



## Identifying Genetic Markers for a Congenital Heart Disorder in Dogs Kathryn M. Meurs, DVM, PhD, North Carolina State University

### RESULTS: Search Continues for the Genetic Mutations for SAS in At-risk Breeds

Subvalvular aortic stenosis (SAS) is the most common congenital heart disorder in dogs, particularly Rottweilers and Golden Retrievers. SAS is a narrowing of the blood outflow tract from the lower left ventricle (chamber) of the heart. As a result, the heart has to contract with greater force to move blood through the small opening. The presence or absence of signs depends on the degree of narrowing; mildly affected dogs may have no signs and live a normal life span but severely affected dogs may exhibit exercise intolerance, endocarditis (infection of the heart valve), congestive heart failure and sudden death. The average life span of severely affected dogs is just 19 months.

Researchers at the North Carolina State University wanted to see if they could find a genetic mutation that was associated with the development of SAS in Rottweilers and Golden Retrievers. They searched the DNA of Rottweilers and Golden Retrievers using specially designed, custom-made DNA sequencing chips that allowed them to look for mutations in a shared genetic region of interest. Using this screening tool to analyze banked DNA samples, they compared DNA from SAS-affected Golden Retrievers and Rottweilers to DNA from dogs without the disease.

Unfortunately, final data analysis did not pinpoint a common causative mutation for SAS in Rottweilers and Golden Retrievers within the specific genetic region studied. However, the research team was able to rule out many genes not associated with this heart disorder in these two breeds and is continuing to search in areas not yet examined.

Identification of causative mutations in breeds at risk for SAS will help researchers develop genetic screening tests to reduce disease prevalence. By identifying affected animals, breeders and veterinarians will have a tool to help gradually eliminate this fatal heart defect from these breeds. (D13CA-071)