

## BREED-RELEVANT CONDITIONS TESTED



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

- ✓ MDR1 Drug Sensitivity (MDR1)
- ✓ Trapped Neutrophil Syndrome (VPS13B)
- ✓ Collie Eye Anomaly, Choroidal Hypoplasia, CEA (NHEJ1)
- ✓ Primary Lens Luxation (ADAMTS17)
- ✓ Neuronal Ceroid Lipofuscinosis 1, NCL 5 (CLN5 Border Collie Variant)
- ✓ Myotonia Congenita (CLCN1 Exon 23)
- ✓ Imlerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 53)

## ADDITIONAL CONDITIONS TESTED



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

- ✓ P2Y12 Receptor Platelet Disorder (P2Y12)
- ✓ 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 (RASGRP1 Exon 5, Basset Hound Variant)
- ✓ Thrombopathia (RASGRP1 Exon 8)
- ✓ Thrombopathia (RASGRP1 Exon 5, American Eskimo Dog Variant)
- ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 4)
- ✓ Von Willebrand Disease Type III, Type III vWD (VWF Exon 7)
- ✓ Von Willebrand Disease Type I (VWF)
- ✓ Von Willebrand Disease Type II, Type II vWD (VWF)
- ✓ Canine Leukocyte Adhesion Deficiency Type I, CLADI (ITGB2)
- ✓ Canine Leukocyte Adhesion Deficiency Type III, CLADIII (FERMT3)
- ✓ Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cavalier King Charles Spaniel Variant)
- ✓ Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cairn and Norfolk Terrier Variant)
- ✓ 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)

## ADDITIONAL CONDITIONS TESTED

- ✓ Pyruvate Kinase Deficiency (PKLR Exon 7 Beagle Variant)
- ✓ Pyruvate Kinase Deficiency (PKLR Exon 10)
- ✓ Ligneous Membranitis, LM (PLG)
- ✓ Platelet factor X receptor deficiency, Scott Syndrome (TMEM16F)
- ✓ Methemoglobinemia CYB5R3
- ✓ Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant)
- ✓ Congenital Hypothyroidism (TPO, Rat, Toy, Hairless 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, prcd (PRCD Exon 1)
- ✓ 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 - crd4/cord1 (RPGRIP1)
- ✓ X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)
- ✓ Progressive Retinal Atrophy, PRA3 (FAM161A)
- ✓ Day blindness, Cone Degeneration, Achromatopsia (CNGB3 Exon 6)

## ADDITIONAL CONDITIONS TESTED

- ✓ 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)
- ✓ Glaucoma (ADAMTS10 Exon 17)
- ✓ Glaucoma (ADAMTS17 Exon 11)
- ✓ Glaucoma (ADAMTS17 Exon 2)
- ✓ Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9 Shepherd Variant)
- ✓ 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)

## ADDITIONAL CONDITIONS TESTED

- ✔ 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)
- ✔ 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 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)

## ADDITIONAL CONDITIONS TESTED

- ✓ Late-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2, Australian Cattle Dog Variant)
- ✓ GM1 Gangliosidosis (GLB1 Exon 15 Shiba Inu Variant)
- ✓ GM1 Gangliosidosis (GLB1 Exon 15 Alaskan Husky Variant)
- ✓ GM1 Gangliosidosis (GLB1 Exon 2)
- ✓ GM2 Gangliosidosis (HEXB, Poodle Variant)
- ✓ GM2 Gangliosidosis (HEXA)
- ✓ Globoid Cell Leukodystrophy, Krabbe disease (GALC Exon 5)
- ✓ Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (Italian Greyhound Variant)
- ✓ Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (Parson Russell Terrier Variant)
- ✓ Persistent Mullerian Duct Syndrome, PMDS (AMHR2)
- ✓ Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS (MYO7A)
- ✓ 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)
- ✓ Hereditary Ataxia (RAB24)
- ✓ Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy (LGI2)
- ✓ Degenerative Myelopathy, DM (SOD1A)
- ✓ Fetal-Onset Neonatal Neuroaxonal Dystrophy (MFN2)
- ✓ Hypomyelination and Tremors (FNIP2)
- ✓ Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome (PLP)

## ADDITIONAL CONDITIONS TESTED

- ✓ Neuroaxonal Dystrophy, NAD (Spanish Water Dog Variant)
- ✓ Neuroaxonal Dystrophy, NAD (Rottweiler Variant)
- ✓ L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH)
- ✓ Neonatal Encephalopathy with Seizures, NEWS (ATF2)
- ✓ 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)
- ✓ Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS (GDNF-AS)
- ✓ Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1, LPN1 (LPN1, ARHGEF10)
- ✓ Juvenile Myoclonic Epilepsy (DIRAS1)
- ✓ Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 2, LPN2 (GJA9)
- ✓ Spongy Degeneration with Cerebellar Ataxia 1, SDCA1, SeSAME/EAST Syndrome (KCNJ10)
- ✓ 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)
- ✓ Limb Girdle Muscular Dystrophy (SGCD, Boston Terrier Variant)
- ✓ Exercise-Induced Collapse (DNM1)
- ✓ Inherited Myopathy of Great Danes (BIN1)
- ✓ Myostatin Deficiency, Bully Whippet Syndrome (MSTN)

## ADDITIONAL CONDITIONS TESTED

- ✓ Myotonia Congenita (CLCN1 Exon 7)
- ✓ Myotubular Myopathy 1, X-linked Myotubular Myopathy, XL-MTM (MTM1, Labrador Variant)
- ✓ Hypocatalasia, Acatalasemia (CAT)
- ✓ Pyruvate Dehydrogenase Deficiency (PDP1)
- ✓ Malignant Hyperthermia (RYR1)
- ✓ Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 8)
- ✓ Lunde hund Syndrome (LEPREL1)
- ✓ Congenital Myasthenic Syndrome (CHAT)
- ✓ Congenital Myasthenic Syndrome (COLQ)
- ✓ Episodic Falling Syndrome (BCAN)
- ✓ Paroxysmal Dyskinesia, PxD (PGIN)
- ✓ Dystrophic Epidermolysis Bullosa (COL7A1)
- ✓ Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1)
- ✓ Ichthyosis, Epidermolytic Hyperkeratosis (KRT10)
- ✓ 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)
- ✓ Oculocutaneous Albinism, OCA2 (Pekingese Type)
- ✓ Bald Thigh Syndrome (IGFBP5)
- ✓ Cleft Lip and/or Cleft Palate (ADAMTS20)
- ✓ Hereditary Vitamin D-Resistant Rickets (VDR)



## ADDITIONAL CONDITIONS TESTED

- ✔ **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)**
- ✔ **Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1)**
- ✔ **Skeletal Dysplasia 2, SD2 (COL11A2)**
- ✔ **Cranio-mandibular Osteopathy, CMO (SLC37A2)**
- ✔ **Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (FGF4 retrogene - CFA12)**
- ✔ **Chondrodystrophy, Norwegian Elkhound and Karelian Bear Dog Variant (ITGA10)**