



United Kingdom Miniature American Shepherd Club

Health Issues and Health Testing in the MAS

As the Kennel Club does not yet recognise the MAS, there are no specific testing requirements or recommendations listed. In the interim UKMASC recommends using the health testing requirements (and tests) of the Australian Shepherd, since that is the breed from which all our stock derives.

Section A: The KC list the following tests as mandatory for Assured Breeders of aussies: BVA/KC Hip Dysplasia Scheme DNA test - HC-HSF4 BVA/KC/ISDS Eye Scheme - annual test
Section B:
In addition, the KC strongly recommended that all breeders should test for
BVA/KC Elbow Dysplasia Scheme
DNA test - CEA/CH
DNA test - prcd-PRA
DNA test - MDR1
DNA test - Cobalamin Malabsorption
The Australian Shepherd Breed Club also recommends testing for
Pelger-Huet Anomaly.

Section A: Mandatory Tests

* BVA Hip Scoring - (British Veterinary Association, UK) or OFA Certification - (Orthopaedic Foundation for Animals, USA)

The term Hip Dysplasia covers a number of developmental problems and degenerative abnormalities involving the hip joint. It's a common disease found in most breeds of dogs and probably has both genetic and environmental components.

BVA (or OFA, if your dog originated in the USA) certification involves a vet taking X-rays of the dog's hip joints, with the dog lying in a specific way. The X-ray is then sent to the BVA or OFA for evaluation by a panel of specialist vets and the dog is given a score or a 'grade'.

With the BVA the dog's X-ray is scored out of 53 points for each hip, giving a total possible score for the dog of 106. Quite simply; the lower the score, the better the hip structure. Scores are in the form (A : B) where A and B are the score for each hip, added together to give a total score of C. The score for each hip (A and B) should match as closely as possible - to balance the dog - and dogs should not be bred from if their total score (C) is significantly above the breed average.

The BVA will accept X-rays for scoring from dogs one year of age and older, although MAS are a slow-maturing breed and X-rays taken before two years of age should not be scored because they do not represent the final, adult structure and the results are rendered less reliable. Once a BVA certificate for hip dysplasia grading has been issued, the dog cannot

be resubmitted for grading.

OFA grades of Excellent, Good, and Fair are all considered normal, non-dysplastic grades. Full OFA certification cannot be done until a dog is two years old, for the reasons mentioned above, although a Preliminary Evaluation can be done prior to that. Some USA breeders regard a 'Prelim' result as sufficient testing to breed from.

BVA figures last updated in November 2011 give the mean average hip score for aussies as a total score (C) of 10. With the health of the breed in mind it would be unwise to breed a MAS with a total score more than a couple of points over this, and a partner with a commensurately low score should always be sought.

* HC - HSF4 - Hereditary Cataracts

Cataracts are often first detected during a routine eye exam on a mature adult, but they can sometimes arise in young adulthood or in early old age. Cataracts in very young puppies are extremely unusual.

The typical inherited cataract in herding dogs involves both eyes and is situated on the back of the lens where they usually begin in the outer layer. Some cataracts progress so slowly that the dogs retain functional vision throughout their lives; others become blind quite quickly. One eye may show signs of the disease before the other, but with hereditary cataracts the other eye will most likely develop one within a few months.

The causes of cataracts are many and varied. The HSF4 gene appears to predispose some dogs to developing a certain type of cataract at some point in the dog's lifetime. Having one or even two copies of the HSF4 gene does not guarantee that the dog will get cataracts, the inheritance is only of the tendency to have them.

* BVA/KC/ISDS Eye Scheme - annual test Eye Examination - British Veterinary Association (UK) or CERF Testing - Canine Eye Registration Foundation (USA)

Genetic eye problems are common in almost all breeds of dogs. A BVA test (or CERF if your dog originated in the USA) is performed by a certified veterinary ophthalmologist. During the test, the dog's pupils are fixed open by eye drops and in a darkened room using special optical equipment and lights, the vet looks for approximately 25 different heritable eye defects. A BVA or CERF rating of 'Normal' means that no signs of any of the defects were found.

Not all the eye problems in dogs have a DNA test and some are degenerative, only starting to show up as the dog matures. It is important to have all breeding dogs' eyes checked anually to ensure that nothing is 'sneaking up' otherwise undetected. A copy of the dog's results are sent to the KC and are published on the health section of their website.

Section B : Recommended Tests

BVA/KC

Elbow Dysplasia Scheme

Just as with the hip dysplasia, the term elbow dysplasia (ED) is an umbrella term used to describe a number of specific elbow abnormalities which affect different sites in the joint. Two x-rays of each elbow are taken by the vet, and submitted to the BVA for scoring. It's usual to have the elbows x-rayed at the same time as the hips, to save the dog having to have two lots of sedative or anaesthetic.

The BVA elbow grading scheme is based on that of the International Elbow Working Group (IEWG). Unlike the hips which are graded out of 53 for each side, (106 total possible score) elbows are scored between 0 and 4 for each side. The overall grade is that of the worse of the two elbows. The KC keep a record of BVA/ED results on their health database.





Grade o = a radiographically normal elbow.

Grade 1 = there is no visible primary lesion but secondary new bone (osteoarthritis) up to 2mm in depth is present at any site around the elbow joint.

Grade 2 = (a) a primary lesion is visible (eg. medial coronoid disease or ununited anconeal process) without visible osteoarthritis OR

(b) no primary lesion is visible but osteoarthritis of more than 2mm and up to 5mm in depth is present at any site around the elbow joint.

Grade 3 = (a) both a primary lesion and any amount of osteoarthritis are visible OR (b) no primary lesion is visible but osteoarthritis over 5mm in depth is

present at any site around the elbow joint.

Unlike the hip scores where ideally you're after a low score below the breed average but one or two points here and there are perfectly acceptable, with elbows you really want a score of o. The BVA itself issues a strong recommendation that breeding stock should only have an overall grade of o.

Dogs with elbow grades of 1 are showing mild or early osteoarthritis, which is likely to be due to ED. They should only be used for breeding with caution, taking into consideration the ED grades of as many relatives as possible, as well as the results of other health tests and characteristics.

Dogs with elbow grades of 2 or 3 have marked osteoarthritis likely to be due to ED, with or without a visible primary lesion. These dogs have a significant chance of passing ED on to their offspring, which is something you really don't want to be doing.

CEA-CH - Collie Eye Anomaly

This is the most common of the eye diseases in herding dogs, identified 50 years ago and subsequently found not only in collies from which it took it's name but in most collie relatives including all sizes of aussie. CEA is a 'syndrome', meaning a group of conditions which appear in conjunction with each other. CEA is a structural defect, so dogs are either born with the problem(s) or they aren't - it's not an issue that suddenly develops later in life.

Choroidal Hypoplasia: This is a recessively inherited eye disorder that causes abnormal development of the choroid - an important layer of tissue containing blood vessels, under the retina of the eye. The choroid is pale and thin, almost transparent, and an ophthalmologist looking at the back of the eye (the fundus) with an ophthalmoscope, will see an area of choroidal thinning that appears like a 'window' to the underlying vessels and eye wall. In severe cases Colobomas (small pits or lesions in the eye tissue layers) are seen at and near the optic nerve head. Colobomas can lead to secondary complications such as partial or complete retinal detachments and/or growth of new, abnormal blood vessels which bleed inside the eye.

CH is the most common form of CEA but most often the least harmful and least severe. Most dogs with CH have little or no ill-effects or loss of vision.

Staphyloma, Coloboma, Ectasia: These terms all refer in general to a cupping or bulging in the eyeball, usually in the area of the optic disc. These conditions may or may not be serious, it all depends on the size and location of the bulge. Large colobomas or severe ectasia can lead to retinal detachment.

Vascular Disease: Defects in the blood vessels of the eye which restrict it's blood supply. Vessels may be malformed, undersized, or even missing.

Retinal Detachment: The loosening or separation of the retina from the inside back wall of the eye. This may involve a tiny area or the entire retina and it can be either one or both eyes. If a retina detaches completely, the dog will be blind in that eye.

Both Vascular Disease and Retinal Detachment are conditions that can deteriorate, causing a gradual loss of vision or eventual blindness.

PRA-PRCD - Progressive Retinal Atrophy - Progressive Rod-Cone Degeneration



PRA is a group of diseases that cause the retina of the eye to degenerate slowly over time. The result is declining vision and eventual blindness. A DNA test determines if the dog is 'Clear'(does not carry the gene for the problem), is a 'Carrier' (has one copy of the gene) or is 'Affected' (has two copies of the gene).'Affected' dogs have a very high risk of developing the disease. Carriers do not suffer from the disease, but can pass the gene on to their offspring.

MDR1 - Multi Drug Resistance Gene Mutation 1

Dogs with the mutated MDR1 gene cannot control the absorption of certain drugs into the brain and central nervous system, nor pump them back out again as a normal dog would. With the body unable to remove them, these drugs stay in the nervous system where they accumulate to toxic levels. This causes abnormal neurological symptoms which may result in hospitalisation or even death. The most common problem drug is Ivermectin which is found in many modern anti-parasitic treatments.

MDR1 affects many dogs in the pastoral group including MAS and collies. A DNA test is available to determine if the dog is 'Normal/Normal' ('Clear' and does not carry the gene), is a 'Mutant/Normal' ('Carrier' which has one copy of the gene), or is 'Mutant/Mutant' ('Affected' and has two copies of the gene). Affected dogs will have a severe (sometimes fatal) reaction if given one of the identified drugs. Since Carrier dogs only have one copy of the mutant gene, they may have a reaction if exposed to the identified drugs but the reaction is less severe. They can also pass the gene on to their offspring. A Normal / Clear dog will not have any adverse reaction to the identified drugs. By extrapolating the data from aussies, we know that approximately half of the MAS population carry at least one copy of the MDR1 gene.

Cobalamin Malabsorption – Vitamin B12 deficiency - Imerslund-Grasbeck syndrome

Dogs with Cobalamin Malabsorption cannot manufacture the special transport chemical that would normally bind with Vitamin B12 in the stomach, carry it across the gut wall and release it into the blood stream. This inability to absorb B12 (Cobalamin) from the diet eventually results in a specific deficiency disease when the reserves of B12 the body was born with, are used up.

VitB12 is a vital component in the manufacture of red blood cells, white blood cells and myelin, the fatty material which surrounds and insulates nerve fibres. Without VitB12 the blood, immune and nervous systems gradually collapse. The body becomes anaemic, suffers immune deficiency disorders and repeated infections, anorexia, nervous system disruption, confusion, mal-coordination and chronic fatigue syndrome. Untreated, the condition is fatal.

CM was first seen in a family of Giant Schnauzers and has since been found in Beagles, German Shepherds, Aussies (6-8%) and Border Collies. The DNA test establishes the genetic status of any given dog and it is known that the problem is inherited as a simple autosomal recessive trait. Affected dogs have inherited two faulty genes, one from each parent. Carriers do not suffer from the disease, but can pass the gene on to their offspring.

Pelger-Huët Anomaly

Pelger-Huët Anomaly (PHA) causes abnormalities in white blood cells called granulocytes. PHA may be mistaken for infection or early stage leukemia, and often only shows up when blood is being examined for other potential problems.

The condition is inherited as an incomplete dominant. Carriers are almost always healthy, but if bred to another carrier the pups that receive two copies of the PHA gene (affected pups) will be reabsorbed, stillborn or die shortly after birth. Occasionally an affected puppy will survive but have severe skeletal deformities and be susceptible to infection. PHA is therefore more a breeder's problem than an owner's as affected puppies almost never survive and if they do, have severe health issues. PHA causes small litters or loss of newborns.

The PHA gene has not yet been found but PHA status of breeding dogs can be determined by examination of a blood smear by a veterinary pathologist. Most PHA carriers have minor anomalies in some of their blood cells. However, not every PHA carrier will exhibit these anomalies so it is possible to receive false negative results from this test. Therefore any breeding dog with near relatives known to be PHA carriers should be tested, and carriers should not be bred to each other. If a breeding dog has extremely variable litter sizes it may be a PHA carrier and should be screened. It is important that anyone buying a pup from a PHA carrier is made aware that the pup may be a carrier.

Because the test for PHA requires the visual examination of a blood smear, this work is not undertaken by the big testing laborotories, but by major veterinary hospitals with the specific expertise. UKMASC is happy to advise if a member would like their dog tested.

Degenerative Myelopathy

Canine Degenerative Myelopathy is an incurable, progressive disease of the spinal cord. It is believed that the body's own immune system starts to attack and destroy the insulation around the nerve fibres of the spinal cord. This breaks the communication between the nerves in lower body of the dog and the brain. The disease typically starts after the age of 7 years and although it is most commonly associated with GSDs, corgis and boxers, the gene thought to be responsible has also been found in over 40 other breeds. DM initially affects the back legs, causing muscle weakness and mal-coordination. The dog may start to stagger or drag it's rear paws, which in the early stages may be misdiagnosed as arthritis. As the disease progresses the condition leads to paralysis of the back legs, incontinence and difficulties with balance and walking. As the disease works it's way up the spine, the dog will start to have problems with it's front legs, extensive muscle loss and progressive paralysis. The final stage involves neural deterioration in the brain, and failure of the respiratory muscles.

Progression of the disease is generally slow but highly variable. The dog could be crippled within a few months or may survive as long as three years or more. A DNA test will reveal Clears, Carriers and Affecteds, but as with the HSF4 gene for cataracts, an affected dog is not guaranteed to suffer from the disease but rather it is predisposed to develop the disease at some point in it's lifetime. It is likely that environmental factors may also play a part in triggering the onset of the disease.

Neuronal Ceroid Lipofuscinosis (NCL)

NCL is a progressive disease which leads to blindness, neurological problems and death. It is a lipid storage disease, meaning that affected dogs have an abnormal ability to store certain fats (lipopigments) in their bodies. There are many biochemical defects believed to be associated with these lipid storage diseases. NCL affects the central nervous system (the brain) and usually causes degeneration of the retina as well.

Affected dogs appear normal at birth but begin to exhibit symptoms early in life - around one to two years of age, although the age of onset and severity of the disease can vary greatly. Symptoms include progressive muscle weakness, loss of coordinated muscle movements, loss of balance, cognitive decline, abnormal behaviour, seizures and dementia.

Although the deteriorating eyesight and blindness may initially resemble PRA, the symptoms of NCL are more often due to a brain dysfunction (abnormal storage of lipoproteins in the brain's visual cortex) rather than a problem in the eye. Due to the severity of the disease, affected dogs rarely survive beyond 26-28 months. There is no treatment or cure at this time. DNA tests reveal normal, carriers or affecteds.

There are other diseases that occasionally occur in Aussies/MAS but at a suffuciently low rate that neither the KC nor any of the breed clubs have included them as recommendations or requirements. If you have cause for concern then DNA tests are also available for

- Canine Cyclic Neutropenia (CCN)
- the Bobtail gene (NBT)
- Cone Degeneration
- Hyperuricosuria (HUU)
- Canine Multi-focal Retinopathy (CMR)



Despite many years of research and some headway being made, there is currently no test for Epilepsy, in any breed of dog. It is currently considered to be polygenic and possibly subject to environmental and other genetic triggers, too.

It's a massively complex subject and until the day when it is understood, all we can do is research our lines and breed as responsibly as possible.

