

Collie Eye Anomaly

Aliases: Choroidal hypoplasia, CEA, CH

Is also known as choroidal hypoplasia (CH), is an inherited disease affecting several dog breeds. The choroid is the layer of tissue in the eye responsible for supplying blood and nutrients to the [Retina](#). In dogs affected with CEA, the choroid does not develop properly and is therefore thinner than normal. The severity of the condition can vary from dog to dog. In mild cases, affected dogs may only show signs of collie eye anomaly on eye exam between about 5 and 12 weeks of age, just prior to normal, age-related pigmentation of the retina which often masks the characteristic, disease-related changes. After this time period, mildly affected dogs may be impossible to distinguish from normal dogs on eye exam (a phenomenon often referred to as "going normal") and may not display obvious vision deficits. In more severely affected dogs, clinical signs include malformations of the eye and/or optic nerve (colobomas), retinal detachment, intraocular bleeding, and subsequent blindness. Both mild and severe forms of CEA are associated with the same NHEJ1 gene [Mutation](#). Therefore, predicting the potential severity of the disease in an affected puppy is difficult as mildly affected parents may produce offspring that are severely affected.

Breed-Specific Information for the Toy Australian Shepherd

Toy Australian shepherd is included as a breed susceptible to collie eye anomaly because of its close relatedness to the Australian shepherd breed, which is known to develop this disease due to [Mutation](#) of the NHEJ1 gene. The frequency of the causal mutation in the general toy Australian shepherd population is unknown.

Testing Tips

Genetic testing of the NHEJ1 gene in toy Australian shepherd dogs will reliably determine whether a dog is a genetic [Carrier](#) of collie eye anomaly. Collie Eye Anomaly is inherited in an [Autosomal Recessive](#) manner in dogs meaning that they must receive two copies of the mutated gene (one from each parent) to develop the disease. In general, carrier dogs do not have features of the disease but when bred with another carrier of the same [Mutation](#), there is a risk of having affected pups. Each pup that is born to this pairing has a 25% chance of inheriting the disease and a 50% chance of inheriting one copy and being a carrier of the NHEJ1 gene mutation. Reliable genetic testing is important for determining breeding practices. In order to eliminate this mutation from breeding lines and to avoid the potential of producing affected pups, breeding of known carriers to each other is not recommended. Toy Australian shepherd dogs that are not carriers of the mutation have no increased risk of having affected pups.

Cone Degeneration

Aliases: Achromatopsia, Rod monochromacy, Day blindness, Hemeralopia, CD

is an inherited eye disease affecting dogs. Affected dogs develop day blindness (blindness in bright light) and [Photophobia](#) (light sensitivity) between 8 to 12 weeks after birth due to degeneration of cells in the eye called cone photoreceptors which are responsible for vision in bright light. Affected dogs have normal vision in low light and structures of the inner eye appear normal on eye exam. Normal cone cell function can be seen on [Electroretinogram](#) (ERG) before six weeks of age, but becomes abnormal between 6 to 12 weeks of age and is completely absent in affected adult dogs signifying complete loss of [Cone Cells](#). The cells responsible for vision in low light called [Rod](#) photoreceptors are not affected and thus, affected dogs will still be able to see normally in low light throughout life.

Breed-Specific Information for the Toy Australian Shepherd

Toy Australian Shepherd is included as a breed susceptible to Cone Degeneration because of its close relatedness to the Miniature Australian Shepherd breed, which is known to develop this disease due to [Mutation](#) of the CNGB3 gene. The frequency of the causal mutation in the general Toy Australian Shepherd population is unknown.

Testing Tips

Genetic testing of the CNGB3 gene in Toy Australian Shepherds will reliably determine whether a dog is a genetic [Carrier](#) of cone degeneration. Cone Degeneration is inherited in an [Autosomal Recessive](#) manner in dogs meaning that they must receive two copies of the mutated gene (one from each parent) to develop the disease. **In general, carrier dogs do not have features of the disease but when bred with another carrier of the same [Mutation](#), there is a risk of having affected pups. Each pup that is born to this pairing has a 25% chance of inheriting the disease and a 50% chance of inheriting one copy and being a carrier of the CNGB3 gene mutation.** Reliable genetic testing is important for determining breeding practices. In order to eliminate this mutation from breeding lines and to avoid the potential of producing affected pups, breeding of known carriers to each other is not recommended. Toy Australian Shepherds that are not carriers of the mutation have no increased risk of having affected pups.

Degenerative Myelopathy

Aliases: Canine degenerative myelopathy, DM

is an inherited neurologic disorder caused by a [Mutation](#) of the SOD1 gene in dogs. This mutation is found in many breeds of dog, though it is not clear for some breeds whether all dogs carrying two copies of the mutation will develop the disease. The variable presentation between breeds suggests that there are environmental or other genetic factors responsible for modifying disease expression. The average age of onset for dogs with degenerative myelopathy is approximately nine years of age. Affected dogs usually present in adulthood with gradual muscle [Atrophy](#) and loss of coordination typically beginning in the hind limbs due to degeneration of the nerves. The condition is not typically painful for the dog, but will progress until the dog is no longer able to walk. The gait of dogs affected with degenerative myelopathy can be difficult to distinguish from the gait of dogs with hip dysplasia, arthritis of other joints of the hind limbs, or intervertebral disc disease. Late in the progression of disease, dogs may lose fecal and urinary continence and the forelimbs may be affected. Affected dogs may fully lose the ability to walk 6 months to 2 years after the onset of symptoms. Affected small breed dogs often progress more slowly than affected large breed dogs and owners may postpone euthanasia until the dog is paraplegic.

Breed-Specific Information for the Toy Australian Shepherd

The toy Australian shepherd is listed as a breed susceptible to degenerative myelopathy because of its close relatedness to the Australian shepherd, which is known to develop this disease due to [Mutation](#) of the SOD1 gene. It is unknown if the toy Australian shepherd develops degenerative myelopathy due to this mutation.

Testing Tips

Genetic testing of the SOD1 gene in toy Australian shepherds will reliably determine whether a dog is a genetic [Carrier](#) of degenerative myelopathy. Degenerative Myelopathy is inherited in an [Autosomal Recessive](#) manner in dogs meaning that they must receive two copies of the mutated gene (one from each parent) to develop the disease. **In general, carrier dogs do not have features of the disease but when bred with another carrier of the same [Mutation](#), there is a risk of having affected pups. Each pup that is born to this pairing has a 25% chance of inheriting the disease and a 50% chance of inheriting one copy and being a carrier of the SOD1 gene mutation.** Reliable genetic testing is important for determining breeding practices. Because symptoms may not appear until adulthood and some at-risk/affected dogs do not develop the disease, genetic testing should be performed before breeding. Until the exact modifying environmental or genetic factor is determined, genetic testing remains the only reliable way to detect neurological disease associated with this mutation prior to death. In order to eliminate this mutation from breeding lines and to avoid the potential of producing affected pups, breeding of known carriers to each other is not recommended. Toy Australian shepherds that are not carriers of the mutation have no increased risk of having affected pups.

Hereditary cataracts

Aliases: Juvenile cataracts, Early onset cataracts, HC, JC, HSF4

(Australian shepherd type) is an inherited eye disease affecting dogs. Cataracts are opacities in the lens of the eye caused by structural changes in lens proteins. A normal lens allows light to pass through it to the [Retina](#) in the back of the eye. Light cannot pass through the parts of the lens affected by cataracts and vision becomes blurry. Dogs with Hereditary cataracts (Australian shepherd type) most commonly present between 2 to 7 years of age with small cataracts that are visible on a veterinary eye exam. In dogs that inherit one copy of the [Mutation](#), cataracts develop slowly, sometimes leading to complete blindness. However, it has been speculated that dogs carrying two copies of the mutation are more likely to develop a more rapidly progressing and severe [Cataract](#). Of note, not all forms of cataracts are inherited and environmental factors such as UV damage can also play a role in the severity of disease. This specific mutation in the HSF4 gene shows [Incomplete Penetrance](#), meaning that not all dogs inheriting two copies of the mutation develop clinical disease. This suggests that other unknown genetic or environmental factors may play a role in modifying disease development and progression.

Breed-Specific Information for the Toy Australian Shepherd

The toy Australian shepherd is included as a breed susceptible to hereditary cataracts (Australian shepherd type) because Australian shepherds are known to develop this disease due to [Mutation](#) of the HSF4 gene. The frequency of the causal mutation in the overall toy Australian shepherd population is unknown. However, in one study of 392 Australian shepherds with and without cataracts from North America and Europe, 25.5% were carriers of the mutation and 3.8% had two copies of the mutation. In this same study, Australian shepherds with this mutation had an approximately 17-fold increased risk of developing cataracts.

Testing Tips

Genetic testing of the HSF4 gene in toy Australian shepherds will reliably determine whether a dog is a genetic [Carrier](#) of hereditary cataracts (Australian shepherd type). **Hereditary cataracts (Australian shepherd type) is inherited in an [Autosomal Dominant](#) manner in dogs meaning that dogs only need to inherit one copy of the mutated gene to be at-risk for the disease. Dogs that inherit two copies of the genetic [Mutation](#) are at-risk of developing a more severe form of the disease. Each pup that is born to a parent carrying one copy of the mutation has a 50% chance of inheriting one copy of the HSF4 gene mutation and being at-risk for the disease. If both parents are affected, the chance of having affected offspring increases to 75-100%.** Because symptoms may not appear until adulthood and not all dogs with the mutation develop disease, genetic testing should be performed before breeding. Reliable genetic testing is important for determining breeding practices. In order to eliminate this mutation from breeding lines and to avoid the potential of producing affected pups, breeding of known carriers is not recommended. Toy **Australian shepherds that are not carriers of the mutation have no increased risk of having affected pups due to this mutation.**

Hyperuricosuria

Aliases: Urolithiasis, HUU

is an inherited condition of the urinary system affecting many breeds of dog, including Australian Shepherds. The SLC2A9 gene codes for a protein that allows the kidneys to transport uric acid from the urine. Dogs with mutations in both copies of the SLC2A9 gene are predisposed to have elevated levels of uric acid in the urine, hence the name hyperuricosuria. Uric acid can form crystals and/or stones (uroliths) in the urinary tract. Dogs with hyperuricosuria most commonly present with symptoms of recurrent urinary tract inflammation, which include frequent urination, blood in the urine, and straining to urinate. They may also have loss of appetite, lethargy, weakness, vomiting and pain. Urinary stones in the bladder can cause urinary tract infections or more seriously, blockage of the [Urethra](#). Both male and female dogs can be affected, but obstruction of urine flow is more common in males due to differences in anatomy. Although an x-ray can be used to exclude other types of stones, urate stones cannot typically be seen using x-rays and must be evaluated by ultrasound. Not all dogs with mutations in both copies of the SLC2A9 gene will have symptoms of disease, though they have increased uric acid excretion in the urine.

Breed-Specific Information for the Toy Australian Shepherd

Toy Australian Shepherd is included as a breed susceptible to hyperuricosuria because of its close relatedness to the Australian Shepherd breed, which is known to develop this disease due to [Mutation](#) of the SLC2A9 gene. The frequency of the causal mutation in the general Toy Australian Shepherd population is unknown.

Testing Tips

Genetic testing of the SLC2A9 gene in Toy Australian Shepherds will reliably determine whether a dog is a genetic [Carrier](#) of hyperuricosuria. Hyperuricosuria is inherited in an [Autosomal Recessive](#) manner in dogs meaning that they must receive two copies of the mutated gene (one from each parent) to develop the disease. **In general, carrier dogs do not have features of the disease but when bred with another carrier of the same [Mutation](#), there is a risk of having affected pups. Each pup that is born to this pairing has a 25% chance of inheriting the disease and a 50% chance of inheriting one copy and being a carrier of the SLC2A9 gene mutation.** Reliable genetic testing is important for determining breeding practices. Because not all affected dogs will have clinical signs associated with hyperuricosuria, genetic testing should be performed before breeding. In order to eliminate this mutation from breeding lines and to avoid the potential of producing affected pups, breeding of known carriers to each other is not recommended. Toy Australian Shepherds that are not carriers of the mutation have no increased risk of having affected pups.

Multifocal Retinopathy 1

Aliases: Canine multifocal retinopathy 1, CMR1

is an inherited disorder of the [Retina](#) affecting multiple breeds of dog. Affected dogs typically present between 11 and 16 weeks of age with multiple discrete circular areas of retinal detachment with underlying fluid accumulation that are visible on an eye exam performed by a veterinarian. These blister-like lesions are typically found in both eyes and can appear gray, tan, orange, or pink and vary in number, size and location. Progression of retinal changes is usually slow and new lesions are not noted after 6 to 12 months of age. Occasionally as affected dogs age, lesions appear to heal and are no longer visible on an eye exam. Generally the dog's vision is not affected although vision loss has been described in some cases of multifocal retinopathy 1.

Breed-Specific Information for the Toy Australian Shepherd

Toy Australian Shepherd is included as a breed susceptible to multifocal retinopathy 1 because of its close relatedness to the Australian Shepherd breed, which is known to develop this disease due to [Mutation](#) of the BEST1 gene. The frequency of the causal mutation in the general Toy Australian Shepherd population is unknown.

Testing Tips

Genetic testing of the BEST1 gene in Toy Australian Shepherds will reliably determine whether a dog is a genetic [Carrier](#) of multifocal retinopathy 1. Multifocal Retinopathy 1 is inherited in an [Autosomal Recessive](#) manner in dogs meaning that they must receive two copies of the mutated gene (one from each parent) to develop the disease. **In general, carrier dogs do not have features of the disease but when bred with another carrier of the same [Mutation](#), there is a risk of having affected pups. Each pup that is born to this pairing has a 25% chance of inheriting the disease and a 50% chance of inheriting one copy and being a carrier of the BEST1 gene mutation.** Reliable genetic testing is important for determining breeding practices. Because visual deficits are generally not noted and lesions can regress as affected dogs age, genetic testing should be performed before breeding. In order to eliminate this mutation from breeding lines and to avoid the potential of producing affected pups, breeding of known carriers to each other is not recommended. Toy Australian Shepherds that are not carriers of the mutation have no increased risk of having affected pups.

Neuronal Ceroid Lipofuscinosis 6

Aliases: Batten disease, Amaurotic idiocy, NCL6, NCL

is a lysosomal storage disease affecting dogs. Affected dogs lack a specific [Enzyme](#) necessary for normal metabolism. As a result, there is an abnormal accumulation of waste compounds primarily in the cells of the nervous system, leading to a range of nervous system disorders. Affected dogs typically present around 1.5 years of age with progressive neurologic disease. Symptoms include loss of vision, behavioral change, anxiety, lack of muscle coordination and abnormal gait. Affected dogs are often humanely euthanized by 2 years of age due to progression of the disease.

Breed-Specific Information for the Toy Australian Shepherd

Toy Australian Shepherd is included as a breed susceptible to neuronal ceroid lipofuscinosis 6 because of its close relatedness to the Australian Shepherd breed, which is known to develop this disease due to [Mutation](#) of the CLN6 gene. The frequency of the causal mutation in the general Toy Australian Shepherd population is unknown.

Testing Tips

Genetic testing of the CLN6 gene in Toy Australian Shepherds will reliably determine whether a dog is a genetic [Carrier](#) of neuronal ceroid lipofuscinosis 6. Neuronal Ceroid Lipofuscinosis 6 is inherited in an [Autosomal Recessive](#) manner in dogs meaning that they must receive two copies of the mutated gene (one from each parent) to develop the disease. In general, carrier dogs do not have features of the disease but when bred with another carrier of the same [Mutation](#), there is a risk of having affected pups. Each pup that is born to this pairing has a 25% chance of inheriting the disease and a 50% chance of inheriting one copy and being a carrier of the CLN6 gene mutation. Reliable genetic testing is important for determining breeding practices. In order to eliminate this mutation from breeding lines and to avoid the potential of producing affected pups, breeding of known carriers to each other is not recommended. Toy Australian Shepherds that are not carriers of this mutation have no increased risk of having affected pups due to this mutation.

Progressive retinal [Atrophy](#), progressive [Rod](#)-cone degeneration

Aliases: PRCD, PRA-PRCD

is a late onset, inherited eye disease affecting many breeds of dog. PRA-prcd occurs as a result of degeneration of both rod and cone type [Photoreceptor Cells](#) of the [Retina](#), which are important for vision in dim and bright light, respectively. Evidence of retinal disease in affected dogs can first be seen on an [Electroretinogram](#) around 1.5 years of age for most breeds, but most affected dogs will not show signs of vision loss until 3 to 5 years of age or later. The rod type cells are affected first and affected dogs will initially have vision deficits in dim light (night blindness) and loss of peripheral vision. Over time affected dogs continue to lose night vision and begin to show visual deficits in bright light. Other signs of progressive retinal atrophy involve changes in reflectivity and appearance of a structure behind the retina called the [Tapetum](#) that can be observed on a veterinary eye exam. Although there is individual and breed variation in the age of onset and the rate of disease progression, the disease eventually progresses to complete blindness in most dogs. Other inherited disorders of the eye can appear similar to PRA-prcd. Genetic testing may help clarify if a dog is affected with PRA-prcd or another inherited condition of the eye.

Breed-Specific Information for the Toy Australian Shepherd

Toy Australian Shepherd is included as a breed susceptible to progressive retinal [Atrophy](#), progressive [Rod](#)-cone degeneration because of its close relatedness to the Miniature Australian Shepherd breed, which is known to develop this disease due to [Mutation](#) of the PRCD gene. The frequency of the causal mutation in the general Toy Australian Shepherd population is unknown.

Testing Tips

Genetic testing of the PRCD gene in Toy Australian Shepherds will reliably determine whether a dog is a genetic [Carrier](#) of PRA-prcd. PRA-prcd is inherited in an [Autosomal Recessive](#) manner in dogs meaning that they must receive two copies of the mutated gene (one from each parent) to develop the disease. In general, carrier dogs do not have features of the disease but when bred with another carrier of the same [Mutation](#), there is a risk of having affected pups. Each pup that is born to this pairing has a 25% chance of inheriting the disease and a 50% chance of being a carrier of the PRCD gene mutation. Reliable genetic testing is important for determining breeding practices. Because symptoms do not appear until adulthood, genetic testing should be performed before breeding. In order to eliminate this mutation from breeding lines and to avoid the potential of producing affected pups, breeding of known carriers to each other is not recommended. Toy Australian Shepherds that are not carriers of the mutation have no increased risk of having affected pups. However, because there are multiple types of PRA caused by mutations in other genes, a normal result in PRCD does not exclude PRA in a pedigree

Multidrug Resistance 1

Aliases: Multidrug sensitivity, Ivermectin sensitivity, MDR1 gene defect, MDR1

is an inherited condition affecting several breeds of dogs, especially herding dogs such as the Toy Australian Shepherd. The [Mutation](#) in the ABCB1 gene associated with MDR1 causes dysfunction of P-glycoprotein, which is responsible for removing certain drugs and toxins from the body. Clinical signs are most commonly associated with distribution of the drug in the central nervous system. If an at-risk dog is treated with one of several common drugs (see below*), they are at risk of developing neurologic symptoms that could range from tremors, excess salivation, anorexia and blindness to coma and even death. Because of the defective ability to metabolize specific drugs, these drugs can be lethal even at low doses. The MDR1 mutation does not cause adverse effects in dogs unless the dog is exposed to these drugs. Therefore, veterinarians should be notified when a dog is at risk for multidrug resistance 1 prior to administration of any medications.

***Drugs known to cause neurological signs related to the MDR1 mutation:**

Acepromazine, butorphanol, doxorubicin, emodepside, erythromycin, ivermectin, loperamide, milbemycin, moxidectin, rifampin, selamectin, vinblastine and vincristine

In addition to this list, there are many other drugs known to be removed from the central nervous system via the P-glycoprotein mechanism in humans. However, reports of neurological dysfunction related to drugs other than those listed here are scarce in dogs. Please consult your veterinarian when giving drugs to known multidrug resistance 1 carriers, affected dogs, or untested dogs of breeds commonly affected with this condition.

Breed-Specific Information for the Toy Australian Shepherd

Toy Australian Shepherd is included as a breed susceptible to multidrug resistance 1 because of its close relatedness to the Miniature Australian Shepherd breed, which is known to develop this disease due to [Mutation](#) of the ABCB1 gene. The frequency of the causal mutation in the general Toy Australian Shepherd population is unknown.

Testing Tips

Genetic testing of the ABCB1 gene in Toy Australian Shepherds will reliably determine whether a dog is a genetic [Carrier](#) of multidrug resistance 1. Multidrug Resistance 1 is inherited in an autosomal incomplete dominant manner in dogs meaning that dogs only need to inherit one copy of the mutated gene to be at an increased risk of developing the disease. Though adverse reactions to certain drugs are most commonly seen in dogs having two copies of the mutated gene, carrier dogs can also experience drug sensitivities and dosages need to be adjusted accordingly. Thus, dogs that have one or two mutant copies of the gene are considered at-risk for adverse drug reactions.

When carriers of this [Mutation](#) are bred with another dog that also is a carrier of the same mutation, there is risk of having affected pups. For each pup that is born

to this pairing, there is a 25% chance that the puppy will inherit two copies of the mutation and a 50% chance that the puppy will inherit one copy of the mutation and, in either case, may be susceptible to having adverse drug reactions. Reliable genetic testing is important for determining breeding practices. Because symptoms do not appear unless dogs are exposed to certain drugs, genetic testing should be performed before breeding. In order to eliminate this mutation from breeding lines and to avoid the potential of producing affected pups, breeding of known carriers is not recommended. Toy Australian Shepherds that are not carriers of the mutation have no increased risk of having affected pups when bred to a dog that is also clear for this mutation.

www.MistysToyAussies.com