



GIZMO



DNA Test Report

Test Date: April 18th, 2019

embk.me/gizmo93

HEALTH

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

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AT RISK

1

CARRIER

CARRIER CONDITIONS

CARRIER status: This indicates the dog has inherited a recessive allele for a genetic trait or mutation. This is not enough to cause symptoms of the disease, but is important to bear in mind if the dog ever has offspring.

 **Carrier**

System: **Neurologic**

Condition: **Degenerative Myelopathy, DM (SOD1A)**



DEGENERATIVE MYELOPATHY, DM

(SOD1A)

Carrier

**SOD1**

GENE NAME

GG

CLEAR

GA

CARRIER

AA

AT RISK

Gizmo is a carrier for a variant in the SOD1 gene. While he or she is unlikely to exhibit signs of disease, as a carrier, he or she will pass the variant on to the next generation. If you choose to breed Gizmo, we highly recommend testing any potential mates for this variant. Breeding to another carrier is not recommended as this will produce a number of affected puppies.

DESCRIPTION

A disease of mature dogs, this is a progressive degenerative disorder of the spinal cord that can cause muscle wasting and gait abnormalities. Affected dogs do not usually show signs until they are at least 8 years old, where the first signs of neural degeneration appear in the nerves that innervate the hind limbs. You may notice your dog scuffing the tops of his or her hind paws, or walking with a hesitant, exaggerated gait. In advanced cases, lower motor neurons are also affected leading to weakness or near-paralysis of all four legs and widespread muscle wasting. Given the advanced age at the time of onset, the treatment for DM is aimed towards making your dog comfortable in his or her old age and includes lifestyle changes and physical therapy. SOD1 codes superoxide dismutase, an enzyme important in neutralizing free radicals and reactive oxygen species, both of which are produced as a byproduct of cell metabolism. If not neutralized, these are injurious to the cell and will cause premature cell death. The first system to show effects of this is the nervous system given the highly specialized and delicate nature of these cells. Please note that these mutations are reported to have incomplete penetrance: that is, while a dog with two copies of this mutation has a much greater chance of developing DM than a dog with one copy of the mutation, or none at all, other genetic and environmental factors will also contribute to whether your dog develops DM.

More information

To learn more about this condition, you can visit <http://www.caninegeneticdiseases.net/DM/basicDM.htm> (<http://www.caninegeneticdiseases.net/DM/basicDM.htm>).

CITATIONS

Awano et al 2009 (<http://www.ncbi.nlm.nih.gov/pubmed/19188595>), Shelton et al 2012 (<http://www.ncbi.nlm.nih.gov/pubmed/22542607>), Capuccio et al 2014 (<http://www.ncbi.nlm.nih.gov/pubmed/24390315>)



OTHER CONDITIONS

Good news! Gizmo tested clear for 14 genetic conditions that are common in his breed mix.

- Von Willebrand Disease Type I (VWF)
- Progressive Retinal Atrophy, prcd
Progressive rod-cone degeneration (PRCD Exon 1)
- Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2 (TTC8)
- GM2 Gangliosidosis (HEXB, Poodle Variant)
- Muscular Dystrophy
Muscular Dystrophy (DMD Golden Retriever Variant)
- Ichthyosis (PNPLA1)
- Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1)
- Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cavalier King Charles Spaniel Variant)
- Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLC4A3)
- Neuronal Ceroid Lipofuscinosis (CLN5 Golden Retriever Variant)
- Neonatal Encephalopathy with Seizures, NEWS (ATF2)
- Dystrophic Epidermolysis Bullosa (COL7A1)
- Osteogenesis Imperfecta, Brittle Bone Disease (COL1A1)
- Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (FGF4 retrogene - CFA12)



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FULL TEST PANEL

Gizmo 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 Gizmo tested clear for.



CLEAR CONDITIONS

- 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 III, Type III vWD (VWF Exon 4) (Chromosome 27)
- Von Willebrand Disease Type II, Type II vWD (VWF) (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, LM (PLG) (Chromosome 1)
- Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant) (Chromosome 17)
- Complement 3 Deficiency, 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, 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)
- Progressive Retinal Atrophy, crd1 (PDE6B) (Chromosome 3)
- Progressive Retinal Atrophy, crd2 (IQCB1) (Chromosome 33)



CLEAR CONDITIONS

- Progressive Retinal Atrophy - crd4/cord1 (RPGRIP1) (Chromosome 15)
- Collie Eye Anomaly, Choroidal Hypoplasia, CEA (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)
- Macular Corneal Dystrophy, MCD (CHST6) (Chromosome 5)
- 2,8-Dihydroxyadenine Urolithiasis, 2,8-DHA Urolithiasis (APRT) (Chromosome 5)
- Cystinuria Type I-A (SLC3A1) (Chromosome 10)
- Cystinuria Type II-A (SLC3A1) (Chromosome 10)
- Cystinuria Type II-B (SLC7A9) (Chromosome 1)
- Hyperuricosuria and Hyperuricemia or Urolithiasis, HUU (SLC2A9) (Chromosome 3)
- Polycystic Kidney Disease, PKD (PKD1) (Chromosome 6)
- Primary Hyperoxaluria (AGXT) (Chromosome 25)
- Protein Losing Nephropathy, PLN (NPHS1) (Chromosome 1)
- X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2) (Chromosome X)
- Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN (COL4A4 Exon 3) (Chromosome 25)
- Primary Ciliary Dyskinesia, PCD (CCDC39 Exon 3) (Chromosome 34)
- Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, Dry Eye Curly Coat Syndrome, CKCSID (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, GSD II (GAA) (Chromosome 9)
- Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA (G6PC) (Chromosome 9)
- Glycogen Storage Disease Type IIIA, GSD IIIA (AGL) (Chromosome 6)
- Mucopolysaccharidosis Type I, MPS I (IDUA) (Chromosome 3)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 1) (Chromosome 9)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 2) (Chromosome 9)



CLEAR CONDITIONS

- Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 5) (Chromosome 6)
- Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 3) (Chromosome 6)
- Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Whippet and English Springer Spaniel Variant) (Chromosome 27)
- Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Wachtelhund Variant) (Chromosome 27)
- Lagotto Storage Disease (ATG4D) (Chromosome 20)
- Neuronal Ceroid Lipofuscinosis 1, NCL 1 (PPT1 Exon 8) (Chromosome 15)
- Neuronal Ceroid Lipofuscinosis 2, NCL 2 (TPP1 Exon 4) (Chromosome 21)
- Neuronal Ceroid Lipofuscinosis 1, Cerebellar Ataxia, NCL-A (ARSG Exon 2) (Chromosome 9)
- Neuronal Ceroid Lipofuscinosis 1, NCL 1 (CLN5 Border Collie Variant) (Chromosome 22)
- Neuronal Ceroid Lipofuscinosis 6, NCL 6 (CLN6 Exon 7) (Chromosome 30)
- Neuronal Ceroid Lipofuscinosis 8, NCL 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, NCL 10 (CTSD Exon 5) (Chromosome 18)
- 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, Familial Enamel Hypoplasia (Italian Greyhound Variant) (Chromosome 13)
- Persistent Mullerian Duct Syndrome, PMDS (AMHR2) (Chromosome 27)
- Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS (MYO7A) (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, NCCD (SPTBN2) (Chromosome 18)
- Cerebellar Ataxia, Progressive Early-Onset Cerebellar Ataxia (SEL1L) (Chromosome 8)
- Cerebellar Hypoplasia (VLDLR) (Chromosome 1)
- Spinocerebellar Ataxia, Late-Onset Ataxia, LoSCA (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, L2HGA (L2HGDH) (Chromosome 0)
- Polyneuropathy, NDRG1 Greyhound Variant (NDRG1 Exon 15) (Chromosome 13)
- Polyneuropathy, NDRG1 Malamute Variant (NDRG1 Exon 4) (Chromosome 13)



CLEAR CONDITIONS

- Narcolepsy (HCRTR2 Intron 6) (Chromosome 12)
- Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 15) (Chromosome 1)
- Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (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, Acral Mutilation Syndrome, AMS (GDNF-AS) (Chromosome 4)
- Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1, LPN1 (LPN1, ARHGEF10) (Chromosome 16)
- Spongy Degeneration with Cerebellar Ataxia 1, SDCA1, SeSAME/EAST Syndrome (KCNJ10) (Chromosome 38)
- Spongy Degeneration with Cerebellar Ataxia 2, SDCA2 (ATP1B2) (Chromosome 5)
- Dilated Cardiomyopathy, DCM1 (PDK4) (Chromosome 14)
- Dilated Cardiomyopathy, DCM2 (TTN) (Chromosome 36)
- 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)
- 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, XL-MTM (MTM1, Labrador Variant) (Chromosome X)
- Hypocatalasia, Acatlasemia (CAT) (Chromosome 18)
- Pyruvate Dehydrogenase Deficiency (PDP1) (Chromosome 29)
- Malignant Hyperthermia (RYR1) (Chromosome 1)
- Imlerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 53) (Chromosome 2)
- Imlerslund-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)
- Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1) (Chromosome 7)
- Ichthyosis, Epidermolytic Hyperkeratosis (KRT10) (Chromosome 9)
- 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, OSD1 (COL9A3, Labrador Retriever) (Chromosome 24)
- Osteogenesis Imperfecta, Brittle Bone Disease (COL1A2) (Chromosome 14)
- Osteogenesis Imperfecta, Brittle Bone Disease (SERPINH1) (Chromosome 21)
- Skeletal Dysplasia 2, SD2 (COL11A2) (Chromosome 12)
- Craniomandibular Osteopathy, CMO (SLC37A2) (Chromosome 5)