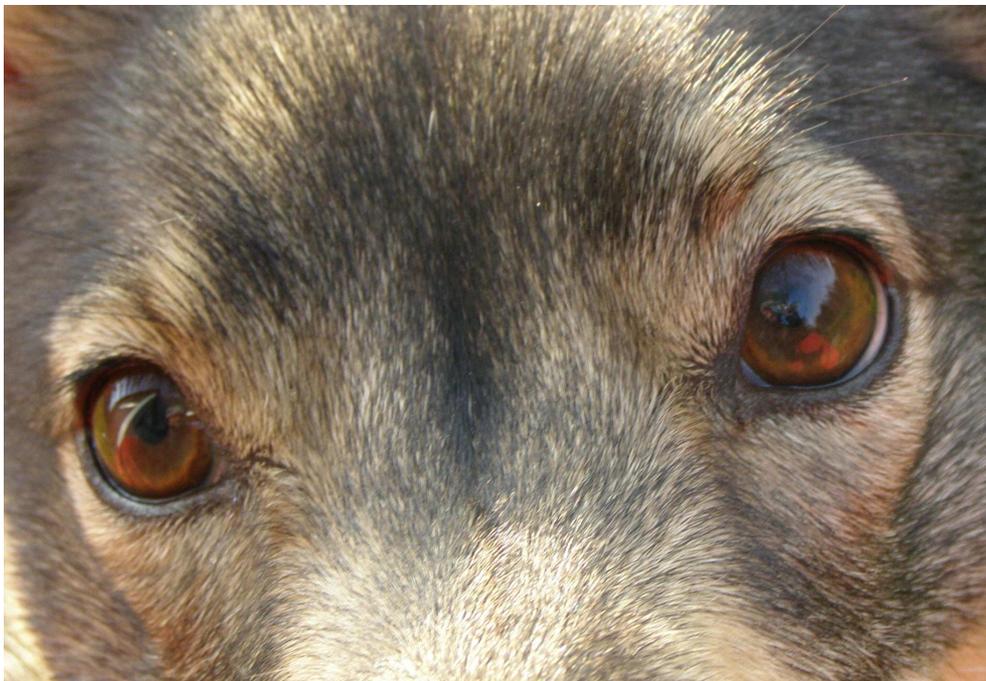


A study of retinopathy in Swedish Vallhunds



Jenny Pleym
2012

Foreword

This is a study of facts, as I interpret them, from available data about the Swedish Vallhund dog breed.

When I first heard about SV retinopathy I immediately thought *'How sad, but that doesn't concern me'*. I was about to breed my bitch. I had previously had her eyes examined, which were found to be clear. Instantly the thought occurred to me, *'What a luck that I had her examined earlier. Now with clear eyes I never need have her examined again, and risk a less favourable result.'* I think it is a normal reaction for someone to bury their head in the sand when it involves a beloved dog, and particularly if there are plans to breed that dog. It is a form of protection against the sorrow that would come with a negative diagnosis. I understand, because this was my reaction.

When I later gained a clearer idea regarding the inheritance of the disease and through curiosity I began to 'dig about' for information. I wanted to ascertain how the affected dogs were related and map out potential carriers. I was shocked as to the extent of the issue in the population! For several days I walked around and did not know how to process my discovery. It was at this time I decided to summarise what knowledge I had gleaned, and I decided that I wanted to share these understandings (along with my own conclusions) in a paper. So it is my hope that this paper is easy to understand as it is important that anyone may absorb and comprehend the content.

I have chosen to publish this study as I think knowledge and openness is the best way to solve a problem held in common. Surely nobody wants to buy, sell or breed dogs that may have a risk of reduced vision? Breeding puppies involves responsibility – for better and for worse. I am aware that I contribute to 'wear a hole' in the myth about the Swedish Vallhund being a particularly sound breed in publishing this paper. However, I am convinced that honesty is the best policy and that there is no point ignoring or hiding actual problems! For the Swedish Vallhund to continue to be a sound breed we need to face the truth, and take seriously matters such as hereditary disease, so that they do not have the potential to develop.

My conclusions are based on the theory that SV retinopathy is inherited in an autosomal recessive mode. I have chosen to interpret that all dogs with a diagnosis have a hereditary type regardless of what assessment the dog had about likely hereditariness. The estimation of which dogs are carriers is based on all information from the SKK Hunddata and KoiraNet where it refers to diagnosis of SV retinopathy that this is; a correct diagnosis; the dog has the disease SV retinopathy; and that degeneration in the dog's eyes has no other cause other than the inherited disease.

Jenny Pleym

Borås, the 31 july 2012

Index

Foreword	1
Index	2
Abstract	3
The disease SV retinopathy	3
Hereditariness of SV retinopathy	4
Why examine the eyes?	5
What measures can we take to defeat the disease?	6
Where does the disease come from? How has it been able to spread so much?	6
References	8
About the author	8
Appendixes (may be mailed on separate order)	9

Abstract

Retinopathy in Swedish Vallhunds, hereinafter referred to as SV retinopathy, is a disease of the eyes that may lead to reduced eyesight and in the worst case to blindness. The disease is hereditary. For a dog to get the disease both parents have to be either carriers of the gene or affected themselves. It can take many years before the disease shows up in a dog. Therefore when you go to breed an adult dog that has been examined and found clear, the disease may not show but manifest later in the dog's life. The only thing you can be sure of is, eye examined dogs which are diagnosed as affected with the SV retinopathy; their parents and all of their offspring are either carriers of the disease gene or affected with the disease.

A dog which is a carrier of the SV retinopathy gene has a single copy of the disease allele and will never develop the disease. It is not possible to find carriers through an eye examination because the disease will never manifest. However, half of the carrier's offspring will carry the disease gene, as they do. Until now it is only possible to find carriers by calculating their relatedness to found affected individuals. To find the affected dogs as many dogs as possible need to be eye examined. For every affected dog found there is on an average six relatives (parents and offspring) that are carriers of the disease.

In Sweden and Finland there is a total of 90 dogs that up to 2011 have been found through examination to be affected with SV retinopathy. When all of those 90 dogs' parents and offspring are counted together, there is a number of 528 more dogs, which are all carriers of the disease. Additionally, there are 355 full siblings to the 90 affected dogs. The probability for the siblings to be affected is that at least one fourth (89 dogs). Half of the siblings are carriers (177 dogs), and the last fourth of the siblings are free from the disease gene altogether (89 dogs).

Far from all dogs are eye examined. This means there are probably many more affected dogs among all Swedish Vallhunds in Sweden and Finland than we know of today. It can be stated that the disease is widely spread in the breed.

The disease SV retinopathy

At the beginning of 2000s in Finland it was discovered that Swedish Vallhunds could be affected by a form of Progressive Retinal Atrophy (P.R.A) which could lead to blindness. The Swedish breed club, the S.K.V, decided to conduct eye examinations on a number of dogs in Sweden also during 2001. About fifty dogs were investigated. At that time eye examination results were not official noted and recorded. The SKV paid a part of the cost of these exams so that they could gain access to the examination results. Those examination results have subsequently disappeared. Some of the examination results contained noted comments about the findings in the dog's eyes. Copies of the results (those which contained noted comments) were sent to veterinary and eye specialist Berith Håkansson Wallin. (Oxbergsblogg, 2007).

A few years later, around 2005-2006, the American eye specialist Andras Komaromy was contacted by a breeder in the USA who wondered about the frequency of noted comments in exam results of Swedish Vallhunds. Komaromy's professional interest extended to how various diseases in dogs are related to human diseases (Sandgren, 2011). Subsequent to the contact Komaromy studied SV retinopathy and has come to some conclusions on the matter.

SV retinopathy develops in three stages. At first red-brown pigment spots or grey mottles

appear in the retina. This doesn't affect the a dogs vision. This stage is most often seen in dogs at 2-3 years of age, but has been seen in dogs from 7 months of age up to 17 years. In the next stage, most often 2-3 years later, larger spots and mottling are seen and also a thinning of the retina. At this stage dogs can show signs of night-blindness. In the last stage degenerative areas expand and the dogs vision in daylight has now decreased and no night-vision is left. (SKV, 2011, Komaromy, 2007)

The development of the disease varies considerably between individuals and most dogs with changes in their eyes at least have some vision up to 10 years of age (Komaromy, 2007). Some dogs around the world have gone blind (WWSVA, 2011). In some dogs the disease stops at the first stage and never develop further (SKV, 2011).

During the years 1990-2011 271 swedish dogs were examined. Of these dogs 180 tested clear and 91 dogs gained some form of noted comment, (i.e. cataract or persistent pupillary membrane (PPM)). Then 31 swedish dogs gained the diagnosis of SV retinopathy. (SKK Avelsdata). In Finland 593 dogs have been examined of which 59 dogs were found to be affected with SV retinopathy (Koiranet). In total there are 90 dogs in Sweden and Finland with SV retinopathy (appendix 1).

Hereditariness of SV retinopathy

In Finland a pedigree of over 1,000 Swedish Vallhunds has been established. In this pedigree there are several affected dogs with non-affected parents. There are an equal number of dogs and bitches affected. This supports the theory that the disease is inherited in an autosomal recessive mode. (Sandgren, 2011). A recessive gene is subordinated to the other (dominant) gene. Only when the recessive genes occur in duplicate, i.e. have been inherited by both the mother and the father, can the disease manifest. Therefore, both the mother and the father of an affected dog have to be either affected themselves, or be carriers of the disease gene (figure 1). (Compare this to the gene for long tail, which is recessive, and for short tail, which is dominant). If a dog carries a single set of the disease gene, it will never be affected by SV retinopathy, However, it is a carrier and may pass the gene on to its offspring.

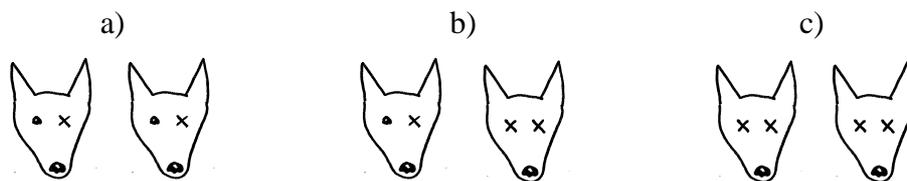


Figure 1. A dog affected by SV retinopathy inherits the disease genes from of it's both parents. Either both parents are carriers (a), or one parent is a carrier and the other is affected by the disease (b), or both parents are affected (c).

If a dog affected by SV retinopathy (having the disease gene in duplicate) and this dog is mated it passes on the disease gene to all of its offspring. Thus, all offspring are carriers of the disease gene. If two carriers of the disease gene are mated, statistically 25 % of the puppies will be affected by the disease, 50 % will be carriers (have the disease gene in single set) and 25 % will be free from the disease gene altogether. (figure 2).

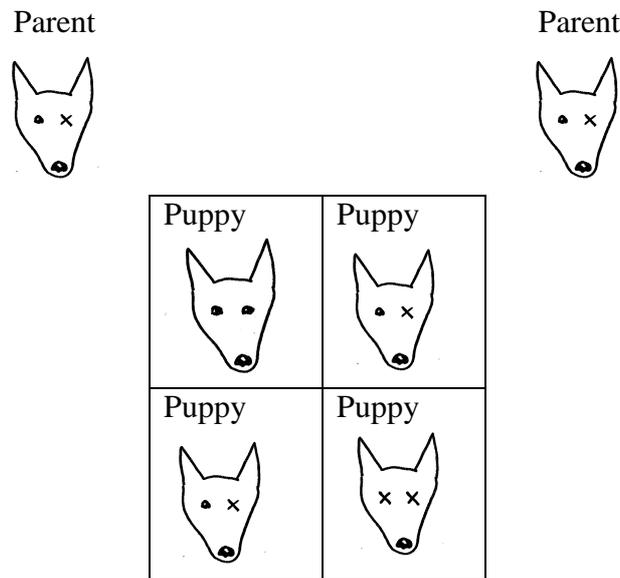


Figure 2. *If two carriers of the disease gene are mated 25 % of the puppies will be affected by the disease, 50 % will be carriers (have a single set of the gene) and 25 % will be free from the disease gene altogether.*

If a dog free from the disease gene is mated to a carrier (having a single set of the disease gene) statistically 50% of the puppies will be free from the disease gene and 50 % will be carriers. None of the puppies will be affected by SV retinopathy.

Why examine the eyes?

Why examine the eyes of dogs when there is no certainty that those affected by SV retinopathy will have the disease show up initially? After all it may take many years before the disease develops, and even then only few of the affected dogs get visual impairment and much fewer go blind! Most dogs gain some visual impairment in very old age anyway, so why does it matter? Why screen dogs for eye disease?

Eye examinations and pedigree studies of the 90 affected dogs has been **the only tool we have to date** for finding carriers and affected dogs among all dogs. When we have the **knowledge** about which dogs are carriers (appendix 9) it will be easier to avoid matings between individuals that we know carry the disease gene.

The very best tool to decide whether a dog is free from the disease gene, carrier of the gene or in the worst case affected by the disease, would of course be to take a simple blood test and do a DNA-test. There is no such test yet. It may come however as Professor Hannes Lohi, Finland, and his team at the University of Helsinki and Folkhälsan Research Center, are working on presenting a DNA-test for SV retinopathy (Koirageenit, 2012).

Because SV retinopathy can have such a slow progress it is especially important to examine the eyes of older dogs, particularly dogs those which have been bred. Every eye examination

is valuable for those that care about the health of the breed! **Thanks to the circumstance that SV retinopathy is inherited in an autosomal recessive mode it is fully possible to eradicate the disease totally without excluding a single dog from being bred!** Although, understandably dogs that are carriers or affected with the disease will have fewer potential partners as breeders would be searching for mates that are not affected or are carrying the disease gene.

What measures can we take to defeat the disease?

Health screen your dogs eyes

Undertake to have your dog's eyes examined! If it is a breeding dog, gain an eye exam before mating. Eye examinations should be done within a year of the matings, because in one year changes may have begun to occur in the eyes already. If you use the dog again a year since the last exam, examine again! Request that the owner of the proposed partner undertakes to gain an eye exam for their dog also. That is not an action of distrust – but an action of care for everybody! Avoid mating your dog to a partner that is not been eye examined within the last year. If you have a companion dog that is not being bred, undertake to gain an eye exam 2-3 times during the dog's life. As a suggestion examine the eyes at 3, 6 and > 8 years of age.

Breeding plans

If you know or suspect that your dog is affected by SV retinopathy or may be a carrier of the disease gene, choose a partner for your dog that likely is not a carrier of the gene. Do not choose another known carrier to help avoid the risk of producing puppies affected by SV retinopathy. Ultimately we may gain a DNA-test that can detect the disease gene. Then it will be easy to test all breeding dogs and all puppies in all litters. Then we will be able to plan the future breeding to decrease the disease gene in the population as much as possible.

Stay up to date

Keep yourself updated in new knowledge about SV retinopathy. Discuss with others. Bring up the subject at meetings and conferences.

Support the research

Contribute to the research about SV retinopathy by sending in blood samples and a copy of the dog's pedigree to the researchers. Blood samples from all Swedish Vallhunds have value, regardless if they are affected by the disease or not. Take the opportunity when visiting the veterinary or at eye examination to have this done. Just as well attach the eye protocol. For Information how to do it, enter the website <http://www.koirangeenit.fi/in-english/participate/>. There is a form and an address to send your sample(s) and how to do it. The blood samples are needed to make DNA-analyses. If you are able to, donate affected dogs eyes to the research when the dog passes away. It is the donated dog's eyes that have thus far contributed to the current knowledge of the disease and it's development.

Attitude

Be open minded. Do not blame. It is **nobody's fault** and no shame if a dog or breeding has been affected by SV retinopathy! The disease is newly discovered and thankfully often is mild, therefore it is not easy to be aware if earlier breeding dogs had visual impairment. Take responsibility. Now when we know that we can take action to limit the disease, apply it!

Where does the disease come from? How has it been able to spread so much?

Right from the beginning when the Swedish Vallhund were rescued from extinction during the 1940-ies, in-breeding and line-breeding was frequently used. This was of course necessary and relatively few dogs from that time are the foundation for all of today's Swedish Vallhunds. Furthermore, that was the way it used to be '*in the old days*'. In-breeding and line-breeding consolidated good quality and created even and type in the dogs.

The disadvantage with in-breeding and line-breeding is that not only are the good qualities consolidated but also the not good also. I believe that SV retinopathy unfortunately is one of the not good elements. I believe it has taken hold and spread, to a large extent, through the population literally due to the concentration of the disease through in-breeding and line-breeding. At some point a Swedish Vallhund of the past has gained the disease gene in it's genome, either because the gene existed in one of the foundation dogs from the 1940-ies, or the gene originated in a dog by mutation. Mutation is an alteration in the DNA that comes into existence through errors in copying of DNA strings. This may happen due to radiation exposure, toxins or viruses at cell proliferation. The fact that SV retinopathy is a unique form of P.R.A that has not been seen in any other breed, is an indication that the disease may have its origin more likely from a mutation.

I have studied the pedigrees of all dogs affected by SV retinopathy in Sweden and Finland but have not been able to come to a conclusion about where the gene originates from. There are a lot of dogs and a lot of blood lines involved. It indicates that the gene has existed in the breed for a very long time, maybe as far back as the 1940-ies. However, it is easy to see an obvious concentration of the gene from a few dogs that were bred prolifically.

When studying the finnish pedigree of more than 1,000 dogs, it is apparent that the lowest common denominator is the dog Ch Eko S44143/77 (Ahonen et al, 2008). All 90 dogs affected by SV retinopathy in Sweden and Finland and totally 520 known carriers of the gene, descend from six of Eko's offspring (appendixes 3-8). Most of them have Eko 6-8 times in their pedigrees about 7-8 generations back. Eko is one of the stud dogs that have had an enormous influence on the breed through his many offspring and many dogs from the following generation which were widely in breeding. Today, in Sweden only a few older dogs have a pedigree where Eko is not found, otherwise he is present in all and every swedish dog's pedigree.

It is probable that the gene for SV retinopathy could have been present in Ch Eko, and because he and his offspring were used so much in breeding (so-called matador breeding effect) the gene is widely spread in the population. In Finland, where the disease was first was discovered, there has been prolific breeding of Eko in the pedigree by necessity, due to the limited breeding options. Most imports from Sweden have been closely related to Eko. One example is the finnish imported Eko-son Västgöttagårdens Viking, who produced one pup affected by SV retinopathy, 5 grand-pups, 13 great-grand-pups and an additional 46 descendants affected by the disease. As Viking had at least one pup affected by SV retinopathy it can be stated that Viking himself was a carrier of the disease gene.

I have found eight carriers of the SV retinopathy gene that are **not** related to Eko. These are Västgöttagårdens Zita, Hildrek Platinum Lady, Hurstfield Kenelm James, Vallmyra Arrow, Bodatorp Danne, Grålötens Ackespett, Grålötens Alva Alvedon and Norsled Emil. These eight dogs all have offspring affected by SV retinopathy and therefore probably carry the gene

themselves. I have not been able to ascertain where the gene might have originated from in either their or Eko's pedigrees, because all Swedish Vallhunds pedigrees lead to the same dogs from the 1940-ies. However, some dogs are over represented in the pedigrees. The English dogs were highly inbred, as an example Norsled Emil has his ancestor Kling S07346/67 **twenty-four times** in his pedigree! It is obvious that the risk of inheriting deleterious genes increase significantly through such in-breeding.

My theory is open to be examined and questioned. No one would be happier than I should someone could prove that my theory is wrong! Or even better, prove that SV retinopathy doesn't exist at all and that I have got everything wrong! Just remember to separate me as a PERSON from my THEORY. If my theory feels terrifying and horrible it doesn't automatically means that I as a person is the same.

References

Ahonen, S., Seppälä, E.H., Vanhapelto, P., Cooper, A., Capaldo, F., Garmer, L., Sjö Dahl-Essén, T., Komaromy, A. and Lohi, H. 2008. *Clinical and genetical studies of progressive retinopathy of Swedish Vallhunds*. Västgötaspetsen 2/2008.

Koirangeenit. 2012. *Canine genetic studies*. University of Helsinki och Folkhälsan. <http://www.koirangeenit.fi/in-english/>

Komaromy, A. 2007. *Abnormal findings in the eyes of Swedish Vallhunds: An update*.

Oxbergsblogg. 2007. Ögon. <http://oxbergsblogg.blogggratis.se/2007/08/14/232058-ogon/>

Sandgren, M. 2011. *A summary of the minutes taken at The Swedish Vallhund World Congress 3-4 june 2011*. <http://svclub.org.nz/resources/SKV%20World%20Congress%20Report.pdf>

Specialklubben för västgötaspets (SKV). 2011. *Västgötaspets – minnesanteckningar under världskongressen 3-4 juni 2011*. http://www.skv76.se/kongr2011_sv.pdf

World wide swedish vallhund alliance (WWSVA), 2011. <http://www.wwsva.com/eye-diseases-in-dogs>

About the author

I, Jenny Pley, have had Swedish Vallhunds since 1990. The first dog, Pyret (1988-2004), was a rescue dog and had no pedigree. However, I have always been curious about pedigrees and lineage and as I had some clues I eventually found her mother, the unregistered Babsan. Babsan worked as a herding dog at a farm in Skåne. Pyret's father was reportedly Vikingatorpets Emil.

The next Swedish Vallhund was Höckgårdens Ida (born 2003), again a rescue dog. She is registered and has a pedigree. I have made a pedigree of all known dogs behind her back to Topsy (whelped, 1930) and all the way to Höckgårdens Ida. The pedigree contains 336 dogs. With that work I became very familiar with the Family Tree of Swedish Vallhunds.



I have a Master of Arts in biology and have studied at the University of Lund. Notwithstanding my studies, which have incorporated the study of genetics and zoophysiology, I consider myself as a happy amateur when it comes to science of heredity. Although I believe I am a little more prepared than the average dog owner. I have always been interested in genetics and veterinary questions.

Appendixes

As an active reader of Västgötaspetsen, blogs, facebook, forums and other sources of discussions about Swedish Vallhunds available on the internet, I have seen many different views of what is presentable to publish. It is extremely easy to be accused for '*hanging out to dry*' people and dogs, or '*search for faults*' in a sound breed and create sorrow, anxiety and dashing breeding plans for others. Many seem to suppose that the one who writes something **negative** about the breed is **evil** and wishes to destroy the good of the breed for puppy buyers, breeders and all others. Anyone that compile (more or less quality examined) facts in lists and spread them, immediately becomes a subject to accusations, threats and hatred. I think that is tiresome, because it makes an open and factual discussion impossible.

Due to this I have chosen not to automatically enclose the appendixes to this study! The appendixes accurately contain lists of affected dogs and known carriers and pedigrees of the relation between affected dogs. I want to emphasize: I love the Swedish Vallhund and do not wish the breed, any individual or any single person any harm! That is NOT the purpose of this study. I want to make this clear!

Anyone who wishes to be privy to the appendixes and my conclusions about the relationship between affected dogs are welcome to order a copy at jenny.pleym@gmail.com, and I will send it as a pdf file.