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ABSTRACT

IARS mutation causes prenatal death in Japanese Black cattle

Takashi Hirano, Tamako Matsuhashi, Kenji Takeda, Hiromi Hara, Naohiko Kobayashi, Kazuo Kita, Yoshikazu Sugimoto, Kei Hanzawa

1Faculty of Agriculture, Tokyo University of Agriculture, Atsugi, Kanagawa, Japan.
2Gifu Prefectural Livestock Research Institute, Takayama, Gifu, Japan.
3Hida Region Livestock Hygiene Service Center, Takayama, Gifu, Japan.
4Shirakawa Institute of Animal Genetics, Nishigo, Fukushima, Japan.

Isoleucyl-tRNA synthetase (IARS) c.235G > C (p.V79L) is a causative mutation for a recessive disease called IARS disorder in Japanese black cattle. The disease is involved in weak calf syndrome and is characterized by low birth weight, weakness and poor suckling. The gestation period is often slightly extended, implying that intrauterine growth is retarded. In a previous analysis of 2597 artificial insemination (AI) procedures, we suggested that the IARS mutation might contribute toward an increase in the incidence of prenatal death. In this study, we extended this analysis to better clarify the association between the IARS mutation and prenatal death. The IARS genotypes of 92 animals resulting from crosses between carrier (G/C) × G/C were 27 normal (G/G), 55 G/C and 10 affected animals (C/C) (expected numbers: 23, 46 and 23, respectively). Compared to the expected numbers, there were significantly fewer affected animals in this population ($P < 0.05$), suggesting that more than half of the affected embryos died prenatally. When the number of AI procedures examined was increased to 11 580, the frequency of re-insemination after G/C × G/C insemination was significantly higher at 61-140 days ($P < 0.001$). The findings suggested that the homozygous IARS mutation not only causes calf death, but also embryonic or fetal death.