

# Certificate of Breed

DOG'S NAME: "Savvy" OWNER'S NAME: Laura Harmon Hickory's Savvy Revelation TEST DATE: May 18th, 2019

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

WOLFINESS 1.5% HIGH

MATERNAL A414/643

**HAPLOTYPE** 

N/A - No "Y' chromosome PATERNAL

**HAPLOTYPE** 

**HEALTH** - Good news! Savvy did not test positive for any of the genetic diseases that Embark screens for.





**LLEWELLIN SETTER** 

Welcome to the Embark family!

Adam Boyko, Ph.D.

Ryan Boyko

CHIEF EXECUTIVE OFFICER





# **GENETIC STATS**

Predicted adult weight: **52 lbs** Genetic age: **8 human years** 

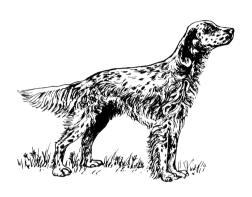
# **TEST DETAILS**

Kit number: EM-8351258 Swab number: 31001809332516





## **LLEWELLIN SETTER**



The Llewellin Setter is widely cherished as one of the best upland bird hunting dogs. A strain of the English Setter, the Llewellin has a great many devotees and is recognized as its own breed by The American Field Dog Stud Book, the oldest dog registry in the USA. These dogs are well-loved for their hunting prowess in intelligence, stamina, nose, biddability, in addition to its gentle nature. While the history of the English Setter goes back to the 1500s, the Llewellin Setter was developed in a breeding program by R. L. Purcell Llewellin in the 1860s, using dogs from Laverack. These dogs were intended to excel in field work, and Llewellin succeeded in that task. Today, a great number of field-type English Setters have Llewellin blood from the historical crossing of the lines.

Fun Fact When a Llewellin Setter smells a bird nearby, rather than barking, they'll start wagging their tail to alert their owner.

## **RELATED BREEDS**







**Irish Setter**Cousin breed





## **MATERNAL LINE**



Through Savvy'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: A1b

This female lineage was very likely one of the original lineages in the wolves that were first domesticated into dogs in Central Asia about 15,000 years ago. Since then, the lineage has been very successful and travelled the globe! Dogs from this group are found in ancient Bronze Age fossils in the Middle East and southern Europe. By the end of the Bronze Age, it became exceedingly common in Europe. These dogs later became many of the dogs that started some of today's most popular breeds, like German Shepherds, Pugs, Whippets, English Sheepdogs and Miniature Schnauzers. During the period of European colonization, the lineage became even more widespread as European dogs followed their owners to farflung places like South America and Oceania. It's now found in many popular breeds as well as village dogs across the world!

#### **HAPLOTYPE: A414/643**

Part of the A1b haplogroup, this haplotype occurs most frequently in the English Springer Spaniels.





## **TRAITS**

#### **Coat Color**

E Locus (Mask, Grizzle, Recessive Red)

K Locus (Dominant Black)

KYKY

A Locus (Agouti, Sable)

D Locus (Dilute, Blue, Fawn)

B Locus (Brown, Chocolate, Liver, Red)

#### **Other Coat Traits**

Furnishings / Improper Coat (RSP02)

Coat Length (FGF5)

TT

Shedding (MC5R)

TT

Coat Texture (KRT71)

CC

Hairlessness (FOXI3)

### **Other Body Features**

Muzzle Length (BMP3) CC
Tail Length (T) CC
Hind Dewclaws (LMBR1) CC
Blue Eye Color (ALX4) N/N

# **Body Size**

Body Size - IGF1

Body Size - IGF1R

GG

Body Size - STC2

TT

Body Size - GHR (E195K)

GG

Body Size - GHR (P177L)

CC

#### Performance

Altitude Adaptation (EPAS1) GG

## **Genetic Diversity**

Inbreeding Coefficient 16%





#### TRAITS: COAT COLOR

Coat Color RESULT

#### E Locus (MC1R)

The E Locus determines if and where a dog can produce dark (black or brown) hair. Dogs with two copies of the recessive **e** allele do not produce dark hairs at all, and will be "red" over their entire body. The shade of red, which can range from a deep copper to yellow/gold to cream, is dependent on other genetic factors including the Intensity (I) Locus, which has yet to be genetically mapped. In addition to determining if a dog can develop dark hairs at all, the E Locus can give a dog a black "mask" or "widow's peak," unless the dog has overriding coat color genetic factors. Dogs with one or two copies of the **Em** allele usually have a melanistic mask (dark facial hair as commonly seen in the German Shepherd and Pug). Dogs with no copies of **Em** but one or two copies of the **Eg** allele usually have a melanistic "widow's peak" (dark forehead hair as commonly seen in the Afghan Hound and Borzoi, where it is called either "grizzle" or "domino").

No dark hairs anywhere (ee)

#### K Locus (CBD103)

The K Locus  $\mathbf{K}^B$  allele "overrides" the A Locus, meaning that it prevents the A Locus genotype from affecting coat color. For this reason, the  $\mathbf{K}^B$  allele is referred to as the "dominant black" allele. As a result, dogs with at least one  $\mathbf{K}^B$  allele will usually have solid black or brown coats (or red/cream coats if they are  $\mathbf{e}\mathbf{e}$  at the E Locus) regardless of their genotype at the A Locus, although several other genes could impact the dog's coat and cause other patterns, such as white spotting. Dogs with the  $\mathbf{k}^{\mathbf{y}}\mathbf{k}^{\mathbf{y}}$  genotype will show a coat color pattern based on the genotype they have at the A Locus.

Not expressed (k<sup>y</sup>k<sup>y</sup>)

## A Locus (ASIP)

The A Locus controls switching between black and red pigment in hair cells, but it will only be expressed in dogs that are not **ee** at the E Locus and are **k**<sup>y</sup>**k**<sup>y</sup> at the K Locus. Sable (also called "Fawn") dogs have a mostly or entirely red coat with some interspersed black hairs. Agouti (also called "Wolf Sable") dogs have red hairs with black tips, mostly on their head and back. Black and tan dogs are mostly black or brown with lighter patches on their cheeks, eyebrows, chest, and legs. Recessive black dogs have solid-colored black or brown coats.

Not expressed (a<sup>t</sup>a<sup>t</sup>)

#### D Locus (MLPH)

Dogs with two copies of the **d** allele will have all black pigment lightened ("diluted") to gray, or brown pigment lightened to lighter brown in their hair, skin, and sometimes eyes. There are many breed-specific names for these dilute colors, such as "blue", "charcoal", "fawn", "silver", and "Isabella". Note that dilute dogs have a higher incidence of Color Dilution Alopecia, especially in certain breeds. Dogs with one copy of the **d** allele will not be dilute, but can pass the **d** allele on to their puppies.

Not expressed (DD)

#### **B Locus (TYRP1)**

to as "chestnut".

Dogs with two copies of the  $\bf b$  allele produce brown pigment instead of black in both their hair and skin. Dogs with one copy of the  $\bf b$  allele will produce black pigment, but can pass the  $\bf b$  allele on to their puppies. E locus  $\bf ee$  dogs that carry two  $\bf b$  alleles will have red or cream coats, but have brown noses, eye rims, and footpads. "Liver" or "chocolate" is the preferred color term for brown in most breeds. In the Llewellin Setter it is referred

Likely brown colored nose/feet (bb)

Registration: FDSB 1684253





## TRAITS: OTHER COAT TRAITS

Other Coat Traits RESULT

#### Furnishings (RSPO2) LINKAGE

Dogs with one or two copies of the **F** allele have "furnishings": the mustache, beard, and eyebrows characteristic of breeds like the Schnauzer, Scottish Terrier, and Wire Haired Dachshund. A dog with two I alleles will not have furnishings, which is sometimes called an "improper coat" in breeds where furnishings are part of the breed standard. The mutation is a genetic insertion which we measure indirectly using a linkage test highly correlated with the insertion.

Likely unfurnished (no mustache, beard, and/or eyebrows) (II)

#### Coat Length (FGF5)

The FGF5 gene is known to affect hair length in many different species, including cats, dogs, mice, and humans. In dogs, the **T** allele confers a long, silky haircoat as observed in the Yorkshire Terrier and the Long Haired Whippet. The ancestral **G** allele causes a shorter coat as seen in the Boxer or the American Staffordshire Terrier. In certain breeds (such as Corgi), the long haircoat is described as "fluff."

Likely long coat (TT)

#### Shedding (MC5R)

Dogs with at least one copy of the ancestral **C** allele, like many Labradors and German Shepherd Dogs, are heavy or seasonal shedders, while those with two copies of the **T** allele, including many Boxers, Shih Tzus and Chihuahuas, tend to be lighter shedders. Dogs with furnished/wire-haired coats caused by RSPO2 (the furnishings gene) tend to be low shedders regardless of their genotype at this gene.

Likely light to moderate shedding (TT)

#### Coat Texture (KRT71)

Dogs with a long coat and at least one copy of the **T** allele have a wavy or curly coat characteristic of Poodles and Bichon Frises, while dogs with two copies of the ancestral **C** allele have a straight coat. Dogs with short coats may carry one or two copies of the **T** allele but still have straight coats.

Likely straight coat (CC)

#### Hairlessness (FOXI3) LINKAGE

A duplication in the FOXI3 gene causes hairlessness over most of the body as well as changes in tooth shape and number. This mutation occurs in Peruvian Inca Orchid, Xoloitzcuintli (Mexican Hairless), and Chinese Crested (other hairless breeds have different mutations). Dogs with the **NDup** genotype are likely to be hairless while dogs with the **NN** genotype are likely to have a normal coat. The **DupDup** genotype has never been observed, suggesting that dogs with that genotype cannot survive to birth. Please note that

Very unlikely to be hairless (NN)





#### TRAITS: OTHER BODY FEATURES

Other Body Features RESULT

#### Muzzle Length (BMP3)

Dogs in medium-length muzzle (mesocephalic) breeds like Staffordshire Terriers and Labradors, and long muzzle (dolichocephalic) breeds like Whippet and Collie have one, or more commonly two, copies of the ancestral **C** allele. Dogs in many short-length muzzle (brachycephalic) breeds such as the English Bulldog, Pug, and Pekingese have two copies of the derived **A** allele. At least five different genes affect muzzle length in dogs, with BMP3 being the only one with a known causal mutation. For example, the skull shape of some breeds, including the dolichocephalic Scottish Terrier or the brachycephalic Japanese Chin, appear to be caused by other genes. Thus, dogs may have short or long muzzles due to other genetic factors that are not yet known to science.

Likely medium or long muzzle (CC)

#### Tail Length (T)

Whereas most dogs have two **C** alleles and a long tail, dogs with one **G** allele are likely to have a bobtail, which is an unusually short or absent tail. This mutation causes natural bobtail in many breeds including the Pembroke Welsh Corgi, the Australian Shepherd, and the Brittany Spaniel. Dogs with **GG** genotypes have not been observed, suggesting that dogs with the **GG** genotype do not survive to birth. Please note that this mutation does not explain every natural bobtail! While certain lineages of Boston Terrier, English Bulldog, Rottweiler, Miniature Schnauzer, Cavalier King Charles Spaniel, and Parson Russell Terrier, and Dobermans are born with a natural bobtail, these breeds do not have this mutation. This suggests that other unknown genetic mutations can also lead to a natural bobtail.

Likely normal-length tail (CC)

#### Hind Dewclaws (LMBR1)

Common in certain breeds such as the Saint Bernard, hind dewclaws are extra, nonfunctional digits located midway between a dog's paw and hock. Dogs with at least one copy of the **T** allele have about a 50% chance of having hind dewclaws. Note that other (currently unknown to science) mutations can also cause hind dewclaws, so some **TT** or **TC** dogs will have hind dewclaws.

Unlikely to have hind dew claws (CC)

#### Blue Eye Color (ALX4) LINKAGE

Embark researchers discovered this large duplication associated with blue eyes in Arctic breeds like Siberian Husky as well as tri-colored (non-merle) Australian Shepherds. Dogs with at least one copy of the duplication (**Dup**) are more likely to have at least one blue eye. Some dogs with the duplication may have only one blue eye (complete heterochromia) or may not have blue eyes at all; nevertheless, they can still pass the duplication and the trait to their offspring. **NN** dogs do not carry this duplication, but may have

Less likely to have blue eyes (NN)





# **TRAITS: BODY SIZE**

Body Size	RESULT
Body Size (IGF1)  The I allele is associated with smaller body size.	Larger (NN)
Body Size (IGFR1)  The A allele is associated with smaller body size.	Larger (GG)
Body Size (STC2)  The A allele is associated with smaller body size.	Larger (TT)
Body Size (GHR - E195K)  The A allele is associated with smaller body size.	Larger (GG)
Body Size (GHR - P177L)  The <b>T</b> allele is associated with smaller body size.	Larger (CC)





## TRAITS: PERFORMANCE

Performance RESULT

Altitude Adaptation (EPAS1)

This mutation causes dogs to be especially tolerant of low oxygen environments (hypoxia), such as those found at high elevations. Dogs with at least one  $\bf A$  allele are less susceptible to "altitude sickness." This mutation was originally identified in breeds from high altitude areas such as the Tibetan Mastiff.

Normal altitude tolerance (GG)



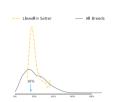


## TRAITS: GENETIC DIVERSITY

Genetic Diversity RESULT

#### **Coefficient Of Inbreeding**

Our genetic COI measures the proportion of your dog's genome where the genes on the mother's side are identical by descent to those on the father's side.



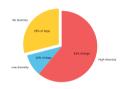
#### MHC Class II - DLA DRB1

A Dog Leukocyte Antigen (DLA) gene, DRB1 encodes a major histocompatibility complex (MHC) protein involved in the immune response. Some studies have shown associations between certain DRB1 haplotypes and autoimmune diseases such as Cushing's disease, but these findings have yet to be scientifically validated.

#### **No Diversity**

16%

How common is this amount of diversity in purebreds:

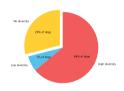


#### MHC Class II - DLA DQA1 and DQB1

DQA1 and DQB1 are two tightly linked DLA genes that code for MHC proteins involved in the immune response. A number of studies have shown correlations of DQA-DQB1 haplotypes and certain autoimmune diseases; however, these have not yet been scientifically validated.

#### No Diversity

How common is this amount of diversity in purebreds:







## **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 (ALT) Activity result: Low Normal

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

The liver enzyme alanine aminotransferase, or ALT, is one of several values your veterinarian measures on routine blood work to gauge 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 your veterinarian may recommend blood work to establish an individualized baseline ALT value during an annual wellness exam or before starting certain medications. You and your veterinarian would then be able to monitor your dog for any deviation from this established baseline. Please note that this mutation should never cause an increase in your dog's ALT activity and does not cause liver disease. If your dog has high ALT activity, please consult your veterinarian.





# **HEALTH**

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

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CARRIER





## **OTHER CONDITIONS**

Good news! Savvy tested clear for 7 other common genetic diseases that Embark tests for.

- MDR1 Drug Sensitivity (MDR1)
- Primary Lens Luxation (ADAMTS17)
- Degenerative Myelopathy (SOD1A)
- Exercise-Induced Collapse (DNM1)

- Progressive Retinal Atrophy prcd
   Progressive rod-cone degeneration (PRCD Exon 1)
- Hyperuricosuria and Hyperuricemia or Urolithiasis (SLC2A9)
- Dilated Cardiomyopathy (PDK4)





## **FULL TEST PANEL**

Savvy is also clear of 163 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 Savvy 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 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 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 (CNGB1) (Chromosome 2)
- Progressive Retinal Atrophy (SAG) (Chromosome 25)
- Golden Retriever Progressive Retinal Atrophy 1 (SLC4A3) (Chromosome 37)





## **CLEAR CONDITIONS**

- 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)
- 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)
- 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)
- 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)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A (SGSH Exon 6 Variant 1) (Chromosome 9)





## CLEAR CONDITIONS

- 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)
- 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 (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)





## CLEAR CONDITIONS

- 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, Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation (POANV)
   (RAB3GAP1, Rottweiler Variant) (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)
- 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)
- 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)
- 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)





## **CLEAR CONDITIONS**

- 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)
- 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)