



8700 2321 4356 718

Gammonwood Siege Master, Mastiff

**Registered Name:** Gammonwood Siege Master

**Call Name:** Siege

**Registration ID:** 2100325167

**Microchip:** 982000123214971

**Breed:** Mastiff

**Gender:** Male

**Owner:** Jennifer Willshire

**Country:** Australia

**Testing date:** 2014/3/20

**DNA identification profile:** Identified with standard ISAG 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	Ocular Disorders	Autosomal Recessive	Clear
Dominant Progressive Retinal Atrophy, (DPRA)	Ocular Disorders	Autosomal Dominant	Clear

## Test results for pharmacogenetics

Disorder	Mode of Inheritance	Result
Malignant Hyperthermia (MH)	Autosomal Dominant	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 not likely to have black coat.
Color Locus A (Agouti)	No call	
Color Locus H (Harlequin)	h/h	The dog doesn't have harlequin pattern.

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**DNA identified with  
identification standard ISAG  
profile:** markers

**Breed:** Mastiff

**Gender:** Male

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

## 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.
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.
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 / "Fluffy" in Welsh Corgi	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
Factor IX Deficiency or Hemophilia B (2 mutations)	X-linked Recessive	Clear
Factor VII Deficiency	Autosomal Recessive	Clear
Factor VIII Deficiency or Hemophilia A; mutation originally found in German Shepherd Dog	X-linked Recessive	Clear
Glanzmann Thrombasthenia Type I, (GT); mutation originally found in Pyrenean Mountain Dog	Autosomal Recessive	Clear
Hereditary Phosphofructokinase (PFK) Deficiency	Autosomal Recessive	Clear
May-Hegglin Anomaly (MHA)	Autosomal Dominant	Clear
Pyruvate Kinase Deficiency (4 mutations)	Autosomal Recessive	Clear
Trapped Neutrophil Syndrome, (TNS)	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type II	Autosomal Recessive	No call



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## 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); mutation originally found in Lapponian Herder	Autosomal Recessive	Clear
Cone Degeneration, (CD) or Achromatopsia; mutation originally found in German Shorthaired Pointer	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
Generalized Progressive Retinal Atrophy	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
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, (XLPR1)	X-linked Recessive	Clear
X-Linked Progressive Retinal Atrophy 2, (XLPR2)	X-linked Recessive	No call



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

### Endocrine Disorders

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

### Immunologic Disorders

Disorder	Mode of Inheritance	Result
Complement 3 (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

### Renal Disorders

<b>Disorder</b>	<b>Mode of Inheritance</b>	<b>Result</b>
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
X-Linked Hereditary Nephropathy, (XLHN)	X-linked Recessive	Clear



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## 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 Acatlasemia	Autosomal Recessive	Clear
Mucopolysaccharidosis Type 3A, (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 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
Duchenne or Dystrophin Muscular Dystrophy, (DMD)	X-linked Recessive	Clear
Myotonia Congenita; mutation originally found in Miniature Schnauzer	Autosomal Recessive	Clear
X-Linked Myotubular Myopathy	X-linked Recessive	Clear





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## 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
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
Hyperekplexia or Startle Disease	Autosomal Recessive	Clear
L-2-Hydroxyglutaric aciduria, (L2HGA) (2 mutations)	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 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	Clear



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

### 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 or Krabbe's Disease, (GLD); Terrier mutation	Autosomal Recessive	Clear

### Skeletal Disorders

Disorder	Mode of Inheritance	Result
Chondrodysplasia; 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

### Dermal Disorders

Disorder	Mode of Inheritance	Result
Dystrophic Epidermolysis Bullosa	Autosomal Recessive	Clear
Epidermolytic Hyperkeratosis	Autosomal Recessive	Clear
Musladin-Lueke syndrome, (MLS)	Autosomal Recessive	Clear

### Other Disorders

Disorder	Mode of Inheritance	Result
Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, (CKCSID)	Autosomal Recessive	Clear
Narcolepsy; mutation originally found in Doberman Pinscher	Autosomal Recessive	Clear
Persistent Müllerian Duct Syndrome, (PMDS); mutation originally found in Miniature Schnauzer	Autosomal Recessive	Clear
Primary Ciliary Dyskinesia, (PCD)	Autosomal Recessive	Clear



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## 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.

## Genoscooper Laboratories - Legal Notice

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