

Genetic Diseases in Poodles

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This article will focus on the known heritable diseases in all three varieties of poodle, as well as current information on mode of inheritance, if available, and any updates on clinical signs and treatments. If any readers have updated information not included in this article, please respond by email to tivin08@yahoo.com. The following diseases will be covered: Addison's disease, Cushing's disease, cerebellar abiotrophy, cerebellar hypoplasia, degenerative joint disease (often manifesting in hip dysplasia), entropion and ectropion, epilepsy, gastric dilatation and volvulus, hypothyroidism, juvenile renal disease, Legg-Calve-Perthes syndrome, monorchid and cryptorchidism, optic nerve hypoplasia, patellar luxation, patent ductus arteriosus, progressive retinal atrophy, sebaceous adenitis, and von Willebrand's disease.

Addison's disease is otherwise known as hypoadrenocorticism, or under production of corticoids, which are important regulatory hormones. There are two classifications of Addison's disease: 1) primary, in which destruction of the adrenal cortex results in inadequate levels of glucocorticoids and mineralocorticoids, and 2) secondary, in which decreased stimulatory hormones from the pituitary gland results in deficiency in cortisol secretion. Most dogs have primary Addison's disease, which is theorized to be the result of immune-mediated destruction of the adrenal glands. Clinical signs include vomiting, lethargy, weight loss, anorexia, and bradycardia (slow heart rate). Some dogs also exhibit increased drinking and urination and severe cases can present with dehydration and shock.

Upon presentation to a veterinarian it is important to distinguish between primary renal (kidney) disease and Addison's disease as clinical signs are very similar. Addisonian patients will typically have secondary renal failure but will respond to intravenous fluids. It is important during the initial presentation to administer steroids as well as potassium-free intravenous fluids as these are the items an Addisonian patient is lacking. The steroids and fluids will treat for shock. The good news is that an Addisonian patient will respond quickly to appropriate treatment and a dog which the owner thought was dying can be up and barking within a few hours or less. The most common test used to confirm Addison's disease is the ACTH stimulation test in which cortisol levels are measured before and 1 hour after the injection of an adrenocortical stimulating hormone. Addison's disease can occur at any age and with either sex, although in dogs it is more common in females.

Addison's disease is very easy to treat with monthly administration of injectible mineralocorticoids and oral prednisone if needed. The prognosis for a normal life expectancy is good as long as the affected dog is treated at appropriate monthly intervals. Unfortunately, the injectible medications can be quite expensive for larger dogs, although the price has decreased over time. There is no published genetic basis for Addison's disease although there are at least two veterinary universities searching for the gene or

genes affected. Both standard poodles and Portuguese water dogs are involved in studies, and the genetic basis is thought to be similar for both breeds. Addison's disease also affects humans, and our past president, John F. Kennedy, was an Addisonian. In the past, human Addison's disease often occurred secondary to tuberculosis. This is a much less common cause today. Addison's disease has been linked to immune-mediated problems and may be polygenic (multiple genes) in nature.

Conversely, Cushing's disease can be thought of as the opposite of Addison's disease. Cushing's is otherwise known as hyperadrenocorticism, or excess of cortisol production. There are three categories of Cushing's disease: 1) pituitary dependent, in which the pituitary gland secretes excess adrenal stimulating hormone which results in the adrenal glands overproducing cortisol. Approximately 95% of the time pituitary dependent Cushing's disease is the result of a slow-growing tumor, commonly an adenoma, on the pituitary gland; 2) functional adrenal tumors, usually on only one of the two glands. The tumor on the adrenal gland does not respond to feedback signals from the pituitary and secretes cortisol at high levels. The non-affected adrenal gland often atrophies, or shrinks, as a result; 3) iatrogenic Cushing's caused by excessive or prolonged administration of steroids (often prednisone) resulting in atrophy of the adrenal glands.

Common clinical signs for Cushing's disease include excessive drinking and urination, increased appetite, pot-belly appearance, muscle weakness, exercise intolerance, panting, lethargy, poor wound healing, hyperpigmentation (increased freckling or darkening) of the skin, hair loss, poor coat condition, and calcinosis cutis (small calcium deposits in the skin). Central nervous system signs can occur in cases which involve large pituitary tumors. Suspicion of Cushing's disease can be seen on a general blood chemistry panel which shows increased alkaline phosphatase (ALKP) and alaminotransferase (ALT), both enzymes involved with the liver, as well as increased cholesterol production, increased glucose, decreased blood urea nitrogen (BUN - kidney enzyme), and decreased thyroid hormone (T4). The ACTH stimulation test mentioned in Addison's disease can also be used to determine if a dog has Cushing's disease. However, a positive result does not tell the veterinarian which type of Cushing's is present. It is recommended to follow a positive ACTH stimulation test with an abdominal ultrasound to visualize the adrenals. The ultrasonographer should be familiar with looking for adrenal glands as they are very small and can be difficult to locate. Once the adrenal glands are visualized it can be seen if there is an adrenal tumor present or if the glands are increased in size due to over stimulation from the pituitary gland. There are also two other tests which can be used to diagnose Cushing's disease – the low dose dexamethasone suppression test and the high dose dexamethasone suppression test. Both of these tests involve administering a specific amount of dexamethasone to the patient and measuring cortisol levels 8 hours later.

While Cushing's disease is less immediately life threatening than Addison's disease, treating it can be trickier. Treatment of pituitary dependent Cushing's involves administering mitotane (Lysodren) which is toxic to particular layers of the adrenal glands. The goal is to administer just enough to decrease the production of cortisol to a

normal level. If too much is administered, the dog can become Addisonian. Administration of lysodren involves dedicated care by the owner as close observation of the dog and multiple veterinary visits are required. Treatment of dogs with adrenal tumors can be successful with surgical removal of the affected gland. If both glands are affected surgery is not recommended. Treatment of iatrogenic Cushing's involves slowly weaning the dog off steroids.

Most Cushingoid animals do not live out the normal life expectancy. There are many side-effects due to increased steroid production, including increased risk for thromboembolism (blood clots in the blood vessels), renal failure, and liver disease. At this time, there does not appear to be a known genetic basis for Cushing's disease although it is theorized that the pituitary tumors may run in families.

One of the most hotly debated subjects amongst dog fanciers is the cause and/or genetic basis of degenerative joint disease (DJD), also known as hip dysplasia. There is no doubt there is some genetic basis for DJD, but it is now widely accepted that there may be multiple contributing factors including a genetic predisposition toward poor joint conformation as well as environmental and dietary factors. Hip dysplasia is a polygenic (more than one gene) hereditary trait that can occur in any breed of dog as well as cats. The hip joint is normal at birth, but develops abnormally resulting in distortion of the acetabulum or "socket" of the joint and subsequent remodeling of the head of the femur or "ball" portion of the joint. This distortion can cause loss of cartilage, severe remodeling of the joint architecture, and destabilization of the joint resulting in pain and inflammation.

Clinical signs include lameness, difficulty getting up or lying down, reluctance to go up and/or down stairs, muscle atrophy, a "hopping" gait, decreased willingness to exercise, and pain upon palpation of the joint. Diagnosis is usually made with radiographs, or x-rays of the pelvis, although physical exam can often provide strong suspicion. Veterinarians will sometimes refer to a positive "Ortolani sign" which refers to the ability to push the femur up and out from the hip socket and feel it snap back into place. A positive Ortolani sign means that the hip joint is excessively lax or loose and hip dysplasia is present. Radiographs will confirm a positive Ortolani sign and will determine the extent of degeneration in the hip joint; this will lead to determining which options for treatment are most feasible. Severely affected animals can show signs of DJD as early as 5-6 months of age.

Treatment of DJD can be divided into medical and surgical options. Medical options include weight control (the thinner the better within healthy limits), pharmaceutical intervention including non-steroidal anti-inflammatories, steroids in severe cases, dietary supplements including glucosamine chondroitin, polyglucosaminoglycans, perna muscle and others, and a low impact exercise program such as swimming.

When medical treatment is not enough to alleviate the pain, surgical options are also available. These include a triple pelvic osteotomy (TPO), total hip replacement, and

femoral head ostectomy (FHO). The triple pelvic osteotomy is a surgery that can only be performed in young animals before significant degenerative changes in the joint have occurred. The pelvis is cut at three sites – the ilium, the ischium, and the pubis, and then rotated so that the hip joint is more stable, i.e. the femur sits more solidly in the socket, or acetabulum. The dog must have strict cage rest for 4-6 weeks following the surgery, and pain management is essential. Prognosis for a normal life with appropriate weight control and exercise is good following most TPOs. This surgery is complex and does require a board certified orthopedic surgeon. Most dogs are too old or have too much damage to the joint when DJD is diagnosed for the TPO to be an option.

The next best surgery when necessary is the total hip replacement. This involves removing the head of the femur and replacing it with a metal implant. Recovery also involves strict cage rest, usually 6-8 weeks, and post-operative pain management. Most dogs return to nearly full function and activity level, with a good quality of life. Failure of the surgery can include luxation or dislocation of the implant from the socket, loosening of the implant from the femoral site, infection of the surgical site, and rejection of the metal implant. Most dogs can do well with only one hip replaced, although a small percentage of dogs do need both hips replaced, in which case the surgeries are typically done one at a time. This surgery does tend to be quite costly, as is the TPO.

The last surgical option, sometimes referred to as a salvage procedure, is the femoral head ostectomy. This involves removing the head and neck of the femur, or the area which usually forms the “ball” part of the ball and socket hip joint. The fascial tissue and overlying muscle of the joint then form a pseudoarthrosis or false joint. By eliminating bony contact between the femur and the pelvis, and allowing formation of a false joint, pain is alleviated. There can be some loss of range of motion and occasionally abnormal gait. It is essential that weight management and low-impact exercise are a part of recovery and post-operative life style for the dog. As with the total hip replacement, most dogs only require one side to be surgically altered to maintain a good quality of life.

There is no genotypic, or DNA test available to determine carriers of DJD. At this time, prevention is encouraged by testing breeding animals with radiographic screening including the Orthopedic Foundation for Animals (OFA) evaluations, or the PennHIP method, which are the two most common in the United States. Canada and Europe sometimes use the Norberg angle or other methods of screening. OFA evaluations must be done at two years of age or older, and involves taking a view of the pelvis and stifles with the legs fully extended and the dog on his/her back. This may be done with or without sedation. The x-rays are reviewed by three board-certified radiologists and given a subjective rating of dysplastic, fair, good, or excellent, depending upon hip conformation. PennHIP screening can be done as early as 4 months of age, and involves taking three radiographic views under sedation. The x-rays include the extended view as taken by the OFA, and two additional views called the compression and distraction radiographs. These views are taken to evaluate the laxity or looseness of the hip joint. The x-rays are then compared and an objective measurement is taken resulting in a percentage rating from a scale of zero to one hundred. The closer to 100%, the tighter the hips are for a given individual.

Another disease with genetic predisposition is entropion and ectropion. Entropion involves turning of the eyelid inward, toward the cornea, often resulting in rubbing of the hair against the cornea and causing inflammation and ulcerations. Ectropion involves turning of the eyelid outward, often resulting in inflammation, decreased tear production, and increased irritation to the cornea and surrounding structures. Both diseases can sometimes occur secondary to other non-genetic causes, such as damage to the eyelid itself (insect bite, laceration), other inflammatory causes such as allergic reactions with subsequent eyelid swelling, and infections of the eyes or ears with subsequent inflammation and itching. If the underlying problem is not genetic in nature, resolving the primary cause will often result in resolution of the entropion or ectropion. If the underlying cause is genetic in nature, usually the only resolution is surgical correction. This involves trimming of the eyelid itself to correct the conformational defect. There is no DNA test for either disease. There is a known genetic predisposition for both diseases.

Epilepsy is a disease that affects a variety of species including humans and dogs. It is known to be a genetic problem in all varieties of poodles and studies are being conducted to determine genotype, or DNA markers. It is believed to be at least partially recessive in nature, but may be polygenic in some cases. There is an ongoing study at Florida State University called the Poodle Epilepsy Project concerning this disease in poodles. The first signs of epilepsy most commonly manifest between one and five years of age. Clinical signs of epilepsy can range from mild seizures that the owner may not even be aware of, to more severe seizures such as petit mal and grand mal types. Before a diagnosis of epilepsy is determined, all other causes of seizures must be ruled out. Other common causes of seizures include metabolic disturbances such as liver insufficiency (which may be secondary to a vascular shunt in newborn and young animals), toxin ingestion, nutritional deficiencies, infectious agents, middle ear infections, and central nervous system (CNS) problems such as brain tumors or hydrocephalus (a congenital defect in which there is an accumulation of fluid in the brain).

Typically, after a thorough physical exam, a veterinarian will run a complete chemistry panel and blood count to rule out metabolic and toxic causes of seizures. If there is a family history of epilepsy, genetic causes may be more likely. In many cases, family history is unknown, and a veterinarian must rely upon available and reasonable testing. For instance, it is possible to run a magnetic resonance imaging (MRI) or computed tomography (CT) scan of the brain, but these procedures require general anesthesia in the dog, and are often prohibitively expensive for owners. Therefore, when all other causes of seizures have been ruled out, and CNS causes are unlikely, the patient is usually considered an epileptic and may be put on anti-seizure medications if indicated.

The most common long term medications for epilepsy include phenobarbital and potassium bromide. If a dog presents to a hospital currently seizing, the veterinarian will administer a fast-acting anti-seizure medication, usually valium or pentobarbital. Occasionally, a patient will require a combination therapy of both phenobarbital and potassium bromide. It is important that, regardless of which type of medication is

prescribed, the dog's seizures are controlled. There should also be six month follow-up visits to measure the therapeutic level of the drug in the bloodstream as well as to compare chemistry panels for monitoring of general organ function. Phenobarbital and to a lesser extent, potassium bromide, can affect liver function. Early adjustment to the drugs can also cause increased drinking and urination, increased appetite, sedation, ataxia (difficulty walking), and hyperexcitability. These side affects usually decrease or disappear over time, as the body adjusts to the drug.

One of the most frightening and fatal diseases seen in poodles of the standard variety is gastric dilatation and volvulus (GDV). This occurs when there is accumulation of gas in the stomach causing distention, sometimes followed by displacement and torsion, or twisting of the stomach. When the stomach twists upon itself, the major blood supply to the lower half of the body, mainly the gut, is cut off. When this happens, the stomach, intestines, and spleen become oxygen deprived and begin to die. If the GDV is not corrected surgically, the dog will die, often in agonizing pain.

Clinical signs for GDV include distended abdomen (often described as the size of a basketball or bigger), difficulty breathing, white or very pale gums, intermittent retching, percussion of the abdomen (pings indicating air filled stomach instead of fluid filled), and shock leading to collapse. Upon presentation, the dog must be stabilized before attempting surgery. This includes decompressing the stomach with a tube if possible, if not than with a large bore needle through the abdominal wall, intravenous fluids, pain medications, oxygen therapy if needed, and electrocardiogram (ECG) monitoring for heart arrhythmias. Once stable, the patient can be prepped for surgery and the stomach is rotated back into place then "tacked" (known as a gastropexy) to prevent future rotation. Occasionally, portions of the stomach may need to be removed if the area has been deprived of oxygen too long, and often a splenectomy, or removal of the spleen, is also performed for the same reasons. Unfortunately, too often the GDV is not discovered in time, and too much of the gut has died to perform surgery. Even when surgical correction is attempted, at least 50% of dogs have poor recovery rates.

The genetic basis for GDV appears to be more conformational than genotypic, i.e. there has not been a discrete gene isolated for the disease. It is most common in large breed, deep-chested dogs. There have been several theories on diet, feeding habits, exercise habits, etc. related to the onset of the disease; at this time many articles have been published, some in direct contradiction of others on advice for decreasing risks for the disease. Several veterinary universities have studied GDV in a variety of breeds, including the poodle. At this time, there is no defined screening test available to determine if your dog will have GDV or produce it.

Hypothyroidism, or decreased production of thyroid hormone, is the next disease to be discussed; it may have underlying causes in autoimmune mediated processes similar in nature to Addison's disease which has already been discussed, and sebaceous adenitis, which will be discussed later. All three of these diseases are most common in the standard poodle. There is no DNA test for hypothyroidism, although it does appear to have familial history. The most common cause of hypothyroidism is destruction of the

thyroid gland itself, resulting in loss of thyroid hormone, and display of related symptoms.

Clinical signs of hypothyroidism include dry, flaky skin, loss of hair (particularly on the hind end), poor hair coat, “hotspots” or skin infections, thickened skin, weight gain, and ear infections. In severe cases the heart can have decreased contractility resulting in a slow heart rate, and neuromuscular signs such as seizures, facial paralysis, and diffuse muscle weakness can occur. Reproductive signs include infertility and lack of libido, as well as lack of heat cycles. Most hypothyroidism is diagnosed by running a thyroid panel including T3 and T4 levels (types of thyroid hormone), thyroid stimulating hormone (TSH) levels, and autoantibodies to T4 and TSH. The disease is very easy to treat, with daily supplementation of thyroid hormone. It is also generally inexpensive, and requires lifelong treatment.

Juvenile renal disease (JRD) is a genetic problem in several breeds, including poodles, that is heartbreaking in nature. Often associated with “fading puppy syndrome” or with the early death of a young dog (usually under 2 years), JRD can be devastating. Clinical signs of JRD include the unexplained death of neonatal and young puppies. Affected puppies often appear normal at birth but then become lethargic, weak, decrease nursing, and become developmentally delayed just seeming to fade away and die. The only way to diagnose JRD in these puppies is through necropsy, or post-mortem pathological examination of the gross and microscopic changes in the kidneys, indicating renal failure. Puppies who survive the neonatal period are often smaller in stature, display increased drinking and urination, increased urinary “accidents” in the house and may have difficulty housebreaking due to the volume of dilute urine produced. As the disease progresses dogs will become lethargic, exercise intolerant with muscle weakness and wasting, vomiting, and anorexic. While a blood chemistry panel can show renal deficiencies, the most accurate method of diagnosing JRD is with a wedge biopsy taken from the kidneys, either surgically or with ultrasound guided biopsy.

Treatment for JRD is more palliative in nature, increasing comfort level for a period of time until renal failure has progressed to its terminal stage. Such treatment includes a low protein and low phosphorus diet, to decrease the work that the kidneys need to perform, as well as increased fluid consumption, either through subcutaneous or intravenous methods, and administration of epogen when anemia becomes a problem. It is possible to perform a kidney transplant, but organ rejection rate is high and few hospitals will perform the procedure. The University of California at Davis Veterinary School is working on improving the success rate of kidney transplants in dogs, and the University of Pennsylvania Veterinary School routinely performs kidney transplants in cats, and rarely dogs. If kidney transplant is an option the owner wants to consider, chances for a successful outcome will increase if a sibling or close relative is available as a donor. The procedure is expensive and requires intensive care post-operatively as well as commitment to close monitoring for the remainder of the dog’s life.

There have not been any DNA markers identified for the gene(s) involved in JRD. It is theorized to be a simple recessive, requiring both parents to be carriers. Pedigree

analysis in various affected breeds does show that JRD will skip generations, be expressed within a litter at a typical recessive percentage, and will recur in repeat breedings of the same sire and dam. Oftentimes a breeder may not realize that he/she has a carrier in her line, if pet owners do not report the diagnosis or death back to the breeder. Of the three varieties, JRD is most common in the standard poodle.

Another orthopedic genetic disease similar to hip dysplasia is Legg-Calve Perthes disease (LCP). In this disease the blood supply to the femoral head of the hip joint is disrupted with resulting death of bone cells and remodeling of the joint. This leads to deterioration of the hip joint with clinical signs of stiffness and pain. This disease appears to affect mainly toy and miniature poodles, and has conflicting theories of being either a simple recessive or polygenic in nature. There is no DNA marker test for LCP at this time.

LCP occurs in humans as well, affecting boys by a large percentage over girls. It is being studied in the medical community and research for DNA markers for the human form of the disease is underway. Diagnosis in both humans and dogs is obtained through x-rays of the hip joints, and OFA has set up a database for breeders. As with severe hip dysplasia, surgery is the most successful form of treatment, however non-steroidal anti-inflammatories and nutritional supplements can be of some help in earlier stages of the disease. As with DJD, clinical signs of LCP can occur as early as 5-6 months of age but may show up as late as 2-3 years. Symptoms of LCP are very similar in nature to DJD with joint pain, stiff gait, difficulty getting up or lying down, exercise intolerance, lameness, and muscle atrophy.

A genetic abnormality which is not life-threatening is monorchidism (only one testicle descended) as well as bilateral cryptorchidism (neither testicle descended). Bilateral cryptorchidism results in sterility as sperm cannot survive in the elevated body temperature of the abdomen versus the scrotum. Monorchids have normal sperm production in the descended testicle, but are sterile in the retained testicle. Monorchids are able to sire puppies if bred, but there is a familial tendency to pass on the trait, and sons of monorchids will often also be monorchid. There is no DNA marker for this abnormality. It is recommended to neuter affected animals, and the AKC does not allow monorchids to compete in conformation events. It is imperative that testicles retained in the abdomen be removed, as the elevated body temperature often induces testicular cancer.

Patellar luxation is another orthopedic disease which can affect all three varieties of poodle, but is most common in toys and miniatures. Like DJD, it is theorized to be a polygenic trait, with resulting conformational abnormalities of the stifle, or knee joint. There are two forms of patellar luxation: 1) medial, where the patella or kneecap luxates, or slides out of joint toward the inside of the leg, and 2) lateral, where the patella luxates out of joint toward the outside of the leg. In either case, clinical signs include intermittent lameness, a hopping gait with the affected leg held up and out, a bow-legged appearance, and occasionally a knock-kneed stance. The majority of patellar luxation cases are medial in nature.

Diagnosis of patellar luxation can be obtained through palpation of the stifle. Either one or both stifles may be affected. There are four categories ranging the severity of the patellar luxation: 1) grade one, intermittent luxation, in which the patella is loose in the groove of the joint, so it luxates with palpation, but mainly stays in the correct position. Surgical correction is not necessary with grade one. 2) grade two, frequent patellar luxation, in which the patella luxates easily out of joint and stays out when the leg is flexed. It pops back into place when the leg is extended, sometimes eliciting a yelp from the dog. Surgical correction may be an option if the dog appears to have increasing discomfort and frequency of luxation. 3) grade three, permanent patellar luxation, in which the patella stays out of the groove on a regular basis, but can be replaced with digital pressure only to relaxate when the pressure is released. The groove of the joint is shallow, and there are often abnormalities of the tibia as well. Although many animals can walk with grade three, surgical correction is recommended as arthritis and pain will result from instability in the joint. 4) grade four, also permanent luxation, in which there are deviations in the tibia and the patella is permanently luxated. The luxation cannot be corrected with digital pressure. The dog cannot walk normally on the affected leg and it is either held upright or the dog walks with a crouched appearance. Again, surgical correction is recommended.

The next disease to be discussed is patent ductus arteriosus (PDA), which also affects all three varieties of poodle, but like patellar luxation is more common in toys and miniatures. PDA is also more common in females than males. Patent ductus arteriosis is a defect in the heart in which a small blood vessel (the ductus arteriosus) which connects the pulmonary artery (which carries blood to the lungs) and the aorta (which carries blood to the rest of the body) does not close as it should. During gestation, the lungs are non-functioning and the fetus gets all of its blood oxygen from the mother. Therefore, the blood must be shunted away from the deflated lungs, which is the function of the ductus arteriosis. Upon birth, when the animal begins to breathe air, there is a shifting of pressure in the lungs and the ductus arteriosus should close. When there is a failure to close a PDA results, and the workload of the heart increases. There is greater pressure for the heart to work against, and untreated, a PDA can lead to heart failure.

Clinical signs include a heart murmur heard upon auscultation with a stethoscope, lethargy, exercise intolerance, panting, and sometimes smaller stature than unaffected littermates. An echocardiogram, or ultrasound, of the heart will confirm a PDA and the degree to which the heart has become affected (enlarged ventricle of the heart muscle, dilation of the left atrium, hypercontractility). Cardiac catheterization and angiogram of the heart are more accurate in diagnosing PDA in smaller breeds, measuring pressure changes and oxygen saturation. There is no DNA marker in PDA, although there is a noted familial tendency and inheritance is theorized to be polygenic.

The only treatment option for extension of life span is surgical correction. Left untreated, a PDA will lead to heart failure and death. Surgery involves ligation, or closure, of the ductus arteriosis vessels. Mild to moderate cases can be treated with a “coil” procedure in which a the patient is placed under general anesthesia and a small coil is inserted via the jugular vein down into the ductus arteriosis and then manipulated in

such a manner as to close the vessel. Occasionally more than one coil is required for complete closure. More severe cases, and cases in which the coil procedure fails, may require open heart surgery to correct. There are many complications with open heart surgery, and most dogs do not live a normal life span post-operatively at this time. Open heart surgery can also be prohibitively expensive to many owners. In mild to moderate cases, prognosis for a normal life span is excellent with surgical correction prior to heart failure.

Another disease which also affects all three varieties of poodle is progressive retinal atrophy (PRA). This is an ophthalmic group of diseases which affects the retina, or imaging layer in the back of the eye. Over time, the size of the reflective layer of the retina increases, which the non-reflective layer atrophies along with the optic disc. Clinical signs can occur as late as 3-4 years of age or older and usually manifest first as decreased night vision or “night blindness” eventually leading to total loss of vision as the disease progresses.

The most common form of PRA in toys and miniatures is progressive rod-cone degeneration (prcd) and can be tested for by DNA markers offered by Optigen. Dogs can be identified as clear (normal vision, cannot pass on to offspring), carriers (normal vision, can pass on the disease to offspring), and affected (progressive blindness, can pass on the disease to offspring). PRA is inherited as a recessive gene, thus carriers are normal appearing individuals but can pass it on to offspring if bred with another carrier. There is no DNA test available for standard poodles at this time, although yearly eye examinations and Canine Eye Registration Foundation (CERF) testing is available for all three varieties. There is no current treatment to slow or stop the progression of PRA.

Optic nerve hypoplasia is another eye disease which results in blindness and may be a heritable condition in all three varieties. At this time, it is more often reported in miniature poodles, but the mode of inheritance is unclear. The disease is an uncommon defect in which the optic disc fails to develop normally, leading to partial or total blindness in the affected eye. Puppies will be born with the defect, and may not exhibit symptoms if only one eye is affected. Puppies are very good at adjusting to one sided vision and often compensate for the unilateral sight, appearing relatively normal. It is important to distinguish optic nerve hypoplasia from micropapilla, in which the optic disc appears smaller but vision is normal. There is no treatment for optic nerve hypoplasia, but whether one or both eyes are affected, dogs can often adjust quite well relying upon their other senses.

Along with Addison's disease, sebaceous adenitis (SA), the next disease to be discussed, is considered to be a possible immune-mediated condition. While present in all three varieties, SA is most often manifest in the standard poodle. There have been suggestions that SA is inherited as a simple recessive, but it appears that the mode of inheritance is likely more complex, and may be polygenic in nature. SA occurs when there is destruction (theorized to be immune-mediated) of the sebaceous glands in the skin, resulting in loss of sebum (a fatty substance which lubricates the skin) and decreased suppleness of the skin itself.

Clinical signs usually appear in young adult dogs 1 year of age or older. SA usually begins with an increase in scale production in the skin, making a flaky or dry skin appearance. Hair loss also occurs with a moth eaten appearance to the coat and eventually thickening of the skin with a rancid or musty odor. If the disease is severe, total hair loss can occur with secondary infections and purulent (green-yellow) discharge, itchiness, inflammation, and pain. Diagnosis is obtained through histological examination of a skin punch biopsy. There is an SA database, and dogs are categorized at the time of biopsy as either clear (no evidence of SA), subclinically affected (appears normal but has some microscopic destruction of sebaceous glands), and affected (has SA).

Treatment for SA depends on the severity of the disease and the patient's response to various therapies. Typically, affected dogs are recommended to be bathed weekly or every other week with an anti-seborrheic shampoo (removes dead hair and scales), be on a fatty acid supplement for the skin, and may include applying a lubricant to the skin in more severe cases.

The last disease to be discussed is von Willebrand's disease (vWD), a loss of von Willebrand's factor, a blood clotting component, which can lead to bleeding disorders. vWD is seen in the standard and miniature varieties of poodle, with the standard being the most common. It is also present in many other breeds, particularly of Germanic origin, and is present in humans as well. The loss or decrease of von Willebrand's factor results in a loss of clotting ability and leads to prolonged bleeding times. Affected dogs will sometimes display nosebleeds, lack of clotting when injured, and increased bleeding during surgical procedures. It is often not recommended to spay, neuter or perform other elective procedures on severely affected dogs only because they may hemorrhage during surgery. In general, it is healthier to spay and neuter non-breeding animals but not in cases of vWD affected patients. If adequate blood products are available for transfusion, surgery can be attempted in emergency situations.

There are two forms of inheritance for vWD. The most common form is type I vWD which has been proposed to be inherited as a trait with incomplete dominance. With incomplete dominance a puppy will inherit the defect if either parent carries the gene, but siblings who have inherited the gene may be affected to different extents depending upon the amount of expression of the gene. It is somewhat like having a dimmer switch on a light source. The same switch can be turned up for bright light or down for low light, with medium light in between the extremes. For vWD, several puppies in a litter may inherit the gene, but depending upon the amount the gene is "turned on" or expressed, the affectedness can range from mild to moderate to severe within one litter. It is important to note that affected animals only need to inherit one copy of the defective gene from either parent for the disease to be expressed. Also, puppies which inherit one copy of the defective gene may not be clinically affected if the gene is only mildly expressed. Puppies that inherit two copies of the defective gene usually die at birth or in the neonatal period.

There is a genetic DNA test available for vWD through VetGen as well as a blood chemistry assay which can determine the percentage of von Willebrand's factor present in the patient's blood at the time of sampling. The DNA test identifies individuals as either clear (do not carry the defective gene), carriers (carry one copy of the defective gene but appear normal), and affected (carry the defective gene and express the disease). In general, dogs with vWD can live a normal life span but may have mild to moderate bleeding episodes, especially with superficial injuries such as lacerations or toenail bleeding. Severe bleeding episodes or traumatic injury will require veterinary care and may result in a blood transfusion.

Overall, the poodle is no more affected by genetic problems than any other purebred dog, and has fewer problems than some. The important point to remember is that breeders should use as many valid genetic tests as are available on their breeding animals and pet owners should see or obtain copies of the tests performed when purchasing a puppy. Pedigree analysis is also useful for those diseases which do not have a genetic test available. On a personal note, I have tried to be as thorough as possible in listing as many of the genetic disorders in poodles that have published results. If I have overlooked current information or have not mentioned a disease that anyone feels I should have mentioned, please contact me at the previously mentioned e-mail address. I hope this article is useful to poodle breeders and pet owners alike.