Paw Print Genetics
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# Laboratory Report

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|---|---|---|---|
| **Laboratory #:** | 190346 | **Call Name:** | Rose |
| **Order #:** | 85226 | **Registered Name:** | The red rose |
| **Ordered By:** | Terri Johnson | **Breed:** | Australian Shepherd |
| **Ordered:** | Aug. 7, 2020 | **Sex:** | Female |
| **Received:** | Aug. 24, 2020 | **DOB:** | May 2020 |
| **Reported:** | Sept. 6, 2020 | **Registration #:** | - |

## Results:

| Disease | Gene | Genotype | Interpretation |
|---|---|---|---|
| Chondrodysplasia (CDPA) | *CFA18 FGF4* | cd/cd | No Leg Shortening Associated with CDPA |
| Chondrodystrophy with Intervertebral Disc Disease Risk Factor (CDDY with IVDD) | *CFA12 FGF4* | WT/WT | Normal (Clear) - No CDDY or Increased IVDD Risk |
| Collie Eye Anomaly | *NHEJ1* | WT/WT | Normal (clear) |
| Cone Degeneration | *CNGB3* | WT/WT | Normal (clear) |
| Craniomandibular Osteopathy | *SLC37A2* | WT/WT | Normal (clear) |
| Degenerative Myelopathy | *SOD1* | WT/WT | Normal (clear) |
| Hereditary Cataracts (Australian Shepherd Type) | *HSF4* | WT/WT | Normal (clear) |
| Hyperuricosuria | *SLC2A9* | WT/WT | Normal (clear) |
| Intestinal Cobalamin Malabsorption (Australian Shepherd Type) | *AMN* | WT/WT | Normal (clear) |
| Intestinal Cobalamin Malabsorption (Border Collie Type) | *CUBN* | WT/WT | Normal (clear) |
| Multidrug Resistance 1 | *ABCB1* | WT/WT | Normal (clear) |
| Multifocal Retinopathy 1 | *BEST1* | WT/WT | Normal (clear) |
| Neuronal Ceroid Lipofuscinosis 6 | *CLN6* | WT/WT | Normal (clear) |
| Neuronal Ceroid Lipofuscinosis 8 (Australian Shepherd Type) | *CLN8* | WT/WT | Normal (clear) |
| Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration | *PRCD* | WT/WT | Normal (clear) |

WT, wild type (normal); M, mutant; Y, Y chromosome (male)

## Interpretation:

Molecular genetic analysis was performed for 15 specific mutations reported to be associated with diseases and traits in dogs. We identified two normal copies of the DNA sequences in the mutations tested.

We identified two normal copies of the DNA sequences for *CFA18 FGF4* associated with CDPA. Thus, this dog does not have shortened legs associated with CDPA. In addition, we identified two normal copies of the DNA sequences for *CFA12 FGF4* associated with CDDY and IVDD risk. Thus, this dog is not affected with CDDY and is not at an increased risk of developing IVDD.

## Recommendations:

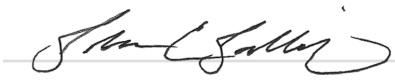
No mutations were identified. Thus, this dog is not at an increased risk for the diseases caused by or associated with the mutations tested. Because this dog is "clear" of these mutations, this dog will only pass the normal genes on to its offspring. Normal results do not exclude inherited mutations not tested in these or other genes that may cause medical problems or may be passed on to offspring.

Two genetic mutations are associated with shortened legs in dogs. Both mutations consist of copied sections (duplication) of the canine *FGF4* gene (called an *FGF4*-retrogene) that have been inserted into two aberrant locations in the genome; one in chromosome 12 (*CFA12 FGF4*; associated with CDDY and IVDD risk) and one in chromosome 18 (*CFA18 FGF4*; associated with chondrodysplasia [CDPA], but not associated with IVDD). Appropriate breeding decisions regarding dogs which have inherited the *CFA12 FGF4* mutation (WT/M or M/M) need to address both the potential loss of genetic diversity in a population which would occur if dogs with this mutation were prohibited from breeding as well as the loss of the short-legged appearance that is a defining physical characteristic for some breeds. In breeds which inherit both mutations, breeders may use genetic testing results to selectively breed for the CDPA (*CFA18 FGF4*) mutation while breeding away from the CDDY and IVDD risk (*CFA12 FGF4*) mutation to reduce IVDD risk and retain the short-legged appearance. However, the frequency of each mutation varies between breeds and, in some cases, may not be conducive to such a breeding strategy. For example, breeds with extreme limb shortening (e.g. Basset hound, Dachshund, Corgi) typically develop their appearance due to inheritance of both the *CFA12 FGF4* and *CFA18 FGF4* mutations. In addition, depending on the breed, offspring born without either the *CFA12 FGF4* or *CFA18 FGF4* mutations may display longer limbs than cohorts and, therefore, not meet specific breed standards.

This dog carries two copies of the cd allele which does not result in leg shortening. However, the actual leg length of the dog is a result of a combination of factors including the mutation associated with CDDY and IVDD risk (*CFA12 FGF4*) as well as variants in other genes. This dog will pass one copy of cd to 100% of its offspring.

No mutation in *CFA12 FGF4* was identified. Thus, this dog is not at an increased risk for Chondrodystrophy (CDDY) and early-onset Intervertebral Disc Disease (IVDD). Because this dog is "clear" of this mutation, this dog will only pass the normal gene on to its offspring. Normal results do not exclude inherited mutations not tested in this gene or other genes that may cause medical problems or may be passed on to offspring.

Paw Print Genetics® has genetic counseling available to you at no additional charge to answer any questions about these test results, their implications and potential outcomes in breeding this dog.



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