

A HEART-TO-HEART CHAT ON

SUBVALVULAR AORTIC STENOSIS

(SAS) IN BERNESE MOUNTAIN DOGS

BY NANCY P MELONE, PHD

It was a beautiful day. Sitting on her patio, she admired the dogs in her back yard as they took turns “owning” a new toy she had just purchased. At that moment, her cell phone rang, breaking the peaceful quiet of the morning. The name of one of her favorite puppy owners appeared across the phone’s display screen. No doubt this would be another call about the latest adventures this wonderful pup had with his owner.

She answered the call. Immediately, she detected that this was not going to be the conversation she had expected. The puppy owner sobbed, “My beautiful puppy has been diagnosed with subvalvular aortic stenosis. They call it SAS.”

The breeder was stunned and, for a moment, speechless. This pup was out of her dream breeding—a gorgeous Top 20 stud dog and her knockout beautiful, Bronze Grand Champion bitch. Both the sire and dam had cleared all of their required health CHIC certifications, including congenital cardiac tests. How could this be? She had never heard of SAS being a problem in Bernese Mountain Dogs.

Following her conversation with the puppy’s owner, the breeder quietly asked around to see if other breeders had encountered this disease. She discovered that while not frequent in the breed, it did happen and had been seen in litters of some lines, including ones she had used. She reflected on the risks of using so-called “popular sires” in any breed and set about learning more about SAS and ways that she might lower the risk of it in her breeding program.

WHILE THIS VIGNETTE is fictional, the story it tells reflects a situation not unlike what a small number of health-conscious breeders and puppy owners have recently experienced. It is a cautionary tale for both breeders and puppy buyers.

Subvalvular Aortic Stenosis (SAS) in Bernese

Although not typically found in Bernese Mountain Dogs, SAS has been diagnosed recently in a small number of puppies/dogs in a few lines. For this reason, it is important that breeders and dog owners become familiar

with the symptoms so that we can monitor the disease to see if SAS is restricted to just these few lines or if it is a more pervasive breed-wide problem that should be addressed more formally.

According to the Berner-Garde database, the first Berner diagnosed anecdotally with SAS was in 1982. The first Berner definitively diagnosed occurred in 1989. Neither bitch appears to be closely related. If we look at the period from 1982 to 2021 (almost 40 years), we see that 88 dogs have been diagnosed with SAS (as of 9/12/2021). Objectively, 88 dogs in 40 years is not a large number over a significant sample of dogs, but certainly worth monitoring.

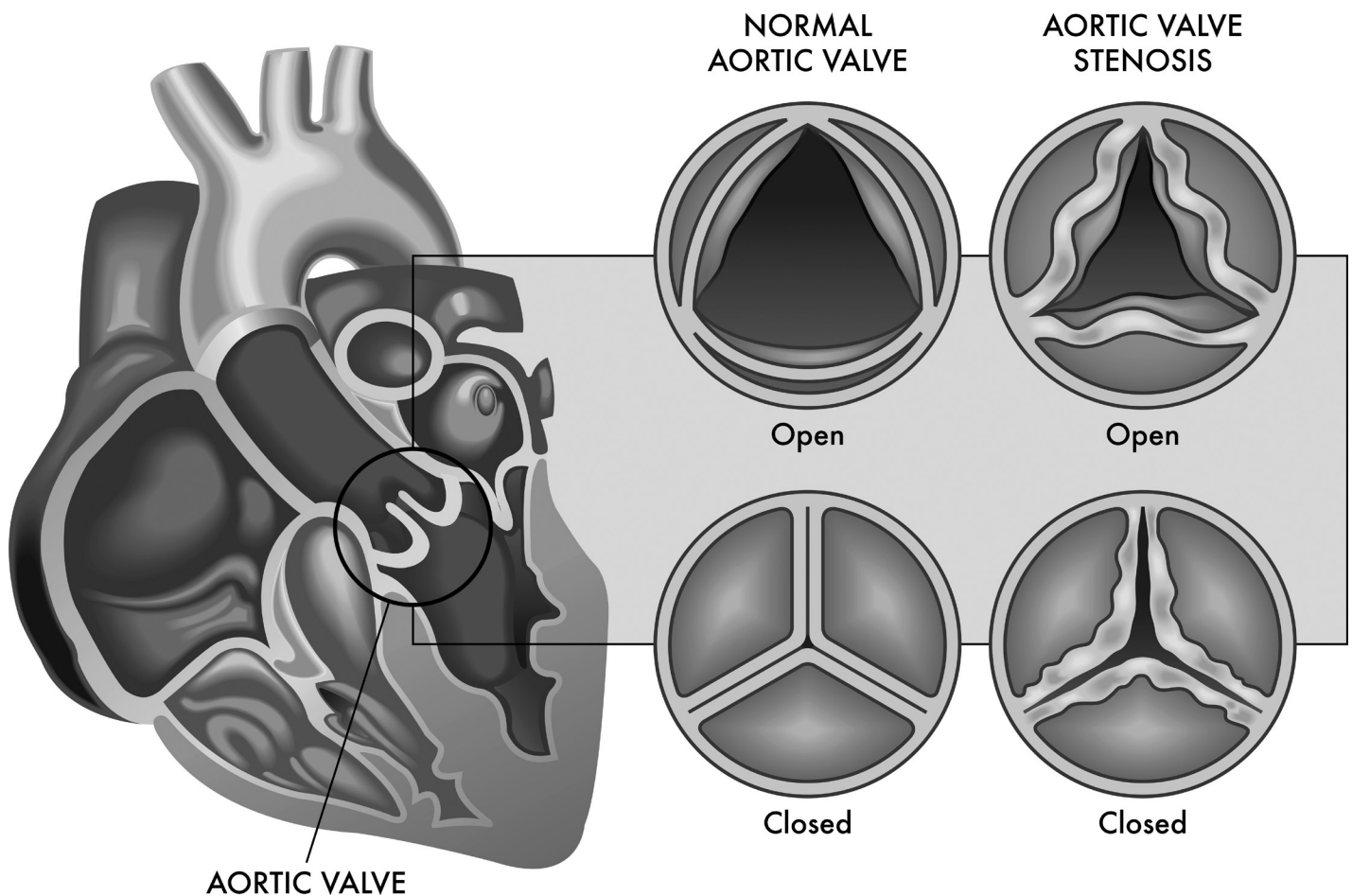


FIGURE 1: Normal and Stenotic Heart Valves

What is Subvalvular Aortic Stenosis (SAS)?

Subvalvular aortic stenosis is a congenital (i.e., present at birth) heart disease in which blood flow from the heart is abnormally reduced or constricted. SAS is typically a disease of large-breed dogs. Breeds in which SAS is commonly found include Newfoundlands, Boxers, Dogues de Bordeaux, Golden Retrievers, German Shepherd Dogs, Bullmastiffs, Bull Terriers, Samoyeds, and Rottweilers. Males seem to be affected slightly more than females. Research in Newfoundlands supports a genetic basis for the disease. This is also suspected to be the case in other breeds, although the mode of inheritance is not clear.

SAS is characterized by a lesion that obstructs or blocks normal blood flow. Most often, there is an abnormal

fibrous ring of tissue in the left ventricle just below the aortic valve, hence the term “subvalvular” (see Figure 1). The lesion typically develops during the first 4 to 8 weeks of a puppy’s life.

The stenosis (or narrowing) can be mild, moderate, or severe. Mild SAS is of no consequence to the dog’s quality of life or lifespan. Treatment is not necessary. A mildly-affected dog tends to live normally, but because of a suspected genetic link with SAS, cardiologists recommend that the dog not be bred.

To the extent the narrowing is considered moderate or severe, the cardiac muscles must work much harder to force blood out through the valve, which in turn abnormally increases the heart’s oxygen needs. The overworked heart muscle thickens (called hypertrophy), resulting in the heart not filling properly. The thicker

heart muscle is also prone to electrical instability, which can interfere with the conduction of electrical signals that trigger the heart to beat. This condition can cause an abnormal heart rhythm and sudden death. These dogs are prone to endocarditis (heart infections) and many must take antibiotics prior to any dental or surgical procedure.

SAS is a progressive disease. As the dog reaches adulthood, the disease can worsen and require treatment. Severely-affected dogs left untreated have an average lifespan of approximately 19 months. This can be extended with treatment to 55 months. Severe SAS may lead to congestive heart failure (CHF) or sudden death. Because of the suspected genetic link, cardiologists strongly advise that these dogs not be used for breeding.

What are the Symptoms of SAS?

Ironically, dogs with SAS may show no obvious signs of disease. SAS is often first detected during a routine veterinary examination when the veterinarian hears a heart murmur using a stethoscope (auscultation). There are two types of murmurs. Systolic murmurs are the most common and occur as the heart contracts. Diastolic murmurs are much less common and occur when the heart relaxes.

Murmurs are graded on a sliding scale from 1 to 6, where 1 is a murmur that can be difficult to hear with a stethoscope in a quiet room and 6 is a murmur so loud it can be heard without the assistance of a stethoscope. Typically grades 1 and 2 are considered mild and do not require treatment. Grades 3 and 4 are considered moderate murmurs, with the dog often showing some clinical signs (e.g., shortness of breath). If the murmur is a grade 3 or above, additional testing may be warranted. Murmurs graded 5 or 6 are considered severe and typically require medical management or surgery.

In puppies with mild cases, the murmur may not be noticed until a puppy is between 6 to 12 months old. In some cases, murmurs in puppies can be “innocent murmurs.” These are murmurs that the puppy eventually outgrows. In these cases, there is a little local turbulence in blood flow, but the heart is able to manage it well. In moderate to severe cases, there may be evidence present at birth. With more severe murmurs, there is the risk

of serious heart damage to the puppy. In ambiguous cases it may be necessary to scan the heart to determine if the murmur is innocent or more serious.

In dogs with moderate to severe SAS, signs of heart dysfunction related to the heart’s failure to circulate blood efficiently may be present. Common symptoms include lethargy, weakness, exercise intolerance, shortness of breath (dyspnea), and collapse or fainting (syncope).

While congestive heart failure (CHF) is typically more common in dogs that have other heart valve issues, it may also be observed in dogs suffering from SAS alone. CHF involves a buildup of fluid in the lungs. Symptoms are consistent with this buildup and include coughing, increased difficulty breathing, reluctance to walk, blue tinge to normally pink gums, racing heart rate, poor appetite, swollen belly, and open-mouthed breathing while at rest.

How is SAS Diagnosed?

A veterinarian may suspect SAS based on a dog’s presenting symptoms, history, and physical examination. Unfortunately, dogs with mild or moderate disease are often asymptomatic. In contrast, dogs with severe SAS may exhibit signs of weakness, exercise intolerance, and potentially sudden death. Often a veterinarian suspecting SAS will find a systolic murmur, a weak femoral (inner thigh) pulse, or an irregular heartbeat. Since SAS has the potential to worsen in severity until a puppy is full-grown, murmurs can become more severe over the first 1.5 years of life.

The current gold standard for definitively diagnosing SAS is an echocardiographic study (i.e., a cardiac ultrasound). This provides a visualization of the ring of tissue below the aortic valve that impedes normal blood flow. Several other abnormalities (e.g., chamber size, heart wall thickness) may be identified as well. During examination, an image of the heart is displayed on a monitor in real time. A doppler study is also part of the echocardiographic examination, measuring the direction and flow of the blood as it moves through the heart. Specifically, it reveals blood flow turbulence and velocity across the ring of tissue blocking the flow. The velocity of the systolic blood flow determines the severity of the stenosis (narrowing). Blood pressure differences across the stenosis indicate how hard the heart is working to move blood through the

narrowing. This pressure difference is called a pressure gradient (PG) and is correlated with survival times. PG differences of 40 mm of Hg are considered mild, whereas a difference of 80 mm of HG is considered severe.

Chest x-rays and electrocardiograms (ECG) can also be useful in providing additional diagnostic information. Electrocardiograms reveal the pattern of electrical activity in the heart, including an irregular heartbeat. Chest x-rays are useful in distinguishing if SAS (or some other disease) is the underlying cause for labored breathing. X-rays also can reveal a fluid buildup in the lungs, seen in congestive heart failure. In a few cases, a 24-hour Holter monitor (a vest worn by the dog for a short duration) may be used to assess the frequency and severity of an irregular heartbeat and to determine if anti-arrhythmia medication should be prescribed.

Is There a Cure or Treatment for SAS?

There is no cure for SAS. Most often the disease is medically managed. Beta-blockers (e.g., atenolol or sotalol) are the current treatments of choice. These drugs reduce symptoms and slow the heart, decreasing the heart's demand for oxygen and consequently, reducing stress on the heart. The jury is still out on the long-term benefits of using beta-blockers as a single treatment; nevertheless, most cardiologists recommend them for dogs with severe SAS. Depending on the diagnosis, veterinarians may also prescribe diuretics, ACE (angiotensin-converting enzyme) inhibitors, inodilators, a reduced sodium/increased protein diet, and/or a modified exercise protocol. CoQ10 as a nutritional supplement has been suggested as beneficial, but there is no published research on its effectiveness.

There is no completely satisfactory corrective surgery for SAS. Surgical treatments have been attempted, although with varied success. There have been attempts to surgically remove the ring of tissue causing stenosis. Unfortunately, the procedure does not improve overall survival times. Standard balloon dilation has also been attempted on dogs with severe SAS, but improvement is only temporary and does not significantly extend survival time in comparison to dogs treated with only beta blockers (e.g., atenolol). A newer minimally-invasive technique using a cutting balloon followed by standard balloon valvuloplasty has been developed for treatment

of severe cases of SAS. Initial reports indicate that this procedure results in a significant decrease in the pressure gradient and helps reduce symptoms of SAS, but again regrowth of the ring of tissue occurs. It is not yet known if this procedure improves survival times. Because survival times for dogs who have undergone surgeries (compared to medical management) are not improved, invasive procedures are usually not recommended.

How Can BMD Breeders Avoid Breeding SAS-affected Puppies?

There is little if any research on SAS in Bernese Mountain Dogs. This is likely because Bernese Mountain Dogs have not been considered a breed in which the disease is frequent, and genetic research typically requires a substantial number of affected dogs to be productive.

Experts believe that control of SAS in any breed will be challenging without a genetic test. No breed currently has such a test, although researchers are working on one for Newfoundlands with varied success. Currently, we have only phenotypic tests (e.g., echocardiograms) to guide us in breeding against SAS. While quite useful, these tests are not as effective as a genetic test would be in efforts to eliminate disease. An analogy to the situation with SAS is hip dysplasia. While very modest progress has been made with phenotypic testing (e.g., hip x-rays), it is less than we would like.

Concerned breeders trying to manage SAS would obviously like to know how SAS is inherited. There have been several efforts to identify modes of inheritance for SAS. In the landmark Pyle and Patterson study of Newfoundlands involving breeding various combinations of affected, affected cross, normal, normal cross, normal non-Newfoundland, etc., the researchers concluded that SAS was indeed genetic in Newfoundlands. Breeding SAS-affected dogs resulted in transmission of the disease to offspring. Unfortunately, establishing the precise mode of inheritance was much less clear. It was hypothesized in that study that the mode of inheritance (based on affected status of offspring at skeletal maturity) was either polygenic or dominant. In contrast, in another study of 230,000 Newfoundlands the mode of inheritance was thought to be autosomal co-dominant. In a more recent study of 45 dogs, the mode of inheri-

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tance was hypothesized to be autosomal dominant or polygenic. Obviously, despite considerable research, the mode of inheritance for SAS in Newfoundlands has not been precisely determined. It could be that the discrepancies across these more recent studies are due to limited data or issues encountered in classifying normal and abnormal dogs. On the positive side, researchers have identified one genetic variant (PICALM) in North American Newfoundlands that has been linked to SAS, and research is underway to determine if this could be a useful test for US dogs. Unfortunately, PICALM was not linked to SAS in European Newfoundlands.

Evaluating the pedigrees of several other breeds (Bullmastiffs, Golden Retrievers, and Rottweilers) suggests an autosomal recessive mode of inheritance. A similar evaluation of the Dogue de Bordeaux breed also suggests an autosomal recessive pattern of inheritance. There have also been reports of possible incomplete penetrance further complicating the issue. So, the bottom line is that mode of inheritance for SAS is still an open question and may vary across breeds.

Given no genetic test to guide breeders and an unknown mode of inheritance, what might a Bernese Mountain Dog breeder do to lower the risk of producing an SAS puppy?

- Do not breed any dog diagnosed with SAS.
- For “popular sires,” obtain Advanced Cardiac Certification after the sire has produced 5 litters and before the 6th. This protects the breed should he be found to have SAS.
- Perform an echocardiogram if you:
 - have a puppy who has a heart murmur after 1.5 years old or
 - suspect SAS in a puppy or dog.
- For breeding pairs that have produced an SAS puppy, obtain an echocardiogram for both the sire/dam before breeding either dog again.
 - For each puppy produced by the pair, perform Basic Cardiac Certification Examination (at 12-24 months)

done by a cardiologist.

- Before breeding a puppy from a litter containing an SAS sibling, obtain an echocardiogram on that puppy.

How Can Puppy Buyers Minimize the Risk of Buying an SAS Puppy?

Education is the best offensive weapon in your effort to purchase a healthy puppy. Familiarize yourself with the various diseases affecting the breed. Finding a quality breeder is your next weapon. Ask your prospective breeder for copies of health certifications of the sire and dam of the puppy that you are purchasing. With reference specifically to SAS, ask if the breeder does basic and advanced cardiac testing of their breeding stock. Ask if they have SAS in their lines. Ask how many litters the sire of your prospective puppy has produced to date. If the sire has produced more than 5 litters, ask if he has had an echocardiogram and been cleared for SAS.

Once you have your new puppy, stay in close contact with your breeder and report any significant health issues that your puppy experiences. Record significant health events in the Berner-Garde database. By remaining in touch with your breeder, you will know about the puppy’s parents as they age and about the health status of your puppy’s siblings.

And So the Story Goes...

Science is an evolving endeavor. If we did not realize this previously, the COVID pandemic has offered a poignant example of the ebb and flow of scientific knowledge, understanding, and progress. The path of science is often slow and torturous. Our knowledge of disease is iterative, expanding, and at times subject to refinement.

Our understanding of SAS in dogs, and specifically in Bernese Mountain Dogs, is a good example of this dynamism. What is critically important in our efforts to inform the Bernese Mountain Dog community is that we do not claim to know more than we know and that we do not cause unnecessary panic.

Care has been taken in this article to differentiate between what is “suspected” from what is “known” about SAS; this includes its presence in our breed. Clearly, SAS is genetically complex and there is much that we do not know, including its true frequency in the population. Other breeds in which SAS is commonly diagnosed have studied SAS for many years. They are just beginning to understand its genetics in their specific breeds.

Nevertheless, to better understand both the frequency of SAS in Bernese and its etiology, fanciers of the breed need to begin tracking instances of the disease by collecting basic cardiac data and documenting these data in the Berner-Garde database, including (importantly) those dogs anecdotally and/or definitively diagnosed with SAS.

While the breed has seen what appears to be a recent uptick in SAS cases in a few lines, there is not enough data to conclude that the problem goes beyond these few lines. Nevertheless, to better understand both the frequency of SAS in Bernese and its etiology, fanciers of the breed need to begin tracking instances of the disease by collecting basic cardiac data and documenting these data in the Berner-Garde database, including (importantly) those dogs anecdotally and/or definitively diagnosed with SAS.

Ideally, breeders and puppy owners should perform inexpensive Basic Cardiac Certification testing on their puppies. In cases where dogs or puppies exhibit SAS symptoms or where dogs are being used for breeding,

it is prudent to perform Advanced Cardiac Certification testing on them. Recording these results in the Berner-Garde database lays the groundwork to get a better handle on modes of inheritance and disease frequencies and, if needed, for any future research.

We can all play a part in the evolving science needed to re-write the story that began this article.

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