

OWNER'S NAME: Misty arnold

DOG'S NAME: Stella

TEST DATE: January 8th, 2019

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

## AUSSIEDOODLE

Welcome to the **Embark** family!



50.0% Australian Shepherd

WOLFINESS **0.9% MEDIUM**

MATERNAL **A226**

HAPLOTYPE



50.0% Poodle (Standard)



Adam Boyko, Ph.D.  
CHIEF SCIENCE OFFICER



Ryan Boyko  
CHIEF EXECUTIVE OFFICER



# STELLA



DNA Test Report

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embk.me/stella277

## BREED MIX

- Australian Shepherd: 50.0%
- Poodle (Standard): 50.0%

## GENETIC STATS

Wolfiness: 0.9 % **MEDIUM**  
 Predicted adult weight: **43 lbs**  
 Genetic age: **18 human years**

## TEST DETAILS

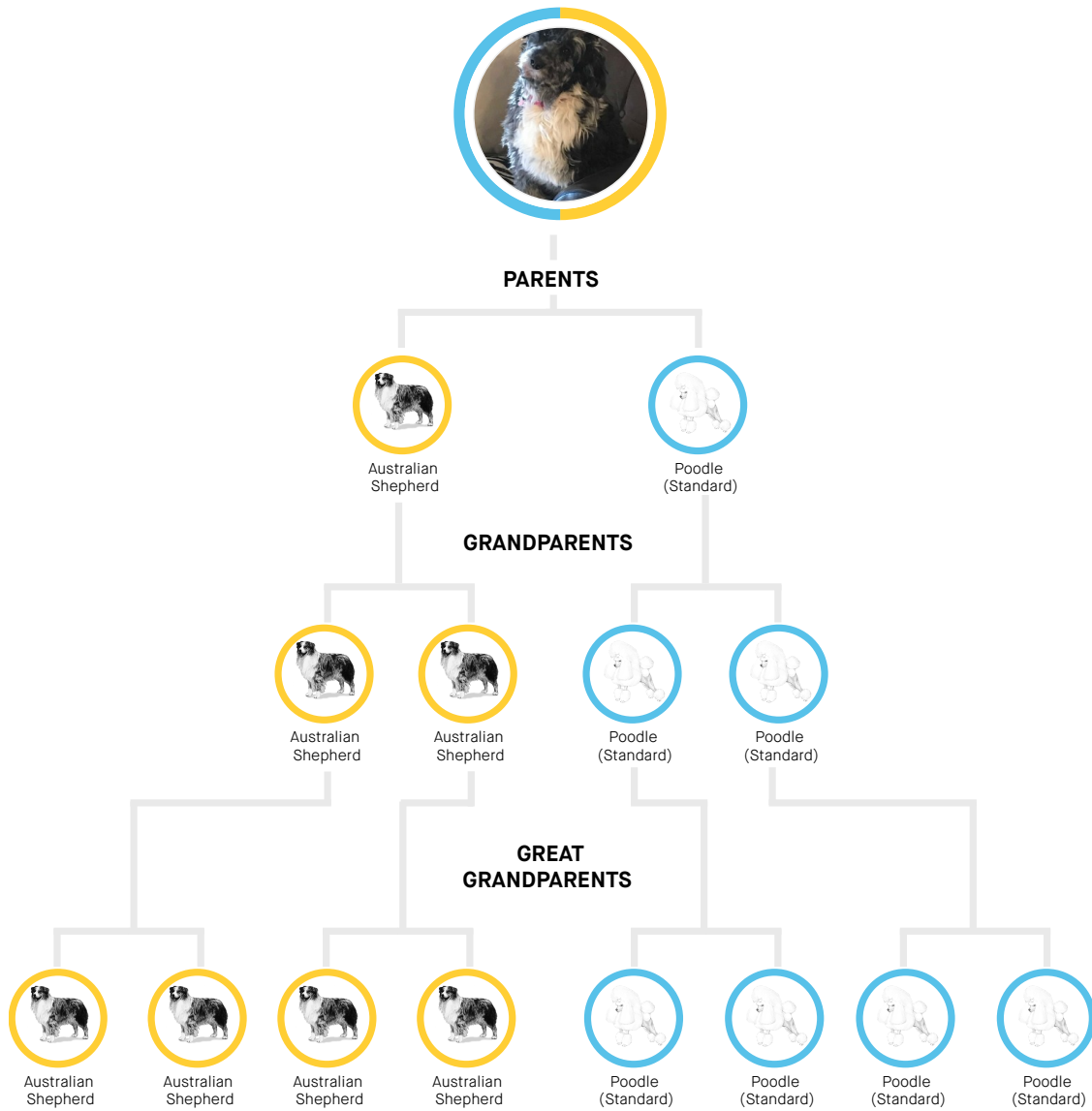
Kit number: EM-4515753  
 Swab number: 31001809341152

## BREED MIX BY CHROMOSOME

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



## FAMILY TREE



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



## AUSTRALIAN SHEPHERD



The Australian Shepherd, or Aussie, is the 17th most popular dog in the United States, and given their intelligence and temperament, it's no wonder they're so well-loved. Despite their name, the Australian Shepherd actually originated from the ranches of the United States around the 1800s. They are praised by stockmen and breeders for their trainability and intelligence. They have a medium build and a wide variation of different coat colors. Australian Shepherds have considerable energy and they usually need a job to do to keep themselves entertained, though they're also happy to spend time with the family and settle down at the end of the day. Australian Shepherds are often employed as guide dogs, rescue dogs, and therapy dogs. In addition to exercising an Aussie, it's equally important to keep their mind occupied, as if an Australian Shepherd gets bored they do have the tendency to invent their own games or activities, which sometimes involve destructive behaviors. This is a breed that thrives on close companionship. Aussies are at times called "Velcro Dogs" for their tendency to stay close to their owner.

**Fun Fact** Australian Shepherds rose to popularity and fame as rodeo stars. After the first World War, people flocked to the west and to watch exhibitions that showcased these very talented canines.

### RELATED BREEDS



**Miniature/MAS-type Australian Shepherd**  
Sibling breed



**Collie**  
Sibling breed



**Shetland Sheepdog**  
Sibling breed



**Border Collie**  
Sibling breed



**Pembroke Welsh Corgi**  
Cousin breed



**Cardigan Welsh Corgi**  
Cousin breed



## POODLE (STANDARD)



The Standard Poodle is a popular, water-loving dog used for centuries as a bird dog and popular pet. Poodles were established in Germany by the 15th century. Oddly enough, they are the national dog breed of France, and they were the most popular breed of dog in the United States throughout the 1960s and 70s. They're still quite popular today, owing to their intelligence, trainability, and non-shedding coats. Although well-known for their fancy fur, they're one of the most intelligent breeds of dog and require a lot of exercise and stimulation.

**Fun Fact** From 1989 to 1991, John Suter raced a team of Poodles in the Iditarod. Although his teams placed in the back half of the pack, he managed to win \$2,000 in prize money before retiring his poodle team. The Iditarod has since changed its rules to specify that only northern dog breeds can compete.

### RELATED BREEDS



**Poodle (Toy)**  
Sibling breed



**Poodle (Miniature)**  
Sibling breed



**Maltese**  
Cousin breed



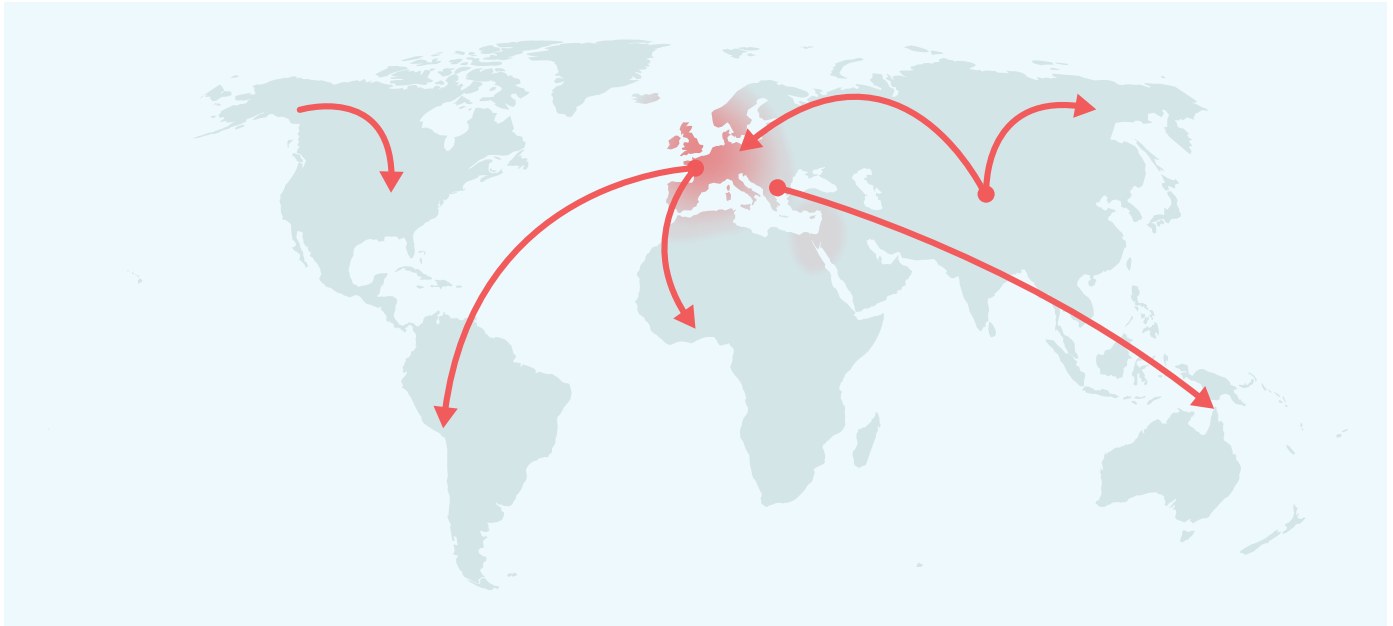
**Havanese**  
Cousin breed



**Bichon Frise**  
Cousin breed



## MATERNAL LINE



Through Stella'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: A226

Part of the large A1e haplogroup, we have spotted this haplotype in village dogs in Central and South America and Papua New Guinea. Among the 10 breeds we have detected it in, we see it most frequently in Border Collies, Doberman Pinschers, and Samoyeds.



## TRAITS

### Coat Color

E Locus (Mask, Grizzle, Recessive Red)	<b>E<sup>m</sup>E</b>
K Locus (Dominant Black)	<b>K<sup>B</sup>k<sup>y</sup></b>
A Locus (Agouti, Sable)	<b>a<sup>w</sup>a</b>
D Locus (Dilute, Blue, Fawn)	<b>DD</b>
B Locus (Brown, Chocolate, Liver, Red, Dudley)	<b>Bb</b>

### Other Coat Traits

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

### Other Body Features

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

### Performance

Altitude Adaptation (EPAS1)	<b>GG</b>
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### Body Size

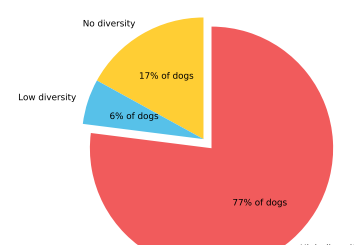
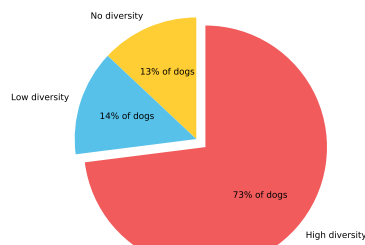
Body Size - IGF1	<b>NI</b>
Body Size - IGF1R	<b>GG</b>
Body Size - STC2	<b>TT</b>
Body Size - GHR (E195K)	<b>GA</b>
Body Size - GHR (P177L)	<b>CC</b>

### 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**

Stella has one copy of a mutation associated with reduced ALT activity as measured on veterinary blood chemistry panels. Please inform your veterinarian that Stella has this genotype, as ALT is often used as an indicator of liver health and Stella is likely to have a lower than average resting ALT activity. As such, an increase in Stella'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! Stella did not test positive for any of the genetic diseases that Embark screens for.

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AT RISK

0

CARRIER



## OTHER CONDITIONS

Good news! Stella tested clear for 15 genetic conditions that are common in her breed mix.

- MDR1 Drug Sensitivity (MDR1)
- Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cavalier King Charles Spaniel Variant)
- Progressive Retinal Atrophy - prcd Progressive rod-cone degeneration (PRCD Exon 1)
- Canine Multifocal Retinopathy cmr1 (BEST1 Exon 2)
- Neuronal Ceroid Lipofuscinosis 6 (CLN6 Exon 7)
- GM2 Gangliosidosis (HEXB, Poodle Variant)
- Neonatal Encephalopathy with Seizures (NEWS) (ATF2)
- Craniomandibular Osteopathy (CMO) (SLC37A2)
- Von Willebrand Disease Type I (VWF)
- Cyclic Neutropenia, Gray Collie Syndrome (AP3B1 Exon 20)
- Collie Eye Anomaly, Choroidal Hypoplasia (NHEJ1)
- Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9 Shepherd Variant)
- Neuronal Ceroid Lipofuscinosis (CLN8)
- Degenerative Myelopathy (SOD1A)
- Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1)



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## **FULL TEST PANEL**

Stella is also clear of 157 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 Stella tested clear for.



## 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)
- 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)
- 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)
- Golden Retriever Progressive Retinal Atrophy 1 (SLC4A3) (Chromosome 37)
- Golden Retriever Progressive Retinal Atrophy 2 (TTC8) (Chromosome 8)



## CLEAR CONDITIONS

- Progressive Retinal Atrophy - crd1 (PDE6B) (Chromosome 3)
- Progressive Retinal Atrophy - crd2 (IQCB1) (Chromosome 33)
- Progressive Retinal Atrophy - crd4/cord1 (RPGRIP1) (Chromosome 15)
- 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 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)
- 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)
- 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)
- 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)



## CLEAR CONDITIONS

- 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 8 (CLN8 Exon 2) (Chromosome 37)
- Neuronal Ceroid Lipofuscinosis (MFSD8) (Chromosome 19)
- Neuronal Ceroid Lipofuscinosis 10 (CTSD Exon 5) (Chromosome 18)
- Neuronal Ceroid Lipofuscinosis (CLN5 Golden Retriever Variant) (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 (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 (LG12) (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)
- 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)



## CLEAR CONDITIONS

- 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)
- 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)
- 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, Acatlasemia (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)
- 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)
- 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)
- Hereditary Vitamin D-Resistant Rickets (VDR) (Chromosome 27)



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- 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)
- Skeletal Dysplasia 2 (COL11A2) (Chromosome 12)