

Dear Elin

Thank you for your email. Cathryn passed your query on to me because I have been working on this research. There is not yet a publication for this study, but a paper is currently in press for the clinical research, and a manuscript is in preparation for the genetic study and should be submitted for publication in the first quarter of next year.

I have attached our announcement for the DNA test for your information, with some details about the research that has been carried out. Four dogs were diagnosed with the disease at the Animal Health Trust, and there have been 8 other affected dogs reported to us by breeders. We have identified the mutation causing the disease, and found that there are carriers within both the contemporary UK and Finnish populations. This disease may not currently have a high prevalence, but could potentially increase in prevalence in the future. If it is indeed rare then it would be possible to prevent it from becoming a major problem in the breed in the future through DNA testing, without having a large effect on the diversity of the gene pool. It would be important to deal with this early to reduce the risk of the prevalence of the mutation increasing within the breed to a point where breeding it out could become more challenging.

To give us an indication if there are any carriers within the Norwegian Buhund population in Norway we could test a representative subset of dogs. I understand that the cataract research you are involved with has resulted in the collection of a large set of DNA samples from Norwegian Buhunds. If a considerable number of these dogs meet the requirements outlined below, we could genotype these dogs, if the Norwegian Buhund Club in Norway could help us to meet the costs incurred. We could do this two different ways:

Firstly, we could issue a summary report, in which the test results of individual dogs would not be provided, but we would be able to inform you of the number of carriers that are present within the set. If you are interested in proceeding with this option and can tell me how many dogs would be genotyped I will be able to let you know how much the genotyping would cost.

Secondly, we could offer a discounted price of £35 per dog (normal cost: £48) for DNA testing if you would like to test a large batch of dogs at once. In this situation we would give individual test results for each dog and a certificate, and would also be able to inform you of the number of carriers that are present within the set. Again for us to be able to draw any meaningful conclusion about whether or not the mutation is present within the Norwegian Buhund population in Norway the dogs included would need to meet the requirements below.

Sample requirements for estimating mutation prevalence:

- We would need to genotype at least 50 dogs, but ideally 100 or more.
- Dogs need to be currently of breeding age (1-7 years), and therefore representative of the contemporary population.
- Each dog cannot share its parents with any of the others within the set.

Thank you for your interest in our research and in our DNA test, please let me know if you have any further questions, would like more information, or are interested in proceeding with either of the options I have outlined in this email.

Kind regards,

Chris

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**From:** Elin Kristiansen [<mailto:> ]

**Sent:** 03 December 2017 11:51

**To:** Cathryn Mellersh  
**Cc:**  
**Subject:** Cerebellar Ataxia in the Norwegian Buhund, UK status

Dear Dr. Mellersh,

I am writing on behalf of the breed council of the Norwegian Buhund Club in Norway, where I as a small animal clinician is the health advisor. I am also conducting research on cataract in the breed, in collaboration with the veterinary college in Norway, NMBU.

It has come to our attention that there has been work done in the UK on Cerebellar Ataxia in the breed, and now released a DNA test for this.

As this is not a condition seen in the breed in Norway, and we have had no reports of either from any of the countries we see Norwegian Buhunds, we would be most interested to hear about your research on this condition, and the relevance for the breed in the UK.

I can't find anything published on this apart from the information on the AHT website, are you publishing your work in the near future?

As the national breed club responsible for maintaining a good health status and a diverse enough genpool in this endangered breed, we now get many concerned buhundowners contacting us with questions on how to approach this.

Look forward to hearing from you.

Best regards,

Elin Kristiansen



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