8700 2396 8742

Athletic Idun, Boerboel

Registered Name:	Athletic Idun	**Owner:**	Linda Stenman
Nickname:	Idun	**Country:**	Sweden
Registration ID:	0000298422	**Testing date:**	2017/11/3
Microchip:	752098100796828	**DNA identification profile:**	Identified with standard ISAG 2006 markers
Breed:	Boerboel		
Gender:	Female		

Dog's identity verified from microchip or tattoo by veterinarian or other authorised 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
Hyperuricosuria, (HUU)	Renal Disorders	Autosomal Recessive	Clear

On behalf of Genoscoper Laboratories,

Jonas Donner

SIGNATURE

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

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Gender:	Female		

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

Test results - Traits - page 1

Coat Type

Trait	Genotype	Description
Coat Length	L/L	The dog is likely to have short-haired coat.
Furnishings / Improper Coat in Portuguese Water Dogs (marker test)	GG/TC	The dog is not genetically likely to express furnishings.
KRT71	C/C	The dog does not carry any copies of the tested allele causing curly coat. The dog most likely has non-curly hair.

On behalf of Genoscoper Laboratories,

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Gender:	Female		

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

Test results - Traits - page 2

Coat Colour

Trait	Genotype	Description
Colour Locus E - Extensions	Em/Em	The dog is likely to have a dark mask.
Colour Locus B - Brown	B/B || B/bd || bd/bd	The dog doesn't have any of the tested b alleles causing brown colour.
Colour Locus K - Dominant Black	KB/ky || kbr/ky || kbr/kbr	The dog is genetically dominant black or brindle.
Colour Locus A - Agouti	ay/ay	The dog is genetically sable.
Colour Locus S - Piebald or extreme white spotting	S/S	The dog is likely to have solid coat colour with minimal white.
Colour Locus H - Harlequin	h/h	The dog doesn't have harlequin pattern.
Colour Locus D - Dilution (marker test available for limited breeds)	D/D	The dog is likely to have non-dilute coat colour.
Merle (M allele)	m/m	This dog is genetically non-merle.
Saddle Tan (*RALY* gene dupl.)	-/-	The dog may have saddle tan pattern if it has also tan point genotype at the A locus.
Albinism (c^{aL}-allele)	C/C	This dog does not carry the tested mutation for albinism.

On behalf of Genoscoper Laboratories,

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Gender:	Female		

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

Test results - Traits - page 3

Body Size

Trait	Genotype	Description
IGF1 (chr15:41221438)	A/G	The dog is heterozygous for the ancestral allele. This means that it carries one copy of the genetic allele typically associated with small body mass and one copy typically associated with large body mass.
IGF1R c.611G>A (p.Arg204His)	G/G	The dog carries two ancestral alleles typically found in larger-sized breeds.
FGF4 insertion	D/D	The dog is homozygous for the ancient allele. The dog is likely to have legs of normal length.
STC2 (chr4:39182836)	T/T	The dog has two copies of the ancestral allele associated with larger body size.
GHR1 (p.E191K)	G/G	The dog has two copies of the ancestral allele associated with larger body size.
GHR2 (p.P177L)	C/C	The dog has two copies of the ancestral allele associated with larger body size.
HMGA2	G/G	The dog has two copies of the ancestral allele associated with larger body size.

On behalf of Genoscoper Laboratories,

Jonas Donner

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Jonas Donner, PhD, Head of Research and Development
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Gender:	Female		

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

Test results - Traits - page 4

Morphology

Trait	Genotype	Description
BMP3 c.1344C>A (p.Phe448Leu)	C/C	The dog does not carry the tested allele typically associated with shortened head (brachycephaly). The dog is more likely to have an elongated head (dolichocephaly).
chr10:11072007	C/C	The dog carries two copies of an allele typically associated with floppy ears. The dog is more likely to have floppy than pricked ears.
T c.189C>G (p.Ile63Met)	C/C	The dog does not carry the tested bobtail-causing genetic variant. The dog is most likely long-tailed.

On behalf of Genoscoper Laboratories,

Jonas Donner

SIGNATURE

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

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Athletic Idun, Boerboel

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, Grey Collie Syndrome, (CN)	Autosomal Recessive	Clear
Canine Leukocyte Adhesion Deficiency (CLAD), type III	Autosomal Recessive	Clear
Canine Scott Syndrome, (CSS)	Autosomal Recessive	Clear
Factor IX Deficiency or Hemophilia B (4 mutations)	X-linked Recessive	Clear
Factor VII Deficiency	Autosomal Recessive	Clear
Factor VIII Deficiency or Hemophilia A (3 mutations)	X-linked Recessive	Clear
Factor XI Deficiency	Autosomal Recessive	Clear
Glanzmann Thrombasthenia Type I, (GT); mutation originally found in Pyrenean Mountain Dog	Autosomal Recessive	Clear
Hereditary Elliptocytosis		Clear
Hereditary Phosphofructokinase (PFK) Deficiency	Autosomal Recessive	Clear
Macrothrombocytopenia; disease-linked variant originally found in Norfolk and Cairn Terrier	Autosomal Recessive	Clear
May-Hegglin Anomaly (MHA)	Autosomal Dominant	Clear
Prekallikrein Deficiency	Autosomal Recessive	Clear
Pyruvate Kinase Deficiency (4 mutations)	Autosomal Recessive	Clear
Thrombopathia (3 mutations)	Autosomal Recessive	Clear
Trapped Neutrophil Syndrome, (TNS)	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 1	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type 3 (3 mutations)	Autosomal Recessive	Clear

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

Ocular Disorders - page 1

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 (3 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 American 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
Congenital Stationary Night Blindness (CSNB)	Autosomal Recessive	Clear
Dominant Progressive Retinal Atrophy, (DPRA)	Autosomal Dominant	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
Primary Open Angle Glaucoma, (POAG); mutation originally found in Norwegian Elkhound	Autosomal Recessive	Clear
Progressive Retinal Atrophy Type III, (PRA type III); mutation originally found in Tibetan Spaniel and Tibetan Terrier	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (CNGA1-PRA); mutation originally found in Shetland Sheepdog	Autosomal Recessive	Clear
Progressive Retinal Atrophy, (PAP1_PRA); mutation originally found in Papillon and Phalene	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

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

Ocular Disorders - page 2

Disorder	Mode of Inheritance	Result
X-Linked Progressive Retinal Atrophy 1, (XLPRA1)	X-linked Recessive	Clear
X-Linked Progressive Retinal Atrophy 2, (XLPRA2)	X-linked Recessive	Clear

Cardiac Disorders

Disorder	Mode of Inheritance	Result
Long QT Syndrome	Autosomal Dominant	Clear

Endocrine Disorders

Disorder	Mode of Inheritance	Result
Congenital Hypothyroidism (2 mutations)	Autosomal Recessive	Clear

Immunological Disorders

Disorder	Mode of Inheritance	Result
Autosomal Recessive Severe Combined Immunodeficiency, (ARSCID)	Autosomal Recessive	Clear
Complement 3 (C3) Deficiency	Autosomal Recessive	Clear
Myeloperoxidase Deficiency		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; mutation originally found in Newfoundland Dog	Autosomal Recessive	Clear
Cystinuria Type II-A; mutation originally found in Australian Cattle Dog	Autosomal Dominant	Clear
Cystinuria, Type II-B; mutation originally found in Miniature Pinscher	Autosomal Dominant	Clear
Familial Nephropathy (FN) (2 mutations)	Autosomal Recessive	Clear
Fanconi Syndrome	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
Protein Losing Nephropathy, (PLN); NPHS1 gene variant		Clear
Renal Cystadenocarcinoma and Nodular Dermatofibrosis, (RCND)	Autosomal Dominant	Clear
X-Linked Hereditary Nephropathy, (XLHN) (2 mutations)	X-linked Recessive	Clear

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) (2 mutations)	Autosomal Recessive	Clear
Mucopolysaccharidosis Type IIIA, (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

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

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); mutation originally found in Golden Retriever	X-linked Recessive	Clear
Duchenne or Dystrophin Muscular Dystrophy, (DMD); mutation originally found in Norfolk Terrier	X-linked Recessive	Clear
Muscular Dystrophy, Ullrich-type; mutation originally found in Landseer	Autosomal Recessive	Clear
Myostatin deficiency (Double Muscling, "Bully")	Autosomal Recessive	Clear
Myotonia Congenita (2 mutations)	Autosomal Recessive	Clear
X-Linked Myotubular Myopathy (2 mutations)	X-linked Recessive	Clear

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

Neurological Disorders - page 1

Disorder	Mode of Inheritance	Result
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
Dandy-Walker-Like Malformation (DWLM); mutation originally found in Eurasier	Autosomal Recessive	Clear
Cerebral Dysfunction; mutation originally found in Friesian Stabyhoun	Autosomal Recessive	Clear
Early-Onset Progressive Polyneuropathy; mutation originally found in Alaskan Malamute	Autosomal Recessive	Clear
Early-Onset Progressive Polyneuropathy; mutation originally found in Greyhound	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; mutation originally found in Weimaraner	Autosomal Recessive	Clear
L-2-Hydroxyglutaric aciduria, (L2HGA); mutation originally found in Staffordshire Bull Terrier	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
Neuroaxonal Dystrophy (NAD); mutation originally found in Spanish Water Dog	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 1, (NCL1); mutation originally found in Dachshund	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 10, (NCL10); mutation originally found in American Bulldog	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 12, (NCL12); mutation originally found in Tibetan Terrier	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 4A, (NCL4); mutation originally found in American Staffordshire Terrier	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 5, (NCL5); mutation originally found in Border Collie	Autosomal Recessive	Clear

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

Neurological Disorders - page 2

Disorder	Mode of Inheritance	Result
Neuronal Ceroid Lipofuscinosis 8, (NCL8); mutation originally found in Alpienländische Dachsbracke (2 mutations)	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis, (NCL7); mutation originally found in Chinese Crested Dog and Chihuahua	Autosomal Recessive	Clear
Progressive Early-Onset Cerebellar Ataxia; mutation originally found in Finnish Hound	Autosomal Recessive	Clear
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

Neuromuscular Disorders

Disorder	Mode of Inheritance	Result
Congenital Myasthenic Syndrome (CMS); mutation originally found in Labrador Retriever	Autosomal Recessive	Clear
Congenital Myasthenic Syndrome, (CMS); mutation originally found in Jack Russell Terrier	Autosomal Recessive	Clear
Congenital Myasthenic Syndrome, (CMS); mutation originally found in Old Danish Pointing Dog	Autosomal Recessive	Clear
GM1 Gangliosidosis (3 mutations)	Autosomal Recessive	Clear
GM2 Gangliosidosis, mutation originally found in Japanese Chin	Autosomal Recessive	Clear
GM2 Gangliosidosis; mutation originally found in Toy Poodle	Autosomal Recessive	Clear
Globoid Cell Leukodystrophy or Krabbe Disease, (GLD); mutation originally found in Terriers	Autosomal Recessive	Clear
Globoid Cell Leukodystrophy or Krabbe Disease, (GLD); mutation originally found in Irish Setter	Autosomal Recessive	Clear

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

Skeletal Disorders

Disorder	Mode of Inheritance	Result
Chondrodysplasia; mutation originally found in Norwegian Elkhound and Karelian Bear Dog	Autosomal Recessive	Clear
Cleft Palate; Cleft Lip and Palate with Syndactyly; ADAMTS20 gene mutation originally found in Nova Scotia Duck Tolling Retriever	Autosomal Recessive	Clear
Cleft Palate; DLX6 gene mutation originally found in Nova Scotia Duck Tolling Retriever	Autosomal Recessive	Clear
Craniomandibular Osteopathy, (CMO); mutation associated with terrier breeds	Autosomal Dominant (Incomplete Penetrance)	Clear
Hereditary Vitamin D-Resistant Rickets, (HVDRR)	Autosomal Recessive	Clear
Oculoskeletal Dysplasia 2 or Dwarfism-Retinal Dysplasia 2, (OSD2)	Autosomal Recessive	Clear
Osteochondrodysplasia; mutation originally found in Miniature Poodle	Autosomal Recessive	Clear
Osteogenesis Imperfecta, (OI); mutation originally found in Beagle		Clear
Osteogenesis Imperfecta, (OI); mutation originally found in Dachshund	Autosomal Recessive	Clear
Skeletal Dysplasia 2, (SD2)	Autosomal Recessive	Clear
Spondylocostal Dysostosis	Autosomal Recessive	Clear
Van den Ende-Gupta Syndrome, (VDEGS)	Autosomal Recessive	Clear

Test results - Additional disorders found in other breeds - page 9

Dermal Disorders

Disorder	Mode of Inheritance	Result
Dystrophic Epidermolysis Bullosa; mutation originally found in Golden Retriever	Autosomal Recessive	Clear
Dystrophic Epidermolysis Bullosa; mutation originally found in Central Asian Ovcharka	Autosomal Recessive	Clear
Epidermolytic Hyperkeratosis	Autosomal Recessive	Clear
Focal Non-Epidermolytic Palmoplantar Keratoderma, (FNEPPK); mutation originally found in Dogue de Bordeaux		Clear
Hereditary Footpad Hyperkeratosis, (HFH)	Autosomal Recessive	Clear
Golden Retriever Ichthyosis	Autosomal Recessive	Clear
Ichthyosis; mutation originally found in Great Dane	Autosomal Recessive	Clear
Lamellar Ichthyosis, (LI)	Autosomal Recessive	Clear
Ligneous Membranitis	Autosomal Recessive	Clear
Musladin-Lueke syndrome, (MLS)	Autosomal Recessive	Clear
X-Linked Ectodermal Dysplasia, (XHED)	X-linked Recessive	Clear

Pharmacogenetics

Disorder	Mode of Inheritance	Result
Malignant Hyperthermia (MH)	Autosomal Dominant	Clear
Multi-Drug Resistance 1, (MDR1)	Autosomal Dominant	Clear

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Athletic Idun, Boerboel

Test results - Additional disorders found in other breeds - page 10

Other Disorders

Disorder	Mode of Inheritance	Result
Amelogenesis Imperfecta, (AI)	Autosomal Recessive	Clear
Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatosis, (CKCSID)	Autosomal Recessive	Clear
Dental Hypomineralisation; mutation originally found in Border Collie	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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