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The reliability of DNA tests for inherited diseases

Which of the internationally offered
DNA tests for inherited diseases are
useful for creating healthy dog
breeds in the Netherlands.



[Maaike Fennema \(3574717\)](#)

Prof. Dr. Jan Rothuizen Dr. Peter Leegwater
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Abstract

The change of the “Wet Dieren”, by the Dutch government, requires the breeders to screen their parental dogs for inherited diseases. Since the breeders and the veterinarians have to apply the DNA tests and they do not have all the knowledge about these tests, this study aims to make a list including all the available DNA tests on the Dutch market. To know if the offered tests are substantiated, they will be evaluated using a set of criteria developed during this project. First, all the DNA tests were gathered from the four biggest laboratories for the Dutch market, Laboklin, VetGen, Van Haeringen and Optigen. This resulted in a list containing 120 different diseases. In this article the focus was on cardiovascular and blood diseases, metabolic and immune problems and eye disorders. If the DNA tests fulfil the criteria they will be published on the website of ‘Expertisecentrum Genetica Gezelschapsdieren’. The most important criterion was that there was a peer reviewed article available about the mutation they tested on. So for all the DNA tests, articles were searched and reviewed. The research resulted in a list of in total 11 cardiovascular and blood diseases, 23 metabolic and immune problems and 27 eye disorders.

Introduction

Inherited diseases are the most common health and welfare problems in purebred animals in The Netherlands. The welfare of animals has become more important the last years since it has become a subject on the political agenda, as a result the 'Wet Dieren' (Lid 2.6 2c.) has been changed. Since the 1st of July 2014 breeders are required to do everything possible to produce healthy offspring. Due to this change, breeders are supposed to screen the parental dogs they use for breeding for inherited diseases. Inherited diseases are caused by a mutation or mutations in the DNA of the dog. DNA tests can show if these mutations are present in individual animals. If the breeders know if the parental dogs have a mutation for a particular disease it is possible to select a combination of parental animals which will produce offspring that is clinically healthy for the examined disease.

There are many DNA tests on the Dutch market, some of these tests are trustworthy but quite a few are not substantiated or even completely unsuited. For the veterinarian and for the breeder, who has to apply the test and has to convert the outcome of the DNA test into a proper breeding policy, it is difficult to know which tests they can use for a reliable outcome. By reviewing all the available tests in a scientifically sound manner it is possible to make a list of reliable tests, which can be used by veterinarians and breeders. The 'Expertisecentrum Genetica Gezelschapsdieren' of the faculty veterinary medicine will publish this list on their website so that the veterinarian and breeder can co-operate to breed purebred animals as healthy as possible. Another important application of DNA diagnostics for veterinarians, is that they can diagnose the genetic susceptibility of individuals in a population at young age, often before the onset of clinical disease. This permits the veterinarian to design an individual health program to prevent or decrease the clinical stage of the disease.

To evaluate if the DNA tests are useful, a list of criteria has been developed. To set up quality criteria for DNA tests it is necessary to understand which types of molecular genetic research exist and how they work. There are two main types of DNA tests. The first is based on a known mutation which causes the disease, which is the best and most used method to diagnose the mutation causing the inherited disease. The second type of test is a marker test which only marks the part of the chromosome where the unknown causal gene lies. This method can be used if the precise mutation is not yet known. It is a less certain method because the gene which is being tested on can no longer be linked to the mutation by recombination of the genes. Therefore the mutations test is preferred over the marker test. There are other factors which play a role in setting up the criteria. The published peer reviewed literature is an imported one. The articles about the mutations confirm that reliable research has been done and that the DNA test is based on valid evidence. It is also necessary to know for which breed the test is useful and how a mutation is inherited.

In this report all the DNA tests offered by the four biggest laboratories for the Netherlands, Laboklin, VetGen, Van Haeringen and Optigen, will be scientifically reviewed to determine the validity of these tests.

This research project aims (1) to develop criteria to review the quality and applicability of DNA tests available, and (2) to apply these criteria for the evaluation of the DNA test for dogs on the Dutch market. In this project the focus was on cardiovascular and blood diseases, metabolic and immune problems and eye disorders.

Material and methods

The project sets up a list of criteria for DNA tests on the Dutch market. A very important criterion which must comply with each test is the basis of a peer reviewed published article. When an article about a mutation is published it may be assumed that fundamental research is done and that methods and results have been accepted by peers in the research field. Tests based on such published results are considered reliable DNA tests. The articles should describe in which breed the mutation occurs and in which country the mutation is found. It is also useful to know how the disease is inherited and if the same disease is due to one mutation into different mutations in the same gene or even two mutations in different genes in different breeds. Because the list produced in this project is only about DNA tests useful in the Netherlands only the breeds that are bred in the Netherlands were included. The "Raad van Beheer" has a list of all dog breeds who are bred in the Netherlands. So this is also a criterion DNA tests in the definitive list must fulfil.

With the knowledge of the criteria the available literature in the PennGen database was used. In this database all the breeds are listed with the inherited diseases they can have and the mutation which can cause the diseases with a reference to the published manuscript. Also PubMed was searched to find other articles about the offered DNA tests and were reviewed to judge the utility and quality of the DNA tests.

To create the definitive list of valid DNA test all the tests available from the four biggest laboratories, active on the Netherlands market, were gathered. The four laboratories which were examined are Laboklin, VetGen, Van Haeringen and Optigen. All DNA tests were ordered by breed. This resulted a good overview of which tests are available. The length of the list required a division into different organ systems. This project focused on cardiovascular and blood diseases, metabolic and immune problems and eye disorders.

To know if all laboratories based their DNA tests on reliable articles a mail was sent to ask on which articles the DNA tests are based. These mails enclosed in the appendix. (Appendix 5) They all used the database of PennGen. After reviewing all these articles about the DNA tests a list with a lot of information about the DNA test was formed. For all offered DNA test it was assessed whether the mutation was known and how the research was done. On the basis of this information all the unsubstantiated DNA tests have been removed from the list so only the reliable tests are still on the list.

Results

By weighing the factors that are useful to determine if the DNA test are valid, the criteria were set up. First we have to know if there is literature available for the mutations where the DNA test are based on. It is also useful to know if a test is a marker or a mutation test. Mutation tests are better because they indicate the mutation itself so it is a 100% sure the tested dog has the mutation. It is also important to know how the effect of the mutation inherited. Is it recessive or dominant and autosomal or X-linked or otherwise? To review all the literature, it was necessary to know which clinical criteria were used to select the case and control groups in DNA research as well as the steps to come to a valid test. It is also necessary to know which breed is used to determine the mutation because in different breeds different mutations may occur. If for example the mutation is found in a Beagle it is not sure if a Labrador with the same disease had the same mutation. Finally, decisions regarding the remaining list should be made based on cost of the DNA test and the time needed for the test result.

The result of gathering all the available DNA tests from Laboklin, VetGen, Van Haeringen and Optigen is a long list with many DNA tests. At this point the list contained 120 different diseases and 188 different breeds. Not all the diseases were offered for each breed but the list at this moment contains 683 combinations of diseases and breeds. (See appendix 1) In this list all available DNA tests are sorted by breed, so it is visible per breed which inherited disease can be tested. In this list there is more information about each test. Mostly this is information that is found on the different sites of the laboratories. This list contains all the DNA tests available and does not say anything about the validity of the DNA tests.

As described above the list is divided into different organ systems because of the length of the list. This article is about blood diseases, metabolic and immune problems and eye disorders. The result of this was three different lists for the three different groups of diseases. (See appendix 2)

In order to obtain more information about the DNA tests and to know if all the available DNA tests are reliable, the literature was reviewed. The response to the mails we had sent to the laboratories was that they all used the database of PennGen for the relevant publications. (Appendix 5). There were many articles in the PennGen database which have been used. Other information was obtained reviewing the literature in PubMed. The result of this research is a new list with a lot more information about the DNA tests. (See Appendix 3)

Some of the test were very well described in the articles, for instance Copper toxicosis in Bedlington Terriers. In this article the mutation is very precisely described for the Bedlington Terriers. The mutation is a deletion in the exon 2 of the COMMD1 gene and this mutation is the main cause for copper toxicosis in the Belington Terrier (Forman, 2005). In this case it was very straightforward that this was a good DNA test. This was the case with many diseases of the list, for instance dry eye curly coat syndrome by the Cavalier King Charles Spaniel: this disease is caused by a deletion of a single base-pair in the FAM83H gene. (Forman, Oliver 2012) and also Pyruvate kinase deficiency in six different dog breeds (Basenjis, Cairn terrier, Westhighland white terrier, Labrador retriever, Pug and Beagle). For all these breeds different mutations were found. For the Labrador retriever, Westhighland white terrier and the Cairn terrier they found the same mutation, a C>T mutation that resulted in an early stop codon. In the Pug and the Beagle they found two different missense

mutations that resulted in loss of enzyme function. (Gultekin, 2012) For the Basenjis they also found a mutation in the gene so also for this breed the DNA test is validated. (Whitney, 1995)

An example of an incorrect test is Achromatopsia type 1 or day blindness. This is a DNA test offered for the Labrador retriever, although there is no peer reviewed published article about any genetic mutation. Therefore this was considered an unsubstantiated DNA test. The same applies for Haemophilia A (Factor VIII deficiency). There is no proof in any article that the mutation for this disease has been found. Therefore this test can be considered unreliable, meaning it should not be offered.

For other diseases the mutation was known for one breed but also offered for different breeds, for example Factor VII deficiency. Factor VII is necessary for the initiation of coagulation of the blood. When a dog is deficient for this factor it will lead to a bleeding disorder. The article about the mutation only concerns the beagle. (Callan, 2006) Nevertheless the DNA test for this mutation is also offered for the giant schnauzer, the Airedale terrier, the Alaskan klee kai and the Scottish deerhound. For these dog breeds there are no scientific articles available, which means the DNA test is not supported in a scientifically manner.

In other cases when a DNA test is offered, the supporting article, which should support the test by describing the mutation, actually states that the study of the mutation has not yet revealed the gene. This is the case for dilated cardiomyopathy in the Doberman pincher. The article is about a large study with 141 Doberman pinchers. Even though they investigated many genes, they did not find the specific gene for this disease. (Mausberg, Theresa-Bernadette 2011) There were other articles available (Meurs, Kathryn 2012). For instance, one article described finding a 16-bp deletion in the PDK4 gene that is associated with cardiomyopathy in Doberman pinschers. However, another article responds to that with a research in the European population where no evidence was found for the PDK4 gene involved. (Owczarek-Lipska, 2013) This inconclusive information indicates the need for more research.

This is also the case for Von-Willebrands Disease. There are three types of this disease of which type 1 is the most common. Type 2 is less common but when a dog is affected the clinical symptoms are more severe. Moreover, type 3 is the rarest form, which is also the worst form a dog can suffer from. The DNA test is offered for all the Von-Willebrands Disease types and for a lot of breeds, especially Von-Willebrands Disease Type 1. This DNA test is available for eleven different dog breeds. The article about the mutation (Rieger, 1998) reveals there is no knowledge about the genetic background of Von-Willebrands Disease Type 1. For the other types a mutation was discovered only for a small number of dog breeds. So the test is available for a lot of breeds but only found in a couple of breeds. This is the same for primary lens luxation. This test is offered for 28 different breeds mostly terriers. In the article about the ADAMTS17 gene they examined 30 different dog breeds. Only 17 of the screened breeds had the mutation in the ADAMTS17 gene. The other breeds do suffer from primary lens luxation but the mutation that causes the disease is not the ADAMTS17 gene or a different mutation is the same gene. (Gould, 2011) The available DNA test for the eleven remaining breeds are not supported by any literature.

A very difficult inherited disease is Progressive Retinal Atrophy. There are a lot of types of PRA and many genes play a role in this disease. One study was performed on the genes of PRA. (Downs, Louise 2014) They screened 231 dogs, representing 36 dog breeds, for 17 different mutations which cause PRA. The result of this large study was that 129 dogs were homozygous, 29 dogs were carriers and the remaining 73 dogs had no mutation on the

tested alleles. From all the 36 tested breed only in 21 breeds a mutations was found. Another interesting aspect is that the disease was found to be heterogeneous in 15 of these 21 breeds, so it is caused by at least two mutations. For 102 dogs the mutations remain unknown. These numbers indicate the complexity of this disease. In appendix 3 all the different types of PRA and the breeds are described.

It was also important to look if the same disease has different mutations in different breeds. This was the case for pyruvate kinase deficiency. The Basenjis, Cairn terrier, Westhighland White Terrier Labrador Retriever have the same mutation but the Pug end the Beagle have two different mutation for the same disease.

Since the list is only for Dutch breeder and veterinarians only the breeds that are registered by the "Raad van Beheer" are included. A couple examples of the breeds, for those who the tests are offered but the breeds are not registered, are the Alaskan klee kai, Italian Greyhound, Small Munsterlander, Polish Lowland Sheepdog. Another types of breeds are the breeds that are called differently in different countries. For instance the American shepherd and the Llewelin Setter. The American shepherd is the American name, whereas in Europe they call this breed the Australian shepherd. The Llewelin Setter is the pure strain form of the English setter. (Sparks, Troy 2002) Most of these breeds, which were not described in the literature, were already removed from the list since they do not have the mutation.

Through this research the 683 original combinations of DNA tests that are offered was reduced to only the good and substantiated DNA test. In appendix 4 the definitive list is included. The list contains eleven of cardiovascular and blood diseases, 23 metabolic and immune problems and 27 eye disorders.

Discussion/Conclusion

After setting the criteria for the DNA test, many criteria seemed very important. During the project the use of all of the criteria was revised, for example ‘in which country is the test designed?’. The tests are mostly designed in other countries than The Netherlands. In the articles it is often described where the dogs come from and therefore the country where the test is most likely made. However, this is difficult to establish for all the DNA tests so this criterion is considered less important. For the best results all the tests have to be tested in The Netherlands, which is an almost impossible task. There is also the criteria of how the DNA tests are developed. This was hard to examine because the literature was often only about the mutation. The laboratories make all the tests themselves. (See the mails in appendix 5). The criteria about how the effects of the mutations inherit is useful because the dominant diseases are only useful to test if the disease only occurs in old aged dogs. Otherwise the disease can be seen before the dog is used for breeding. The DNA tests are particularly useful in order to detect carriers of recessive diseases. The criteria of what kind of DNA test is conducted, marker of mutation test was a very important criteria. When the project progressed it appeared that all the offered DNA tests were mutation tests. The two criteria that are the most important are: (1) if there is literature available and (2) for which breed is the DNA test created? These were the two criteria to which all the DNA tests must comply.

The reviewing of the literature indicated some interesting findings. When gathering all the information from the laboratories sometimes two different names for the same disease were used. Mostly they had the same mutation so we could combine them. For example Van Haeringen called a disease Multidrug Resistance 1 while Laboklin called it Ivermectin hypersensitivity (MDR1 gene defect). Also the data base of PenGenn did not always provide the proper information. Some articles did not exist anymore so other articles were sought in PubMed. There were a couple of diseases which were offered for all breeds for example Thrombasthenia 2. The article is only about the Pyrenean Mountain Dog so all the other breeds are not supported. There were also difficulties with some breeds, for instance the collies. In America the collies are not divided in different breeds but in Europe there are a lot of different collies. Other breeds are not registered by the ‘‘Raad van Beheer’’ so we did not include them in the definitive list. Sometimes the laboratories claim they found a mutation by themselves for a particular breed. In this case there is no publication available so we cannot know whether these claims are substantiated. Von-Willebrand disease type 1 is a not so severe disease that is very common. Every dog, with the risk of this disease could be tested but that is a little exaggerated. It is also advisable only to test the dogs who undergo surgery for example.

The overall conclusion of this project is that a lot of research has been done on DNA tests and many of the offered DNA tests were valuable tests. However there are a lot of tests, which are not based on any peer reviewed articles or the articles were easily misinterpreted. In some cases it looked like there was a mutation found but actually there is more research necessary. Since the laboratories are still doing a lot of investigation on DNA tests, the list will be longer and more complete over the course of the next few years.

Reference

- Aguirre, G. D., Baldwin, V., Pearce Kelling, S., Narfström, K., Ray, K., & Acland, G. M. (1998). Congenital stationary night blindness in the dog: Common mutation in the RPE65 gene indicates founder effect. *Molecular Vision*, 4, 23.
- Ahonen, S., Arumilli, M., & Lohi, H. (2013). A CNGB1 frameshift mutation in papillon and phalène dogs with progressive retinal atrophy. *Plos One*, 8(8), e72122. doi:10.1371/journal.pone.0072122
- Ameratunga, R., Winkelstein, J. A., Brody, L., Binns, M., Cork, L. C., Colombani, P., et al. (1998). Molecular analysis of the third component of canine complement (C3) and identification of the mutation responsible for hereditary canine C3 deficiency. *The Journal of Immunology*, 160(6), 2824-30.
- Aronovich, E. L., Carmichael, K. P., Morizono, H., Koutlas, I. G., Deanching, M., Hoganson, G., et al. (2000). Canine heparan sulfate sulfamidase and the molecular pathology underlying sanfilippo syndrome type A in dachshunds. *Genomics*, 68(1), 80-4. doi:10.1006/geno.2000.6275
- Benson. (2003). Mutations associated with neutropenia in dogs and humans disrupt intracellular transport of neutrophil elastase. *Nature Genetics*, 35(1), 90-96.
- Benson, K., Li, F., Person, R., Albani, D., Duan, Z., Wechsler, J., et al. (2003). Mutations associated with neutropenia in dogs and humans disrupt intracellular transport of neutrophil elastase. *Nature Genetics*, 35(1), 90-6. doi:10.1038/ng1224
- Beurlet, S., Krief, P., Sansonetti, A., Briend Marchal, A., Kiladjian, J., Padua, R., et al. (2011). Identification of JAK2 mutations in canine primary polycythemia. *Experimental Hematology*, 39(5), 542-5. doi:10.1016/j.exphem.2011.02.003
- Boudreaux. (2011). P2Y12 receptor gene mutation associated with postoperative hemorrhage in a greater swiss mountain dog. *Veterinary Clinical Pathology*, 40(2), 202-206.
- Boudreaux, M. K., & Catalfamo, J. L. (2001). Molecular and genetic basis for thrombasthenic thrombopathia in otterhounds. *American Journal of Veterinary Research*, 62(11), 1797-804.
- Boudreaux, M. K., Wardrop, K. J., Kiklevich, V., Felsburg, P., & Snekvik, K. (2010). A mutation in the canine kindlin-3 gene associated with increased bleeding risk and susceptibility to infections. *Thrombosis and Haemostasis*, 103(2), 475-7. doi:10.1160/TH09-09-0571
- Boudreaux, M., Catalfamo, J., & Klok, M. (2007). Calcium-diacylglycerol guanine nucleotide exchange factor I gene mutations associated with loss of function in canine platelets. *Translational Research*, 150(2), 81-92. doi:10.1016/j.trsl.2007.03.006
- Boudreaux, M., & Martin, M. (2011). P2Y12 receptor gene mutation associated with postoperative hemorrhage in a greater swiss mountain dog. *Veterinary Clinical Pathology*, 40(2), 202-6. doi:10.1111/j.1939-165X.2011.00318.x

- Brooks, M., Gu, W., Barnas, J., Ray, J., & Ray, K. (2003). A line 1 insertion in the factor IX gene segregates with mild hemophilia B in dogs. *Mammalian Genome*, 14(11), 788-95. doi:10.1007/s00335-003-2290-z
- Callan, M. B., Aljamali, M. N., Margaritis, P., Griot Wenk, M. E., Pollak, E. S., Werner, P., et al. (2006). A novel missense mutation responsible for factor VII deficiency in research beagle colonies. *Journal of Thrombosis and Haemostasis*, 4(12), 2616-22. doi:10.1111/j.1538-7836.2006.02203.x
- Cameron, J., Maj, M., Levandovskiy, V., MacKay, N., Shelton, G. D., & Robinson, B. (2007). Identification of a canine model of pyruvate dehydrogenase phosphatase 1 deficiency. *Molecular Genetics and Metabolism*, 90(1), 15-23. doi:10.1016/j.ymgme.2006.09.011
- Database of PennGen. (2015). Retrieved June, 2015, from <http://research.vet.upenn.edu/penngen/AvailableTests/TestsAvailableatLabsWorldwide/tabid/7620/Default.aspx>
- Davis, B., Toivio Kinnucan, M., Schuller, S., & Boudreaux, M. K. (2008). Mutation in beta1-tubulin correlates with macrothrombocytopenia in cavalier king charles spaniels. *Journal of Veterinary Internal Medicine*, 22(3), 540-5. doi:10.1111/j.1939-1676.2008.0085.x
- Dekomien, G., Runte, M., Gödde, R., & Epplen, J. T. (2000). Generalized progressive retinal atrophy of sloughi dogs is due to an 8-bp insertion in exon 21 of the PDE6B gene. *Cytogenetics and Cell Genetics*, 90(3-4), 261-7.
- Dodgson, S. E., Day, R., & Fyfe, J. C. (2012). Congenital hypothyroidism with goiter in tenterfield terriers. *Journal of Veterinary Internal Medicine*, 26(6), 1350-7. doi:10.1111/j.1939-1676.2012.01015.x
- Downs, L. M., Bell, J. S., Freeman, J., Hartley, C., Hayward, L. J., & Mellersh, C. S. (2013). Late-onset progressive retinal atrophy in the gordon and irish setter breeds is associated with a frameshift mutation in C2orf71. *Animal Genetics*, 44(2), 169-77. doi:10.1111/j.1365-2052.2012.02379.x
- Downs, L., Hitti, R., Pregolato, S., & Mellersh, C. (2014). Genetic screening for PRA-associated mutations in multiple dog breeds shows that PRA is heterogeneous within and between breeds. *Veterinary Ophthalmology*, 17(2), 126-30. doi:10.1111/vop.12122
- Erkens. (2009). Presence of the ABCB1 (MDR1) deletion mutation causing ivermectin hypersensitivity in certain dog breeds in belgium. *Vlaams Diergeneeskundig Tijdschrift*, 78(4), 256-260.
- Evans, J. P., Brinkhous, K. M., Brayer, G. D., Reisner, H. M., & High, K. A. (1989). Canine hemophilia B resulting from a point mutation with unusual consequences. *Proceedings of the National Academy of Sciences of the United States of America*, 86(24), 10095-9.
- Expertisecentrum genetica gezelschapsdieren. (2015). Retrieved June, 2015, from <http://www.diergeneeskunde.nl/klinieken/expertisecentrum-genetica-gezelschapsdieren/>
- Forman, O. P., Bournsnel, M. E. G., Dunmore, B. J., Stendall, N., van den Sluis, B., Fretwell, N., et al. (2005). Characterization of the COMMD1 (MURR1) mutation causing copper toxicosis in bedlington terriers. *Animal Genetics*, 36(6), 497-501. doi:10.1111/j.1365-2052.2005.01360.x

- Forman, O., Penderis, J., Hartley, C., Hayward, L., Ricketts, S., & Mellersh, C. (2012). Parallel mapping and simultaneous sequencing reveals deletions in BCAN and FAM83H associated with discrete inherited disorders in a domestic dog breed. *PLOS Genetics*, *8*(1), e1002462. doi:10.1371/journal.pgen.1002462
- Fyfe, J. C., Hemker, S. L., Venta, P. J., Stebbing, B., & Giger, U. (2014). Selective intestinal cobalamin malabsorption with proteinuria (imerslund-gräsbeck syndrome) in juvenile beagles. *Journal of Veterinary Internal Medicine*, *28*(2), 356-62. doi:10.1111/jvim.12284
- Fyfe, J. C., Hemker, S. L., Venta, P. J., Fitzgerald, C. A., Outerbridge, C. A., Myers, S. L., et al. (2013). An exon 53 frameshift mutation in CUBN abrogates cubam function and causes imerslund-gräsbeck syndrome in dogs. *Molecular Genetics and Metabolism*, *109*(4), 390-6. doi:10.1016/j.ymgme.2013.05.006
- Gerber, K., Harvey, J., D'Agorne, S., Wood, J., & Giger, U. (2009). Hemolysis, myopathy, and cardiac disease associated with hereditary phosphofructokinase deficiency in two whippets. *Veterinary Clinical Pathology*, *38*(1), 46-51. doi:10.1111/j.1939-165X.2008.00089.x
- Giger, U., Smith, B. F., Woods, C. B., Patterson, D. F., & Stedman, H. (1992). Inherited phosphofructokinase deficiency in an american cocker spaniel. *Journal of the American Veterinary Medical Association*, *201*(10), 1569-71.
- Goldstein, O., Guyon, R., Kukekova, A., Kuznetsova, T. N., Pearce-Kelling, S. E., Johnson, J., et al. (2010). COL9A2 and COL9A3 mutations in canine autosomal recessive oculoskeletal dysplasia. *Mammalian Genome*, *21*(7-8), 398-408. doi:10.1007/s00335-010-9276-4
- Goldstein, O., Jordan, J. A., Aguirre, G. D., & Acland, G. M. (2013). A non-stop S-antigen gene mutation is associated with late onset hereditary retinal degeneration in dogs. *Molecular Vision*, *19*, 1871-84.
- Goldstein, O., Jordan, J., Aguirre, G., & Acland, G. (2013). A non-stop S-antigen gene mutation is associated with late onset hereditary retinal degeneration in dogs. *Molecular Vision*, *19*, 1871-84.
- Goldstein, O., Mezey, J., Boyko, A., Gao, C., Wang, W., Bustamante, C., et al. (2010). An ADAM9 mutation in canine cone-rod dystrophy 3 establishes homology with human cone-rod dystrophy 9. *Molecular Vision*, *16*, 1549-69.
- Goldstein, O., Mezey, J., Schweitzer, P., Boyko, A., Gao, C., Bustamante, C., et al. (2013). IQCB1 and PDE6B mutations cause similar early onset retinal degenerations in two closely related terrier dog breeds. *Investigative Ophthalmology & Visual Science*, *54*(10), 7005-19. doi:10.1167/iovs.13-12915
- Gornik. (2014). Canine multifocal retinopathy caused by a BEST1 mutation in a boerboel. *Veterinary Ophthalmology*, *17*(5), 368-372.
- Gould, D., Pettitt, L., McLaughlin, B., Holmes, N., Forman, O., Thomas, A., et al. (2011). ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Veterinary Ophthalmology*, *14*(6), 378-84. doi:10.1111/j.1463-5224.2011.00892.x

- Gould, D. (2011). ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. *Veterinary Ophthalmology*, 14(6), 378-384. doi:10.1111/j.1463-5224.2011.00892.x
- Gregory, B. L., Shelton, G. D., Bali, D. S., Chen, Y., & Fyfe, J. C. (2007). Glycogen storage disease type IIIa in curly-coated retrievers. *Journal of Veterinary Internal Medicine*, 21(1), 40-6.
- Gu, W., Brooks, M., Catalfamo, J., Ray, J., & Ray, K. (1999). Two distinct mutations cause severe hemophilia B in two unrelated canine pedigrees. *Thrombosis and Haemostasis*, 82(4), 1270-5.
- Gultekin, G. I., Raj, K., Foureman, P., Lehman, S., Manhart, K., Abdulmalik, O., et al. (2012). Erythrocytic pyruvate kinase mutations causing hemolytic anemia, osteosclerosis, and secondary hemochromatosis in dogs. *Journal of Veterinary Internal Medicine*, 26(4), 935-44. doi:10.1111/j.1939-1676.2012.00958.x
- Guziewicz, K. E., Zangerl, B., Lindauer, S. J., Mullins, R. F., Sandmeyer, L. S., Grahn, B. H., et al. (2007). Bestrophin gene mutations cause canine multifocal retinopathy: A novel animal model for best disease. *Investigative Ophthalmology & Visual Science*, 48(5), 1959-67. doi:10.1167/iovs.06-1374
- Guziewicz, K., Zangerl, B., Lindauer, S., Mullins, R., Sandmeyer, L., Grahn, B., et al. (2007). Bestrophin gene mutations cause canine multifocal retinopathy: A novel animal model for best disease. *Investigative Ophthalmology & Visual Science*, 48(5), 1959-67. doi:10.1167/iovs.06-1374
- Hoffmann, I., Guziewicz, K., Zangerl, B., Aguirre, G., & Mardin, C. (2012). Canine multifocal retinopathy in the australian shepherd: A case report. *Veterinary Ophthalmology*, 15 Suppl 2, 134-8. doi:10.1111/j.1463-5224.2012.01005.x
- Hytonen. (2012). A novel GUSB mutation in brazilian terriers with severe skeletal abnormalities defines the disease as mucopolysaccharidosis VII. *Plos One*, 7(7), e40281.
- Inal Gultekin, G., Raj, K., Lehman, S., Hillström, A., & Giger, U. (2012). Missense mutation in PFKM associated with muscle-type phosphofructokinase deficiency in the wachtelhund dog. *Molecular and Cellular Probes*, 26(6), 243-7. doi:10.1016/j.mcp.2012.02.004
- Kijas, J. M., Bauer, T. R., Gäfvert, S., Marklund, S., Trowald Wigh, G., Johannisson, A., et al. (1999). A missense mutation in the beta-2 integrin gene (ITGB2) causes canine leukocyte adhesion deficiency. *Genomics*, 61(1), 101-7. doi:10.1006/geno.1999.5948
- Kishnani, P. S., Bao, Y., Wu, J. Y., Brix, A. E., Lin, J. L., & Chen, Y. T. (1997). Isolation and nucleotide sequence of canine glucose-6-phosphatase mRNA: Identification of mutation in puppies with glycogen storage disease type ia. *Biochemical and Molecular Medicine*, 61(2), 168-77.
- Kramer, J. W., Venta, P. J., Klein, S. R., Cao, Y., Schall, W. D., & Yuzbasiyan Gurkan, V. (2004). A von willebrand's factor genomic nucleotide variant and polymerase chain reaction diagnostic test associated with inheritable type-2 von willebrand's disease in a line of german shorthaired pointer dogs. *Veterinary Pathology*, 41(3), 221-8. doi:10.1354/vp.41-3-221
- Kuchtey, J., Olson, L., Rinkoski, T., Mackay, E., Iverson, T. M., Gelatt, K., et al. (2011). Mapping of the disease locus and identification of ADAMTS10 as a candidate gene in a canine model of primary open angle glaucoma. *PLOS Genetics*, 7(2), e1001306. doi:10.1371/journal.pgen.1001306

- Kukekova, A. V., Goldstein, O., Johnson, J. L., Richardson, M. A., Pearce-Kelling, S. E., Swaroop, A., et al. (2009). Canine RD3 mutation establishes rod-cone dysplasia type 2 (rcd2) as ortholog of human and murine rd3. *Mammalian Genome*, 20(2), 109-23. doi:10.1007/s00335-008-9163-4
- Kukekova, A., Goldstein, O., Johnson, J., Richardson, M., Pearce Kelling, S., Swaroop, A., et al. (2009). Canine RD3 mutation establishes rod-cone dysplasia type 2 (rcd2) as ortholog of human and murine rd3. *Mammalian Genome*, 20(2), 109-23. doi:10.1007/s00335-008-9163-4
- LeVine, D. N., Zhou, Y., Ghiloni, R. J., Fields, E. L., Birkenheuer, A. J., Gookin, J. L., et al. (2009). Hereditary 1,25-dihydroxyvitamin D-resistant rickets in a pomeranian dog caused by a novel mutation in the vitamin D receptor gene. *Journal of Veterinary Internal Medicine*, 23(6), 1278-83. doi:10.1111/j.1939-1676.2009.0405.x
- Lipscomb, D. L., Bourne, C., & Boudreaux, M. K. (2000). Two genetic defects in alphaIIb are associated with type I glanzmann's thrombasthenia in a great pyrenees dog: A 14-base insertion in exon 13 and a splicing defect of intron 13. *Veterinary Pathology*, 37(6), 581-8.
- Mausberg, T., Wess, G., Simak, J., Keller, L., Drögemüller, M., Drögemüller, C., et al. (2011). A locus on chromosome 5 is associated with dilated cardiomyopathy in doberman pinschers. *Plos One*, 6(5), e20042. doi:10.1371/journal.pone.0020042
- Mauser, A. E., Whitlark, J., Whitney, K. M., & Lothrop, C. D. (1996). A deletion mutation causes hemophilia B in Ihasa apso dogs. *Blood*, 88(9), 3451-5.
- Mealey, K., & Meurs, K. (2008). Breed distribution of the ABCB1-1Delta (multidrug sensitivity) polymorphism among dogs undergoing ABCB1 genotyping. *Journal of the American Veterinary Medical Association*, 233(6), 921-4. doi:10.2460/javma.233.6.921
- Meek, K., Kienker, L., Dallas, C., Wang, W., Dark, M. J., Venta, P. J., et al. (2001). SCID in jack russell terriers: A new animal model of DNA-PKcs deficiency. *The Journal of Immunology*, 167(4), 2142-50.
- Mellersh, C., Pettitt, L., Forman, O., Vaudin, M., & Barnett, K. (2006). Identification of mutations in HSF4 in dogs of three different breeds with hereditary cataracts. *Veterinary Ophthalmology*, 9(5), 369-78. doi:10.1111/j.1463-5224.2006.00496.x
- Mellersh, C. (2012). DNA testing and domestic dogs. *Mammalian Genome*, 23(1-2), 109-23. doi:10.1007/s00335-011-9365-z
- Meurs, K. M., Lahmers, S., Keene, B. W., White, S. N., Oyama, M. A., Mauceli, E., et al. (2012). A splice site mutation in a gene encoding for PDK4, a mitochondrial protein, is associated with the development of dilated cardiomyopathy in the doberman pinscher. *Human Genetics*, 131(8), 1319-25. doi:10.1007/s00439-012-1158-2
- Mischke, R., Kühnlein, P., Kehl, A., Langbein Detsch, I., Steudle, F., Schmid, A., et al. (2011). G244E in the canine factor IX gene leads to severe haemophilia B in rhodesian ridgebacks. *The Veterinary Journal*, 187(1), 113-8. doi:10.1016/j.tvjl.2010.01.017
- Miyadera, K., Kato, K., Aguirre Hernández, J., Tokuriki, T., Morimoto, K., Busse, C., et al. (2009). Phenotypic variation and genotype-phenotype discordance in canine cone-rod dystrophy with an RPGRIP1 mutation. *Molecular Vision*, 15, 2287-305.

- Miyadera, K., Kato, K., Aguirre-Hernández, J., Tokuriki, T., Morimoto, K., Busse, C., et al. (2009). Phenotypic variation and genotype-phenotype discordance in canine cone-rod dystrophy with an RPGRIP1 mutation. *Molecular Vision*, *15*, 2287-305.
- Mizukami, K., Chang, H., Ota, M., Yabuki, A., Hossain, M. A., Rahman, M. M., et al. (2012). Collie eye anomaly in hokkaido dogs: Case study. *Veterinary Ophthalmology*, *15*(2), 128-32. doi:10.1111/j.1463-5224.2011.00950.x
- Mizukami, K., Shoubudani, T., Nishimoto, S., Kawamura, R., Yabuki, A., & Yamato, O. (2012). Trapped neutrophil syndrome in a border collie dog: Clinical, clinico-pathologic, and molecular findings. *Journal of Veterinary Medical Science*, *74*(6), 797-800.
- Okawa, T., Yanase, T., Shimokawa Miyama, T., Hiraoka, H., Baba, K., Tani, K., et al. (2011). Prekallikrein deficiency in a dog. *Journal of Veterinary Medical Science*, *73*(1), 107-11.
- Olson, L. M., Rinkoski, T., Mackay, E. O., Iverson, T. M., Gelatt, K. N., Haines, J. L., et al. (2011). Mapping of the disease locus and identification of ADAMTS10 as a candidate gene in a canine model of primary open angle glaucoma. *PLOS Genetics*, *7*(2), e1001306. doi:10.1371/journal.pgen.1001306
- Owczarek-Lipska, M., Mausberg, T., Stephenson, H., Dukes-McEwan, J., Wess, G., & Leeb, T. (2013). A 16-bp deletion in the canine PDK4 gene is not associated with dilated cardiomyopathy in a european cohort of doberman pinschers. *Animal Genetics*, *44*(2), 239. doi:10.1111/j.1365-2052.2012.02396.x
- Parker, H., Kukekova, A., Akey, D., Goldstein, O., Kirkness, E., Baysac, K., et al. (2007). Breed relationships facilitate fine-mapping studies: A 7.8-kb deletion cosegregates with collie eye anomaly across multiple dog breeds. *Genome Research*, *17*(11), 1562-71. doi:10.1101/gr.6772807
- Perryman, L. E. (2004). Molecular pathology of severe combined immunodeficiency in mice, horses, and dogs. *Veterinary Pathology*, *41*(2), 95-100. doi:10.1354/vp.41-2-95
- Petersen-Jones, S. M., Entz, D. D., & Sargan, D. R. (1999). cGMP phosphodiesterase-alpha mutation causes progressive retinal atrophy in the cardigan welsh corgi dog. *Investigative Ophthalmology & Visual Science*, *40*(8), 1637-44.
- Pilorge. (2007). Advantages and limitations of DNA tests in veterinary ophthalmology in dogs. *Le Nouveau Praticien Veterinaire Canine - Feline*, (33), 27-31.
- Rieger, M. (1998). Identification of mutations in the canine von willebrand factor gene associated with type III von willebrand disease. *Thrombosis and Haemostasis*, *80*(2), 332-337.
- Seppälä, E. H., Reuser, A. J. J., & Lohi, H. (2013). A nonsense mutation in the acid a-glucosidase gene causes pompe disease in finnish and swedish lapphunds. *Plos One*, *8*(2), e56825. doi:10.1371/journal.pone.0056825
- Sidjanin, D., Lowe, J., McElwee, J., Milne, B., Phippen, T., Sargan, D., et al. (2002). Canine CNGB3 mutations establish cone degeneration as orthologous to the human achromatopsia locus ACHM3. *Human Molecular Genetics*, *11*(16), 1823-33.

- Silverstein Dombrowski, D., Carmichael, K. P., Wang, P., O'Malley, T., Haskins, M., & Giger, U. (2004). Mucopolysaccharidosis type VII in a german shepherd dog. *Journal of the American Veterinary Medical Association*, 224(4), 553-7, 532.
- Skelly, B. J., Sargan, D. R., Winchester, B. G., Smith, M. O., Herrtage, M. E., & Giger, U. (1999). Genomic screening for fucosidosis in english springer spaniels. *American Journal of Veterinary Research*, 60(6), 726-9.
- Skelly, B. J., Wallace, M., Rajpurohit, Y. R., Wang, P., & Giger, U. (1999). Identification of a 6 base pair insertion in west highland white terriers with erythrocyte pyruvate kinase deficiency. *American Journal of Veterinary Research*, 60(9), 1169-72.
- Slutsky, J., Raj, K., Yuhnke, S., Bell, J., Fretwell, N., Hedhammar, A., et al. (2013). A web resource on DNA tests for canine and feline hereditary diseases. *The Veterinary Journal*, 197(2), 182-187. doi:<http://dx.doi.org/10.1016/j.tvjl.2013.02.021>
- Smith, B. F., Stedman, H., Rajpurohit, Y., Henthorn, P. S., Wolfe, J. H., Patterson, D. F., et al. (1996). Molecular basis of canine muscle type phosphofructokinase deficiency. *Journal of Biological Chemistry*, 271(33), 20070-4.
- Sparks, T. D. (2002). *About llewellin setter*. Retrieved June 25, 2015, from http://www.nalba.org/llewellin_setters.htm
- Veranderingen wet dieren 1 juli 2014*. (2014). Retrieved June, 2015, from <https://www.knmvd.nl/actueel/nieuws/item/10848379/Veranderingen-Wet-Dieren-1-juli-2014>
- Verfuurden, B., Wempe, F., Reinink, P., van Kooten, P J S, Martens, E., Gerritsen, R., et al. (2011). Severe combined immunodeficiency in frisian water dogs caused by a RAG1 mutation. *Genes and Immunity*, 12(4), 310-3. doi:10.1038/gene.2011.6
- Website of laboklin*. (2015). Retrieved June, 2015, from <http://www.laboklin.de/index.php?link=labogen/pages/html/en/home.html>
- Website of optigen*. (2015). Retrieved June, 2015, from https://www.optigen.com/opt9_test.html
- Website of van Haeringen*. (2015). Retrieved June, 2015, from <https://www.vhlgenetics.com/Webshop/Dieren/Hond/tabid/256/catid/6/language/nl-NL/Default.aspx>
- Website of VetGen*. (2015). Retrieved June, 2015, from <http://vetgen.com/AllBreedsList.aspx>
- Wet dieren*. (2014). Retrieved June, 2015, from http://wetten.overheid.nl/BWBR0030250/Hoofdstuk2/1/Artikel26/geldigheidsdatum_26-06-2015
- Whitney, K. M., & Lothrop, C. D. (1995). Genetic test for pyruvate kinase deficiency of basenjis. *Journal of the American Veterinary Medical Association*, 207(7), 918-21.

- Winkler, P., Gornik, K., Ramsey, D., Dubielzig, R., Venta, P., Petersen Jones, S., et al. (2014). A partial gene deletion of SLC45A2 causes oculocutaneous albinism in doberman pinscher dogs. *Plos One*, 9(3), e92127. doi:10.1371/journal.pone.0092127
- Zangerl, B., Goldstein, O., Philp, A. R., Lindauer, S. J. P., Pearce-Kelling, S. E., Mullins, R. F., et al. (2006). Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*, 88(5), 551-63. doi:10.1016/j.ygeno.2006.07.007
- Zangerl, B., Goldstein, O., Philp, A., Lindauer, S. J. P., Pearce Kelling, S., Mullins, R., et al. (2006). Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. *Genomics*, 88(5), 551-63. doi:10.1016/j.ygeno.2006.07.007
- Zeng, R., Coates, J. R., Johnson, G. C., Hansen, L., Awano, T., Kolicheski, A., et al. (2014). Breed distribution of SOD1 alleles previously associated with canine degenerative myelopathy. *Journal of Veterinary Internal Medicine*, 28(2), 515-521. Retrieved from SCOPUS database.
- Zhang, Q., Acland, G. M., Wu, W. X., Johnson, J. L., Pearce-Kelling, S., Tulloch, B., et al. (2002). Different RPGR exon ORF15 mutations in canids provide insights into photoreceptor cell degeneration. *Human Molecular Genetics*, 11(9), 993-1003.
- Zhang, Q., Acland, G., Wu, W., Johnson, J., Pearce Kelling, S., Tulloch, B., et al. (2002). Different RPGR exon ORF15 mutations in canids provide insights into photoreceptor cell degeneration. *Human Molecular Genetics*, 11(9), 993-1003.

Appendix 1

Table 1 List with all the diseases.

<i>Breeds</i>	<i>Disease</i>	<i>Mono/poly, recessive/dominant</i>	<i>Mutation</i>	<i>Which lab.</i>	<i>Results Time/Price</i>
<i>All Breeds</i>	Canine degenerative myelopathy (DM)	Autosomaal recessive	SOD1-gene	Laboklin	3-5 days
			SOD1-gene	Van Haeringen	<20 days € 79,50
	Hiplaxity 1/2	Multifactorial origin		Van Haeringen	<10 days € 39,50
	Hyperuricemia (HUU)	Autosomal recessive	SLC2A9 Gene	Van Haeringen	<10 days € 39,50
				Laboklin	3-5 days
	Malignant hyperthermia (MH)	Autosomal dominant		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
	Multidrug Resistance 1 (MDR1)	Autosomal recessive	MDR1 Gene	Van Haeringen	<10 days € 80,00
	Polycythemia	Autosomal dominant	JAK2 Gene	Van Haeringen	<10 days € 39,50
Thrombasthenia 2	Autosomal recessive		Van Haeringen	<10 days € 39,50	
<i>Airedale Terrier</i>	Faktor VII - Deficiency	Autosomal recessive		Laboklin	3-5 days
				VetGen	\$65.00 USD
	Hemophilia B (Facort IX deficiency)	X-linked recessive		VetGen	\$65.00 USD
<i>Alaskan Klee Kai</i>	Faktor VII - Deficiency	Autosomal recessive		Laboklin	3-5 days
				VetGen	\$65.00 USD
<i>Alaskan malamute</i>	Polyneuropathy 1	Autosomal recessive		Laboklin	1-2 weeks
				Van Haeringen	<10 days € 39,50
<i>American Bulldog</i>	Cone Degeneration (CD)	Autosomal recessive	CNGB3 Gene	Optigen	
	Neuronal ceroid lipofuscinosis (NCL) 10	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				VetGen	\$65.00 USD
Canine Multi-focal Retinopathy (CMR)	Autosomal recessive	VMD2 Gene	Optigen	US\$95.00	

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price
American Cocker Spaniel	Phosphofructokinase deficiency (PFKD)	Autosomal recessive		Laboklin	1-2 weeks
				VetGen	\$65.00 USD
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
American Eskimo Dog	FN (Familial Nephropathy)	Autosomal recessive		Van Haeringen	<10 days € 110,00
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
			prcd Gene	Optigen	\$195
	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
			2-20% of carriers will develop condition	Optigen	\$90
American Hairless Terrier	Thrombopathia 2	Autosomal recessive		Van Haeringen	<10 days € 39,50
	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
American Pitbull Terrier				Optigen	\$90
	Cerebellar Ataxia / Neuronal ceroid lipofuscinosis (NCL), 4A	Autosomal recessive		Optigen	3-4 weeks \$150
	Cone Rod Dystrophy 2 (CRD2)	Autosomal recessive		Optigen	<2 weeks \$120
American Staffordshire Terrier	Cerebellar Ataxia / Neuronal ceroid lipofuscinosis (NCL), 4A	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				Optigen	3-4 weeks \$150
	Cone Rod Dystrophy 2 (CRD2)	Autosomal recessive		Van Haeringen	<25 days € 100,00
	Hyperuricosuria (HU)	Autosomal recessive		VetGen	\$65.00 USD
Australian Cattle Dog	Cystinuria	Autosomal dominant		Laboklin	1-2 weeks
				Van Haeringen	<10 days € 39,50
	Myotonia Congenita 2	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
			prcd Gene	Optigen	\$195
	rcd4 Progressive Retinal Atrophy (rcd4 PRA)	Autosomal recessive		Laboklin	1-2 weeks
		C2orf71 Gene	Optigen	\$95	

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price
<i>(sequel)Australian Cattle Dog</i>	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
		Carriers have a small chance of getting sick.		VetGen	\$65.00 USD
		2-20% of carriers will develop condition		Optigen	\$90
<i>Australian Shepherd</i>	CMR1 (Canine Multifocal Retinopathy)	Autosomal recessive		Van Haeringen	<10 days € 39,50
			BEST1 gene	VetGen	\$65.00 USD
			VMD2 Gene	Optigen	US\$95.00
	Collie Eye Anomalie (CEA)	Autosomal recessive		Laboklin	4-6 weeks
			chromosome number 37	Optigen	\$180
	Cyclic Neutropenia (CN)	Autosomal recessive		VetGen	\$65.00 USD
	Hereditary Cataract (HC)	Autosomal dominant	HSF4 Gene	Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
		Autosomal co-dominant	HSF4-2 Gene	Optigen	\$100
	Hyperuricosuria (HU)	Autosomal recessive		VetGen	\$65.00 USD
	Ivermectin hypersensitivity (MDR1 gene defect)	Autosomal recessive	mdr1 gene	Laboklin	1-2 weeks
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
		prcd Gene	Optigen	\$195	
Cone Degeneration (CD)	Autosomal recessive	CNGB3 Gene	Optigen	\$160	
<i>Australian Silky Terrier</i>	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
<i>Australian Stumpy Tail Cattle Dog</i>	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive	prcd Gene	Optigen	\$195
<i>Basenjis</i>	Pyruvate kinase deficiency (PK)	Autosomal recessive		Laboklin	1-2 weeks
				VetGen	\$65.00 USD
				Optigen	\$80
	Basenji Progressive Retinal Atrophy (bas PRA)	Autosomal recessive		Optigen	\$95

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price		
Basset	Thrombopathia	Autosomal recessive		Van Haeringen	<10 days € 39,50		
				Laboklin	1-2 weeks		
	X-linked severe combined Immunodeficiency (X-SCID)	X-Chromosomal		Van Haeringen	<10 days € 39,50		
				Laboklin	1-2 weeks		
Beagle	Factor VII deficiency	Autosomal recessive		Van Haeringen	<10 days € 39,50		
				Laboklin	3-5 days		
				VetGen	\$65.00 USD		
	Primary open angle glaucoma (POAG)	Autosomal recessive			Van Haeringen	<10 days € 39,50	
					Laboklin	1-2 weeks	
					ADAMTS10	Optigen	\$95
	Musladin-Lueke syndrome (MLS)	Autosomal recessive			Van Haeringen	<10 days € 39,50	
					Laboklin	3-5 days	
						Laboklin	3-5 weeks
	Neonatal cortical cerebellar atrophy (NCCD)	Autosomal recessive			Laboklin	1-2 weeks	
						Van Haeringen	<10 days € 39,50
						Van Haeringen	<10 days € 39,50
Osteogenesis Imperfecta	Autosomal dominant			Van Haeringen	<10 days € 39,50		
					Van Haeringen	<10 days € 39,50	
					Van Haeringen	<10 days € 39,50	
Pyruvate kinase Deficiency 3 (PKDef)	Autosomal recessive			Laboklin	1-2 weeks		
					VetGen	\$65.00 USD	
					Laboklin	4-6 weeks	
Bearded Collie	Collie Eye Anomalie (CEA)	Autosomal recessive		Laboklin	4-6 weeks		
					Van Haeringen	<25 days € 140,00	
Bedlington Terriers	Copper toxicosis	Autosomal recessive		chromosome number 37	Optigen	\$180	
					Laboklin	1-2 weeks	
					Van Haeringen	<10 days € 39,50	
				Commd1	VetGen	\$65.00 USD	

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price
Bernese Mountain Dog	Von-Willebrands Disease Type 1	Autosomal dominant variable penetrance		Van Haeringen	<10 days € 89,00
				Laboklin	3-5 days
				VetGen	Binnen 2 weeks \$65.00 USD
Bichon Frise	Macrothrombocytopenia (MTC)	Autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD
Black Russian Terrier	hyperuricosuria (HU)	Autosomal recessive		VetGen	\$65.00 USD
Boerboel	CMR1 (Canine Multifocal Retinopathy)	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Optigen	US\$95.00
	hyperuricosuria (HU)	Autosomal recessive		VetGen	\$65.00 USD
Bolonka Zwetna	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive	prcd Gene	Optigen	\$195
Border Collie	Collie Eye Anomalie (CEA)	Autosomal recessive		Laboklin	4-6 weeks
				Van Haeringen	<25 days € 140,00
				Optigen	\$180
				Laboklin	1-2 weeks
				chromosome number 37	
				Mdr1 gene	1-2 weeks
				Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				Optigen	\$95
				Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				Optigen	\$95
	Cobalamin Malabsorption/cubilin deficiency	Autosomal recessive	cubilin gene	Optigen	\$95
Boston Terrier	Hereditary Cataract 2 (HSF4)	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				VetGen	\$65.00 USD
				Optigen	\$100
	Cobalamin Malabsorption/cubilin deficiency	Autosomal recessive		Laboklin	3-5 weeks
Boxer	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD

<i>Breeds</i>	<i>Disease</i>	<i>Mono/poly, recessive/dominant</i>	<i>Mutation</i>	<i>Which lab.</i>	<i>Results Time/Price</i>
<i>Boykin Spaniel</i>	Collie Eye Anomalie (CEA)	Autosomal recessive		Laboklin	4-6 weeks
			chromosome number 37	Optigen	\$180
<i>Brazilian Terrier</i>	Exercise induced collapse (EIC)	Autosomal recessive	DNM1 gene	Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
<i>Briard</i>	Congenital stationary night blindness (CSNB)	Autosomal recessive	RPE65 gene	Laboklin	1-2 weeks
				Van Haeringen	<10 days € 39,50
<i>Braittany Spaniel</i>	C3 Deficiency	Autosomal recessive	RPE65 Gene	Optigen	\$135
				Van Haeringen	<10 days € 39,50
<i>Bull Dog</i>	Canine Multifocal Retinopathy (CMR1 & CMR2)	Autosomal recessive	BEST1 gene	VetGen	\$65.00 USD
	hyperuricosuria (HU)	Autosomal recessive		VetGen	\$65.00 USD
<i>Bull Mastiff</i>	Canine Multifocal Retinopathy (CMR1 & CMR2)	Autosomal recessive	BEST1 gene	VetGen	\$65.00 USD
				VMD2 Gene	Optigen
	Dominant Progressive Retinal Atrophy (PRA)	Autosomal dominant		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				Optigen	ong.2 weeks \$120
<i>Bull Terrier</i>	Polycystic kidney disease (PKD1)	Autosomal dominant		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
	Hemophilia B (Facort IX deficiency)	X-linked recessive		VetGen	\$65.00 USD
<i>Cairn Terrier</i>	Globoid Cell Leukodystrophy / Krabbes Disease	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
	Pyruvate kinase deficiency (PK)	Autosomal recessive		Laboklin	1-2 weeks
				VetGen	\$65.00 USD
<i>Cane Corsos</i>	Hemophilia B (Facort IX deficiency)	X-linked recessive		VetGen	\$65.00 USD
				van Haeringen	<10 days € 39,50
	CMR1 (Canine Multifocal Retinopathy)	Autosomal recessive			
			BEST1 gene	VetGen	\$65.00 USD
			VMD2 Gene	Optigen	US\$95.00

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price	
Cardigan Welsh Corgi	rcd3 Progressive Retinal Atrophy (rcd3 PRA)	Autosomal recessive		van Haeringen	<10 days € 39,50	
				VetGen	\$65.00 USD	
				PDE6A Gene	Optigen	\$80
Cavalier King Charles Spaniel	Dry eye curly coat syndrome (CCS)	Autosomal recessive		Laboklin	3-5 days	
				Van Haeringen	<10 days € 39,50	
	Episodic Falling (EF)	Autosomal recessive		Laboklin	3-5 days	
				Van Haeringen	<20 days € 59,50	
	Muscular dystrophy (MD)	X-chromosomal-recessive		Laboklin	1-2 weeks	
	Thrombocytopaenia	Autosomal recessive		Van Haeringen	<10 days € 39,50	
	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD	
Chesapeake Bay Retriever	Ectodermal dysplasia/Skin fragility syndrome (ED/SFS)	Autosomal recessive		Laboklin	1-2 weeks	
				DNM1 gene	Laboklin	3-5 days
	Exercise induced collapse (EIC)	Autosomal recessive		DNM1 gene	Van Haeringen	<20 days € 59,50
				prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive	
				prcd Gene	Optigen	\$195
Chihuahua	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD	
Chinese Crested Dog	Canine Multiple System Degeneration (CMSD)	Autosomal recessive		Laboklin	1-2 weeks	
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00	
				prcd Gene	Optigen	\$195
	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days	
				Van Haeringen	<10 days € 39,50	
			Carriers have a small chance of getting sick.	VetGen	\$65.00 USD	
			2-20% of carriers will develop condition	Optigen	\$90	
	Von-Willebrands Disease Type 2	Autosomal recessive		VetGen	\$65.00 USD	
	rcd3 Progressive Retinal Atrophy (rcd3 PRA)	Autosomal recessive	PDE6A Gene	Optigen	\$80	
	Chinese Foo Dog	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
Optigen					\$90	

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price			
Clumber Spaniel	Pyruvate Dehydrogenase Phosphatase 1 (PDP1)	Autosomal recessive		Van Haeringen	<10 days € 39,50			
				Laboklin	1-2 weeks			
				VetGen	\$65.00 USD			
Cockapoo	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive	prcd Gene	Van Haeringen	<25 days € 150,00			
				Optigen	\$195			
				Optigen	\$80			
English Cocker Spaniel	Familial Nephropathy (FN)	Autosomal recessive		Van Haeringen	<10 days € 110,00			
				prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive	prcd Gene	Optigen	\$195
				Phosphofruktokinase deficiency (PFKD)	Autosomal recessive		VetGen	\$65.00 USD
Collies	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	Optigen	\$80			
				VetGen	\$65.00 USD			
	Gray Collie Syndrome (Cyclic Neutropenia)	Autosomal recessive			Van Haeringen	<10 days € 39,50		
					Collie Eye Anomaly (CEA)	Autosomal recessive		Van Haeringen
	Ivermectin hypersensitivity (MDR1 gene defect)	Autosomal recessive		mdr1 gene	Laboklin	1-2 weeks		
					chromosome number 37	Optigen	\$180	
					Van Haeringen	<25 days € 187,50		
					Laboklin	1-2 weeks		
					Optigen	\$180		
					Von-Willebrands Disease Type 2	Autosomal recessive		VetGen

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price	
Coton de Tulear	Bandara's Neonatal Ataxia (BNAt)	Autosomal recessive	GRM1 gene	VetGen	\$65.00 USD	
				Van Haeringen	<10 days € 39,50	
	CMR2 (Canine Multifocal Retinopathy)	Autosomal recessive	BEST1 gene	van Haeringen	<10 days € 39,50	
				VetGen	\$65.00 USD	
				VMD2 Gene	Optigen	US\$95.00
	Von-Willebrands Disease Type 1	Autosomal dominant (variable penetrance)		Van Haeringen	<10 days € 89,00	
				Laboklin	3-5 days	
VetGen				Binnen 2 weeks \$65.00 USD		
Curly Coated Retrievers	Exercise induced collapse (EIC)	Autosomal recessive	DNM1 gene	Laboklin	3-5 days	
			DNM1 gene	Van Haeringen	<20 days € 59,50	
	Glycogen Storage Disease GSD Type IIIa (GSDIIIa)	Autosomal recessive	AGL Gene	Van Haeringen	<20 days € 44,50	
				Laboklin	1-2 weeks	
	Cone-Rod Dystrophy 1-PRA (Cord1-PRA)	Autosomal recessive		VetGen	\$65.00 USD	
Czechoslovakian Wolfdog	Pituitary dwarfism	Autosomal recessive		Van Haeringen	<20 days € 69,50	
				Laboklin	1-2 weeks	
Dachshund	Osteogenesis Imperfecta	Autosomal recessive		Laboklin	1-2 weeks	
				Van Haeringen	<10 days € 39,50	
		Cone Rod Dystrophy 4-PRA (CRD4-PRA)	Autosomal recessive		Van Haeringen	<10 days € 39,50
		Cone-Rod Dystrophy 1-PRA (Cord1-PRA)	Autosomal recessive		VetGen	\$65.00 USD
		Progressive retinal atrophy (crd-PRA)	Autosomal recessive	NPHP4 gene	Laboklin	1-2 weeks
		Mucopolysaccharidosis Type IIIa	Autosomal recessive		Van Haeringen	<10 days € 39,50
		Narcolepsy	Autosomal recessive		Van Haeringen	<20 days € 49,50
	Hcrtr2 Gene				Optigen	\$130
		Neuronal ceroid lipofuscinosis (NCL) 1/2	Autosomal recessive		Van Haeringen	<10 days € 39,50
					Laboklin	1-2 weeks
Dalmation	hyperuricosuria (HU)	Autosomal recessive		VetGen	\$65.00 USD	

<i>Breeds</i>	<i>Disease</i>	<i>Mono/poly, recessive/dominant</i>	<i>Mutation</i>	<i>Which lab.</i>	<i>Results Time/Price</i>	
<i>Doberman Pincher</i>	Albinism (White)	Autosomal recessive	OCA4 gene	VetGen	\$65.00 USD	
	Dilated Cardiomyopathy	Autosomal recessive		Van Haeringen	<10 days € 39,50	
				VetGen	\$65.00 USD	
	Narcolepsy	Autosomal recessive		Van Haeringen	<20 days € 49,50	
				Laboklin	1-2 weeks	
	Von-Willebrands Disease Type 1	Autosomal dominant (variable penetrance)		Hcrtr2 Gene	Optigen	\$130
				Van Haeringen	<10 days € 89,00	
				VetGen	Binnen 2 weeks \$65.00 USD	
Laboklin				3-5 days		
<i>Dogue de Bordeaux</i>	Canine Multi-focal Retinopathy (CMR)	Autosomal recessive	VMD2 Gene	Optigen	US\$95.00	
<i>Drentsche Patrijshond</i>	Von-Willebrands Disease Type 1	Autosomal dominant (variable penetrance)		Van Haeringen	<10 days € 89,00	
				VetGen	Binnen 2 weeks \$65.00 USD	
				Laboklin	3-5 days	
<i>Dutch Kooiker</i>	Von Willebrand disease 3 - 2	Autosomal recessive		Van Haeringen	<10 days € 39,50	
				Laboklin	1-2 weeks	
				VetGen	\$65.00 USD	
<i>Dwarf Poodle</i>	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00	
			prcd Gene	Optigen	\$195	
<i>English Bulldog</i>	Canine Multifocal Retinopathy (CMR1 & CMR2)	Autosomal recessive	BEST1 gene	VetGen	\$65.00 USD	
<i>English Cocker Spaniel</i>	Familial Nephropathy (FN)	Autosomal recessive		Van Haeringen	<10 days € 110,00	
				Laboklin	1-2 weeks	
				Optigen	\$95	
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		prcd Gene	Optigen	\$195
				Van Haeringen	<25 days € 150,00	
			prcd Gene	Optigen	\$195	

<i>Breeds</i>	<i>Disease</i>	<i>Mono/poly, recessive/dominant</i>	<i>Mutation</i>	<i>Which lab.</i>	<i>Results Time/Price</i>
<i>(sequel) English Cocker Spaniel</i>	Phosphofructokinase deficiency (PFKD)	Autosomal recessive		VetGen	\$65.00 USD
				Optigen	\$80
<i>English Mastiff</i>	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD
	Dominant Progressive Retinal Atrophy (PRA)	Autosomal dominant		Laboklin	1-2 weeks
<i>English Setter</i>	Canine Multifocal Retinopathy (CMR1 & CMR2)	Autosomal recessive	BEST1 gene	VetGen	\$65.00 USD
	Neuronal ceroid lipofuscinosis (NCL) 8	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				VetGen	\$65.00 USD
rcd4 Progressive Retinal Atrophy (rcd4 PRA)	Autosomal recessive		Laboklin	1-2 weeks	
<i>English Shepherd</i>	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive	C2orf71 Gene	Optigen	\$95
			prcd Gene	Optigen	\$195
<i>English Springer Spaniel</i>	Fucosidosis	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	102 weeks
	Familial Nephropathy (FN)	Autosomal recessive		Laboklin	1-2 weeks
	Phosphofructokinase deficiency (PFKD)	Autosomal recessive		Laboklin	1-2 weeks
				VetGen	\$65.00 USD
				Optigen	\$80
	Cone-Rod Dystrophy 1-PRA (Cord1-PRA)	Autosomal recessive		VetGen	\$65.00 USD
Tremor, X-linked	X-Chromosomal		Van Haeringen	<10 days € 39,50	
<i>English Toy Spaniel</i>	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD
<i>Entlebucher Mountain Dog</i>	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
				Optigen	\$195
<i>Farm Collie</i>	rcd2 Progressive Retinal Atrophy (rcd2 PRA)	Autosomal recessive		Van Haeringen	<25 days € 187,50
<i>Finnish Hound</i>	Cerebellar Ataxia, progressive early-onset	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
	Glycogen storage disease type II (Pompe Disease)	Autosomal recessive	SEL1L gene	Laboklin	1-2 weeks

<i>Breeds</i>	<i>Disease</i>	<i>Mono/poly, recessive/dominant</i>	<i>Mutation</i>	<i>Which lab.</i>	<i>Results Time/Price</i>		
<i>Finnish Lapphund</i>	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00		
			prcd Gene	Optigen	\$195		
<i>French Bulldog</i>	Hereditary cataract (HC)	Autosomal recessive	HSF4 gene	Laboklin	1-2 weeks		
			HSF4 Gene	VetGen	\$65.00 USD		
			HSF4-1 Gene	Optigen	\$100		
<i>Frisian Water Dogs</i>	SCID 2	Autosomal recessive		Van Haeringen	<10 days € 39,50		
<i>Fox Terrier</i>	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days		
<i>German Pinscher</i>	Von-Willebrands Disease Type 1	Autosomal dominant (variable penetrance)		Van Haeringen	<10 days € 89,00		
				VetGen	Binnen 2 weeks \$65.00 USD		
				Laboklin	3-5 days		
<i>German Pointer</i>	Junctional epidermolysis bullosa (JEB)	Autosomal recessive		Laboklin	1-2 weeks		
	Von-Willebrands Disease Type 2	Autosomal recessive		Van Haeringen	<10 days € 69,00		
				VetGen	\$65.00 USD		
				Laboklin	3-5 days		
	Hemophilia B (Facort IX deficiency)	X-linked recessive		VetGen	\$65.00 USD		
<i>German Shepherd</i>	Canine Leukocyte Adhesion Deficiency (CLAD), Type 3	Autosomal recessive		Van Haeringen	<10 days € 39,50		
	hyperuricosuria (HU)	Autosomal recessive		VetGen	\$65.00 USD		
	Mucopolysaccharidosis Type VII	Autosomal recessive		Van Haeringen	<10 days € 39,50		
				Laboklin	1-2 weeks		
			Ivermectin hypersensitivity (MDR1 gene defect)	Autosomal recessive	mdr1 gene	Laboklin	1-2 weeks
	Pituitary dwarfism	Autosomal recessive			Van Haeringen	<20 days € 69,50	
					Laboklin	1-2 weeks	
			Renal Cystadenocarcinoma and Nodular Dermatofibrosis	Autosomal dominant		VetGen	\$65.00 USD
					Van Haeringen	<10 days € 39,50	
	<i>German Shorthaired Pointer</i>	Cone Degeneration	Autosomal recessive		Van Haeringen	<10 days € 39,50	
CNGB3 Gene				Optigen	\$160		

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price	
German Spaniel	Phosphofruktokinase deficiency (PFKD)	Autosomal recessive		Laboklin	1-2 weeks	
German Spitz	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive	prcd Gene	Optigen	\$195	
German Wirehaired Pointer	Hemophilia B (Facort IX deficiency)	X-linked recessive		VetGen	\$65.00 USD	
	Von-Willebrands Disease Type 2	Autosomal recessive		VetGen	\$65.00 USD	
Giant Schnauzer	Factor VII - Deficiency	Autosomal recessive		Laboklin	3-5 days	
				VetGen	\$65.00 USD	
	hyperuricosuria (HU)	Autosomal recessive		VetGen	\$65.00 USD	
	Neuroaxonal dystrophy (NAD)	Autosomal recessive		Van Haeringen	<10 days € 39,50	
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00	
				prcd Gene	Optigen	\$195
				ADAM9 Gene	Optigen	\$120
Glen of Imaal Terrier	Cone Rod Dystrophy 3 (CRD3)	Autosomal recessive		Van Haeringen	<25 days € 100,00	
				ADAM9 Gene	Optigen	\$120
Golden Retriever	Epidermolysis bullosa, dystrophic (RDEB)	Autosomal recessive		Van Haeringen	<10 days € 39,50	
	GR PRA1 (Progressive Retinal Atrophy)	Autosomal recessive		Van Haeringen	<10 days € 39,50	
				Laboklin	1-2 weeks	
				Optigen	\$100	
	GR PRA2 (Progressive Retinal Atrophy)	Autosomal recessive		Optigen	\$100	
	Ichthyosis 2	Autosomal recessive		Van Haeringen	<10 days € 39,50	
				Optigen	\$120	
	Muscular Dystrophy (GRMD)	X-Chromosomal		Van Haeringen	<10 days € 39,50	
				Laboklin	1-2 weeks	
		prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
			prcd Gene	Optigen	\$195	
Gordon Setter	Cerebellar Ataxia 2	Autosomal recessive		Van Haeringen	<10 days € 39,50	
	rcd4 Progressive Retinal Atrophy (rcd4 PRA)	Autosomal recessive		van Haeringen	<10 days € 39,50	
				Laboklin	1-2 weeks	
				C2orf71 Gene	Optigen	\$95

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price
Great Dane	Centronuclear Myopathy (cnm)	Autosomal recessive		Laboklin	3-5 days
Great Pyrenees	CMR1 (Canine Multifocal Retinopathy)	Autosomal recessive		van Haeringen	<10 days € 39,50
			VMD2 Gene	Optigen	US\$95.00
Great Swiss Mountain Dog	Bleeding disorder due to P2RY12 defect	Mono, autosomaal recessief	P2RY12 Gene	Van Haeringen	<10 days € 39,50
Greyhound	Hereditary polyneuropathy (HN)	Autosomal recessive		Laboklin	1-2 weeks
				Van Haeringen	<10 days € 39,50
				VetGen	\$65.00 USD
			NDRG1 gene	Optigen	\$95
Havanese	Haemophilia A (Factor VIII)	X-Chromosomal		Van Haeringen	<20 days € 49,50
				Laboklin	2-5 days
	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD
Hokkaido	Collie Eye Anomalie (CEA)	Autosomal recessive		Laboklin	4-6 weeks
				Van Haeringen	<25 days € 140,00
			chromosome number 37	Optigen	\$180
Husky	GM1-Gangliosidosis	Autosomal recessive		Laboklin	1-2 weeks
Irish Setters	Canine Leukocyte Adhesion Deficiency (CLAD), Type 1	Autosomal recessive		Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
				Optigen	\$135
	Globoid cell leukodystrophy (Krabbe disease)	Autosomal recessive		Laboklin	1-2 weeks
	Neuronal ceroid lipofuscinosis (NCL) 8	Autosomal recessive		Van Haeringen	<10 days € 39,50
	rcd1 Progressive Retinal Atrophy (rcd1 PRA)	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				Optigen	\$120
			PDEB gene	VetGen	\$65.00 USD
	rcd4 Progressive Retinal Atrophy (rcd4 PRA)	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
			C2orf71 Gene	Optigen	\$95

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price	
Irish Terrier	Digital Hyperkeratosis (Corny Feet)	Autosomal recessive		Laboklin	1-2 weeks	
				Van Haeringen	<10 days € 39,50	
Irish Wolfhound	Startle Disease or Hyperekplexia	Autosomal recessive		Laboklin	1-2 weeks	
Italian Greyhound	IG PRA1 (Progressive Retinal Atrophy)	Autosomal Dominant with Incomplete Penetrance		Optigen	\$105	
Jack Russell Terrier	Late onset ataxia (LOA)	Autosomal recessive	(CAPN1)-gene	Laboklin	1-2 weeks	
	Primary Lens Luxation (PLL)	Autosomal recessive		Van Haeringen	<10 days € 39,50	
				Laboklin	3-5 days	
			2-20% of carriers will develop condition		Optigen	\$90
	SCID	Autosomal recessive		Van Haeringen	<10 days € 39,50	
	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD	
	Spinocerebellar ataxia (SCA)	Autosomal recessive		Laboklin	3-5 days	
Jagd Terrier	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days	
				Van Haeringen	<10 days € 39,50	
			Carriers have a small chance of getting sick.		VetGen	\$65.00 USD
			2-20% of carriers will develop condition		Optigen	\$90
Japanese chin/spitz						
Karerlian Beardog	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00	
				Optigen	\$195	
Kerry Blue Terrier	Canine Multiple System Degeneration (CMSD)	Autosomal recessive		Laboklin	1-2 weeks	
	Von-Willebrands Disease Type 1	Autosomal dominant (variable penetrance)		Van Haeringen	<10 days € 89,00	
				VetGen	Binnen 2 weeks \$65.00 USD	
					Laboklin	3-5 days
Kelpie	Cerebellar Abiotrophy	Autosomal recessive		Van Haeringen	<10 days € 39,50	
Kromfohländer	Digital Hyperkeratosis (Corny Feet)	Autosomal recessive		Laboklin	1-2 weeks	
				Van Haeringen	<10 days € 39,50	
Kuvasz	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00	
				Optigen	\$195	

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price
Labrador Retriever	Centronuclear Myopathy (CNM or HMLR)	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	3-5 days
	Cystinuria	Autosomal recessive		Laboklin	1-2 weeks
				Van Haeringen	<10 days € 39,50
	Exercise induced collapse (EIC)	Autosomal recessive	DNM1 gene	Laboklin	3-5 days
			DNM1 gene	Van Haeringen	<20 days € 59,50
	Hereditary Nasal Parakeratosis (HNPK)	Autosomal recessive		Van Haeringen	<20 days € 91,50
				Laboklin	3-5 days
			SUV39H2 gene	Optigen	\$120
	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD
	Myotubular myopathy (MTM)	X-Chromosomal		Van Haeringen	<10 days € 39,50
	Narcolepsy	Autosomal recessive		Van Haeringen	<20 days € 49,50
				Laboklin	1-2 weeks
			Hctr2 Gene	Optigen	\$130
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
			prcd Gene	Optigen	\$195
	Pyruvate kinase Deficiency (PKDef)	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				VetGen	\$65.00 USD
	Retinal Dysplasia Retinal Folds+OculoSkeletal Dysplasia (RD+OSD) 1	Autosomal dominant (incomplete penetrance)		Van Haeringen	<10 days € 39,50
			Laboklin	4-6 weeks	
			Optigen	\$160	
Skeletal Dysplasia 2 (SD2)	Autosomal recessive		Van Haeringen	<10 days € 39,50	
			Laboklin	1-2 weeks	
Achromatopsia Type 1/Day Blindness	Autosomal recessive		Optigen	\$100	

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price
Lagotto Romagnolo	Juvenile epilepsy	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	3-5 werkdays
				Optigen	\$95
Lakeland Terrier	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
				Optigen	\$90
Lancashire Heeler	Collie Eye Anomalie (CEA)	Autosomal recessive		Laboklin	4-6 weeks
				Van Haeringen	<25 days € 140,00
				Optigen	\$180
				Van Haeringen	<10 days € 39,50
				Laboklin	3-5 days
				VetGen	\$65.00 USD
Landseer	Cystinuria	Autosomal recessive		Optigen	\$90
				Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
				Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				VetGen	\$65.00 USD
Lapponian Herder	Glycogen storage disease type II (Pompe Disease)	Autosomal recessive		Laboklin	1-2 weeks
				Van Haeringen	<25 days € 150,00
				Optigen	\$195
				Optigen	US\$95.00
Large munsterlander	hyperuricosuria (HU)	Autosomal recessive		VetGen	\$65.00 USD
				VetGen	\$65.00 USD
Leonberger	Leonberger Polyneuropathy 1 (LPN1)	Autosomal recessive		Laboklin	1-2 weeks
Lhasa Apso	Hemophilia B (Facort IX deficiency)	X-linked recessive		VetGen	\$65.00 USD
Llewelin Setter	rcd4 Progressive Retinal Atrophy (rcd4 PRA)	Autosomal recessive	C2orf71 Gene	Optigen	\$95
Longhaired Whippet	Ivermectin hypersensitivity (MDR1 gene defect)	Autosomal recessive	mdr1 gene	Laboklin	1-2 weeks
				Optigen	\$180
	Collie Eye Anomaly (CEA)	Autosomal recessive	chromosome number 37	Optigen	\$180

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price	
Lucas Terrier	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days	
		2-20% of carriers will develop condition		Optigen	\$90	
Maltese	Glycogen Storage Disease Type I (GSD I)	Autosomal recessive		Van Haeringen	<10 days € 39,50	
	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD	
Maltipoo	prcd Progressice Retinal Atrophy (prcd PRA)	Autosomal recessive	prcd Gene	Optigen	\$195	
Manchester Terrier	Von-Willebrands Disease Type 1	Autosomal dominant (variable penetrance)		Van Haeringen	<10 days € 89,00	
				VetGen	Binnen 2 weeks \$65.00 USD	
				Laboklin	3-5 days	
Markiesje	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00	
			prcd Gene	Optigen	\$195	
Mastiffs	CMR1 (Canine Multifocal Retinopathy)	Autosomal recessive		Van Haeringen	<10 days € 39,50	
			VMD2 Gene	Optigen	US\$95.00	
	Dominant Progressive Retinal Atrophy (PRA)	Autosomal dominant		Van Haeringen	<10 days € 39,50	
				Optigen	ong.2 weeks \$120	
McNab	Ivermectin hypersensitivity (MDR1 gene defect)	Autosomal recessive		Laboklin	1-2 weeks	
			mdr1 gene			
Miniature American Shepherd	CMR1 (Canine Multifocal Retinopathy)	Autosomal recessive		Van Haeringen	<10 days € 39,50	
			VMD2 Gene	Optigen	US\$95.00	
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00	
				prcd Gene	Optigen	\$195
	Cone Degeneration (CD)	Autosomal recessive		CNGB3 Gene	Optigen	\$160
	Collie Eye Anomaly (CEA)	Autosomal recessive		chromosome number 37	Optigen	\$180

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price
Miniature Australian Shepherd	Canine Multi-focal Retinopathy (CMR)	Autosomal recessive	VMD2 Gene	Optigen	US\$95.00
	Cone Degeneration (CD)	Autosomal recessive	CNGB3 Gene	Optigen	\$160
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive	prcd Gene	Optigen	\$195
	Collie Eye Anomaly (CEA)	Autosomal recessive	chromosome number 37	Optigen	\$180
	Hereditary Cataract (HD)	Autosomal co-dominant	HSF4-2 Gene	Optigen	\$100
Miniature Bull Terrier	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
		Carriers have a small chance of getting sick.		VetGen	\$65.00 USD
		2-20% of carriers will develop condition		Optigen	\$90
Miniature Pinscher	Cystinuria	Autosomal dominant		Laboklin	1-2 weeks
				Van Haeringen	<10 days € 39,50
Miniature Poodle	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
			prcd Gene	Optigen	\$195
	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD
Miniature Schnauzer	Myotonia Congenita	Autosomal recessive		Van Haeringen	<20 days € 49,50
				Laboklin	3-5 days
	Type A Progressive Retinal Atrophy (Type A PRA)	Autosomal recessive		Van Haeringen	<25 days € 132,50
				Optigen	\$160
	Persistent Muellerian Duct Syndrome (PMDS)	sex-limited autosomal recessive trait	MISR11 Gene	Optigen	\$95
Moyen Poodle	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
Newfoundland	Cystinuria	Autosomal recessive		Laboklin	3-5 days
			Gene SLC3A1	Van Haeringen	<10 days € 39,50
				VetGen	\$65.00 USD
				Optigen	\$80
Norfolk Terrier	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
Norwegian Elkhound	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
			prcd Gene	Optigen	\$195

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price		
Norwich Terrier	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days		
			2-20% of carriers will develop condition	Optigen	\$90		
Nova Scotia Duck Tolling Retriever	Collie Eye Anomalie (CEA)	Autosomal recessive		Laboklin	4-6 weeks		
				Van Haeringen	<25 days € 140,00		
				chromosome number 37	Optigen	\$180	
		prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00	
				prcd Gene	Optigen	\$195	
Old Danish Pointer	Congenital Myasthenic Syndrome	Autosomal recessive		Van Haeringen	<10 days € 39,50		
Old English Sheepdog (Bobtail)	Cerebellar Ataxia 2	Autosomal recessive		Van Haeringen	<10 days € 39,50		
		Exercise induced collapse (EIC)	Autosomal recessive	DNM1 gene	Laboklin	3-5 days	
		Ivermectin hypersensitivity (MDR1 gene defect)	Autosomal recessive	mdr1 gene	Laboklin	1-2 weeks	
		Primary ciliary Dyskinesia (PCD)	Autosomal recessive		Laboklin	1-2 weeks	
					Van Haeringen	<10 days € 39,50	
Otterhound	Collie Eye Anomalie (CEA)	Autosomal recessive		Van Haeringen	<25 days € 140,00		
		Thrombasthenia	Autosomal recessive	Van Haeringen	<10 days € 39,50		
		Papillion	Pap-Progressive Retinal Atrophy 1 (Pap-PRA1)	Autosomal recessive		Van Haeringen	<25 days € 100,00
						Laboklin	1-2 weeks
						Optigen	\$90
	Cone-Rod Dystrophy 1-PRA (Cord1-PRA)	Autosomal recessive		VetGen	\$65.00 USD		
Parson Russell Terrier	Von-Willebrands Disease Type 1	Autosomal dominant (variable penetrance)		Van Haeringen	<10 days € 89,00		
				VetGen	Binnen 2 weeks \$65.00 USD		
				Laboklin	3-5 days		
		Late onset ataxia (LOA)	Autosomal recessive	CAPN1 -gene	Laboklin	1-2 weeks	
		hyperuricosuria (HU)	Autosomal recessive		VetGen	\$65.00 USD	

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price
<i>(sequel) Parson Russell Terrier</i>	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
				VetGen	\$65.00 USD
				Optigen	\$90
<i>Patterdale Terrier</i>	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
				VetGen	\$65.00 USD
				Optigen	\$90
<i>Pembroke Welsh Corgi</i>	Exercise induced collapse (EIC)	Autosomal recessive	DNM1 gene	Laboklin	3-5 days
				Van Haeringen	<10 days € 89,00
				VetGen	Binnen 2 weeks \$65.00 USD
				Laboklin	3-5 days
<i>Perro de Presa Canarios</i>	CMR1 (Canine Multifocal Retinopathy)	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Optigen	US\$95.00
				Laboklin	1-2 weeks
				Optigen	\$90
<i>Phalene</i>	Pap-Progressive Retinal Atrophy 1 (Pap-PRA1)	Autosomal recessive		Van Haeringen	<25 days € 100,00
				Laboklin	1-2 weeks
				Optigen	\$90
				Optigen	\$90
<i>Pitbull Terrier</i>	Cone Rod Dystrophy 2 (CRD2)	Autosomal recessive		Van Haeringen	<25 days € 100,00
				VetGen	\$65.00 USD
				Laboklin	1-2 weeks
				Optigen	\$95
<i>Polish Lowland Sheepdog</i>	rcd4 Progressive Retinal Atrophy (rcd4 PRA)	Autosomal recessive		Laboklin	1-2 weeks
				Optigen	\$95
				Van Haeringen	<10 days € 39,50
				Optigen	\$95
<i>Pomeranian</i>	Vitamin D-deficiency rickets, type II	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				VetGen	\$65.00 USD
				Optigen	\$95
<i>Poodle</i>	Neonatal Encephalopathy	Autosomal recessive	ATF2 Gene	Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				VetGen	\$65.00 USD
				Optigen	\$95

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price
<i>(sequel) Poodle</i>	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD
	Von-Willebrands Disease Type 1	Autosomal dominant (variable penetrance)		Van Haeringen	<10 days € 89,00
				VetGen	Binnen 2 weeks \$65.00 USD
				Laboklin	3-5 days
<i>Portuguese Water Dog</i>	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive	prcd Gene	Optigen	\$195
	GM1-Gangliosidosis	Autosomal recessive		Laboklin	1-2 weeks
				Optigen	\$120
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
<i>Pug</i>			prcd Gene	Optigen	\$195
	Necrotizing Meningoencephalitis (NME)	Autosomal recessive (with variable penetrance)		Laboklin	1-2 weeks
	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
	Pyruvate kinase Deficiency 2 (PKDef)	Autosomal recessive		Van Haeringen	<10 days € 39,50
<i>Rat Terrier</i>				Laboklin	1-2 weeks
	Congenital Hypothyroidism (CHG) 3	Autosomal recessive		Van Haeringen	<10 days € 39,50
	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
<i>Rhodesian Ridgeback</i>				VetGen	\$65.00 USD
		Carriers have a small chance of getting sick.		VetGen	\$65.00 USD
		2-20% of carriers will develop condition		Optigen	\$90
	Haemophilia B (factor IX deficiency)	X-chromosomal-recessive		Laboklin	3-5 days
<i>Rough Collie</i>				VetGen	\$65.00 USD
	rcd2 Progressive Retinal Atrophy (rcd2 PRA)	Autosomal recessive		Van Haeringen	<25 days € 187,50
				Optigen	\$180
<i>Saarloos Wolfdog</i>	Collie Eye Anomaly (CEA)	Autosomal recessive	chromosome number 37	Optigen	\$180
	Pituitary dwarfism	Autosomal recessive		Van Haeringen	<20 days € 69,50
				Laboklin	1-2 weeks

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price	
Samoyed	Familial Nephropathy (FN)	X-chromosomal-recessive		Laboklin	1-2 weeks	
				VetGen	\$65.00 USD	
	Retinal Dysplasia Retinal Folds+OculoSkeletal Dysplasia (RD+OSD) 2	Autosomal recessive		Van Haeringen	<25 days € 132,50	
				Laboklin	4-6 weeks	
	X Linked Progressive Retinal Atrophy 1 (XL PRA1)	X-Chromosomal		Optigen	\$160	
				Van Haeringen	<10 days € 39,50	
			Optigen	\$150		
Schapendoes	gPRA (Progressive Retinal Atrophy)	Autosomal recessive		Van Haeringen	<10 days € 39,50	
				Laboklin	1-2 weeks	
Schipperke	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00	
				prcd Gene	Optigen	\$195
Scottish Deerhound	Faktor VII - Deficiency	Autosomal recessive		Laboklin	3-5 days	
				VetGen	\$65.00 USD	
Scottish Terrier	Von-Willebrands Disease Type 3	Autosomal recessive		Van Haeringen	<10 days € 49,50	
				VetGen	\$65.00 USD	
				Laboklin	1-2 weeks	
Sealyham Terrier	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days	
				Van Haeringen	<10 days € 39,50	
				Carriers have a small chance of getting sick.	VetGen	\$65.00 USD
				2-20% of carriers will develop condition	Optigen	\$90
Shetland Sheepdog	Collie Eye Anomalie (CEA)	Autosomal recessive		Laboklin	4-6 weeks	
				Van Haeringen	<25 days € 140,00	
	Ivermectin hypersensitivity (MDR1 gene defect)	Autosomal recessive		chromosome number 37	Optigen	\$180
				mdr1 gene	Laboklin	1-2 weeks
				Von-Willebrands Disease Type 3	Autosomal recessive	Laboklin
			VetGen	\$65.00 USD		

<i>Breeds</i>	<i>Disease</i>	<i>Mono/poly, recessive/dominant</i>	<i>Mutation</i>	<i>Which lab.</i>	<i>Results Time/Price</i>
<i>Shiba Inu</i>	GM1 Gangliosidosis	Autosomal recessive		Van Haeringen	<10 days € 39,50
<i>Shih Tzu</i>	Prekallikrein deficiency	Autosomal recessive		Van Haeringen	<10 days € 39,50
	Macrothrombocytopenia (MTC)	autosomal dominant	beta-1 tubulin gene	VetGen	\$65.00 USD
<i>Siberian Husky</i>	X Linked Progressive Retinal Atrophy 1 (XL PRA1)	X-Chromosomal		Van Haeringen	<10 days € 39,50
				Optigen	\$150
<i>Silken Windhound</i>	Ivermectin hypersensitivity (MDR1 gene defect)	Autosomal recessive	mdr1 gene	Laboklin	1-2 weeks
	Collie Eye Anomaly (CEA)	Autosomal recessive	chromosome number 37	Optigen	\$180
<i>Silky Terrier</i>	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive	prcd Gene	Optigen	\$195
<i>Sloughi</i>	rcd1a Progressive Retinal Atrophy (rcd1a PRA)	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
			PDE6B gene	Optigen	\$80
<i>Small Munsterlander</i>	rcd4 Progressive Retinal Atrophy (rcd4 PRA)	Autosomal recessive		Laboklin	1-2 weeks
			C2orf71 Gene	Optigen	\$95
<i>Smooth Collies</i>	rcd2 Progressive Retinal Atrophy (rcd2 PRA)	Autosomal recessive		Van Haeringen	<25 days € 187,50
				Optigen	\$180
	Collie Eye Anomaly (CEA)	Autosomal recessive	chromosome number 37	Optigen	\$180
<i>Soft-Coated Wheaten Terrier</i>	Protein losing nephropathy (PLN)	Autosomal recessive		Laboklin	3-5 days
<i>Spaniel breeds</i>	Phosphofructokinase deficiency (PFK)	Autosomal recessive		Van Haeringen	<10 days € 39,50
<i>Spanish Water Dog</i>	Congenital Hypothyreosis (CHG)	Autosomal recessive		Laboklin	1-2 weeks
				Van Haeringen	<10 days € 39,50
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
			prcd Gene	Optigen	\$195
<i>Stabijhoun</i>	Von-Willebrands Disease Type 1	Autosomal dominant (variable penetrance)		Van Haeringen	<10 days € 89,00
				VetGen	Binnen 2 weeks \$65.00 USD
				Laboklin	3-5 days

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price
Staffordshire Bull Terrier	Hereditary Cataract 2 (HSF4)	Autosomal recessive	HSF4 Gene	Van Haeringen	<10 days € 39,50
			HSF4 Gene	VetGen	\$65.00 USD
			HSF4 gene	Laboklin	1-2 weeks
			HSF4-1 Gene	Optigen	\$100
	L2-Hydroxyglutaric aciduria (L2-HGA)	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	3-5 days
Sussex Spaniel	Pyruvate Dehydrogenase Phosphatase 1 (PDP1)	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
				VetGen	\$65.00 USD
Swedish Lapphund	Glycogen storage disease type II (Pompe Disease)	Autosomal recessive		Laboklin	1-2 weeks
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
			prcd Gene	Optigen	\$195
Teddy Roosevelt Terrier	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
			2-20% of carriers will develop condition	Optigen	\$90
Tenterfield Terrier	Congenital Hypothyroidism (CHG) 2	Autosomal recessive		Van Haeringen	<10 days € 39,50
	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
		Carriers have a small chance of getting sick.	VetGen	\$65.00 USD	
		2-20% of carriers will develop condition	Optigen	\$90	
Tibetan Terrier	Neuronal ceroid lipofuscinosis (NCL)	Autosomal recessive		Laboklin	1-2 weeks
	Primary Lens Luxation (PLL)	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	3-5 days
		Carriers have a small chance of getting sick.	VetGen	\$65.00 USD	
		2-20% of carriers will develop condition	Optigen	\$90	
	rcd4 Progressive Retinal Atrophy (rcd4 PRA)	Autosomal recessive		Laboklin	1-2 weeks
			C2orf71 Gene	Optigen	\$95

Breeds	Disease	Mono/poly, recessive/dominant	Mutation	Which lab.	Results Time/Price
Toy Fox Terrier	Congenital Hypothyroidism (CHG) 3	Autosomal recessive		Van Haeringen	<10 days € 39,50
	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
		Carriers have a small chance of getting sick.		VetGen	\$65.00 USD
		2-20% of carriers will develop condition		Optigen	\$90
Toy Poodle	Gangliosidosis, GM2, type II	Autosomal recessive		Van Haeringen	<10 days € 39,50
	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
			prcd Gene	Optigen	\$195
Volpino Italiano	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
		Carriers have a small chance of getting sick.		VetGen	\$65.00 USD
		2-20% of carriers will develop condition		Optigen	\$90
Wäller	Hereditary cataract (HC)	Autosomal dominant	HSF4 gene	Laboklin	1-2 weeks
Weimaraner	Hypomyelination (Shaking Puppy Syndrome)	Autosomal recessive		Laboklin	1-2 weeks
Welsh Corgi	Muscular Dystrophy, Duchenne type (MDM)	X-Chromosomal		Van Haeringen	<10 days € 39,50
	rcd3 Progressive Retinal Atrophy (rcd3 PRA)	Autosomal recessive		Laboklin	1-2 weeks
	X-linked severe combined Immunodeficiency (X-SCID)	X-Chromosomal		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
Welsh Terrier	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
		Carriers have a small chance of getting sick.		VetGen	\$65.00 USD
		2-20% of carriers will develop condition		Optigen	\$90
Westhighland White Terrier	Globoid Cell Leukodystrophy / Krabbes Disease	Autosomal recessive		Van Haeringen	<10 days € 39,50
				Laboklin	1-2 weeks
	Pyruvate kinase deficiency (PK)	Autosomal recessive		Laboklin	1-2 weeks
				VetGen	\$65.00 USD
Westphalia Terrier	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days

<i>Breeds</i>	<i>Disease</i>	<i>Mono/poly, recessive/dominant</i>	<i>Mutation</i>	<i>Which lab.</i>	<i>Results Time/Price</i>
<i>Whippet</i>	Collie Eye Anomalie (CEA)	Autosomal recessive		Laboklin	4-6 weeks
				Optigen	\$180
	Muscular Hypertrophy	Autosomal dominant		Van Haeringen	<10 days € 39,50
		Autosomal recessive		Laboklin	1-2 weeks
	Phosphofruktokinase deficiency (PFKD)	Autosomal recessive		Laboklin	1-2 weeks
			VetGen	\$65.00 USD	
<i>White Shepherd</i>	Ivermectin hypersensitivity (MDR1 gene defect)	Autosomal recessive	mdr1 gene	Laboklin	1-2 weeks
<i>Whire-haired Fox Terrier</i>	Primary lens luxation (PLL)	Autosomal recessive		Van Haeringen	<10 days € 39,50
		Carriers have a small chance of getting sick.		VetGen	\$65.00 USD
		2-20% of carriers will develop condition		Optigen	\$90
<i>Wire-haired Pointer</i>	Exercise induced collapse (EIC)	Autosomal recessive	DNM1 gene	Laboklin	3-5 days
<i>Wolfdog</i>	Dilated Cardiomyopathy	Autosomal recessive		Van Haeringen	<10 days € 39,50
<i>Yorkshire Terrier</i>	prcd Progressive Retinal Atrophy (prcd PRA)	Autosomal recessive		Van Haeringen	<25 days € 150,00
			prcd Gene	Optigen	\$195
	Primary lens luxation (PLL)	Autosomal recessive		Laboklin	3-5 days
				Van Haeringen	<10 days € 39,50
		Carriers have a small chance of getting sick.		VetGen	\$65.00 USD
		2-20% of carriers will develop condition		Optigen	\$90
		L2-Hydroxyglutaric Aciduria	Autosomal recessive		VetGen

Appendix 2

Table 2 Cardiovascular and blood diseases

<i>Diseases</i>	<i>Breeds</i>
<i>Bleeding disorder due to P2RY12 defect</i>	Great Swiss Mountain Dog
<i>Cyclic Neutropenia (CN) (Gray Collie Syndrome)</i>	Australian Shepherd Collies
<i>Dilated Cardiomyopathy</i>	Doberman Pincher, Wolfdog
<i>Factor VII - Deficiency</i>	Giant Schnauzer Beagle Airedale Terrier
	Alaskan Klee Kai Scottish Deerhound
<i>Haemophilia A (Factor VIII)</i>	Havanese
<i>Haemophilia B (Factor IX deficiency)</i>	Rhodesian Ridgeback Lhasa Apso German Wirehaired Pointer
	Airedale Terrier Bull Terrier Cairn Terrier
	German Pointer
<i>Macrothrombocytopenia (MTC)</i>	Boxer Labrador Retriever Bichon Frise
	Cavalier King Charles Spaniel Shih Tzu Poodle
	Chihuahua Parson Russell Terrier Miniature Poodle
	English Cocker Spaniel Mastiffs Maltese
	English Toy Spaniel Jack Russell Terrier Havanese
<i>Polycythemia</i>	All Breeds
<i>Thrombasthenia</i>	Otterhound
<i>Thrombasthenia 2</i>	All Breeds
<i>Thrombocytopaenia</i>	Cavalier King Charles Spaniel
<i>Thrombopathia</i>	Basset Landseer American Eskimo Dog
<i>Von-Willebrands Disease Type 1</i>	Bernese Mountain Dog Stabijhoun Poodle
	Coton de Tulear Pembroke Welsh Corgi Papillion
	Doberman Pincher Manchester Terrier Kerry Blue Terrier
	Drentsche Pattrijshond German Pinscher
<i>Von-Willebrands Disease Type 2</i>	Chinese Crested Dog German Wirehaired Pointer German Pointer
	Collies
<i>Von-Willebrands Disease Type 3</i>	Dutch Kooiker Scottish Terrier Shetland Sheepdog

Table 3 Metabolic and Immune problems

<i>Diseases</i>	<i>Breeds</i>
<i>C3 Deficiency</i>	Brittany Spaniel
<i>Canine Leukocyte Adhesion Deficiency (CLAD), Type 1</i>	Irish Setters
<i>Canine Leukocyte Adhesion Deficiency (CLAD), Type 3</i>	German Shepherd
<i>Cobalamin Malabsorption/cubilin deficiency</i>	Beagle Border Collie Boston Terrier
<i>Congenital Hypothyreosis (CHG)</i>	Spanish Water Dog
<i>Congenital Hypothyreosis (CHG) 2</i>	Tenterfield Terrier Rat Terrier
<i>Congenital Hypothyreosis (CHG) 3</i>	Toy Fox Terrier
<i>Copper toxicosis</i>	Bedlington Terriers
<i>Fucosidosis</i>	English Springer Spaniel
<i>Glycogen Storage Disease GSD Type IIIa (GSDIIIa)</i>	Curly Coated Retrievers

<i>Glycogen Storage Disease Type I (GSD I)</i>	Maltese		
<i>Ivermectin hypersensitivity (MDR1 gene defect)</i>	Australian Shepherd	White Shepherd	Silken Windhound
	Bobtail	Shetland Sheepdog	McNab
	Border Collie	Longhaired Whippet	German Shepherd
	Collies	All Breeds	
<i>Mucopolysaccharidose Type VII - 2</i>	Brazilian Terrier		
<i>Mucopolysaccharidosis Type IIIa</i>	Dachshund		
<i>Mucopolysaccharidosis Type VII</i>	German Shepherd		
<i>Phosphofructokinase deficiency (PFKD)</i>	Spaniel breeds	Whippet	German Spaniel
	American Cocker Spaniel	English Springer Spaniel	English Cocker Spaniel
	Cockapoo		
<i>Prekallikrein deficiency</i>	Shih Tzu		
<i>Pyruvate Dehydrogenase Phosphatase 1 (PDP1)</i>	Clumber Spaniel	Sussex Spaniel	
<i>Pyruvate kinase Deficiency (PKDef)</i>	Basenjis	Labrador Retriever	Westhighland White Terrier
	Cairn Terrier		
<i>Pyruvate kinase Deficiency 2 (PKDef)</i>	Pug		
<i>Pyruvate kinase Deficiency 3 (PKDef)</i>	Beagle		
<i>Severe combined Immunodeficiency (X-linked-SCID)</i>	Basset	Welsh Corgi	
<i>Severe combined Immunodeficiency (SCID)</i>	Jack Russell Terrier		
<i>Severe combined Immunodeficiency 2(SCID2)</i>	Frisian Water Dogs		
<i>Vitamin D-deficiency rickets, type II</i>	Pomeranian		
<i>Glycogen storage disease type II (Pompe Disease)</i>	Finnish Hound	Laponian Herder	Swedish Lapphund

Table 4 Eye disorders

Diseases	Breeds		
<i>Achromatopsia Type 1/Day Blindness</i>	Labrador Retriever		
<i>Albinism (White)</i>	Doberman Pincher		
<i>Canine Multi-focal Retinopathy (CMR 1)</i>	Australian Shepherd	American Bulldog	Dogue de Bordeaux
	Boerboel	Miniature Australian Shepherd	Laponian Herder
	Cane Corsos	Perro de Presa Canarios	Miniature American Shepherd
	Great Pyrenees	Mastiffs	
<i>Canine Multi-focal Retinopathy (CMR 2)</i>	Coton de Tulear		
<i>Canine Multi-focal Retinopathy (CMR1 & CMR2)</i>	Bull Dog	Bull Mastiff	English Bulldog
	English Mastiff		
<i>Collie Eye Anomalie (CEA)</i>	Australian Shepherd	Miniature Australian Shepherd	Miniature American Shepherd
	Bearded Collie	Rough Collie	Silken Windhound
	Border Collie	Longhaired Whippet	Whippet
	Boykin Spaniel	Shetland Sheepdog	Collies
	Hokkaido	Sheepdogs	Nova Scotia Duck Tolling Retriever
	Lancashire Heeler	Smooth Collies	
<i>Cone Degeneration (CD)</i>	German Shorthaired Pointer	Miniature Australian Shepherd	Miniature American Shepherd

	Alaskan malamute	Australian Shepherd	
<i>Cone Rod Dystrophy 2 (CRD2)</i>	American Pitbull Terrier	Glen of Imaal Terrier	Pitbull Terrier
	American Staffordshire Terrier		
<i>Cone Rod Dystrophy 4-PRA (CRD4-PRA)</i>	Dachshund		
<i>Cone-Rod Dystrophy 1-PRA (Cord1-PRA)</i>	Curly Coated Retrievers	Papillion	English Springer Spaniel
	Dachshund		
<i>Congenital stationary night blindness (CSNB)</i>	Briard		
<i>Dry eye curly coat syndrome (CCS)</i>	Cavalier King Charles Spaniel		
	Australian Shepherd	Wäller	Miniature Australian Shepherd
<i>Hereditary Cataract (HC)</i>	French Bulldog		
<i>Hereditary Cataract 2 (HSF4)</i>	Boston Terrier	Staffordshire Bull Terrier	
<i>Primary lens luxation (PLL)</i>	American Eskimo Dog	Pug	Rat Terrier
	American Hairless Terrier	Sealyham Terrier	Teddy Roosevelt Terrier
	Australian Cattle Dog	Tenterfield Terrier	Tibetan Terrier
	Chinese Crested Dog	Patterdale Terrier	Parson Russell Terrier
	Chinese Foo Dog	Norwich Terrier	Norfolk Terrier
	Fox Terrier	Miniature Bull Terrier	Toy Fox Terrier
	Jack Russell Terrier	Volpino Italiano	Welsh Terrier
	Jagd Terrier	Westphalia Terrier	Whire-haired Fox Terrier
	Lakeland Terrier	Yorkshire Terrier	Lucas Terrier
	Lancashire Heeler		
<i>Primary open angle glaucoma (POAG)</i>	Beagle		
<i>Progressive Retinal Atrophy (prcd PRA)</i>	Maltipoo	Norwegian Elkhound	Finnish Lapphund
	American Cocker Spaniel	Nova Scotia Duck Tolling Retriever	German Spitz
	American Eskimo Dog	Poodle	Giant Schnauzer
	Australian Cattle Dog	Portuguese Water Dog	Golden Retriever
	Australian Shepherd	Schipperke	Karelian Beardog
	Australian Silky Terrier	Silky Terrier	Kuvasz
	Australian Stumpy Tail Cattle Dog	Spanish Water Dog	Labrador Retriever
	Bolonka Zwetna	Swedish Lapphund	Laponian Herder
	Chesapeake Bay Retriever	Toy Poodle	Markiesje
	Chinese Crested Dog	Yorkshire Terrier	Miniature American Shepherd
	Cockapoo	Entlebucher Mountain Dog	Miniature Australian Shepherd
	Dwarf Poodle	English Shepherd	Miniature Poodle
	English Cocker Spaniel	Moyen Poodle	
<i>Progressive Retinal Atrophy -Dominant- (PRA)</i>	Bull Mastiff	Mastiffs	English Mastiff
<i>Progressive Retinal Atrophy (Basenji PRA)</i>	Basenjjs		
<i>Progressive Retinal Atrophy (crd-PRA)</i>	Dachshund		
<i>Progressive Retinal Atrophy (gPRA)</i>	Schapendoes		
<i>Progressive Retinal Atrophy (GR PRA1 and 2)</i>	Golden Retriever		
<i>Progressive Retinal Atrophy (IG PRA1)</i>	Italian Greyhound		
<i>Progressive Retinal Atrophy (rcd1 PRA)</i>	Irish Setters	Sloughi	

<i>Progressive Retinal Atrophy (rcd2 PRA)</i>	Collies	Smooth Collies	Rough Collie
	Farm Collie		
<i>Progressive Retinal Atrophy (rcd3 PRA)</i>	Cardigan Welsh Corgi	Chinese Crested Dog	Welsh Corgi
<i>Progressive Retinal Atrophy (rcd4 PRA)</i>	Australian Cattle Dog	Tibetan Terrier	Small Munsterlander
	English Setter	Polish Lowland Sheepdog	Llewellyn Setter
	Gordon Setter	Irish Setters	
<i>Progressive Retinal Atrophy (Type A PRA)</i>	Miniature Schnauzer		
<i>Progressive Retinal Atrophy 1 (Pap-PRA1)</i>	Papillion	Phalene	
<i>Progressive Retinal Atrophy 1 (X Linked PRA1)</i>	Samoyed	Siberian Husky	
<i>Retinal Dysplasia Retinal Folds OculoSkeletal Dysplasia (RD+OSD) 1 and 2</i>	Labrador Retriever	Samoyed	

Appendix 3

Cardiovascular system and blood disorders

Bleeding disorder due to P2RY12 defect

Great Swiss Mountain Dog Mutation described in article. (Boudreaux, 2011)

Cyclic Neutropenia (CN)

Australian Shepherd No article found about the mutation.

Collies Mutation described in article. (Benson, 2003)

Dilated Cardiomyopathy

Doberman Pincher This article is about that they do not know the mutation yet.

Wolfhound No article found about the mutation.
(Mausberg, Theresa-Bernadette 2011)

Factor VII - Deficiency

Giant Schnauzer No article found about the mutation

Beagle Mutation described in article (Callan, M B 2006)

Airedale Terrier No article found about the mutation

Alaskan Klee Kai Breed is not registered by the "Raad van Beheer"

Scottish Deerhound The mutation is only described for the Beagle

Haemophilia A (Factor VIII)

Havanese No article found about the mutation

Haemophilia B (Factor IX deficiency)

Rhodesian Ridgeback Mutation described in article. (Mischke, 2011)

Airedale Terrier Mutation described in article. (Mauser, 1996)

Bull Terrier Mutation described in article. (Mauser, 1996)

Cairn Terrier Mutation described in article but no breed. (Evans, 1989)

German Pointer No article found about the mutation

German Wirehaired Pointer Mutation described in article. (Brooks, Marjory 2003)

Lhasa Apso Mutation described in article. (Mauser, 1996)

Macrothrombocytopenia (MTC)

Boxer Article in GenPenn is about another disease no other article found.

Cavalier King Charles Spaniel Mutation described in article (Davis, 2008)

Chihuahua, English Cocker Spaniel, English Toy Spaniel, Havanese, Jack Russell Terrier, Maltese, Mastiffs, Miniature Poodle, Parson Russell Terrier, Poodle, Shih Tzu, Bichon Frise, Labrador Retriever
The mutation only described in the Cavalier King Charles Spaniel.

Polycythemia

All Breeds Only tested on a couple of breeds

Maltese, poodle, Yorkshire Terrier and West Highland White Terrier Mutation described in article.
(Beurlet, Stephanie 2011)

Thrombasthenia

Otterhound Mutation described in article. (Boudreaux, 2001)

Thrombasthenia 2

All Breeds **No article found about the mutation about all breeds.**
Pyreneese Berghond **Mutation described in article.** (Lipscomb, 2000)

Thrombocytopaenia

Cavalier King Charles Spaniel **No article found about the mutation.**

Thrombopathia

Basset, American Eskimo Dog, Landseer **Mutation described in article** (Boudreaux, Mary 2007)

Von-Willebrands Disease Type 1

Bernese Mountain Dog, Coton de Tulear, Doberman Pincher, Drentsche Patrijshond, German Pinscher, Kerry Blue Terrier, Manchester Terrier, Papillion, Pembroke Welsh Corgi, Poodle, Stabyhoun. **Article describes no mutation is yet found.** (Rieger,M. 1998)

Von-Willebrands Disease Type 2

Chinese Crested Dog, Collies, German Pointer **No article found about the mutation.**
German wirehaired pointer **Mutation described in article** (Kramer, 2004)

Von-Willebrands Disease Type 3

Dutch Kooiker, Scottish Terrier, Shetland Sheepdog **Mutation described in article** (Rieger,M. 1998)

Metabolic and immune diseases

C3 Deficiency

Brittany Spaniel **Mutation described in article** (Ameratunga, 1998)

Canine Leukocyte Adhesion Deficiency (CLAD), Type 1

Irish Setters **Mutation described in article** (Kijas, 1999)

Canine Leukocyte Adhesion Deficiency (CLAD), Type 3

German shepherd **Mutation described in article** (Boudreaux, 2010)

Cobalamin Malabsorption/cubilin deficiency

Beagle **Mutation described in article** (Fyfe, 2014)
Border Collie **Mutation described in article** (Fyfe, John 2013)
Boston Terrier **No article found about the mutation.**

Congenital Hypothyreosis (CHG) 1,2 and 3

Spanish Water Dog, Rat Terrier, Toy Fox Terrier **No article found about the mutation.**
Tenterfield Terriër **Mutation described in article** (Dodgson, 2012)

Copper toxicosis

Bedlington Terriers **Mutation described in article** (Forman, 2005)

Fucosidosis

English springer spaniel **Mutation described in article** (Skelly, 1999)

Glycogen Storage Disease Type I (GSD I)

Maltese **Mutation described in article** (Kishnani, 1997)

Glycogen Storage Disease GSD Type IIIa (GSDIIIa)

Curly Coated Retrievers **Mutation described in article** (Gregory, Brittany 2007)

Ivermectin hypersensitivity (MDR1 gene defect)

All Breed	Only tested on a couple of breeds
Australian Shepherd, Collies, Border Collie, Shetland Sheepdog, White Shepherd, German Shepherd	
Longhaired Whippet, Silken Windhound	Mutation described in article (Mealey, Katrina 2008)
Bobtail	No article found about the mutation.
McNab	Breed is not registered by the "Raad van Beheer".

Mucopolysaccharidose Type VII - 2

Brazilian Terrier Mutation described in article (Hytonen, 2012)

Mucopolysaccharidosis Type VII

German Shepherd Mutation described in article (Silverstein Dombrowski, Deborah 2004)

Mucopolysaccharidosis Type IIIa

Dachshund Mutation described in article (Aronovich, E L 2000)

Phosphofruktokinase deficiency (PFKD)

American Cocker Spaniel, German Spaniel, Whippetn	Mutation described in article (Giger, U 1992)
Cockapoo	No article found about the mutation.
English Cocker Spaniel	No article found about the mutation.
English Springer Spaniel	Mutation described in article (Smith, 1996)

Prekallikrein deficiency

Shih Tzu Mutation described in article (Okawa, Takumi 2011)

Pyruvate Dehydrogenase Phosphatase 1 (PDP1)

Clumber Spaniel, Sussex Spaniel Mutation described in article (Cameron, Jessie 2007)

Pyruvate kinase Deficiency (PKDef)

Basenjis	Mutation described in article (Whitney, 1995)
Cairn Terrier	Mutation described in article (Gultekin, 2012)
Westhighland White Terrier	Mutation described in article (Skelly, 1999)
Labrador Retriever	Mutation described in article (Gultekin, 2012)
Type 2 Pug	Mutation described in article (Gultekin, 2012)
Type 3 Beagle	Mutation described in article (Gultekin, 2012)

Severe combined Immunodeficiency (X-linked-SCID)

Basset, Welsh Corgi Mutation described in article (Perryman, 2004)

Severe combined Immunodeficiency (SCID)

Jack Russell Terrier Mutation described in article (Meek, 2001)

Severe combined Immunodeficiency 2(SCID2)

Frisian Water Dogs Mutation described in article (Verfuurden, 2011)

Trapped Neutrophil Syndrome (TNS)

Border Collie Mutation described in article (Mizukami, Keijiro 2012)

Vitamin D-deficiency rickets, type II

Pomeranian Mutation described in article. (LeVine, 2009)

Glycogen storage disease type II (Pompe Disease)

Finnish Hound	No article found about the mutation
Lapponian Herder, Swedish Lapphund	Mutation described in article (Seppälä, Eija 2013)

Eye diseases

Achromatopsia Type 1/Day Blindness

Labrador Retriever **No article found about the mutation**

Albinism (White)

Doberman Pincher **Mutation described in article** (Winkler, Paige 2014)

Canine Multi-focal Retinopathy (CMR)

Australian Shepherd **Mutation described in article** (Hoffmann, Ingo 2012)

Boerboel **Mutation described in article** (Gornik 2014)

Great Pyrenees, Mastiffs, **Mutation described in article** (Guziewicz, Karina 2007)

American Bulldog, Cane Corso, Dogue de Bordeaux, Miniature Australian Shepherd, Laponian Herder, Bull Dog, Bull Mastiff, English Bulldog, English Mastiff

No article found about the mutation

Miniature American Shepherd, Perro de Presa Canarios **Breeds are not registered by the "Raad van Beheer"**

Canine Multi-focal Retinopathy (CMR 2)

Coton de Tulear **Mutation described in article** (Guziewicz, Karina 2007)

Collie Eye Anomalie (CEA)

Australian Shepherd, Bearded Collie, Border Collie, Boykin Spaniel, Lancashire Heeler, Nova Scotia Duck Tolling Retriever, Sheepdogs, Shetland Sheepdog, Whippet, Collies, Longhaired Whippet, Miniature Australian Shepherd, Rough Collie, Smooth Collies **Mutation described in article** (Parker, Heidi 2007)

Hokkaido **Mutation described in article.** (Mizukami, Keijiro 2012)

Silken Windhound **No article found about the mutation**

Cone Degeneration (CD)

German Shorthaired Pointer, Alaskan malamute, **Mutation described in article** (Sidjanin, Duska 2002)

Australian Shepherd, Miniature Australian Shepherd **No article found about the mutation**

Cone Rod Dystrophy 2 (CRD2)

American Pitbull Terrier, American Staffordshire Terrier **Mutation described in article.** (Goldstein, Orly 2013)

Pitbull Terrier **No article found about the mutation**

Cone Rod Dystrophy 3 (CRD3)

Glen of Imaal Terrier **Mutation described in article.** (Goldstein, Orly 2010)

Cone Rod Dystrophy 4-PRA (CRD4-PRA)

Dachshund **No article found about the mutation**

Cone-Rod Dystrophy 1-PRA (Cord1-PRA)

Dachshund **Mutation described in article** (Miyadera, Keiko 2009)

English Springer Spaniel **Mutation described in article** (Downs, Louise 2014)

Papillon, Curly Coated Retriever **No article found about the mutation**

Congenital stationary night blindness (CSNB)

Briard **Mutation described in article** (Aguirre, 1998)

Dry eye curly coat syndrome (CCS)

Cavalier King Charles Spaniel **Mutation described in article** (Forman, Oliver 2012)

Hereditary Cataract (HC)

Australian Shepherd [Mutation described in article](#) (Mellersh, Cathryn 2006)

French Bulldog, Wäller, Miniature Australian Shepherd [No article found about the mutation](#)

Hereditary Cataract 2 (HSF4)

Boston Terrier, Staffordshire Bull Terrier [Mutation described in article](#) (Mellersh, Cathryn 2006)

Primary lens luxation (PLL)

Australian Cattle Dog, Chinese Crested Dog, Miniature Bull Terrier, Jack Russell Terrier, Jagd Terrier, Lancashire Heeler, , Parson Russell Terrier, Patterdale Terrier, Rat Terrier, Sealyham Terrier, Tenterfield Terrier, Tibetan Terrier, Toy Fox Terrier, Volpino Italiano, Welsh Terrier, Whire-haired Fox Terrier, Yorkshire Terrier

[Mutation described in article](#) (Gould, David 2011)

American Eskimo Dog, American Hairless Terrier, Chinese Foo Dog, Fox Terrier, Lakeland Terrier, Lucas Terrier, Norfolk Terrier, Norwich Terrier, Pug, Teddy Roosevelt Terrier, Westphalia Terrier.

[No article found about the mutation](#)

Primary open angle glaucoma (POAG)

Beagle [Mutation described in article](#) (Olson, Lana M 2011)

Progressive Retinal Atrophy (prcd PRA)

Maltipoo, Australian Silky Terrier, Bolonka Zwetna, English Shepherd, German Spitz, Giant Schnauzer, Schipperke [No article found about the mutation](#)

American Cocker Spaniel, American Eskimo Dog, Australian Cattle Dog, Australian Shepherd, Australian Stumpy Tail Cattle Dog, Chesapeake Bay Retriever, Chinese Crested Dog, Cockapoo, Dwarf Poodle, English Cocker Spaniel, Entlebucher Mountain Dog, Finnish Lapphund, Golden Retriever, Karelian Beardog, Kuvasz, Labrador Retriever, Lapponian Herder, Markiesje, Miniature Australian Shepherd, Miniature Poodle, Moyen Poodle, Norwegian Elkhound, Nova Scotia Duck Tolling Retriever, Poodle, Portuguese Water Dog, Silky Terrier, Spanish Water Dog, Swedish Lapphund, Toy Poodle, Yorkshire Terrier [Mutation described in article](#) (Zangerl, Barbara 2006)

Progressive Retinal Atrophy (Dominant-PRA)

Bull Mastiff, English Mastiff [Mutation described in article.](#) (Downs, Louise 2014)

Progressive Retinal Atrophy (Basenji PRA)

Basenjis [Mutation described in article](#) (Goldstein, Orly 2013)

Progressive Retinal Atrophy (gPRA)

Schapendoes [Mutation described in article](#) (Downs, Louise 2014)

Progressive Retinal Atrophy (GR PRA1)

Golden Retriever [Mutation described in article](#) (Downs, Louise 2014)

Progressive Retinal Atrophy (GR PRA2)

Golden Retriever [Mutation described in article](#) (Downs, Louise 2014)

Progressive Retinal Atrophy (IG PRA1)

Italian Greyhound [No article found about the mutation](#)

Progressive Retinal Atrophy (rcd1 PRA)

Irish Setters [Mutation described in article](#) (Downs, Louise 2014)

Sloughi [Mutation described in article](#) (Dekomien, G 2000)

Progressive Retinal Atrophy (rcd2 PRA)

Farm Collie, Rough Collies, Smooth Collies [Mutation described in article](#) (Kukekova, Anna V 2009)

Progressive Retinal Atrophy (rcd3 PRA)

Cardigan Welsh Corgi, Chinese Crested Dog [Mutation described in article](#) (Petersen-Jones, S M 1999)

Welsh Corgi [No article found about the mutation](#)

Progressive Retinal Atrophy (rcd4 PRA)

Australian Cattle Dog, Llewelin Setter, Polish Lowland Sheepdog, Small Munsterlander **No article found about the mutation.**

English setter

No article found about the mutation.

Gordon setter, Irish Setters

Mutation described in article (Downs, 2013)

Tibetan terrier

Mutation described in article. (Downs, Louise 2014)

Progressive Retinal Atrophy (Type A PRA)

Miniature Schnauzer

Mutation described in article (Downs, Louise 2014)

Progressive Retinal Atrophy 1 (Pap-PRA1)

Papillion, Phalene

Mutation described in article (Ahonen, Saija 2013)

Progressive Retinal Atrophy 1 (X Linked PRA1)

Samoyed, Siberian Husky

Mutation described in article (Zhang, Qi 2002)

Retinal Dysplasia Retinal Folds OculoSkeletal Dysplasia (RD+OSD) 1

Labrador Retriever, Samoyed

Mutation described in article (Goldstein, Orly 2010)

Appendix 4

Cardiovascular system and blood disorders

Table 5 Definitive list for good DNA tests

<i>Disease</i>	<i>Breed</i>
<i>Bleeding disorder due to P2RY12 defect</i>	Great Swiss Mountain Dog
<i>Cyclic Neutropenia (CN)</i>	Collies
<i>Factor VII - Deficiency</i>	Beagle
<i>Haemophilia B (Factor IX deficiency)</i>	Rhodesian ridgeback
	Airedale Terrier
	Bull Terrier
	German wirehaired pointer
	Lhasa Apso
<i>Macrothrombocytopenia (MTC)</i>	Boxer
	Cavalier King Charles Spaniel
<i>Polycythemia</i>	Maltese
	Poodle
	Yorkshire Terrier
	West Highland White Terrier
<i>Thrombasthenia</i>	Otterhound
<i>Thrombasthenia 2</i>	Pyreneese Berghond
<i>Thrombopathia</i>	Basset
	American Eskimo Dog
	Landseer
<i>Von-Willebrands Disease Type 2</i>	German Wirehaired Pointer
<i>Von-Willebrands Disease Type 3</i>	Dutch Kooiker
	Scottish Terrier
	Shetland Sheepdog

Metabolic and immune diseases

Table 6 Definitive list for good DNA tests

<i>Disease</i>	<i>Breed</i>
<i>C3 Deficiency</i>	Brittany Spaniel
<i>Canine Leukocyte Adhesion Deficiency (CLAD), Type 1</i>	Irish Setters
<i>Canine Leukocyte Adhesion Deficiency (CLAD), Type 3</i>	German shepherd
<i>Cobalamin Malabsorption/cubilin deficiency</i>	Beagle
	Border Collie
<i>Congenital Hypothyreosis (CHG) 2</i>	Tenterfield Terriër
<i>Copper toxicosis</i>	Bedlington Terriers
<i>Fucosidosis</i>	English springer spaniel
<i>Glycogen Storage Disease Type I (GSD I)</i>	Maltese
<i>Glycogen Storage Disease GSD Type IIIa (GSDIIIa)</i>	Curly Coated Retrievers
<i>Ivermectin hypersensitivity (MDR1 gene defect)</i>	Australian Shepherd
	Collies
	Border Collie
	Shetland Sheepdog
	White Shepherd
	German Shepherd
	Longhaired Whippet
	Silken Windhound

<i>Mucopolysaccharidose Type VII - 2</i>	Brazilian Terrier
<i>Mucopolysaccharidosis Type VII</i>	German Shepherd
<i>Mucopolysaccharidosis Type IIIa</i>	Dachshund
<i>Phosphofructokinase deficiency (PFKD)</i>	American Cocker Spaniel
	German Spaniel
	Whippet
	English Springer Spaniel
<i>Prekallikrein deficiency</i>	Shih Tzu
<i>Pyruvate Dehydrogenase Phosphatase 1 (PDP1)</i>	Clumber Spaniel, Sussex Spaniel
<i>Pyruvate kinase Deficiency (PKDef)</i>	Basenjis
	Cairn Terrier
	Westhighland White Terrier
	Labrador Retriever
	Type 2 Pug
	Type 3 Beagle
<i>Severe combined Immunodeficiency (X-linked-SCID)</i>	Basset
	Welsh Corgi
<i>Severe combined Immunodeficiency (SCID)</i>	Jack Russell Terrier
<i>Severe combined Immunodeficiency 2(SCID2)</i>	Frisian Water Dogs
<i>Trapped Neutrophil Syndrome (TNS)</i>	Border Collie
<i>Vitamin D-deficiency rickets, type II</i>	Pomeranian
<i>Glycogen storage disease type II (Pompe Disease)</i>	Lapponian Herder
	Swedish Lapphund

Eye diseases

Table 7 Definitive list for good DNA tests

<i>Disease</i>	<i>Breed</i>
<i>Albinism (White)</i>	Doberman Pincher
<i>Canine Multi-focal Retinopathy (CMR)</i>	Australian Shepherd
	Boerboel
	Great Pyrenees
	Mastiffs
<i>Canine Multi-focal Retinopathy (CMR 2)</i>	Coton de Tulear
<i>Collie Eye Anomalie (CEA)</i>	Australian Shepherd
	Bearded Collie
	Border Collie
	Boykin Spaniel
	Hokkaido
	Lancashire Heeler
	Nova Scotia Duck Tolling Retriever
	Sheepdog
	Shetland Sheepdog
	Whippet
	Collies
	Longhaired Whippet
	Miniature Australian Shepherd
	Rough Collies
	Smooth Collies
<i>Cone Degeneration (CD)</i>	German Shorthaired Pointer
	Alaskan malamute
<i>Cone Rod Dystrophy 2 (CRD2)</i>	American Pitbull Terrier
	American Staffordshire Terrier
<i>Cone Rod Dystrophy 3 (CRD3)</i>	Glen of Imaal Terrier
<i>Cone-Rod Dystrophy 1-PRA (Cord1-PRA)</i>	Dachshund

Congenital stationary night blindness (CSNB)	English Springer Spaniel		
	Briard		
	Dry eye curly coat syndrome (CCS)	Cavalier King Charles Spaniel	
		Australian Shepherd	
	Hereditary Cataract (HC)	Boston Terrier	
		Staffordshire Bull Terrier	
	Hereditary Cataract 2 (HSF4)	Australian Cattle Dog	
		Chinese Crested Dog	
		Miniature Bull Terrier	
		Jack Russell Terrier	
		Jagd Terrier	
		Lancashire Heeler	
		Parson Russell Terrier	
		Patterdale Terrier	
		Rat Terrier	
		Sealyham Terrier	
		Tenterfield Terrier	
		Tibetan Terrier	
	Primary lens luxation (PLL)	Toy Fox Terrier	
		Volpino Italiano	
Welsh Terrier			
Whire-haired Fox Terrier			
Yorkshire Terrier			
Primary open angle glaucoma (POAG)		Beagle	
		Progressive Retinal Atrophy (prcd PRA)	American Cocker Spaniel
			American Eskimo Dog
			Australian Cattle Dog
			Australian Shepherd
			Australian Stumpy Tail Cattle Dog
			Chesapeake Bay Retriever
	Chinese Crested Dog		
	Cockapoo		
	Dwarf Poodle		
	English Cocker Spaniel		
	Entlebucher Mountain Dog		
Finnish Lapphund			
Golden Retriever			
Karelian Beardog			
Kuvasz			
Labrador Retriever			
Lapponian Herder			
Markiesje			
Miniature Australian Shepherd			
Miniature Poodle			
Moyen Poodle			
Norwegian Elkhound			
Nova Scotia Duck Tolling Retriever			
Poodle			
Portuguese Water Dog			
Silky Terrier			
Spanish Water Dog			
Swedish Lapphund			
Toy Poodle			
Yorkshire Terrier			
Progressive Retinal Atrophy -Dominant- (PRA)	Bull Mastiff		
	English Mastiff		
Progressive Retinal Atrophy (gPRA)	Schapendoes		

<i>Progressive Retinal Atrophy (GR PRA1)</i>	Golden Retriever
<i>Progressive Retinal Atrophy (GR PRA2)</i>	Golden Retriever
<i>Progressive Retinal Atrophy (rcd1 PRA)</i>	Irish Setters
	Sloughi
<i>Progressive Retinal Atrophy (rcd2 PRA)</i>	Farm Collie
	Rough Collies
	Smooth Collies
<i>Progressive Retinal Atrophy (rcd3 PRA)</i>	Cardigen Welsh Corgi
	Chinese Crested Dog
<i>Progressive Retinal Atrophy (rcd4 PRA)</i>	Gordon setter
	Irish Setters
	Tibetan terrier
<i>Progressive Retinal Atrophy (Type A PRA)</i>	Miniature Schnauzer
<i>Progressive Retinal Atrophy 1 (Pap-PRA1)</i>	Papillion,
	Phalene
<i>Progressive Retinal Atrophy 1 (X Linked PRA1)</i>	Samoyed
	Siberian Husky
<i>Retinal Dysplasia Retinal Folds OculoSkeletal Dysplasia (RD+OSD) 1</i>	Labrador Retriever
	Samoyed

Appendix 5

The mail sent to the Laboratories

Questions about DNA tests for inherited diseases in dogs (research internship)



 **Maaïke Fennema** <m.fennema@students.uu.nl>

11 mei ☆



aan vetgen ▾

Dear Sir/Madam,

We are two Veterinary Medicine students at the University of Utrecht. We are doing a research internship with the subject 'DNA tests for inherited diseases; which ones are applicable for the market in the Netherlands?' under supervision of prof. dr. Jan Rothuizen and dr. Peter Leegwater. The goal is to produce a list of DNA tests which are, in our opinion, scientifically valid and suited for purebred dog populations in the Netherlands. Dog breeders and veterinarians can use this list to choose a reliable test for specific breeds and diseases. Advice of veterinarians will be used by dog breeders and the Kennel club to define a breeding program to systematically improve the health status of these populations and test the health of the actual offspring.

During our search on the internet we identified your company as one of the largest suppliers of DNA tests for inherited diseases in the Netherlands. Your support to our study will therefore be of great importance. To be able to evaluate the tests you offer we have the following questions relating to essential information we will need to evaluate the tests which is not available on your website.

We saw on your website many tests for different diseases. The questions we want to ask you are:

- What type of test is used for each disease, a mutation test or a marker test?
- Is each test based on published peer reviewed information? If so, can you please indicate the reference to the key publications on which your test is based?
- If the test is offered for more than one breed than those mentioned in the key publications, can you inform us about additional information you may have about the applicability in other breeds?

We would be most grateful to receive your answers to our questions. If you require any further information, feel free to contact us.

Yours sincerely,

Amy Koning (A.J.Koning@uu.nl)

Maaïke Fennema (M.Fennema@uu.nl)


Prof. dr. Jan Rothuizen (J.Rothuizen@uu.nl)

Dr. Peter Leegwater (P.A.J.Leegwater@uu.nl)

Answer from OptiGen

Re: Questions about DNA tests for inherited diseases in dogs (research internship)



 **Sue Pearce-Kelling** <suepk@optigen.com>

11 mei ★



aan mij ▾

Dear Maaïke,

Thank you for contacting OptiGen about your internship project and for taking on this worthwhile project. I have provided brief replies to your questions below in **red text**. If further details are needed, please feel free to contact me.

Best regards,
Sue PK

Sue Pearce-Kelling
President and Manager, OptiGen, LLC
Cornell Business & Technology Park
767 Warren Road, Suite 300
Ithaca, NY 14850
www.optigen.com
ph: [\(607\) 257-0301](tel:(607)257-0301)
fax: [\(607\) 257-0353](tel:(607)257-0353)

We saw on your website many tests for different diseases. The questions we want to ask you are:

- What type of test is used for each disease, a mutation test or a marker test? Currently (as of 5/11/15), ALL of OptiGen's DNA tests are mutation, not marker, based.

- Is each test based on published peer reviewed information? If so, can you please indicate the reference to the key publications on which your test is based? Most of OptiGen's tests are based on published peer-reviewed information and all in that category are referenced in the WSAVA database: <http://research.vet.upenn.edu/Default.aspx?TabId=7620> If you are not familiar with this useful site, I think you may find it very informative. Unfortunately, there is currently one important piece of information missing from this database—the mention of Intellectual Property (patents & licensing). As you may be aware, some of the DNA tests are governed by patents and licenses are required in order for a laboratory to use/sell the tests. You can find information on OptiGen's licensed tests on our webpage here: http://www.optigen.com/opt9_patent.html

- If the test is offered for more than one breed than those mentioned in the key publications, can you inform us about additional information you may have about the applicability in other breeds? All of the tests that Optigen currently offers are mutation based and to the best of our knowledge, are typically fully penetrant, regardless of breed background. The prcd-PRA mutation, for example, has been shown to cause PRA in many more breeds than were initially known to carry the mutation at the time of the research paper publication (in 2005). We are aware that there can be some variations in age of onset and rate of disease progression, particularly in a couple of breeds that carry prcd. English Cocker Spaniels (ECS) that are homozygous for the prcd mutation often do not show clinical symptoms of PRA until they are over 7 years of age whereas most breeds that are homozygous for prcd show clinical symptoms of early-stage retinal degeneration by the time dogs are 3-4 years of age. We are very interested in understanding what modifiers in the ECS genetic background cause this delayed/slower retinal disease progression.


We would be most grateful to receive your answers to our questions. If you require

any further information, feel free to contact us.

Answer from VetGen

Questions about DNA tests for inherited diseases in dogs (research internship)



 **VetGen Laboratory** <vetgen@vetgen.com>
aan mij ▾

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FROM THE GENETICIST

Dear Amy and Maaïke,

Our web site is a constant work in progress, but as for the current listings we can tell you that some have been developed in house while most are based on research done elsewhere. All of the primary publications for each test may be found on the WSAVA database hosted by UPenn.

<http://research.vet.upenn.edu/Default.aspx?TabId=7620>

Here you can search by disease or breed, and primary publications where available will be listed. In the cases where we have breeds not listed in the primary publication, it is due either to reference in secondary publications as with many of the eye diseases, or detection of the mutation in our own research samples. In the case of all of the type I vWD breeds, the additional breeds were added after correlation between the presence of the mutation and known bleeders with low ELISA numbers for most breeds.

Let us know if you have questions about any specific tests.


VetGen Customer Service

vetgen@vetgen.com

Answer from Laboklin

Questions about DNA tests for inherited diseases in dogs (research internship)



 **Maaïke Fennema** Dear Sir/Madam, We are two Veterinary Medicine students at the University of ... 11 mei ☆

 **LABOKLIN Niederlande** <service.nl@laboklin.com> 11 mei ★  

aan mij ▾

Beste Maaïke,

Ik ga eens kijken wat ik voor jullie kan doen. Het is zo dat wij alleen testen aanbieden als wij ergens een bewijzend onderzoek hebben gevonden. Maar bel mij maar even wanneer jullie tijd hebben.

Met vriendelijke groet,
Alexandra Knossenburg
Dierenarts

LABOKLIN GmbH&Co.KG
Verlengde Klinkertstraat 6
6433 PL Hoensbroek (NL)
Tel: 0031 (0)85 4890580
eMail service.nl@laboklin.com

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