BOGEY

HEALTH REPORT

Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (FGF4 retrogene - CFA12)

Bogey inherited one copy of the variant we tested
Bogey is at increased risk for Type I IVDD

How to interpret this result
Bogey has one copy of an FGF4 retrogene on chromosome 12. In some breeds such as Beagles, Cocker Spaniels, and Dachshunds (among others) this variant is found in nearly all dogs. While those breeds are known to have an elevated risk of IVDD, many dogs in those breeds never develop IVDD. For mixed breed dogs and purebreds of other breeds where this variant is not as common, risk for Type I IVDD is greater for individuals with this variant than for similar dogs.

What is Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD?
Type I Intervertebral Disc Disease (IVDD) is a back/spine issue that refers to a health condition affecting the discs that act as cushions between vertebrae. With Type I IVDD, affected dogs can have a disc event where it ruptures or herniates towards the spinal cord. This pressure on the spinal cord causes neurologic signs which can range from a wobbly gait to impairment of movement. Chondrodystrophy (CDDY) refers to the relative proportion between a dog's legs and body, wherein the legs are shorter and the body longer. There are multiple different variants that can cause a markedly chondrodystrophic appearance as observed in Dachshunds and Corgis. However, this particular variant is the only one known to also increase the risk for IVDD.

When signs & symptoms develop in affected dogs
Signs of CDDY are recognized in puppies as it affects body shape. IVDD is usually first recognized in adult dogs, with breed specific differences in age of onset.

Signs & symptoms
Research indicates that dogs with one or two copies of this variant have a similar risk of developing IVDD. However, there are some breeds (e.g. Beagles and Cocker Spaniels, among others) where this variant has been passed down to nearly all dogs of the breed and most do not show overt clinical signs of the disorder. This suggests that there are other genetic and environmental factors (such as weight, mobility, and family history) that contribute to an individual dog's risk of developing clinical IVDD. Signs of IVDD include neck or back pain, a change in your dog's walking pattern (including dragging of the hind limbs), and paralysis. These signs can be mild to severe, and if your dog starts exhibiting these signs, you should schedule an appointment with your veterinarian for a diagnosis.

How vets diagnose this condition
For CDDY, dogs with one copy of this variant may have mild proportional differences in their leg length. Dogs with two copies of this variant will often have visually longer bodies and shorter legs. For IVDD, a neurological exam will be performed on any dog showing suspicious signs. Based on the result of this exam, radiographs to detect the presence of calcified discs or advanced imaging (MRI/CT) to detect a disc rupture may be recommended.

How this condition is treated
IVDD is treated differently based on the severity of the disease. Mild cases often respond to medical management which includes cage rest and pain management, while severe cases are often treated with surgical intervention. Both conservative and surgical treatment should be followed up with rehabilitation and physical therapy.

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HEALTH REPORT

Progressive Retinal Atrophy, prcd (PRCD Exon 1)

Bogey inherited one copy of the variant we tested

What does this result mean?
This result should not impact Bogey'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 Progressive Retinal Atrophy, prcd?
PRA-prcd is a retinal disease that causes progressive, non-painful vision loss. The retina contains cells, called photoreceptors, that collect information about light and send signals to the brain. There are two types of photoreceptors: rods, for night vision and movement, and cones, for day vision and color. This type of PRA leads to early loss of rod cells, leading to night blindness before day blindness.

When signs & symptoms develop in affected dogs
The age affected dogs will first show signs of visual impairment varies by breed. However, most begin showing clinical signs in early adulthood.

How vets diagnose this condition
Veterinarians use a focused light to examine the pupils. In affected dogs, the pupils will appear more dilated and slower to contract. Your vet may also use a lens to visualize the retina at the back of the eye to look for changes in the optic nerve or blood vessels. You may be referred to a veterinary ophthalmologist for a definitive diagnosis.

How this condition is treated
Currently, there is no definitive treatment for PRA. Supplements, including antioxidants, have been proposed for management of the disease, but have not been scientifically proven effective.

Actions to take if your dog is affected

- Careful monitoring by your veterinarian will be required for the rest of your affected dog's life as secondary complications, including cataracts, can develop.
- With blind dogs, keeping furniture in the same location, making sure they are on a leash in unfamiliar territory, and training them to understand verbal commands are some of the ways to help them at home.

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DNA Test Report | Test Date: February 25th, 2021 | embk.me/bogey112

BREED-RELEVANT CONDITIONS TESTED

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

- Von Willebrand Disease Type I (VWF)
- Autosomal Recessive Hereditary Nephropathy, Familial Nephropathy, ARHN (COL4A4 Exon 3)
- Glycogen storage disease Type VII, Phosphofructokinase Deficiency, PFK Deficiency (PFKM Whippet and English Springer Spaniel Variant)
- GM2 Gangliosidosis (HEXB, Poodle Variant)
- Neonatal Encephalopathy with Seizures, NEWS (ATF2)
- Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS (GDNF-AS)
- Exercise-Induced Collapse (DNM1)
- Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1)

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ADDITIONAL CONDITIONS TESTED

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

- MDR1 Drug Sensitivity (ABCB1)
- 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 I, CLADI (ITGB2)
- Canine Leukocyte Adhesion Deficiency Type III, CLADIII (FERMT3)
- Congenital Macrothrombocytopenia (TUBB1 Exon 1, Cairn and Norfolk Terrier Variant)
- 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)

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ADDITIONAL CONDITIONS TESTED

- Pyruvate Kinase Deficiency (PKLR Exon 10)
- Trapped Neutrophil Syndrome (VPS13B)
- Ligneous Membranitis, LM (PLG)
- Platelet factor X receptor deficiency, Scott Syndrome (TMEM16F)
- Methemoglobinemia CYB5R3
- Congenital Hypothyroidism (TPO, Tenterfield Terrier Variant)
- Congenital Hypothyroidism (TPO, Rat, Toy, Hairless 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)
- Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLC4A3)
- Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2 (TTC8)
- Progressive Retinal Atrophy, crd1 (PDE6B)
- Progressive Retinal Atrophy - crd4/cord1 (RPGRIP1)
- X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGR)
- Progressive Retinal Atrophy, PRA3 (FAM161A)
- Collie Eye Anomaly, Choroidal Hypoplasia, CEA (NHEJ1)
- Day blindness, Cone Degeneration, Achromatopsia (CNGB3 Exon 6)

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ADDITIONAL CONDITIONS TESTED

- 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)
- Glaucoma (ADAMTS10 Exon 9)
- Glaucoma (ADAMTS10 Exon 17)
- Glaucoma (ADAMTS17 Exon 11)
- Glaucoma (ADAMTS17 Exon 2)
- Goniodysgenesis and Glaucoma (OLFM3)
- Hereditary Cataracts, Early-Onset Cataracts, Juvenile Cataracts (HSF4 Exon 9 Shepherd Variant)
- Primary Lens Luxation (ADAMTS17)
- Congenital Stationary Night Blindness (RPE65)
- Congenital Stationary Night Blindness (LRIT3)
- 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)

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ADDITIONAL CONDITIONS TESTED

- Primary Ciliary Dyskinesia, PCD (CCDC39 Exon 3)
- Primary Ciliary Dyskinesia, PCD (NME5)
- 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 IIIA, Sanfilippo Syndrome Type A, MPS IIIA (SGSH Exon 6 Variant 1)
- 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 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)
- Neuronal Ceroid Lipofuscinosis (CLN5 Golden Retriever Variant)

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ADDITIONAL CONDITIONS TESTED

- Adult-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2, Tibetan Terrier Variant)
- Late-Onset Neuronal Ceroid Lipofuscinosis (ATP13A2, Australian Cattle Dog 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)
- Autosomal Recessive Amelogenesis Imperfecta, Familial Enamel Hypoplasia (Parson Russell Terrier 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)
- Neonatal Interstitial Lung Disease (LAMP3)
- 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)
- Hereditary Ataxia (RAB24)
- Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy (LGI2)
- Degenerative Myelopathy, DM (SOD1A)
- Fetal-Onset Neonatal Neuroaxonal Dystrophy (MFN2)
- Hypomyelination and Tremors (FNIP2)

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ADDITIONAL CONDITIONS TESTED

- Shaking Puppy Syndrome, X-linked Generalized Tremor Syndrome (PLP)
- Neuroaxonal Dystrophy, NAD (Spanish Water Dog Variant)
- Neuroaxonal Dystrophy, NAD (Rottweiler Variant)
- L-2-Hydroxyglutaricaciduria, L2HGA (L2HGDH)
- Polyneuropathy, NDRG1 Malamute Variant (NDRG1 Exon 4)
- Narcolepsy (HCRTR2 Intron 6)
- Narcolepsy (HCRTR2 Exon 1)
- 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)
- Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1, LPN1 (LPN1, ARHGEF10)
- Juvenile Myoclonic Epilepsy (DIRAS1)
- Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 2, LPN2 (GJA9)
- 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)
- Long QT Syndrome (KCNQ1)
- Cardiomyopathy and Juvenile Mortality (YARS2)
- Muscular Dystrophy (DMD, Cavalier King Charles Spaniel Variant 1)
- Muscular Dystrophy (DMD Golden Retriever Variant)
- Limb Girdle Muscular Dystrophy (SGCD, Boston Terrier Variant)
- Ulrich-like Congenital Muscular Dystrophy (COL6A3, Labrador Variant)
- Centronuclear Myopathy (PTPLA)
- Inherited Myopathy of Great Danes (BIN1)

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ADDITIONAL CONDITIONS TESTED

- 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)
- Inflammatory Myopathy (SLC25A12)
- 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)
- Inherited Selected Cobalamin Malabsorption with Proteinuria (CUBN)
- Lundehund Syndrome (LEPREL1)
- Congenital Myasthenic Syndrome (CHAT)
- Congenital Myasthenic Syndrome (COLQ)
- Congenital Myasthenic Syndrome (CHRNE)
- Congenital Myasthenic Syndrome (COLQ)
- Myasthenia Gravis Like Syndrome (CHRNE)
- Episodic Falling Syndrome (BCAN)
- Paroxysmal Dyskinesia, PxD (PGIN)
- Demyelinating Polyneuropathy (SBF2/MTRM13)
- Dystrophic Epidermolysis Bullosa (COL7A1)
- Dystrophic Epidermolysis Bullosa (COL7A1)
- Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1)
- Ichthyosis, Epidermolytic Hyperkeratosis (KRT10)
- Ichthyosis (PNPLA1)

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ADDITIONAL CONDITIONS TESTED

- Ichthyosis (SLC27A4)
- Ichthyosis (NIPAL4)
- Hereditary Footpad Hyperkeratosis (FAM83G)
- Hereditary Footpad Hyperkeratosis (DSG1)
- Hereditary Nasal Parakeratosis (SUV39H2)
- Musladin-Lueke Syndrome (ADAMTSL2)
- Oculocutaneous Albinism, OCA (Pekingese Type)
- Bald Thigh Syndrome (IGFBP5)
- Lethal Acrodermatitis (MKLN1)
- Ehlers Danlos (Doberman) (ADAMTS2)
- Cleft Lip and/or Cleft Palate (ADAMTS20)
- Hereditary Vitamin D-Resistant Rickets (VDR)
- Osteogenesis Imperfecta, Brittle Bone Disease (COL1A2)
- Osteogenesis Imperfecta, Brittle Bone Disease (SERPINH1)
- Osteogenesis Imperfecta, Brittle Bone Disease (COL1A1)
- Skeletal Dysplasia 2, SD2 (COL11A2)
- Craniomandibular Osteopathy, CMO (SLC37A2)
- Raine Syndrome, Canine Dental Hypomineralization Syndrome (FAM20C)
- Chondrodystrophy, Norwegian Elkhound and Karelian Bear Dog Variant (ITGA10)