994). Jakubiczka et al. (2000) reported on the incidental prenatal detection of an interstitial X-chromosomal deletion in a human male fetus and his mother by fetal sexing with a primer pair recognizing an X–Y homologous locus (DXYS19).