



## **BREED MIX**



## **GENETIC STATS**

Wolfiness: 1.8 % **HIGH**Predicted adult weight: **24 lbs** 

Predicted adult weight: **24 lbs**Genetic age: **17 human years** 

## **TEST DETAILS**

Kit number: EM-3983627

Swab number: 31001805240069

## **BREED MIX BY CHROMOSOME**

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

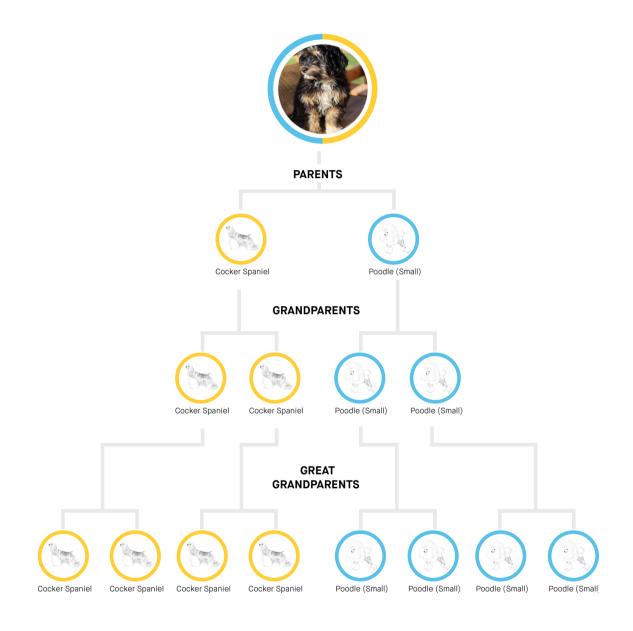








# **FAMILY TREE**



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





DNA Test Report

embk.me/montanasmountainecho

Fun Fact A Cocker Spaniel named Lupo is the pet of the Duke and Duchess of Cambridge, also known as Prince William and Kate Middleton.

#### **COCKER SPANIEL**

Test Date: December 5th, 2018

A beloved family dog, the popular Cocker Spaniel is a cheerful and merry breed. The Cocker Spaniel derives from the Spaniel family, originating in Spain with references dating back to the 14th century. By the 1800s, Spaniels were split between toy dogs and large hunting dogs. The Cocker Spaniel was named after its excellence in hunting woodcock. Although Spaniels were located in England for hundreds of years, they were not considered an individual breed until 1892 when the English Kennel Club recognized the Cocker Spaniel as an official breed. However, before this, English Cockers were being imported to America and were recognized by the AKC as an official breed in 1887. It was not until 1936 that they gained recognition by the AKC as English Cocker Spaniels, which were bigger than the American-type Cocker Spaniels. In 1938, a motion was passed not to breed the two types of Spaniels, which finally led to the distinction of Cocker Spaniels as their own breed. It may come as a surprise the Cocker Spaniels are sporting dogs, whose ability to adapt to household living may deceive you of their impressive agility and obedience skills. They are strong performers in conformation shows, while also capable of field work, portraying their well-rounded nature. The Cocker Spaniel is a soft and affectionate breed, that appreciates time and attention with the family. It should be noted that although they thrive on human interaction, the Cocker Spaniel's hunting instincts can kick in when out exercising so remember to keep them on a leash in a non-enclosed area. Due to their soft and gentle nature, a Cocker Spaniel can easily become nervous in unknown scenarios or with harsher training methods which can result in barking and sometimes submissive urination (be prepared!). The Cocker Spaniel is a visually impressive breed, whose thick and heavy coat requires constant grooming to prevent knots and tangles developing. It is common for Cocker Spaniels to seek professional assistance in keeping their pooch well groomed. This beloved family dog ranks as the 30th most popular breed.

#### **RELATED BREEDS**



English Cocker Spaniel Sibling breed



Sussex Spaniel
Cousin breed



English Springer
Spaniel
Cousin breed



Cavalier King
Charles Spaniel
Cousin breed





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# **POODLE (SMALL)**

Miniature and toy poodles are varieties of the poodle breed which originated in Germany in the 15th century. Unlike the larger standard poodle (>15 inches tall), these small poodles were not developed for hunting---except for truffles!---and were generally used as lap dogs and companions. Small poodles are frequently used to create designer dogs like Schnoodles and Maltipoos with low-shedding, hypoallergenic coats. All poodles are highly intelligent and energetic, and need daily exercise and stimulation. They are overall healthy dogs, although heritable eye disease, epilepsy and allergies are relatively common, and toy poodles also have a heightened risk of accidents/trauma due to their small size.

#### **Alternative Names**

Toy Poodle, Miniature Poodle

**Fun Fact** Although Toy Poodles are the most popular dog breed in Japan, Poodles as a group are the eight most popular breed in the US, with miniature poodles being the most common variety.

#### RELATED BREEDS



Poodle (Standard) Sibling breed



**Maltese** Cousin breed



Havanese Cousin breed



**Bichon Frise**Cousin breed







## **MATERNAL LINE**



Through Echo'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: A1a**

A1a is the most common maternal lineage among Western dogs. This lineage traveled from the site of dog domestication in Central Asia to Europe along with an early dog expansion perhaps 10,000 years ago. It hung around in European village dogs for many millennia. Then, about 300 years ago, some of the prized females in the line were chosen as the founding dogs for several dog breeds. That set in motion a huge expansion of this lineage. It's now the maternal lineage of the overwhelming majority of Mastiffs, Labrador Retrievers and Gordon Setters. About half of Boxers and less than half of Shar-Pei dogs descend from the A1a line. It is also common across the world among village dogs, a legacy of European colonialism.

#### **HAPLOTYPE: A17**

Part of the large A1a haplogroup, this common haplotype is found in village dogs across the globe. Among breed dogs, we find it most frequently in Labrador Retrievers, Boxers, and Mastiffs.







## **TRAITS**

## **Coat Color**

E Locus (Mask, Grizzle, Recessive Red)	Ee
K Locus (Dominant Black)	$\mathbf{k}^{\mathbf{y}}\mathbf{k}^{\mathbf{y}}$
A Locus (Agouti, Sable)	a <sup>t</sup> a <sup>t</sup>
D Locus (Dilute, Blue, Fawn)	DD
B Locus (Brown, Chocolate, Liver, Red, Dudley)	ВВ
Saddle Tan	NI

#### **Other Coat Traits**

FI	Brachycephaly (BMP3)	CC
TT	Natural Bobtail (T)	CC
СТ	Hind Dewclaws (LMBR1)	CC
СТ	Blue Eye Color	N/N
N/N		
NN	Performance	
	TT CT CT N/N	TT Natural Bobtail (T)  CT Hind Dewclaws (LMBR1)  CT Blue Eye Color  N/N

N/N

**Other Body Features** 

Altitude Adaptation (EPAS1)

Oculocutaneous Albinism Type 2 -

OCA2, Doberman Z Factor Albinism

(SLC45A2)

#### **Body Size**

Body Size - IGF1	II
Body Size - IGF1R	GA
Body Size - STC2	AA
Body Size - GHR (E195K)	GA
Body Size - GHR (P177L)	СС

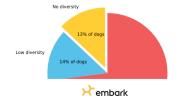
#### **Genetic Diversity**

Inbreeding Coefficient 1%



MHC Class II - DLA DRB1

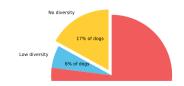
No Diversity



MHC Class II - DLA DQA1 and DQB1

No Diversity

GG







#### **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

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







# **HEALTH**

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

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CARRIER





#### OTHER CONDITIONS

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

- Von Willebrand Disease Type I (VWF)
- Progressive Retinal Atrophy prcd
   Progressive rod-cone degeneration (PRCD Exon 1)
- Glycogen storage disease Type VII, Phosphofructokinase deficiency
   (PFKM Whippet and English Springer Spaniel Variant)
- Neonatal Encephalopathy with Seizures (NEWS) (ATF2)
- Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1)

- Congenital Macrothrombocytopenia
   (TUBB1 Exon 1, Cavalier King Charles Spaniel Variant)
- Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy (COL4A4 Exon 3)
- GM2 Gangliosidosis
   (HEXB, Poodle Variant)
- Degenerative Myelopathy (SOD1A)
- Exercise-Induced Collapse (DNM1)





# **FULL TEST PANEL**

Echo is also clear of 159 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 Echo tested clear for.







- MDR1 Drug Sensitivity (MDR1) (Chromosome 14)
- P2Y12 Receptor Platelet Disorder (P2RY12) (Chromosome 23)
- Factor IX Deficiency, Hemophilia B (F9 Exon 7, Terrier Variant) (Chromosome X)
- Factor IX Deficiency, Hemophilia B (F9 Exon 7, Rhodesian Ridgeback Variant) (Chromosome X)
- Factor VII Deficiency (F7 Exon 5) (Chromosome 22)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 10, Boxer Variant) (Chromosome X)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 11, Shepherd Variant 1) (Chromosome X)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 1, Shepherd Variant 2) (Chromosome X)
- Thrombopathia (RASGRP2 Exon 5, Basset Hound Variant) (Chromosome 18)
- Thrombopathia (RASGRP2 Exon 8) (Chromosome 18)
- Thrombopathia (RASGRP2 Exon 5, American Eskimo Dog Variant) (Chromosome 18)
- Von Willebrand Disease Type II (VWF Exon 28) (Chromosome 27)
- Von Willebrand Disease Type III (VWF Exon 4) (Chromosome 27)
- 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)





- Golden Retriever Progressive Retinal Atrophy 1 (SLC4A3) (Chromosome 37)
- 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)
- Primary Lens Luxation (ADAMTS17) (Chromosome 3)
- Congenital stationary night blindness (RPE65) (Chromosome 6)
- 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)
- Primary Ciliary Dyskinesia (CCDC39 Exon 3) (Chromosome 34)
- Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatosis (CKCSID), Dry Eye Curly Coat Syndrome (FAM83H Exon 5)
   (Chromosome 13)
- X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia (EDA Intron 8) (Chromosome X)
- Renal Cystadenocarcinoma and Nodular Dermatofibrosis (RCND) (FLCN Exon 7) (Chromosome 5)
- Glycogen Storage Disease Type II, Pompe's Disease (GAA) (Chromosome 9)
- Glycogen Storage Disease Type Ia, Von Gierke Disease (G6PC) (Chromosome 9)
- Glycogen Storage Disease Type IIIa (GSD IIIa) (AGL) (Chromosome 6)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A (SGSH Exon 6 Variant 1) (Chromosome 9)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A (SGSH Exon 6 Variant 2) (Chromosome 9)





- Mucopolysaccharidosis Type VII, Sly Syndrome (GUSB Exon 5) (Chromosome 6)
- Mucopolysaccharidosis Type VII, Sly Syndrome (GUSB Exon 3) (Chromosome 6)
- Glycogen storage disease Type VII, Phosphofructokinase deficiency (PFKM 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 (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)
- 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)
- 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)
- 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)
- 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)
- Skeletal Dysplasia 2 (COL11A2) (Chromosome 12)
- Craniomandibular Osteopathy (CMO) (SLC37A2) (Chromosome 5)