

Detection of c.3149\_3150insC mutation in the C17H2orf71 gene causing RCD4 in several dog breeds

**Sample**

Sample: 08-12345  
Name: Lassie DEMO  
Breed: ---  
Tattoo number: 1392013  
Microchip: 123 456 789 012 345  
Reg. number: REGQ12345  
Date of birth: 31.12.1909  
Sex: female  
Date received: 25.11.2008  
Sample type: blood  
The identity of the animal has been checked.

**Customer**

Jan Novák  
Dlouhá 1  
30000 Plzeň  
Czech Republic

**Result: Mutation was not detected (N/N)**

**Legend:** N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

**Explanation**

Presence or absence of c.3149\_3150insC mutation in the C17H2orf71 gene causing rode-cone dysplasia (RCD4) was tested. The mutation is related with RCD4 in breeds: Australian Cattle Dog, English Setter, Gordon Setter, Irish Red & White Setter, Irish Setter, Polish Lowland Sheepdog, Small Munsterlander, Standard Poodle and Tibetan Terrier. RCD4 is a form of progressive retinal atrophy (PRA) characterized by the degeneration of the photoreceptors in the retina. It results in vision loss and eventually complete blindness. The average age of RCD4 diagnosis is 10 years.

Mutation that causes RCD4 is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. The dogs with N/P genotype are considered carriers of the disease (heterozygotes), they are healthy but they can transmit the mutation on their offspring. In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N, 50 % N/P and 25 % P/P.

In individuals with N/N and N/P result might develop any different form of PRA, due to mutations that were not detected by this test.

Method: SOP171-RCD4, fragment analysis

Report date: 25.11.2008

Responsible person: Mgr. Martina Šafrová, Laboratory Manager



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