

0 At Risk

We did not detect any at risk genetic conditions in Pike's DNA.

0 Notable

We did not detect any notable genetic variants in Pike's DNA.

213 Clear

### 2,8-dihydroxyadenine (DHA) Urolithiasis

2,8-dihydroxyadenine (DHA) urolithiasis is a genetic disorder caused by a mutation in the APRT gene. A defective APRT enzyme can result in the formation of 2,8-dihydroxyadenine (DHA) urinary stones which can lead to urinary tract obstruction.

### Acral Mutilation Syndrome

Acral Mutilation Syndrome (AMS) causes insensitivity to pain, leading to a tendency to lick or bite paws excessively. This often results in loss of toenails, fractures, and toe amputation.

### Acute Respiratory Distress Syndrome

Acute respiratory distress syndrome (ARDS) is a rare, life-threatening disorder that causes a rapid severe respiratory failure that is unresponsive to treatment.

### Alaskan Husky Encephalopathy

Alaskan Husky Encephalopathy (AHE) is a severe, early-onset disorder of the nervous system that causes wide-ranging clinical signs, including seizures, behavioral changes, and incoordination.

### Alexander Disease

Alexander disease is a rare, fatal neurological disease, causing progressive weakness in all limbs.

### Amelogenesis Imperfecta

Amelogenesis Imperfecta (AI) is a disorder that affects the hard enamel coating of the teeth. Dogs with this condition suffer from enamel thinning and roughening and discoloration of the teeth.

### Bandera's Neonatal Ataxia

Bandera's Neonatal Ataxia (BNA), discovered in the Coton de Tulear, is a severe brain problem that affects a puppy's ability to move its body properly.

### Benign Familial Juvenile Epilepsy

Benign Familial Juvenile Epilepsy (BFJE) is a neurological disorder causing epileptic seizures.

### Canine Leukocyte Adhesion Deficiency (CLAD), type III

Canine Leukocyte Adhesion Deficiency (CLAD) is a disease causing fever, mucosal hemorrhaging, marked dental disease, poor wound healing, and lameness.

### Canine Multifocal Retinopathy 1

Canine Multifocal Retinopathy 1 (CMR1) is an eye disorder that can cause retinal decay which may impact vision, but very rarely results in blindness.

### Canine Multifocal Retinopathy 2

Canine Multifocal Retinopathy 2 (CMR2) is an eye disorder that can cause retinal decay which may impact vision, but very rarely results in blindness.

## Canine Multifocal Retinopathy 3

Canine Multifocal Retinopathy 3 (CMR3) is an eye disorder that can cause retinal decay which may impact vision, but very rarely results in blindness.

## Canine Scott Syndrome

Canine Scott Syndrome (CSS) is a bleeding disorder caused by a hereditary defect affecting blood platelets, which are important in the blood clotting process. This means that clots form more slowly at a site of bleeding.

## Centronuclear Myopathy (Discovered in the Great Dane)

Centronuclear Myopathy (CNM) is a type of muscle disease causing weakness and muscle wasting.

## Centronuclear Myopathy (Discovered in the Labrador Retriever)

Centronuclear Myopathy (CNM) is a type of muscle disease causing weakness and muscle wasting.

## Cerebellar Ataxia

Cerebellar Ataxias (CAs) are a group of disorders in which there is a degeneration in the movement center of the brain, known as the cerebellum. The degeneration leads to a lack of coordination in movements.

## Cerebellar Cortical Degeneration

Cerebellar cortical degeneration (CDD) is a disease causing incoordination and impaired balance.

## Cerebellar Hypoplasia

Cerebellar Hypoplasia (CH) causes difficulty controlling bodily movements (ataxia). The severity of the ataxia can vary between affected dogs and does not worsen with age.

## Cerebral Dysfunction

Cerebral Dysfunction is a brain disorder that causes affected puppies to show severe mental depression, odd behaviors, and limb weakness.

## Chondrodysplasia

Chondrodysplasia is a skeletal disorder that leads to the development of shorter than normal legs.

## Cleft Lip & Palate with Syndactyly

Cleft Lip & Palate with Syndactyly (CLPS) is a disorder that causes an abnormal hole in the roof of the mouth (cleft palate), a hole between the lip and the nose (cleft lip), and joined toes.

## Cleft Palate

Cleft Palate (CP) a disorder that causes an abnormal hole in the roof (palate) of the mouth. Puppies with this condition can also have a smaller than normal lower jaw bone.

## Collie Eye Anomaly (CEA)

Collie Eye Anomaly (CEA) is an eye disease where the layer in the eye supplying blood and nutrients to the retina is thinner than normal, resulting in visual defects in more severely affected dogs. CEA is most commonly found in breeds of herding descent.

## Complement 3 Deficiency

Complement 3 Deficiency (C3D) is a disease that causes severe immunodeficiency, leaving affected dogs vulnerable to infections.

### **Cone Degeneration (Discovered in the Alaskan Malamute)**

Cone Degeneration (CD), also called "day-blindness" is an inherited eye disorder causing light-sensitivity (photophobia) and an inability to see in bright light.

### **Cone Degeneration (Discovered in the German Shepherd Dog)**

Cone Degeneration (CD), also called "day-blindness" is an inherited eye disorder causing light-sensitivity (photophobia) and an inability to see in bright light.

### **Cone Degeneration (Discovered in the German Shorthaired Pointer)**

Cone Degeneration (CD), also called "day-blindness" is an inherited eye disorder causing light-sensitivity (photophobia) and an inability to see in bright light.

### **Cone-Rod Dystrophy**

Cone-Rod Dystrophy (CRD) is an eye disorder resulting in degeneration of the retina at the back of the eye at a young age, causing progressive vision loss.

### **Cone-Rod Dystrophy 1**

Cone-Rod Dystrophy (CRD1) is an eye disorder resulting in degeneration of the retina at the back of the eye at a young age, causing progressive vision loss.

### **Cone-Rod Dystrophy 2**

Cone-Rod Dystrophy (CRD2) is an eye disorder resulting in degeneration of the retina at the back of the eye at a young age, causing progressive vision loss.

### **Congenital Dyshormonogenic Hypothyroidism with Goiter (Discovered in the Shih Tzu)**

Congenital Dyshormonogenic Hypothyroidism (CDH) is a rare condition causing a defect in iodide transport and a subsequent reduction in the synthesis of thyroid hormones, resulting in hypothyroidism and goiter.

### **Congenital Hypothyroidism (Discovered in the Tenterfield Terrier)**

Congenital Hypothyroidism is a disease of insufficient thyroid hormone production. As this hormone is important in many aspects of the metabolism and development, the result is a wide variety of signs including slow growth, dwarfism, and mental impairment.

### **Congenital Hypothyroidism (Discovered in the Toy Fox and Rat Terrier)**

Congenital Hypothyroidism is a disease of insufficient thyroid hormone production. As this hormone is important in many aspects of the metabolism and development, the result is a wide variety of signs including slow growth, dwarfism, and mental impairment.

### **Congenital Myasthenic Syndrome (Discovered in the Golden Retriever)**

Congenital myasthenic syndromes (CMSs) are a group of inherited neuromuscular disorders. The CMS in Golden Retrievers is characterized by generalized muscle weakness and abnormal gait. The causative gene for CMS in Golden Retrievers is COLQ.

### **Congenital Myasthenic Syndrome (Discovered in the Jack Russell Terrier)**

Congenital Myasthenic Syndrome (CMS) is a neuromuscular disorder. Affected dogs suffer from exercise intolerance and collapse after 5-30 minutes of exercise.

### **Congenital Myasthenic Syndrome (Discovered in the Labrador Retriever)**

Congenital Myasthenic Syndrome (CMS) is a neuromuscular disorder. Affected dogs suffer from exercise intolerance and collapse after 5-30 minutes of exercise.

### **Congenital Myasthenic Syndrome (Discovered in the Old Danish Pointer)**

Congenital Myasthenic Syndrome (CMS) is a neuromuscular disorder. Affected dogs suffer from exercise intolerance and collapse after 5-30 minutes of exercise.

### **Congenital Stationary Night Blindness (CSNB)**

Congenital Stationary Night Blindness (CSNB) is an eye (retinal) disorder that causes non-progressive or very slowly progressing loss of night vision.

### **Cranio-mandibular Osteopathy**

Cranio-mandibular Osteopathy (CMO), also known as "lion's jaw", is a disorder of the skull where the jaw bones show swelling and thickening, causing pain, drooling, and difficulties in eating.

### **Cystic Renal Dysplasia and Hepatic Fibrosis**

Cystic renal dysplasia and hepatic fibrosis is a disorder causing renal (kidney failure) and liver scarring (fibrosis)

### **Cystinuria Type I-A**

Dogs with Cystinuria are not able to reabsorb the amino acid cystine in their kidneys and therefore high concentrations can accumulate in the urinary tract resulting in formation of cystine crystals and stones that can cause obstruction.

### **Cystinuria Type II-A**

Dogs with Cystinuria are not able to reabsorb the amino acid cystine in their kidneys and therefore high concentrations can accumulate in the urinary tract resulting in formation of cystine crystals and stones that can cause obstruction.

### **Deafness and Vestibular Dysfunction (Discovered in Doberman Pinscher)**

Dogs with this condition show deafness in one or both ears. Other signs include head tilt, circling, lack of coordination, and uncontrolled eye movements.

### **Degenerative Myelopathy**

Degenerative Myelopathy (DM) is a neurological disorder, usually affecting dogs in their senior years. Loss of hind limb coordination is an early sign of disease, and as the condition progresses the hind limbs of affected dogs become increasingly weak.

### **Demyelinating Neuropathy**

Demyelinating Neuropathy is a progressive neurodegenerative disease, causing noisy breathing, regurgitation and mild exercise intolerance.

### **Dental Hypomineralization**

Dental Hypomineralization is a disease that causes abnormal mineralization of the teeth, resulting in a brownish discoloration and abnormal wear of the teeth.

### **Dilated Cardiomyopathy (Discovered in the Schnauzer)**

Dilated cardiomyopathy (DCM) is a cardiac disorder causing heart failure.

### **Dominant Progressive Retinal Atrophy**

Dominant Progressive Retinal Atrophy (DPRA) is an eye disease resulting in vision loss and eventual blindness.

### **Dystrophic Epidermolysis Bullosa (Discovered in the Central Asian Ovcharka)**

Dystrophic Epidermolysis Bullosa is a skin disorder that causes blistering of the skin and irritations in the oral cavity and upper digestive tract. These disease signs may diminish around 8 months of age.

### **Dystrophic Epidermolysis Bullosa (Discovered in the Golden Retriever)**

Dystrophic Epidermolysis Bullosa is a skin disorder that causes blistering of the skin and irritations in the oral cavity and upper digestive tract. These disease signs may diminish around 8 months of age.

### **Early Adult Onset Deafness For Border Collies only (Linkage test)**

Early Adult Onset Deafness is a disease of gradual hearing loss affecting both ears. This test is a linkage test and relevant for the Border Collie breed only. The disease causing variant has not yet been identified.

### **Early Retinal Degeneration (Discovered in the Norwegian Elkhound)**

Early retinal degeneration (ERD) is an eye disorder characterized by abnormal development followed by degeneration of photoreceptors in the eye. The disorder causes early onset blindness in Norwegian Elkhounds.

### **Early-Onset Progressive Polyneuropathy (Discovered in the Alaskan Malamute)**

Early-Onset Progressive Polyneuropathy is characterized by the dysfunction and breakdown of multiple nerve types, which causes generalized weakness, and related clinical signs.

### **Early-Onset Progressive Polyneuropathy (Discovered in the Greyhound)**

Early-Onset Progressive Polyneuropathy is characterized by the dysfunction and breakdown of multiple nerve types, which causes generalized weakness, and related clinical signs.

### **Early-onset PRA (Discovered in the Portuguese Water Dog)**

Progressive retinal atrophy (PRA) is a disorder where the light sensing retina at the back of the eye degenerates resulting in vision loss.

### **Enamel Hypoplasia (Discovered in the Parson Russell Terrier)**

Amelogenesis imperfecta, also known as enamel hypoplasia, is a disorder where the hard enamel covering of the teeth is thin, roughened and discolored.

### **Epidermolytic Hyperkeratosis**

Epidermolytic Hyperkeratosis is a skin disorder that causes the skin to be fragile and easily damaged from birth followed by thickening of the skin in adulthood.

### **Episodic Falling Syndrome**

Episodic Falling Syndrome (EFS) is a condition causing the temporary inability to relax muscles resulting in collapse episodes which are usually a few seconds to several minutes long.

### **Exercise-Induced Collapse**

Exercise-Induced Collapse (EIC) is a neuromuscular disorder which can cause incoordination and weakness, resulting in collapse, after periods of strenuous exercise.

### **Factor VII Deficiency**

Factor VII Deficiency is an inherited blood clotting disorder that results in excessive bleeding occurring after a severe trauma or surgery. The signs of the disease are typically mild but can vary in severity in different affected dogs.

---

### **Factor XI Deficiency**

Factor XI Deficiency is a hereditary disorder that impacts blood clotting. The disease usually causes a mild, spontaneous bleeding disorder but more severe bleeding may occur following surgery. Many dogs that are at risk will not show any signs of disease.

---

### **Fanconi Syndrome**

Fanconi Syndrome is a disorder of kidney function. For dogs affected with the syndrome the kidney's ability to reabsorb essential metabolites such as glucose, electrolytes, amino acids, and proteins in the urine, is impaired causing a wide variety of signs including frequent drinking and urinating, weight loss, and poor coat condition.

---

### **Fetal Onset Neuroaxonal Dystrophy**

Fetal Onset Neuroaxonal Dystrophy (FNAD) is a disorder that disturbs the development of motor (movement) nerves in the central nervous system. Signs include abnormal curvature of the spine and contracted joints. Affected puppies to have no voluntary movement of limbs. Respiratory deficiencies (difficulties breathing) cause affected puppies to die at birth.

---

### **Focal Non-Epidermolytic Palmoplantar Keratoderma**

Focal Non-Epidermolytic Palmoplantar Keratoderma (FNEPPK) is an inherited skin disorder that causes a type of hereditary footpad hyperkeratosis (HFH). Signs include hard, thickened, and cracked footpads.

---

### **GM1 Gangliosidosis (Discovered in the Portuguese Water Dog)**

GM1 gangliosidosis is a disorder of progressive nervous system degeneration, resulting in vision impairment, head tremor, involuntary eye movements, limb weakness with difficulties in balancing and fatigue.

---

### **GM1 Gangliosidosis (Discovered in the Shiba)**

GM1 gangliosidosis is a disorder of progressive nervous system degeneration, resulting in vision impairment, head tremor, involuntary eye movements, limb weakness with difficulties in balancing and fatigue.

---

### **GM2 Gangliosidosis (Discovered in the Japanese Chin)**

GM2 Gangliosidosis, is a disorder of progressive nervous system degeneration, with signs including incoordination of movements and tremors.

---

### **GM2 Gangliosidosis (Discovered in the Toy Poodle)**

GM2 Gangliosidosis, is a disorder of progressive nervous system degeneration, with signs including incoordination of movements and tremors.

---

### **Generalized Progressive Retinal Atrophy (Discovered in the Schapendoes)**

Generalized progressive retinal atrophy (GPRA) is an eye disorder where the light sensing retina at the back of the eye degenerates, causing progressive vision loss and eventual blindness.

---

### **Glanzmann Thrombasthenia Type I**

Glanzmann thrombasthenia (GT) causes susceptibility to bleeding due to poor blood platelet aggregation.

---

## **Glanzmann Thrombasthenia Type I (Discovered in Great Pyrenees)**

Glanzmann Thrombasthenia (GT) Type I is a blood disorder characterized by poor blood platelet aggregation. Platelet in the blood are needed to help start clot formation, so as a result of this disorder bleeding may be prolonged.

## **Globoid Cell Leukodystrophy (Discovered in Terriers)**

Globoid Cell Leukodystrophy (GLD) is a disorder which results in degeneration of the nervous system. GLD is characterized by muscle weakness, tremors, and ataxia (uncoordinated movement). Signs of the disease also include behavioral changes, incoherence, blindness, and deficits in normal reflexes.

## **Globoid Cell Leukodystrophy (Discovered in the Irish Setter)**

Globoid Cell Leukodystrophy (GLD) is a disorder which results in degeneration of the nervous system. GLD is characterized by muscle weakness, tremors, and ataxia (uncoordinated movement). Signs of the disease also include behavioral changes, incoherence, blindness, and deficits in normal reflexes.

## **Glycogen Storage Disease Type IIIa, (GSD IIIa)**

Glycogen storage disease (GSD) type IIIa is a disorder that affects the metabolism of glycogen.

## **Glycogen Storage Disease Type Ia**

Glycogen Storage Disease (GSD) Type Ia is a severe metabolic disorder causing critically low blood sugar levels.

## **Goniodysgenesis and Glaucoma (Discovered in the Border Collie)**

Goniodysgenesis and glaucoma is an eye disorder where the pressure inside the eye increases to higher than normal levels, and can lead to eye damage and blindness.

## **Hemophilia A (Discovered in Old English Sheepdog)**

Hemophilia A, also known as Factor VIII Deficiency, is a blood clotting disorder, which can cause bruising or abdominal bleeding without apparent reason. The disease is more commonly seen in male dogs.

## **Hemophilia A (Discovered in the Boxer)**

Hemophilia A, also known as Factor VIII Deficiency, is a blood clotting disorder, which can cause bruising or abdominal bleeding without apparent reason. The disease is more commonly seen in male dogs.

## **Hemophilia A (Discovered in the German Shepherd Dog - Variant 1)**

Hemophilia A, also known as Factor VIII Deficiency, is a blood clotting disorder, which can cause bruising or abdominal bleeding without apparent reason. The disease is more commonly seen in male dogs.

## **Hemophilia A (Discovered in the German Shepherd Dog - Variant 2)**

Hemophilia A, also known as Factor VIII Deficiency, is a blood clotting disorder, which can cause bruising or abdominal bleeding without apparent reason. The disease is more commonly seen in male dogs.

## **Hemophilia A (Discovered in the Havanese)**

Hemophilia A, also known as Factor VIII Deficiency, is a blood clotting disorder, which can cause bruising or abdominal bleeding without apparent reason. The disease is more commonly seen in male dogs.

## Hemophilia B

Hemophilia B, also known as Factor IX Deficiency, is a blood clotting disorder most commonly seen in male dogs, which can result in prolonged bleeding after an injury or a surgical procedure.

### Hemophilia B (Discovered in the Airedale Terrier)

Hemophilia B, also known as Factor IX Deficiency, is a blood clotting disorder most commonly seen in male dogs, which can result in prolonged bleeding after an injury or a surgical procedure.

### Hemophilia B (Discovered in the Lhasa Apso)

Hemophilia B, also known as Factor IX Deficiency, is a blood clotting disorder most commonly seen in male dogs, which can result in prolonged bleeding after an injury or a surgical procedure.

## Hereditary Ataxia (Discovered in the Norwegian Buhund)

Hereditary ataxia is a disorder of the nervous system leading to uncoordinated movement and head tremors.

## Hereditary Elliptocytosis

Hereditary Elliptocytosis is a disease where the red blood cells are abnormally oval shape, rather than the characteristic biconcave or dumbbell shape that is normally seen.

## Hereditary Footpad Hyperkeratosis

Hereditary Footpad Hyperkeratosis (HFH) is a skin disorder, with disease signs including hard, thickened, and cracked footpads and an abnormal coat.

## Hereditary Nasal Parakeratosis (Discovered in the Greyhound)

Hereditary nasal parakeratosis (HNPK) is a disorder causing crusting and dryness of the nose, leading to soreness and irritation.

## Hereditary Nasal Parakeratosis (Discovered in the Labrador Retriever)

Hereditary nasal parakeratosis (HNPK) is a disorder causing crusting and dryness of the nose, leading to soreness and irritation.

## Hereditary Vitamin D-Resistant Rickets Type II

Hereditary Vitamin D-Resistant Rickets (HVDRR) is a bone defect where vitamin D cannot be delivered to the bones effectively. This prevents normal bone mineralization, leading to softening and bending of the bones and skeletal problems.

## Hyperekplexia or Startle Disease

Hyperekplexia or Startle Disease is a rare muscle cramping disorder. Signs of the disease can be seen in very young puppies, and include muscle stiffness and tremors when handled.

## Hyperuricosuria

Hyperuricosuria (HUU) is a condition that predisposes affected dogs to the formation of urinary stones, such as kidney or bladder stones.

## Hypocatalasia

Hypocatalasia is the deficiency of an enzyme called catalase in red blood cells. The catalase enzyme plays an important role in the cells defense against a type of chemical damage, known as oxidative damage. The disorder is characterized by ulcers and progressive gangrene (tissue death) of the mouth.

## Hypomyelination

Hypomyelination is a neurological disorder causing muscle tremors and movement difficulties.

### **Hypophosphatemia**

This early onset condition originally discovered in Karelian Bear Dogs is a metabolic bone disease that disturbs skeletal mineralization.

### **Ichthyosis (Discovered in the American Bulldog)**

Ichthyosis is a skin condition causing generalized scaling of the skin and abdominal redness.

### **Ichthyosis (Discovered in the Great Dane)**

Ichthyoses is a disease of skin cornification. This particular mutation causes severe thickening, drying and scaling of the skin, which can lead to secondary infections.

### **Intestinal Cobalamin Malabsorption (Discovered in the Beagle)**

Intestinal Cobalamin Malabsorption (ICM) or Imerslund-Gräsbeck Syndrome is a metabolic disorder resulting from a failure to absorb vitamin B12 in the small intestine of the gut causing retarded growth, a low count of the oxygen carrying red blood cells (anemia), and a low count of white blood cells (immune system cells).

### **Intestinal Cobalamin Malabsorption (Discovered in the Border Collie)**

Intestinal Cobalamin Malabsorption (ICM) or Imerslund-Gräsbeck Syndrome is a metabolic disorder resulting from a failure to absorb vitamin B12 in the small intestine of the gut causing retarded growth, a low count of the oxygen carrying red blood cells (anemia), and a low count of white blood cells (immune system cells).

### **Intestinal Cobalamin Malabsorption (Discovered in the Komondor)**

Intestinal Cobalamin Malabsorption (ICM) or Imerslund-Gräsbeck Syndrome is a disorder where the body is unable to absorb cobalamin (vitamin B12) in the small intestine. This results in weakness and failure to thrive, vomiting, diarrhea, anemia, and decreased numbers of white blood cells (immune system cells).

### **Juvenile Encephalopathy (Discovered in the Parson Russell Terrier)**

Juvenile encephalopathy is a brain disorder causing epileptic seizures and progressive brain damage.

### **Juvenile Laryngeal Paralysis and Polyneuropathy**

Juvenile laryngeal paralysis and polyneuropathy (JLPP) causes difficulty breathing and swallowing due to voice box (larynx) paralysis.

### **Juvenile Myoclonic Epilepsy**

Juvenile Myoclonic Epilepsy is a specific form of epilepsy first described in Rhodesian Ridgebacks, causing muscle jerks and twitches, that can progress to generalized seizures.

### **L-2-Hydroxyglutaric Aciduria**

L-2-Hydroxyglutaric Aciduria (L2HGA) is a metabolic disease caused by a fault in the enzyme that breaks down a chemical in the body known as L-2-hydroxyglutaric acid, which increases to toxic levels. This causes damage to the nervous system and results in incoordination, muscle stiffness during exercise or times of excitement, and altered behavior or epileptic seizures.

### **L-2-Hydroxyglutaric Aciduria (Discovered in the Westie)**

L-2-Hydroxyglutaric Aciduria (L2HGA) is a metabolic disease that causes damage to the nervous system and results in incoordination, muscle stiffness during exercise or times of excitement, and altered behavior or epileptic seizures.

### **Lagotto Storage Disease**

Lagotto Storage Disease (LSD) is a neurological disorder resulting in a progressive incoordination, poor balance and behavior changes.

### **Lamellar Ichthyosis**

Lamellar Ichthyosis is a skin disorder causing severe thickening and scaling of the skin, leading to secondary bacterial and yeast skin infections.

### **Lethal Acrodermatitis (Discovered in the Bull Terrier)**

Lethal acrodermatitis (LAD) is a disease characterized by poor growth, immune system deficiency, skin problems and infections.

### **Ligneous Membranitis**

Ligneous Membranitis is a rare inflammatory disease of the mucous membranes. The disease causes conjunctivitis, mouth ulcers and swollen gums. Other clinical signs include nasal discharge, loud breathing and enlarged lymph nodes.

### **Lung Developmental Disease (Discovered in the Airedale Terrier)**

Lung developmental disease in Airedale Terriers is characterized by lethal hypoxic respiratory distress and failure that occur within the first days or weeks of life in affected puppies.

### **MDR1 Medication Sensitivity**

The MDR1 gene mutation causes a defect to a drug pumping protein that plays an important role in limiting drug absorption and distribution (particularly to the brain). Dogs with the MDR1 mutation may have severe adverse reactions to some commonly used medications.

### **Macrothrombocytopenia**

Macrothrombocytopenia is a blood disorder characterized by oversized blood platelets, which play an important role in blood clotting when a blood vessel is injured.

### **May-Hegglin Anomaly**

May-Hegglin Anomaly (MHA) is a blood disorder that causes deficiency and abnormal shaping of the blood platelets which are important in the clotting process.

### **Microphthalmia (Discovered in the Soft-Coated Wheaten Terrier)**

Microphthalmia is an eye disorder present from birth where affected puppies to have abnormally small and underdeveloped eyes.

### **Mucopolysaccharidosis Type IIIA (Discovered in the Dachshund)**

Mucopolysaccharidosis Type IIIA (MSP IIIA) is a disease of progressive incoordination, first in the pelvic legs and later progressing to all four legs. Leg movements become erratic when walking and affected dogs have difficulty balancing.

### **Mucopolysaccharidosis Type IIIA (Discovered in the New Zealand Huntaway)**

Mucopolysaccharidosis Type IIIA (MSP IIIA) is a disease of progressive incoordination, first in the hindlimbs and later progressing to all four legs. Leg movements become erratic when walking and affected dogs have difficulty balancing.

### **Mucopolysaccharidosis Type VII (Discovered in the Brazilian Terrier)**

Mucopolysaccharidosis Type VII (MPS VII) is a disorder causing severe changes in skeletal structure, with joint hyperlaxity.

### **Mucopolysaccharidosis Type VII (Discovered in the German Shepherd Dog)**

Mucopolysaccharidosis Type VII (MPS VII) is a disorder causing hind limb weakness (progressing to incoordination in all legs), growth retardation, facial and other skeletal abnormalities, and corneal clouding.

### **Muscular Dystrophy (Discovered in the Cavalier King Charles Spaniel)**

Muscular Dystrophy is a severe disorder that causes muscle breakdown and weakness. DMD is characterized by a curve to the dog's back and a crouched posture. It typically affects males, although some females may also show some muscle weakness as well.

### **Muscular Dystrophy (Discovered in the Golden Retriever)**

Muscular Dystrophy is a severe disorder that causes muscle degeneration and weakness due to the formation of excess connective tissue in the muscle.

### **Muscular Dystrophy (Discovered in the Landseer)**

Muscular Dystrophies are a group of progressive disorders leading to muscular dysfunction. This form leads to movement difficulties from a young age due to increasing muscle weakness.

### **Muscular Dystrophy (Discovered in the Norfolk Terrier)**

This form of Muscular Dystrophy is a disease of progressive muscular weakness, respiratory problems, and cardiomyopathy (heart disease).

### **Muscular Hypertrophy (Double Muscling)**

Muscular Hypertrophy (Double Muscling) is a condition where the muscles of the body become much larger and more pronounced than normal, due to a defect in a gene that helps to regulate muscle growth.

### **Musladin-Lueke Syndrome**

Musladin-Lueke Syndrome (MLS) is a disorder affecting the development and structure of connective tissue that is characterized by stiff joints, an abnormal facial expression, and thick, tight skin.

### **Myeloperoxidase Deficiency**

Myeloperoxidase Deficiency is a condition causing an increased susceptibility to fungal and bacterial infections. This is due to a defect of an important enzyme present in the white blood cells of the immune system.

### **Myotonia Congenita**

Myotonia Congenita is a muscle disorder affecting dogs from birth. The condition causes affected dogs to have muscles that contract and cramp easily.

### **Myotonia Congenita (Discovered in the Labrador Retriever)**

Myotonia congenita is a muscle disorder that affects dogs from birth, causing stiff movement and delayed relaxation of muscles after exercise.

### **Myotonia Congenita (Discovered in the Miniature Schnauzer)**

Myotonia congenita is a muscle disorder that affects dogs from birth, causing stiff movement and delayed relaxation of muscles after exercise.

### **Myotubular Myopathy**

Myotubular Myopathy is a disorder that affects the muscle cells. This starts with an early-onset hind limb weakness, progressing to an inability to move.

### **Narcolepsy (Discovered in the Dachshund)**

Narcolepsy is a sleep disorder that causes sudden attacks of sleep due to the brain's inability to regulate REM sleep.

### **Narcolepsy (Discovered in the Labrador Retriever)**

Narcolepsy is a sleep disorder that causes sudden attacks of sleep due to the brain's inability to regulate REM sleep.

### **Nemaline Myopathy**

Nemaline myopathy is a muscle disorder characterized by defects to muscle fibers causing weakness and tremors.

### **Neonatal Cerebellar Cortical Degeneration**

Neonatal Cerebellar Cortical Degeneration (NCCD) is a disease of uncoordinated movements and loss of balance that is present from birth.

### **Neonatal Encephalopathy with Seizures**

Neonatal Encephalopathy with Seizures (NEWS) is a disorder that affects the development of the balance and movement center of the brain.

### **Neuroaxonal Dystrophy**

Neuroaxonal Dystrophy is a neurodegenerative disorder. Signs of this disease include movement abnormalities, abnormal vocalization, incontinence, and behavioral changes.

### **Neuroaxonal Dystrophy (Discovered in the Papillon)**

Neuroaxonal dystrophy (NAD) comprises a group of rare neurodegenerative disorders. NAD in the Papillon is characterized by an abnormal gait, blindness, tremors, and collapse.

### **Neuroaxonal Dystrophy (Discovered in the Rottweiler)**

Neuroaxonal dystrophy is a neurodegenerative disorder causing uncoordinated movement and gait (walking/running pattern) changes.

### **Neuronal Ceroid Lipofuscinosis 1**

Neuronal Ceroid Lipofuscinosis 1 (NCL1) is a neurological disease, with typical signs of rapidly progressing vision impairment, ataxia (uncontrolled movements), and behavioral changes, such as anxiety, sound sensitivity, and inability to recognize familiar individuals.

### **Neuronal Ceroid Lipofuscinosis 12 (Discovered in the Australian Cattle Dog)**

Neuronal Ceroid Lipofuscinosis 12 (NCL12) is a late onset progressive disease causing uncoordinated movements and behavioral changes.

### **Neuronal Ceroid Lipofuscinosis 7**

Neuronal Ceroid Lipofuscinosis 7 (NCL7) is a progressive disease affecting behavior, movement, and vision, with a risk of epileptic seizures in later stages.

### **Neuronal Ceroid Lipofuscinosis 8 (Discovered in the Alpine Dachsbracke)**

Neuronal Ceroid Lipofuscinosis 8 (NCL8) is a progressive disease causing uncoordinated movements, behavioral changes, vision loss, and epileptic seizures.

### **Neuronal Ceroid Lipofuscinosis 8 (Discovered in the Australian Shepherd)**

Neuronal Ceroid Lipofuscinosis 8 (NCL8) is a progressive disease causing uncoordinated movements, behavioral changes, vision loss, and epileptic seizures.

### **Neuronal Ceroid Lipofuscinosis 8 (Discovered in the English Setter)**

Neuronal Ceroid Lipofuscinosis 8 (NCL8) is a progressive disease causing uncoordinated movements, behavioral changes, vision loss, and epileptic seizures.

### **Neuronal Ceroid Lipofuscinosis 8 (Discovered in the Saluki)**

Neuronal Ceroid Lipofuscinosis 8 (NCL8) is a progressive neurologic disease characterized by uncoordinated movements, behavioral changes, vision loss, and seizures.

### **Obesity risk (POMC)**

A number of factors can affect appetite and risk of obesity. This particular test detects a mutation in the POMC gene, known to regulate appetite, that has been associated with increased appetite and obesity in dogs.

### **Osteochondrodysplasia**

Osteochondrodysplasia is a disorder of bone and cartilage development resulting in stunted growth, misshapen limbs, and abnormal movement.

### **Osteochondromatosis (Discovered in the American Staffordshire Terrier)**

Osteochondromatosis is a condition causing benign bone tumors, called osteochondromas, to form during puppyhood.

### **Osteogenesis Imperfecta (Discovered in the Beagle)**

Osteogenesis Imperfecta (OI) is a disease of fragile bones and loose joints.

### **Osteogenesis Imperfecta (Discovered in the Dachshund)**

Osteogenesis Imperfecta (OI) is a disease of fragile bones and loose joints.

### **P2RY12-associated Bleeding Disorder**

This is a bleeding disorder due to a blood protein (P2RY12) defect and was first described in Greater Swiss Mountain Dogs.

### **Paroxysmal Dyskinesia**

Paroxysmal dyskinesia (PxD) is a neurological disorder causing episodes of abnormal tone or movement of limbs. Affected dogs seem normal between these episodes.

### **Persistent Müllerian Duct Syndrome**

Persistent Müllerian Duct Syndrome (PMDS) is a disorder of sexual development affecting male dogs, where a uterus and other female sex organs develop in otherwise externally normal-appearing male dogs.

### **Phosphofructokinase Deficiency**

Phosphofructokinase Deficiency (PFK) is a disorder where an enzyme important in the production of energy from sugars is lacking, resulting in weakness, muscle cramps, discolored urine, anemia, and jaundice.

### **Polycystic Kidney Disease**

Polycystic Kidney Disease (PKD) is a condition causing formation of cysts in the kidneys which leads to chronic kidney failure.

### **Prekallikrein Deficiency**

Prekallikrein Deficiency is a disorder which causes blood to take a longer time to clot.

### **Primary Ciliary Dyskinesia**

Primary Ciliary Dyskinesia (PCD) is a disorder found to affect formation of the tiny hairs in the respiratory system called cilia, resulting in recurrent respiratory tract infections. As cilia throughout the various body systems are affected, it can also impact hearing and cause infertility in males.

### **Primary Ciliary Dyskinesia (Discovered in the Alaskan Malamute)**

Primary ciliary dyskinesia (PCD) is a disorder that causes a defect in the formation of cilia, tiny hair-like projections, found on cells in the respiratory system, reproductive system, ear and nervous system. Clinical signs of PCD are recurrent infections of the respiratory tract and fertility problems.

### **Primary Lens Luxation**

Primary Lens Luxation (PLL) is a condition that can cause the lens of the eye to become loose and eventually displace. The disorder is caused by degeneration of the fibers that hold the lens in place.

### **Primary Open Angle Glaucoma (Discovered in Basset Fauve de Bretagne)**

Primary Open Angle Glaucoma (POAG) is a disorder that results in an increase in eye pressure, leading to damage to the optic nerve and retina, and blindness if left untreated.

### **Primary Open Angle Glaucoma (Discovered in Petit Basset Griffon Vendéen)**

Primary Open Angle Glaucoma (POAG) is a disorder that results in an increase in eye pressure, leading to damage to the optic nerve and retina, and blindness if left untreated.

### **Primary Open Angle Glaucoma and Lens Luxation (Discovered in Chinese Shar-Pei)**

Primary Open Angle Glaucoma (POAG) is a disease where the pressure in the eye increases, causing pain and leading to blindness if untreated. The same mutation can also cause lens luxation (LL), or displacement of the lens in the eye.

### **Progressive Early-Onset Cerebellar Ataxia**

Progressive early-onset cerebellar ataxia is a disorder of the nervous system that causes ataxia (uncoordinated movements) and loss of balance.

### **Progressive Retinal Atrophy**

Progressive Retinal Atrophy (PRA) causes the light sensitive photoreceptor cells in the eye to degenerate, leading to night blindness.

### **Progressive Retinal Atrophy (Discovered in the Basenji)**

Progressive retinal atrophy (PRA) is an eye disorder where the light sensing retina at the back of the eye degenerates, causing progressive vision loss and eventual blindness.

## Progressive Retinal Atrophy (Discovered in the Golden Retriever - GR-PRA1 variant)

Golden Retriever progressive retinal atrophy 1 (GR-PRA1) is an inherited eye disorder which causes degeneration of the light sensing retina at the back of the eye, resulting in progressive loss of vision.

## Progressive Retinal Atrophy (Discovered in the Lhasa Apso)

Progressive retinal atrophy (PRA) is a disorder where the light sensing retina at the back of the eye degenerates resulting in vision loss.

## Progressive Retinal Atrophy (Discovered in the Papillon and Phalène)

Progressive Retinal Atrophy (PRA) is a disorder that causes the degeneration of the light sensing retina at the back of the eye, resulting in vision loss.

## Progressive Retinal Atrophy (Discovered in the Shetland Sheepdog - BBS2 variant)

Progressive Retinal Atrophy (PRA) is an eye disease resulting in gradual loss of vision. Dogs with this form of the disease can show additional features including a wavy, atypical coat texture, an upturned nose and dental defects.

## Progressive Retinal Atrophy 1 (Discovered in the Italian Greyhound)

Progressive retinal atrophy (PRA) is a disorder where the light sensing retina at the back of the eye degenerates, resulting in vision loss.

## Progressive Retinal Atrophy Type III

Progressive Retinal Atrophy (PRA) is a disorder that causes the degeneration of the light sensing retina at the back of the eye, resulting in vision loss.

## Progressive Rod Cone Degeneration (prcd-PRA)

PRCD is a particularly common form of progressive retinal atrophy (PRA) and is found in many breeds and mixed breed dogs. PRA is caused by the degeneration of photoreceptor cells of the retina resulting in progressive vision loss and eventual blindness.

## Protein Losing Nephropathy

Protein Losing Nephropathy (PLN) is a disorder in which affected dogs lose protein through their kidneys, leading to kidney failure over time.

## Pyruvate Dehydrogenase Phosphatase 1 Deficiency

Pyruvate Dehydrogenase Phosphatase 1 (PDP1) Deficiency is a metabolic disorder characterized by exercise intolerance.

## Pyruvate Kinase Deficiency (Discovered in the Basenji)

Pyruvate kinase deficiency (PKD) is a disorder that causes anemia due to the breakdown of red blood cells.

## Pyruvate Kinase Deficiency (Discovered in the Beagle)

Pyruvate Kinase Deficiency (PKD) is a disorder that causes anemia due to the breakdown of red blood cells.

## Pyruvate Kinase Deficiency (Discovered in the Pug)

Pyruvate Kinase Deficiency (PKD) is a disorder that causes anemia due to the breakdown of red blood cells.

### **Pyruvate Kinase Deficiency (Discovered in the West Highland White Terrier)**

Pyruvate Kinase Deficiency (PKD) is a disorder that causes anemia due to the breakdown of red blood cells.

### **QT Syndrome**

Long QT Syndrome (LQTS) is a rare cardiac disease that causes an irregular heart beat and has been associated with sudden death in the English Springer Spaniel.

### **Renal Cystadenocarcinoma and Nodular Dermatofibrosis**

Renal Cystadenocarcinoma and Nodular Dermatofibrosis (RCND) is a canine kidney cancer syndrome.

### **Rod-Cone Dysplasia 1**

Rod-Cone Dysplasia 1 (rcd1) is an inherited eye disorder that results in blindness, and was identified in Irish Setters and Irish Red and White Setters.

### **Rod-Cone Dysplasia 1a**

Rod-Cone Dysplasia (rcd1a) is an eye disorder affecting the development of retinal photoreceptors at the back of the eye, resulting in vision loss.

### **Rod-Cone Dysplasia 3**

Rod-Cone Dysplasia 3 (rcd3) is an eye disorder affecting the development of retinal photoreceptors at the back of the eye, resulting in vision loss.

### **Sensory Ataxic Neuropathy**

Sensory Ataxic Neuropathy (SAN) is a slowly progressive neurologic disorder causing uncoordinated movements and impaired balance, that was first found in Golden Retrievers.

### **Sensory Neuropathy**

Sensory neuropathy is a rare, severe neurological disorder caused by the degeneration of nerve cells. Affected dogs lack pain sensation, resulting in injury and self harm.

### **Severe Combined Immunodeficiency**

Autosomal Recessive Severe Combined Immunodeficiency (ARSCID) is a severe immunodeficiency disorder discovered in Jack Russell Terriers, that leaves affected dogs prone to infection.

### **Severe Combined Immunodeficiency (Discovered in Frisian Water Dogs)**

Severe Combined Immunodeficiency (SCID) is a dysfunction of the immune system, that leaves affected dogs prone to infection.

### **Shaking Puppy Syndrome (Discovered in the Border Terrier)**

Spongiform LeucoEncephaloMyelopathy (SLEM) is a disorder commonly known as shaking puppy syndrome. Affected pups show severe body tremors, which are generally first noted when the puppy begins to try to walk.

### **Skeletal Dysplasia 2**

Skeletal Dysplasia 2 (SD2) is an abnormality of the skeleton that results in mild dwarfism.

### **Spinocerebellar Ataxia (Late-Onset Ataxia)**

Spinocerebellar Ataxia also known as Late Onset Ataxia (LOA) is a disease of the nervous system characterized by uncoordinated movements and impaired balance.

### **Spinocerebellar Ataxia with Myokymia and/or Seizures**

Spinocerebellar Ataxia with Myokymia and/or Seizures (SAMS) is a disease of the nervous system characterized by uncoordinated movements and impaired balance. This particular form may present with muscle twitching and seizures.

### **Spondylocostal Dysostosis**

Spondylocostal Dysostosis is a developmental disease that leads to skeletal abnormalities often noted at birth.

### **Spongy Degeneration with Cerebellar Ataxia**

Spongy Degeneration with Cerebellar Ataxia (SDCA) is an early onset disease of poor coordination, muscle twitching and seizures.

### **Spongy Degeneration with Cerebellar Ataxia (Discovered in Belgian Malinois)**

Spongy Degeneration with Cerebellar Ataxia (SDCA2) is an early onset disease of loss of coordination, seizures, circling, and blindness.

### **Stargardt Disease (Discovered in the Labrador Retriever)**

Stargardt Disease (STGD) is a form of late onset retinal degeneration, resulting in loss of the light detecting cells at the back of the eye and progressive vision loss.

### **Trapped Neutrophil Syndrome**

Trapped Neutrophil Syndrome (TNS) is a disorder of the white blood cells first identified in Border Collies.

### **Van den Ende-Gupta Syndrome**

Van den Ende-Gupta Syndrome (VDEGS) is a condition that can cause severe skeletal abnormalities. The most common ones noted are severe patella luxation (dislocated kneecap) and severe underbite.

### **X-Linked Ectodermal Dysplasia**

X-linked Ectodermal Dysplasia (XHED) is a disorder causing skin, dental, and immune system problems.

### **X-Linked Hereditary Nephropathy (Discovered in the Navasota Dog)**

X-Linked Hereditary Nephropathy (XLHN) is a kidney disorder, leading to kidney failure.

### **X-Linked Hereditary Nephropathy (Discovered in the Samoyed)**

X-Linked Hereditary Nephropathy (XLHN) is a kidney disorder, leading to kidney failure.

### **X-Linked Myotubular Myopathy**

X-linked Myotubular Myopathy (XLMTM) is a condition of the muscles that results in trouble eating, breathing, and moving around.

### **X-Linked Progressive Retinal Atrophy 1**

X-Linked Progressive Retinal Atrophy 1 (XLPRA1) causes degeneration of the light detecting retina at the back of the eye, resulting in loss of vision.

### **X-Linked Progressive Retinal Atrophy 2**

X-Linked Progressive Retinal Atrophy 2 (XLPRA2) causes early-onset degeneration of the light detecting retina at the back of the eye, resulting in loss of vision.

## X-Linked Severe Combined Immunodeficiency (Discovered in the Basset Hound)

X-linked Severe Combined Immunodeficiency (XSCID) is a severe dysfunction of the immune system, that leaves affected dogs prone to infection.

## X-Linked Severe Combined Immunodeficiency (Discovered in the Cardigan Welsh Corgi)

X-linked Severe Combined Immunodeficiency (XSCID) is a severe dysfunction of the immune system, that leaves affected dogs prone to infection.

## X-Linked Tremors

X-Linked Tremors is a neurologic disorder where affected dogs lack the protective covering over the nerves of the central nervous system, resulting in tremors and often leading to premature death.

## Xanthinuria (Discovered in a mixed breed dog)

Xanthinuria can cause formation of stones throughout the upper and lower urinary tracts, including the urethra, bladder, ureters, and kidneys. These stones can result in pain, bloody urine, infection and blockage of the urinary tract.

## Xanthinuria (Discovered in the Cavalier King Charles Spaniel)

Xanthinuria can cause formation of stones throughout the upper and lower urinary tracts, including the urethra, bladder, ureters, and kidneys. These stones can result in pain, bloody urine, infection and blockage of the urinary tract.

## Xanthinuria (Discovered in the Toy Manchester Terrier)

Xanthinuria can cause formation of stones throughout the upper and lower urinary tracts, including the urethra, bladder, ureters, and kidneys. These stones can result in pain, bloody urine, infection and blockage of the urinary tract.

## von Willebrand's Disease, type 1

von Willebrand's Disease (vWD) type 1 is a clotting disorder that usually causes mild bleeding tendencies in affected dogs though some may have more severe signs. The low level of von Willebrand's factor impacts the bloods clotting ability.

## von Willebrand's Disease, type 2

von Willebrand's Disease (vWD) type 2 is a blood clotting disorder that causes moderate to severe bleeding tendencies due to low level and abnormal structure of von Willebrand's factor.

## von Willebrand's Disease, type 3 (Discovered in the Kooiker Hound)

von Willebrand's Disease (vWD) Type 3 is a clotting disorder that causes severe bleeding tendencies in affected dogs.

## von Willebrand's Disease, type 3 (Discovered in the Scottish Terrier)

von Willebrand's Disease (vWD) Type 3 is a clotting disorder that causes severe bleeding tendencies in affected dogs.

## von Willebrand's Disease, type 3 (Discovered in the Shetland Sheepdog)

von Willebrand's Disease (vWD) Type 3 is a clotting disorder that causes severe bleeding tendencies in affected dogs.

