Cheek Swab Collection and DNA Extraction

Mark Neff, PhD, Veterinary Genetics Laboratory, University of California - Davis
Invited by Eddie Dziuk, Delegate, National Beagle Club

Recently, the Broad Institute of MIT/Harvard has presented the canine genetics community with a whole genome sequence of the dog. This is an unprecedented advance in the field – the genome sequence will enable researchers to transition from family-based studies that identify the determinants of simple traits, to more powerful population-based approaches that can elucidate the genetic components of multi-factorial traits (i.e., complex diseases).

Mapping genes with populations requires two key resources. First, digitized whole-breed genealogies must be assembled to optimize selection of animals for inclusion in the study (this allows researchers to calibrate the size and intensity of the genome signal that they are searching for). The AKC has recently made electronic pedigree data accessible to academic researchers to facilitate this step. Second, DNA must be collected from a large number of animals (typically, greater than 200 dogs) for both the case (affected) and control (unaffected) groups that make up a population-based project. Cheek swabs currently represent the best means for achieving the necessary degree of breeder participation.

Cheek swabs strike an important balance – these samples provide ample DNA of sufficient quality for the most common laboratory method of DNA fingerprinting. At the same time, the ease and low cost of collecting swabs dramatically lower the barriers to breeder participation (relative to blood collections). One or more swabs can be obtained from a dog in less than five minutes without the need for a visit to a veterinary clinic. The swabs can then be shipped in an envelope by regular post to the laboratory, where they can be stored at room temperature indefinitely. Ongoing studies indicate that swabs can provide DNA for at least eight years, so long as ambient humidity is not too high.

The DNA extracted from a single swab is sufficient for running 200 polymerase chain reactions (PCRs), with as many as eight genetic markers per reaction. This marker density is at the threshold needed for population-based mapping; multiple swabs can be collected per dog to ensure that enough genetic material has been collected (roughly 2% of swabs fail to provide DNA, so additional swabs are always recommended). Cheek swab DNA can be arrayed in conventional laboratory plates and allowed to dessicate, which facilitates high-throughput DNA analysis.

There are new technologies on the horizon for which cheek swab DNA may not be a suitable substrate (namely, detecting single base differences using high density DNA chips). Most genetic research, however, emphasizes the most recent generations of dogs, primarily so that accurate and standardized methods of phenotyping (i.e., diagnostics) can be applied during sample ascertainment. Older generations of dogs add power to studies, but this is usually to augment or confirm a finding. If emergent technologies turn out to require DNA derived from blood samples, these samples can still be readily collected from extant dogs. Cheek swab samples from older, possibly deceased dogs will still retain their value and provide the needed data for strengthening or confirming genetic associations.