

Canine genetic diseases screened for by chip microarray, according to

[Donner et al. \(2018\).](#)

Alaskan Husky Encephalopathy (AHE)
Amelogenesis Imperfecta (AI)
Autosomal Recessive Severe Combined Immunodeficiency (ARSCID)
Bandera's Neonatal Ataxia (BNAt)
Benign Familial Juvenile Epilepsy or Remitting Focal Epilepsy (BFJE)
Bleeding disorder due to P2RY12 defect
Canine Cyclic Neutropenia (CN)
Canine Multifocal Retinopathy 2 (CMR2); mutation originally found in Coton de Tulear
Canine Multifocal Retinopathy 3 (CMR3); mutation originally found in Lapponian Herder
Cavalier King Charles Spaniel Muscular Dystrophy (CKCS-MD)
Centronuclear Myopathy (CNM); mutation originally found in Great Dane
Centronuclear Myopathy (CNM); mutation originally found in Labrador Retriever
Chondrodysplasia; mutation originally found in Norwegian Elkhound and Karelian Bear Dog
Cleft Palate / Cleft Lip and Palate with Syndactyly; DLX6 gene mutation originally found in Nova Sc
Collie Eye Anomaly (CEA)
Complement 3 (C3) Deficiency
Cone Degeneration (CD) or Achromatopsia
Cone Degeneration (CD) or Achromatopsia; mutation originally found in German Shorthaired Poin
Cone-Rod Dystrophy (cord1-PRA / crd4)
Cone-Rod Dystrophy 1 (crd1); mutation originally found in American Staffordshire Terrier
Cone-Rod Dystrophy 2 (crd2); mutation originally found in Pit Bull Terrier
Cone-Rod Dystrophy of Standard Wirehaired Dachshund (crd SWD)
Congenital Hypothyroidism; mutation originally found in Toy Fox- and Rat Terrier
Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis (CKCSID)
Congenital Myasthenic Syndrome (CMS); mutation originally found in Old Danish Pointing Dog
Congenital Stationary Night Blindness (CSNB)
Cranio-mandibular Osteopathy (CMO)
Cystinuria Type I-A; mutation originally found in Newfoundland Dog
Cystinuria Type II-A; mutation originally found in Australian Cattle Dog
Cystinuria Type II-B; mutation originally found in Miniature Pinscher
Degenerative Myelopathy (DM)
Dominant Progressive Retinal Atrophy (DPRA)
Duchenne or Dystrophin Muscular Dystrophy (DMD); mutation originally found in Golden
Retrieve Early Retinal Degeneration (erd); mutation originally found in Norwegian Elkhound
Early-Onset Progressive Polyneuropathy; mutation originally found in Alaskan Malamute
Early-Onset Progressive Polyneuropathy; mutation originally found in Greyhound
Epidermolysis bullosa dystrophic; mutation originally found in Golden Retriever
Epidermolytic Hyperkeratosis
Episodic Falling (EF)
Exercise-Induced Collapse (EIC)

Factor IX Deficiency or Hemophilia B; mutation Gly379Glu
Factor IX Deficiency or Hemophilia B; mutation originally found in Airedale Terrier
Factor IX Deficiency or Hemophilia B; mutation originally found in Lhasa Apso
Factor IX Deficiency or Hemophilia B; mutation originally found in Rhodesian Ridgeback
Factor VII Deficiency
Factor VIII Deficiency or Hemophilia A; mutation originally found in Boxer
Factor VIII Deficiency or Hemophilia A; mutation originally found in German Shepherd Dog
Factor VIII Deficiency or Hemophilia A; p.Cys548Tyr mutation originally found in German Shepherd
Familial Nephropathy (FN); mutation originally found in English Cocker Spaniel
Familial Nephropathy (FN); mutation originally found in English Springer Spaniel
Fetal Onset Neuroaxonal Dystrophy (FNAD)
Generalized Progressive Retinal Atrophy
Glanzmann Thrombasthenia Type I (GT); mutation originally found in Pyrenean Mountain Dog
Globoid Cell Leukodystrophy or Krabbe's Disease (GLD); mutation originally found in Irish Setter
Globoid Cell Leukodystrophy or Krabbe's Disease (GLD); mutation originally found in Terriers
Glycogen Storage Disease Type Ia (GSD Ia)
Glycogen Storage Disease Type II or Pompe's Disease (GSD II)
Glycogen Storage Disease Type IIIa (GSD IIIa)
GM1 Gangliosidosis; mutation originally found in Alaskan Husky
GM1 Gangliosidosis; mutation originally found in Portuguese Water Dog
GM1 Gangliosidosis; mutation originally found in Shiba Dog
GM2 Gangliosidosis; mutation originally found in Japanese Chin
GM2 Gangliosidosis; mutation originally found in Toy Poodle
Golden Retriever Ichthyosis
Golden Retriever Progressive Retinal Atrophy 1 (GR_PRA 1)
Golden Retriever Progressive Retinal Atrophy 2 (GR_PRA 2)
Hereditary Ataxia or Cerebellar Ataxia; mutation originally found in Old English Sheepdog and Gor
Hereditary Elliptocytosis
Hereditary Footpad Hyperkeratosis (HFH)
Hereditary Nasal Parakeratosis (HNPK)
Hereditary Phosphofructokinase (PFK) Deficiency
Hereditary Vitamin D-Resistant Rickets (HVDRR)
Hyperekplexia or Startle Disease
Hyperuricosuria (HUU)
Hypocatalasia or Acatlasemia
Hypomyelination; mutation originally found in Weimaraner
Imerslund-Gräsbeck Syndrome (IGS); mutation originally found in Beagle
Imerslund-Gräsbeck Syndrome (IGS); mutation originally found in Border Collie
L-2-Hydroxyglutaric aciduria (L2HGA); mutation originally found in Staffordshire Bull Terrier
Lagotto Storage Disease (LSD)
Lamellar Ichthyosis (LI)
Macrothrombocytopenia; disease-linked variant originally found in Norfolk and Cairn Terrier
Malignant Hyperthermia (MH)

May-Hegglin Anomaly (MHA)
Mucopolysaccharidosis Type IIIA (MPS IIIA); mutation originally found in Dachshund
Mucopolysaccharidosis Type IIIA (MPS IIIA); mutation originally found in New Zealand Huntaway
Mucopolysaccharidosis Type VII (MPS VII); mutation originally found in Brazilian Terrier
Mucopolysaccharidosis Type VII (MPS VII); mutation originally found in German Shepherd
Multidrug Resistance 1 (MDR1 gene variant)
Muscular Hypertrophy (Double Muscling)
Musladin-Lueke syndrome (MLS)
Myotonia Congenita; mutation originally found in Australian Cattle Dog
Myotonia Congenita; mutation originally found in Miniature Schnauzer
Narcolepsy; mutation originally found in Dachshund
Narcolepsy; mutation originally found in Doberman Pinscher
Narcolepsy; mutation originally found in Labrador Retriever
Neonatal Cerebellar Cortical Degeneration or Cerebellar Abiotrophy (NCCD)
Neonatal Encephalopathy with Seizures (NEWS)
Neuronal Ceroid Lipofuscinosis 1 (NCL1); mutation originally found in Dachshund
Neuronal Ceroid Lipofuscinosis 10 (NCL10); mutation originally found in American Bulldog
Neuronal Ceroid Lipofuscinosis 4A (NCL4); mutation originally found in American Staffordshire Terrier
Neuronal Ceroid Lipofuscinosis 5 (NCL5); mutation originally found in Border Collie
Neuronal Ceroid Lipofuscinosis 8 (NCL8); mutation originally found in Australian Shepherd mix
Neuronal Ceroid Lipofuscinosis 8 (NCL8); mutation originally found in English Setter
Oculoskeletal Dysplasia 2 or Dwarfism-Retinal Dysplasia 2 (OSD2)
Osteochondrodysplasia; mutation originally found in Miniature Poodle
Osteogenesis Imperfecta (OI); mutation originally found in Dachshund
Persistent Müllerian Duct Syndrome (PMDS); mutation originally found in Miniature Schnauzer
Polycystic Kidney Disease in Bull Terriers (BTPKD)
Prekallikrein Deficiency
Primary Ciliary Dyskinesia (PCD)
Primary Hereditary Cataract (PHC); mutation originally found in Australian Shepherd
Primary Hereditary Cataract (PHC); mutation originally found in Terriers
Primary Lens Luxation (PLL)
Primary Open Angle Glaucoma (POAG); mutation originally found in Beagle
Primary Open Angle Glaucoma (POAG); mutation originally found in Norwegian Elkhound
Progressive Retinal Atrophy (PAP1_PRA); mutation originally found in Papillon and Phalene
Progressive Retinal Atrophy (PRA); mutation originally found in Basenji
Progressive Retinal Atrophy Type III (PRA type III); mutation originally found in Tibetan Spaniel and
Progressive Rod-Cone Degeneration (PRCD)
Protein Losing Nephropathy (PLN); NPHS1 gene variant
Pyruvate Dehydrogenase Phosphatase 1 (PDP1) Deficiency
Pyruvate Kinase Deficiency; mutation originally found in Basenji
Pyruvate Kinase Deficiency; mutation originally found in Beagle
Pyruvate Kinase Deficiency; mutation originally found in Pug
Pyruvate Kinase Deficiency; mutation originally found in West Highland White Terrier

Renal Cystadenocarcinoma and Nodular Dermatofibrosis (RCND)
Rod-Cone Dysplasia 1 (rcd1); mutation originally found in Irish Setter
Rod-Cone Dysplasia 1a (rdc1a); mutation originally found in Sloughi
Rod-Cone Dysplasia 3 (rcd3)
Severe Combined Immunodeficiency in Frisian Water Dogs (SCID)
Skeletal Dysplasia 2 (SD2)
Spinal Dysraphism
Spinocerebellar Ataxia / Late-Onset Ataxia (SCA/LOA)
Spinocerebellar Ataxia with Myokymia and/or Seizures (SCA)
Thrombopathia; mutation originally found in Basset Hound
Thrombopathia; mutation originally found in Eskimo Spitz
Thrombopathia; mutation originally found in Landseer
Trapped Neutrophil Syndrome (TNS)
Von Willebrand's Disease Type 1 (vWD 1)
Von Willebrand's Disease Type 2 (vWD 2)
Von Willebrand's Disease Type 3 (vWD 3); mutation originally found in Kooikerhondje
Von Willebrand's Disease Type 3 (vWD 3); mutation originally found in Scottish Terrier
Von Willebrand's Disease Type 3 (vWD 3); mutation originally found in Shetland Sheepdog
X-Linked Ectodermal Dysplasia (XHED)
X-Linked Hereditary Nephropathy (XLHN)
X-Linked Myotubular Myopathy
X-Linked Progressive Retinal Atrophy 1 (XLPRA1)
X-Linked Progressive Retinal Atrophy 2 (XLPRA2)
X-linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Basset Hound
X-linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Cardigan Welsh
X-Linked Tremors; mutation originally found in English Springer Spaniel