

## Research on Berger Picard PRA du Berger Picard : CNRS April 19<sup>th</sup> 2016

Currently, the Biobank Cani-DNA contains at the Rennes laboratory 68 blood samples from Bergers Picards (26 males and 42 females) with clinical and genealogical data for most of the samples. Of these 68 dogs, 35 had a dated ophthalmological examination, 31 exams on dogs less than 5 years old and 3 others on dogs between 5 and 8 years old. Ocular examinations are fundamental to carry out the project. Indeed, in order to obtain optimal clinical information, dogs should be examined clinically by an ophthalmologist at least once, ideally several times in their life, given the PRA's late declaration and its evolution, depending on dogs. The genetic study will be effective only with eye examinations carried out on as many dogs as possible, preferably after 4 or 5 years. Of the 68 dogs whose DNA is entered in Cani-DNA, 3 are affected (3 females with 11 years, 8 years and 1.5 years) and 2 are suspect (1 female and 1 male of 4 years). In addition, a female is affected by multifocal retinopathy at 1 year. Currently, only two disease free dogs over the age of five at the time of diagnosis could serve as controls for genetic studies. On the other hand, 9 Picards belonging to the pedigree could serve as controls if they are examined in 2016 and if they are still alive (age > 10 years). A first family tree including 154 Picards was constructed and includes all affected and suspect cases of PRA. Detailed analysis of the pedigree makes it possible to make the hypothesis that the Picard APR is transmitted in a monogenic (a single cause: a single mutated gene), autosomal (on a non-sexual chromosome - chromosome 1 to 38) and recessive (the 2 copies of the gene must be mutated for that the dog to be affected) mode. This pedigree should be supplemented by new dogs whose blood can be taken and the eye examinations carried out; other affected dogs and disease free dogs linked to the pedigree will be able to participate in the judicious choice of new dogs to sequence, to optimize the chances of finding the cause of this PRA in the Picard and especially to validate it on a larger number of dogs.

The aim of this research is to develop a genetic diagnostic and screening test for veterinarians and breeders and to propose new candidate genes for human retinopathies.

### Actions to be taken

It is important to continue collecting blood samples from affected dogs and their parents, but also from old dogs that are clinically free, ie with a CEO at an advanced age (> 9 years), to serve as controls in the Genetic analyzes.

Partner associations will try to identify owners of affected dogs or siblings of affected dogs and their parents to encourage eye examinations and blood sampling. Oral specimens are possible and sometimes easier to perform but are not always the best because the amount and quality of DNA extracted are not optimal for whole genome sequencing. Frozen sperm is also an interesting source of DNA, especially for champion dogs that have largely reproduced and if eye examinations and clinical information are available from these dogs.

It is highly desirable that the blood samples be accompanied by additional clinical information (CEO, result of dysplasia), and any other indications, with photos that could benefit from genetic analyzes (eg long hair, lack of doodle, brittle nails etc.)....).

For more information, see our website : <http://dog-genetics.genouest.org>

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