

## Spinal muscular atrophy in Maine Coon Cats (SMA)\*

The genetic cause of spinal muscular atrophy (SMA) in Maine coon cats (MC) was determined in May 2005, culminating a cooperative, 8-year effort of breeders, veterinarians, and researchers at Michigan State University, University of California at San Diego, the National Center for Biotechnology Information, and the National Cancer Institute. A convenient and reliable DNA based carrier test is now available.

SMA is a disorder caused by death of spinal cord neurons that activate skeletal muscles of the trunk and limbs. Loss of neurons in the first few months of life leads to muscle weakness and atrophy that first becomes apparent at 3-4 months of age. Affected kittens develop an odd gait with a sway of the hindquarters and stand with the hocks nearly touching. They may also stand with toes out in the front. By 5-6 months of age they are too weak in the hindquarters to readily jump up on furniture and often have a clumsy landing when jumping down. The long hair MC may hide it, but careful feeling of the limbs will reveal reduced muscle mass. Affected kittens are not in pain, they eat and play avidly, they are not incontinent, and most live very comfortably as indoor cats for many years. As of this writing, the oldest affected cats of which we know are 8-9 years of age. Known affected kittens have occurred in breeding programs across the United States, and, in retrospect, likely carriers have been exported widely.

Studies have demonstrated that the disorder is inherited as a simple autosomal recessive trait. For a kitten to have the SMA disorder, it must receive the mutated copy (allele) of the disease gene from **both** parents, and male and female kittens are equally affected. The parents of affected kittens show no outward signs of disease, but they are obligate carriers, by definition. In a breeding program, both male and female carriers will pass on their mutant alleles to 50% of all their offspring, on average. When two carriers are inadvertently mated, on average 25% of the kittens will have SMA. That means that in litters from such matings, there may be some combination of SMA and normal kittens, all SMA kittens, or all normal kittens. Unidentified carriers in breeding programs continue to spread the mutant gene throughout the MC breed.

SMA in MC is caused by large deletion on cat chromosome A1 that removed 2 genes. Identification of the SMA mutation has allowed design of a laboratory test to detect the deletion in DNA from cat cheek cells, blood, or frozen semen. DNA isolated from samples is subjected to a polymerase chain reaction (PCR) to amplify the portion of the genome harboring the deletion. Normal and deleted alleles are differentiated by the size of the amplification products separated by agarose gel electrophoresis. Carrier cats can be positively identified in the laboratory on the basis of both the mutant and normal allele PCR products amplifying from their DNA. If one thinks a kitten may be affected, the test can also be used for definitive diagnostic confirmation.

The SMA test is offered to MC breeders, owners, and veterinarians through the Laboratory of Comparative Medical Genetics at Michigan State University in hopes that it will be used to stop the spread of SMA. Testing results are kept strictly confidential and are returned only to the person submitting the cat's sample. Test results for each cat will be returned as a printed certificate by US post and also by email if desired.

The cost of the test is \$50/cat. To obtain cheek brushes and instructions for sample collection, please contact the laboratory by email ([fyfe@cvm.msu.edu](mailto:fyfe@cvm.msu.edu)) indicating how many cats you wish to test and providing a mailing address. For additional information, please contact Dr. John C. Fyfe by email or by mail at Laboratory of Comparative Medical Genetics, 2209 Biomedical Physical Sciences, Michigan State University, East Lansing, MI 48824

\*Article obtained from the Maine Coon Breeders and Fanciers Association web site. To visit the MCBFA web site, go here:

<http://www.mcbfa.org/index.html>