“SKIPPER”

HILLTOP PUPS SKIPPER

DNA Test Report | Test Date: March 17th, 2019 | embk.me/hilltoppupsskipper

GENETIC STATS

Wolfiness: 1.2 % **MEDIUM**
Predicted adult weight: **15 lbs**
Genetic age: **17 human years**

TEST DETAILS

Kit number: EM-1603259
Swab number: 31001809340146

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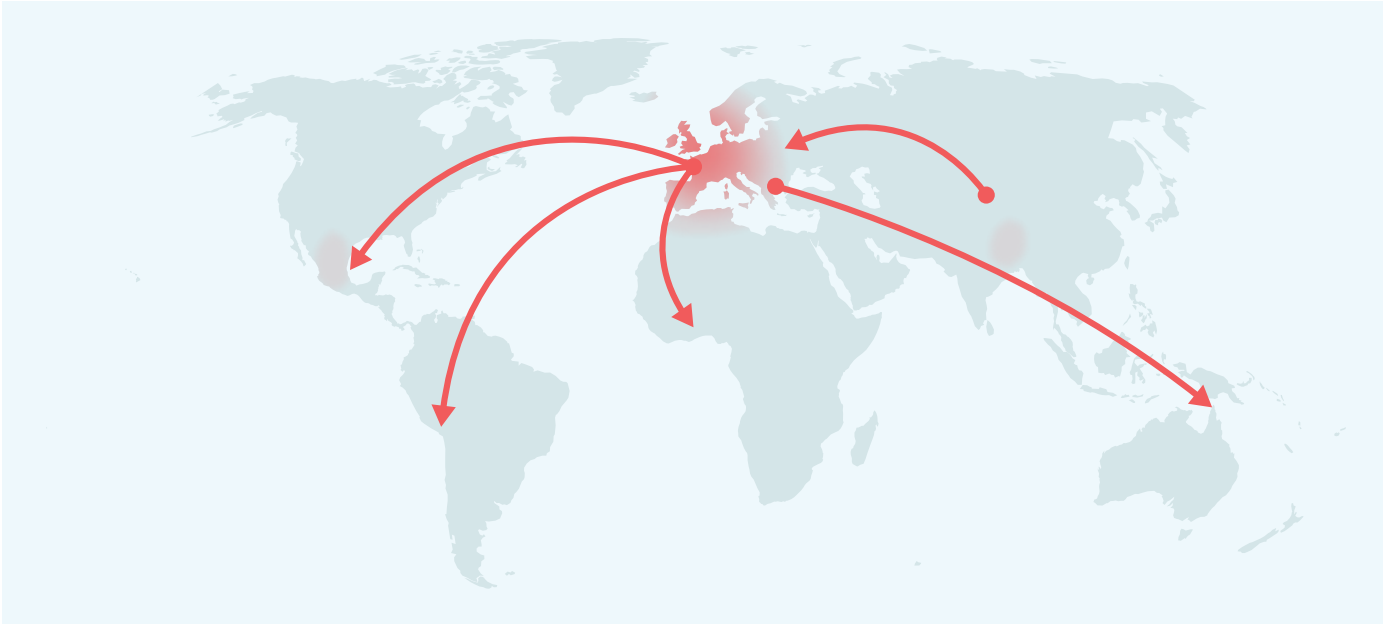
MATERNAL LINE



Through Skipper’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: B1

B1 is the second most common maternal lineage in breeds of European or American origin. It is the female line of the majority of Golden Retrievers, Basset Hounds, and Shih Tzus, and about half of Beagles, Pekingese and Toy Poodles. This lineage is also somewhat common among village dogs that carry distinct ancestry from these breeds. We know this is a result of B1 dogs being common amongst the European dogs that their conquering owners brought around the world, because nowhere on earth is it a very common lineage in village dogs. It even enables us to trace the path of (human) colonization: Because most Bichons are B1 and Bichons are popular in Spanish culture, B1 is now fairly common among village dogs in Latin America.

HAPLOTYPE: B84

Part of the large B1 haplogroup, this haplotype occurs most frequently in Golden Retrievers, Beagles, and Staffordshire Terriers.

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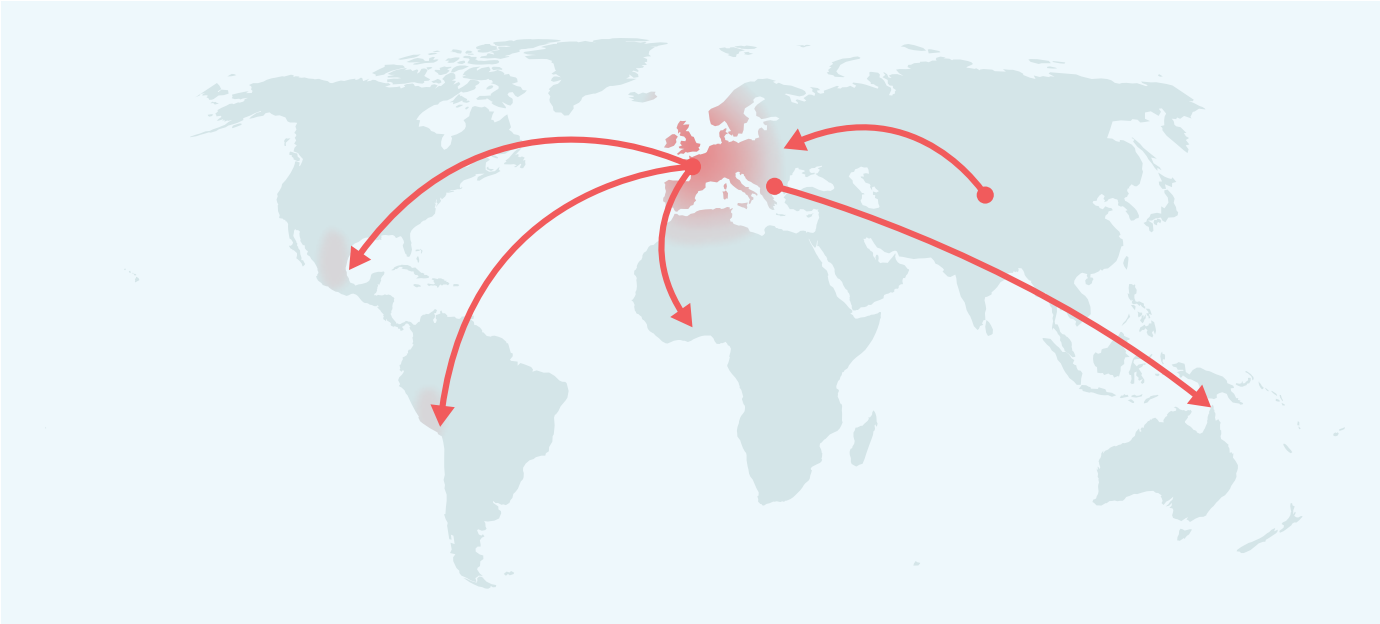
PATERNAL LINE



Through Skipper’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: A1a

Some of the wolves that became the original dogs in Central Asia around 15,000 years ago came from this long and distinguished line of male dogs. After domestication, they followed their humans from Asia to Europe and then didn't stop there. They took root in Europe, eventually becoming the dogs that founded the Vizsla breed 1,000 years ago. The Vizsla is a Central European hunting dog, and all male Vizslas descend from this line. During the Age of Exploration, like their owners, these pooches went by the philosophy, "Have sail, will travel!" From the windy plains of Patagonia to the snug and homey towns of the American Midwest, the beaches of a Pacific paradise, and the broad expanse of the Australian outback, these dogs followed their masters to the outposts of empires. Whether through good fortune or superior genetics, dogs from the A1a lineage traveled the globe and took root across the world. Now you find village dogs from this line frolicking on Polynesian beaches, hanging out in villages across the Americas, and scavenging throughout Old World settlements.

HAPLOTYPE: H1a.8/32/44

Part of the A1a haplogroup, this haplotype occurs most frequently in mixed-breed dogs.

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TRAITS

Coat Color

E Locus (Mask, Grizzle, Recessive Red)	**ee**
K Locus (Dominant Black)	**$K^B k^y$**
A Locus (Agouti, Sable)	**$a^y a^t$**
D Locus (Dilute, Blue, Fawn)	**DD**
B Locus (Brown, Chocolate, Liver, Red, Dudley)	**BB**
Saddle Tan	**NI**

Other Coat Traits

Furnishings / Improper Coat (RSPO2)	**FI**
Long Haircoat (FGF5)	**TT**
Shedding (MC5R)	**TT**
Curly Coat (KRT71)	**CT**
Hairlessness (FOXI3)	**N/N**
Hairlessness (SGK3)	**NN**
Oculocutaneous Albinism Type 2 - OCA2, Doberman Z Factor Albinism (SLC45A2)	**N/N**

Other Body Features

Brachycephaly (BMP3)	**AC**
Natural Bobtail (T)	**CC**
Hind Dewclaws (LMBR1)	**CT**
Blue Eye Color	**N/N**

Performance

Altitude Adaptation (EPAS1)	**GG**

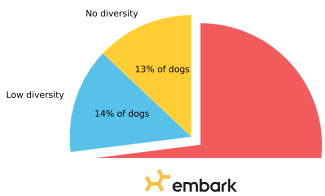
Body Size

Body Size - IGF1	**II**
Body Size - IGF1R	**GG**
Body Size - STC2	**TA**
Body Size - GHR (E195K)	**AA**
Body Size - GHR (P177L)	**CC**

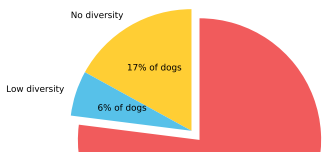
Genetic Diversity

Inbreeding Coefficient **5%**

MHC Class II - DLA DRB1
High Diversity



MHC Class II - DLA DQA1 and DQB1
High Diversity



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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: Low Normal

Hilltop Pups Skipper has one copy of a mutation associated with reduced ALT activity as measured on veterinary blood chemistry panels. Please inform your veterinarian that Hilltop Pups Skipper has this genotype, as ALT is often used as an indicator of liver health and Hilltop Pups Skipper is likely to have a lower than average resting ALT activity. As such, an increase in Hilltop Pups Skipper’s ALT activity could be evidence of liver damage, even if it is within normal limits by standard ALT reference ranges.

More information on Alanine Aminotransferase Activity:
This result helps your vet understand what your dog's baseline ALT activity is. The enzyme alanine aminotransferase, or ALT, is commonly used to evaluate liver health. Dogs with one or more copies of the "A" allele are likely to have a lower baseline ALT activity ("low normal") than dogs with zero copies of the "A" allele ("normal"). This means that you and your vet might adjust what you consider your dog's baseline ALT levels to be, and consider deviations from this as "abnormal." Please note that this mutation should never increase your dog's ALT activity. If your dog has high ALT activity, please consult your veterinarian.

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HEALTH

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

0	1
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)**

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DEGENERATIVE MYELOPATHY

(SOD1A)

Carrier

SOD1	GG	**GA**	AA
GENE NAME	**CLEAR**	**CARRIER**	**AT RISK**

Hilltop Pups Skipper 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 Hilltop Pups Skipper, 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 8 years old, where the first signs of neural degeneration appear 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 includes lifestyle changes and physical therapy. SOD1 codes superoxide dismutase, an enzyme important in neutralizing free radicals and reactive oxygen species, both of which are produced as a 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)

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OTHER CONDITIONS

Good news! Skipper tested clear for 13 genetic conditions that are common in his breed mix.

- Von Willebrand Disease Type I
  (VWF)
- Progressive Retinal Atrophy - prcd
  Progressive rod-cone degeneration (PRCD Exon 1)
- Golden Retriever Progressive Retinal Atrophy 2
  (TTC8)
- GM2 Gangliosidosis
  (HEXB, Poodle Variant)
- Muscular Dystrophy
  Muscular Dystrophy (DMD Golden Retriever Variant)
- Ichthyosis
  (PNPLA1)
- Osteochondrodysplasia, Skeletal Dwarfism
  (SLC13A1)
- Congenital Macrothrombocytopenia
  (TUBB1 Exon 1, Cavalier King Charles Spaniel Variant)
- Golden Retriever Progressive Retinal Atrophy 1
  (SLC4A3)
- Neuronal Ceroid Lipofuscinosis
  (CLN5 Golden Retriever Variant)
- Neonatal Encephalopathy with Seizures (NEWS)
  (ATF2)
- Dystrophic Epidermolysis Bullosa
  (COL7A1)
- Osteogenesis Imperfecta, Brittle Bone Disease
  (COL1A1)

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FULL TEST PANEL

Skipper is also clear of 158 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 Skipper tested clear for.

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CLEAR CONDITIONS

- 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)
- Canine Leukocyte Adhesion Deficiency Type III (LAD3) (FERMT3) (Chromosome 18)
- Canine Elliptocytosis (SPTB Exon 30) (Chromosome 8)
- Cyclic Neutropenia, Gray Collie Syndrome (AP3B1 Exon 20) (Chromosome 31)
- 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 - rcd3 Rod-cone dysplasia, rcd3 (PDE6A) (Chromosome 4)
- Progressive Retinal Atrophy - CNGA (CNGA1 Exon 9) (Chromosome 13)
- Progressive Retinal Atrophy (CNGB1) (Chromosome 2)
- Progressive Retinal Atrophy (SAG) (Chromosome 25)

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CLEAR CONDITIONS

- 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)
- 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)
- Glaucoma Primary Open Angle Glaucoma (ADAMTS17 Exon 2) (Chromosome 3)
- 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)
- Canine Fucosidosis (FUCA1) (Chromosome 2)
- 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 I (IDUA) (Chromosome 3)

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- 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 Whippet and English Springer Spaniel Variant) (Chromosome 27)
- Glycogen storage disease Type VII, Phosphofructokinase deficiency (PFKM Wachtelhund Variant) (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 Border Collie Variant) (Chromosome 22)
- Neuronal Ceroid Lipofuscinosis 6 (CLN6 Exon 7) (Chromosome 30)
- Neuronal Ceroid Lipofuscinosis 8 (CLN8 English Setter Variant) (Chromosome 37)
- Neuronal Ceroid Lipofuscinosis (MFSD8) (Chromosome 19)
- Neuronal Ceroid Lipofuscinosis (CLN8 Australian Shepherd Variant) (Chromosome 37)
- Neuronal Ceroid Lipofuscinosis 10 (CTSD Exon 5) (Chromosome 18)
- Adult-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2) (Chromosome 2)
- GM1 Gangliosidosis (GLB1 Exon 15 Shiba Inu Variant) (Chromosome 23)
- GM1 Gangliosidosis (GLB1 Exon 15 Alaskan Husky Variant) (Chromosome 23)
- GM1 Gangliosidosis (GLB1 Exon 2) (Chromosome 23)
- 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)
- Deafness and Vestibular Syndrome of Dobermans (DVDob, DINGS) (Chromosome 21)
- Shar-Pei Autoinflammatory Disease (SPAID, Shar-Pei Fever) (MTBP) (Chromosome 13)
- 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)

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- 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)
- Spongy Degeneration with Cerebellar Ataxia 1 (SDCA1), SeSAME/EAST (KCNJ10) (Chromosome 38)
- Spongy Degeneration with Cerebellar Ataxia 2 (SDCA2) (ATP1B2) (Chromosome 5)
- 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 Corgi Variant) (Chromosome X)
- Centronuclear Myopathy (PTPLA) (Chromosome 2)
- Exercise-Induced Collapse (DNM1) (Chromosome 9)
- Inherited Myopathy of Great Danes (BIN1) (Chromosome 19)
- Myostatin Deficiency, Bully Whippet Syndrome (MSTN) (Chromosome 37)
- 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)
- Malignant Hyperthermia (RYR1) (Chromosome 1)
- 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)
- Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1) (Chromosome 7)
- Ichthyosis, Epidermolytic Hyperkeratosis (KRT10) (Chromosome 9)
- Ichthyosis (SLC27A4) (Chromosome 9)
- Ichthyosis (NIPAL4) (Chromosome 4)
- 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)

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CLEAR CONDITIONS

- Hereditary Vitamin D-Resistant Rickets (VDR) (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)
- Skeletal Dysplasia 2 (COL11A2) (Chromosome 12)
- Craniomandibular Osteopathy (CMO) (SLC37A2) (Chromosome 5)