

Molecular Diagnosis of the COMMD1 Gene in Korean Bedlington Terriers

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Purpose: The deletion of exon2 of the copper metabolism domain containing? 1 (COMMD1) gene(formerly MURR1) causes copper toxicosis in Bedlington terriers. Bedlington terriers with this autosomal recessive disorder were shown to have the elevated liver copper levels and suffer from hepatitis, progressive cirrhosis of the liver and even death in severe cases. In order to survey the frequency of COMMD1 mutation in Korea, we have screened 31 Bedlington terriers for the COMMD1 mutation.

Materials and Methods: Blood samples were collected from 31 Bedlington terriers of pet dog clubs in Korea. We extracted the genomic DNA from blood using the genomic DNA extraction kit(G-DEX II b, iNtRON Bio, Korea). Mutation status of the blood donors have been screened by multiplex-PCR method. Three pairs of primer sets were designed to amplify DNA fragments from inside the deleted region and across the deletion break points.

Results: Of the 31 samples, 15 were wild type homozygous for the normal COMMD1 gene, 13 were heterozygous, having both normal and mutated copy of the COMMD1 gene. And 3 were mutant type homozygous.

Conclusion: The screening multiplex-PCR for COMMD1 mutation could help establish a structured selective breeding program to prevent COMMD1 mutation in Bedlington terriers in Korea.

Key words: COMMD1, copper toxicosis, PCR, Bedlington terrier

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