OAKLEY

HEALTH REPORT

Ichthyosis (PNPLA1)

Oakley inherited one copy of the variant we tested

What does this result mean?
This result should not impact Oakley's health but it could have consequences for siblings or other related dogs if they inherited two copies of the variant. We recommend discussing this result with their owners or breeders if you are in contact.

Impact on Breeding
Your dog carries this variant and will pass it on to ~50% of his offspring.

What is Ichthyosis?
This skin disorder gets its name from the thick, darkly pigmented scales of skin ("ichthys" is Greek for "fish") that affected dogs display on their noses, paw pads, and muzzles.

When signs & symptoms develop in affected dogs
As puppies, affected dogs can show signs of scaling. This disease tends to worsen with age.

How vets diagnose this condition
Examining the characteristic lesions is the first step in diagnosing Ichthyosis. Confirmatory genetic testing and/or skin biopsies can also be performed.

How this condition is treated
There is no definitive treatment for ichthyosis: typically, ichthyotic dogs are maintained on a continuous treatment of mild anti-dandruff shampoos and moisturizing rinses. This is a chronic and frustrating condition to manage.

Actions to take if your dog is affected

- Following your veterinarian's advice on skin care and nutrition is the best way to manage ichthyosis.

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BREED-RELEVANT CONDITIONS TESTED

Oakley did not have the variants that we tested for, that are relevant to his breeds:

- Von Willebrand Disease Type I (VWF)
- Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cavalier King Charles Spaniel Variant)
- Progressive Retinal Atrophy, prcd (PRCD Exon 1)
- Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLC4A3)
- Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2 (TTC8)
- Neuronal Ceroid Lipofuscinosis (CLN5 Golden Retriever Variant)
- GM2 Gangliosidosis (HEXB, Poodle Variant)
- Degenerative Myelopathy, DM (SOD1A)
- Neonatal Encephalopathy with Seizures, NEWS (ATF2)
- Muscular Dystrophy (DMD Golden Retriever Variant)
- Dystrophic Epidermolysis Bullosa (COL7A1)
- Osteogenesis Imperfecta, Brittle Bone Disease (COL1A1)
- Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1)
- Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (FGF4 retrogene - CFA12)

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ADDITIONAL CONDITIONS TESTED

Oakley did not have the variants that we tested for, in the following conditions that the potential effect on dogs with Oakley's breeds may not yet be known.

- MDR1 Drug Sensitivity (MDR1)
- P2Y12 Receptor Platelet Disorder (P2Y12)
- Factor IX Deficiency, Hemophilia B (F9 Exon 7, Terrier Variant)
- Factor IX Deficiency, Hemophilia B (F9 Exon 7, Rhodesian Ridgeback Variant)
- Factor VII Deficiency (F7 Exon 5)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 10, Boxer Variant)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 11, Shepherd Variant 1)
- Factor VIII Deficiency, Hemophilia A (F8 Exon 1, Shepherd Variant 2)
- Thrombopathia (RASGRP1 Exon 5, Basset Hound Variant)
- Thrombopathia (RASGRP1 Exon 8)
- Thrombopathia (RASGRP1 Exon 5, American Eskimo Dog Variant)
- Von Willebrand Disease Type III, Type III vWD (VWF Exon 4)
- Von Willebrand Disease Type III, Type III vWD (VWF Exon 7)
- Von Willebrand Disease Type II, Type II vWD (VWF)
- Canine Leukocyte Adhesion Deficiency Type III, CLADIII (FERMT3)
- Canine Elliptocytosis (SPTB Exon 30)
- Glanzmann's Thrombasthenia Type I (ITGA2B Exon 12)
- May-Hegglin Anomaly (MYH9)
- Prekallikrein Deficiency (KLKB1 Exon 8)
- Pyruvate Kinase Deficiency (PKLR Exon 5)
- Pyruvate Kinase Deficiency (PKLR Exon 7 Labrador Variant)
- Pyruvate Kinase Deficiency (PKLR Exon 7 Pug Variant)
- Pyruvate Kinase Deficiency (PKLR Exon 7 Beagle Variant)

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ADDITIONAL CONDITIONS TESTED

- Ligneous Membranitis, LM (PLG)
- Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant)
- Complement 3 Deficiency, C3 Deficiency (C3)
- Severe Combined Immunodeficiency (PRKDC)
- Severe Combined Immunodeficiency (RAG1)
- X-linked Severe Combined Immunodeficiency (IL2RG Variant 1)
- X-linked Severe Combined Immunodeficiency (IL2RG Variant 2)
- Progressive Retinal Atrophy, rcd1 (PDE6B Exon 21 Irish Setter Variant)
- Progressive Retinal Atrophy, rcd3 (PDE6A)
- Progressive Retinal Atrophy, CNGA (CNGA1 Exon 9)
- Progressive Retinal Atrophy (CNGB1)
- Progressive Retinal Atrophy (SAG)
- Progressive Retinal Atrophy, crd1 (PDE6B)
- Progressive Retinal Atrophy, crd2 (IQCB1)
- Progressive Retinal Atrophy - crd4/cord1 (RPGRIP1)
- Collie Eye Anomaly, Choroidal Hypoplasia, CEA (NHEJ1)
- Achromatopsia (CNGA3 Exon 7 German Shepherd Variant)
- Achromatopsia (CNGA3 Exon 7 Labrador Retriever Variant)
- Autosomal Dominant Progressive Retinal Atrophy (RHO)
- Canine Multifocal Retinopathy (BEST1 Exon 2)
- Canine Multifocal Retinopathy (BEST1 Exon 5)
- Canine Multifocal Retinopathy (BEST1 Exon 10 Deletion)
- Canine Multifocal Retinopathy (BEST1 Exon 10 SNP)
- Glaucoma (ADAMTS10 Exon 9)
- Glaucoma (ADAMTS17 Exon 11)

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ADDITIONAL CONDITIONS TESTED

- Glaucoma (ADAMTS17 Exon 2)
- Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9 Shepherd Variant)
- Primary Lens Luxation (ADAMTS17)
- Congenital Stationary Night Blindness (RPE65)
- Macular Corneal Dystrophy, MCD (CHST6)
- 2,8-Dihydroxyadenine Urolithiasis, 2,8-DHA Urolithiasis (APRT)
- Cystinuria Type I-A (SLC3A1)
- Cystinuria Type II-A (SLC3A1)
- Cystinuria Type II-B (SLC7A9)
- Hyperuricosuria and Hyperuricemia or Urolithiasis, HUU (SLC2A9)
- Polycystic Kidney Disease, PKD (PKD1)
- Primary Hyperoxaluria (AGXT)
- Protein Losing Nephropathy, PLN (NPHS1)
- X-Linked Hereditary Nephropathy, XLHN (COL4A5 Exon 35, Samoyed Variant 2)
- Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN (COL4A4 Exon 3)
- Primary Ciliary Dyskinesia, PCD (CCDC39 Exon 3)
- Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatosis, Dry Eye Curly Coat Syndrome, CKCSID (FAM83H Exon 5)
- X-linked Ectodermal Dysplasia, Anhidrotic Ectodermal Dysplasia (EDA Intron 8)
- Renal Cystadenocarcinoma and Nodular Dermatofibrosis, RCND (FLCN Exon 7)
- Canine Fucosidosis (FUCA1)
- Glycogen Storage Disease Type II, Pompe's Disease, GSD II (GAA)
- Glycogen Storage Disease Type IA, Von Gierke Disease, GSD IA (G6PC)
- Glycogen Storage Disease Type IIIA, GSD IIIA (AGL)
- Mucopolysaccharidosis Type I, MPS I (IDUA)
- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 1)

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ADDITIONAL CONDITIONS TESTED

- Mucopolysaccharidosis Type IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 2)
- Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 5)
- Mucopolysaccharidosis Type VII, Sly Syndrome, MPS VII (GUSB Exon 3)
- Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Whippet and English Springer Spaniel Variant)
- Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Wachtelhund Variant)
- Lagotto Storage Disease (ATG4D)
- Neuronal Ceroid Lipofuscinosis 1, NCL 1 (PPT1 Exon 8)
- Neuronal Ceroid Lipofuscinosis 2, NCL 2 (TPP1 Exon 4)
- Neuronal Ceroid Lipofuscinosis 1, Cerebellar Ataxia, NCL4A (ARSG Exon 2)
- Neuronal Ceroid Lipofuscinosis 1, NCL 5 (CLN5 Border Collie Variant)
- Neuronal Ceroid Lipofuscinosis 6, NCL 6 (CLN6 Exon 7)
- Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 English Setter Variant)
- Neuronal Ceroid Lipofuscinosis (MFSD8)
- Neuronal Ceroid Lipofuscinosis (CLN8 Australian Shepherd Variant)
- Neuronal Ceroid Lipofuscinosis 10, NCL 10 (CTSD Exon 5)
- Adult-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2, Tibetan Terrier Variant)
- GM1 Gangliosidosis (GLB1 Exon 15 Shiba Inu Variant)
- GM1 Gangliosidosis (GLB1 Exon 15 Alaskan Husky Variant)
- GM1 Gangliosidosis (GLB1 Exon 2)
- GM2 Gangliosidosis (HEXA)
- Globoid Cell Leukodystrophy, Krabbe disease (GALC Exon 5)
- Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (Italian Greyhound Variant)
- Persistent Mullerian Duct Syndrome, PMDS (AMHR2)
- Deafness and Vestibular Syndrome of Dobermans, DVDob, DINGS (MYO7A)
- Shar-Pei Autoinflammatory Disease, SPAID, Shar-Pei Fever (MTBP)

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ADDITIONAL CONDITIONS TESTED

- Alaskan Husky Encephalopathy, Subacute Necrotizing Encephalomyelopathy (SLC19A3)
- Alexander Disease (GFAP)
- Cerebellar Abiotrophy, Neonatal Cerebellar Cortical Degeneration, NCCD (SPTBN2)
- Cerebellar Ataxia, Progressive Early-Onset Cerebellar Ataxia (SEL1L)
- Cerebellar Hypoplasia (VLDLR)
- Spinocerebellar Ataxia, Late-Onset Ataxia, LoSCA (CAPN1)
- Spinocerebellar Ataxia with Myokymia and/or Seizures (KCNJ10)
- Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy (LGI2)
- Fetal-Onset Neonatal Neuroaxonal Dystrophy (MFN2)
- Hypomyelination and Tremors (FNIP2)
- Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome (PLP)
- Neuroaxonal Dystrophy, NAD (Spanish Water Dog Variant)
- L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH)
- Polyneuropathy, NDRG1 Greyhound Variant (NDRG1 Exon 15)
- Polyneuropathy, NDRG1 Malamute Variant (NDRG1 Exon 4)
- Narcolepsy (HCRTR2 Intron 6)
- Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 15)
- Progressive Neuronal Abiotrophy, Canine Multiple System Degeneration, CMSD (SERAC1 Exon 4)
- Juvenile Laryngeal Paralysis and Polyneuropathy, Polyneuropathy with Ocular Abnormalities and Neuronal Vacuolation, POANV (RAB3GAP1, Rottweiler Variant)
- Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS (GDNF-AS)
- Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1, LPN1 (LPN1, ARHGEF10)
- Spongy Degeneration with Cerebellar Ataxia 1, SDCA1, SeSAME/EAST Syndrome (KCNJ10)
- Spongy Degeneration with Cerebellar Ataxia 2, SDCA2 (ATP1B2)
- Dilated Cardiomyopathy, DCM1 (PDK4)
- Dilated Cardiomyopathy, DCM2 (TTN)

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ADDITIONAL CONDITIONS TESTED

- Long QT Syndrome (KCNQ1)
- Muscular Dystrophy (DMD, Cavalier King Charles Spaniel Variant 1)
- Muscular Dystrophy (DMD Pembroke Welsh Corgi Variant)
- Centronuclear Myopathy (PTPLA)
- Exercise-Induced Collapse (DNM1)
- Inherited Myopathy of Great Danes (BIN1)
- Myostatin Deficiency, Bully Whippet Syndrome (MSTN)
- Myotonia Congenita (CLCN1 Exon 7)
- Myotonia Congenita (CLCN1 Exon 23)
- Myotubular Myopathy 1, X-linked Myotubular Myopathy, XL-MTM (MTM1, Labrador Variant)
- Hypocatalasia, Acatalasemia (CAT)
- Pyruvate Dehydrogenase Deficiency (PDP1)
- Malignant Hyperthermia (RYR1)
- Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 53)
- Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 8)
- Congenital Myasthenic Syndrome (CHAT)
- Congenital Myasthenic Syndrome (COLQ)
- Episodic Falling Syndrome (BCAN)
- Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1)
- Ichthyosis, Epidermolytic Hyperkeratosis (KRT10)
- Ichthyosis (SLC27A4)
- Ichthyosis (NIPAL4)
- Focal Non-Epidermolytic Palmoplantar Keratoderma, Pachyonychia Congenita (KRT16)
- Hereditary Footpad Hyperkeratosis (FAM83G)
- Hereditary Nasal Parakeratosis (SUV39H2)

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ADDITIONAL CONDITIONS TESTED

- **Musladin-Lueke Syndrome (ADAMTSL2)**
- **Cleft Lip and/or Cleft Palate (ADAMTS20)**
- **Hereditary Vitamin D-Resistant Rickets (VDR)**
- **Oculoskeletal Dysplasia 1, Dwarfism-Retinal Dysplasia, OSD1 (COL9A3, Labrador Retriever)**
- **Osteogenesis Imperfecta, Brittle Bone Disease (COL1A2)**
- **Osteogenesis Imperfecta, Brittle Bone Disease (SERPINH1)**
- **Skeletal Dysplasia 2, SD2 (COL11A2)**
- **Craniomandibular Osteopathy, CMO (SLC37A2)**
- **Chondrodystrophy, Norwegian Elkhound and Karelian Bear Dog Variant (ITGA10)**

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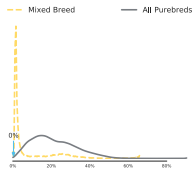
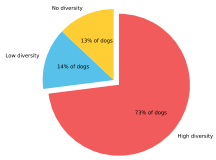
DNA Test Report | Test Date: April 12th, 2019 | embk.me/oakley92

INBREEDING AND DIVERSITY

CATEGORY	RESULT
Coefficient Of Inbreeding Our genetic COI measures the proportion of your dog's genome where the genes on the mother's side are identical by descent to those on the father's side.	**0%** 
MHC Class II - DLA DRB1 A Dog Leukocyte Antigen (DLA) gene, DRB1 encodes a major histocompatibility complex (MHC) protein involved in the immune response. Some studies have shown associations between certain DRB1 haplotypes and autoimmune diseases such as Addison's disease (hypoadrenocorticism) in certain dog breeds, but these findings have yet to be scientifically validated.	**High Diversity** How common is this amount of diversity in mixed breed dogs: 
MHC Class II - DLA DQA1 and DQB1 DQA1 and DQB1 are two tightly linked DLA genes that code for MHC proteins involved in the immune response. A number of studies have shown correlations of DQA-DQB1 haplotypes and certain autoimmune diseases; however, these have not yet been scientifically validated.	**High Diversity** How common is this amount of diversity in mixed breed dogs: