

## 鹿児島大学と共同研究を実施した学術論文が 日本獣医学雑誌に掲載決定

鹿児島大学と共同研究を実施した「猫のピルビン酸キナーゼ欠損症」の学術論文が日本獣医学雑誌 (The Journal of Veterinary Medical Science, JVMS) に受理され、2015年,77巻(6月号)に掲載が決定致しましたので、お知らせ致します。

[なお、論文の内容の要旨(日本語)は、Bio Plus ニュース(14),September 12,2014を参考にして下さい。]



NOTE *Internal Medicine*

### Real-time PCR genotyping assay for feline erythrocyte pyruvate kinase deficiency and mutant allele frequency in purebred cats in Japan

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**ABSTRACT.** Erythrocyte pyruvate kinase (PK) deficiency is an inherited glycolytic erythroenzymopathy caused by mutations of the *PKLR* gene. A causative mutation of the feline *PKLR* gene was originally identified in Abyssinian and Somali cats in the U.S.A. In the present study, a TaqMan probe-based real-time PCR genotyping assay was developed and evaluated for rapid genotyping and large-scale screening for this mutation. Furthermore, a genotyping survey was carried out in a population of four popular purebred cats in Japan to determine the current mutant allele frequency. The assay clearly displayed all genotypes of feline PK deficiency, indicating its suitability for large-scale survey as well as diagnosis. The survey demonstrated that the mutant allele frequency in Abyssinian and Somali cats was high enough to warrant measures to control and prevent the disease. The mutant allele frequency was relatively low in Bengal and American shorthair cats; however, the testing should still be carried out to prevent the spread of the disease. In addition, PK deficiency should always be considered in the differential diagnosis of anemia in purebred cats in Japan as well as worldwide.

**KEY WORDS:** anemia, feline *PKLR* gene, mutant allele frequency, pyruvate kinase deficiency, real-time PCR genotyping

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