

Swab code: bru100100_swab
Swab activated on 7/28/2022
Results completed on 7/28/2022
Report accessed on 7/28/2022
Ordered by Lorenz Connelly

vetsupport@embarkvet.com
1-855-203-8271

Patient Information

Bruce

0 yrs 2 mths - M
Genetic Age: 4 human years
Predicted Adult Weight: 44 lbs

Client Information

Omar W

oww@example.com
555-555-0598

Breed Information

100.0% Border Collie



1 Increased Risk Result

Collie Eye Anomaly, Choroidal Hypoplasia, CEA [Page 2](#)



219 Clear Results

Bruce is not at increased risk for 219 of the genetic health variants that Embark tests. [Page 5](#)



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1 Increased Risk Result

Collie Eye Anomaly, Choroidal Hypoplasia, CEA

How to interpret this result

Bruce has two copies of this recessive variant in the NHEJ1 gene and is considered at risk for developing Collie Eye Anomaly, Choroidal Hypoplasia, CEA. Collie Eye Anomaly (CEA) is a complex congenital defect of the retina, although retinal involvement is secondary to the primary ocular defects associated with this disorder. There is no cure for CEA, although surgical intervention may be of value depending on the clinical presentation.

You can learn more about clinical signs, treatment, and care for Bruce below or email vetsupport@embarkvet.com should you desire to speak with a genetic counselor.

What is Collie Eye Anomaly, Choroidal Hypoplasia, CEA?

Collie Eye Anomaly (CEA) is a complex congenital defect of the retina, although retinal involvement is secondary to the primary ocular defects associated with this disorder. The primary phenotypic element of the disorder is regional hypoplasia of the choroid, the highly vascular layer underlying the retina; the choroid anchors the retina to the underlying structures and supplies it with oxygen and nourishment. Disease phenotype is variable in both the severity of its clinical manifestations and the ophthalmic lesions present, ranging from choroidal hypoplasia, scleral coloboma, to retinal detachment, although most affected dogs are not visually impaired. Tortuous retinal vessels and multiple retinal folds may be observed in a minority of cases. Dogs presenting with optic nerve head coloboma or large lesions are often most severely affected and at risk of retinal detachment and blindness.

Age of Onset of Clinical Signs or Symptoms

CEA is present from birth, however, by three months of age the puppy fundus changes from its blue tapetal color to the adult green-yellow appearance. At the same time minor chorioretinal changes can be masked by the development of more pigment in the RPE, in which case affected animals are classified as so-called "go normal" animals.

Variant Info

NHEJ1 Intron 4

Recessive inheritance

2 copies of the variant

Keep in mind, while an individual dog may no longer have an easily detectable CEA phenotype, they will pass on this variant to their offspring, and there is no guarantee that at risk progeny will have a mild phenotype.

Clinical Signs

The ophthalmoscopic changes consist of focal hypoplasia or absence of the tapetum and a focal lack of pigment of the retinal pigmented epithelium allowing the visualization of abnormal choroidal blood vessels.

Dogs with poor vision will often act hesitant on stairs, bump into doorways or walls, and be disoriented if furniture is moved.

Follow-up Diagnostics to Consider

A comprehensive ophthalmological examination is recommended, however, its diagnostic value depends on the age of the dog.

Treatment and Management Options

- There is no cure for CEA, although surgical intervention can help mitigate the signs of the disease in severe cases. If surgery is not an option, lifestyle changes can be made to help blind dogs adapt to their condition.
- Surgical treatment may consist of retinal reattachment (in the case of detached retinas), though the success rate is decreased if there is no retinal pigment. If the retina is not yet detached and there is retinal pigment present, a laser retinopexy around the coloboma may be an option.
- Owners should help affected dogs navigate their homes and the outside world by keeping furniture in the same location, making sure they are on a leash when in unfamiliar territory, and training them to understand verbal commands or using scent markers.

More Information

Choroidal hypoplasia is always bilaterally present, but to varying degrees between affected dogs and even within the same individual. However, no matter how extensive, choroidal hypoplasia appears to be of no clinical significance in terms of an effect on sight.

Some studies suggest that NEHJ1 intronic deletion is not the causative variant but, rather, a marker linked to the locus underlying the trait in some populations but apparently linked to both the wild-type and CH-causing locus in other populations.

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References

Parker HG, Kukekova AV, Akey DT, et al. Breed relationships facilitate fine-mapping studies: a 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. *Genome Res.* 2007;17(11):1562-1571. doi:10.1101/gr.6772807"

Fredholm M, Larsen RC, Jonsson M, Soderlund MA, Hardon T, Proschowsky HF. Discrepancy in compliance between the clinical and genetic diagnosis of choroidal hypoplasia in Danish Rough Collies and Shetland Sheepdogs. *Anim Genet.* 2016;47(2):250-252. doi:10.1111/age.12405"



Mizukami K, Chang HS, Ota M, et al. Collie eye anomaly in Hokkaido dogs: case study. *Vet Ophthalmol.* 2012;15(2):128-132. doi:10.1111/j.1463-5224.2011.00950.x"

Mellersh CS. The genetics of eye disorders in the dog. *Canine Genet Epidemiol.* 2014;1:3. Published 2014 Apr 16. doi:10.1186/2052-6687-1-3"

All Conditions Tested



To view COI and traits information, log into your account.

Auditory (2)



	Gene	Copies	Results
 Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS	MYO7A	0	Clear
 Early Onset Adult Deafness, EOAD - Rhodesian Ridgeback Variant	EPS8L2 Deletion Exon 12	0	Clear

Cardiac (4)

Dilated Cardiomyopathy




	Gene	Copies	Results
 Dilated Cardiomyopathy, DCM1 - Doberman Pinscher Variant 1	PDK4	0	Clear
 Dilated Cardiomyopathy, DCM2 - Doberman Pinscher Variant 2	TTN	0	Clear

Other

	Gene	Copies	Results
 Cardiomyopathy and Juvenile Mortality - Belgian Shepherd Variant	YARS2	0	Clear
 Long QT Syndrome - English Springer Spaniel Variant	KCNQ1	0	Clear


Endocrine (3)

Hypothyroidism




	Gene	Copies	Results
 Congenital Dyshormonogenic Hypothyroidism with Goiter - Shih Tzu Variant	SLC5A5	0	Clear
 Congenital Hypothyroidism - Rat, Toy Fox, and Hairless Terrier Variant	TPO Exon 3	0	Clear
 Congenital Hypothyroidism - Tenterfield Terrier Variant	TPO Exon 9	0	Clear

Gastrointestinal (4)

Gastroenteropathy












	Gene	Copies	Results
 Lundehund Syndrome	LEPREL1	0	Clear

Malabsorptive Disorder

	Gene	Copies	Results
 Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption - Beagle Variant	CUBN Exon 8	0	Clear
 Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption - Border Collie Variant	CUBN Exon 53	0	Clear
 Inherited Selected Cobalamin Malabsorption with Proteinuria - Komondor Variant	CUBN	0	Clear

Hematologic (32)

Coagulopathy

	Gene	Copies	Results
 Bernard-Soulier Syndrome, BSS - Cocker Spaniel Variant	GP9	0	Clear
 Congenital Macrothrombocytopenia - Cairn and Norfolk Terrier Variant	TUBB1 Exon 1	0	Clear
 Factor IX Deficiency, Hemophilia B - Rhodesian Ridgeback Variant	F9 Exon 7	0	Clear
 Factor IX Deficiency, Hemophilia B - Terrier Variant	F9 Exon 7	0	Clear
 Factor VII Deficiency	F7 Exon 5	0	Clear
 Factor VIII Deficiency, Hemophilia A - Boxer Variant	F8 Exon 10	0	Clear
 Factor VIII Deficiency, Hemophilia A - German Shepherd Variant 1	F8 Exon 11	0	Clear
 Factor VIII Deficiency, Hemophilia A - German Shepherd Variant 2	F8 Exon 1	0	Clear
 Glanzmann's Thrombasthenia Type I - Great Pyrenees Variant	ITGA2B Exon 13	0	Clear
 Glanzmann's Thrombasthenia Type I - Otterhound Variant	ITGA2B Exon 12	0	Clear
 May-Hegglin Anomaly - Pug Variant	MYH9	0	Clear

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	Gene	Copies	Results
✔ P2Y12 Receptor Platelet Disorder - Greater Swiss Mountain Dog Variant	P2Y12	0	Clear
✔ Platelet Factor X Receptor Deficiency, Scott Syndrome - German Shepherd Dog Variant	TMEM16F	0	Clear
✔ Prekallikrein Deficiency - Shih Tzu Variant	KLKB1 Exon 8	0	Clear
✔ Thrombopathia - American Eskimo Dog Variant	RASGRP1 Exon 5	0	Clear
✔ Thrombopathia - Basset Hound Variant	RASGRP1 Exon 5	0	Clear
✔ Thrombopathia - Landseer Variant	RASGRP1 Exon 8	0	Clear
✔ Von Willebrand Disease Type I, Type I vWD	VWF	0	Clear
✔ Von Willebrand Disease Type II, Type II vWD - Pointer Variant	VWF	0	Clear
✔ Von Willebrand Disease Type III, Type III vWD - Shetland Sheepdog Variant	VWF Exon 7	0	Clear
✔ Von Willebrand Disease Type III, Type III vWD - Terrier Variant	VWF Exon 4	0	Clear

Red Blood Cell Abnormality

	Gene	Copies	Results
✔ Canine Elliptocytosis - Labrador Retriever Variant	SPTB Exon 30	0	Clear
✔ Methemoglobinemia - Pomeranian Variant	CYB5R3	0	Clear
✔ Pyruvate Kinase Deficiency - Basenji Variant	PKLR Exon 5	0	Clear
✔ Pyruvate Kinase Deficiency - Beagle Variant	PKLR Exon 7	0	Clear
✔ Pyruvate Kinase Deficiency - Labrador Retriever Variant	PKLR Exon 7	0	Clear
✔ Pyruvate Kinase Deficiency - Pug Variant	PKLR Exon 7	0	Clear
✔ Pyruvate Kinase Deficiency - Terrier Variant	PKLR Exon 10	0	Clear

White Blood Cell Abnormality

	Gene	Copies	Results
✔ Canine Leukocyte Adhesion Deficiency Type I, CLAD I - Setter Variant	ITGB2 Exon 3	0	Clear
✔ Canine Leukocyte Adhesion Deficiency Type III, CLAD III - German Shepherd Variant	FERMT3	0	Clear
✔ Trapped Neutrophil Syndrome, TNS	VPS13B Exon 19	0	Clear

Other

	Gene	Copies	Results
✔ Ligneous Membranitis, LM - Scottish Terrier Variant	PLG	0	Clear

Immunologic (6)

	Gene	Copies	Results
✔ Complement 3 Deficiency, C3 Deficiency - Brittany Variant	C3	0	Clear
✔ Severe Combined Immunodeficiency, SCID - Terrier Variant	PRKDC	0	Clear
✔ Severe Combined Immunodeficiency, SCID - Wetterhoun Variant	RAG1	0	Clear
✔ Shar-Pei Autoinflammatory Disease, SPAID, Shar-Pei Fever	MTBP	0	Clear
✔ X-linked Severe Combined Immunodeficiency, X-SCID - Basset Hound Variant	IL2RG Exon 1	0	Clear
✔ X-linked Severe Combined Immunodeficiency, X-SCID - Corgi Variant	IL2RG	0	Clear

Integument (18)**Collagen Abnormality**

	Gene	Copies	Results
✔ Dystrophic Epidermolysis Bullosa - Central Asian Shepherd Dog Variant	COL7A1	0	Clear
✔ Dystrophic Epidermolysis Bullosa - Golden Retriever Variant	COL7A1 Exon 68	0	Clear

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	Gene	Copies	Results
✓ Ehlers Danlos - Doberman Pinscher Variant	ADAMTS2	0	Clear
✓ Musladin-Lueke Syndrome, MLS - Beagle Variant	ADAMTSL2 Exon 7	0	Clear

Keratin Abnormality

	Gene	Copies	Results
✓ Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, Dry Eye Curly Coat Syndrome, CKCSID - Cavalier King Charles Spaniel Variant	FAM83H	0	Clear
✓ Focal Non-Epidermolytic Palmoplantar Keratoderma, Pachyonychia Congenita - Dogue de Bordeaux Variant	KRT16 Exon 6	0	Clear
✓ Hereditary Footpad Hyperkeratosis - Rottweiler Variant	DSG1	0	Clear
✓ Hereditary Footpad Hyperkeratosis - Terrier and Kromfohrlander Variant	FAM83G	0	Clear
✓ Hereditary Nasal Parakeratosis, HNPk - Labrador Retriever Variant	SUV39H2	0	Clear
✓ Ichthyosis, Epidermolytic Hyperkeratosis - Terrier Variant	KRT10 Intron 5	0	Clear
✓ Ichthyosis, ICH1 - Golden Retriever Variant	PNPLA1 Exon 8	0	Clear
✓ Ichthyosis - American Bulldog Variant	NIPAL4 Exon 6	0	Clear
✓ Ichthyosis - Great Dane Variant	SLC27A4	0	Clear

Other

	Gene	Copies	Results
✓ Bald Thigh Syndrome - Greyhound Variant	IGFBP5	0	Clear
✓ Ectodermal Dysplasia, Skin Fragility Syndrome - Chesapeake Bay Retriever Variant	PKP1 Intron 1	0	Clear
✓ Lethal Acrodermatitis, LAD - Bull Terrier Variant	MKLN1	0	Clear

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	Gene	Copies	Results
✓ Oculocutaneous Albinism, OCA - Small Breed Variant	SLC45A2	0	Clear
✓ X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia, XHED - German Shepherd Dog Variant	EDA	0	Clear

Metabolic (33)

Enzyme Deficiency

	Gene	Copies	Results
✓ Hypocatalasia, Acatalasemia - Beagle Variant	CAT	0	Clear
✓ L-2-Hydroxyglutaricaciduria, L2HGA - Staffordshire Bull Terrier Variant	L2HGDH	0	Clear
✓ Pyruvate Dehydrogenase Deficiency - Spaniel Variant	PDP1	0	Clear

Storage Disease

	Gene	Copies	Results
✓ Canine Fucosidosis - English Springer Spaniel Variant	FUCA1	0	Clear
✓ GM1 Gangliosidosis - Alaskan Husky Variant	GLB1 Exon 15	0	Clear
✓ GM1 Gangliosidosis - Portuguese Water Dog Variant	GLB1 Exon 2	0	Clear
✓ GM1 Gangliosidosis - Shiba Inu Variant	GLB1 Exon 15	0	Clear
✓ GM2 Gangliosidosis - Japanese Chin Variant	HEXA	0	Clear
✓ GM2 Gangliosidosis - Poodle Variant	HEXB Exon 3	0	Clear
✓ Globoid Cell Leukodystrophy, Krabbe Disease - Terrier Variant	GALC Exon 5	0	Clear
✓ Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA - Maltese Variant	G6PC	0	Clear
✓ Glycogen Storage Disease Type II, Pompe's Disease, GSD II - Finnish and Swedish Lapphund, Lapponian Herder Variant	GAA Exon 15	0	Clear
✓ Glycogen Storage Disease Type IIIA, GSD IIIA - Curly Coated Retriever Variant	AGL GDE	0	Clear

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	Gene	Copies	Results
✓ Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency - Wachtelhund Variant	PFKM Exon 8	0	Clear
✓ Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency - Whippet and English Springer Spaniel Variant	PFKM Exon 21	0	Clear
✓ Lagotto Storage Disease	ATG4D Exon 10	0	Clear
✓ Late-Onset Neuronal Ceroid Lipofuscinosis, NCL12 - Australian Cattle Dog Variant	ATP13A2	0	Clear
✓ Mucopolysaccharidosis IIIB, Sanfilippo Syndrome Type B, MPS IIIB - Schipperke Variant	NAGLU	0	Clear
✓ Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA - Dachshund Variant	SGSH	0	Clear
✓ Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA - New Zealand Huntaway Variant	SGSH	0	Clear
✓ Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII - German Shepherd Variant	GUSB	0	Clear
✓ Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII - Terrier Brasileiro Variant	GUSB	0	Clear
✓ Neuronal Ceroid Lipofuscinosis 1, NCL1 - Dachshund Variant	PPT1 Exon 8	0	Clear
✓ Neuronal Ceroid Lipofuscinosis 10, NCL10 - American Bulldog Variant	CTSD Exon 5	0	Clear
✓ Neuronal Ceroid Lipofuscinosis 2, NCL2 - Dachshund Variant	TPP1 Exon 4	0	Clear
✓ Neuronal Ceroid Lipofuscinosis 5, NCL5 - Border Collie and Australian Cattle Dog Variant	CLN5 Exon 4	0	Clear
✓ Neuronal Ceroid Lipofuscinosis 5, NCL5 - Golden Retriever Variant	CLN5 Exon 4	0	Clear
✓ Neuronal Ceroid Lipofuscinosis 6, NCL6 - Australian Shepherd Variant	CLN6 Exon 7	0	Clear
✓ Neuronal Ceroid Lipofuscinosis 7, NCL7 - Chihuahua and Chinese Crested Variant	MFSD8	0	Clear
✓ Neuronal Ceroid Lipofuscinosis 8, NCL8 - Australian Shepherd and German Shorthaired Pointer Variant	CLN8	0	Clear

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	Gene	Copies	Results
✓ Neuronal Ceroid Lipofuscinosis 8, NCL8 - English Setter Variant	CLN8 Exon 2	0	Clear
✓ Neuronal Ceroid Lipofuscinosis 8, NCL8 - Saluki Variant	CLN8	0	Clear
✓ Neuronal Ceroid Lipofuscinosis, Cerebellar Ataxia, NCL4A - American Staffordshire Terrier Variant	ARSG Exon 2	0	Clear

Muscular (13)

Movement Disorder

	Gene	Copies	Results
✓ Myotonia Congenita - Australian Cattle Dog Variant	CLCN1 Exon 23	0	Clear
✓ Myotonia Congenita - Miniature Schnauzer Variant	CLCN1 Exon 7	0	Clear

Muscular Dystrophy




	Gene	Copies	Results
✓ Limb Girdle Muscular Dystrophy - Boston Terrier Variant	SGCD	0	Clear
✓ Muscular Dystrophy - Cavalier King Charles Spaniel Variant 1	DMD	0	Clear
✓ Muscular Dystrophy - Golden Retriever Variant	DMD	0	Clear
✓ Ullrich-like Congenital Muscular Dystrophy - Labrador Retriever Variant 1	COL6A3 Exon 10	0	Clear

Myopathy

	Gene	Copies	Results
✓ Centronuclear Myopathy, CNM - Labrador Retriever Variant	PTPLA	0	Clear
✓ Exercise-Induced Collapse, EIC	DNM1	0	Clear
✓ Inflammatory Myopathy - Dutch Shepherd Variant	SLC25A12	0	Clear
✓ Inherited Myopathy of Great Danes	BIN1	0	Clear







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	Gene	Copies	Results
 Myotubular Myopathy 1, X-linked Myotubular Myopathy, XL-MTM - Labrador Retriever Variant	MTM1 Exon 7	0	Clear
 Nemaline Myopathy - American Bulldog Variant	NEB	0	Clear
Other			
 Myostatin Deficiency, Bully Whippet Syndrome	MSTN	0	Clear

Neurologic (32)

Brain or Seizure Disorder

	Gene	Copies	Results
 Alaskan Husky Encephalopathy, Subacute Necrotizing Encephalomyelopathy	SLC19A3 Exon 2	0	Clear
 Alexander Disease - Labrador Retriever Variant	GFAP Exon 4	0	Clear
 Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy - Lagotto Romagnolo Variant	LG12 Exon 8	0	Clear
 Cerebellar Abiotrophy, Neonatal Cerebellar Cortical Degeneration, NCCD - Beagle Variant	SPTBN2	0	Clear
 Cerebellar Hypoplasia - Eurasier Variant	VLDLR	0	Clear
 Hereditary Ataxia, Cerebellar Degeneration - Old English Sheepdog and Gordon Setter Variant	RAB24 Exon 1	0	Clear
 Neonatal Encephalopathy with Seizures, NEWS - Poodle Variant	ATF2	0	Clear
 Progressive Early-Onset Cerebellar Ataxia - Finnish Hound Variant	SEL1L	0	Clear
 Spinocerebellar Ataxia with Myokymia and/or Seizures - Terrier Variant 2	KCNJ10	0	Clear
 Spinocerebellar Ataxia, Late-Onset Ataxia, LoSCA - Terrier Variant 1	CAPN1	0	Clear

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	Gene	Copies	Results
✓ Spongy Degeneration with Cerebellar Ataxia 1, SDCA1, SeSAME/EAST Syndrome - Shepherd Variant 1	KCNJ10	0	Clear
✓ Spongy Degeneration with Cerebellar Ataxia 2, SDCA2 - Shepherd Variant 2	ATP1B2	0	Clear

Movement Disorder

	Gene	Copies	Results
✓ Degenerative Myelopathy, DM	SOD1A	0	Clear
✓ Hypomyelination and Tremors - Weimaraner Variant	FNIP2	0	Clear
✓ Juvenile Myoclonic Epilepsy - Rhodesian Ridgeback Variant	DIRAS1	0	Clear
✓ Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD - Chinese Crested Variant	SERAC1	0	Clear
✓ Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD - Kerry Blue Terrier Variant	SERAC1	0	Clear
✓ Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome - English Springer Spaniel Variant	PLP1	0	Clear

Narcolepsy

	Gene	Copies	Results
✓ Narcolepsy - Dachshund Variant	HCRT2	0	Clear
✓ Narcolepsy - Doberman Pinscher Variant	HCRT2	0	Clear
✓ Narcolepsy - Labrador Retriever Variant	HCRT2	0	Clear

Neurodegenerative Disorder

	Gene	Copies	Results
✓ Fetal-Onset Neonatal Neuroaxonal Dystrophy - Giant Schnauzer Variant	MFN2	0	Clear
✓ Neuroaxonal Dystrophy, NAD - Rottweiler Variant	VPS11	0	Clear
✓ Neuroaxonal Dystrophy, NAD - Spanish Water Dog Variant	TECPR2	0	Clear

Neuropathy

	Gene	Copies	Results
✓ Alaskan Malamute Polyneuropathy, AMPN	NDRG1	0	Clear
✓ Demyelinating Polyneuropathy - Miniature Schnauzer Variant	SBF2/MTRM13	0	Clear
✓ Juvenile Laryngeal Paralysis and Polyneuropathy, Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation, POANV - Rottweiler Variant	RAB3GAP1	0	Clear
✓ Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1, LPN1	ARHGEF10 Exon 17	0	Clear
✓ Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 2, LPN2	GJA9	0	Clear
✓ Laryngeal Paralysis - Miniature Bull Terrier Variant	RAPGEF6	0	Clear

Sensory Neuropathy

	Gene	Copies	Results
✓ Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS - Spaniel and Pointer Variant	GDNF-AS	0	Clear
✓ Sensory Neuropathy - Border Collie Variant	FAM134B	0	Clear

Neuromuscular (7)**Junctionopathy**

	Gene	Copies	Results
✓ Congenital Myasthenic Syndrome, CMS - Golden Retriever Variant	COLQ Exon 13	0	Clear
✓ Congenital Myasthenic Syndrome, CMS - Heideterrier Variant	CHRNE	0	Clear
✓ Congenital Myasthenic Syndrome, CMS - Jack Russell Terrier Variant	CHRNE Exon 7	0	Clear
✓ Congenital Myasthenic Syndrome, CMS - Labrador Retriever Variant	COLQ Exon 14	0	Clear
✓ Congenital Myasthenic Syndrome, CMS - Old Danish Pointing Dog Variant	CHAT Exon 6	0	Clear

Movement Disorder

	Gene	Copies	Results
✓ Episodic Falling Syndrome - Cavalier King Charles Spaniel Variant	BCAN Exons 1-4	0	Clear
✓ Paroxysmal Dyskinesia, PxD - Soft Coated Wheaten Terrier Variant	PIGN	0	Clear

Ophthalmologic (31)**Glaucoma**

	Gene	Copies	Results
✓ Goniodysgenesis and Glaucoma, Pectinate Ligament Dysplasia, PLD - Border Collie Variant	OLFML3	0	Clear
✓ Primary Open Angle Glaucoma and Primary Lens Luxation - Chinese Shar-Pei Variant	ADAMTS17	0	Clear
✓ Primary Open Angle Glaucoma - Basset Fauve de Bretagne Variant	ADAMTS17	0	Clear
✓ Primary Open Angle Glaucoma - Beagle Variant	ADAMTS10	0	Clear
✓ Primary Open Angle Glaucoma - Norwegian Elkhound Variant	ADAMTS10	0	Clear

Iris or Lens

	Gene	Copies	Results
✓ Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts - Australian Shepherd Variant	HSF4	0	Clear
✓ Primary Lens Luxation	ADAMTS17	0	Clear

Retinopathy

	Gene	Copies	Results
✓ Achromatopsia - German Shepherd Variant	CNGA3 Exon 7	0	Clear
✓ Achromatopsia - Labrador Retriever Variant	CNGA3 Exon 7	0	Clear
✓ Autosomal Dominant Progressive Retinal Atrophy - English Mastiff and Bullmastiff Variant	RHO Exon 1	0	Clear
✓ Canine Multifocal Retinopathy, cmr1	BEST1/VMD2 Exon 2	0	Clear

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	Gene	Copies	Results
✓ Canine Multifocal Retinopathy, cmr2 - Coton de Tulear Variant	BEST1/VMD2 Exon 5	0	Clear
✓ Canine Multifocal Retinopathy, cmr3 - Finnish and Swedish Lapphund, Lapponian Herder Variant	BEST1/VMD2 Exon 10	0	Clear
⚠ Collie Eye Anomaly, Choroidal Hypoplasia, CEA	NHEJ1 Intron 4	2	At risk
✓ Congenital Stationary Night Blindness - Beagle Variant	LRIT3	0	Clear
✓ Congenital Stationary Night Blindness - Briard Variant	RPE65	0	Clear
✓ Day Blindness, Cone Degeneration, Achromatopsia - Alaskan Malamute Variant	CNGB3 Deletion	0	Clear
✓ Day Blindness, Cone Degeneration, Achromatopsia - German Shorthaired Pointer Variant	CNGB3 Exon 6	0	Clear
✓ Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1	SLC4A3 Exon 16	0	Clear
✓ Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2	TTC8 Exon 8	0	Clear
✓ Macular Corneal Dystrophy, MCD - Labrador Retriever Variant	CHST6	0	Clear
✓ Progressive Retinal Atrophy, CNGA - Shetland Sheepdog Variant	CNGA1 Exon 9	0	Clear
✓ Progressive Retinal Atrophy, Cone-Rod Dystrophy 1, crd1 - American Staffordshire Terrier Variant	PDE6B	0	Clear
✓ Progressive Retinal Atrophy, Cone-Rod Dystrophy 4, crd4/crd1	RPGRIP1 Exon 2	0	Clear
✓ Progressive Retinal Atrophy, PRA1 - Papillon Variant	CNGB1	0	Clear
✓ Progressive Retinal Atrophy, PRA3 - Tibetan Spaniel and Terrier Variant	FAM161A	0	Clear
✓ Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration, prcd	PRCD Exon 1	0	Clear
✓ Progressive Retinal Atrophy, Rod-Cone Dysplasia 1, rcd1 - Irish Setter Variant	PDE6B Exon 21	0	Clear
✓ Progressive Retinal Atrophy, Rod-Cone Dysplasia 3, rcd3 - Corgi Variant	PDE6A	0	Clear

	Gene	Copies	Results
✓ Progressive Retinal Atrophy - Basenji Variant	SAG	0	Clear
✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 - Samoyed and Husky Variant	RPGR Exon 15	0	Clear

Oral Cavity (4)

Developmental Disorder

	Gene	Copies	Results
✓ Cleft Lip and/or Cleft Palate - Nova Scotia Duck Tolling Retriever Variant	ADAMTS20	0	Clear

Tooth Structure Defect

	Gene	Copies	Results
✓ Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia - Italian Greyhound Variant	ENAM	0	Clear
✓ Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia - Parson Russell Terrier Variant	ENAM	0	Clear
✓ Raine Syndrome, Canine Dental Hypomineralization Syndrome - Border Collie Variant	FAM20C	0	Clear

Personalized Medicine (3)

	Gene	Copies	Results
✓ Alanine Aminotransferase Activity	GPT	0	Clear
✓ MDR1 Drug Sensitivity	ABCB1	0	Clear
✓ Malignant Hyperthermia	RYR1	0	Clear

Pulmonary (4)

	Gene	Copies	Results
✓ Neonatal Interstitial Lung Disease - Airedale Terrier Variant	LAMP3	0	Clear

	Gene	Copies	Results
✔ Primary Ciliary Dyskinesia, PCD - Alaskan Malamute Variant	NME5	0	Clear
✔ Primary Ciliary Dyskinesia, PCD - Old English Sheepdog Variant	CCDC39	0	Clear
✔ Recurrent Inflammatory Pulmonary Disease, RIPD - Rough Collie Variant	AKNA	0	Clear

Skeletal (10)

Chondrodystrophy

	Gene	Copies	Results
✔ Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD - Retrogene	FGF4 - chr12	0	Clear
✔ Chondrodystrophy - Norwegian Elkhound and Karelian Bear Dog Variant	ITGA10	0	Clear
✔ Oculoskeletal Dysplasia 2, Dwarfism-Retinal Dysplasia 2, drd2, OSD2 - Samoyed Variant	COL9A2 5' UTR	0	Clear
✔ Osteochondrodysplasia, Skeletal Dwarfism - Miniature Poodle Variant	SLC13A1	0	Clear
✔ Skeletal Dysplasia 2, SD2 - Labrador Retriever Variant	COL11A2	0	Clear

Decreased Bone Strength

	Gene	Copies	Results
✔ Hereditary Vitamin D-Resistant Rickets - Pomeranian Variant	VDR Exon 4	0	Clear
✔ Osteogenesis Imperfecta, Brittle Bone Disease - Beagle Variant	COL1A2	0	Clear
✔ Osteogenesis Imperfecta, Brittle Bone Disease - Dachshund Variant	SERPINH1 Exon 5	0	Clear
✔ Osteogenesis Imperfecta, Brittle Bone Disease - Golden Retriever Variant	COL1A1 Exon 18	0	Clear

Other

	Gene	Copies	Results
✔ Craniomandibular Osteopathy, CMO - Terrier and Australian Shepherd Variant	SLC37A2 Exon 15	0	Clear

Urogenital (14)

Nephropathy

	Gene	Copies	Results
✓ Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN - Cocker Spaniel Variant	COL4A4 Exon 3	0	Clear
✓ Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN - English Springer Spaniel Variant	COL4A4 Exon 30	0	Clear
✓ Fanconi Syndrome - Basenji Variant	FAN1	0	Clear
✓ Polycystic Kidney Disease, PKD - Bull Terrier Variant	PKD1 Exon 29	0	Clear
✓ Protein Losing Nephropathy, PLN - Soft Coated Wheaten and Airedale Terrier Variant	NPHS1	0	Clear
✓ X-Linked Hereditary Nephropathy, XLHN - Samoyed Variant 2	COL4A5 Exon 35	0	Clear

Urolithiasis

	Gene	Copies	Results
✓ 2,8-Dihydroxyadenine Urolithiasis, 2,8-DHA Urolithiasis - American Indian Dog Variant	APRT Exon 3	0	Clear
✓ Cystinuria Type I-A - Newfoundland Variant	SLC3A1 Exon 2	0	Clear
✓ Cystinuria Type II-A - Australian Cattle Dog Variant	SLC3A1 Exon 6	0	Clear
✓ Cystinuria Type II-B - Miniature Pinscher Variant	SLC7A9 Exon 9	0	Clear
✓ Hyperuricosuria and Hyperuricemia or Urolithiasis, HUU	SLC2A9 Exon 5	0	Clear
✓ Primary Hyperoxaluria - Coton de Tulear Variant	AGXT Exon 2	0	Clear

Other

	Gene	Copies	Results
✓ Persistent Mullerian Duct Syndrome, PMDS - Miniature and Standard Schnauzer Variant	AMHR2	0	Clear
✓ Renal Cystadenocarcinoma and Nodular Dermatofibrosis, RCND - German Shepherd Dog Variant	FLCN	0	Clear

Glossary

Key Terms

Increased Risk Result

The dog is at risk for showing clinical signs (phenotype) of a given condition. For recessive conditions, this means a dog has inherited two copies of an associated variant. For dominant, codominant, and additive conditions, this means a dog has inherited at least one copy of the variant. X-linked conditions will vary based on sex of the dog.

A dog's breed(s) and genetic background are also considered in this assessment. Genetic testing is an assessment of risk and not a clinical diagnosis, and not all dogs in this category will develop clinical signs.

Notable Result

A result may be notable for several reasons. The variant may not induce a disease state but rather inform patient care (this may include the tests listed under Personalized Medicine). The dog may have only one copy of a variant with a recessive mode of inheritance (meaning the dog is a carrier and is not expected to show the phenotype associated with the variant). The impact of the variant may also be influenced by a dog's breed(s). Based on the available research within the breed or related breeds, you will see more specific text within the results.

Clear Result

A dog with two healthy copies of a gene sequence is not at risk for developing the associated disease due to that variant. Many diseases can manifest as a result of other unknown genetic variants and/or environmental factors.

Variant

An alteration in the DNA with the potential to cause a change in phenotype (i.e. disease). A report may state that the dog has zero, one, or two copies of the variant for which we test. The term "variant" may be used interchangeably with "mutation."

Genotype

The genetic code related to the variant being present or absent in the dog's DNA.

Phenotype

The physical impact or appearance directed by the genotype. The phenotype is often described as an expression of the genotype.

Complex Phenotype

The condition, appearance, or other physical expression of the genotype controlled by both genetic and environmental factors.

Penetrance

Proportion of dogs with a particular genotype that expresses the associated phenotype. There are two types of penetrance.

1. Incomplete penetrance means that not all dogs with the genotype will develop the clinical signs of the phenotype.
2. Complete penetrance means that all dogs with the genotype will develop the clinical signs of the phenotype.

Carrier

This term has traditionally been used to describe a dog that has one copy of the variant but is not expected to show the phenotype associated with the variant (this is applicable to variants with a recessive mode of inheritance (MOI) as described below). If used in a breeding pair, a carrier may pass the variant to its litter.

At-risk

This indicates that the dog may manifest the disease and generally is used when a dog has two copies of the variant (but this depends on the MOI).

Embark uses the term "at-risk" and not "affected" because genetic testing is an assessment of risk and not a clinical diagnosis.

Linkage Disequilibrium Test

When a causal variant cannot be identified or when the variant is incompatible with the genotyping platform constraints, allelic association or linkage disequilibrium (LD) tests can be utilized. This is typically done to assist dog breeders in selectively breeding out a deleterious condition. LD tests are based on a statistical association between two loci that are physically very close in the DNA. The coupling of the chosen proxy marker to the causal variant is known mathematically for the most relevant populations.

LD-based tests have a slightly increased incidence of false positives and false negatives, which are test-specific and known. Embark offers limited numbers of these tests. Embark continuously works to refine LD-based tests by assaying the direct variant in a subset of dogs using alternative methods. These inputs help to refine the tests over time.

Provisional Result

Embark combines random sampling and sequencing with the use of blinded controls to confirm that each test is performing to standard at >99% genotyping accuracy and reproducibility. Our standard health tests have been validated using known heterozygous and homozygous samples to ensure design accuracy and use multiple probes per condition to ensure reproducibility. Provisional tests are for rare disorders for which DNA samples from carrier and/or at-risk individuals are not available for calculating test reliability, or for structural variants where more testing is needed to ensure the same level of accuracy.

If you have access to DNA from carrier or at-risk individuals and are interested in helping us validate a test, please contact us at vetsupport@embarkvet.com

Modes of Inheritance

Recessive

A dog is thought to need two copies of a variant to be considered at-risk for the clinical disease or to have the visible phenotype for traits. This may apply to autosomal or X-linked variants, however. Read below for additional details regarding X-linked variants.

Dominant

A dog is thought to need only one copy of the variant to be considered at-risk for the clinical disease or to have the visible phenotype for traits.

Codominant/Additive

In general, these terms are used to describe variants in which dogs with one copy of the variant have a different phenotype compared to dogs with zero or two copies of the variant (although there is a slight difference between the two terms).

X-linked

The variant resides on the X chromosome, and male dogs need just one copy of the variant to be considered at-risk. For recessively inherited X-linked conditions, female dogs typically require two copies of the variant to be considered at-risk. Female dogs who have one copy of a recessively inherited X-linked variant are often referred to as carriers, but they can exhibit signs of disease that range from clinically asymptomatic to fully affected. This is due to a normal phenomenon known as X-chromosome inactivation, where one X chromosome is silenced in each cell.

Weight

The Embark DNA test provides a genetic size based not just on breed ancestry but on over a dozen genes known to influence a dog's weight, as well as sex and breed-specific modifiers.

Our algorithm explains over 85% of the variance in healthy adult weight. However, due to a few as-yet-undiscovered genes and genetic interactions that affect size, this algorithm sometimes under or over-predicts weight.

Genetic Age

Dogs age at very different rates due to a number of genetic and environmental factors. Embark's genetic age calculates how old a dog would be if he or she were aging at an average human rate (using humans in the USA as the baseline). This measure is more personalized than "one dog year = seven human years".

[View the patient's profile see the personalized genetic age table for this dog.](#)

We start by asking the dog's approximate calendar age. We then calculate genetic age by factoring a dog's breed composition along with information from genes that affect size, sex, and the dog's inbreeding coefficient (COI).

Impact of Breed

When determining whether or not a variant is expected to have a clinical impact for a breed, we have taken into account research either published, internal, or otherwise presented by a subject matter authority as our primary criteria. So, while a dog may have the variant associated with a disease (one or two copies for dominant variants and two copies for autosomal recessive variants), he or she may not be known to be at significant clinical risk from that variant.

Based on the available research within the breed or highly related breeds, you may see text similar to the following options:

1. This genetic variant is not likely to significantly increase the risk that this dog will develop the clinical disease.
2. This genetic variant is associated with an increased risk that this dog will develop the clinical disease.
3. We do not know whether this variant increases the risk that this dog will develop the clinical disease.

Embark is continuing to explore the relationship of genotype to phenotype, and risk assessment may be updated as more data is reviewed. You can contact vetsupport@embarkvet.com or call 1-855-203-8271 to report any clinical diagnoses.