

## Health Report – Sept 2015

Thanks to the participation by many Picard fanciers around the world, as of Sept 18<sup>th</sup> there are 275 samples from 267 individual Picards DNA banked for any reason. About 2/3 of these are from the USA, and the remaining 1/3 from Europe & Canada. Most of these dogs were clinically normal, with no disease conditions reported at the time the sample was banked, although we do have some reported with various eye, heart, orthopedic, gastric, allergic, and neurologic problems. There is no one condition rampant or overly common in the breed, but with the research emphasis on PRA (progressive retinal atrophy) and CMR (canine multifocal retinopathy), there are a few dogs reported with these conditions. If a banked dog is diagnosed with ANY condition that may be heritable, please report this so the record can be updated – this is very important as research moves forward on many fronts.

Currently there are 9 Picards reported by their owners to be affected with PRA with DNA banked at U of MO. Most are onset of clinical signs and blindness by 2-4yrs of age, and 1 was not showing clinical signs until 8yrs of age. One dog had very damaged retinas at the first exam, so it was difficult to know precisely when and where the disease started on that individual. There are 4 dogs reported with CMR, but there may be others in the collection that have not been reported as CMR-affected. If you have a dog diagnosed with geographic retinal dysplasia or CMR, please make sure to report this.

Berger Picards are one of the few breeds to have multiple individuals that have been Whole Genome Sequenced. Thanks to the efforts and funding of many individuals and clubs, there is WGS data for one Picard diagnosed with CMR, one Picard diagnosed with early-onset (blind at 2yrs of age) PRA, and one Picard reported to be clinically normal. Just within the past 2 weeks, the research team at the University of Missouri College of Veterinary Medicine has new alignments and mutation reports for the Picard WGS's, and the research team has been studying these new reports for mutations of interest. There are mutations indicated in genes known to cause early and late onset PRAs, and another one that may be involved in CMR. These mutations are being analyzed, and all reported affected dogs are in the process of being tested to see if they have these mutations. It would be very helpful for the research team to have more affected individuals to include in this analysis, to help rule in or rule out these mutations as causative. If you own a dog diagnosed with either condition, please report this as soon as possible to the research team! If the dog has not been DNA banked yet, please send that sample right away.

In addition, the research team is doing what they call “reverse genetics” – usually researchers have a disease, and go in search of mutations that may be causing that disease. With the vast amount of info from the WGS's, they now can see many mutations in genes that have a known function reported in other species (usually human or laboratory mouse/rat), and are looking to see if these diseases appear in dogs (and in this case, specifically in Picards). In one or more of the Picard WGS's, there are mutations in genes known to cause the conditions listed below. The dogs that were sequenced are generally heterozygous (or carrier – one normal copy of the gene, one mutated copy of the gene) for these mutations, so they would not show the condition. However, there could be Picards that have 2 copies of the mutation and show the disease. It's also possible that these may be mutations that do not actually have an effect, so although there are many conditions listed here, this should not be cause for panic. However, if any Picard breeder or owner has seen any of these conditions in a Picard, the research team would very much appreciate knowing about it, and having DNA from the individual showing that condition.

Mutations have been found in WGS of Picards which may cause the following conditions:

- Abnormal brittle hair & nails
- Abnormal and/or discolored teeth
- ALS (non-SOD1 Degenerative Myelopathy)
- Anxiety
- Chronic pulmonary disease
- Congenital ichthyosis
- Congenital myasthenia (muscle wasting – 2 genes with mutations identified)
- Diabetes
- Dilated cardiomyopathy
- Epilepsy (2 genes with mutations identified)
- Erythrocytosis (abnormally high blood cell count)
- Extra toes, renal disease (unusual combination for one gene but this is what has been reported)
- Eye color (may determine light vs dark eyes)
- Hemophilia
- Hypertrophic cardiomyopathy
- Multiple developmental defects (possible cause of fading puppies?)
- Muscle weakness (3 genes with mutations identified)
- Neonatal diarrhea & jaundice (2 mutations identified)
- Paroxysmal dyskinesia (uncoordinated movement)
- Peripheral neuropathy (may not know where feet are, or tingling sensation in extremities)
- PRA (early onset, late onset, geographic retinopathy mutations identified)
- Sensory neuropathy (similar to peripheral neuropathy – lack sensation in feet, limbs)
- Stiff joints & skin nodules

Again, at this point the research team does not know if these conditions appear in Picards (other than the PRA & retinopathy mutations found – which may or may not be the right genes), or if the mutations identified are actually causing any disease. However, if there are Picards with any of these issues, please report this to the research team as soon as possible so they can be evaluated. This could potentially have a great impact on Picard health, so please spread word of this to other fanciers.

The U of MO research team can be reached by emailing Liz Hansen at [HansenL@missouri.edu](mailto:HansenL@missouri.edu), or call 573-884-3712. Thank you for your cooperation and support of the research in the past, and for the future!

**Liz Hansen**

Animal Molecular Genetics Laboratory

University of Missouri - College of Veterinary Medicine

Columbia, MO 65211

573-884-3712

[HansenL@missouri.edu](mailto:HansenL@missouri.edu)