

HEALTH AND GENETICS IN THE BORDER COLLIE

A Buyers/Breeders Guide

To be considered a genetic disorder, a health problem needs to have been demonstrated to be heritable, that is, passed on through one or both parents. Some disorders have high heritability, which means if the genes are present, the individual will have the disorder, and some disorders have low heritability, meaning both genetic and environmental factors are involved in whether the disorder occurs. It is generally easier to control disorders with high heritability because all individuals with the genetic makeup for the disorder can usually be identified. The term heritable disorder should be distinguished from the term congenital disorder, or problems that are present from birth, which may or may not be heritable.

Many diseases are inherited in a recessive manner (eg NCL, CEA and TNS). This means that the disease can be carried in perfectly healthy, normal animals without detection. If two animals that are carriers of the disease are bred, a proportion of pups in the litter may be affected with the disease. On average 25% of the litter may be affected with disease, whilst approximately 50% of pups in the litter will be asymptomatic carriers of the disease. Only 25% of the litter, on average, will not carry disease genes.

Border Collies are considered to be a generally healthy breed. However, as in all animals, there are some potential health problems. This information is presented to help both breeders and buyers to become more aware of some of the health and genetic issues in the breed at this time.

1. Clear or normal – means the sample submitted from the animal does not contain the gene responsible for the hereditary disorder tested, and the animal will not develop the condition, nor pass it on to any progeny
2. Carrier – means the sample submitted from the animal carries one copy only of the gene responsible for the hereditary disorder tested, the animal will not develop the condition, but may pass a copy of the gene on to any progeny
3. Affected – means the sample submitted from the animal carries two copies of the gene responsible for the hereditary disorder, and the animal will develop the condition, and will pass a copy of the gene on to any progeny
4. Clear by parentage – means the animal has been DNA profiled to confirm its parentage, and that both parents are also profiled, and are clear or normal for the hereditary disorder tested
5. Clear by pedigree – means the animal via pedigree is from parents that have been DNA tested, or from DNA tested stock that have been tested as clear or normal. This claim relies on the integrity of the registration papers for the animal and its ancestors. The parentage for the animal has not been verified by DNA.

Inherited disorders reported in the breed include the following:

NEURONAL CEROID LIPOFUSCINOSIS (NCL – formerly referred to as CL)

NCL is a type of storage disease affecting Border Collies. NCL has been published as CL or NCL in scientific literature. This is because it is a storage disease which affects the nervous tissue, or neurons, of the brain.

Inheritance patterns and percentage of affected pups in litters indicate that NCL is inherited in a recessive manner. The gene responsible for disease causes a faulty protein to be made in the neurons, this causes an accumulation of waste products inside the tissues of the brain and the function of the neuron is destroyed. Symptoms of NCL usually commence with behavioural changes - hyperactivity and aggression, progressing to loss of learned behaviour, dementia and blindness. Clinical signs usually commence between 16 months and 23 months. Therefore the problem occurs well after puppies are sold and it is heartbreaking for owners as well as breeders. There is no cure or treatment for affected animals, and all are eventually euthanased on humane grounds.

Since 1980 a total of 26 litters have been identified as having NCL affected puppy/s. When compared with the vast number of litters bred in this time, it can be seen that this disorder is actually very rare in the breed.

In 2006 a peer reviewed genetic test became available for diagnosing the NCL status of Border Collies. Without the research, dedication and fund raising of Border Collie clubs from around the world over the previous 17 yrs this would not have been achieved. The genetic NCL status of any dog or bitch used for breeding should be known. This involves submitting either blood or cheek swab tests to a laboratory capable of testing for the disease. The individual will be classified as either clear (normal) for NCL, a carrier of NCL or affected by NCL. If both the parents of an individual have been tested as normal for NCL, then the individual is classified as “clear by parentage” or “clear by pedigree”(see definitions on previous page) The Border Collie Club of Victoria does not support the breeding of two NCL carriers together, and recommend that this is never done. The advent of genetic testing means that no Border Collie breeder should ever breed or sell a puppy that is affected with NCL again.

For pet owners it should be emphasised that:

1. All Border Collies used for breeding should have their NCL status classified either by genetic test or by parentage
2. Carrier puppies live perfectly normal, healthy lives and will never show any symptoms of NCL. A NCL carrier animal should only ever be bred to a NCL clear (by DNA) animal.

Trapped Neutrophil Syndrome (TNS)

Trapped Neutrophil Syndrome- TNS (or Hereditary Neutropenia) is a more recently described disease in Border Collies. Inheritance patterns and percentage of affected pups in litters indicate that TNS is inherited in a recessive manner (like NCL) with both parents having to be carriers of the disease to produce an affected pup.

Symptoms may be first seen in puppies as young as 2 weeks or as old as 7 months of age. Affected puppies are usually smaller, have slower growth rates, and can appear to have a “ferret like” head and a poor haircoat. Some puppies are not obviously different until they become ill. Other symptoms of disease include lameness, inappetence, diarrhoea and a high temperature. Sometimes pups become ill after their first vaccination. Both male and female puppies of any colour can be affected. TNS is a disease which ultimately causes a deficiency of the immune system, so symptoms can vary between pups. Affected animals are eventually euthanased on humane grounds.

Blood tests may reveal a low neutrophil count, nucleated red blood cells in circulation despite a non-regenerative anaemia, a fasting hypercholesterolaemia, decreased serum albumin and increased serum alkaline phosphatase. X-rays of limb bones may show reduced density and thin cortices and in some cases fractures in metaphyses. Sclerotic bone may be present in areas adjacent to the fractures.

Diagnosis of TNS requires three criteria. Firstly, the pups need to show clinical symptoms consistent with TNS Secondly, blood tests which confirm a low neutrophil count are suspicious of TNS but do not provide a diagnosis. Puppies will occasionally develop low neutrophil counts for other reasons, eg. viral or bacterial infections, and should not be condemned on this basis. The third criteria- bone marrow biopsy- will give accurate diagnosis. Pups with TNS have been found to have an increased “myeloid to erythroid ratio” in their bone marrow. This means there are more white blood cells precursors in the bone marrow than there should be- hence the name “trapped neutrophil syndrome” (neutrophils trapped in the bone marrow) was made.

A genetic mutation test for TNS is now available (2007) which involves submitting a blood sample or cheek swab. This test has not been peer reviewed Results will classify each animal as clear for TNS, TNS carrier or TNS affected. If both the parents of an individual have been tested as clear for TNS, then the individual is classified as “clear by parentage” or “clear by pedigree The Border Collie Club of Victoria does not support the breeding of two TNS carriers together, and recommend that this is never done. The advent of genetic testing means that no Border Collie breeder should ever breed or sell a puppy affected with TNS again.

For pet owners it should be emphasised that:

1. All Border Collies used for breeding should have their TNS status classified either by genetic test or by parentage.
2. Carrier puppies live perfectly normal, healthy lives and will never show any symptoms of TNS. A TNS carrier animal should only ever be bred to a TNS clear (by DNA) animal.

COLLIE EYE ANOMALY (CEA)

Collie Eye Anomaly (CEA), a hidden cause of blindness, is a recessively inherited congenital disease. Collie breeds are affected - Rough and Smooth Collies, Shetland Sheepdogs (Shelties) and Border Collies are the predominant breeds in which this condition is seen. Border collies show a very low incidence of affected CEA puppies compared to other breeds where this condition is present.

In mild cases of CEA pale areas may be seen at the back of the eye (called choroidal hypoplasia). Other changes that may be seen in the eye with CEA include increased tortuosity of retinal blood vessels (colobomas) in the optic nerve head, and occasional retinal haemorrhage and detachment. Animals with CEA do not necessarily show all of these changes, but may only have slight changes. Poor vision occurs with CEA but is often difficult to detect especially if the clinical changes in the eye are mild. Owners usually report poor vision in animals which have large pits in the optic nerve head. Blindness may occur if there is retinal haemorrhage and detachment.

As a disease, CEA is easiest to detect in young pups at approximately 6 – 8 weeks of age by a veterinary ophthalmology examination. This is the best time to check for choroidal hypoplasia. Colobomas are checked at around 14 weeks of age or later. After 14 weeks of age it is not possible to detect all cases of CEA. The development of retinal pigment can cover the signs of CEA. In this case the dog can appear normal (hence the term “go normal”), but it can still transmit the CEA gene(s) Carrier animals will show no clinical signs on examination.

In 2006 a peer reviewed genetic test became available for diagnosing the CEA status of Border Collies. The genetic CEA status of any dog or bitch used for breeding should be known. This involves submitting either blood or cheek swab tests to a laboratory capable of testing for the disease. The individual will be classified as either clear for CEA, a carrier of CEA, or affected by CEA. If both the parents of an individual have been tested as clear for CEA, then the individual is classified as “clear by parentage” or “clear by pedigree. The Border Collie Club of Victoria does not support the breeding of two CEA carriers together, nor should a CEA affected animal ever be bred to a CEA carrier animal, as these genetic combinations will produce CEA affected animals. The advent of genetic testing means that no Border Collie breeder should ever breed or sell a puppy affected with CEA again.

For pet owners it should be emphasised that:

1. All Border Collies used for breeding should have their CEA status classified either by genetic test or by parentage
2. CEA carriers live perfectly normal, healthy lives and will never show any symptoms of CEA. A CEA carrier animal should only ever be bred to a CEA clear (by DNA) animal.

HIP DYSPLASIA (HD)

Canine hip dysplasia (HD) is a genetic disorder that is inherited in a complex manner because it is influenced by more than one gene. The mode of inheritance is called polygenic. It was first recorded in the 1930s, the incidence in Border Collies is lower than in many other breeds. All individuals which are to be bred from should have their hips examined and scored by radiographic assessment (x-ray) prior to breeding.

With dogs that have HD the hip ball is not a neat fit in the socket joint. The joint is subsequently loose and becomes subject to excessive wear and tear. This results in the development of degenerative joint disease (arthritis). It is the arthritis that causes the joint pain and lameness. Radiographs (x-rays) of the hip joint will demonstrate laxity in the joint as well as assess the amount of degenerative joint disease at that time. Dogs should be scored by the most commonly used system in Australia (AVA) after 12 months of age. For more information on the grading system used by the AVA please go to www.ava.com.au. Alternatively they may be assessed by the PennHIP System after 16 weeks of age. For more information on the PennHIP system please go to www.pennhip.org. The age at which a dog with HD shows clinical signs of lameness and the severity of the lameness is highly variable, this disease may occur at almost any lifestage.

Other environmental factors can impact on the degree of wear and tear the joint is subjected to. The two main ones are the weight of the dog and the amount and type of exercise the dog takes. Obviously the heavier the dog the more stress is put on the hip joint so large heavy dogs are more likely to become lame because of hip dysplasia than small light ones. Dogs that get a lot of exercise at fast gaits such as galloping behind a car or bicycle and dogs that do a lot of jumping or turning and stopping such as when fetching balls or sticks stress the hip joints and are therefore more likely to become lame. A normal hip joint can sustain these sorts of stresses without developing arthritis, but a loose dysplastic joint does not cope as well. Another environmental factor for HD is feeding poor quality puppy diets with excessive calcium, or over supplementing calcium, which make pups grow rapidly, giving bones and joints less time to develop properly. Whilst calcium is very important in a puppy's diet, commercial foods these days do not require supplementation if the puppy is being predominantly fed these diets.

For pet owners it should be emphasised that:

1. HD cannot be accurately predicted in pups- but you minimise your risks of having problems with HD by purchasing a pup from a breeder who is scoring breeding animals and breeding only from animals with acceptable scores.
2. Puppy owners should be aware that they should
 - a) Not allow their puppy to get overweight
 - b) not subject young growing joints of their pup to repetitive hard exercise especially running and twisting on hard surfaces
 - c) take care with their puppy's nutrition
 - d) only breed their dog after having the hips radiographed and scored, and then only breed if the score is acceptable to other approved animals

OSTEOCHONDRITIS DISSECANS (OCD)

OCD is a disorder of cartilage inside joints, where cartilage precursors at the joint surface fail to develop into bone. This ultimately causes areas of thickened cartilage at the joint surface, which are prone to mechanical trauma during normal movement, and flaps of cartilage will lift away from the surface of the joint which causes arthritis and soreness. The flaps can break away and float around inside the joint, becoming "joint mice", causing chronic arthritis and lameness. Genetics clearly play a role in this disease- it occurs much more commonly in certain breeds (more often large/giant breeds), as well as at specific sites of different joints for different breeds. However like HD, other factors are thought to contribute to the severity of disease; trauma, rapid growth rates, increased weight bearing, and nutrition to name some.

OCD is thought to be polygenic. Problems usually present in affected animals between 4-7 months of age, sometimes characterised by sudden onset of significant lameness, other times discomfort of the joint and reduced range of motion may be seen. Commonly affected joints in the Border Collie include the shoulder, elbow, and hock. OCD has also been seen in the hip, stifle, and in some breeds the vertebrae of the neck may be affected.

A diagnosis of OCD may be quite difficult. Radiographs of affected and opposite joints allow for comparison and may give a definitive diagnosis. These need to be taken with the animal under heavy sedation or general anaesthetic so that the joints can be stretched and positioned accurately. Sometimes lesions are obvious, other times the changes are much more subtle and difficult to detect.

Screening radiographs of the elbows are recommended to be taken at the same time the dog is assessed for HD. The elbows are then assessed for any signs of joint disease. This provides information with regard to OCD that has occurred in the elbow, but will also rule out other types of inherited elbow dysplasias.

For pet owners it should be emphasised that:

1. OCD cannot be accurately predicted in pups- but you minimise your risks of having problems with OCD by purchasing a pup from a breeder who is breeding only from sound animals with no history of OCD in their lines, and who is also elbow scoring .
2. Puppy owners should be aware that they should
 - a) not allow their puppy to get overweight
 - b) not subject young growing joints of their pup to repetitive hard exercise especially running and twisting on hard surfaces
 - c) take care with their puppy's nutrition
 - d) not breed from an animal diagnosed with OCD

In summary, this brochure is meant to provide a brief description of some common conditions and some current knowledge about their heritability in our breed. Please remember, conditions present in Border Collies are not limited to those discussed here. Keep track of updated and more in depth Border Collie health and genetics information by checking the Victorian Border Collie Club Web Site on www.bordercollie.org.au.

This document was prepared by members of the Hereditary Diseases and Disorders Advisory Panel (HDDAP) of the BCCV Inc. as an information guide only. The BCCV makes no representation or warranty on the accuracy of the science and DNA testing described herein, but makes this guide available to members in good faith based on information available at time of publishing (June 2010)

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