

OWNER'S NAME: Barbara Willemse

DOG'S NAME: "UK"
Joanna From Galia's Den

TEST DATE: October 22nd, 2018

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

MIXED BREED

Welcome to the
Embark family!

50.0% German Shepherd Dog



WOLFINESS **0.6% LOW**

MATERNAL HAPLOTYPE **A25**



50.0% Irish Wolfhound


Adam Boyko, Ph.D.
CHIEF SCIENCE OFFICER


Ryan Boyko
CHIEF EXECUTIVE OFFICER



"UK"

JOANNA FROM GAIA'S DEN



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BREED MIX

- German Shepherd Dog: 50.0%
- Irish Wolfhound: 50.0%

GENETIC STATS

Wolfiness: 0.6 % **LOW**
 Predicted adult weight: **83 lbs**
 Genetic age: **3 human years**

TEST DETAILS

Kit number: EM-4545405
 Swab number: 31001806010176

BREED MIX BY CHROMOSOME

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





"UK"

JOANNA FROM GAIJA'S DEN

FAMILY TREE



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



“UK”

JOANNA FROM GAIJA'S DEN

GERMAN SHEPHERD DOG



The German Shepherd dog is the second most popular dog breed in the United States, and the fourth most popular in the United Kingdom (where it is known as the Alsatian). This breed was standardized in Germany at the end of the 19th century from local dogs used for herding and livestock guarding. Their confidence, courageousness and keen sense of smell coupled with their notable intelligence make them highly suited to police work, military roles, and search and rescue. German Shepherds require regular physical and mental exercise and have a heavy shedding coat that comes in both short and long varieties. They were first recognized by the AKC in 1908 and later became fashionable as soldiers returning from WWI spoke highly of the German dogs and Hollywood popularized the breed with stars like Strongheart and Rin Tin Tin.

Fun Fact Despite being sometimes called the "Alsatian wolf dog", German Shepherds are not true wolf dogs—they are 100% dog. Nevertheless, German shepherds were crossed with wolves in the past to form the Czechoslovakian and Saarloos wolfdog breeds. German Shepherds, along with other breeds and sled dogs, were also used in the creation of the Chinook breed.

RELATED BREEDS



White Shepherd
Sibling breed



Belgian Sheepdog
Cousin breed



Belgian Tervuren
Cousin breed



Belgian Malinois
Cousin breed



Dutch Shepherd
Cousin breed



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IRISH WOLFHOUND



Fun Fact Irish Wolfhounds are mentioned in the writings of Julius Caesar, who praised them as excellent dogs of war.

Irish Wolfhounds are best known for their extraordinary size—they seem more equine than canine. They aren't especially heavy dogs, but they are exceptionally tall. Irish Wolfhounds are an extremely ancient and well-known breed of dog from Ireland. Based on their name, people can see why Irish Wolfhounds had to be tall and imposing: Their main purpose was to hunt wolves. They have a history spanning from as early as 600 CE. Ancient wooden carvings of dogs that look very much like Irish Wolfhounds have been dated even farther back to 200 BCE. Traditionally, Irish Wolfhounds were only allowed to be owned by noblemen and royalty. In modern times, however, Irish Wolfhounds are extremely popular all around the world and are known for being loving companion dogs. Irish Wolfhounds are extremely interesting dogs. They are introverted, intelligent, and reserved. They are known for having individual personalities, thus making it hard to give an accurate description of the breed's temperament. They don't like being left alone. They become extremely attached to their families, human and otherwise, and might not do well if separated from them. If prospective owners adopt an Irish Wolfhound that isn't a puppy and has had a previous family, they'll need to do a lot of work to bond with the new dog. They tend to be standoffish with strangers; however, once they've met someone once or twice they should be friendly. Because they are so large, Irish Wolfhounds need a lot of space and aren't the best choice for apartment dwellers. If owners have the space, though, Irish Wolfhounds can thrive in any environment. They love children—especially children who are members of their family—and they do well with other dogs. Owners need to keep in mind the size of Irish Wolfhounds—they might accidentally harm or frighten a much smaller dog or child.

RELATED BREEDS



Scottish Deerhound
Sibling breed



Whippet
Cousin breed



Greyhound
Cousin breed



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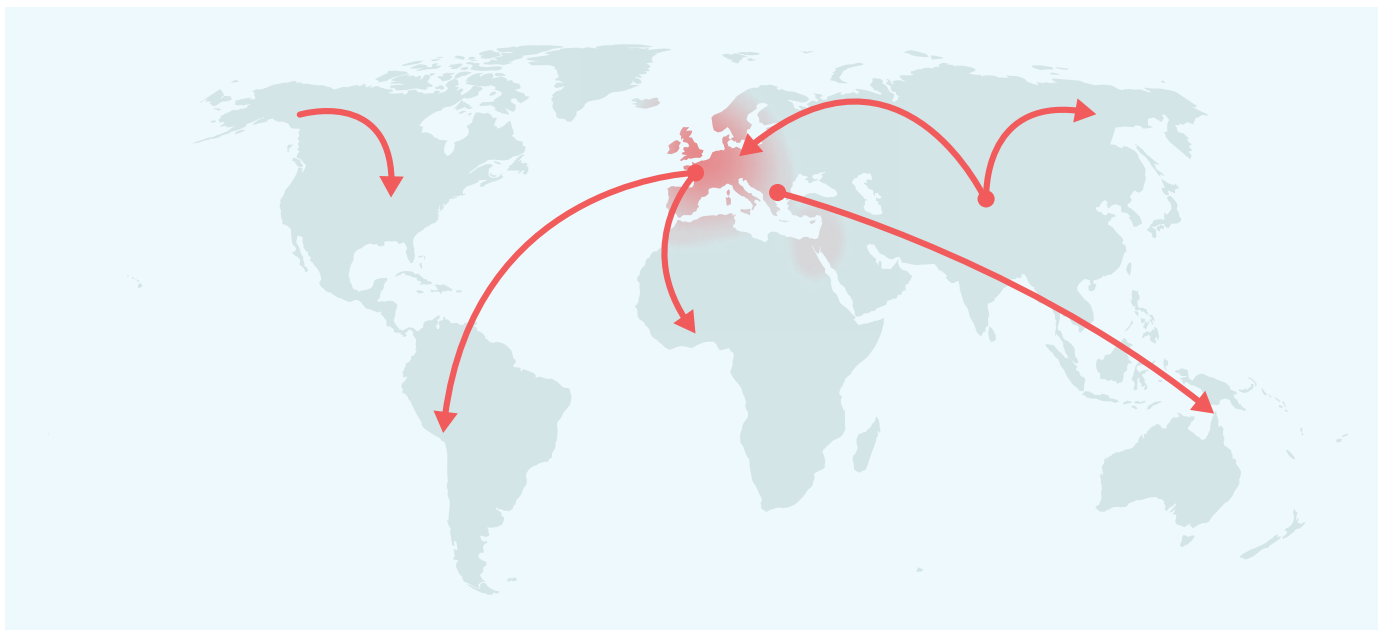


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MATERNAL LINE



Through Uk's mitochondrial DNA we can trace her mother's ancestry back to where dogs and people first became friends. This map helps you visualize the routes that her 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: A25

Part of the large A1e haplogroup, we have detected this haplotype in village dogs in Mexico. We also see it in Irish Wolfhounds, Great Pyrenees, Brittanys, and Labrador Retrievers.





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TRAITS

Coat Color

E Locus (Mask, Grizzle, Recessive Red)	E^mE^m
K Locus (Dominant Black)	K^Bk^y
A Locus (Agouti, Sable)	a^wa^t
D Locus (Dilute, Blue, Fawn)	DD
B Locus (Brown, Chocolate, Liver, Red)	BB

Other Coat Traits

Furnishings / Improper Coat (RSPO2)	FI
Long Haircoat (FGF5)	GT
Shedding (MC5R)	CT
Curly Coat (KRT71)	CC

Other Body Features

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

Body Size

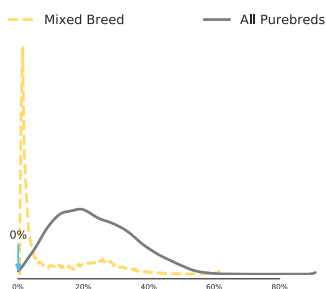
Body Size - IGF1	NN
Body Size - IGF1R	GG
Body Size - STC2	TT
Body Size - GHR (E195K)	GG
Body Size - GHR (P177L)	CC

Performance

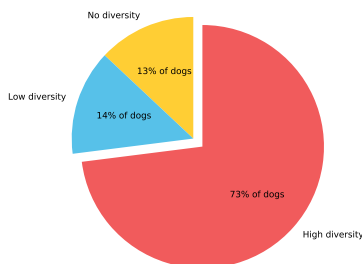
Altitude Adaptation (EPAS1)	GG
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Genetic Diversity

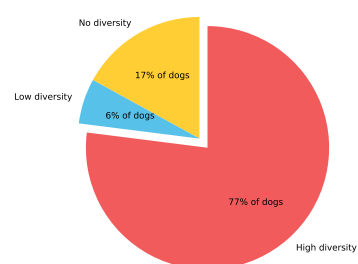
Inbreeding Coefficient **0%**



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

Joanna From Gaia's Den has one copy of a mutation associated with reduced ALT activity as measured on veterinary blood chemistry panels. Please inform your veterinarian that Joanna From Gaia's Den has this genotype, as ALT is often used as an indicator of liver health and Joanna From Gaia's Den is likely to have a lower than average resting ALT activity. As such, an increase in Joanna From Gaia's Den'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:

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.



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HEALTH

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

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AT RISK

1

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: **Ophthalmologic**

Condition: **Canine Multifocal Retinopathy cmr3 (BEST1 Exon 10 SNP)**



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CANINE MULTIFOCAL RETINOPATHY

cmr3 (BEST1 Exon 10 SNP)

Carrier



BEST1/VMD2 (Exon 10)

GENE NAME

GG

CLEAR

GT

CARRIER

TT

AT RISK

Joanna From Gaia's Den is a carrier for a mutation in the BEST1 gene. As a carrier he or she is unlikely to show signs of the disease. If you choose to breed Joanna From Gaia's Den, we highly recommend genotyping any potential mates as breeding to another carrier or an affected dog could produce affected puppies.

DESCRIPTION

This is a nonprogressive retinal disease that, in rare cases, can lead to vision loss. All known mutations lie in the BEST1 gene and are inherited in an autosomal recessive manner. We test for mutations that have been identified in Bulldog and Mastiff breeds, the Coton de Tulear, the Lapponian herder, the Finnish Lapphund, and the Swedish Lapphund. In all of these breeds, CMR is typically only identified when a vet examines the eye with an ophthalmoscope: this magnifies and illuminates the retina which, in dogs with CMR, exhibits multiple surface abnormalities that range from small, flat folds (called "retinal folds") to larger, irregularly edged raised lesions (called "geographic lesions"). Dogs with larger lesions can suffer from vision loss. CMR is fairly non-progressive; new lesions will typically stop forming by the time a dog is an adult, and some lesions will even regress with time.

CITATIONS

Zangerl et al 2010 (<http://www.ncbi.nlm.nih.gov/pubmed/21197113>), <http://omia.org/OMIA001554/9615/> ()



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OTHER CONDITIONS

Good news! Uk tested clear for 10 genetic conditions that are common in her breed mix.

- MDR1 Drug Sensitivity (MDR1)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 1, Shepherd Variant 2)
- Achromatopsia (CNGA3 Exon 7 German Shepherd Variant)
- Renal Cystadenocarcinoma and Nodular Dermatofibrosis (RCND) (FLCN Exon 7)
- Malignant Hyperthermia (RYR1)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 11, Shepherd Variant 1)
- Canine Leukocyte Adhesion Deficiency Type III (LAD3) (FERMT3)
- X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia (EDA Intron 8)
- Mucopolysaccharidosis Type VII, Sly Syndrome (GUSB Exon 3)
- Degenerative Myelopathy (SOD1A)



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FULL TEST PANEL

Uk is also clear of 156 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 Uk tested clear for.



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CLEAR CONDITIONS

- 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)
- 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)
- 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 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 - prcd 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)



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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 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)
- 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 Dermatitis (CKCSID), Dry Eye Curly Coat Syndrome (FAM83H Exon 5) (Chromosome 13)
- 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)
- 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)



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- 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 15 Alaskan Husky 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)
- 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)
- 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)



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- 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)
- 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)
- 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)
- 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, Acatlasemia (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)
- 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)



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- 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)