### PWD Health diseases and tests (Updated Sept 2023)

Genetic DNA tests are available for a number of diseases that have been found in PWD. The interpretation of the results and the subsequent actions of a breeder depend upon understanding some basic genetics. Dogs inherit 2 copies of each gene, one from each parent. If a dog inherits a faulty gene from one parent then the effect on the dog depends on whether that gene is dominant or recessive. If dominant it overrides the healthy gene and the dog will be “affected” . If recessive then the healthy gene overrides the faulty gene and the dog will be a “carrier”. If a dog inherits 2 faulty genes, one from each parent, then it will be affected regardless of whether the faulty gene is dominant or recessive.

CLEAR Clear means the dog has been tested for the disease-causing mutation and it has not been found. The dog will not develop the disorder and cannot pass them onto their offspring.

CARRIER A carrier is a dog which has inherited one mutated copy of a gene and one normal copy. Where the gene is recessive this means The dog will be healthy but can pass on the mutated copy to any offspring. A carrier should always be mated to a Clear partner, and never to another carrier.

AFFECTED Where the gene is dominant the dog will be affected even with one copy of the gene and has a 50% chance of passing it on to their offspring. Where the dog carries 2 copies of either a recessive or dominant gene it will be affected and will pass one copy on to its offspring.

Screening tests (as opposed to DNA tests) can only give breeders the tools to reduce the probability of a puppy being unhealthy. This applies to Hips, Elbows and Eye screening.

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| Screening test | **The disease** | **The Result of testing** | **How to get it done** |
| Hip score  Mandatory for Kennel Club Assured breeders, Strongly advised by the Kennel Club for other breeders. | Hip dysplasia is a complex inherited condition where the hip joint does not develop correctly. As a dog gets older, the joint undergoes wear and tear and deteriorates, leading to a loss of function. This can cause varying degrees of pain, discomfort, stiffness and lameness. | [The British Veterinary Association (BVA)](https://www.bva.co.uk/) and The Royal Kennel Club (RKC) Hip Dysplasia Scheme assess dogs' x-rays to look for abnormalities in hip joints. Hips are scored out of a possible score of 106. The current average score for dogs tested over the last 5 years is 11 at December 2022. The aim is to keep this score as low as possible when selecting a mate. | Contact your vet or another local vet and ask for the BVA hip score, your dog will need an xray, take the RKC registration document with you. The vet will submit the Xray to the BVA panel and advise you of the result. The BVA panel will also advise the Royal Kennel Club who will update your dog’s registration record. |

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| Screening test | **The disease** | **The Result of testing** | **How to get it done** |
| Elbow Score  Mandatory for Kennel Club Assured breeders, Strongly advised by the Kennel Club for other breeders. | Elbow dysplasia is a complex inherited condition where the elbow joint does not develop correctly. As a dog gets older, the joint undergoes wear and tear and deteriorates, leading to a loss of function. This can cause varying degrees of pain, discomfort, stiffness and lameness. | As for hips above there is a [The British Veterinary Association (BVA)](https://www.bva.co.uk/) and The Royal Kennel Club (RKC) Elbow Dysplasia Scheme which also uses an Xray. Elbows are graded 0 to 3. 0 being not affected. At Dec 2022 for submissions over the previous 15 years scores are  91% score 0, 7% score 1, 2% score 2, 0% score 3 (0 being not affected, 3 being badly affected) | As for hips above, it is recommended to get this done at the same time as the hip scores so only one anaesthesia/sedation is required. |
| BVA Eye screen | How an eye condition is inherited varies dramatically. Some eye conditions may be controlled by a number of different genes, as well as environmental factors. Others may be entirely controlled by just one gene and some may not be inherited at all. DNA testing alone will not identify all possible eye diseases. | The BVA /KC/ International Sheep Dog Society (ISDS) Eye Scheme offers breeders the opportunity of screening for inherited eye disease by examination of the eye. Examination under the eye scheme is not restricted to the identification of inherited eye disease, but also includes general assessment of the health of the eye and adnexa (eyelids, tear ducts and other parts around the eye ball). | [Eye panellists](https://www.bva.co.uk/canine-health-schemes/eye-scheme/find-an-eye-panellist/) appointed by the British Veterinary Association can carry out eye tests and can issue certificates under the scheme. Owners make an appointment with one of the eye panellists directly, or through their own veterinary surgeon. Often, breed clubs and general championship shows will arrange for a BVA panellist to attend their shows. This enables many dogs to be examined on one occasion at a reduced rate. A list of panellists can be found on the Royal Kennel Club Web site |
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| DNA test | **The disease** | **The Result** | **How to get it done** |
| JDCM (recessive) Juvenile Dilated Cardiomyopathy | Juvenile Dilated Cardiomyopathy is a rare, fatal condition caused by a recessive gene. It affects young dogs, who succumb to heart failure before reaching adulthood, normally at around 12 weeks. As a simple recessive gene, it had been difficult to identify and was particularly heart breaking as seemingly healthy puppies would suddenly die, often shortly after joining their adopted families. Since a recessive gene is responsible, that means if at least one parent is "clear (or normal)" (that is, it does not carry a copy of the cardio version of the gene), its offspring cannot contract the disease. | In 2007 a genetic linkage test became available which links certain sections of DNA to the probability of being carriers of JDCM. The test is not 100% definitive. However if the result is “Probable Normal” then there is only a slight chance that the dog could be a carrier and an even smaller chance it could be affected. It is safe to mate a carrier with a probable normal dog as puppies can only be carriers or probable normal. | The test is only available at PennVet, in the USA. Search for PennGen DNA tests. Book the test on line. **Sample Types:**Fresh EDTA blood or Cheek brushes/swabs  The Micropthalmia test below can be done from the same sample. |
| MO-PWD Microphthalmia Syndrome (recessive) | This disease is characterized by microphthalmia (abnormally small eyes) and other ocular abnormalities, such as persistent pupillary membranes, glaucoma, retinal degeneration and cataracts. Other signs may include stunted growth, decreased platelet counts, anaemia, and possibly behavioural abnormalities. Affected puppies are often euthanized due to the lack of vision, and in some instances to behavioural problems. | As a recessive gene two copies would be required to be affected. It is safe to mate a carrier with a clear dog as puppies can only be carriers or clear. | Also available ONLY from PennVet, in the USA. Search for PennGen DNA tests. Book the test on line. **Sample Types:**Fresh EDTA blood or Cheek brushes/swabs  The JDCM test above can be done from the same sample. |

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| DNA test | **The disease** | **The Result** | **How to get it done** |
| GM1 (recessive) Ganglidiosis  Sep 23 Recommended by the RKC. Recorded on Registration records by the Kennel Club | GM1 Storage Disease, or gangliosidosis, is a recessive, genetic disorder that is inevitably fatal. It is caused by a deficiency of beta-galactosidase, with resulting abnormal storage of acidic lipid materials in cells of the central and peripheral nervous systems, but particularly in the nerve cells. Thanks to the availability of a genetic test PWDs who are GM1 Storage Disease carriers are able to be genetically identified, and the condition has now been almost entirely eliminated from the breed. It is a fatal nerve disease that typically appears when a puppy is approximately six months of age. The affected puppy will show clinical signs of cerebellar dysfunction including ataxia, tremors, paresis, and seizures. The pet may also exhibit a change in temperament. Lesions of the retina and clouding of the cornea may occur. | Thanks to the availability of a genetic test PWDs who are GM1 Storage Disease carriers are able to be genetically identified, and the condition has now been almost entirely eliminated from the breed. As a recessive gene two copies of the gene would be required to be affected. It is safe to mate a carrier with a clear dog as puppies can only be carriers or clear. | Several laboratories offer this test including:  Laboklin and MydogDNA Both of these labs also do CDDY, CDPA, EO-PRA, prcd-PRA  The Royal Kennel Cub (which also do prcd-PRA  Cheek swab or Blood sample required. |

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| DNA test | **The disease** | **The Result** | **How to get it done** |
| Prcd-PRA (recessive) Progressive Retinal Atrophy  Sep 23, Mandatory for litter registrations. Recorded on Registration records by the Kennel Club | The cells of the retina receive light from the external environment and transmit the information to the brain where it is interpreted to become vision. PRA causes cells in the retina at the back of the eye to degenerate and die, even though the cells may have developed normally early in life. The cells of the retina receive light from the external environment and transmit the information to the brain where it is interpreted to become vision. Owners of affected dogs first notice that their dog becomes night blind, but this eventually progresses to total [blindness](https://www.thekennelclub.org.uk/health-and-dog-care/health/health-and-care/a-z-of-health-and-care-issues/a-guide-to-blindness-in-dogs/). The age of onset of first signs varies from breed to breed, however, in all cases puppies are born with perfect vision and their sight begins to degenerate later in life, from around 3 years of age or later. | As a recessive gene two copies would be required to be affected. It is safe to mate a carrier with a clear dog as puppies can only be carriers or clear. | Several laboratories offer this test including:  Laboklin and MydogDNA Both labs also do CDDY, CDPA, EO-PRA, GM1  The Royal Kennel Cub (which also do GM1  Cheek swab or Blood sample required. |
| EO-PRA (recessive) Early onset Progressive retinal atrophy  Recorded on Registration records by the Kennel Club | Early onset PRA has the same effect as prcd-PRA above but affects younger dogs, usually around the age of two to three years old. | As a recessive gene two copies would be required to be affected. It is safe to mate a carrier with a clear dog as puppies can only be carriers or clear. | Several laboratories offer this test including:  Laboklin and MydogDNA  Both labs also do CDDY, CDPA, GM1, prcd-PRA  Cheek swab or Blood sample required. |
| CDPA Chondrodysplasia (recessive) | Chondrodysplasia is an inherited disorder that affects the way that bones develop. The condition is a type of dwarfism that causes affected dog's limbs to be noticeably shorter than unaffected dogs. | As a recessive gene two copies would be required to be affected. It is safe to mate a carrier with a clear dog as puppies can only be carriers or clear. | Offered by UC Davis in the USA who are conducting a study with PWD, and also Laboklin and MydogDNA (both of these also do CDDY, prcd-PRA and EO-PRA, and GM1) |
| DNA test | **The disease** | **The Result** | **How to get it done** |
| CDDY (dominant) Chondrodystrophy with IVDD risk | Chondrodystrophy  is a trait that is common to many dog breeds and it is characterised by shorter legs due to shorter long bones. It also changes the character of all of the intervertebral discs at a young age. The discs have abnormal degeneration of the nucleus pulposus, which is the centre of the intervertebral disc that normally provides cushion and flexibility to the back. The end of the degeneration process is a mineralized or calcified disc. The change in the cellular structure of the disc is what predisposes it to herniate (move into the spinal canal impinging on the spinal cord). The chondrodystrophic degenerative phenotype is evident in all the intervertebral discs as early as 10 weeks of age in dogs carrying either one or two copies of the CDDY gene but is absent in those without it. (Murphy et al., 2019). This has only very recently been discovered in PWD but is endemic in some short legged breeds like Beagles and Dachshunds where every dog carries 2 copies of the gene.  As short legs are not required in PWD this is not a gene that we need!  Therefore the aim of breeders is to eliminate this gene within the next 2 or 3 generations at most.  Although there have been some affected dogs that have been put to sleep due to spine degeneration, there are many more who have lived with no apparent symptoms well into old age. It is unclear if there may be a masking gene or some other characteristic of PWD which limits the effect of this disease. | The CDDY gene is dominant so a dog with only one copy is affected. It is not yet endemic in PWD so we have the opportunity to eliminate it within 2 or 3 generations by careful breeding without discarding breeding dogs. However it requires all breeders to co-operate. The PWD Club of GB have looked at the recommendations of scientists, note ably at UC Davis and recommend that, if it is avoidable, breeders should not breed from affected dogs with one copy of the gene and definitely do not breed from a dog with 2 copies of the gene. If an affected dog is bred from only use a clear mate and any puppies that may be used in a breeding program in the future must be tested and so far as possible, only clear puppies retained for breeding.  The club’s view is that to eliminate all affected dogs would be detrimental to the gene pool, therefor breeders are asked to follow the above advice. | Offered by UC Davis in the USA who are conducting a study with PWD, and also Laboklin and MydogDNA (both of these also do CDPA, prcd-PRA and EO-PRA, and GM1) |

## Laboratories that The Royal Kennel Club record and publish the results from

The KC have a criteria that they request DNA testing laboratories meet to enable them to record their results, helping to maintain and protect the integrity of results that appear on a dog’s record.  
  
The KC strongly advise that customers ensure their chosen laboratory is included on their list below if they wish for them to record and publish the results. Results from laboratories not included on this list will not be recorded.

Read the full list of UK laboratories

* [Animal DNA Diagnostics](http://www.animaldnadiagnostics.co.uk/) (UK)
* [Animal Genetics](http://www.animalgenetics.eu/) (UK)
* [Canine Genetic Testing](http://www.cagt.co.uk/) (UK)
* [Laboklin](http://www.laboklin.co.uk/)(UK)
* [Pinmoore Animal Laboratory Services](http://www.palsvetlab.co.uk/) (UK)
* [The Kennel Club's DNA Testing Services](https://www.thekennelclub.org.uk/shop/health/) (UK)

Read the full list of overseas laboratories

* [Alfort School of Veterinary Medicine](http://www.vet-alfort.fr/)(France)
* [Antagene](http://www.antagene.com/)(France)
* [Auburn University](https://www.auburn.edu/)(USA)
* [Bochum University](https://www.ruhr-uni-bochum.de/en)(Germany)
* [Cornell University](https://ahdc-portal.vet.cornell.edu/#!/test_fee/search)(USA)
* [Embark](https://embarkvet.com/)(USA)
* [FERAGEN Genetic Laboratory](http://www.feragenlab.com/dog-dna-testing/)(Austria and Germany)
* [Genetic Technologies Ltd (Animal Network)](http://www.animalnetwork.com.au/tests/)- Australia
* [Genindexe](http://www.genindexe.com/)(France)
* [Genomia](http://www.genomia.cz/)(Czech Republic)
* [HealthGene](http://www.healthgene.com/)(Canada)
* [Hospital for Sick Children](http://www.sickkids.ca/)(Canada)
* [Michigan State University](http://www.mmg.msu.edu/)(USA)
* [MyDogDNA](https://www.mydogdna.com/)(Finland)
* [Orthopedic Foundation for Animals](https://www.ofa.org/diseases/dna-tested-diseases)(USA)
* [Paw Print Genetics](http://www.pawprintgenetics.com/)(USA)
* [PennGEN Laboratories](http://research.vet.upenn.edu/Default.aspx?alias=research.vet.upenn.edu/penngen)(USA)
* [University College, Dublin](http://www.ucd.ie/)(Ireland)
* [University of Bern](http://www.unibe.ch/eng)(Switzerland)
* [University of California - Davis Veterinary Genetics Laboratory](http://www.vgl.ucdavis.edu/services/dog.php)(USA)
* [University of Minnesota - Veterinary Diagnostic Laboratory](https://www.vdl.umn.edu/)(USA)
* [University of New South Wales](http://www.unsw.edu.au/)(Australia)
* [University of Pennsylvania](http://research.vet.upenn.edu/AvailableTests/TestsAvailableatPennGen/tabid/8242/Default.aspx)(USA)
* [University of Utrecht](http://www.uu.nl/en)(Holland)
* [Van Haeringen](http://www.vhlgenetics.com/)(Holland)
* [Veterinary Diagnostics Centre (DDC)](http://www.vetdnacenter.com/)(USA)