AVAILABLE TESTS FOR GENE-BASED EYE DISEASES

Authors: Roberta Corvi, DMV, PhD , Simon Petersen-Jones, DVM, PhD, Dipl.ECVO
Co-Authors: Adolfo Guandalini, DMV, PhD, Dipl.ECVO, Gilles Chaudieu, DVM, Dipl.ECVO

SYNDROMIC CONDITIONS

Bardet-Biedl Syndrome
GENE: Bardet-Biedl Syndrome 4 gene (BBS4)
MUTATION: Base Substitution A > T in exon 2
Puli (LABOKLIN)

Chew T et al. A Coding Variant in the Gene Bardet-Biedl Syndrome 4 (BBS4) Is Associated with a Novel Form of Canine Progressive Retinal Atrophy. G3 (Bethesda). 2017; 7(7): 2327–2335.

Curly Coat Dry Eye Syndrome (CKSID)
GENE: Family With Sequence Similarity 83 Member H (FAM83H)
MUTATION: 1 bp deletion (del C) exon 5
Australian Cobberdog (PPG)
Bernardoodle (ORIVET)
Cavador (ORIVET)
Cavalier King Charles Spaniel (ANTAGENE, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG)
Cavapoo (PPG)
Cavoodle (ORIVET)

Hartley C et al. Congenital keratoconjunctivitis sicca and ichthyosiform dermatosis in Cavalier King Charles spaniel dogs. Part II: candidate gene study. Vet Ophthalmol. 2012; 15: 327-332.
Forman OP et al. Parallel mapping and simultaneous sequencing reveals deletions in BCAN and FAM83H associated with discrete inherited disorders in a domestic dog breed. PLoS Genet. 2012; 8: e1002462.
Drogemuller M et al. A mutation in the FAM83G gene in dogs with hereditary footpad hyperkeratosis (HFH). PLoS Genet. 2014; 10: e1004370.

Gangliosidosis 1 (GM1) Alaskan Husky Type
GENE: Galactosidase Beta 1 (GLB1)
MUTATION: 19 bp duplication (dup TCCCAGACTTGCCCCAGGA)
Alaskan Husky (GENEFAST, LABOKLIN, PPG)

Muller G et al. GM-1-gangliosidosis in Alaskan Huskies: Clinical and pathological findings. Vet Pathol. 2001;38(3):281-90
Kreutzer R et al. A duplication in the canine beta-galactosidase gene GLB1 causes exon skipping and GM1-gangliosidosis in Alaskan huskies. Genetics. 2005; 170(4):1857-61.

Kreutzer R et al. Impact of beta-galactosidase mutations on the expression of the canine lysosomal multienzyme complex. Biochim Biophys Acta. 2009;1792(10):982-7.

Gangliosidosis 1 (GM1) Portuguese Water Dog Type
GENE: Galactosidase Beta 1 (GLB1)
MUTATION: Base Substitution G>A
Porti-Doodle, (ORIVET)
Portuguese Water Dog (GENEFAST, GENOMIA, LABOKLIN, ORIVET, PPG))
* included in WISDOM PANEL Premium Kit

Saunders GK et al. GM1 gangliosidosis in Portuguese water dogs: pathologic and biochemical findings. Vet Pathol. 1988 ;25(4):265-9
Wang, ZH. Isolation and characterization of the normal canine BETA-galactosidase gene and its mutation in a dog model of GM1-gangliosidosis. J Inherit Metab Dis. 2000; 23;593-606.

Gangliosidosis 1 (GM1) Shiba Inu Type
GENE: Galactosidase Beta 1 (GLB1)
MUTATION: 1 bp deletion (delC) in exon 15
Shiba Inu (GENEFAST, GENOMIA, LABOKLIN, ORIVET, PPG, UCD)
Shih tzu (ORIVET)
* included in WISDOM PANEL Premium Kit

Yamato O et al. A novel mutation in the gene for canine acid beta-galactosidase that causes GM1-gangliosidosis in Shiba dogs. J Inherit Metab Dis. 2002; 25: 525-526.
Uddin MM et al. Molecular epidemiology of canine GM1 gangliosidosis in the Shiba Inu breed in Japan: relationship between regional prevalence and carrier frequency. BMC Vet Res. 2013; 9: 132.

Gangliosidosis 2 (GM2) Japanese Chin Type
GENE: Hexosaminidase Subunit Alpha (HEXA)
MUTATION: Base Substitution G>A
Japanese Chin (LABOKLIN, ORIVET, PPG, RCGHA)
* included in WISDOM PANEL Premium Kit
**a test for mutation in HEXB instead of HEXA is offered by this Lab

Sanders DN et al. GM2 gangliosidosis associated with a HEXA missense mutation in Japanese Chin dogs: a potential model for Tay Sachs disease. Mol Genet Metab. 2013; 108: 70-75.

Gangliosidosis 2 (GM2) Poodle Type
GENE: Hexosaminidase Subunit Beta (HEXB)

MUTATION: 1 bp deletion (delG)
Aussiedoodle (PPG)
Australian Cobberdog (ORIVET)
Australian Labradoodle (ORIVET, PPG)
Bernedoodle (PPG)
Bordoodle (PPG)
Cavapoo (PPG)
Cockapoo (ORIVET, PPG)
Dwarf Poodle (ORIVET)
Goldendoodle (ORIVET, PPG)
Groodle (ORIVET)
Labradoodle (ORIVET, PPG)
Maltipoo (PPG)
Miniature Poodle (LABOKLIN, PPG)
Newfypoo (PPG)
Standard Poodle (ORIVET, PPG, RCGHA)
Retrodoodle (ORIVET)
Schnoodle(ORIVET, PPG)
Sheepadoodle (PPG)
Toy Poodle (LABOKLIN, ORIVET, PPG, RCGHA)
* included in WISDOM PANEL Premium Kit

Rahman MM et al. A frameshift mutation in the canine HEXB gene in toy poodles with GM2 gangliosidosis variant 0 (Sandhoff disease). The Vet J. 2012; 194(3);412–416.

Gangliosidosis 2 (GM2) Shiba Inu Type
GENE: Hexosaminidase Subunit Beta (HEXB)
MUTATION: 3 bp deletion (delCCT) in exon 3
Shiba Inu (GENOMIA, LABOKLIN, ORIVET, PV)

Kolicheski A et al.GM2 Gangliosidosis in Shiba Inu Dogs with an In-Frame Deletion in HEXB. J Vet Intern Med. 2017; Sep;31(5):1520-1526

Lagotto Storage Disease (LSD)
GENE: Autophagy Related 4D Cysteine Peptidase (ATG4D)
MUTATION: chr20:50618958-50618958: G/ A
Lagotto Romagnolo (GENEFAST, LABOKLIN, RCGHA)
*included in WISDOM PANEL Premium Kit

Kyöstilä K et al. A missense change in the ATG4D gene links aberrant autophagy to a neurodegenerative vacuolar storage disease. PLoS Genet. 2015;15;11(4):e1005169.
Syrjä P et al. Basal Autophagy Is Altered in Lagotto Romagnolo Dogs with an ATG4D Mutation. Vet Pathol. 2017;54(6):953-963.

Mucopolysaccharidosis Type I (Plott Hound Type)
GENE: Alpha-L-Iduronidase (IDUA)
MUTATION: Base Substitution G>A
Plott Hound (ORIVET, PPG, PV)

Menon KP et al. Architecture of the canine IDUA gene and mutation underlying canine mucopolysaccharidosis I. Genomics. 1992; 14;763-768.

Mucopolysaccharidosis Type I (Boston Terrier Type)
GENE: Alpha-L-Iduronidase (IDUA)
MUTATION: 8 bp insertion (ins CGGCCCCC)
Boston Terrier (PV)

Menon KP et al. Architecture of the canine IDUA gene and mutation underlying canine mucopolysaccharidosis I. Genomics. 1992; 14;763-768.

Mucopolysaccharidosis IIIA (Dachshund Type)
GENE: N-Sulfoglucosamine Sulfohydrolase (SGSH)
MUTATION: 3 bp deletion (del CCA)
Dachshund Standard/Miniature Long/Smooth/Wirehaired (LABOKLIN**, PPG, PV, RCGHA**)
* included in WISDOM PANEL Premium Kit
** this Lab doesn't specify if different mutations are searched in Dachshund and New Zealand Huntway dogs)

Aronovich EL et al. Canine heparan sulfate sulfamidase and the molecular pathology underlying Sanfilippo syndrome type A in Dachshunds. Genomics. 2000; 68:80-84
Crawley AC et al. Enzyme replacement reduces neuropathology in MPS IIIA dogs. Neurobiol Dis. 2011; 43:422-434

Mucopolysaccharidosis IIIA (New Zealand Huntway Type)
GENE: N-Sulfoglucosamine Sulfohydrolase (SGSH)
MUTATION: 1 bp insertion (ins A)
New Zealand Huntaway (LABOKLIN**, ORIVET**, PPG, PV)
* included in WISDOM PANEL Premium Kit
** ** this Lab doesn't specify if different the mutations are searched in Dachshund and New Zealand Huntway dogs)

Yogalingam G et al. Identification of a mutation causing mucopolysaccharidosis type IIA in New Zealand Huntaway dogs. Genomics. 2002 ;79(2):150-153.
Crawley AC et al. Enzyme replacement reduces neuropathology in MPS IIIA dogs. Neurobiol Dis. 2011; 43:422-434

Mucopolysaccharidosis IIIB
GENE: N-Acetyl-Alpha-Glucosaminidase (NAGLU)
MUTATION: 40-70 bp insertion (and 11 bp duplication) in exon 6
Schipperke (GENEFAST, LABOKLIN, PV)

Raj K et al. An exonic insertion in the NAGLU gene causing Mucopolysaccharidosis IIIB in Schipperke dogs. Sci Rep. 2020;10(1):3170.

Mucopolysaccharidosis VI (Great Dane Type)
GENE: Arylsulfatase B (ARSB)
MUTATION: Base Substitution C>T
Great Dane (ORIVET)

Ping W et al. Mucopolysaccharidosis Type VI in a Great Dane Caused by a Nonsense Mutation in the ARSB Gene. Vet Pathol. 2018;55(2):286-293

Mucopolysaccharidosis VI (Miniature Pinscher Type)
GENE: Arylsulfatase B (ARSB)
MUTATION: Base Substitution G>A in exon 5
Miniature Pinscher (GENEFAST, PV)

Raj K et al. ARSB gene variants causing Mucopolysaccharidosis VI in Miniature Pinscher and Miniature Schnauzer dogs. Anim Genet. 2020;51(6):982-986.

Mucopolysaccharidosis VI (Miniature Schnauzer Type)
GENE: arylsulphatase B (ARSB)
MUTATION: 56 bp deletion at the junction of exon 1
Miniature Schnauzer (PV)

Raj K et al. ARSB gene variants causing Mucopolysaccharidosis VI in Miniature Pinscher and Miniature Schnauzer dogs. Anim Genet. 2020;51(6):982-986.

Mucopolysaccharidosis VI (Poodle Type)
GENE: Arylsulfatase B (ARSB)
MUTATION: 22bp deletion in exon 1
Australian Cobberdog (ORIVET)
Australian Labradoodle (ORIVET)
Bernardoodle (ORIVET)
Cavoodle (ORIVET)
Goldendoodle (ORIVET)

Groodle (ORIVET)
Harlequin Pinscher (ORIVET)
Labradoodle (ORIVET)
Miniature Poodle (ORIVET, PV)
Moodle (ORIVET)
Schipperke (ORIVET)
Toy Poodle (ORIVET, PV)

Jolly RD et al. Mucopolysaccharidosis type VI in a Miniature Poodle-type dog caused by a deletion in the arylsulphatase B gene. N Z Vet J. 2012; 60:183-8

Mucopolysaccharidosis VII (Brazilian Terrier Type)
GENE: Glucuronidase Beta (GUSB)
MUTATION: Base Substitution C>T
Brazilian Terrier (LABOKLIN, RCGHA, PPG, UCD)
* included in WISDOM PANEL Premium Kit

Hytönen MK et al. A Novel GUSB Mutation in Brazilian Terriers with Severe Skeletal Abnormalities Defines the Disease as Mucopolysaccharidosis. PLoS One. 2012; 7:e40281

Mucopolysaccharidosis VII (German Shepherd Type)
GENE: Glucuronidase Beta (GUSB)
MUTATION: Base Substitution G>A
Belgian Groenendael Shepherd (ORIVET)
Belgian Lakenois Shepherd (ORIVET)
Belgian Malinois Shepherd (ORIVET, PPG)
Belgian Tervueren Shepherd (ORIVET)
German Shepherd Dog (GENEFAST, LABOKLIN, ORIVET, PPG, PV, RCGHA, UCD)
Shiloh Sheperd (PPG)
White Sheperd Dog (PPG)
* included in WISDOM PANEL Premium Kit

Ray J et al. Cloning of the canine beta-glucuronidase cDNA, mutation identification in canine MPS VII, and retroviral vector-mediated correction of MPS VII cells. Am J Vet Res. 1998; 59;1092–1095.

Muslandin-Lueke Syndrome (MLS)
GENE: ADAMTS Like 2 (ADAMTSL2)
MUTATION: Base Substitution C>T
Beagle (AG, ANTAGENE, GENOMIA, PPG, RCGHA, UCD)
* included in WISDOM PANEL Premium Kit

Bader HL et al. An ADAMTSL2 Founder Mutation Causes Musladin-Lueke Syndrome, a Heritable Disorder of Beagle Dogs, Featuring Stiff Skin and Joint Contractures. PLoS ONE. 2010; 5(9): e12817.

Neuronal Ceroid Lipofuscinosis 1 (Cane Corso Type)
GENE: Palmitoyl-Protein Thioesterase 1 (PPT1)
MUTATION: chr15:2860424-2860424: G/A
Cane Corso Italiano (PPG)

Kolicheski A et al. Homozygous PPT1 Splice Donor Mutation in a Cane Corso Dog With Neuronal Ceroid Lipofuscinosis. J Vet Intern Med. 2017 ;31(1):149-157

Neuronal Ceroid Lipofuscinosis 1 (Dachshund Type)
GENE: Palmitoyl-Protein Thioesterase 1 (PPT1)
MUTATION: 1 bp insertion (ins C)
Dachshund Standard Short/Long/Smooth/Wire Hair (ORIVET, PPG, RCGHA)
Miniature Dachshund Short/Long/Smooth/Wire Hair (ORIVET, PPG, RCGHA)
* included in WISDOM PANEL Premium Kit

Sanders DN et al. A mutation in canine PPT1 causes early onset neuronal ceroid lipofuscinosis in a Dachshund. Mol Genet and Metab. 2010; 100(4);349–356

Neuronal Ceroid Lipofuscinosis 2 (Dachshund Type)
GENE: Palmitoyl-Protein Thioesterase 1 (PPT1)
MUTATION: 1 bp deletion (del C)
Dachshund Standard Short/Long/Smooth/Wire Hair (PPG)
Miniature Dachshund Short/Long/Smooth/Wire Hair (PPG)

Awano T et al. A frame shift mutation in canine TPP1 (the ortholog of human CLN2) in a juvenile Dachshund with neuronal ceroid lipofuscinosis. Mol Genet Metab. 2006; 89(3):254-60

Neuronal Ceroid Lipofuscinosis 4A (American Bully Type)
GENE: Arylsulfatase G (ARSG)
MUTATIONn: Base Substitution G>A
American Bully (PPG)
American Pit Bull Terrier (PPG)
American Staffordshire Terrier (PPG)
Bull Terrier (PPG)

Abitbol M et al. A canine Arylsulfatase G (ARSG) mutation leading to a sulfatase deficiency is associated with neuronal ceroid lipofuscinosis. Proc Natl Acad Sci USA. 2010 ; 107(33):14775-80

Neuronal Ceroid Lipofuscinosis 5 (Border Collie Type)
GENE: CLN5 Intracellular Trafficking Protein (CLN5)
MUTATION: Base Substitution C>T
Australian Cattle Dog (GENOMIA, ORIVET, PPG)
Australian Stumpy Tail Cattle Dog (ORIVET, PPT)
Border Collie (AG, GENOMIA, ORIVET,PPG)
Bordoodle (PPG)
Koolie (ORIVET, PPG)
Miniature Australian Cattle Dog (PPG)

Melville SA et al. A mutation in canine CLN5 causes neuronal ceroid lipofuscinosis in Border collie dogs. Genomics. 2005; 86;287-294.

Neuronal Ceroid Lipofuscinosis 5 (Golden Retriever Type)
GENE: CLN5 Intracellular Trafficking Protein (CLN5)
MUTATION: 2 bp deletion (del AG)
Golden Retriever (AG, ANTAGENE, GENOMIA, PPG, UCD)
Goldendoodle (PPG, UCD)

Gilliam D et al. Golden Retriever dogs with neuronal ceroid lipofuscinosis have a two-base-pair deletion and frameshift in CLN5. Mol Genet Metab. 2015; 115:101-9

Neuronal Ceroid Lipofuscinosis 6 (Australian Shepherd Type)
GENE: CLN6 Transmembrane ER Protein (CLN6)
MUTATION: Base Substitution T>C
Aussiedoodle (PPG)
Australian Shepherd (ORIVET, GENOMIA, PPG)
Miniature American Shepherd (ORIVET, PPG)
Miniature Australian Shepherd (PPG)
New Zealand Heading Dog (ORIVET)
Toy Australian Shepherd (PPG)

Katz ML et al. A missense mutation in canine CLN6 in an Australian shepherd with neuronal ceroid lipofuscinosis. J Biomed Biotechnol. 2011; 2011: 198042

Neuronal Ceroid Lipofuscinosis 7 (Chinese Crested Type)
GENE: Major Facilitator Superfamily Domain Containing 8 (MFSD8)
MUTATION: 1 bp deletion (delT)

Chihuahua (ANTAGENE, ORIVET, PPG, RCGHA)
Chinese Crested (ANTAGENE, ORIVET, PPG, RCGHA)
* included in WISDOM PANEL Premium Kit

Guo J et al. A rare homozygous MFSD8 single-base-pair deletion and frameshift in the whole genome sequence of a Chinese Crested dog with neuronal ceroid lipofuscinosis. BMC Vet Res. 2015;10;960.
Ashwini A et al. Neuronal ceroid lipofuscinosis associated with an MFSD8 mutation in Chihuahuas. Mol Genet Metab. 2016;118(4):326-32.
Faller KME et al. The Chihuahua Dog: A New Animal Model for Neuronal Ceroid Lipofuscinosis CLN7 Disease? J Neurosci Res. 2016;94(4):339-47

Neuronal Ceroid Lipofuscinosis 8 (Alpine Dachsbracke Type)
GENE: CLN8 Transmembrane ER And ERGIC Protein (CLN8)
MUTATION: g.30852988_30902901del
Alpine Dachsbracke (ANTAGENE, GENOMIA, RCGHA)
* included in WISDOM PANEL Premium Kit

Hirz M et al. Neuronal ceroid lipofuscinosis (NCL) is caused by the entire deletion of CLN8 in the Alpenländische Dachsbracke dog. Molecular Genetics and Metabolism. 2017; 120:269–277

Neuronal Ceroid Lipofuscinosis 8 (Australian Sheperd Type)
GENE: CLN8 Transmembrane ER And ERGIC Protein (CLN8)
MUTATION: Base Substitution G>A
Aussiedoodle (PPG)
Australian Cattle Dog (RCGHA)
Australian Sheperd (RCGHA, PPG)
Miniature American Sheperd (RCGHA, PPG)
Miniature Australian Sheperd (PPG)
Toy Australian Sheperd (PPG)
* included in WISDOM PANEL Premium Kit

Guo J et al. A CLN8 nonsense mutation in the whole genome sequence of a mixed breed dog with neuronal ceroid lipofuscinosis and Australian Shepherd ancestry. Mol Genet Metab 2014; 112:302-9

Neuronal Ceroid Lipofuscinosis 8 (English Setter Type)
GENE: CLN8 Transmembrane ER And ERGIC Protein (CLN8)
MUTATION: Base Substitution C>T
English Setter (ANTAGENE, GENEFAST, GENOMIA, ORIVET, PPG, RCGHA, TIHO)
Gordon Setter (ANTAGENE, ORIVET, PPG)

Irish Setter (ORIVET)
* included in WISDOM PANEL Premium Kit

Katz ML et al. A mutation in the CLN8 gene in English Setter dogs with neuronal ceroid-lipofuscinosis. Biochem Biophys Res Com. 2005; 327;541-547.

Neuronal Ceroid Lipofuscinosis 8 (Saluki Type)
GENE: CLN8 Transmembrane ER And ERGIC Protein (CLN8)
MUTATION: 1 bp insertion (c.349dupT) in exon 2
Saluki (GENOMIA)
* included in WISDOM PANEL Premium Kit

Lingaas F et al. Neuronal ceroid lipofuscinosis in Salukis is caused by a single base pair insertion in CLN8. Anim Genet. 2018; 49:52-58

Neuronal Ceroid Lipofuscinosis 10 (American Bulldog Type)
GENE: Cathepsin D (CTSD)
MUTATION: Base Substitution c.597G>A
American Bulldog (ANTAGENE, GENOMIA, ORIVET,PPG, TIHO)
American Bully (ANTAGENE, ORIVET, PPG)
American Pit Bull Terrier (ORIVET)

Awano T et al. A mutation in the cathepsin D gene (CTSD) in American Bulldogs with neuronal ceroid lipofuscinosis. Mol Genet Metab. 2006; 87(4);341–348.

Neuronal Ceroid Lipofuscinosis 12 (Australian Cattledog Type)
GENE: ATPase Cation Transporting 13A2 (ATP13A2)
MUTATION: Base Substitution C>T
Australian Cattledog (GENOMIA)
* included in WISDOM PANEL Premium Kit

Schmutz I et al. ATP13A2 missense variant in Australian Cattle Dogs with late onset neuronal ceroid lipofuscinosis. Mol Genet Metab. 2019; 127:95-106

Neuronal Ceroid Lipofuscinosis 12 (Tibetan Terrier Type)
GENE: ATPase Cation Transporting 13A2 (ATP13A2)
MUTATION: 1 bp deletion (delG)n exon 16
Tibetan Terrier (ANTAGENE, GENOMIA, ORIVET, PPG, TIHO)

Farias FG et al. A truncating mutation in ATP13A2 is responsible for adult-onset neuronal ceroid lipofuscinosis in Tibetan terriers, Neurobiol. Dis. 2011; doi:10.1016/j.nbd.2011.02.009

Wohlke A et al. A one base pair deletion in the canine ATP13A2 gene causes exon skipping and late-onset neuronal ceroid lipofuscinosis in the Tibetan terrier. PLoS Genet. 2011; 7: e1002304.

Neuronal Ceroid Lipofuscinosis A (American Staffordshire and Pit Bull Terriers Type)
GENE: Arylsulphatase G (ARSG gene)
MUTATION: Base Substitution G>A
American Staffordshire Terrier (GENEFAST, GENOMIA)
Pitt Bull Terrier (GENOMIA)

Abitol M at al. A canine Arylsulfatase G (ARSG) mutation leading to a sulfatase deficiency is associated with neuronal ceroid lipofuscinosis. Proc Natl Acad Sci USA. 2010; 17;107(33):14775-80

Oculoskeletal dysplasia (RD/OSD - Drd1)
GENE: COL9A3
MUTATION: 1 bp insertion (ins G) in exon 1
Labradoodle (PPG)
Labrador Retrievers (GENOMIA, LABOKLIN, PPG)

Goldstein O et al. COL9A2 and COL9A3 mutations in canine autosomal recessive oculoskeletal dysplasia. Mamm Genome. 2010; 21: 398-408
Smit JJ et al. Evaluation of candidate genes as a cause of chondrodysplasia in Labrador retrievers. Vet J. 2011; 187: 269- 271.

Oculoskeletal dysplasia (RD/OSD – Drd1, Northern Inuit Type)
GENE: Collagen Type IX Alpha 3 Chain (COL9A3)
MUTATION: nonsense single nucleotide polymorphism in exon 14
Northern Inuit (LABOKLIN)
Tamaskan (LABOKLIN)

Stavinohova R et al. Clinical, histopathological and genetic characterisation of oculoskeletal dysplasia in the Northern Inuit Dog. PloS One. 2019;14(8):e0220761

Oculoskeletal dysplasia 2 (RD/OSD - Drd2)
GENE: Collagen Type IX Alpha 2 Chain (COL9A2)
MUTATION: 1267 bp deletion
Samoyedo (LABOKLIN, PPG)

Goldstein O et al. COL9A2 and COL9A3 mutations in canine autosomal recessive oculoskeletal dysplasia. Mamm Genome. 2010; 21: 398-408.

Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation (POANV)
GENE: RAB3 GTPase Activating Protine Catalytic Subunit 1 (RAB3GAP1)
MUTATION: 1 bp deletion
Black Russian Terrier (UCD)
Rottweiler (UCD)

Mhlanga-Mutangadura T et al. A mutation in the Warburg syndrome gene, RAB3GAP1, causes a similar syndrome with polyneuropathy and neuronal vacuolation in Black Russian Terrier dogs. Neurobiol Dis. 2016;86:75-85.
Mhlanga-Mutangadura T et al. A Homozygous RAB3GAP1:c.743delC Mutation in Rottweilers with Neuronal Vacuolation and Spinocerebellar Degeneration. J Vet Intern Med. 2016 ; 30(3): 813–818.

GLOBE

Congenital Eye Malformations
GENE: SIX Homeobox 6 (SIX6)
MUTATION: Base Substitution C>T
Golden Retriever (ORIVET)
Goldendoodle (ORIVET)
Groodle (ORIVET)

Hug P et al. A SIX6 Nonsense Variant in Golden Retrievers with Congenital Eye Malformations. Genes. 2019; 10, 454;1-11

Microphthalmia, Anophthalmia and Coloboma (MO-SCWT)
GENE: Retinol Binding Protein 4 (RBP4)
MUTATION: 3 bp deletion (delAAG)
Soft Coated Wheaten Terrier (LABOKLIN, ORIVET, PV)
* included in WISDOM PANEL Premium Kit

Kaukonen M et al. Maternal Inheritance of a Recessive RBP4 Defect in Canine Congenital Eye Disease. Cell Rep. 2018; 23:2643-2652

Microphthalmia (MOS-PWD)
GENE: UNPUB
MUTATION: UNPUB
Portuguese Water Dog (PV)

Shaw GC et al. Microphthalmia With Multiple Anterior Segment Defects in Portuguese Water Dogs. Vet Pathol. 2019;56(2):269-273

CONJUNCTIVA

Ligneous membranitis (Maltese Type)
GENE: UNPUB
MUTATION: UNPUB
Maltese (GENEFAST)

UNPUB

Ligneous membranitis (Scottish Terrier Type)
GENE: Plasminogen (PLG)
MUTATION: crh1:49514382-49514382: T/ A
Scottish Terrier (RCGHA)
* included in WISDOM PANEL Premium Kit

Ainsworth S et al. Ligneous membranitis in Scottish Terriers is associated with a single nucleotide polymorphism in the plasminogen (PLG) gene. Anim Genet. 2015;46(6):707-710

CORNEA

Macular Corneal Dystrophy (MCD)
GENE: Carboydrate Sulfotransferase 6 (CHST6)
MUTATION: Base Substitution C>A
Australian Labradoodle (ORIVET, PPG)
Goldendoodle (ORIVET)
Labradoodle (ORIVET, PPG)
Retrodoodle (ORIVET)
Labrador Retriever (ORIVET, PPG)
*included in EMBARK Breed + Health Kit

Pont TR et al. A Carbohydrate Sulfotransferase-6 (CHST6) gene mutation is associated with Macular Corneal Dystrophy in Labrador Retrievers. Vet Ophthalmol. 2016; 19:488-492
Busse C et al. Phenotype of macular corneal dystrophy in Labrador Retrievers: A multicenter study. Vet Ophthalmol. 2019; 22(3):294-304

LENS

Hereditary Cataract, Early-onset Cataract, Juvenile Cataract (Boston Terrier Type)
GENE: Heat Shock Transcription Factor 4 (HSF4)

MUTATION: 1 bp insertion (insC)
American Bully (ORIVET)
Australian Labradoodle (PPG)
Boston Terrier (AG, ANTAGENE, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, PV, UCD)
French Bulldogs (AG, GENSOL, LABOKLIN, ORIVET, PPG, PV, UCD)
Goldendoodle (PPG)
Labradoodle (PPG)
Miniature Poodle (PPG)
Shorty Bull (ORIVET, PPG)
Staffordshire Bull Terrier (AG, ANTAGENE, GENEFAST, GENSOL, LABOKLIN, ORIVET, PGG, PV, UCD)
Standard Poodle (PPG)
Toy Poodle (PPG)
Wallace Bulldog (ORIVET)
* included in EMBARK Breed + Health Kit

Mellersh CS et al. Identification of mutations in HSF4 in dogs of three different breeds with hereditary cataracts. Vet Ophthalm. 2006; 9(5);369-378.
Mellersh CS et al. Mutation in HSF4 associated with early but not late-onset hereditary cataract in the Boston Terrier. J Hered. 2007;98(5):531-3

Hereditary Cataract, Early-onset Cataract, Juvenile Cataract (Australian Shepherd Type)
GENE: Heat Shock Transcription Factor 4 (HSF4)
MUTATION: 1 bp deletion (delC)
Aussiedoodle (PPG)
Australian Shepherd (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, UCD)
Miniature American Sheperd (PPG)
Miniature Australian Sheperd (LABOKLIN, PPG)
Toy Australian Sheperd (PPG)
Wäller (LABOKLIN)
* included in EMBARK Breed + Health Kit

Mellersh CS. et al. Identification of mutations in HSF4 in dogs of three different breeds with hereditary cataracts. Vet Ophthalm. 2006; 9(5);369-378.
Mellersh CS et al. Mutation in HSF4 is associated with hereditary cataract in the Australian Shepherd. Vet Ophthalmol. 2009; 12(6):372-8.

Hereditary Cataract in the Lagotto Romagnolo
GENE:
MUTATION:
Lagotto Romagnolo (TIHO**)
** for more information: http://www.greccio.de/seiten/katarakt.html

UNPUB

Primary Lens Luxation (PLL)
GENE: ADAM Metallopeptidase With Thrombospondin Type 1 Motif 17 (ADAMTS17)
MUTATION: Base Substitution G>A splice-donor-site mutation at the 5' end of intron 10
American Eskimo Dog (AG, GENEFAST, GENSOL, LABOKILN, ORIVET, PPG, UCD)
American Hairless (Rat) Terrier (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA)
Australian Cattle Dog (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKILN, ORIVET, PPG, RCGHA, UCD)
Australian Kelpie (ORIVET)
Australian Stumpy Tail Cattle Dog (ORIVET, PPG)
Australian Terrier (ORIVET)
Biewer (GENSOL, PPG)
Border Collie (ORIVET)
Bull Terrier (ORIVET, PPG)
Carlin Pinscher (PPG)
Chinese Crested (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG ,RCGHA, UCD)
Chinese Foo (AG, GENEFAST, GENSOL, PPG, TIHO, UCD)
Danish/Swedish Farmdog (LABOKLIN, PPG, RCGHA, UCD)
Fox Terrie (AG, GENEFAST, GENOMIA, LABOKLIN, PPG, RCGHA, UCD)
Harlequin Pinscher (ORIVET)
Irish Terrier (ORIVET)
Italian Greyhound (UCD)
Jack Russell Terrier (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, TIHO, UCD)
Jagd Terrier (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, UCD)
Koolie (ORIVET, PPG)
Lakeland Terrier (AG, ANTAGENE, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA)
Lancashire Heeler (AG, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, TIHO, UCD)
Lucas Terrier (AG, GENEFAST, GENSOL, LABOKLIN, PPG)
Miniature Bull Terrier (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, TIHO, UCD)
Miniature Fox Terrier (ORIVET, PPG)
Norfolk Terrier (GENEFAST, GENSOL, LABOKLIN, PPG)
Norwich Terrier (AG, GENEFAST, GENSOL, LABOKLIN, PPG, RCGHA)
Parson Russell Terrier (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, TIHO, UCD)
Patterdale Terrier (AG, GENOMIA, GENEFAST, LABOKLIN, PPG, RCGHA, TIHO, UCD)
Pug (AG GENEFAST)
Puli (PPG)

Pumi (PPG, RCGHA)
Rat Terrier (AG, GENOMIA, GENEFAST, GENSOL, ORIVET, PPG, RCGHA, TIHO, UCD)
Sealyham Terrier (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, TIHO, UCD)
Shar Pei (ORIVET)
Teddy Roosvelt Terrier (AG, GENEFAST, LABOKLIN, PPG)
Tenterfield Terrier (AG, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCHGA, UCD)
Tibetan Spaniel (ORIVET)
Tibetan Terrier (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, TIHO, UCD)
Toy Fox Terrier (AG, GENOMIA, GENEFAST, GENSOL , LABOKLIN, PPG, RCGHA, TIHO, UCD)
Volpino Italiano (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, TIHO, UCD)
Welsh Terrier (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, TIHO, UCD)
Westphalia Terrier (GENEFAST)
Yorkshire Terrier (AG, GENOMIA, GENEFAST, GENSOL, LABOKLIN, PPT, RCGHA, UCD)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Farias FH et al. An ADAMTS17 splice donor site mutation in dogs with primary lens luxation. Invest Ophthalmol Vis Sci. 2010; 51(9);4716-4721.
Gould D et al. ADAMTS17 mutation associated with primary lens luxation is widespread among breeds. Vet Ophthalmol. 2011; 14: 378-384

Primary Open Angle Glaucoma and Primary Lens Luxation POAG-PLL
SEE: GLAUCOMA

GLAUCOMA

Goniodysgenesis and Glaucoma
GENE: Olfactomedin Like 3 (OLFML3)
MUTATION: Base Substitution G>A
Border Collie (AG, GENOMIA, GENSOL, LABOKLIN, ORIVET)
Koolie (ORIVET)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Pugh CA et al. Arginine to glutamine mutation in olfactomedin-like 3 (OLFML3) is a candidate for severe goniodysgenesis and glaucoma in the Border Collie dog breed. G3 (Bethesda). 2019; 9(3):943-954

Primary Closed Angle Glaucoma (PCAG)

GENE:
MUTATION: 2 linked variants have been identified
Italian Greyhound (UCD)

UNPUB (by UCD Davis canine researchers Dr. Niels Pedersen and Hongwei Liu)

Primary Open Angle Glaucoma POAG (Basset Fauve de Bretagne Type)
GENE: ADAM Metallopeptidase With Thrombospondin Type 1 Motif 17 (ADAMTS17)
MUTATION: Missense Mutation (G519S) in exon 11
Basset Fauve de Bretagne (GENEFAST, LABOKLIN, PPG, RCGHA)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Oliver JA et al. Two Independent Mutations in ADAMTS17 Are Associated with Primary Open Angle Glaucoma in the Basset Hound and Basset Fauve de Bretagne Breeds of Dog. PLoS One. 2015; 10: e0140436.

Primary Open Angle Glaucoma POAG (Basset Hound Type)
GENE: ADAM Metallopeptidase With Thrombospondin Type 1 Motif 17 (ADAMTS17)
MUTATION: 19 bp deletion in exon 2
Basset Hound (GENEFAST, LABOKLIN, PPG)
* included in EMBARK Breed + Health Kit

Oliver JA et al. Two Independent Mutations in ADAMTS17 Are Associated with Primary Open Angle Glaucoma in the Basset Hound and Basset Fauve de Bretagne Breeds of Dog. PLoS One. 2015; 10: e0140436.

Primary Open Angle Glaucoma POAG (Beagle Type)
GENE: ADAM Metallopeptidase With Thrombospondin Ype 1 Motif 10 (ADAMTS10)
MUTATION: Base Substitution G>A
Beagle (AG, GENEFAST, GENOMIA, LABOKLIN, ORIVET, UCD)
* included in EMBARK Breed + Health Kit

Kuchtey J et al. Mapping of the disease locus and identification of ADAMTS10 as a candidate gene in a canine model of primary open angle glaucoma. PLoS Genet. 2011; 7(2):e1001306

Primary Open Angle Glaucoma POAG (Norwegian Elkhound Type)
GENE: ADAM Metallopeptidase With Thrombospondin Ype 1 Motif 10 (ADAMTS10)
MUTATION: missense mutation (p.A387T) in exon 9
Norwegian Elkhound (LABOKLIN, ORIVET, PPG)
* included in EMBARK Breed + Health Kit

Ahonen SJ et al. A novel missense mutation in ADAMTS10 in Norwegian Elkhound primary glaucoma. PLoS One. 2014; 9: e111941

Primary Open Angle Glaucoma POAG (Petit Basset Griffon Vendéen Type)
GENE: ADAM Metallopeptidase With Thrombospondin Type 1 Motif 17 (ADAMTS17)
MUTATION: Inversion Mutation
Petit Basset Griffon Vendéen (GENOMIA, RCGHA)
* included in WISDOM PANEL Premium Kit

Bedford PG. Open-angle glaucoma in the Petit Basset Griffon Vendeen. Vet Ophthalmol. 2017; 20:98-102

Primary Open Angle Glaucoma and Primary Lens Luxation POAG-PLL
GENE: ADAM Metallopeptidase With Thrombospondin Type 1 Motif 17 (ADAMTS17)
MUTATION: 6 bp deletion (delCGTGGT) in exon 22
Sharpei (ANTAGENE, GENOMIA, LABOKLIN, TIHO)
* included in WISDOM PANEL Premium Kit

James AC et al. A novel ADAMTS17 mutation is associated with primary open angle glaucoma and primary lens luxation in the Shar Pei; The 9th international conference on canine and feline genetics and genomics

FUNDUS/RETINA (NON-SYNDROMIC)

Basenji Progressive Retinal Atrophy (Bas_PRA)
GENE: S-Antigen Visual Arrestin (SAG)
MUTATION: Base Substitution T>C
Basenji (ANTAGENE, GENOMIA, LABOKLIN, ORIVET, PPG, RCGHA)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Goldstein O et al. A non-stop S-antigen gene mutation is associated with late onset hereditary retinal degeneration in dogs. Mol Vis 2013; 19;1871-1884.

Canine Multifocal Retinopathy (CMR1)
GENE: Bestrophin 1 (BEST1)
MUTATION: Base Substitution C>T in exon 2
American Bulldog (AG, ANTAGENE, GENOMIA, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, UCD)
American Bully (PPG, UCD)
American Pit Bul Terrier (RCGHA)

Aussiedoodle (PPG, UCD)
Australian Bulldog (GENEFAST, ORIVET)
Australian Koolie (PPG, UCD)
Australian Shepherd (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, PV, RCGHA, UCD)
Boerboel (ANTAGENE, LABOKLIN, ORIVET, PPG, RCGHA, UCD)
Brazilian Terrier (PPG, UCD)
British Bulldog (ORIVET)
Bullmastiff (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, PV, RCGHA, UCD)
Cane Corso Italiano (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, UCD)
Dogo Canario (AG, ANTAGENE, GENOMIA, GENSOL, LABOKLIN, PPG, RCGHA)
Dogue de Bordeaux (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, PV, RCGHA, UCD)
English Bulldog (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, PPG, PV, RCGHA, UCD)
French Bulldog (GENEFAST, LABOKLIN, ORIVET, PPG, PV, RCGHA, UCD)
Great Pyrenees (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, PV, RCGHA, UCD)
Havanese (ORIVET, UCD)
Koolie (ORIVET, PPG, UCD)
Mastiff (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, PPG, PV, RCGHA, UCD)
Miniature American Sheperd (ANTAGENE, PPG, RCGHA, UCD)
Miniature Australian Sheperd (LABOKLIN, PPG, UCD)
Old English Mastiff (AG, GENOMIA, GENEFAST, GENSOL, ORIVET, PV, UCD)
Shorty Bull (PPG, UCD)
Toy Australian Sheperd (PPG, UCD)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Guziewicz KE et al. Bestrophin gene mutations cause canine multifocal retinopathy: a novel animal model for best disease. Invest Ophthalmol Vis Sci. 2007; 48(5);1959-1967.
Hoffmann I et al. Canine multifocal retinopathy in the Australian Shepherd: a case report. Vet Ophthalmol. 2012; 15 Suppl 2: 134-138.
Gornik KR et al. Canine multifocal retinopathy caused by a BEST1 mutation in a Boerboel. Vet Ophthalmol. 2014; 17: 368-372.

Canine Multifocal Retinopathy (CMR2)
GENE: Bestrophin 1 (BEST1)
MUTATION: Base Substitution G>A in exon 5
Coton De Tulear (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA, UCD)
* included in WISDOM PANEL Premium Kit

* included in EMBARK Breed + Health Kit

Guziewicz KE et al. Bestrophin gene mutations cause canine multifocal retinopathy: a novel animal model for best disease. Invest Ophthalmol Vis Sci. 2007;48(5);1959-1967.

Canine Multifocal Retinopathy (CMR3)
GENE: Bestrophin 1 (BEST1)
MUTATION: Base Substitution and Nucleotide deletion (G>T and delC)
Finnish Lapphund (LABOKLIN, ORIVET, PPG, UCD)
Lapponian Herder (LABOKLIN, ORIVET, PPG, RCGHA, UCD)
Swedish Lapphund (LABOKLIN, ORIVET, PPG, UCD)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Zangerl B et al. Assessment of canine BEST1 variations identifies new mutations and establishes an independent bestrophinopathy model (cmr3). 2010; Mol Vis, 16:2791-2804.

Collie Eye Anomaly-Choroidal Hypoplasia (CEA-CH)
GENE: Non-Homologous End Joining Factor 1 (NHEJ1)
MUTATION: 7799 bp deletion in Intron 4
Aussiedoodle (GENSOL, PPG)
Australian Kelpie (LABOKLIN, ORIVET)
Australian Koolie (PPG)
Australian Shepherd (AG, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG)
Bearded Collie (GENSOL, LABOKLIN, ORIVET, PPG)
Border Collie (AG, ANAGENE, GENOMIA, GENEFAST, GENSOL, ORIVET, PPG)
Bordoodle (PPG)
Boykin Spaniel (GENSOL, LABOKLIN, ORIVET, PPG)
Collie Rough “Long Haired” (AG, ANAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG)
Collie Smooth “Short Haired” (AG, ANAGENE, GENOMIA, GENSOL, GENEFAST, LABOKLIN, ORIVET, PPG)
English Sheperd (GENEFAST, GENSOL, PPG)
Hokkaido (GENSOL, LABOKLIN, PPG)
Koolie (ORIVET, PPG)
Lancashire Heeler (AG, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG)
Long-haired Wippet (GENSOL, LABOKLIN, PPG)
Miniature American Shepherd (ANAGENE, GENEFAST, GENSOL, ORIVET, PPG)
Miniature Australian Sheperd (GENEFAST, GENSOL, LABOKLIN, PPG)
Nova Scotia Duck Tolling Retriever (ANTAGENE, GENOMIA, GENSOL, LABOKILN, PPG)
Old English Sheepdog (ORIVET)
Old-Time Scotch Collie (PPG)
Scottish Collie (PPG)

Shetland Sheepdog (AG, ANAGENE, GENOMIA, GENSOL, LABOKLIN, ORIVET, PPG)
Silken Windhound (GENSOL, LABOKLIN, PPG)
Smithfield, Whippet (ORIVET)
Toy Australian Sheperd (GENSOL, PPG)
Whippet (GENEFAST)
* included in EMBARK Breed + Health Kit

Lowe JK et al. Linkage mapping of the primary disease locus for collie eye anomaly. Genomics. 2003; 82: 86-95
Parker HG et al. Breed relationships facilitate fine-mapping studies: A 7.8-kb deletion cosegregates with Collie eye anomaly across multiple dog breeds. Genome Res. 2007; 17: 1562-1571
Mizukami K et al. Collie eye anomaly in Hokkaido dogs: case study. Vet Ophthalmol. 2012; 15: 128-132.

Cone Degeneration (CD-Achromatopsia)
GENE: Cyclic Nucleotide Gated Channel Subunit Alpha 3 (CNGA3)
MUTATION: chr10:44234861-44234861: C/T
German Shepherd (LABOKLIN, RCGHA, PPG)
Shilou Sheperd (PPG)
White Sheperd Dog (PPG)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Tanaka N et al. Canine CNGA3 Gene Mutations Provide Novel Insights into Human Achromatopsia-Associated Channelopathies and Treatment. PLoS One. 2015; 10(9):e0138943.

Cone Degeneration 1 (CD1-Achromatopsia)
GENE: Cyclic Nucleotide Gated Channel Subunit Beta 3 (CNGB3)
MUTATION: Whole-gene deletion, at least 140 kb
Alaskan Husky (PPG)
Alaskan Malamute (AG, GENOMIA, ORIVET, PPG, RCGHA, UCD)
Alaskan Sled Dog (PPG, UCD)
Aussidoodle (PPG)
Australian Shepherd (AG, GENOMIA, ORIVET, PPG, RCGHA, UCD)
German Shorthaired Pointer (AG**, GENOMIA**, UCD**)
Labradoodle (ORIVET)
Miniature American Shepherd (ORIVET, PPG, RCGHA)
Miniature Australian Sheperd (GENOMIA, PPG, UCD)
Pomsky (PPG)
Samoyedo (GENOMIA)
Siberian Husky (GENOMIA, ORIVET, PPG, UCD)
Toy Australian Sheperd (PPG)

*included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit
**not specified if a different mutation is searched in this breed by this Lab

Sidjanin DJ et al. Canine CNGB3 mutations establish cone degeneration as orthologous to the human achromatopsia locus ACHM3. Human Molecular Genetics. 2002; 11: 1823-1833.
Yeh et al. Genomic deletion of CNGB3 is identical by descent in multiple canine breeds and causes achromatopsia. BMC Genetics. 2013; 14:27.

Cone Degeneration 2 (CD2-Achromatopsia)
GENE: Cyclic Nucleotide Gated Channel Subunit Beta 3 (CNGB3)
MUTATION: Base Substitution G>A
Alaskan Husky (PPG)
Alaskan Sled Dog (PPG)
Deutsch Kurzhaar (PPG)
German Shorthaired Pointer (ANTAGENE, ORIVET, PPG, RCGHA)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Sidjanin JD et al. Canine CNGB3 mutations establish cone degeneration as orthologous to the human achromatopsia locus ACHM3. 2002; Hum Mol Genet, 11;16, 1823-1833.

Cone Degeneration Labrador Type (Achromatopsia)
GENE: Cyclic Nucleotide Gated Channel Subunit Alpha 3 (CNGA3)
MUTATION: 3bp deletion (del TGG)
Australian Labradoodle (PPG)
Labradoodle (LABOKLIN, PPG)
Labrador Retriever (LABOKLIN, PPG)
* included in EMBARK Breed + Health Kit

Tanaka N et al. Canine CNGA3 Gene Mutations Provide Novel Insights into Human Achromatopsia-Associated Channelopathies and Treatment. PLoS One. 2015; 10(9):e0138943.

Cone-Rod Dystrophy 1 – crd1
GENEPhosphodiesterase 6B (PDE6B)
MUTATION: 3 bp deletion (del GTT)
American Bully (ORIVET, PPG)
American Pit Bull Terrier (ORIVET, PPG)
American Staffordshire Terrier (LABOKLIN, ORIVET, PPG, RCGHA, UCD)
* included in WISDOM PANEL Premium Kit

* included in EMBARK Breed + Health Kit

Goldstein O et al. IQCB1 and PDE6B mutations cause similar early onset retinal degenerations in two closely related terrier dog breeds. Invest Ophthalmol Vis Sci. 2013; 54(10);7005-7019.

NOTE: PDE6B gene is only expressed in rods and not in cones, the disease is indeed a rod cone dysplasia and therefore should be renamed

Cone-Rod dystrophy 2 – crd2
GENE**:** IQ Motif Containing B1 (IQCB1)
MUTATION: 1 bp insertion (ins C)
Anerican Bully (PPG)
American Pit Bull Terrier (LABOKLIN, PPG, RCGHA, UCD)
American Staffordshire Terrier (PPG)
* included in WISDOM PANEL Premium Kit

Goldstein O et al. IQCB1 and PDE6B mutations cause similar early onset retinal degenerations in two closely related terrier dog breeds. Invest Ophthalmol Vis Sci. 2013; 54(10);7005-7019.

Cone-Rod dystrophy 3 – crd3
GENE**: ADAM Metallopeptidase Domain 9 (**ADAM9)
MUTATION: 40 kb deletion
Glen of Imaal Terrier (PPG)

Goldstein O et al. An ADAM9 mutation in canine cone-rod dystrophy 3 establishes homology with human cone-rod dystrophy 9. Mol Vis. 2010 ;16:1549-69
Kropatsch R et al. Generalized progressive retinal atrophy in the Irish Glen of Imaal Terrier is associated with a deletion in the ADAM9 gene. Mol Cell Probes. 2010; 24(6):357-63

Cone-Rod Dystrophy 4 (crd4/CORD1) **NOT ADVISED**
GENE: RPGR Interacting Protein 1 (RPGRIP1)
MUTATION: 44 bp insertion or 59 bp insertion in exon 2
American Bulldog (ORIVET)
American Bully (ORIVET)
Australian Cobberdog (ORIVET, PPG)
Australian Labradoodle (ORIVET, PPG)
Beagle (LABOKLIN, ORIVET, PPG)
Bolnka Zwenta (LABOKLIN)
Boykin Spaniel (PPG)
Carlin Pinscher (PPG)

Chihuahua (PPG)
Clumber Spaniel (LABOKLIN)
Curly Coated Retriever (GENEFAST, GENOMIA, LABOKLIN, ORIVET)
Dachshund Standard Long/Short/Smooth/Wire Hair (GENSOL, LABOKLIN, ORIVET, PPG)
English Springer Spaniel (GA, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG)
Field Spaniel (PPG)
French Bulldog (ORIVET, PPG)
Kaninchen Dachshund (ORIVET)
Labradoodle (ORIVET, PPG)
Labrador Retriever (PPG)
Miniature Dachshund Long/Short/Smooth/Wire Hair (AG, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG)
Papillon (ORIVET)
Pembroke Welsh Corgi (ORIVET)
Portuguese Podengo Pequeno (PPG)
Retrodoodle (ORIVET)
Short Bully (ORIVET, PPG)
Spoodle (ORIVET)
* included in EMBARK Breed + Health Kit

Mellersh CS et al. Canine RPGRIP1 mutation establishes cone–rod dystrophy in miniature longhaired dachshunds as a homologue of human Leber congenital amaurosis. 2006; Genomics, 88(3);293-301.
Kuznetsova T et al. Exclusion of RPGRIP1 ins44 from Primary Causal Association with Early-Onset Cone-Rod Dystrophy in Dogs. Investigative Ophthalmology and Visual Science. 2012; 53: 5486-5501.
Forman OP et al.. Canine genome assembly correction facilitates identification of a MAP9 deletion as a potential age of onset modifier for RPGRIP1-associated canine retinal degeneration. Mamm Genome. 2016; 27: 237-245.
Miyadera et al. Phenotypic variation and genotypephenotype discordance in canine cone-rod dystrophy with an RPGRIP1 mutation. Mol Vis. 2009; 15: 2287-2305. 64.
Miyadera K et al. Genome-wide association study in RPGRIP1(-/-) dogs identifies a modifier locus that determines the onset of retinal degeneration. Mamm Genome. 2012; 23: 212-223.
Narfstrom K et al. Assessment of hereditary retinal degeneration in the English springer spaniel dog and disease relationship to an RPGRIP1 mutation. Stem Cells Int. 2012; 2012 685901.

NOTE: genetic changes underlying CORD1 are more complex than initially thought. Not all dogs in the pet population with RPGRIP1 mutation developed clinical signs of PRA. A second modifying locus influencing the expression of the RPGRIP1 mutation was then identified. Ongoing studies are attempting to fully understand the genotype-phenotype correlation and interaction between the two gene variations. In some breeds the RPGRIP1 mutation does not segregate with PRA and therefore the RPGRIP1 genotyping test needs to be interpreted with caution.

Cone-Rod Dystrophy (crd SWD, CORD2)
GENE: Nephrocystin 4 (NPHP4)
MUTATION: 180 bp deletion (exon 5/intron 5)
Miniature Dachshund Wire Hair (GENEFAST, ORIVET, PPG, RCGHA, TIHO)
Dachshund Standard Long/Short/Smooth/Wire Hair (GENEFAST, GENOMIA, LABOKLIN, PPG, RCGHA, TIHO)
* included in WISDOM PANEL Premium Kit

Wiik AC et al. A deletion in nephronophthisis 4 (NPHP4) is associated with recessive cone-rod dystrophy in standard wire-haired dachshund. Gen Res. 2008; 18(9);1415-1421.
Wiik AC et al. A population study of a mutation allele associated with cone-rod dystrophy in the standard wire-haired dachshund. Anim Genet. 2009 ; 40(4):572-4

RPE65 retinal dystrophy)
GENE: Retinoid Isomerohydrolase RPE65 (RPE65)
MUTATION: 4 bp deletion (del AAGA) in exon 5
Beagle (ORIVET)
Briard (ANTAGENE, GENOMIA, LABOKLIN, ORIVET, PPG)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit
(RPE65 mutation in Briards was original classified as a congenital stationary night blindness. However it is not a real congenital stationary night blindness, the condition also affects day vision and is slowly progessive. Therefore the disease should be renamed)

Narfström K et al.The Briard dog: A new animal model of congenital stationary night blindness. Ophthalmology 1998; 73:750-756.
Aguirre GD et al. Congenital stationary night blindness in the dog: common mutation in the RPE65 gene indicates founder effect. Molecular Vision. 1998; 4: 23.
Veske A et al. Retinal dystrophy of swedish Briard/Briard-beagle dogs is due to a 4-bp deletion in RPE65. Genomics. 1999; 57: 57-61.

Congenital Stationary Night Blindness 2 (CSNB)
GENE: Leucine Rich Repeat, Ig-Like and Transmembrane Domains 3 (LRIT3)
MUTATION: 1 bp deletion
* included in EMBARK Breed + Health Kit

Das RG et al. Genome-wide association study and whole-genome sequencing identify a deletion in *LRIT3* associated with canine congenital stationary night blindness. Sci Rep. 2019;9(1):14166

Dominant Progressive Retinal Atrophy (PRA-D)

GENE: Rhodopsin (RHO)
MUTATION: Base Substitution C>G
Bullmastiff (AG, ANAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA)
Mastiff (AG, ANAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA)
Old English Mastiff (AG, GENOMIA, GENEFAST, ORIVET)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Kijas JW et al. Naturally occurring rhodopsin mutation in the dog causes retinal dysfunction and degeneration mimicking human dominant retinitis pigmentosa. Proc Natl Acad Sci USA. 2002; 99: 6328-6333
Kijas JW et al. Canine Models of Ocular Disease: Outcross Breedings Define a Dominant Disorder Present in the English Mastiff and Bull Mastiff Dog Breeds. J Hered. 2003; 94;1, 27–30

Early-onset PRA (Portuguese Water Dog)
GENE: Colied-Coil Domain Containing 66 (CCDC66)
MUTATION: 1 bp insertion (ins A)
Portuguese Water Dog (LABOKLIN)
Spanish Water Dog (LABOKLIN)
* included in WISDOM PANEL Premium Kit

Murgiano L et al. CCDC66 frameshift variant associated with a new form of early-onset progressive retinal atrophy in Portuguese Water Dogs. Sci Rep. 2020;10(1):21162

Early Retinal Degeneration (arPRA-erd)
GENE: Serine/THronine Kinase 38 Like (STK38L)
MUTATION: 15 bp duplication (dup GGAAACAGAGTTCTT), 229 bp insertion
Norwegian Elkhound (ORIVET, PPG)
* included in WISDOM PANEL Premium Kit

Acland GM, Aguirre GD. Retinal degenerations in the dog: IV. Early retinal degeneration (erd) in Norwegian elkhounds. Exp Eye Res. 1987;44(4):491-521.
Goldstein O et al. Exonic SINE insertion in STK38L causes canine early retinal degeneration (erd). Genomics. 2010; 96(6):362-8.
Berta ÁI et al. Photoreceptor cell death, proliferation and formation of hybrid rod/S-cone photoreceptors in the degenerating STK38L mutant retina. PLoS One. 2011; 6(9):e24074.

Golden Retriever Generalised PRA 1 (GR_PRA 1)
GENE: Solute Carrier Family 4 Member 3 (SLC4A3)

MUTATION: 1 bp insertion (ins C)
Golden Retriever (AG, ANTAGENE, GENEFAST, GENOMIA, LABOKLIN, ORIVET, PPG, UCD)
Goldendoodle (ORIVET, PPG, UDC)
Groodle (ORIVET)
Labrador Retriever (UCD)
Lhasa Apso (PPG)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Downs et al. A frameshift mutation in golden retriever dogs with progressive retinal atrophy endorses SLC4A3 as a candidate gene for human retinal degenerations. PLoS One. 2011;6(6):e21452

Golden Retriever Generalised PRA 2 (GR_PRA 2)
GENE: Tetratricopeptide Repeat Domain 8 (TTC8)
MUTATION: 1 bp deletion (del A)
Australian Labradoodle (PPG)
Golden Retriever (AG, GENEFAST, GENOMIA, GENSOL, LABOKLIN, ORIVET, PPG, UCD)
Goldendoodle (ORIVET, PPG, UCD)
Groodle (ORIVET)
Labradoodle (PPG)
Labrador Retriever (PPG, UCD)
* included in EMBARK Breed + Health Kit

Downs LM et al. A novel mutation in TTC8 is associted with progressive retinal atrophy in the golden retriever. Canine Genetics and Epi. 2014; 1:4 1-12.

Papillon Progressive Retinal Atrophy 1 (Pap_PRA1)
GENE: Cyclic Nucleotide Gated Channel Subunit Beta 1 (CNGB1)
MUTATION: 1 bp deletion (del A) and 6 bp insertion
Papillon (ANTAGENE, GENEFAST, GENOMIA, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA)
Phalene (GENOMIA, LABOKLIN, PPG, RCGHA)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Winkler PA et al. A large animal model for CNGB1 autosomal recessive retinitis pigmentosa. PLoS One. 2013; 8(8); e72229.
Ahonen SJ et al. A CNGB1 frameshift mutation in Papillon and Phalene dogs with progressive retinal atrophy. PLoS One. 2013; 8: e72122.

Progressive Retinal Atrophy (Giant Schnauzer Type)
GENE**:** NECAP Endocytosis Associated 1 (NECAP1)
MUTATION: Base Substitution c.544G> A (p.Gly182Arg).

Giant Schnauzer (GENOMIA, LABOKLIN)

Hitti RJ et al. Whole Genome Sequencing of Giant Schnauzer Dogs with Progressive Retinal Atrophy Establishes NECAP1 as a Novel Candidate Gene for Retinal Degeneration. Genes (Basel). 2019; 10(5): 385

Progressive Retinal Atrophy IG1 (Italian Greyhound Type, sub-divided in PRA-IG1a, 1b, and 1c based on risk genotypes)
GENE:
MUTATION: associated variants at 5 loci have been identified
Italian Greyhound (UCD)
* included in WISDOM PANEL Premium Kit

UNPUB (by UCD Davis canine researchers Dr. Niels Pedersen and Hongwei Liu)

Progressive Retinal Atrophy (Lhasa Apso Type)
GENE: Interphotoreceptor Matrix Proteoglycan 2 (IMPG2)
MUTATION: line-1 insertion
Lhasa Apso
*included in WISDOM PANEL Premium Kit

Hitti-Malin RJ et al. A LINE-1 insertion situated in the promoter of IMPG2 is associated with autosomal recessive progressive retinal atrophy in Lhasa Apso dogs. BMC Genet. 2020;21(1):100.

Progressive Retinal Atrophy (Puli Type)
GENE: Bardet-Biedl Syndrome 4 (BBS4)
MUTATION: Base Substitution A>T
Puli (ORIVET)

Chew T et al. A Coding Variant in the Gene Bardet-Biedl Syndrome 4 (*BBS4*) Is Associated with a Novel Form of Canine Progressive Retinal Atrophy. G3 (Bethesda). 2017; 5;7(7):2327-2335.

Progressive Retinal Atrophy g-PRA (Schapendoes Type)
GENE: Colied-Coil Domain Containing 66 (CCDC66)
MUTATION: 1 bp insertion in exon 6
Schapendoes Dog (GENEFAST, LABOKLIN, RCGHA)
* included in WISDOM PANEL Premium Kit

Dekomien G et al. Progressive retinal atrophy in Schapendoes dogs: mutation of the newly identified CCDC66 gene. Neurogenetics. 2010; 11(2):163-174

Progressive Retinal Atrophy (Shetland Sheepdog Type-BBS2 variant)
GENE: Bardet-Biedl Syndrome 2 (BBS2)
MUTATION: Base Substitution G>C
Shetland Sheepdog
* included in WISDOM PANEL Premium Kit

Hitti-Malin RJ. Elucidating the Genetic Basis of Canine Progressive Retinal Atrophies in Several Breeds of Dog (Doctoral thesis 2020). ttps://doi.org/10.17863/CAM.58690

Progressive Retinal Atrophy (Shetland Sheepdog Type-CNGA1 variant)
GENE: Cyclic Nucleotide Gated Channel Subunit Alpha 1 (CNGA1)
MUTATION:4 bp deletion (delAACT) in exon 9
Shetland Sheepdog (AG, ANTAGENE, GENOMIA, LABOKLIN, ORIVET, PPG, RCGHA)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Wiik AC et al. Progressive retinal atrophy in Shetland sheepdog is associated with a mutation in the CNGA1 gene. Anim Genet. 2015; 46: 515-521.

Progressive Retinal Atrophy (Weimaraner Type, X-linked)
GENE: Retinitis Pigmentosa GTPase Regulator (RPGR)
MUTATION: first 4 exons deletion
Weimaraner (BOCHUM UNIVERSITY**)
**test not avaiable for commercial use (https://www.ruhr-uni-bochum.de/en)

Kropatsch R et al. A large deletion in RPGR causes XLPRA in Weimaraner dogs. Canine Genet Epidemiol. 2016; 3:7

Progressive Retinal Atrophy 3 (PRA3)
GENE: FAM161 Centrosomal Protein A (FAM161A)
MUTATION: nucleotide Insertion
Tibetan Terriers (GENOMIA, ORIVET, PPG, RCGHA)
Tibetan Spaniel (GENOMIA, PPG, RCGHA)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Downs LM et al. An Intronic SINE Insertion in FAM161A that Causes Exon-Skipping Is Associated with Progressive Retinal Atrophy in Tibetan Spaniels and Tibetan Terriers. PLoS One. 2014; 9(4)e93990.

Progressive Rod Cone Degeneration (prcd-PRA, arPRA)
GENE: Photoreceptor disc component (PRCD)
MUTATION: Base Substitution G>A
American Cocker Spaniel (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
American Eskimo Dog (AG, GENSOL, LABOKLIN, ORIVET, PPG)
American Hairless (Rat) Terrier (ANTAGENE,ORIVET, PPG)
Aussiedoodle (GENSOL, PPG)
Australian Cattle Dog (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
Australian Cobber Dog (AG, ORIVET, PPG)
Australian Labradoodle (ORIVET)
Australian Shepherd (AG, GENSOL, LABOKLIN, ORIVET, PPG)
Australian Silky Terrier (ANTAGENE, LABOKLIN)
Australian Stumpy Tail Cattle Dog (AG, GENSOL, LABOKLIN, ORIVET, PPG)
Australian Terrier (ORIVET)
Barbet (AG, ANTAGENE, PPG)
Bearded Collie (LABOKLIN)
Bernardoodle (ORIVET, PPG)
Biewel (GENSOL, PPG)
Black Russian Terrier (ORIVET)
Bolognese (LABOKLIN)
Bolonka Zwenta (AG, LABOKLIN, PPG)
Bordoodle (PPG)
Boykin Spaniel (GENSOL, PPG)
Cao de Agua (ANTAGENE)
Cavapoo (PPG)
Cavoodle (ORIVET)
Chesapeake Bay Retriever (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
Chihuahua (AG, GENSOL, LABOKLIN, ORIVET, PPG)
Chinese Crested (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
Cockapoo (AG, GENSOL)
Coton De Tulear (GENSOL, ORIVET)
English Cocker Spaniel (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
English Sheperd (AG, GENSOL, LABOKLIN, PPG)
Entlebuchen (AG, ANTAGENE, GENSOL, LABOKLIN, PPG)
Finnish Lapphund (AG, GENSOL, LABOKLIN, ORIVET, PPG)
French Water Dog (LABOKLIN)
German Spitz (AG, GENSOL, LABOKLIN, ORIVET)
Giant Schnauzer (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
Giant Spitz (LABOKLIN)
Golden Retriever (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
Goldendoodle (AG, GENSOL, ORIVET, PPG)
Groodle (ORIVET)
Hungarian Kuvasz (ORIVET)
Jack Russel Terrier (LABOKLIN)
Karelian Bear Dog (AG, GENSOL, LABOKLIN, PPG)
Koolie (ORIVET, PPG)

Kuvasz (AG, ANTAGENE, GENSOL, LABOKLIN, PPG)
Labradoodle (AG, GENSOL, ORIVET, PPG)
Labrador Retriever (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
Lagotto Romagnolo (LABOKLIN)
Lapponian Herder (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
Maltipoo (AG, PPG)
Markiesje (AG, GENSOL, LABOKLIN, PPG)
Miniature American Shepherd (AG, ANTAGENE, GENSOL, ORIVET, PPG)
Miniature Australian Sheperd (GENSOL, LABOKLIN, PPG)
Miniature Fox Terrier (ORIVET)
Miniature Poodle (AG, GENSOL, LABOKLIN, ORIVET, PPG)
Moodle (ORIVET)
Newfypoo (GENSOL, PPG)
Norwegian Elkhound (AG, ANTAGENE, GESOL, LABOKLIN, ORIVET, PPG)
Nova Scotia Duck Tolling Retriever (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
Old English Sheepdog (ORIVET)
Parson Russel Terrier (LABOKLIN)
Pembroke Welsh Corgi (AG)
Perro de Agua Espanol (ANTAGENE)
Poodle (GENSOL, LABOKLIN, ORIVET, PPG)
Pomeranian (ORIVET)
Poruguese Podengo Pequeno (PPG)
Portuguese Water Dog (AG, GENSOL, LABOKLIN, ORIVET, PPG)
Rat Terrier (ORIVET, PPG)
Russian Tsvetnaya Bolonka (PPG)
Schipperke (AG, GENSOL, LABOKLIN, ORIVET, PPG)
Schnoodle (PPG)
Sheepadoodle (PPG)
Silky Terrier (AG, GENSOL, ORIVET, PPG)
Spanish Water Dog (AG, GENSOL, LABOKLIN, PPG)
Spoodle (ORIVET)
Standard Poodle (AG, GENSOL, LABOKLIN, ORIVET, PPG)
Swedish Jamthund (AG)
Swedish Lapphund (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
Tenterfield Terrier (ORIVET)
Toy Australian Sheperd (GENSOL, PPG)
Toy Poodle (AG, GENSOL, LABOKLIN, ORIVET, PPG)
Wäller (LABOKLIN)
Yorkshire Terrier (AG, ANTAGENE, GENSOL, LABOKLIN, ORIVET, PPG)
*offered in many breeds by GENOMIA and performed by partner laboratory
* included in EMBARK Breed + Health Kit
* suggested for all canine breeds by GENEFAST
* suggested for all canine breeds by UCD

Acland GM et al. Linkage analysis and comparative mapping of canine progressive rod-cone degeneration (prcd) establishes potential locus homology with retinitis pigmentosa (RP17) in humans. Proc Natl Acad Sci U S A. 1998; 95: 3048-3053.
Acland GM et al. A novel retinal degeneration locus identified by linkage and comparative mapping of canine early retinal degeneration. Genomics. 1999; 59: 134-142.
Zangerl B et al. Identical mutation in a novel retinal gene causes progressive rod-cone degeneration in dogs and retinitis pigmentosa in humans. Genomics. 2006; 88: 551-563.

Retinopathy of the Swedish Vallhund
GENE: MER Proto-Oncogene, Tyrosine Kinase (MERTK)
MUTATION: line-1 insertion
Swedish Vallhund (RCGHA)
* included in WISDOM PANEL Premium Kit

Cooper AE et al. A Novel Form of Progressive Retinal Atrophy in Swedish Vallhund Dogs. PLoS ONE. 2014; 9(9): e106610
Ahonen SJ et al. Increased Expression of MERTK is Associated with A Unique Form of Canine Retinopathy. PloS ONE, 2014; 17;9(12): e114552
Everson R et al. An intronic LINE-1 insertion in MERTK is strongly associated with retinopathy in Swedish Vallhund dogs. PLoS One. 2017; 16;12(8):e0183021.

Rod-Cone Dysplasia 1 (arPRA - rcd1)
GENE: Phosphodiesterase 6B (PDE6B)
MUTATION: Base Substitution G>A
Irish setter (AG, ANTAGENE, GENEFAST, GENSOL, LABOKLIN, PPG, RCGHA, UCD)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Suber ML et al. Irish setter dogs affected with rod-cone dysplasia contain a nonsense mutation in the rod cGMP phosphodiesterase beta-subunit gene. Proceedings of the National Academy of Sciences of the United States of America. 1993; 90: 3968-3972.
Clements PJ et al. Confirmation of the rod cGMP phosphodiesterase beta subunit (PDE beta) nonsense mutation in affected rcd-1 Irish setters in the UK and development of a diagnostic test. Current Eye Research. 1993; 12: 861- 866

Rod-Cone Dysplasia 1a (arPRA - rcd1a)
GENE: Phosphodiesterase 6B (PDE6B)
MUTATION: 8 bp insertion (ins TGAAGTCC)
Sloughi (ANTAGENE, GENOMIA, LABOKLIN, ORIVET, PPG, RCGHA, UCD)
* included in WISDOM PANEL Premium Kit

Dekomien G et al. 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. 2000; 90: 261-267

Rod-Cone Dysplasia 2 (arPRA - rcd2)
GENE: RD3 Regulator of GUCY2D (RD3)
MUTATION: 22 bp insertion of exon 4
Collie Rough "Long Haired" (GENOMIA, LABOKLIN, PPG)
Collie Smooth "Short Haired" (GENOMIA, LABOKLIN, PPG)
Old-time Scotch Collie (PPG)
Scottish Collie (PPG)

Friedman JS et al. Premature truncation of a novel protein, RD3, exhibiting subnuclear localization is associated with retinal degeneration. Am J Hum Genet. 2006;79:1059-1070
Vilboux T et al. Progressive Retinal Atrophy in Border Collie: A new XLPRA (BMC Vet Res. 2008;3;4:10
Kukekova AV et al. Canine RD3 mutation establishes rod-cone dysplasia type 2 (rcd2) as ortholog of human and murine rd3. Mamm Genome. 2009;20(2):109-23

Rod-Cone Dysplasia 3 - rcd3
GENE: Phosphodiesterase 6A (PDE6A)
MUTATION: 1 bp deletion (del A)
Cardigan Welsh Corgi (AG, ANTAGENE, GENOMIA, GENEFAST, GENSOL, LABOKLIN, ORIVET, PPG, RCGHA)
Chinese Crested (ANTAGENE, GENOMIA, GENSOL, LABOKLIN, ORIVET, PPG)
Pembroke Welsh Corgi (GENSOL, LABOKLIN, PPG, RCGHA)
Pomeranian (GENOMIA, GENSOL, LABOKLIN, ORIVET, PPG)
Pomsky (PPG)
* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Petersen-Jones SM et al. cGMP phosphodiesterase-alpha mutation causes progressive retinal atrophy in the Cardigan Welsh corgi dog. Invest Ophthalmol Vis Sci. 1999; 40(8):1637-1644
Petersen-Jones SM, Entz DD. An improved DNA-based test for detection of the codon 616 mutation in the alpha cyclic GMP phosphodiesterase gene that causes progressive retinal atrophy in the Cardigan Welsh Corgi; Vet Ophthalm. 2002; 5, 2, 103-106

Rod-Cone Dysplasia 4 - rcd4
GENE: Photoreceptor Cilium Actin Regulator (PCARE) (formerly C2orf71)
MUTATION: 1 bp insertion (ins C)
Australian Cattle Dog (ANTAGENE, GENOMIA, LABOKLIN, PPG)

English Setter (ANTAGENE, GENOMIA, LABOKLIN, PPG, TIHO)
Goldendoodle (PPG)
Gordon Setter (ANTAGENE, GENOMIA, LABOKLIN, PPG, TIHO)
Irish Setter (ANTAGENE, GENOMIA, LABOKLIN, PPG, TIHO)
Japanese Spitz (PPG)
Labradoodle (PPG)
Llewelyn Setter (PPG)
Miniature Poodle (LABOKLIN, PPG)
Old Danish Pointer (LABOKLIN, PPG, TIHO)
Polish Lowland Shepdog (ANTAGENE, GENOMIA, LABOKLIN, PPG, TIHO)
Small Muensterlander (ANTAGENE, GENOMIA, LABOKLIN, PPG)
Standard Poodle (GENOMIA, LABOKLIN, PPG)
Tatra Sheperd Dog (LABOKLIN)
Tibetan Terrier (ANTAGENE, GENOMIA, LABOKLIN, PPG)
Toy Poodle (LABOKLIN, PPG)

Downs LM et al. Late-onset progressive retinal atrophy in the Gordon and Irish Setter breeds is associated with a frameshift mutation in C2orf71. Anim Genet. 2013; 44: 169-177
Svensson M et al. Progressive retinal atrophy in the Polski Owczarek Nizinny dog: a clinical and genetic study. Vet Ophthalmol. 2016; 19: 195-205.

Stargardt Disease, Juvenile Macular Degeneration (STGD)
GENE: ATP Binding Cassette Subfamily A Member 4 (ABCA4)
MUTATION: 1 bp insertion (ins C)
Labradoodle (PPG)
Labrador Retriever (LABOKLIN, PPG)
* included in WISDOM PANEL Premium Kit

Makelainen S et al. An ABCA4 loss-of-function mutation causes a canine form of Stargardt disease. PLoS Genet. 2019 ;15(3):e1007873.

Type A PRA, XLPRA2 (Miniature Schnauzer Type) (**NOT ADVISED**)
GENE: Phosducin (PDC) on Chromosome 7
MUTATION: Base Substitution C>G
Miniature Schnauzer (ORIVET)

Zhang Q et al. Characterization of canine photoreceptor phosducin cDNA and identification of a sequence variant in dogs with photoreceptor dysplasia. Gene. 1998; 215;231-239

NOTE: Studies suggested that the Phosducin variant described in Zhang et al was not causal for PRA in the Miniature Schnauzer and that Photoreceptor Dysplasia (Type A PRA) of Miniature Schnauzers was due to a mutation in RPGR which is on the X

chromosome. This gave rise to X-linked condition (XLPRA2) in which males were affected and carrier females had a milder disease. However this condition appears to be either extremely rare or non existant. The Phosducin gene variant does not appear to be disease-associated (Murgiano et al 2019).

Type B PRA (Miniature Schnauzer Type)
Variants in linked loci (HIVEP3/PPT1-PRA instead of Type B PRA or type 1 PRA to refer to this form of PRA of Miniature Schnauzers.
GENE: Palmitoyl-Protein Thioesterase 1 (PPT1)
Miniature Schnauzer (ORIVET)

Murgiano L et al. Complex Structural PPT1 Variant Associated with Non-syndromic Canine Retinal Degeneration: GENES, GENOMES, GENETICS. 2019; 9:2, 425-437

NOTE: Only whole-genome sequencing can be utilized to reliably genotype PPT1 and determine disease status. Test is no longer available in other Labs and it has been replaced with a test using a linked HIVEP3 variant (below)

Type 1 PRA (Miniature Schnauzer Type). Note this appears to the the same as Type-B PRA above.
Currently it is suggested that this form of PRA in Miniature Schnauzers is called HIVEP3/PPT1-PRA instead of Type B PRA or type 1 PRA.
GENE: HIVEP Zinc Finger 3 (HIVEP3)
MUTATION: Base Substitution G>A
Miniature Schnauzer (LABOKLIN, ORIVET, GENOMIA)

Kaukonen M et al. A putative silencer variant in a spontaneous canine model of retinitis pigmentosa. PLoS Genet. 2020; 16(3):e1008659

NOTE: The HIVEP3 variant is currently used for testing Miniature Schnautzer with this form of PRA although the HIVEP3 variant could be a linked marker. The linked PPT1 variant is not suited for use in a commercial test. Further studies are needed to see if either of the PPT1 or HIVEP3 variants are disease-causing or whether a different closely linked variant is causal. Until there is further clarification it is recommended that the HIVEP3 variant is used in DNA testing.
Aguirre GD, Lohi H, Kaukonen M, Murgiano L. Formal commentary. *PLoS Genet*. 2020; 16: e1009059.

X-Linked PRA1
GENE: Retinitis Pigmentosa GTPase Regulator (RPGR) on Chromosome X
MUTATION: 5 bp beletion (delGAGAA) in ORF15 exon
Pomsky (ANTAGENE)
Samoyed (ANTAGENE, GENOMIA, GENEFAST, LABOKLIN, ORIVET, PPG)
Siberian Husky (ANTAGENE, GENOMIA, GENEFAST, LABOKLIN, ORIVET, PPG)

* included in WISDOM PANEL Premium Kit
* included in EMBARK Breed + Health Kit

Zeiss CJ et al. Retinal pathology of canine X-linked progressive retinal atrophy, the locus homologue of RP3. Investigative Ophthalmology and Visual Science. 1999; 40: 3292-3304.
Zeiss CJ et al. Mapping of X-linked progressive retinal atrophy (XLPRA), the canine homolog of retinitis pigmentosa 3 (RP3). Human Molecular Genetics. 2000; 9: 531-537.
Zhang Q, et al. Different RPGR exon ORF15 mutations in Canids provide insights into photoreceptor cell degeneration. Hum Mol Genet. 2002; 11(9);993-1003.

X-Linked PRA2
GENE: Retinitis Pigmentosa GTPase Regulator (RPGR)
MUTATION: 2bp del in RPGR: c. 1084–1085delGA
Mixed breed (RCGHA) – derived from Miniature Schnauzer with Type A PRA (see above)
* included in WISDOM PANEL Premium Kit

Zangerl B et al. Independent Origin and Restricted Distribution of RPGR Deletions causing XLPRA. J Hered. 2007; 98(5):526-530
Summarized in Introduction to Murgiano L et al. Complex Structural PPT1 Variant Associated with Non-syndromic Canine Retinal Degeneration: GENES, GENOMES, GENETICS. 2019; 9:2, 425-437

DNA Testing Laboratories

ANIMAL GENETICS (AG):
https://www.animalgenetics.us/Canine/Genetic_Disease/Disease_Index.asp

ANTAGENE: https://www.antagene.com/it/sante/test-genetique

EMBARK: https://embarkvet.com/products/dog-health/health-conditions/
* offers a "Breed + Health" Dog DNA Kit including tests for detecting genetic conditions affecting eyes and vision

GENEFAST:
https://www.genefast.com/downloads/categorie/file/PRICE%20LIST%202018%20rev.0.pdf

GENOMIA: https://www.genomia.cz/en/veterinarni/psi/#section0

GENSOL: https://www.gensoldx.com/test-information/

LABOKLIN: https://laboklin.com/en/products/genetics/hereditary-diseases/dog/

ORIVET: https://www.orivet.com/store/canine-disease

PAW PRINT GENETICS (PPG): https://www.pawprintgenetics.com/products/tests/index/

PENN VET (PV): https://www.vet.upenn.edu/research/academic-departments/clinical-sciences-advanced-medicine/research-labs-centers/penngen/penngen-tests/genetic-tests

ROYAL CANIN Genetic Health Analysis (RCGHA): http://marsveterinary.force.com/royalcaningha/

UC DAVIS (UCD): https://vgl.ucdavis.edu/tests?field_species_target_id=231

UNIVERSITY OF HANNOVER (TIHO) : https://www.tiho-hannover.de/en/kliniken-institute/institute/institute-for-animal-breeding-and-genetics/services/genetic-testing/genetic-testing-dog

WISDOM PANEL: https://www.wisdompanel.com/en-us/products
*offers a "Premium" Dog DNA Collection Kit including tests for detecting genetic conditions affecting eyes and vision