



BR00 196

Josie's Malachai, Mastiff

Registered Name: Josie's Malachai

Owner: Joseph Moody

Call Name: Malachai

Country: United States

Registration ID: WS31358510

Testing date: 2016/2/26

Breed: Mastiff

Gender: Male

Dog's identity verified from microchip or tattoo by veterinarian or other authorized person during sample taking: **No**

Test results - Known disorders in the breed

Disorder	Type	Mode of Inheritance	Result
Canine Multifocal Retinopathy 1, (CMR1); Mastiff-related breeds mutation	Ocular Disorders	Autosomal Recessive	Clear
Degenerative Myelopathy, (DM)	Neurologic Disorders	Autosomal Recessive (Incomplete Penetrance)	Clear
Dominant Progressive Retinal Atrophy, (DPRA)	Ocular Disorders	Autosomal Dominant	Clear

Test results for pharmacogenetics

Disorder	Mode of Inheritance	Result
Multidrug resistance 1 (MDR1 gene mutation)	Autosomal Recessive	Clear

On behalf of Genoscooper Laboratories,

When obtaining a carrier or at risk test result, we recommend that you contact your veterinarian for more detailed information on the condition and possible treatment.

SIGNATURE

Jonas Donner, PhD, Head of Research and Development at Genoscooper Laboratories



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Test results - Traits - page 1

Trait	Genotype	Description
Color Locus E - Extensions	Em/Em	The dog is likely to have a dark mask.
Color Locus B - Brown	B/B B/bd bd/bd	The dog doesn't have any of the tested b alleles causing brown color.
Color Locus K - Dominant Black	ky/ky	The dog is likely to express the coat color defined by the color locus A.
Color Locus A - Agouti	ay/ay	The dog is genetically sable.
Color Locus S - Piebald or extreme white spotting	S/S	The dog is likely to have solid coat color with minimal white.
Color Locus H - Harlequin	h/h	The dog doesn't have harlequin pattern.

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Test results - Traits - page 2

Trait	Genotype	Description
Furnishings / Improper Coat in Portuguese Water Dogs (marker test)	GG/TT	The dog is not genetically likely to express furnishings.
Color Pattern - Saddle Tan	-/-	The dog may have saddle tan pattern if it has also tan point genotype at the A locus.
Body mass, insulin-like growth factor 1 (IGF1) gene variant	A/G	The dog is heterozygous for this variant. This means that it carries one copy of the genetic variant typically associated with small body mass and one copy typically associated with large body mass. This genotype is often observed e.g. in Shetland Sheepdog, Border Collie and Welsh Corgi.
Snout/skull length (shortened head versus elongated head), bone morphogenetic protein 3 (BMP3) gene variant	C/C	Your dog is homozygous for the genetic variant typically found in breeds with an elongated head (e.g. Saluki, Collie, Irish Wolfhound).
Ear erectness (pricked ears versus floppy ears), variant chr10:11072007	C/C	Your dog is homozygous for (carries two copies of) a genetic variant typically associated with floppy ears. This genotype is common in breeds like English Springer Spaniel, Leonberger, Saluki, and Dachshunds. Interestingly, the C-allele of this variant is the ancestral allele frequent in wolf.
Bobtail	C/C	The dog does not carry any copy of the bobtail mutation. It therefore likely has a long-tailed phenotype.
Curly coat	C/C	The dog is genetically non-curly.
Coat length /	G/G	The dog carries two copies of the genetic variant typically associated with a short-haired coat.
Tiny size, insulin-like growth factor 1 receptor (IGF1R) gene variant	G/G	Your dog is homozygous for a genetic variant typically found in larger-sized breeds (height at the withers > 25.4 cm (10 inches)).

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Test results - Additional disorders found in other breeds - page 1

Blood Disorders

Disorder	Mode of Inheritance	Result
Bleeding disorder due to P2RY12 defect	Autosomal Recessive	Clear
Canine Cyclic Neutropenia, Cyclic Hematopoiesis, Gray Collie Syndrome, (CN)	Autosomal Recessive	Clear
Canine Leucocyte Adhesion Deficiency (CLAD), type III	Autosomal Recessive	Clear
Factor IX Deficiency or Hemophilia B (3 mutations)	X-linked Recessive	Clear
Factor VII Deficiency	Autosomal Recessive	Clear
Factor VIII Deficiency or Hemophilia A (3 mutations)	X-linked Recessive	Clear
Glanzmann Thrombasthenia Type I, (GT) (2 mutations)	Autosomal Recessive	Clear
Hereditary Elliptocytosis		Clear
Hereditary Phosphofructokinase (PFK) Deficiency (2 mutations)	Autosomal Recessive	Clear
Macrothrombocytopenia (2 mutations)	Autosomal Recessive	No call
May-Hegglin Anomaly (MHA)	Autosomal Dominant	Clear
Prekallikrein Deficiency	Autosomal Recessive	Clear
Pyruvate Kinase Deficiency (9 mutations)	Autosomal Recessive	Clear
Thrombopathia (2 mutations)	Autosomal Recessive	Clear
Trapped Neutrophil Syndrome, (TNS)	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type II	Autosomal Recessive	Clear



Test results - Additional disorders found in other breeds - page 2

Ocular Disorders

Disorder	Mode of Inheritance	Result
Canine Multifocal Retinopathy 2, (CMR2); mutation originally found in Coton de Tulear	Autosomal Recessive	Clear
Canine Multifocal Retinopathy 3, (CMR3) (2 mutations)	Autosomal Recessive	No call
Cone Degeneration, (CD) or Achromatopsia (2 mutations)	Autosomal Recessive	Clear
Cone-Rod Dystrophy 1, (crd1); mutation originally found in American Staffordshire Terrier	Autosomal Recessive	Clear
Cone-Rod Dystrophy 2, (crd2); mutation originally found in Pit Bull Terrier	Autosomal Recessive	Clear
Cone-Rod Dystrophy, (cord1-PRA / crd4)	Autosomal Recessive (Incomplete Penetrance)	Clear
Cone-Rod Dystrophy, Standard Wirehaired Dachshund, (crd SWD)	Autosomal Recessive	Clear
Early Retinal Degeneration, (erd); mutation originally found in Norwegian Elkhound	Autosomal Recessive	Clear
Glaucoma in the Norwegian Elkhound	Autosomal Recessive	Clear
Golden Retriever Progressive Retinal Atrophy 1, (GR_PRA 1)	Autosomal Recessive	Clear
Primary Hereditary Cataract (PHC); mutation originally found in Australian Shepherd	Autosomal Dominant (Incomplete Penetrance)	Clear
Primary Lens Luxation, (PLL)	Autosomal Recessive	Clear
Primary Open Angle Glaucoma, (POAG); mutation originally found in Beagle	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (PAP1_PRA) (2 mutations)	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (PRA); mutation originally found in Basenji	Autosomal Recessive	Clear
Rod-Cone Dysplasia 1, (rcd1) and Rod-Cone Dysplasia 1a, (rdc1a) (2 mutations)	Autosomal Recessive	Clear
Rod-Cone Dysplasia 3, (rcd3)	Autosomal Recessive	Clear
X-Linked Progressive Retinal Atrophy 1, (XLPRA1)	X-linked Recessive	Clear
X-Linked Progressive Retinal Atrophy 2, (XLPRA2)	X-linked Recessive	Clear



Test results - Additional disorders found in other breeds - page 3

Endocrine Disorders

Disorder	Mode of Inheritance	Result
Congenital Hypothyroidism (3 mutations)	Autosomal Recessive	No call

Immunologic Disorders

Disorder	Mode of Inheritance	Result
Autosomal Recessive Severe Combined Immunodeficiency, (ARSCID)	Autosomal Recessive	Clear
Complement 3 (C3) Deficiency	Autosomal Recessive	Clear
Severe Combined Immunodeficiency in Frisian Water Dogs, (SCID)	Autosomal Recessive	Clear
X-linked Severe Combined Immunodeficiency (XSCID) (2 mutations)	X-linked Recessive	Clear



Test results - Additional disorders found in other breeds - page 4

Renal Disorders

Disorder	Mode of Inheritance	Result
Cystinuria Type I-A (4 mutations)	Autosomal Recessive	Clear
Cystinuria Type II-A and Cystinuria Type II-B (2 mutations)	Autosomal Dominant	Clear
Hyperuricosuria, (HUU)	Autosomal Recessive	Clear
Polycystic Kidney Disease in Bull Terriers, (BTPKD)	Autosomal Dominant	Clear
Primary Hyperoxaluria, (PH); mutation originally found in Coton de Tulear	Autosomal Recessive	Clear
Renal Cystadenocarcinoma and Nodular Dermatofibrosis, (RCND)	Autosomal Dominant	Clear
X-Linked Hereditary Nephropathy, (XLHN) (2 mutations)	X-linked Recessive	Clear



Test results - Additional disorders found in other breeds - page 5

Metabolic Disorders

Disorder	Mode of Inheritance	Result
Glycogen Storage Disease Type II or Pompe's Disease, (GSD II)	Autosomal Recessive	Clear
Glycogen Storage Disease Type IIIa, (GSD IIIa)	Autosomal Recessive	Clear
Glycogen Storage Disease Type Ia, (GSD Ia)	Autosomal Recessive	Clear
Hypocatalasia or Acatalasemia	Autosomal Recessive	Clear
Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS) (4 mutations)	Autosomal Recessive	Clear
Mucopolysaccharidosis Type 3A, (MPS IIIA) (2 mutations)	Autosomal Recessive	Clear
Mucopolysaccharidosis Type VII, (MPS VII) (2 mutations)	Autosomal Recessive	Clear
Pyruvate Dehydrogenase Phosphatase 1 (PDP1) Deficiency	Autosomal Recessive	Clear

Muscular Disorders

Disorder	Mode of Inheritance	Result
Cavalier King Charles Spaniel Muscular Dystrophy (CKCS-MD)	X-linked Recessive	Clear
Centronuclear Myopathy, (CNM) (2 mutations)	Autosomal Recessive	Clear
Duchenne or Dystrophin Muscular Dystrophy, (DMD)	X-linked Recessive	Clear
Muscular Hypertrophy (Double Muscling)	Autosomal Recessive	Clear
Myotonia Congenita; mutation originally found in Australian Cattle Dog	Autosomal Recessive	Clear
X-Linked Myotubular Myopathy	X-linked Recessive	Clear



Test results - Additional disorders found in other breeds - page 6

Neurologic Disorders

Disorder	Mode of Inheritance	Result
Adult-Onset Neuronal Ceroid Lipofuscinosis, (Adult-onset NCL), mutation originally found in Tibetan terrier	Autosomal Recessive	Clear
Alaskan Husky Encephalopathy, (AHE)	Autosomal Recessive	Clear
Bandera's Neonatal Ataxia, (BNAt)	Autosomal Recessive	Clear
Benign Familial Juvenile Epilepsy or Remitting Focal Epilepsy	Autosomal Recessive	Clear
Early-Onset Progressive Polyneuropathy (2 mutations)	Autosomal Recessive	Clear
Fetal Onset Neuroaxonal Dystrophy, (FNAD)	Autosomal Recessive	Clear
Hereditary Ataxia or Cerebellar Ataxia; mutation originally found in Old English Sheepdog and Gordon Setter	Autosomal Recessive	Clear
Hyperekplexia or Startle Disease	Autosomal Recessive	Clear
Hypomyelination in Weimaraners	Autosomal Recessive	Clear
L-2-Hydroxyglutaric aciduria, (L2HGA) (3 mutations)	Autosomal Recessive	Clear
Lagotto Storage Disease, (LSD)	Autosomal Recessive	Clear
Neonatal Cerebellar Cortical Degeneration or Cerebellar Abiotrophy, (NCCD)	Autosomal Recessive	Clear
Neonatal Encephalopathy with Seizures, (NEWS)	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 8, (NCL8); rare variant	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis Type 1, (NCL1)	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis Type 10, (NCL10)	Autosomal Recessive	Clear
Progressive early-onset cerebellar ataxia; mutation originally found in Finnish Hound	Autosomal Recessive	No call
Spinal Dysraphism	Autosomal Recessive	Clear
Spinocerebellar Ataxia with Myokymia and/or Seizures (SCA)	Autosomal Recessive	Clear
Spinocerebellar Ataxia/ Late-Onset Ataxia (SCA, LOA)	Autosomal Recessive	Clear
X-Linked Tremors; mutation originally found in English Springer Spaniel	X-linked Recessive	Clear



Test results - Additional disorders found in other breeds - page 7

Neuromuscular Disorders

Disorder	Mode of Inheritance	Result
Congenital Myasthenic Syndrome, (CMS)	Autosomal Recessive	Clear
Episodic Falling, (EF)	Autosomal Recessive	Clear
Exercise-Induced Collapse, (EIC)	Autosomal Recessive (Incomplete Penetrance)	Clear
GM2 Gangliosidosis or Sandhoff Disease (2 mutations)	Autosomal Recessive	Clear
Globoid Cell Leukodystrophy or Krabbe's Disease, (GLD) (2 mutations)	Autosomal Recessive	Clear

Skeletal Disorders

Disorder	Mode of Inheritance	Result
Chondrodysplasia; mutation originally found in Norwegian Elkhound and Karelian Bear Dog	Autosomal Recessive	Clear
Cranio-mandibular Osteopathy, (CMO)	Autosomal Dominant (Incomplete Penetrance)	Clear
Hereditary Vitamin D-Resistant Rickets, (HVDRR)	Autosomal Recessive	Clear
Oculoskeletal Dysplasia 2 or Dwarfism-Retinal Dysplasia 2	Autosomal Recessive	Clear
Osteochondrodysplasia in Miniature Poodles	Autosomal Recessive	Clear
Osteogenesis Imperfecta, (OI) found in the Beagle		Clear
Osteogenesis Imperfecta, (OI) or Brittle Bone Disease; mutation originally found in Dachshund	Autosomal Recessive	Clear
Skeletal Dysplasia 2, (SD2)	Autosomal Recessive	Clear



Test results - Additional disorders found in other breeds - page 8

Dermal Disorders

Disorder	Mode of Inheritance	Result
Dystrophic Epidermolysis Bullosa	Autosomal Recessive	Clear
Epidermolytic Hyperkeratosis	Autosomal Recessive	Clear
Hereditary Footpad Hyperkeratosis, (HFH)	Autosomal Recessive	Clear
Lamellar Ichthyosis, (LI)	Autosomal Recessive	Clear
Musladin-Lueke syndrome, (MLS)	Autosomal Recessive	Clear
X-Linked Ectodermal Dysplasia, (XHED)	X-linked Recessive	Clear

Other Disorders

Disorder	Mode of Inheritance	Result
Amelogenesis Imperfecta, (AI)	Autosomal Recessive	Clear
Cleft palate; mutation originally found in Nova Scotia Duck Tolling Retriever, reverse assay	Autosomal Recessive	Clear
Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, (CKCSID)	Autosomal Recessive	Clear
Narcolepsy (3 mutations)	Autosomal Recessive	Clear
Persistent Müllerian Duct Syndrome, (PMDS); mutation originally found in Miniature Schnauzer	Autosomal Recessive	Clear
Primary Ciliary Dyskinesia, (PCD)	Autosomal Recessive	Clear



APPENDIX

Explanation of the results of the tested disorders

Autosomal recessive inheritance (ARI)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - A dog carries one copy of the tested mutation. Carriers typically have a normal, healthy appearance but pass on the mutation to approximately 50% of their offspring.

At risk - A dog carries two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

Autosomal dominant inheritance (ADI)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

At risk - A dog carries one or two copies of the tested mutation and is at high or increased risk of developing the disease/condition.

X-linked recessive inheritance (X-linked)

Clear - A dog carries no copies of the tested mutation and has no or reduced likelihood of developing and passing on the disease/condition.

Carrier - Female carriers typically have a normal, healthy appearance but carry one copy of the tested mutation on one of their X chromosomes. As males only have one X chromosome, there are no male carriers.

At risk - Female dogs at risk carry two mutated copies of the tested mutation. Males carry one copy of the tested mutation on their single X chromosome. Dogs at risk are at high or increased risk of developing the disease/condition.

Please note that the descriptions above are generalized based on typically observed inheritance patterns. When obtaining a 'carrier' or 'at risk' test result, always refer to the corresponding online test documentation for more detailed information on the condition and any exceptions.

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