

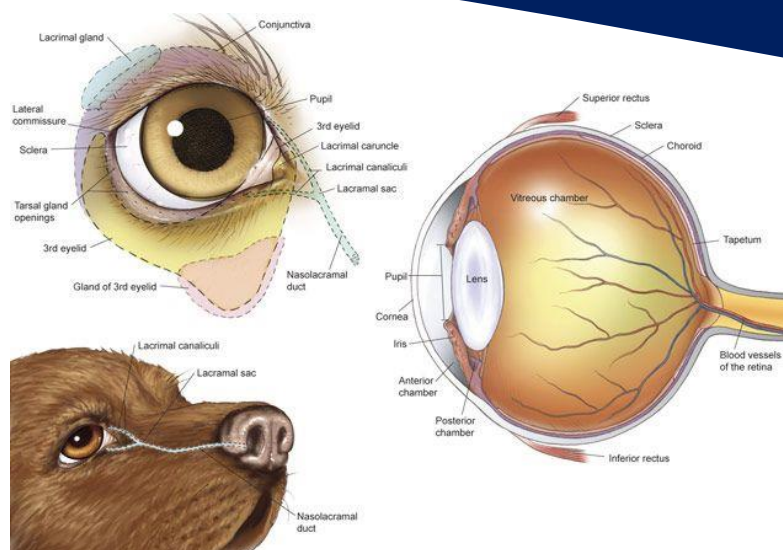
## Hereditary Eye problems

There are various eye conditions that Golden Retrievers are susceptible to some serious and sight affecting such as, Progressive Retinal Atrophy (PRA) and Hereditary Cataracts (HC) and other conditions that don't affect the sight such as, Multifocal Retinal Dysplasia (MRD) and Post Polar Cataract (PPC). In Australia, there is a yearly screening program for eyes and breeders are strongly recommended to use this.

## Progressive retinal atrophy

**Progressive retinal atrophy (PRA)** is a group of inherited eye disorders that occur in many different dog breeds that is characterised by vision loss due to degeneration of the photoreceptor cells in the retina, eventually leading to complete blindness. To date, more than 20 different mutations causing canine-PRA have been described and several breeds including the Golden Retriever are affected by more than one form of PRA (*André C et al 2008*). Genetically distinct forms of PRA may have different clinical characteristics such as rate of progression and age of onset. In the Golden Retriever it is thought that PRA is caused by at least four mutations three of which have been reported to cause PRA in Golden Retrievers (prcd-PRA, GR\_PRA1 and GR\_PRA2).

Most PRA cases in the Golden Retriever are clinically indistinguishable from PRA cases of other breeds. The mode of inheritance appears to be consistent with autosomal recessive and the age of diagnosis is most commonly at approximately 5 years of age (*Downs et al 2011*).



The most common form of PRA identified in dogs in general is caused by a mutation in the Progressive rod cone degeneration gene (Prcd-PRA) (*Zangerl B, et al 2006*). The PRCD-mutation has also been associated with PRA in a small number of PRA-affected Golden Retrievers. However, the majority (61%) of cases of PRA in the Golden Retriever are caused by a mutation in the SLC4A3 gene and known as GR\_PRA1 (*Downs et al 2011*). Recently, a single base deletion in the TTC8 gene was identified to cause PRA in the Golden Retriever and is known as GR\_PRA2. This third mutation which is fully penetrant and a common cause of PRA in the breed does not explain all remaining cases, suggesting that there is at least one more genetically distinct form of PRA in the Golden Retriever (*Downs et al 2014*).

Patented commercial genetic tests for both prcd-PRA and GR\_PRA1 are available. These tests identify those dogs with one copy of the mutation (carrier) and dogs with two copies of the mutation (affected). By ensuring that at least one parent is Normal/Clear of GR\_PRA1 or prcd-PRA then no GR\_PRA1/prcd-PRA affected offspring will be produced in a mating. See the chart below for expected breeding outcomes when the GR\_PRA1/prcd-PRA status has been determined by DNA testing.

Expected Breeding Outcomes with known pcrd-PRA/GR1_PRA status			
Parent 1 Status	Parent 2 Status		
	Normal/Clear	Carrier	Affected
Normal/Clear	All Normal	50% Normal 50% Carrier	All Carrier
Carrier	50% Normal 50% Carrier	25% Normal 50% Carrier 25% Affected	50% Carrier 50% Affected
Affected	All Carrier	50% Carrier 50% Affected	All Affected

## Hereditary Cataracts

**Hereditary Cataracts (HC)** Cataracts are simply defined as opacities of the lens and can develop for a variety of reasons, including advanced age and the secondary effects of other diseases such as diabetes or progressive retinal atrophy, and trauma. Fortunately, the age of onset, appearance and evolution of the cataracts which are hereditary are usually quite specific, enabling inherited cataracts to be distinguished from other non-inherited types of cataract. This condition generally results in an inability to see clearly and can cause total blindness. They usually start by being small and grow progressively, though the speed of growth is highly variable. Some cataracts will grow so slowly that the dog's vision remains relatively clear, while others will grow such a way that the dog will quickly go blind. Corrective surgery is possible, though it is costly and is not always effective (Rubin, 1989).

## Posterior polar subcapsular cataracts

Posterior polar subcapsular cataracts (PPC) better known as the star cataract is a hereditary cataract seen in the golden retriever. It is usually in both eyes and becomes apparent between 6 to 18 months of age. Some dogs may develop this problem as late as 6 to 7 years of age. This cataract may be slowly progressive, but rarely interferes with vision. It is advised not to breed from affected animals as mating's with affected dogs have produced litters of blind pups. Despite the large number of breeds affected by HC only a single gene, the transcription factor *HSF4*, has been implicated in the development of cataracts in dogs to date. However, *HSF4* has been excluded from involvement in the development of PPC in the Golden Retriever (Nakai et al 1997).

## Multifocal Retinal Dysplasia

**Retinal Dysplasia (RD)** is a congenital/neonatal condition characterised by abnormal development of the retina. The disorder can be inherited, or it can be acquired as a result of a viral infection or some other event before the pups were born. The normal retina lines the back of the eye. The retinal cells receive light stimuli from the external environment and transmit the information to the brain where it is interpreted to become vision. Retinal dysplasia results in extremely varied clinical and microscopic appearances so that, for example, folds, ridges, rosettes, geographic abnormalities and localised detachments are all possible manifestations of retinal dysplasia.

### **Multifocal Retinal Dysplasia (MRD)**

although not painful nor progressive, refers to small folds or rosettes within the retinal tissue, either singularly or multiple. Litter screening is useful, although subtle changes are not always clearly defined. In older animals remodelling of some or all multifocal lesions may result in them becoming less obvious, even disappearing, over time. Irregularly shaped (geographic) areas of retinal dysplasia may also be encountered instead of or alongside of folds in the retinal tissue. While focal or multifocal folds may lessen or disappear as the dog ages, geographic retinal lesions will not (Crispin et al., 1999; Long et al., 1999).

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