

Certificate of Breed

OWNER'S NAME: Brookside Pomsky

DOG'S NAME: Jaguar

TEST DATE: June 23rd, 2018

POMSKY

This certifies the authenticity of Jaguar's canine genetic background as determined following careful analysis of more than 200,000 genetic markers.



WOLFINESS 0.6% LOW

MATERNAL A241

HAPLOTYPE

PATERNAL **Ha.4** HAPLOTYPE

50.0% Pomeranian

50.0% Siberian Husky

Welcome to the **Embark** family!

Adam Boyko, Ph.D.

Ryan Boyko
CHIEF EXECUTIVE OFFICER





BREED MIX



GENETIC STATS

Wolfiness: 0.6 % LOW

Predicted adult weight: 22 lbs Genetic age: 16 human years

TEST DETAILS

Kit number: EM-6015661

Swab number: 31001804054280

BREED MIX BY CHROMOSOME

Our advanced test identifies from where Jaguar inherited every part of the chromosome pairs in his genome.

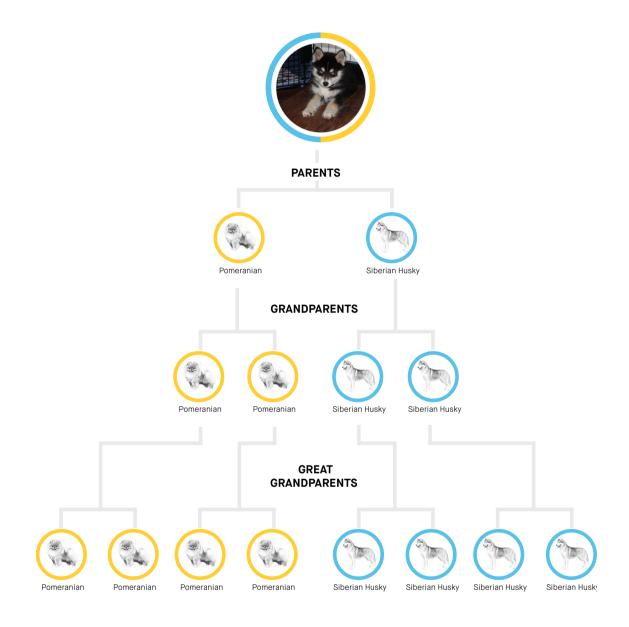








FAMILY TREE



Our algorithms predict this is the most likely family tree to explain Jaguar's breed mix, but this family tree may not be the only possible one.









Fun Fact Pomeranians boast one of the widest variety of color options in one breed. The American Kennel Club lists 23 accepted colors

POMERANIAN

Cute, feisty and furry, Poms are intelligent and loyal to their families. Don't let their cuteness fool you, however. These independent, bold dogs have minds of their own. They are alert and curious about the world around them. Unfortunately, in their minds, they are much larger than they really are, which can sometimes lead them to harass and even attack much larger dogs. Luckily, if they are properly socialized with other dogs and animals, they generally get along quite well with them. Poms take their name from the province of Pomerania, in Germany. They became especially popular when Queen Victoria allowed some of her Pomeranians to be shown in a conformation show, the first Pomeranians ever to be shown. Pomeranians make excellent pets for older people and those who are busy, because they aren't an overly dependent breed. They are also good for apartment dwellers or homes that don't have a backyard. Because of their small size, they aren't recommended for families with small children who might injure them accidentally. While Poms are good with children, they are not a good choice for very young or highly active children because of their small size. Never let your small children and your Pom play without supervision. Because they are so small, Poms can be perceived as prey by owls, eagles, hawks, coyotes, and other wild animals. Never leave them outside unattended, and be watchful if there are predatory birds in your location. If this is the case, stay close to your Pom to discourage birds from trying to carry them off!

RELATED BREEDS



American Eskimo

Dog

Cousin breed







SIBERIAN HUSKY

The Siberian Husky originated from the extreme north east of Siberia. They were initially domesticated by the Chukchi –an ancient population that thrived by herding reindeer and moving with each season to new grazing regions. They came to America in 1909 and found their place in the Alaskan wilderness. They love to be out in cold weather and are known to be the ideal sled dog. They have strong insulated paws that are perfect for traction in the snow. The Siberian Husky also has two layers in their coat that protects them from Arctic winters.

Fun Fact In 1925 a team of Siberian Huskies saved Nome, Alaska by carrying the serum to cure diphtheria a considerable distance by sled. The run was done in the middle of a blizzard and in conditions below -23 degrees Fahrenheit. The run is remembered by the annual Iditarod Trail Sled Race, and Balto, the famous sled dog who led his team through the final leg.

RELATED BREEDS



Alaskan Malamute Sibling breed



Greenland DogSibling breed



Samoyed Cousin breed







MATERNAL LINE



Through Jaguar's mitochondrial DNA we can trace his mother's ancestry back to where dogs and people first became friends. This map helps you visualize the routes that his ancestors took to your home. Their story is described below the map.

HAPLOGROUP: A1e

This female lineage likely stems from some of the original Central Asian wolves that were domesticated into modern dogs starting about 15,000 years ago. It seemed to be a fairly rare dog line for most of dog history until the past 300 years, when the lineage seemed to "explode" out and spread quickly. What really separates this group from the pack is its presence in Alaskan village dogs and Samoyeds. It is possible that this was an indigenous lineage brought to the Americas from Siberia when people were first starting to make that trip themselves! We see this lineage pop up in overwhelming numbers of Irish Wolfhounds, and it also occurs frequently in popular large breeds like Bernese Mountain Dogs, Saint Bernards and Great Danes. Shetland Sheepdogs are also common members of this maternal line, and we see it a lot in Boxers, too. Though it may be all mixed up with European dogs thanks to recent breeding events, its origins in the Americas makes it a very exciting lineage for sure!

HAPLOTYPE: A241

Part of the large A1e haplogroup, this haplotype occurs most commonly in village dogs up in Alaska.







PATERNAL LINE



Through Jaguar's Y chromosome we can trace his father's ancestry back to where dogs and people first became friends. This map helps you visualize the routes that his ancestors took to your home. Their story is described below the map.

HAPLOGROUP: A1b

For most of dog history, this haplogroup was probably quite rare. However, a couple hundred years ago it seems to have found its way into a prized male guard dog in Europe who had many offspring, including the ancestors of many European guard breeds such as Doberman Pinchers, St. Bernards, and Great Danes. Despite being rare, many of the most imposing dogs on Earth have it; strangely, so do many Pomeranians! Perhaps this explains why some Poms are so tough, acting like they're ten times their actual size! This lineage is most commonly found in working dogs, in particular guard dogs. With origins in Europe, it spread widely across other regions as Europeans took their dogs across the world.

HAPLOTYPE: Ha.4

Part of the A1b haplogroup, this haplotype is found in village dogs in North America and Africa. As for breeds, it occurs most frequently in Miniature Pinscher, Great Dane, and Poodle.







TRAITS

Coat Color

E Locus (Mask, Grizzle, Recessive Red)	Ee
K Locus (Dominant Black)	k ^y k ^y
A Locus (Agouti, Sable)	a ^w a ^t
D Locus (Dilute, Blue, Fawn)	Dd
B Locus (Brown, Chocolate, Liver, Red)	ВВ

Other Coat Traits

Furnishings / Improper Coat (RSPO2)	II	Brachycephaly (BMP3)	AC
Long Haircoat (FGF5)	GT	Natural Bobtail (T)	cc
Shedding (MC5R)	cc	Hind Dewclaws (LMBR1)	cc
Curly Coat (KRT71)	cc	Blue Eye Color	N/Dup
Hairlessness (FOXI3)	N/N		

Body Size

Body Size - IGF1	NI
Body Size - IGF1R	GA
Body Size - STC2	TA
Body Size - GHR (E195K)	GA
Body Size - GHR (P177L)	СТ

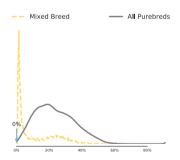
Altitude Adaptation (EPAS1) GG

Other Body Features

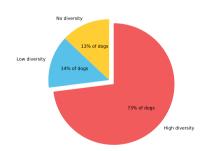
Performance

Genetic Diversity

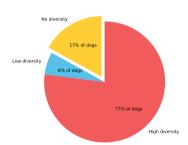
Inbreeding Coefficient 0%



MHC Class II - DLA DRB1 **High Diversity**



MHC Class II - DLA DQA1 and DQB1 **No Diversity**









CLINICAL TRAITS

These clinical genetic traits can inform clinical decisions and diagnoses. These traits do not predict a disease state or increased risk for disease. We currently assess one clinical trait: Alanine Aminotransferase Activity.

Alanine Aminotransferase Activity result: Normal

Jaguar has two normal alleles at ALT.

More information on Alanine Aminotransferase Activity:

Known to be highly expressed in liver cells, activity levels of alanine aminotransferase, or ALT, is a common value on most blood chemistry panels and is known to be a sensitive measure of liver health. Dogs with two ancestral G alleles show "normal" activity. Dogs that have one or two copies of the derived A allele may have lower resting levels of ALT activity, known as "low normal". If your dog's result is "low normal" then when a blood chemistry panel is being interpreted the values that you and your veterinarian consider "normal" may need to be adjusted. Please note that neither a "normal" nor a "low normal" result for this predicts a disease state or increased risk for liver disease. Moreover, this mutation does not associate with increased levels of ALT: If your dog has high ALT levels, please consult your veterinarian.







HEALTH

Good news! Jaguar did not test positive for any of the genetic diseases that Embark screens for.

O AT RISK CARRIER

CARRIER CONDITIONS

CARRIER status: This indicates the dog has inherited a recessive allele for a genetic trait or mutation. This is not enough to cause symptoms of the disease, but is important to bear in mind if the dog ever has offspring.

Carrier

System: Neurologic

Condition: Degenerative Myelopathy (SOD1A)





DEGENERATIVE MYELOPATHY

(SOD1A)

Carrier



SOD1 GG GA AA
GENE NAME CLEAR CARRIER AT RISK

Jaguar is a carrier for a mutation in the SOD1 gene. While he or she is unlikely to exhibit signs of disease, as a carrier, he or she will pass the mutation on to the next generation. If you choose to breed Jaguar, we highly recommend testing any potential mates for this mutation. Breeding to another carrier is not recommended as this will produce a number of affected puppies.

DESCRIPTION

A disease of mature dogs, this is a progressive degenerative disorder of the spinal cord that can cause muscle wasting and gait abnormalities. Affected dogs do not usually show signs until they are at least eight years old, where the first signs of neural degeneration appears in the nerves that innervate the hind limbs. You may notice your dog scuffing the tops of his or her hind paws, or walking with a hesitant, exaggerated gait. In advanced cases, lower motor neurons are also affected leading to weakness or near-paralysis of all four legs and widespread muscle wasting. Given the advanced age at the time of onset, the treatment for DM is aimed towards making your dog comfortable in his or her old age and include lifestyle changes and physical therapy. All known predisposing mutations for DM lie in the SOD1 gene, and have been identified in many breeds including the Boxer, Pembroke Welsh Corgi, German Shepherd Dog, Rhodesian Ridgeback, Chesapeake Bay Retriever, and Bernese Mountain Dog. SOD1 codes superoxide dismutase, an enzyme important in neutralizing free radicals and reactive oxygen species, both of which are produced as byproduct of cell metabolism. If not neutralized, these are injurious to the cell and will cause premature cell death. The first system to show effects of this is the nervous system given the highly specialized and delicate nature of these cells. Please note that these mutations are reported to have incomplete penetrance: that is, while a dog with two copies of this mutation has a much greater chance of developing DM than a dog with one copy of the mutation, or none at all, other genetic and environmental factors will also contribute to whether your dog develops DM.

More information

To learn more about this condition, you can visit http://www.caninegeneticdiseases.net/DM/basicDM.htm (http://www.caninegeneticdiseases.net/DM/basicDM.htm).

CITATIONS

Awano et al 2009 (http://www.ncbi.nlm.nih.gov/pubmed/19188595), Shelton et al 2012 (http://www.ncbi.nlm.nih.gov/pubmed/22542607), Capuccio et al 2014 (http://www.ncbi.nlm.nih.gov/pubmed/24390315)







OTHER CONDITIONS

Good news! Jaguar tested clear for 4 genetic conditions that are common in his breed mix.

- Progressive Retinal Atrophy rcd3
 Rod-cone dysplasia, rcd3 (PDE6A)
- Malignant Hyperthermia (RYR1)

- GM1 Gangliosidosis
 (GLB1 Exon 15 Alaskan Husky Variant)
- Hereditary Vitamin D-Resistant Rickets (VDR)





FULL TEST PANEL

Jaguar is also clear of 160 other genetic health conditions that Embark tests for.

To help ensure healthy breeds, every test includes analysis of our full panel of over 160 genetic health conditions.

The following pages list out all the other genetic health conditions that Jaguar tested clear for.





- MDR1 Drug Sensitivity (MDR1) (Chromosome 14)
- P2Y12 Receptor Platelet Disorder (P2RY12) (Chromosome 23)
- Factor IX Deficiency, Hemophilia B (F9 Exon 7, Terrier Variant) (Chromosome X)
- Factor IX Deficiency, Hemophilia B (F9 Exon 7, Rhodesian Ridgeback Variant) (Chromosome X)
- Factor VII Deficiency (F7 Exon 5) (Chromosome 22)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 10, Boxer Variant) (Chromosome X)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 11, Shepherd Variant 1) (Chromosome X)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 1, Shepherd Variant 2) (Chromosome X)
- Thrombopathia (RASGRP2 Exon 5, Basset Hound Variant) (Chromosome 18)
- Thrombopathia (RASGRP2 Exon 8) (Chromosome 18)
- Thrombopathia (RASGRP2 Exon 5, American Eskimo Dog Variant) (Chromosome 18)
- Von Willebrand Disease Type II (VWF Exon 28) (Chromosome 27)
- Von Willebrand Disease Type III (VWF Exon 4) (Chromosome 27)
- Von Willebrand Disease Type I (VWF) (Chromosome 27)
- Canine Leukocyte Adhesion Deficiency Type III (LAD3) (FERMT3) (Chromosome 18)
- · Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cavalier King Charles Spaniel Variant) (Chromosome 24)
- Canine Elliptocytosis (SPTB Exon 30) (Chromosome 8)
- Cyclic Neutropenia, Gray Collie Syndrome (AP3B1 Exon 20) (Chromosome 31)
- Glanzmann's Thrombasthenia Type I (ITGA2B Exon 13) (Chromosome 9)
- Glanzmann's Thrombasthenia Type I (ITGA2B Exon 12) (Chromosome 9)
- May-Hegglin Anomaly (MYH9) (Chromosome 10)
- Prekallikrein Deficiency (KLKB1 Exon 8) (Chromosome 16)
- Pyruvate Kinase Deficiency (PKLR Exon 5) (Chromosome 7)
- Pyruvate Kinase Deficiency (PKLR Exon 7 Labrador Variant) (Chromosome 7)
- Pyruvate Kinase Deficiency (PKLR Exon 7 Pug Variant) (Chromosome 7)
- Pyruvate Kinase Deficiency (PKLR Exon 7 Beagle Variant) (Chromosome 7)
- Pyruvate Kinase Deficiency (PKLR Exon 10) (Chromosome 7)
- Trapped Neutrophil Syndrome (VPS13B) (Chromosome 13)
- Ligneous Membranitis (PLG) (Chromosome 1)
- Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant) (Chromosome 17)
- Complement 3 (C3) deficiency (C3) (Chromosome 20)
- Severe Combined Immunodeficiency (PRKDC) (Chromosome 29)
- Severe Combined Immunodeficiency (RAG1) (Chromosome 18)
- X-linked Severe Combined Immunodeficiency (IL2RG Variant 1) (Chromosome X)
- X-linked Severe Combined Immunodeficiency (IL2RG Variant 2) (Chromosome X)
- Progressive Retinal Atrophy rcd1 Rod-cone dysplasia, rcd1 (PDE6B Exon 21 Irish Setter Variant) (Chromosome 3)
- Progressive Retinal Atrophy Rod-cone dysplasia, rcd1a (PDE6B Exon 21 Sloughi Variant) (Chromosome 3)
- Progressive Retinal Atrophy CNGA (CNGA1 Exon 9) (Chromosome 13)





- Progressive Retinal Atrophy prod Progressive rod-cone degeneration (PRCD Exon 1) (Chromosome 9)
- Progressive Retinal Atrophy (CNGB1) (Chromosome 2)
- Progressive Retinal Atrophy (SAG) (Chromosome 25)
- Golden Retriever Progressive Retinal Atrophy 2 (TTC8) (Chromosome 8)
- Progressive Retinal Atrophy crd1 (PDE6B) (Chromosome 3)
- Progressive Retinal Atrophy crd2 (IQCB1) (Chromosome 33)
- Progressive Retinal Atrophy crd4/cord1 (RPGRIP1) (Chromosome 15)
- Collie Eye Anomaly, Choroidal Hypoplasia (NHEJ1) (Chromosome 37)
- Day blindness, Achromatopsia, Cone Degeneration (CNGB3 Exon 6) (Chromosome 29)
- Achromatopsia (CNGA3 Exon 7 German Shepherd Variant) (Chromosome 10)
- Achromatopsia (CNGA3 Exon 7 Labrador Retriever Variant) (Chromosome 10)
- Autosomal Dominant Progressive Retinal Atrophy (RHO) (Chromosome 20)
- Canine Multifocal Retinopathy cmr1 (BEST1 Exon 2) (Chromosome 18)
- Canine Multifocal Retinopathy cmr2 (BEST1 Exon 5) (Chromosome 18)
- Canine Multifocal Retinopathy cmr3 (BEST1 Exon 10 Deletion) (Chromosome 18)
- Canine Multifocal Retinopathy cmr3 (BEST1 Exon 10 SNP) (Chromosome 18)
- Glaucoma Primary Open Angle Glaucoma (ADAMTS10 Exon 9) (Chromosome 20)
- Glaucoma Primary Open Angle Glaucoma (ADAMTS10 Exon 17) (Chromosome 20)
- Glaucoma Primary Open Angle Glaucoma (ADAMTS17 Exon 11) (Chromosome 3)
- Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9 Boston Terrier Variant) (Chromosome 5)
- Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9 Shepherd Variant) (Chromosome 5)
- Primary Lens Luxation (ADAMTS17) (Chromosome 3)
- Congenital stationary night blindness (RPE65) (Chromosome 6)
- Macular Corneal Dystrophy (MCD) (CHST6) (Chromosome 5)
- 2,8-Dihydroxyadenine (2,8-DHA) Urolithiasis (APRT) (Chromosome 5)
- Cystinuria Type I-A (SLC3A1) (Chromosome 10)
- Cystinuria Type II-A (SLC3A1) (Chromosome 10)
- Cystinuria Type I-A (SLC7A9) (Chromosome 1)
- Hyperuricosuria and Hyperuricemia or Urolithiasis (SLC2A9) (Chromosome 3)
- Polycystic Kidney Disease (PKD1) (Chromosome 6)
- Primary Hyperoxaluria (AGXT) (Chromosome 25)
- Protein Losing Nephropathy (NPHS1) (Chromosome 1)
- X-Linked Hereditary Nephropathy (Samoyed Variant 2) (COL4A5 Exon 35) (Chromosome X)
- · Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy (COL4A4 Exon 3) (Chromosome 25)
- Primary Ciliary Dyskinesia (CCDC39 Exon 3) (Chromosome 34)
- Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatosis (CKCSID), Dry Eye Curly Coat Syndrome (FAM83H Exon 5)
 (Chromosome 13)
- X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia (EDA Intron 8) (Chromosome X)
- Renal Cystadenocarcinoma and Nodular Dermatofibrosis (RCND) (FLCN Exon 7) (Chromosome 5)





- Glycogen Storage Disease Type II, Pompe's Disease (GAA) (Chromosome 9)
- Glycogen Storage Disease Type Ia, Von Gierke Disease (G6PC) (Chromosome 9)
- Glycogen Storage Disease Type IIIa (GSD IIIa) (AGL) (Chromosome 6)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A (SGSH Exon 6 Variant 1) (Chromosome 9)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A (SGSH Exon 6 Variant 2) (Chromosome 9)
- Mucopolysaccharidosis Type VII, Sly Syndrome (GUSB Exon 5) (Chromosome 6)
- Mucopolysaccharidosis Type VII, Sly Syndrome (GUSB Exon 3) (Chromosome 6)
- · Glycogen storage disease Type VII, Phosphofructokinase deficiency (PFKM Exon 21) (Chromosome 27)
- Glycogen storage disease Type VII, Phosphofructokinase deficiency (PFKM Exon 8) (Chromosome 27)
- Lagotto Storage Disease (ATG4D) (Chromosome 20)
- Neuronal Ceroid Lipofuscinosis 1 (PPT1 Exon 8) (Chromosome 15)
- Neuronal Ceroid Lipofuscinosis 2 (TPP1 Exon 4) (Chromosome 21)
- Neuronal Ceroid Lipofuscinosis 1, Cerebellar Ataxia NCL-A (ARSG Exon 2) (Chromosome 9)
- Neuronal Ceroid Lipofuscinosis 1 (CLN5 Exon 4 Variant 1) (Chromosome 22)
- Neuronal Ceroid Lipofuscinosis 6 (CLN6 Exon 7) (Chromosome 30)
- Neuronal Ceroid Lipofuscinosis 8 (CLN8 Exon 2) (Chromosome 37)
- Neuronal Ceroid Lipofuscinosis (MFSD8) (Chromosome 19)
- Neuronal Ceroid Lipofuscinosis (CLN8) (Chromosome 37)
- Neuronal Ceroid Lipofuscinosis 10 (CTSD Exon 5) (Chromosome 18)
- Neuronal Ceroid Lipofuscinosis (CLN5 Exon 4 Variant 2) (Chromosome 22)
- Adult-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2) (Chromosome 2)
- GM1 Gangliosidosis (GLB1 Exon 15 Shiba Inu Variant) (Chromosome 23)
- GM1 Gangliosidosis (GLB1 Exon 2) (Chromosome 23)
- GM2 Gangliosidosis (HEXB, Poodle Variant) (Chromosome 2)
- GM2 Gangliosidosis (HEXA) (Chromosome 30)
- Globoid Cell Leukodystrophy, Krabbe disease (GALC Exon 5) (Chromosome 8)
- Autosomal Recessive Amelogenesis Imperfecta (Italian Greyhound Variant) (Chromosome 13)
- Persistent Mullerian Duct Syndrome (AMHR2) (Chromosome 27)
- Alaskan Husky Encephalopathy, Subacute Necrotizing Encephalomyelopathy (SLC19A3) (Chromosome 25)
- Alexander Disease (GFAP) (Chromosome 9)
- Cerebellar Abiotrophy, Neonatal Cerebellar Cortical Degeneration (SPTBN2) (Chromosome 18)
- Cerebellar Ataxia, Progressive Early-Onset Cerebellar Ataxia (SEL1L) (Chromosome 8)
- Cerebellar Hypoplasia (VLDLR) (Chromosome 1)
- Spinocerebellar Ataxia, Late-Onset Ataxia (CAPN1) (Chromosome 18)
- Spinocerebellar Ataxia with Myokymia and/or Seizures (KCNJ10) (Chromosome 38)
- Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy (LGI2) (Chromosome 3)
- Fetal-Onset Neonatal Neuroaxonal Dystrophy (MFN2) (Chromosome 2)
- Hypomyelination and Tremors (FNIP2) (Chromosome 15)





- Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome (PLP) (Chromosome X)
- L-2-Hydroxyglutaricaciduria (L2HGDH) (Chromosome 0)
- Neonatal Encephalopathy with Seizures (NEWS) (ATF2) (Chromosome 36)
- Polyneuropathy, NDRG1 Greyhound Variant (NDRG1 Exon 15) (Chromosome 13)
- Polyneuropathy, NDRG1 Malamute Variant (NDRG1 Exon 4) (Chromosome 13)
- Narcolepsy (HCRTR2 Intron 6) (Chromosome 12)
- · Progressive Neuronal Abiotrophy (Canine Multiple System Degeneration) (SERAC1 Exon 15) (Chromosome 1)
- Progressive Neuronal Abiotrophy (Canine Multiple System Degeneration) (SERAC1 Exon 4) (Chromosome 1)
- Juvenile Laryngeal Paralysis and Polyneuropathy (RAB3GAP1) (Chromosome 19)
- · Hereditary Sensory Autonomic Neuropathy (HSAN), Acral Mutilation Syndrome (GDNF-AS) (Chromosome 4)
- Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1 (LPN1, ARHGEF10) (Chromosome 16)
- Dilated Cardiomyopathy (PDK4) (Chromosome 14)
- · Long QT Syndrome (KCNQ1) (Chromosome 18)
- Muscular Dystrophy Cavalier King Charles Spaniel Variant 1 (Chromosome X)
- Muscular Dystrophy Muscular Dystrophy (DMD Pembroke Welsh Corqi Variant) (Chromosome X)
- Muscular Dystrophy Muscular Dystrophy (DMD Golden Retriever Variant) (Chromosome X)
- Centronuclear Myopathy (PTPLA) (Chromosome 2)
- Exercise-Induced Collapse (DNM1) (Chromosome 9)
- Inherited Myopathy of Great Danes (BIN1) (Chromosome 19)
- Myotonia Congenita (CLCN1 Exon 7) (Chromosome 16)
- Myotonia Congenita (CLCN1 Exon 23) (Chromosome 16)
- Myotubular Myopathy 1, X-linked Myotubular Myopathy (MTM1) (Chromosome X)
- Hypocatalasia, Acatalasemia (CAT) (Chromosome 18)
- Pyruvate Dehydrogenase Deficiency (PDP1) (Chromosome 29)
- Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 53) (Chromosome 2)
- Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 8) (Chromosome 2)
- Congenital Myasthenic Syndrome (CHAT) (Chromosome 28)
- Congenital Myasthenic Syndrome (COLQ) (Chromosome 23)
- Episodic Falling Syndrome (BCAN) (Chromosome 7)
- Dystrophic Epidermolysis Bullosa (COL7A1) (Chromosome 20)
- Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1) (Chromosome 7)
- Ichthyosis, Epidermolytic Hyperkeratosis (KRT10) (Chromosome 9)
- Ichthyosis (PNPLA1) (Chromosome 12)
- Ichthyosis (SLC27A4) (Chromosome 9)
- Focal Non-Epidermolytic Palmoplantar Keratoderma, Pachyonychia Congenita (KRT16) (Chromosome 9)
- Hereditary Footpad Hyperkeratosis (FAM83G) (Chromosome 5)
- Hereditary Nasal Parakeratosis (SUV39H2) (Chromosome 2)
- Musladin-Lueke Syndrome (ADAMTSL2) (Chromosome 9)





- Cleft Lip and/or Cleft Palate (ADAMTS20) (Chromosome 27)
- Oculoskeletal Dysplasia 1, Dwarfism-Retinal Dysplasia (COL9A3, Labrador Retriever) (Chromosome 24)
- Osteogenesis Imperfecta, Brittle Bone Disease (COL1A2) (Chromosome 14)
- Osteogenesis Imperfecta, Brittle Bone Disease (SERPINH1) (Chromosome 21)
- Osteogenesis Imperfecta, Brittle Bone Disease (COL1A1) (Chromosome 9)
- Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1) (Chromosome 14)
- Skeletal Dysplasia 2 (COL11A2) (Chromosome 12)
- Craniomandibular Osteopathy (CMO) (SLC37A2) (Chromosome 5)