

I have received a few pedigrees of beagles that are either affected with PRA (via genetic testing) (cone Rod dysptrophy 4) or carriers. While this condition has not been known to cause blindness in beagles so far, I would suggest that we do test for it and do our best to not breed affected to affected. This is not something to get hysterical over at all, but just another consideration when breeding. With the pedigrees I have seen there are probably a substantial number of carriers and affected already in the US bloodlines. While the test may show "affected" we may never see actual clinical expression. This is very much like the F7 test and may only be a laboratory finding vs clinical expression. A good thought is to CERF older dogs and again probably will not find any clinical expression.

If you have an affected beagle that has developed clinical expression of disease please contact me privately at beagleinfo@mchsi.com.

This is the information I received from Optigen in 2017

Following up on our conversation, please see the information below I received earlier today from a well informed veterinary ophthalmologist who has done extensive research on RPGRIP1 and its association with retinal disease and who assists in advising OptiGen on these matters. Please see her response on this matter below. If you should learn of any PRA-affected beagles (regardless of their RPGRIP1 status) both Dr. Miyadera and OptiGen would be interested in obtaining a sample from such dogs for inclusion in OptiGen's PRA research/free DNA testing program. If you have any questions, please feel free to contact me.

As chair of the health and genetics committee for Beagles, I hope you may help spread the word on OptiGen's PRA/Free DNA testing program and we can help solve one more mystery of an inherited disease in dog.

Best regards,

Sue PK

Susan Pearce-Kelling
President and Manager, OptiGen, LLC

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From Dr. Keiko Miyadera:

Beagles have the insertion mutation in RPGRIP1 at the same position as the Dachshunds have. Just that their mutation is 15bp longer than that of the Dachshunds - so Beagles have a 44bp insertion, rather than 29bp in the Dachshunds (Published in Molecular Vision 2009; <http://www.molvis.org/molvis/v15/a246/index.html>).

Molecular Vision: Phenotypic variation and genotype ...

www.molvis.org

Phenotypic variation and genotype-phenotype discordance in canine cone-rod dystrophy with an RPGRIP1 mutation . Keiko Miyadera, 1, 2 Kumiko Kato, 2 Jesús Aguirre ...

I suspect this mutation is not uncommon at least among the laboratory Beagles based on my screening in Japan and in the US.

Of note, the homozygous affecteds do not show PRA clinical signs. My colleague Dr. Kumiko Kato has been following up on these "affected" Beagles for almost 10 years repeating ERG. While these dogs initially showed reduced cone ERG, from what I hear, over the years, their ERGs are some how "recovering".

In short, indeed Beagles do have a variation of the RPGRIP1 insertion mutation. *The "affected" Beagles, although they may show some initial abnormality that is only detectable via ERG, they never seem to show signs of PRA clinically.*