24/9/2015

8700 2321 7010 726

Gammonwood Harmony's Diadem, Mastiff

Registered name: Gammonwood Harmony's Diadem
Nickname: Harmony
Registration ID: WS44812801
Microchip: 982000167855832
Breed: Mastiff
Gender: Female

Owner: Joseph Moody
Country: United States
Testing date: 25/11/2014
DNA identification profile: Identified with standard AKC markers

Dog's identity verified from microchip or tattoo by veterinarian or other authorized person during sample taking: **Yes**

Test results - Known disorders in the breed

Disorder	Type	Mode of inheritance	Result
Canine Multifocal Retinopathy 1 (CMR1), Mastiff-related breeds mutation	Eye disorders	Autosomal Recessive	Clear
Autosomal Dominant Progressive Retinal Atrophy (ADPRA)	Eye disorders	Autosomal Dominant	Clear
Malignant Hyperthermia (MH)	Pharmacogenetics	Autosomal Dominant	Clear

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

On behalf of Genoscoper Laboratories,

Jonas Donner

SIGNATURE

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

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Test results - Traits - page 1/2

Trait	Genotype	Description
Colour Locus E (Extensions)	Em/Em	The dog is homozygous for EM allele.
Colour Locus B (Brown)	B/B || B/bd || bd/bd	The dog does not carry any of the tested b alleles.
Colour Locus K (Dominant Black)	ky/ky	The dog is homozygous for ky allele.
Colour Locus A (Agouti)	ay/ay	The dog is homozygous for ay-allele.
Colour Locus H (Harlequin)	h/h	The dog is likely to be non-harlequin.

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Test results - Traits - page 2/2

Trait	Genotype	Description
Furnishings / Improper Coat in Portuguese Water Dogs (marker test)	GG/TT	The dog is not genetically likely to express furnishings.
Body mass, insulin-like growth factor 1 (IGF1) gene variant	G/G	The dog is homozygous for the genetic variant typically associated with large body mass. This genotype is common e.g. in Great Dane, Newfoundland Dog and Greater Swiss Mountain Dog.
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.
Natural Bobtail (T-box mutation)	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 / "Fluffy" in Welsh Corgi	T/G	The dog carries one copy of the genetic variant typically associated with a short-haired coat, and one copy of the variant typically associated with a long-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 1/8

Blood disorders

Disorder	Mode of inheritance	Result
Bleeding disorder due to P2RY12 defect	Autosomal Recessive	Clear
Canine Cyclic Neutropenia (Gray Collie Syndrome)	Autosomal Recessive	Clear
Factor IX Deficiency or Haemophilia B (2 mutations)	X-linked Recessive	Clear
Factor VII Deficiency	Autosomal Recessive	Clear
Factor VIII deficiency or Haemophilia A; mutation originally found in German Shepherd Dog	X-linked Recessive	Clear
Glanzmann Thrombasthenia (GT), Type I; mutation originally found in Pyrenean Mountain Dog	Autosomal Recessive	Clear
Glycogen Storage Disease VII or Hereditary Phosphofructokinase (PFK) Deficiency	Autosomal Recessive	Clear
May-Hegglin Anomaly (MHA)	Autosomal Dominant	Clear
Pyruvate Kinase Deficiency of Erythrocyte (4 mutations)	Autosomal Recessive	Clear
Trapped Neutrophil Syndrome (TNS)	Autosomal Recessive	Clear

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Gammonwood Harmony's Diadem, Mastiff

Test results - Additional disorders found in other breeds - page 2/8

Eye disorders

Disorder	Mode of inheritance	Result
Achromatopsia or Cone Degeneration (CD); mutation originally found in German Shorthaired Pointer	Autosomal Recessive	Clear
Canine Multifocal Retinopathy 2 (CMR2); mutation originally found in Coton de Tulear	Autosomal Recessive	Clear
Canine Multifocal Retinopathy 3 (CMR3); mutation originally found in Lapponian Herder	Autosomal Recessive	Clear
Cone-rod Dystrophy (cord1-PRA / crd4)	Autosomal Recessive (Incomplete Penetrance)	Clear
Cone-rod dystrophy (crd SWD); mutation originally found in Standard Wire-haired Dachshund	Autosomal Recessive	Clear
Generalized Progressive Retinal Atrophy (gPRA)	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; mutation originally found in Beagle	Autosomal Recessive	Clear
Rod-Cone Dysplasia 1 (rcd1) and Rod-Cone Dysplasia 1a, (rcd1a) (2 mutations)	Autosomal Recessive	Clear
Rod-Cone Dysplasia 3 (rcd3)	Autosomal Recessive	Clear
X-Linked Progressive Retinal Atrophy 1 (XLPRA1)	X-linked Recessive	Clear

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Test results - Additional disorders found in other breeds - page 3/8

Endocrine disorders

Disorder	Mode of inheritance	Result
Congenital hypothyroidism; mutation originally found in Toy Fox- and Rat Terrier	Autosomal Recessive	Clear

Immunological disorders

Disorder	Mode of inheritance	Result
C3 deficiency	Autosomal Recessive	Clear
X-linked Severe Combined Immunodeficiency (XSCID) (2 mutations)	X-linked Recessive	Clear

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Test results - Additional disorders found in other breeds - page 4/8

Kidney disorders

Disorder	Mode of inheritance	Result
Hyperuricosuria and Hyperuricemia (HUU) or Urolithiasis	Autosomal Recessive	Clear
Polycystic Kidney Disease (PKD)	Autosomal Dominant	Clear
Primary hyperoxaluria (PH); mutation originally found in Coton de Tulear	Autosomal Recessive	Clear
X-linked Hereditary Nephropathy (XLHN)	X-linked Recessive	Clear

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Test results - Additional disorders found in other breeds - page 5/8

Metabolic disorders

Disorder	Mode of inheritance	Result
Glycogen Storage Disease Type II (GSD II), or Pompe's disease	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
Mucopolysaccharidosis Type IIIA (MPS IIIA); mutation originally found in Dachshund	Autosomal Recessive	Clear
Mucopolysaccharidosis Type VII (MPS VII); mutation originally found in Brazilian Terrier	Autosomal Recessive	Clear
Pyruvate Dehydrogenase Deficiency	Autosomal Recessive	Clear

Muscular disorders

Disorder	Mode of inheritance	Result
Cavalier King Charles Spaniel Muscular Dystrophy (CKCS-MD)	X-linked Recessive	Clear
Duchenne-like Muscular Dystrophy, Pembroke Welsh Corgi-type	X-linked Recessive	Clear
Muscular Dystrophy, Duchenne type or Golden Retriever Muscular Dystrophy (GRMD)	X-linked Recessive	Clear
Myotonia; mutation originally found in Miniature Schnauzer	Autosomal Recessive	Clear
Myotubular Myopathy 1 or X-linked Myotubular Myopathy	X-linked Recessive	Clear

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Test results - Additional disorders found in other breeds - page 6/8

Neurological disorders

Disorder	Mode of inheritance	Result
Benign Familial Juvenile Epilepsy or Remitting Focal Epilepsy	Autosomal Recessive	Clear
Cerebellar abiotrophy or neonatal cerebellar cortical degeneration (NCCD)	Autosomal Recessive	Clear
Fetal-onset Neuroaxonal Dystrophy (FNAD)	Autosomal Recessive	Clear
Hyperekplexia or Startle Disease	Autosomal Recessive	Clear
L-2-Hydroxyglutaric aciduria (L2HGA) (2 mutations)	Autosomal Recessive	Clear
Neonatal Encephalopathy with Seizures (NEWS)	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis, type 12, mutation originally found in Tibetan terrier	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 1 (NCL1)	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 10 (NCL10)	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 6 (NCL6)	Autosomal Recessive	Clear
Polyneuropathy (2 mutations)	Autosomal Recessive	Clear
Progressive early-onset cerebellar ataxia; mutation originally found in Finnish Hound	Autosomal Recessive	Clear

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Test results - Additional disorders found in other breeds - page 7/8

Neuromuscular disorders

Disorder	Mode of inheritance	Result
Episodic falling (EF)	Autosomal Recessive	Clear
GM1 Gangliosidosis (3 mutations)	Autosomal Recessive	Clear
GM2 Gangliosidosis; mutation originally found in Toy Poodle	Autosomal Recessive	Clear
Globoid Cell Leukodystrophy (GLD) or Krabbe's disease, Terrier mutation	Autosomal Recessive	Clear

Skeletal disorders

Disorder	Mode of inheritance	Result
Chondrodysplasia (dwarfism); mutation originally found in Norwegian Elkhound and Karelian Bear Dog	Autosomal Recessive	Clear
Craniomandibular Osteopathy (CMO)	Autosomal Dominant (Incomplete Penetrance)	Clear
Osteogenesis imperfecta (OI) or Brittle Bone Disease; mutation originally found in Dachshund	Autosomal Recessive	Clear
Skeletal Dysplasia 2 (SD2)	Autosomal Recessive	Clear

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

Skin disorders

Disorder	Mode of inheritance	Result
Epidermolytic Hyperkeratosis or Ichthyosis in Norfolk Terrier	Autosomal Recessive	Clear
Musladin-Lueke syndrome (MLS)	Autosomal Recessive	Clear

Other disorders

Disorder	Mode of inheritance	Result
Congenital Keratoconjuctivitis Sicca and Ichthyosiform Dermatosis (CKCSID) or Dry Eye Curly Coat Syndrome	Autosomal Recessive	Clear
Narcolepsy	Autosomal Recessive	Clear
Persistant Müllerian Duct Syndrome (PMDS), mutation originally found in Miniature Schnauzer	Autosomal Recessive	Clear
Primary Ciliary Dyskinesia (PCD)	Autosomal Recessive	Clear

On behalf of Genoscoper Laboratories,

Jonas Donner

SIGNATURE

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

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.
Affected - 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.
Affected - 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.
Affected - Affected female dogs carry two mutated copies of the tested mutation. Affected males carry one copy of the tested mutation on their single X chromosome. Affected dogs 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 affected test result, always refer to the corresponding online test documentation for more detailed information on the condition and any exceptions.

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