GENOMIA
genetic laboratory

**Result certificate #029914:**

**Detection of c.660C>T mutation in ADAMTSL2 gene causing Musladin-Lueke syndrom in Beagle breed**

**Sample**
Sample: 13-03509
Name: Harley z Kunčiček
Breed: Beagle
Microchip: 900008800238492
Date of birth: 24.6.2012
Sex: male
Date received: 12.02.2013
Sample type: buccal swab
Sample certified by Vet/Tech or witness.

**Customer**
Taťána Letáčková
Albrechtičky 170
74255 Albrechtičky
Czech Republic

Result: Mutation was not detected (N/N)

**Explanation**

Presence or absence of c.660C>T mutation in exon 7 of ADAMTSL2 gene causing Musladin-Lueke syndrom (MLS) in Beagle breed was tested. ADAMTSL2 gene is coding the fibrillin-1 protein which forms the main part of the tissue microfibriles. MLS is an inherited disease that affects development and structure of connective tissues and manifests itself by extensive skin and joint fibrosis and affects even heart.

Mutation that causes MLS in Beagle breed is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P (positive/positive) genotype only. The dogs with N/P (negative/positive) genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP80
Sensitivity (probability of correct identification of the defective form of the gene in heterozygous or mutated homozygous) is higher than 99%. Specificity (probability of correct identification of the normal form of the gene in a normal homozygous or heterozygous) is higher than 99%.

Report date: 18.02.2013
Responsible person: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

Dajbychová

Genomia s.r.o, Janáčkova 51, 32300 Plzeň, Czech Republic, VAT#: CZ25212991
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

 