Benign Familial Juvenile Epilepsy (BFJE) in Lagotto Romagnolo

Benign Familial Juvenile Epilepsy was first recognized and reported in the Lagotto Romagnolo as early as 2002. The disease is described as inherited benign juvenile epilepsy. A similar disease is described in human medicine. It is an autosomal recessive gene that causes the disorder. The affected puppies usually show evidence of seizures and ataxia from 5 to 9 weeks of age and these resolve spontaneously by 8 to 13 weeks of age. Research at the University of Helsinki identified the causative gene in 2007, showing an autosomal recessive mode of inheritance.

Genetic testing for BFJE is available and it is recommended that all breeding stock be tested prior to breeding. Testing is available from Optigen in USA, Laboklin in Germany or Genoscoper in Finland.

Many countries have submitted samples and now, thousands of Lagotto have been tested for the BFJE gene. The data shows the following results of that testing: 3% affected; 46% carriers; 51% normal.

To better explain what these results mean, please see below for definitions:

- **NORMAL**: The dog carries two copies of the normal gene and therefore has no predisposition to epilepsy.
- **CARRIER**: The dog carries one mutant and one normal copy of the epilepsy gene. The dog does not develop the disease but can transfer a gene defect to approximately 50% of its offspring.
- **AFFECTED**: The dog carries two mutant copies of the epilepsy gene and will likely develop the disease in early puppyhood. If bred from the dog will transfer the gene defect to its entire offspring.

Carrier dogs should only be mated to clear dogs so as to avoid producing BFJE affected puppies. It is **NOT** recommended to withdraw carrier dogs from breeding, as this would greatly reduce the already small Lagotto gene pool and not necessary when mating to a clear or normal dog. This concept and practice is consistent with the other Lagotto Romagnolo clubs and breeders worldwide, and follows the guidelines of genetics research and data.

For more information, please visit the open access article on PLOS Genetics Website: [http://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1002194](http://journals.plos.org/plosgenetics/article?id=10.1371/journal.pgen.1002194)