

Swiss Researchers Partner with Westie Foundation to Find Genetic Cause of CMO

Mutation Present in 100% of Study's Diagnosed Cases in West Highland White, Cairn and Scottish Terriers

By Teresa Barnes, VP Communications

The Westie Foundation of America (WFA), the University of Bern Switzerland's Department of Veterinary Biosciences and Research Programs Unit, Molecular Medicine, and the University of Helsinki and Folkhalsan Research Center, Finland announced today the discovery of the genetic mutation that causes Craniomandibular Osteopathy (CMO), a debilitating head and jaw bone condition that occurs in West Highland White, Cairn and Scottish Terrier breeds of dog.

Scientists reported a mutation on chromosome 5 caused the condition in 100 percent of the CMO affected dogs in the study.

Veterinary Scientists mapped a causative mutation to a small region of the genome on dog chromosome 5 via a genome-wide association study using DNA from 10 CMO-affected dogs and 41 unaffected or control dogs of West Highland White Terriers or Westies. Further experimentation confirmed the same mutation or CMO associated chromosome 5 haplotype was present in all CMO cases in Cairn, Scottish and West Highland White Terriers. The majority of the dogs held two copies of the mutation while a smaller number had at least one copy.

"This is really big milestone for the WFA and Westie breeders. We are grateful to our donors who have supported our research efforts for hanging in there with us and supporting our choices of research projects to fund and with which to provide biologic samples," said Bebe Pinter, President of the Westie Foundation of America. "Our efforts in CMO have definitely paid off."

With this discovery, we can now work with owners and breeders to prevent the

condition from being inherited in next generations of these dogs."

Dogs suffering from the disease often experience severe pain and lose the ability to open their mouths, resulting in some cases death from malnutrition or from euthanasia due to the condition.

CMO occurs when the angular processes of the mandible and tympanic bullae become so thickened they cannot be opened. The disease is seldom recognized until signs of discomfort due to chewing or eating are noticeable, typically between the ages of four and seven months of age in the dogs. More importantly from a breeder's standpoint, animals may be affected without showing significant clinical signs. This makes the DNA screening test that much more important to clear the problem in the breed.

"We particularly encourage owners of clinically affected dogs to submit samples," said Cord Drogenmuller, Professor at the Institute for Genetics, University of Berne, Switzerland. "This information will help us to evaluate the penetrance level of the mutation, to possibly refine the risk prediction in the future, if necessary, and to start to investigate possible additional cause(s) of CMO in the three terrier breeds."

Blood samples from affected dogs that are submitted with the x-rays, will be tested free of charge, according to the scientist.

"We are very pleased with the findings of this study and believe it will have a rapid impact on the health and viability of the Westie breed and the other terrier breeds," said Dr. Kay McGuire, a veterinarian and member of board of directors of

the Westie Foundation of America who assisted with the study.

Because of the diagnostic value of the identified mutation, a direct DNA test for CMO will be launched in Switzerland on December 1, 2012 at a cost of just over \$100 U.S. dollars. Full details are available at http://www.genetics.unibe.ch/content/service/dog/index_eng.html. The test will also be available soon in the U.S. Please contact the Westie Foundation of America for more information at (888) 928-3843, (281) 326-3843 or health@westiefoundation.org.

This is wonderful news! :)



"Our rescue westie, Hallie, was a participant in the CMO study and The Westie Foundation included her story and photo in their article on the study back in 2010. So glad this has worked and holds much promise! Hopefully soon no other dog will suffer from this awful disease."

Jackie Myers

