

BELLA

Veterinary Report by Embark

embarkvet.com
Test Date: March 3rd, 2020

Customer-supplied information

Owner Name: Guthrey Preston	Breed type: purebred
Dog Name: Bella	Breed: Bernese mountain dog
Sex: Female (intact)	Breed registration: aka ws67024202
Date of birth: 12/20/18	Microchip#: n/a

Genetic summary

Genetic breed identification: Bernese Mountain Dog	Predicted adult weight: 86 lbs Calculated from 17 size genes.
Breed mix: Bernese Mountain Dog: 100.0%	Genetic age: 28 human years Human equivalent age based on size, date of birth provided, and other factors

Clinical Tools

These clinical genetic tools can inform clinical decisions and diagnoses. These tools do not predict increased risk for disease.

Alanine Aminotransferase Activity (GPT)

Bella's baseline ALT level is Low Normal

Why is this important to your vet?

Bella has one copy of a variant associated with reduced ALT activity as measured on veterinary blood chemistry panels. Please inform your veterinarian that Bella has this genotype, as ALT is often used as an indicator of liver health and Bella is likely to have a lower than average resting ALT activity. As such, an increase in Bella's ALT activity could be evidence of liver damage, even if it is within normal limits by standard ALT reference ranges.

What is Alanine Aminotransferase Activity?

Alanine aminotransferase (ALT) is a clinical tool that can be used by veterinarians to better monitor liver health. This result is not associated with liver disease. ALT is one of several values that veterinarians measure on routine blood work to evaluate the liver. It is a naturally occurring enzyme located in liver cells that helps break down protein. When the liver is damaged or inflamed, ALT is released into the bloodstream.

How vets diagnose this condition

Genetic testing is the only way to provide your veterinarian with this clinical tool.

How this condition is treated

Veterinarians may recommend blood work to establish a baseline ALT value for healthy dogs with one or two copies of this variant.

Health Report

How to Interpret Bella's genetic health results:
If Bella inherited any of the variants that we tested, they will be listed at the top of the Health Report section, along with a description of how to interpret this result. We also include all of the variants that we tested Bella for that we did not detect the risk variant for.

A genetic test is not a diagnosis

This genetic test does not diagnose a disease. Please talk to your vet about your dog's genetic results, or if you think that your pet may have a health condition or disease.

Bella inherited one variant that you should learn more about.

Degenerative Myelopathy, DM	<input checked="" type="checkbox"/>
Breed-Relevant Genetic Conditions	1 variant not detected <input checked="" type="checkbox"/>
Additional Genetic Conditions	193 variants not detected <input checked="" type="checkbox"/>

Health Report

Degenerative Myelopathy, DM (SOD1A)
 Bella inherited one copy of the variant we tested

What does this result mean?

This result should not impact Bella'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 her offspring.

What is Degenerative Myelopathy, DM?

The dog equivalent of Amyotrophic Lateral Sclerosis, or Lou Gehrig's disease, DM is a progressive degenerative disorder of the spinal cord. Because the nerves that control the hind limbs are the first to degenerate, the most common clinical signs are back muscle wasting and gait abnormalities.

When signs & symptoms develop in affected dogs

Affected dogs do not usually show signs of DM until they are at least 8 years old.

How vets diagnose this condition

Definitive diagnosis requires microscopic analysis of the spinal cord after death. However, veterinarians use clues such as genetic testing, breed, age, and other diagnostics to determine if DM is the most likely cause of your dog's clinical signs.

How this condition is treated

As dogs age seniors at the time of onset, the treatment for DM is aimed towards increasing their comfort through a combination of lifestyle changes, medication, and physical therapy.

Actions to take if your dog is affected

- Giving your dog the best quality of life for as long as possible is all you can do after receiving this diagnosis.

Breed-Relevant Conditions Tested

Bella did not have the variants that we tested for, that are relevant to her breed:

Von Willebrand Disease Type I (VWF)

Additional Conditions Tested

Bella did not have the variants that we tested for, in the following conditions that the potential effect on dogs with Bella's breed 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 11, Shepherd Variant 2)
- Thrombopathia (RASGRP1 Exon 5, Bassett Hound Variant)
- Thrombopathia (RASGRP1 Exon 8)
- Thrombopathia (RASGRP1 Exon 5, American Eskimo Dog Variant)
- Von Willebrand Disease Type II, Type III vWD (VWF Exon 4)
- Von Willebrand Disease Type II, 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 II, CLADII (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)
- Pyruvate Kinase Deficiency (PKLR Exon 7 Pug Variant)

Additional Conditions Tested

- Pyruvate Kinase Deficiency (PKLR Exon 7 Beagle Variant)
- Pyruvate Kinase Deficiency (PKLR Exon 10)
- Trapped Neutrophil Syndrome (VPS13B)
- Ligneeux Membranitis, LM (PLG)
- Platelet factor X receptor deficiency, Scott Syndrome (TMEM16F)
- Methemoglobinemia CY5R3
- 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 (L2R9 Variant 1)
- X-linked Severe Combined Immunodeficiency (L2R9 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, prcd (PRCD Exon 1)
- Progressive Retinal Atrophy (CNGB1)
- Progressive Retinal Atrophy (SAG)
- Golden Retriever Progressive Retinal Atrophy 1, GR-PRA1 (SLCA43)
- Golden Retriever Progressive Retinal Atrophy 2, GR-PRA2 (TTC8)
- Progressive Retinal Atrophy, rcd1 (PDE6B)
- Progressive Retinal Atrophy - crd4/cord1 (RPGRI1)
- X-Linked Progressive Retinal Atrophy 1, XL-PRA1 (RPGRI)

Additional Conditions Tested

- Progressive Retinal Atrophy, PRA3 (FAM161A)
- Collie Eye Anomaly, Choroidal Hypoplasia, CEA (NHEJ1)
- Day blindness, Cone Degeneration, Achromatopsia (CNGB3 Exon 6)
- 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 (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)
- 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)

Additional Conditions Tested

- Polycystic Kidney Disease, PKD (PKD1)
- Primary Hyperoxaluria (AGXT)
- Protein Losing Nephropathy, PLN (NPHIS1)
- 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 Dermatitis, Dry Eye Cuity Coat Syndrome, CKCSID (FAM53H 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)
- 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 (TPPI Exon 4)
- Neuronal Ceroid Lipofuscinosis 1, Cerebellar Ataxia, NCL4A (ARSG Exon 2)

Additional Conditions Tested

- Neuronal Ceroid Lipofuscinosis 1, NCL 5 (CLN5 Border Collie Variant)
- Neuronal Ceroid Lipofuscinosis 6, NCL 6 (CLN6 English)
- Neuronal Ceroid Lipofuscinosis 8, NCL 8 (CLN8 English Setter Variant)
- Neuronal Ceroid Lipofuscinosis (MFSDB8)
- Neuronal Ceroid Lipofuscinosis (CLN8 Australian Shepherd Variant)
- Neuronal Ceroid Lipofuscinosis 10, NCL 10 (CTSD Exon 5)
- Neuronal Ceroid Lipofuscinosis (CLN5 Golden Retriever Variant)
- 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 (HEXB, Poodle Variant)
- 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, DV/DB, DINGS (MYO7A)
- Shar-Pei Autoinflammatory Disease, SPAID, Shar-Pei Fever (MTBP)
- 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)

Additional Conditions Tested

- Cerebellar Hypoplasia (VLDLR)
- Spinocerebellar Ataxia, Late-Onset Ataxia, LSCA (CAPN1)
- Spinocerebellar Ataxia with Myokymia and/or Seizures (KCNJ10)
- Hereditary Ataxia (RAB24)
- Benign Familial Juvenile Epilepsy, Remitting Focal Epilepsy (LG12)
- 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)
- Neuroaxonal Dystrophy, NAD (Rottweiler Variant)
- L-2-Hydroxyglutaricaciduria, LHGA (L2HGDH)
- Neonatal Encephalopathy with Seizures, NEWS (ATF2)
- Polynuropathy, NDRG1 Greyhound Variant (NDRG1 Exon 15)
- Polynuropathy, NDRG1 Malamute Variant (NDRG1 Exon 4)
- Narcolepsy (HCRTR2 Intron 6)
- Narcolepsy (NCRTR2 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)
- Hereditary Sensory Autonomic Neuropathy, Acral Mutilation Syndrome, AMS (DDNF-AS)
- Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 1, LPN1 (LPN1, ARHGFE10)
- Juvenile Myoclonic Epilepsy (DIRAS1)
- Juvenile-Onset Polyneuropathy, Leonberger Polyneuropathy 2, LPN2 (GJA9)
- Spongy Degeneration with Cerebellar Ataxia 1, SDCA1, SesAME/EAST Syndrome (KCNJ10)

Additional Conditions Tested

- Spongy Degeneration with Cerebellar Ataxia 2, SDCA2 (ATP1B2)
- Dilated Cardiomyopathy, DCM1 (PDK4)
- Dilated Cardiomyopathy, DCM2 (TTN)
- Long QT Syndrome (KCNQ1)
- Congenital Dystrophy (DMD, Cavalier King Charles Spaniel Variant 1)
- Muscular Dystrophy (DMD Pembroke Welsh Corgi Variant)
- Limb Dystrophy (DMD Golden Retriever Variant)
- Limb Girdle Muscular Dystrophy (SGCG, Boston Terrier 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)
- Hypocatalasemia, Acatlasemia (CAT)
- Pyruvate Dehydrogenase Deficiency (PDP1)
- Malignant Hyperthermia (RYR1)
- Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 5)
- Imerslund-Grasbeck Syndrome, Selective Cobalamin Malabsorption (CUBN Exon 8)
- Inherited Selected Cobalamin Malabsorption with Proteinuria (CUBN)
- Lundehund Dystrophy (LEPREL1)
- Congenital Myasthenic Syndrome (CHAT)
- Congenital Myasthenic Syndrome (COLG)

Additional Conditions Tested

- Congenital Myasthenic Syndrome (CHRN)
- Episodic Falling Syndrome (BCAN)
- Paroxysmal Dyskinesia, PxD (PGIN)
- Dystrophic Epidermolysis Bullosa (COL7A1)
- Dystrophic Epidermolysis Bullosa (COL7A1)
- Ectodermal Dysplasia, Skin Fragility Syndrome (PKP1)
- Ichthyosis, Epidermolytic Hyperkeratosis (KRT10)
- Ichthyosis (PNPLA1)
- Ichthyosis (SLC27A4)
- Ichthyosis (NIPAL4)
- Hereditary Footpad Hyperkeratosis (FAM830)
- Hereditary Nasal Parakeratosis (SLU39H2)
- Mustadlin-Lueke Syndrome (ADMTSL2)
- Oculocutaneous Albinism, OCA (Pekingesee Type)
- Bald Thigh Syndrome (IGFBP5)
- 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)
- Osteochondrodysplasia, Skeletal Dwarfism (SLC13A1)
- Skeletal Dysplasia 2, SD2 (COL11A2)
- Craniofacial Dysplasia, CMO (SLC37A2)
- Chondrodystrophy and Intervertebral Disc Disease, CDDY/IVDD, Type I IVDD (FGF4 retrogene - CFA12)

Additional Conditions Tested

- Chondrodystrophy, Norwegian Elkhound and Karelian Bear Dog Variant (TTGA10)

Genetic Diversity and Inbreeding

Coefficient of Inbreeding (COI)
Genetic Result: 31%

Your genetic COI measures the proportion of your dog's genome (her genes) where the genes on the mother's side are identical by descent to those on the father's side. The higher your dog's coefficient of inbreeding (the percentage), the more inbred your dog is.

Your Dog's COI

Bernese Mountain Dog All Breeds

This graph represents where your dog's inbreeding levels fall on a scale compared to both dogs with a similar breed makeup to her (the yellow dotted line) and all purebred dogs (the grey line).

Genetic Diversity and Inbreeding

More on the Science
Embark scientists, along with our research partners at Cornell University, have shown the impact of inbreeding on longevity and fertility and developed a state-of-the-art, peer-reviewed method for accurately measuring COI and predicting average COI in litters.

Citations

Sains & Boyko 2019 "Fine-Scale Resolution of Runs of Homozygosity Reveal Patterns of Inbreeding and Substantial Overlap with Recessive Disease Genotypes in Domestic Dogs"

Chu et al 2019 "Inbreeding depression causes reduced fecundity in Golden Retrievers"

Yordy et al 2019 "Body size, inbreeding, and lifespan in domestic dogs"

About Embark

Embark Veterinary is a canine genetics company offering research-grade genetic tests to pet owners and breeders. Every Embark test examines over 200,000 genetic markers, and provides results for over 200 genetic health conditions, breed identification, clinical tools, and more.

Embark is a research partner of the Cornell University College of Veterinary Medicine and collaborates with scientists and registries to accelerate genetic research in canine health. We make it easy for customers and vets to understand, share and make use of their dog's unique genetic profile to improve canine health and happiness.

Learn more at embarkvet.com

Veterinarians and hospitals can send inquiries to veterinarians@embarkvet.com.