BORDER COLLIE CLUB
OF SOUTH AUSTRALIA

The Border Collie Club of South Australia Inc.

Inherited Disorders in Border Collies – Testing Information for Breeders and Puppy Buyers

Border Collies are generally considered to be a relatively healthy dog breed. However, like all animals, they are susceptible to conditions that can affect their health and welfare. Many of these disorders are inherited – passed on to a dog from its parents.

Fortunately, within the last 30 years, researchers have developed tests for a large number of diseases that Border Collies can inherit. DNA testing can identify which dogs are affected by (display themselves) or carry (not affected, but can pass on the genetic mutation to puppies) these diseases. X-Rays can also detect some structural disorders. An ethical breeder uses both DNA testing and X-Rays to determine which dogs should or should not be bred together, to have the greatest chance of producing healthy puppies.

This document will list the most current DNA and X-Ray tests available for Border Collies (as of December 2021). Some tests have been available for many years, whilst some have emerged only recently. A more detailed list of conditions will be given below the table. For breeders, links for laboratories providing tests will also be provided at the end of the document.

It should be noted, however, that some conditions are not yet testable, as the genetic mutations that are responsible for causing these is not yet known. As a result, these conditions cannot be fully prevented by DNA testing. Ethical breeders who have a deep understanding of the ancestry, or lines, of their breeding dogs can try to prevent these disorders by avoiding using affected dogs or lines with related symptoms. These conditions will also be listed.

The Border Collie Club of South Australia (BCCSA) recommends that:

1. Ethical breeders undertake all key tests, avoid breeding dogs who are both carriers of a single condition or a carrier to an affected dog, and begin to adopt emerging tests into their breeding practises to do their best to avoid producing affected puppies.
2. Ethical breeders carefully research and know their lines in an attempt to avoid producing dogs with untestable conditions.
3. Puppy buyers look for breeders who can provide evidence of testing their parent dogs for the key conditions at minimum, so they can be assured their new puppy won't be affected by most preventable conditions.

Key Tests (as of December 2021)

Condition Name	*Directly Preventable (Autosomal Recessive)? (Yes or No)*	*Organisations Offering Tests (info for breeders)*
CEA – Collie Eye Anomaly (DNA)	Yes	Orivet BC Panel / MyDogDNA Panel / PPG / Embark
TNS – Trapped Neutrophil Syndrome (DNA)	Yes	Orivet BC Panel / MyDogDNA Panel / PPG / Embark
NCL5 – Neuronal Ceroid Lipofuscinosis Type 5 (DNA)	Yes	Orivet BC Panel / Animal Genetics / Laboklin. PawPrint Genetics (PPG) / Embark, NOT currently part of MyDogDNA Panel
MDR1 – Multiple Drug Resistance Type 1 (DNA)	Preventable Yes (although is autosomal incomplete dominant)	Orivet BC Panel / MyDogDNA Panel / PPG / Embark
IGS/ICM – Imerslund-Gräsbeck Syndrome / Cobalamin Malabsorption (DNA)	Yes	Orivet BC Panel / MyDogDNA Panel / PPG / Embark
DM - Degenerative Myelopathy (DNA)	Yes	Orivet BC Panel / MyDogDNA Panel / PPG / Embark
GGD – Glaucoma and Goniodysgenesis (DNA)	In part – Genetic component Yes, Environmental component No	Orivet BC Panel / MyDogDNA Panel / PPG / Embark
RS/DH – Raine Syndrome / Dental Hypomineralisation (DNA)	Yes	Orivet BC Panel / MyDogDNA Panel / PPG / Embark
SN – Sensory Neuropathy (DNA)	Yes	MyDogDNA Panel / Orivet *Add-On* / PPG / Embark
Hip Dysplasia (Hip X-Rays)	No – genetic component suggested, but other factors can also cause or contribute	X-Ray by vet, interpretation by CHED Panellist vet or PennHip
Elbow Dysplasia (Elbow X-Rays)	No – genetic component suggested, but other factors can also cause or contribute	X-Ray by vet, interpretation by CHED Panellist vet

Emerging Tests (as of December 2021)

Condition Name	*Directly Preventable (Autosomal Recessive)? (Yes or No)*	*Organisations Offering Tests (info for breeders)*
EAOD - Early Adult Onset Deafness (DNA)	In part – linkage marker test only at present, but 'At Risk' puppies can be prevented	MyDogDNA Panel – 4 markers Orivet BC Panel – 1 marker

CA - Cerebellar Abiotrophy (DNA)	In part – linkage marker tests only at present, but 'At Risk' puppies can be prevented	Working Dog CA Panel - Dog Breeding Science Australia
MC/MH – Myotonia Congenita / Myotonia Hereditaria (DNA)	Yes	Orivet BC Panel / MyDogDNA Panel / PPG
OCD – Osteochondritis Dissecans (Shoulder X-Rays)	No – genetic component suggested, but other factors can also cause or contribute	X-Ray by vet
RPED – Retinal Pigment Epithelial Dystrophy, otherwise known as CPRA – Central Progressive Retinal Atrophy (Ophthalmological Exam)	Partially – Although this is believed to be an autosomal recessive condition, annual screenings are recommended to detect this	Ophthalmological Examination by ACES Panellist vet
Eye Examination, including Gonioscopy, for various eye conditions, including Primary Lens Luxation (PLL) (Ophthalmological Exam)	Partially – Although these are autosomal recessive conditions, annual screenings are recommended to detect these diseases	Ophthalmological Examination by ACES Panellist vet
CSD – Congenital Sensorineural Deafness (BAER Testing) ***Testing is recommended for both breeding dogs and litters of puppies***	No – Mode of inheritance is presently unknown, but strong links to parent hearing status	BAER Testing by vet

Untestable Conditions (as of December 2021)

Condition Name	Risk Factors (Aside from Family History)
Idiopathic Epilepsy	Unknown
DLE/SLE – Lupus (Discoid Lupus Erythematosus / Systemic Lupus Erythematosus)	DLE anecdotally more likely in chocolate or dilute-based lines
BCC – Border Collie Collapse	Unknown
PRA – Progressive Retinal Atrophy (Border Collie Type)	Questionable ophthalmic exam results. Males are more likely to be affected, whilst females can be hidden carriers. Known issue in European BCs at least.
EPI – Exocrine Pancreatic Insufficiency	Combination of nutritional, intestinal microbiome, immune, and genetic factors.
MG – (Acquired) Myasthenia Gravis	Combination of environmental and hormonal factors, as well as potential infection.

Unvalidated Tests (as of December 2021)

Condition Name	*Reason*	*Appears on Breed Panels from these Organisations*
vWD - von Willebrand Disease (Type II) (DNA)	Detects the causal mutation in German Shorthaired Pointers only	Orivet BC Panel
Cystinuria (Type IIA) (DNA)	Limited evidence to suggest this exists in Border Collies	Orivet BC Panel / MyDogDNA Panel / PPG / Embark
PLL – Primary Lens Luxation (via DNA Test)	Relevant for other breeds, but does not detect the causal mutation in Border Collies	Orivet BC Panel / MyDogDNA Panel / PPG / Embark

Detailed List of Conditions

Key Tests

CEA – Collie Eye Anomaly - is an autosomal recessive inherited gene and one of the most common inherited diseases. It can be quite mild or severe. CEA affects the choroid and blood vessels to the retina and is often not detected until the dog displays signs of blindness.

TNS – Trapped Neutrophil Syndrome – is an autosomal recessive gene causing disorder of the immune system, preventing affected dogs' neutrophils being released from the bone marrow to fight infections properly. Puppies are prone to infections, and affected puppies rarely survive past 6 months of age. There is no cure; antibiotics and steroids may help in the short term, but humane euthanasia is recommended.

NCL 5 – Neuronal Ceroid Lipofuscinosis 5 – is a neurodegenerative disorder where a specific enzyme for cell metabolism is missing. That leads to nervous system cell function being disrupted. Dogs present with progressively worsening neurological symptoms, from about 15 months, of disorientation, behavioural changes, aggression, seizures, abnormal gait and balance problems, blindness, and dementia. Affected dogs rarely survive past 28 months of age.

IGS/CM – Imerslund-Gräsbeck Syndrome / Cobalamin Malabsorption – is a disorder that causes affected dogs to be unable to produce a protein that is required for the absorption of the vitamin cobalamin (B12) in the intestinal tract. Dogs with this disorder can suffer from anorexia, lethargy, poor muscle mass, and painful ulcers, and in some cases, can develop severe neurological symptoms leading to seizures, coma, or death. There is no cure, although dogs can be treated with a lifelong B12 supplementation via injections.

MDR1 – Multi Drug Resistance 1 – is a mutation that causes dogs to be sensitive to certain drugs. An affected dog's brain cannot filter some drugs in the brain properly, meaning these drugs build up in the brain, resulting in seizures. Ivermectin, a drug often used in heartworm treatments, is one example of a drug to be avoided by affected dogs. Dogs with two copies of the mutation are particularly at risk, although dogs with only one copy (carriers) may also show signs of sensitivity. It is not recommended to breed a dog with even one copy of the mutation.

RS/DH - Raine Syndrome / Dental Hypomineralisation – is a disorder that causes extreme tooth wear at an early age. Affected dogs will develop pulpitis, and require extraction of teeth to prevent further pain. There is no cure for this condition.

DM – Degenerative Myelopathy – is a neurological disorder where affected dogs suffer muscle wastage and lose coordination due to spinal nerve damage. This disorder generally affects dogs older than 8 years of age, and affected dogs may lose the ability to walk and bladder control between 6 months to 2 years after the first symptoms appear. Treatments are available, but their effectiveness is limited if nerve damage has already occurred. Although DM is inherited when two copies of the defect are passed on to the puppy by its parents, it has 'incomplete penetrance', which means not all dogs with 2 copies of this gene will develop the condition.

GGD – Glaucoma and Goniodysgenesis – Goniodysgenesis is a disorder that affects the development of the eye. This puts pressure on the optic nerve. Typical symptoms include sore or watery eyes, light sensitivity, and eyelid spasms. Without treatment, GGD often results in 'primary' glaucoma (glaucoma from a hereditary condition), and eventual blindness. It should be noted that 'secondary' glaucoma can develop in any dog. This is caused by an environmental factor such as trauma to the eye, and is not genetically linked.

SN – Sensory Neuropathy – is a neurological disorder that affects the nervous system, particularly nerve cells that control muscle movement (motor neurons) and feeling (sensory neurons). Symptoms typically appear before 7 months of age, and include loss of coordination, feet that knuckle over, and urinary incontinence. Self-mutilation of the feet may occur due to tingling or loss of feeling in the paws. There is no cure, and affected dogs are usually euthanised before they reach 2 years of age.

HD - Hip Dysplasia – is an abnormal formation of the hip socket and is variable in severity. It causes progressive pain, lameness and arthritis, and may be treatable with nonsteroidal anti-inflammatory medications. The worst form is non formation of the hip sockets. Pups from parents with good hip scores are less likely to develop HD, although there is no guarantee, as environmental factors can also influence the development of the disease. Hip X-Rays are taken by a reputable vet, and are then sent to a CHED (Canine Hip and Elbow Dysplasia) scorer. The breed average is currently 7.7. See the ANKC ORCHID (Officially Registered Canine Health Information Database) website for contact details for scorers: http://orchid.ankc.org.au

ED – Elbow Dysplasia – is an abnormal formation of the elbow socket. It causes progressive pain, lameness and arthritis, and may be treatable with nonsteroidal anti-inflammatory medications. Pups from parents with good elbow scores are less likely to develop ED, although there is no guarantee, as environmental factors can also influence the development of the disease. Elbow X-Rays are taken by a reputable vet, and are then sent to a CHED (Canine Hip and Elbow Dysplasia) scorer. The breed average is currently 0. See the ANKC ORCHID (Officially Registered Canine Health Information Database) website for contact details for scorers: http://orchid.ankc.org.au

Emerging Tests

EAOD – Early Onset Adult Deafness – is a condition where dogs lose their hearing typically when they are around 3-5 years old. This is before the expected 'old age onset' of hearing loss in dogs, which tends to occur at around 10 years of age. Research is developing for this condition, but it does appear to be genetically linked. The mutation/s that cause EAOD have not yet been determined. However, researchers have developed 'marker tests' that can identify genetic markers that have been found in dogs that have lost their hearing early. Currently, two 'marker tests' exist, one offered by Genoscoper (through MyDogDNA, which has recently been acquired by Mars' Wisdom brand), and the other offered by Orivet. The Genoscoper test looks for 4 markers, whereas the Orivet test looks for 1 which is unrelated to the Genoscoper markers. Evidence currently suggests a good correlation between the Genoscoper test and EAOD, as all EAOD-diagnosed dogs that have been tested have been found to have the markers and were given an 'At Risk' result.
As the test is in the emerging stage and the exact mutation causing EAOD has not yet been found, the American Border Collie Association recommends that the results of this test be used carefully. They advise that any dogs who test as 'Notable' (carrier) or 'At Risk' (potentially affected) not be discarded from a breeding program simply because of this result, only that they be bred to a dog tested as 'Clear' for the EAOD marker tests in future. This will assist breeders to avoid producing any 'At Risk' puppies, and reduce the chances of deafness occurring in their litters.

CA – Cerebellar Abiotrophy – is a neurological disease that causes deterioration and loss of neuron cells in the cerebellum; the area of the brain that controls balance and coordination. This condition affects many breeds, including Border Collies and Kelpies. There are at least two main types that affect BCs: Early Onset, and Late Onset. For the early onset type, puppies can display symptoms between 4-16 weeks of age, whilst late onset typically occurs between 3-8 months but can appear in dogs up to two years of age. Dogs affected by CA have balance problems, an awkward gait, poor muscle control, head or neck tremors, and can have poor depth perception. Severely affected dogs are generally euthanised, but individuals with mild to moderate symptoms can survive into adulthood.
Research is developing to detect the Border Collie and Kelpie variant of this condition, but it does appear to be genetically linked. The mutation/s that cause CA have not yet been determined. However, researchers have developed two 'marker tests' that can identify genetic markers that have been found in dogs affected by CA; LINGO3 for early onset, and VMP1 for late onset. Currently, these marker tests are offered by Dog Breeding Science Australia. A LINGO3 'At Risk' result does not guarantee that a dog will develop early onset CA, but correlates well with clinical symptoms. VMP1 'At Risk' results have correlated quite accurately with dogs showing late onset clinical symptoms. Breeders are recommended to breed a 'Carrier' or 'Affected' dog to a 'Clear' dog for both types of CA.

MC/MH – Myotonia Congenita / Myotonia Hereditaria – is a genetic muscular disorder that results in delays to the relaxation of the skeletal muscles. The muscles remain contracted, which results in muscle stiffness and hypertrophy (excessive muscle size). MC/MH is rare in Border Collies, but has been detected in some individuals as part of a genetic testing panel.

OCD - Osteochondritis Dissecans – is an abnormal development of cartilage on the end of a bone, which impacts on the joint between bones. OCD typically occurs within the shoulder joint, but can also affect the hip, stifle (knee), or elbow. The exact cause is unknown, but OCD can be linked to both genetic and non-genetic factors, including rapid bone development, improper diet, and trauma. Symptoms of OCD include pain reactions, swollen joints, and lameness. Shoulder X-Rays are taken by a reputable vet, and examined to detect any lesions. Pups from

parents with clear shoulder X-Rays are less likely to develop OCD, although there is no guarantee.

RPED – Retinal Pigment Epithelial Dystrophy, otherwise known as CPRA – Central Progressive Retinal Atrophy – is a disorder that affects the retinal pigment cells of the eye. The coating of the retina cannot efficiently break down used parts of the photoreceptors, and so lipopigment builds up and moves into the retina itself. The photoreceptors that detect light and colour also break down. Affected dogs initially struggle to see in bright light. Loss of vision may slowly develop, but does not usually progress to complete blindness. Environmental factors such as a poor quality diet may influence the severity of the disease, and Vitamin E supplements are sometimes recommended as a treatment option. Onset may begin from 3-4 months of age. RPED/CPRA is diagnosed through eye examination by a veterinary ophthalmologist, and although it can be detected early at 12 months in some dogs, it is usually detected from 18 months of age. Breeding dogs are recommended to have an eye exam each year up until 8 years of age.

Eye Examinations (including Gonioscopies) – are able to detect abnormalities and changes in the structure or function of the eye. In Border Collies, there are four main conditions that eye examinations and gonioscopies screen for: Collie Eye Anomaly (CEA), Glaucoma and Goniodysgenesis (GGD), Primary Lens Luxation (PLL), and Retinal Pigment Epithelial Dystrophy (RPED), which is otherwise known as Central Progressive Retinal Atrophy (CPRA). Although CEA and GGD have reliable DNA tests, the Border Collie type of PLL and RPED/CPRA do not. Eye examinations are currently the only way of detecting these conditions. As PLL and RPED/CPRA are genetically inherited, it is important that these conditions are detected and that affected dogs are not bred together. For greater detail on each of these conditions, find them listed individually in this document.

Australian Canine Eye Scheme (ACES) Panellists are veterinary ophthalmologists who can issue ACES certificates to record eye abnormalities and certify that a dog does not display signs of these conditions at the time of examination. *Important: As these conditions can develop as a dog ages, the Australian Veterinary Association (AVA) recommends assessment from one year of age and then annual reassessment throughout the dog's breeding life.*

See the ANKC ORCHID (Officially Registered Canine Health Information Database) website for contact details for ACES Panellists: http://orchid.ankc.org.au

CSD - Congenital Sensorineural Deafness **–** is a condition that causes a loss of hearing receptors in affected dogs. The resulting deafness can occur unilaterally (deaf in one ear), or bilaterally (deaf in both ears). Onset of inherited CSD typically occurs in the first few weeks of a puppy's life, but can develop in dogs up to 6 months of age. No treatment is available, and hearing loss is permanent. Evidence suggests CSD is genetically inherited, although the genes involved are not yet known. However, known risk factors for increased likelihood of CSD in Border Collies include merle colouring, significant white areas on the head, blue eye pigmentation, and parent dogs with affected hearing. It should also be noted that deafness can also be acquired, rather than genetically inherited, through exposure to environmental factors such as infections in utero or as puppies.
As dogs can adapt to hearing loss, particularly in dogs with unilateral hearing, deafness can be difficult to identify. Brainstem Auditory Evoked Response (BAER) testing can be used to objectively assess a dog for deafness, either unilateral or bilateral. It is recommended that parent dogs are assessed by a veterinarian with experience in BAER testing, and that only bilateral hearing dogs are bred to reduce the likelihood of producing pups with CSD, although

there is no guarantee. It is also recommended that litters of pups are BAER tested for CSD at around 5.5-6.5 weeks of age to determine their hearing status.

Untestable Conditions

Dogs with these conditions in the immediate family history should not be bred from; however, by the time these diseases have occurred, the dog may have already been bred. A breeder should have a good depth of knowledge about the health of the dogs they are breeding in order to prevent these conditions from arising in puppies they produce – not just their own dogs, but the parents, grandparents, great grandparents, and the litter siblings at each of these generations.

Idiopathic Epilepsy – is a neurological disorder where abnormal electrical signals in the brain cause spasms, convulsions, and can result in a loss of consciousness. Seizures can result from non-genetic causes such as illness, trauma, or poisons, but epilepsy without a known cause is called 'idiopathic'. Idiopathic epilepsy is believed to be genetic, and is unusually common in Border Collies. First signs usually occur between 6 months to 5 years of age, but onset may occur later in life. Most seizures occur at night or mornings, whilst the dog is asleep. Epilepsy is not curable, but can be treated with medications to varying degrees of success.

DLE/SLE – Discoid Lupus Erythematosus / Systemic Lupus Erythematosus – are genetic diseases of the immune system. Affected dogs' immune systems react against their own DNA and cause inflammatory reactions. DLE causes inflammation of the skin, and generally affects the nose leather or far less commonly, the ears and mouth. It causes loss of nose pigment, which can progress to skin cracking and painful ulceration. A biopsy is needed to confirm the diagnosis. SLE is far more severe, and causes inflammation across multiple organs. Arthritis, lameness, blood chemistry problems and seizures can all result from SLE. Exposure to sunlight often aggravates both conditions. Neither condition has a cure, but they can be managed with sunlight avoidance and medications, depending on the severity of the condition. Although no research seems to have been done, anecdotally, DLE has a higher incidence of appearing in chocolate or dilute (blue or lilac)-based lines. It is recommended that dogs with DLE/SLE should not be bred.

BCC – Border Collie Collapse – is a disorder of the nervous system that is triggered by exercise. During strenuous activity, affected dogs can suffer a BCC episode, during which they may experience disorientation, cognitive delay, staggered gait, a dragging of the legs, and may fall over. This is sometimes confused with heat exhaustion, but occurs only 5-15 minutes into exercise. Their body temperature is not raised any higher than an unaffected dog doing the same activity, and recovery tends to occur within 5-30 minutes without any organ damage or blood abnormalities; much faster than a dog with heat exhaustion and associated internal chemistry changes. Although Labrador Retrievers suffer from the similar condition Exercise Induced Collapse (EIC), the genetic mutation that causes EIC does not correlate with BCC. BCC-affected dogs are often retired from strenuous activities and are carefully managed during hot weather.

PRA – Progressive Retinal Atrophy (Border Collie Type) – is a genetic disorder of the retina in the eye. The retina contains photoreceptor cells, which receive light from the lens, and then convert the light into electrical signals to send to the brain. PRA causes lesions in both eyes (bilateral), which result in a loss of night vision. This further progresses to the dog losing its sight entirely. The Border Collie type, currently labelled XLPRA3, does not currently have a genetic test, and can only be diagnosed through an ophthalmic exam and electroretinography. Onset has occurred in dogs as early as 6 months old, but average is at 4 years of age. This type is believed to be X-linked, which means that males are much more likely to be affected by the disease than females. This is problematic, as a female's carrier status is not likely to be detected unless she is bred to a carrier or affected male and subsequently produces affected puppies. In countries

such as France, where the PRA-affected rate is predicted to be 10-15%, breeding dogs are recommended to have an eye exam each year up until 8 years of age.

EPI – Exocrine Pancreatic Insufficiency – is a disorder that results in the pancreas not being able to produce digestive enzymes that break down fats, carbohydrates, and proteins. Affected dogs cannot digest food and nutrients, and so lose weight despite being fed well. Symptoms include a ravenous appetite, and an increased amount of pale, fatty, and 'pudding-like' fecal matter. EPI can be inherited genetically, but currently appears to be more commonly 'acquired' in Border Collies. The acquired form develops from events such as Pancreatic Acinar Atrophy (PAA) - where the immune system attacks its own pancreas cells - or from bouts of pancreatic inflammation (chronic pancreatitis). The exact causes of these events are unknown, but are believed to be related to the dog's nutrition, intestinal microbiome, and immune and genetic factors. EPI is diagnosed via a blood test, and is treated using life-long enzyme replacement medication, as well as vitamin B12 and/or vitamin E supplements if recommended by the veterinarian.

MG – (Acquired) Myasthenia Gravis – is an autoimmune disease that affects the transmission of signals between the nerves and muscles. Affected dogs fatigue easily, have difficulty with drooling and blinking, show muscle weakness, and may collapse during exercise. It is commonly associated with megaoesophagus, where food is often brought up without passing into the stomach. Although MG is inherited in some breeds, Border Collies are usually affected by the acquired form, which is believed to be caused by a mixture of environmental and hormonal factors, and/or infection. However, there are breeds that are predisposed to developing acquired MG, which could suggest a genetic component as well. Onset of acquired MG generally occurs between 1-4 years of age, but can be diagnosed in dogs as old as 9-13. Treatment consists of medication to improve nerve signal transmission, but megaoesophagus issues could necessitate intensive care.

Unvalidated Tests for Border Collies

vWD – von Willebrand Disease (Type II) – is a blood clotting disorder where a factor within the blood of affected dogs is structurally abnormal, and so does not clot properly. This can lead to bleeding episodes, or severe and even life-threatening bleeding as a result of even minor trauma. Blood or plasma transfusions to induce clotting may be required for treatment. vWD is rare in Border Collies, but has been detected in some individuals.
The vWD(II) test has received criticism since 2017. A study (Vos-Loohuis et al., 2017) tested German Wirehaired Pointers, a breed at particular risk. It was found that the vWD(II) test produced false positive 'affected' results for dogs that had no signs of a bleeding disorder. Another study that reviewed data from genetic testing (Donner et al., 2018) noted an unusually high percentage of dogs across all breeds testing as 'affected' to vWD(II). Blood samples were taken from some of these 'affected' dogs to test for the vWF antigen (present in a truly vWD affected dog). Results for tested dogs were either within normal range, or in the borderline range, lending weight to the suggestion that the vWD(II) test produces false positive 'affected' results. This test is only relevant for German Shorthaired Pointers, and not other breeds.

Cystinuria (Type IIA) – is a condition that affects a dog's ability to filter cystine and other amino acids from urine. The amino acids cannot be transported by the kidneys, and so they build up and crystalise to form bladder stones. Blockages can cause urinary tract infections or kidney failure, and without surgical intervention, can be fatal.
Although the genetic test is commonly offered as part of a breed panel, and having the results does no harm, there is limited evidence to suggest Cystinuria is inherited in the Border Collie breed.

PLL via DNA Test – Primary Lens Luxation – is a disease resulting in breakdown of ligaments to the eye, partially dislocating the lens of the eye. The movement of the lens into other areas can cause dislocation of other parts of the eye, potential pressure build-up leading to secondary glaucoma, and blindness. PLL affected dogs often experience symptom onset between 3-6 years of age. PLL is generally thought to be autosomal recessive, which means dogs who inherit 2 copies of the mutation are at risk; however, some carriers of only one copy of the mutation have also developed PLL. It should be noted that secondary lens luxation can develop in any dog. This is caused by an environmental factor such as trauma to the eye, and is not genetically linked.

The current PLL DNA test, based on the ADAMTS17 mutation, is not relevant to the Border Collie breed (Gould et al., 2011; Crispin, 2018). As such, until the mutation causing PLL in Border Collies is found, the only way to detect PLL in the breed is to schedule regular ocular examinations by an Australian Canine Eye Scheme (ACES) Panellist. The Kennel Club (UK) and the International Sheep Dog Society (ISDS) require yearly eye certification to check for conditions such as PLL, and ACES also recommends yearly tests. See the ANKC ORCHID (Officially Registered Canine Health Information Database) website for ACES Panellist contact details: http://orchid.ankc.org.au

Laboratories offering DNA Testing

MyDogDNA: https://mydogdna.com/products/mydogdna
Dog Breeding Science Australia (CA testing panel): https://breeding.dog/index.php?test=cam
Orivet (BC Breed Profile): https://www.orivet.com/store/canine-full-breed-profile/border-collie---full-breed-profile
Orivet (Sensory Neuropathy add-on): https://www.orivet.com/store/canine-disease/sensory-neuropathy-border-collie-type---single-assay-test
Animal Genetics (NCL5 test): https://www.animalgenetics.us/Canine/Genetic_Disease/NCL5.asp
Laboklin (including NCL5 test):
https://www.laboklin.co.uk/laboklin/GeneticDiseases.jsp?speciesID=BorderCollie&catID=DogsGD
PawPrint Genetics (including NCL5 test):
https://www.pawprintgenetics.com/products/tests/index/#idV
Embark (including NCL5 test:
https://embarkvet.com/breeders/

Panellists offering Hip and Elbow Scoring

ANKC Canine Hip and Elbows Dysplasia Scheme (CHEDS): http://orchid.ankc.org.au/
PennHip - Veterinarians taking X-Rays can submit PennHip-style radiographs to PennHip for a PennHip score. For more information about PennHip, see their website:
https://antechimagingservices.com/antechweb/pennhip

Panellists offering Ophthalmological Screening

ANKC Australian Canine Eye Scheme (ACES): http://orchid.ankc.org.au/