

INHERITED DISEASES

While the genetics of many inherited conditions are simple recessives as, not all are.

Let's start with the ones that are autosomal recessive traits.

Bleeding Disorders

Von Willebrand Disease Type I (VWF).

This disease is inherited as an **autosomal recessive**. BUT while it is an inherited bleeding disorder and does affect many breeds of dogs. not all are affected the same. Those affected with VWDI have less than half of the normal level of **von Willebrand coagulation factor (vWf)**, an essential protein needed for normal blood clotting.

Not all dogs with two copies of the mutation are equally affected. Dogs that have less than 35% of the normal amount of vWf usually have mild to moderate signs and may bruise easily, have frequent nosebleeds, bleed from the mouth when teething, and have prolonged bleeding after surgery or trauma. Rarely is the bleeding severe enough to cause death. Most dogs will have a normal lifespan despite increased blood clotting times.

Factor VII Deficiency (F7 Exon 5)

This also an **autosomal recessive**. Affected dogs do not make enough Factor VII protein, an essential for the blood coagulation process. Affected dogs may not show any symptoms normally, but may bleed excessively after surgery or an injury.

Ophthalmic Conditions

Many ophthalmic conditions are also **autosomal recessives**. This includes the two mentioned by Beth in the Newsletter : **Canine Multifocal Retinopathy cmr1** (BEST1 Exon 2) and **Progressive Retinal Atrophy - prcd** Progressive rod-cone degeneration (PRCD Exon 1)

Canine Multifocal Retinopathy

This mutation causes raised lesions on the retina which changes the appearance in the inside of the eye, but rarely affects sight. This usually appears when puppies are only a few months old and generally do not worsen over time. The lesions may result in minor folding of the retina or disappear.

Progressive Retinal Atrophy – prcd Progressive rod-cone degeneration (PRCD Exon 1)

PRA stands for a group of disease that cause the retina to degenerate over time. This results in declining vision and eventually blindness. Prcd refers to progressive rod-cone degeneration, a type of PRA.

With this problem, cells in the back of the eye on the retina that seem to develop normally early in life, degenerate and die. Rod cells are the first to lose normal function. The result is night

blindness as these cells operate in low levels of light. The Cone cells function in full light and when they fail, the dog will eventually be blind. Blindness from other conditions can appear similar to PRA so an exam by a veterinary ophthalmologist as well as the genetic test will help with the diagnosis. There is no treatment.

The Other Autosomal recessive Condition

Juvenile laryngeal paralysis and polyneuropathy polyneuropathy complex, congenital laryngeal paralysis or JLPP.

This is also an autosomal recessive condition. When excited or exercised affected dogs have difficulty breathing, may have a change in their bark and develop weakness with loss of coordination in the hind legs and then also the front legs also. Difficulty swallowing followed by choking or pneumonia may occur. Eye problems are common. There is no treatment and most dogs die by 6 months or are euthanized.

The Autosomal Dominant one

Malignant Hyperthermia (MH)

Malignant Hyperthermia is an autosomal dominant genetic mutation that causes a dog to have dangerous physical reactions in response to specific triggers which include exposure to certain drugs, most notably the inhaled anesthetics like halothane. Other triggers include the ingestion of food ingredients such as caffeine and hops as well as too much exercise. Because the condition can become apparent when a dog is under stress or over-stimulated MH is also known as "canine stress syndrome". Because the mutation is autosomal dominant, only one copy of the mutation is necessary to produce the problem.

When the affected dog experiences one of the triggers it can have extreme muscle contractions, increased metabolism, rapid heartbeat and elevated body temperature. The body produces too much carbon dioxide, and enters a hypermetabolic state. Muscles become rigid and stiff and a seizure may result.

A dog with the MH mutation may not survive undergoing anesthesia using halothane or other types of inhaled anesthesia. There are some forms of anesthesia that can be safely used to sedate dogs affected with MH. There is no cure for Malignant Hyperthermia, but dogs with this mutation can avoid stressful situations, intense exercise, and food and drugs that trigger symptoms.

For information on DNA testing check the AKC-CHF site including the article [Responsible Use of DNA Testing](http://www.akcchf.org/news-events/news/responsible-use-of-dna.html) at <http://www.akcchf.org/news-events/news/responsible-use-of-dna.html>