Genetic Diseases

Collie Eye Anomaly (CEA) aka choroidal hypoplasia
(NHEJ1 Gene mutation)

The choroid of the eye is thinner than normal, varying in severity from dog to dog. In mild cases signs only show on eye exams under 12 weeks of age, after this it is difficult sometimes impossible to distinguish these dogs from normal dogs on eye exams. This is referred to as going normal.

In dogs with more severe cases the signs are more serious such as malformations of the eye or optic nerve. Signs can be colobomas, intraocular bleeding, retinal detachment, and blindness. It's impossible to tell the severity in which a dog will be effected just by testing. But testing can help to prevent continuing to reproduce this mutation in future dogs. Carriers should not be bred to other carriers or affected dogs.

Cone Degeneration (CD) aka Day blindness
(CNGB3 Gene mutation)

Affected dogs develop Blindness in bright lights and light sensitivity around 8 to 12 weeks of age. This is due to the degeneration of cells in the eye. Affected dogs (Those carrying two copies of the mutation) are able to see normally in low lighting throughout life. Avoid breeding carriers to other carriers or affected dogs.

Hereditary Cataracts (HC)
(HSF4 Gene Mutation)

Cataracts are opacities (cloudy/non clear spots) in the eye's lens caused by structural changes in lens proteins. These block the flow of light causing blurred vision and overtime can lead to blindness. Dogs carrying one copy of this should not be bred and are at risk of cataracts. Dogs carrying two copies are at risk of developing fast developing severe cataracts.

Degenerative Myelopathy (DM)
(SOD1 Gene Mutation)

Degenerative Myelopathy is a neurological disorder that generally appears around 9 years of age. It leads to fecal and urinary incontinence, and eventually loss of the ability to walk. It progresses slower is small breeds such as the toy Australian Shepherd usually taking several years. Because of this late onset dogs with DM typically will pass from old age before it progresses to losing the ability to walk, and becoming incontinent. Dogs carrying two copies are considered affected, while one copy is a carrier. A carrier should not be bred to another carrier or affected dog.

Progressive Retinal Atrophy, Progressive Rod-Cone Degeneration (PRA-PRCD)
(PRCD Gene Mutation)

PRA-PRCD is an eye disease that has a late onset in dogs, typically around age 5 or later. Generally the dogs will become night blind (have difficulty seeing is low light) and lose peripheral vision. As it progresses it can lead to total blindness. To prevent producing dogs affected (carrying 2 copies of the mutation) by PRA-PRCD it is important to avoid breeding carriers to other carriers or affected dogs.

Multifocal Retinopathy 1 (CMR1)
(BEST1 Gene Mutation)

CMR1 usually presents itself between 12 and 16 weeks, it is a disorder of the retina starting with blister like lesions developing in areas where the retinas detach. Usually progression of the disease stops around one year of age. In most cases it will not result in blindness, but it is possible. To prevent producing dogs affected (carrying 2 copies of the mutation) by CMR1 it is important to avoid breeding carriers to other carriers or affected dogs.

Hyperuricosuria (HUU)
(SLC2A9 Gene Mutation)

Affected dogs have elevated levels of uric acid in the urine, which can lead to crystals and stones forming in the urinary tract. Signs are usually difficulty urinating, urinating frequently, or blood in the urine.

MDR1: Multi-Drug Resistance Gene
(ABCB1 Gene Mutation)

Short Version:

MDR1 is a gene that causes a dangerous sensitivity to some commonly used medications such as Ivermectin. Ivermectin is found in commonly used medications such as Heartgard. About 50% of Australian Shepherds are affected by MDR1.

Longer more detailed version:

MDR1 is a gene responsible for the production of a protein, P-glycoprotein (P-GP). P-GP is a drug efflux pump that aids in controlling drug absorption and distribution specifically in the brain. This causes increased levels of the medications inside the cells.

Imagine a partially clogged drain; when you run water into the sink, the water will build up in the sink similar to the way the medications do in a cells of an MDR1 affected animal.

***Please refer to MDR1 file for a list of medications to avoid

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