



Canine Genetic Disorders

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The genetics of dogs is a complex and fascinating field. While selective breeding has led to the diverse range of breeds we see today, it has also predisposed certain breeds to specific genetic diseases. Genetic diseases in dogs are caused by changes or mutations in their DNA, and can lead to a wide range of health problems. These diseases can affect various organs, tissues, and systems within a dog's body, and can be inherited from their parents. Due to the complex genetic factors involved, it can be difficult to identify and manage genetic diseases in dogs, making genetic testing an important tool for responsible breeding and pet ownership.

Causes of Genetic Disease in Dogs

Genetic diseases in dogs are conditions caused by genetic predispositions and result from mutations within an individual dog's DNA. These diseases affect the health and lifespan of the canine population. Some genetic diseases are inherited, meaning one parent passes down a defective gene to offspring, while others are acquired, meaning the dog acquires the mutation after birth.

Some of the common disorders of dog are as follows:

Cancer: A familial, or breed related incidence of cancer is being investigated in several breeds. These include the common disorders of lymphoma, osteosarcoma, hemangiosarcoma, melanoma, and mast cell cancer. Osteosarcoma is most prevalent in Great Danes, Saint Bernards, Doberman Pinchers, and Labradors. Skin and soft-tissue cancers are most prevalent in Saint Bernards, Bassett Hounds, German Shepherds, Golden Retrievers, English Setters, Great Danes, Pointers, and Flat-Coated Retrievers. Breast tumors are most prevalent in Pointers, Poodles, Cocker Spaniels, German Shorthaired Pointers, and Boston Terriers. Melanomas are most prevalent in Scottish Terriers, German Shorthaired Pointers, Cocker Spaniels, Pointers, Golden Retrievers, and Boxers.

Progressive Retinal Atrophy: There are several inherited PRAs identified in dogs. The most common is an autosomal recessive, late-onset progressive rod cone degeneration. The mutation causing this disease occurred long before the differentiation of many breeds, so it is shared across many breed lines. **Progressive Retinal Atrophy (PRA)** is an umbrella term for a group of genetic disorders that affect the retina, leading to a progressive loss of vision and ultimately blindness in dogs. This condition is inherited and is prevalent in several breeds, including but not limited to Cocker Spaniels, Labrador Retrievers, Poodles, and many others.

Hereditary Epilepsy occurs in many breeds, and represents a diverse group of recurring seizure conditions. There are no tests available to diagnose hereditary epilepsy. When diagnosing epilepsy, other non-hereditary seizure disorders must be ruled out. The onset of hereditary epilepsy can be neonatal, juvenile, or adult, although most dogs have their first seizure sometime after their first birthday. While most hereditary epilepsies cause recurring seizure episodes throughout life, some can cause only one or two seizures, and never occur again.

Hip Dysplasia: Hip dysplasia is a condition that primarily affects the hip joint, which is a ball-and-socket joint. In a healthy joint, the ball at the top of the thigh bone (femur) fits snugly into the socket in the pelvis, allowing for smooth movement. This disorder of malformation and hip joint laxity occurs across all breeds. The breeds with the highest frequency are; Bulldog (73.6%), Pug (61.7%), Otterhound (50.6%), Neopolitan Mastiff (48.5%), and St. Bernard (46.7%). Breeders must use breadth and depth of pedigree normalcy to select against this disorder.

Brachycephalic Syndrome: Brachycephalic breeds are popular for their distinctive appearance, characterized by broad, short skulls, flat faces, and short noses. This facial structure, while appealing to many,

predisposes these breeds to a range of health issues collectively referred to as brachycephalic syndrome. Bulldogs, Pugs, Boston Terriers, French Bulldogs, and Shih Tzus are among the breeds most commonly affected.

Bladder Stones: Bladder or urine stones are concentrated, crystallized minerals that build up in a dog's urine. The amino acid cystine is a common culprit in developing bladder stones. Bladder stones can be extremely painful and impair the dog's urinary function. While they can affect any dog, there are certain dog breeds that tend to display genetic markers predisposing them to the development of bladder stones, including Dalmatians, Newfoundlands, Bichon Frise, Miniature Schnauzers, Labrador Retrievers, Australian Cattle Dogs, Miniature Pinschers, Mastiffs, American Pit Bull Terriers, English Bulldogs, French Bulldogs, Chihuahuas, Rottweilers, Dachshunds, and Scottish Terriers, among others.

Hypothyroidism: Hypothyroidism is caused by autoimmune thyroiditis; an inherited autoimmune disorder where the thyroid gland is destroyed by autoantibodies. In order to diagnose the disease, you have to identify the autoantibodies. A thyroid profile is a snapshot of a moving picture of the thyroid health of a dog. An affected dog will begin to produce thyroid autoantibodies usually between 1 and 3 years of age. The breeds with the highest percentages are; English Setter (33.5%), Polish Lowland Sheepdog (30.7%), Havanese (25.6%), Old English Sheepdog (22.8%), and Boxer (19.7%). For mixed breed dogs, 11.5% of 49,126 dogs tested positive for thyroid autoantibodies.

Congenital Heart Anomalies: Several breeds of dogs and cats have hereditary congenital heart anomalies. These include patent ductus arteriosus (PDA), aortic stenosis, ventricular septal defect, and ventricular stenosis. Problems with managing these disorders include missed diagnoses on subclinically affected animals, and not utilizing breadth of pedigree in counseling breeders. If a breeder is concerned about carrying genes for the disorder, all related animals should be screened by Doppler echocardiography. This includes both pet and breeding siblings.

Atopic/Allergic Skin Disease: The heritability of atopic disease in Labrador and Golden retrievers is estimated at 47%, which is higher than many polygenically inherited disorders, including hip dysplasia. The breeds with the highest incidence of atopic skin disease are; West Highland White Terrier, Cairn Terrier, English Setter, Irish Setter, and Dalmatian.

Patella Luxation: This disorder is much more common in the small stature breeds. However, as many of these small dogs do not develop significant arthritis and discomfort from the condition, many breeders do not track the disorder or forward the results of patella evaluations to the OFA. The OFA patella database reports an average of 5.55% of submitted dogs with patella luxation. The breeds with the highest incidence are Pomeranian (47.9%), Chow Chow (29.5%), and Cocker Spaniel (27.2%).

Elbow Dysplasia: This disorder is classically defined as one of three disorders; ununited anconeal process, fractured coronoid process, or osteochondritis dessicans of the elbow joint. More recent research indicates that elbow dysplasia may actually be a disorder of uncoordinated growth of the radius and ulna. When the radius grows longer than the ulna allows, it causes elbow joint incongruity.

Gastric Dilatation/Volvulus (Bloat): Bloat occurs primarily in the large and giant breeds. Dr. Larry Glickman at Purdue University conducted an epidemiological survey, and found that the Great Dane has the highest average lifetime risk of a bloat episode of 42.4%. Other breeds at higher-than average risk include the Bloodhound, Irish Wolfhound, Irish Setter, Akita, standard Poodle, German Shepherd Dog, and Boxer.

von Willebrand's disease (vWD): Autosomal recessive vWD is the most common canine hereditary bleeding disorder, and has been reported in over 50 different breeds of dogs. Blood assays for vWD factor shows that the disorder is most prevalent in the Corgi, Doberman Pinscher, German Shepherd Dog, German Shorthaired Pointer, Golden Retriever, Shetland Sheepdog, and Standard Poodle.

Drug Sensitivity/Ivermectin Sensitivity: The defect causing ivermectin sensitivity in Collies and other breeds has been identified as a mutation in the MDR1 or multi-drug resistance gene. This defective gene can also cause neurotoxicity from loperamide, vincristine, and other drugs, through alterations in the blood brain barrier. A genetic test is available, and the following are results of testing in several breeds (%homozygous/%heterozygous): Collie (32%/46%), Australian Shepherd (2%/30%), Old English Sheepdog (1%/9%), Shetland Sheepdog (2%/17%), Longhaired Whippet (16%/52%), English Shepherd (<1%/14%).

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