UPDATE
Fanconi Syndrome

By Lisa Auerbach

On September 12, 2011, Dr. Gary Johnson publicly announced the identification in his laboratory of a genetic mutation that causes Fanconi Syndrome.

At the same time, he announced the availability of a new DNA direct test for the disease. The new direct DNA test, which is done through OFA (as was the old linked marker test), had already started to be used on August 29, 2011, after Dr. Johnson became confident the genetic mutation had been identified.

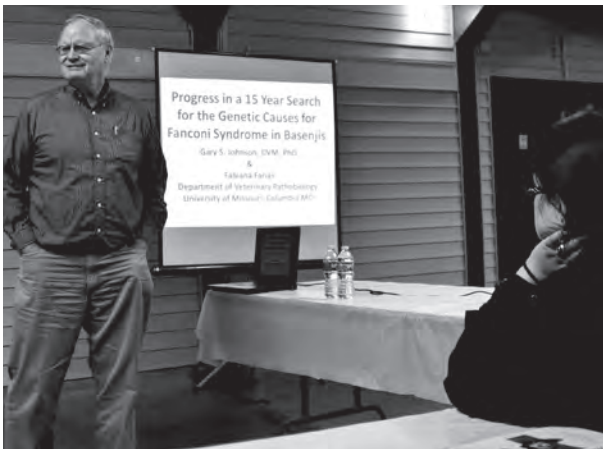
What will change? The new test tests for the mutation itself, not for markers linked to the mutation. Thus, the terminology associated with results will change. The linked marker results were given as probably clear/normal, probably carrier, probably affected, and indeterminate. The new test results will be clear/normal, carrier, or affected. The word "probably" will no longer appear, and there will no longer be an indeterminate category.

Dr. Johnson is recommending that all breeding stock be retested with the new DNA direct test. The new direct test can now be ordered from OFA, and the old test is no longer available. The direct test will cost $65 for a new test, with the option, starting November 1, of a price of $50 for retests. For retests, the Basenji Health Endowment will pay the other $15.

In reporting his research laboratory's work to the BCOA, Dr. Johnson emphasized the work done by Ms. Fabiana Farias, who will receive her doctorate this winter with her work on Fanconi Syndrome as an integral part of her PhD thesis. Dr. Johnson also made a point of noting the contributions of Jon Curby, who he described as having "played an essential role in all aspects of the research."

At the Nationals, Dr. Johnson reviewed the history of his work with Fanconi. His first Fanconi grant was from the AKC Canine Health Foundation, with matching funds from the Basenji Club of America and the Basenji Health Endowment. Subsequent work was supported by the Basenji Health Endowment and the AKC Canine Health Foundation. The last part of the research was funded by the University of Missouri with a $50,000 Mizzou Advantage Grant.

Samples from affected dogs and their relatives were used to help identify the approximate location of the mutation, by using marker assays. In 2007, these assays yielded results indicating a location on chromosome 3. Dr. Johnson was asked by BCOA and BHE if he could make available an interim test for Basenji breeders for the 2007 breeding season. At that time, Dr. Johnson, through the OFA offered a linked marker test as an interim aid to Basenji breeders when planning litters to try and avoid producing Fanconi Syndrome affected puppies.



Dr. Gary Johnson at the 2011 BCOA National Specialty. Photo by George Woodard

The next step was a whole genome sequence from an affected Basenji, to compare to the known Boxer genome. This led to the identification of a 370 base pair deletion in a gene on canine chromosome 3 that Dr. Johnson and his group believe is the gene associated with Basenji Fanconi Syndrome. Dr. Johnson believes Fanconi Syndrome is the first canine inherited disease whose gene has been identified with whole genome sequencing.

The Fanconi story is not yet finished – Dr. Johnson is interested in studying how the mutation causes disease, in hopes of providing treatments or preventative measures that might delay onset or lessen severity of disease.

If you'd like more details on the research, Dr. Johnson's presentation at Nationals is online at www.basenji.org/ClubDocs/Johnson_2011_Presentation.pdf