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## Animal Specialty Hospital of Florida

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### FAQs about Breed Predispositions for Acquired Cardiac Disease and Congenital Cardiac Defects

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#### *What is a genetic predisposition?*

A genetic predisposition means that an individual may have a tendency to develop certain diseases as a consequence of inherited defects in the genes. A *gene* is a segment of a chromosome, which is the portion of a cell that contains the instructions to produce certain substances, commonly proteins. These proteins have a job to do, and abnormal proteins may be unable to perform their job correctly. This can ultimately lead to acquired cardiac (heart) disease.

#### *What is a breed predisposition?*

Since breeds are created by allowing closely-related individuals to procreate, a genetic predisposition for heart disease may develop. While certain traits and characteristics of the breed may be desirable, the tendency toward developing heart disease is not. Conscientious breeders do their best to eliminate individuals known to have heart disease from the breeding pool, however many breeds are well-known for their association with heart disease. Ongoing research is being performed in order to try to identify specific gene defects associated with acquired heart disease in certain breeds, and may someday allow us to offer genetic testing to help us identify young animals likely to develop heart disease later in life or breeding animals likely to have offspring at risk for heart disease. This may ultimately allow us to screen these individuals out from the breeding population.

#### *Are certain breeds more likely to have a predisposition to develop heart disease?*

Absolutely. A number of breeds we see commonly develop chronic mitral valvular disease (CMVDz, myxomatous valvular degeneration/MVD, mitral “endocardiosis”- all equivalent terms). **Cavalier King Charles Spaniels (CKCS)** are particularly afflicted, and are commonly used as models for studies of CMVDz. Polygenic modes of inheritance are suspected in CKCS and Dachshunds. Poodles, Papillion, Maltese and Chihuahuas are all commonly reported to develop CMVDz, but really *any* breed can be affected. In general, small breeds tend to get CMVDz, and large to giant breeds get dilated cardiomyopathy, but this is a generalization, and overlap can occur.

The high prevalence of dilated cardiomyopathy (**DCM**) in Doberman Pinschers, Boxer dogs, American Cocker Spaniels, Newfoundlands, Irish Wolfhounds, Portuguese Water Dogs (PWDs), Mastiffs, and Great Danes suggest a genetic basis for the disease, arguably autosomal dominant or recessive. An X-linked autosomal dominant pattern has been identified in Great Danes, and an autosomal recessive pattern has been identified in PWDs. Recently, a specific gene mutation (autosomal dominant) has been identified in Boxers with cardiomyopathy. The trendy term right now for Boxer cardiomyopathy is **ARVC** (arrhythmogenic right ventricular cardiomyopathy), but it is arguable

whether or not Boxers have the same exact disease that people have with ARVC. This is especially the case since the particular gene mutant identified in Boxers is not a mutation associated with the disease in people. However, many different defects may ultimately result in phenotypic dilated cardiomyopathy. Other commonly affected breeds with DCM include Labrador Retrievers, Golden Retrievers, Saint Bernards, Springer Spaniels, English Sheepdogs, Afghan Hounds, Scottish Deerhounds, terriers, and English Cocker Spaniels, with males being more likely to develop severe disease in Dobermans, and conflicting reports of sex predilection or lack thereof in other breeds.

Most commonly, hypertrophic cardiomyopathy (**HCM**) is reported in domestic short hair (DSH) cats, but strong evidence exists for a familial inheritance in Maine Coon cats, American Shorthairs, as well as Persians. Males are greatly overrepresented, and the nonobstructive form is most common. A particular gene defect in protein myosin C has been identified in the colony of Maine Coons at Davis. HUNDREDS of different genetic defects have been associated with humans with HCM, so we're pretty sure more than one defect in cats may be at work.

### **What is a congenital heart defect?**

A congenital heart defect means that a problem occurred with the development of the heart when an individual was growing inside the womb. These defects commonly result from a mutation in one or more genes, and are often heritable. This means they can be passed from one or both of the parents to the puppy or kitten. These defects are usually detectable at birth or a very young age.

### **Are certain breeds more likely to have congenital heart defects?**

Absolutely. Different breeds may be predisposed to developing different cardiac defects. Generally, the most common congenital defects in dogs include patent ductus arteriosus and subaortic stenosis. The most common congenital cardiac defects in cats tend to be septal defects and valvular dysplasias.

Patent ductus arteriosus (**PDA**): Commonly reported breeds that may be predisposed to PDA include Bichon Frise, Chihuahuas, Cocker Spaniels, Collies, English Springer Spaniels, **German Shepherds**, Keeshonds, **Labrador Retrievers**, **Maltese**, **Pomeranians**, **Poodles**, Shetland Sheepdogs, Welsh Corgis, and Yorkshire Terriers. **Newfoundlands** and Cavalier King Charles Spaniels may also be predisposed. Depending on the geographic region, PDA may be the #1 or #2 most commonly reported congenital cardiac defect in dogs.

Subaortic stenosis (**SAS**): Reported breed predilections include Newfoundland, **Boxers**, Rottweilers, **Golden Retrievers**, Bouvier des Flandres, and **German Shepherds**. The trait has been characterized with an autosomal dominant pattern with gene modifiers or polygenic pattern in Newfoundlands. Bull Terriers often have concurrent mitral valve abnormalities. Other cardiac defects have been associated with SAS and include mitral valvular dysplasia, PDA and aortic arch abnormalities. Studies in Newfoundlands have documented the development of progressive lesions in the early postnatal period, suggesting that "congenital SAS," at least in Newfoundlands, is technically a misnomer. Depending on the geographic location, SAS may be the #1 or #2 most commonly reported congenital cardiac defect in dogs.

Ventricular septal defects (**VSDs**) are familial in English Springer Spaniels, and an autosomal dominant pattern with incomplete penetrance or a polygenic trait is suggested. Probably the 3<sup>rd</sup> or 4<sup>th</sup> most commonly reported congenital cardiac defect in dogs.

Pulmonic stenosis (**PS**): Reported breed predilections include Beagles, **Keeshonds**, **English Bulldogs**, Mastiffs, Samoyed, Miniature Schnauzers, Cocker Spaniels, West Highland White terriers, Chihuahuas, **Newfoundlands**, Bassets and Chows. Risks may be higher for male English Bulldogs and Bull Mastiffs. Probably 3<sup>rd</sup> or 4<sup>th</sup> most commonly reported congenital cardiac defect in dogs.

Tricuspid valvular dysplasia (**TVD**) is a congenital malformation of the tricuspid apparatus. The defect is common in Labrador Retrievers, Old English Sheepdogs, Great Danes, German Shepherds, and Irish Setters. NOT UNCOMMON.

Mitral valvular dysplasia (**MVD**) is a congenital malformation of the mitral valve apparatus and is common in cats, Great Danes, German Shepherds, and Afghan Hounds. In Bull Terriers mitral dysplasia is **X-linked** (carried by females) and the defect is typically seen in males. RARE.

Atrial septal defect (**ASD**) is common in Boxers, Doberman Pinschers, old English Sheepdogs and Samoyeds. Standard Poodles may also be predisposed. RARE.

Tetralogy of Fallot (**TofF**): (0.25 per 1000 dogs), usually associated with other defects including tracheal hypoplasia, peritoneal-pericardial diaphragmatic hernia (PPDH), pectus excavatum, PDA, atrial septal defect (ASD) or persistent right aortic arch (PRAA), and is **autosomal recessive** in Keeshonds. RARE.

**Do mongrel dogs/cats (mutts) develop acquired heart disease or have heart defects?**

Absolutely. Acquired heart disease may develop in any dog or cat, and mixed breed animals may deliver puppies or kittens with congenital heart defects. Overall, the *risk* for developing heart disease or having heart defects is probably less in mixed breed animals. Generally, if an individual dog or cat is known to have offspring with congenital heart defects or have developed acquired cardiac disease, they should not be allowed to breed in the future.