



# "KENZIE"

## LAKESHORE'S EXQUISITE EXAMPLE



DNA Test Report

Test Date: April 12th, 2019

[embk.me/lakeshoresexquisiteexample](http://embk.me/lakeshoresexquisiteexample)

### BREED-RELEVANT CONDITIONS TESTED



**Kenzie did not have the variants that we tested for, that are relevant to her breed:**

- ✓ Von Willebrand Disease Type I (VWF)
- ✓ Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cavalier King Charles Spaniel Variant)
- ✓ Progressive Retinal Atrophy, prcd (PRCD Exon 1)
- ✓ GM2 Gangliosidosis (HEXB, Poodle Variant)
- ✓ Degenerative Myelopathy, DM (SOD1A)
- ✓ Neonatal Encephalopathy with Seizures, NEWS (ATF2)
- ✓ Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1)
- ✓ Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (FGF4 retrogene - CFA12)



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### ADDITIONAL CONDITIONS TESTED



**Kenzie did not have the variants that we tested for, in the following conditions that the potential effect on dogs with Kenzie's breed may not yet be known.**

- ✓ MDR1 Drug Sensitivity (MDR1)
- ✓ P2Y12 Receptor Platelet Disorder (P2RY12)
- ✓ Factor IX Deficiency, Hemophilia B (F9 Exon 7, Terrier Variant)
- ✓ Factor IX Deficiency, Hemophilia B (F9 Exon 7, Rhodesian Ridgeback Variant)
- ✓ Factor VII Deficiency (F7 Exon 5)
- ✓ Factor VIII Deficiency, Hemophilia A (F8 Exon 10, Boxer Variant)
- ✓ Factor VIII Deficiency, Hemophilia A (F8 Exon 11, Shepherd Variant 1)
- ✓ Factor VIII Deficiency, Hemophilia A (F8 Exon 1, Shepherd Variant 2)
- ✓ Thrombopathia (RASGRP2 Exon 5, Basset Hound Variant)
- ✓ Thrombopathia (RASGRP2 Exon 8)
- ✓ Thrombopathia (RASGRP2 Exon 5, American Eskimo Dog Variant)
- ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4)
- ✓ Von Willebrand Disease Type II, Type II vWD (VWF)
- ✓ Canine Leukocyte Adhesion Deficiency Type III, CLAD3 (FERMT3)
- ✓ Canine Elliptocytosis (SPTB Exon 30)
- ✓ Glanzmann's Thrombasthenia Type I (ITGA2B Exon 12)
- ✓ May-Hegglin Anomaly (MYH9)
- ✓ Prekallikrein Deficiency (KLKB1 Exon 8)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 5)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 7 Labrador Variant)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 7 Pug Variant)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 7 Beagle Variant)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 10)
- ✓ Trapped Neutrophil Syndrome (VPS13B)



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- ✔ Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant)
- ✔ Complement 3 Deficiency, C3 Deficiency (C3)
- ✔ Severe Combined Immunodeficiency (PRKDC)
- ✔ Severe Combined Immunodeficiency (RAG1)
- ✔ X-linked Severe Combined Immunodeficiency (IL2RG Variant 1)
- ✔ X-linked Severe Combined Immunodeficiency (IL2RG Variant 2)
- ✔ Progressive Retinal Atrophy, rcd1 (PDE6B Exon 21 Irish Setter Variant)
- ✔ Progressive Retinal Atrophy, rcd3 (PDE6A)
- ✔ Progressive Retinal Atrophy, CNGA (CNGA1 Exon 9)
- ✔ Progressive Retinal Atrophy (CNGB1)
- ✔ Progressive Retinal Atrophy (SAG)
- ✔ Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLC4A3)
- ✔ Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2 (TTC8)
- ✔ Progressive Retinal Atrophy, crd1 (PDE6B)
- ✔ Progressive Retinal Atrophy, crd2 (IQCB1)
- ✔ Progressive Retinal Atrophy - crd4/cord1 (RPGRIP1)
- ✔ Collie Eye Anomaly, Choroidal Hypoplasia, CEA (NHEJ1)
- ✔ Achromatopsia (CNGA3 Exon 7 German Shepherd Variant)
- ✔ Achromatopsia (CNGA3 Exon 7 Labrador Retriever Variant)
- ✔ Autosomal Dominant Progressive Retinal Atrophy (RHO)
- ✔ Canine Multifocal Retinopathy (BEST1 Exon 2)
- ✔ Canine Multifocal Retinopathy (BEST1 Exon 5)
- ✔ Canine Multifocal Retinopathy (BEST1 Exon 10 Deletion)
- ✔ Canine Multifocal Retinopathy (BEST1 Exon 10 SNP)
- ✔ Glaucoma (ADAMTS10 Exon 9)



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- ✓ Glaucoma (ADAMTS10 Exon 17)
- ✓ Glaucoma (ADAMTS17 Exon 11)
- ✓ Glaucoma (ADAMTS17 Exon 2)
- ✓ Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9 Shepherd Variant)
- ✓ Primary Lens Luxation (ADAMTS17)
- ✓ Congenital Stationary Night Blindness (RPE65)
- ✓ Macular Corneal Dystrophy, MCD (CHST6)
- ✓ 2,8-Dihydroxyadenine Urolithiasis, 2,8-DHA Urolithiasis (APRT)
- ✓ Cystinuria Type I-A (SLC3A1)
- ✓ Cystinuria Type II-A (SLC3A1)
- ✓ Cystinuria Type II-B (SLC7A9)
- ✓ Hyperuricosuria and Hyperuricemia or Urolithiasis, HUU (SLC2A9)
- ✓ Polycystic Kidney Disease, PKD (PKD1)
- ✓ Primary Hyperoxaluria (AGXT)
- ✓ Protein Losing Nephropathy, PLN (NPHS1)
- ✓ X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)
- ✓ Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN (COL4A4 Exon 3)
- ✓ Primary Ciliary Dyskinesia, PCD (CCDC39 Exon 3)
- ✓ Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, Dry Eye Curly Coat Syndrome, CKCSID (FAM83H Exon 5)
- ✓ X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia (EDA Intron 8)
- ✓ Renal Cystadenocarcinoma and Nodular Dermatofibrosis, RCND (FLCN Exon 7)
- ✓ Canine Fucosidosis (FUCA1)
- ✓ Glycogen Storage Disease Type II, Pompe's Disease, GSD II (GAA)
- ✓ Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA (G6PC)
- ✓ Glycogen Storage Disease Type IIIA, GSD IIIA (AGL)

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## ADDITIONAL CONDITIONS TESTED

- ✔ Mucopolysaccharidosis Type I, MPS I (IDUA)
- ✔ Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 1)
- ✔ Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 2)
- ✔ Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 5)
- ✔ Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 3)
- ✔ Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Whippet and English Springer Spaniel Variant)
- ✔ Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Wachtelhund Variant)
- ✔ Lagotto Storage Disease (ATG4D)
- ✔ Neuronal Ceroid Lipofuscinosis 1, NCL 1 (PPT1 Exon 8)
- ✔ Neuronal Ceroid Lipofuscinosis 2, NCL 2 (TPP1 Exon 4)
- ✔ Neuronal Ceroid Lipofuscinosis 1, Cerebellar Ataxia, NCL4A (ARSG Exon 2)
- ✔ Neuronal Ceroid Lipofuscinosis 1, NCL 5 (CLN5 Border Collie Variant)
- ✔ Neuronal Ceroid Lipofuscinosis 6, NCL 6 (CLN6 Exon 7)
- ✔ Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 English Setter Variant)
- ✔ Neuronal Ceroid Lipofuscinosis (MFSD8)
- ✔ Neuronal Ceroid Lipofuscinosis (CLN8 Australian Shepherd Variant)
- ✔ Neuronal Ceroid Lipofuscinosis 10, NCL 10 (CTSD Exon 5)
- ✔ Neuronal Ceroid Lipofuscinosis (CLN5 Golden Retriever Variant)
- ✔ Adult-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2, Tibetan Terrier Variant)
- ✔ GM1 Gangliosidosis (GLB1 Exon 15 Shiba Inu Variant)
- ✔ GM1 Gangliosidosis (GLB1 Exon 15 Alaskan Husky Variant)
- ✔ GM1 Gangliosidosis (GLB1 Exon 2)
- ✔ GM2 Gangliosidosis (HEXA)
- ✔ Globoid Cell Leukodystrophy, Krabbe disease (GALC Exon 5)
- ✔ Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (Italian Greyhound Variant)

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### ADDITIONAL CONDITIONS TESTED

- ✓ Persistent Mullerian Duct Syndrome, PMDS (AMHR2)
- ✓ Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS (MY07A)
- ✓ Shar-Pei Autoinflammatory Disease, SPAID, Shar-Pei Fever (MTBP)
- ✓ Alaskan Husky Encephalopathy, Subacute Necrotizing Encephalomyelopathy (SLC19A3)
- ✓ Alexander Disease (GFAP)
- ✓ Cerebellar Abiotrophy, Neonatal Cerebellar Cortical Degeneration, NCCD (SPTBN2)
- ✓ Cerebellar Ataxia, Progressive Early-Onset Cerebellar Ataxia (SEL1L)
- ✓ Cerebellar Hypoplasia (VLDLR)
- ✓ Spinocerebellar Ataxia, Late-Onset Ataxia, LoSCA (CAPN1)
- ✓ Spinocerebellar Ataxia with Myokymia and/or Seizures (KCNJ10)
- ✓ Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy (LG12)
- ✓ Fetal-Onset Neonatal Neuroaxonal Dystrophy (MFN2)
- ✓ Hypomyelination and Tremors (FNIP2)
- ✓ Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome (PLP)
- ✓ Neuroaxonal Dystrophy, NAD (Spanish Water Dog Variant)
- ✓ L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH)
- ✓ Polyneuropathy, NDRG1 Greyhound Variant (NDRG1 Exon 15)
- ✓ Polyneuropathy, NDRG1 Malamute Variant (NDRG1 Exon 4)
- ✓ Narcolepsy (HCRTR2 Intron 6)
- ✓ Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 15)
- ✓ Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 4)
- ✓ Juvenile Laryngeal Paralysis and Polyneuropathy, Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation, POANV (RAB3GAP1, Rottweiler Variant)
- ✓ Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS (GDNF-AS)
- ✓ Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1, LPN1 (LPN1, ARHGEF10)
- ✓ Spongy Degeneration with Cerebellar Ataxia 1, SDCA1, SeSAME/EAST Syndrome (KCNJ10)

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## ADDITIONAL CONDITIONS TESTED

- ✔ Spongy Degeneration with Cerebellar Ataxia 2, SDCA2 (ATP1B2)
- ✔ Dilated Cardiomyopathy, DCM1 (PDK4)
- ✔ Dilated Cardiomyopathy, DCM2 (TTN)
- ✔ Long QT Syndrome (KCNQ1)
- ✔ Muscular Dystrophy (DMD, Cavalier King Charles Spaniel Variant 1)
- ✔ Muscular Dystrophy (DMD Pembroke Welsh Corgi Variant )
- ✔ Muscular Dystrophy (DMD Golden Retriever Variant)
- ✔ Centronuclear Myopathy (PTPLA)
- ✔ Exercise-Induced Collapse (DNM1)
- ✔ Inherited Myopathy of Great Danes (BIN1)
- ✔ Myostatin Deficiency, Bully Whippet Syndrome (MSTN)
- ✔ Myotonia Congenita (CLCN1 Exon 7)
- ✔ Myotonia Congenita (CLCN1 Exon 23)
- ✔ Myotubular Myopathy 1, X-linked Myotubular Myopathy, XL-MTM (MTM1, Labrador Variant)
- ✔ Hypocatalasia, Acatlasemia (CAT)
- ✔ Pyruvate Dehydrogenase Deficiency (PDP1)
- ✔ Malignant Hyperthermia (RYR1)
- ✔ Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 53)
- ✔ Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 8)
- ✔ Congenital Myasthenic Syndrome (CHAT)
- ✔ Congenital Myasthenic Syndrome (COLQ)
- ✔ Episodic Falling Syndrome (BCAN)
- ✔ Dystrophic Epidermolysis Bullosa (COL7A1)
- ✔ Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1)
- ✔ Ichthyosis, Epidermolytic Hyperkeratosis (KRT10)



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### ADDITIONAL CONDITIONS TESTED

- ✓ Ichthyosis (PNPLA1)
- ✓ Ichthyosis (SLC27A4)
- ✓ Ichthyosis (NIPAL4)
- ✓ Focal Non-Epidermolytic Palmoplantar Keratoderma, Pachyonychia Congenita (KRT16)
- ✓ Hereditary Footpad Hyperkeratosis (FAM83G)
- ✓ Hereditary Nasal Parakeratosis (SUV39H2)
- ✓ Musladin-Lueke Syndrome (ADAMTSL2)
- ✓ Cleft Lip and/or Cleft Palate (ADAMTS20)
- ✓ Hereditary Vitamin D-Resistant Rickets (VDR)
- ✓ Oculoskeletal Dysplasia 1, Dwarfism-Retinal Dysplasia, OSD1 (COL9A3, Labrador Retriever)
- ✓ Osteogenesis Imperfecta, Brittle Bone Disease (COL1A2)
- ✓ Osteogenesis Imperfecta, Brittle Bone Disease (SERPINH1)
- ✓ Osteogenesis Imperfecta, Brittle Bone Disease (COL1A1)
- ✓ Skeletal Dysplasia 2, SD2 (COL11A2)
- ✓ Craniomandibular Osteopathy, CMO (SLC37A2)
- ✓ Chondrodystrophy, Norwegian Elkhound and Karelian Bear Dog Variant (ITGA10)

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