Optimal Selection™

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BR02 040

Klein Sandfontein Judge, Boerboel

Registered Name: Klein Sandfontein Judge
Call Name: Judge
Registration ID: 0000291503
Microchip: 900250000319225
Breed: Boerboel
Gender: Male

Owner: Shelby Flynn
Country: United States
Testing date: 2017/3/31

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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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.
Curly coat	C/C	The dog is genetically non-curly.

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Test results - Traits - page 2

Coat Color

Trait	Genotype	Description
Color Locus E - Extensions	Em/E	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.
Color Locus D - Dilution (marker test available for limited breeds)	D/d	The dog is likely to be non-dilute. The dog carries dilute coat color.
Color Locus C - Albinism (c^{aL}-allele)	C/C	This dog does not carry the tested mutation for albinism.
Color Pattern (RALY gene): Saddle Tan	-/-	The dog may have saddle tan pattern if it has also tan point genotype at the A locus.

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Test results - Traits - page 3

Body Size

Trait	Genotype	Description
Chondrodysplasia; breed-defining trait	D/D	The dog is homozygous for the ancient allele. The dog is likely to have legs of normal length.
Tiny size, insulin-like growth factor 1 receptor (IGF1R) gene variant	G/G	The dog carries two ancestral alleles typically found in larger-sized breeds.
Body mass, insulin-like growth factor 1 (IGF1) gene variant	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.
Body size, STC2 gene variant chr4:39182836	T/T	The dog has two copies of the ancestral allele associated with larger body size.
Body size, GHR1 gene variant E191K	G/G	The dog has two copies of the ancestral allele associated with larger body size.
Body size, GHR2 gene variant P177L	C/C	The dog has two copies of the ancestral allele associated with larger body size.
Body size, HMGA2 gene variant	G/G	The dog has two copies of the ancestral allele associated with larger body size.

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Test results - Traits - page 4

Morphology

Trait	Genotype	Description
Ear erectness (pricked ears versus floppy ears), variant chr10:11072007	T/T	The dog is homozygous and carries two copies of a genetic variant typically associated with pricked ears. This genotype is common in breeds like Finnish Spitz, German Shepherd, Samoyed, Terriers and in Collie-related breeds.
Bobtail	C/C	The dog does not carry any copy of the bobtail mutation. It therefore likely has a long-tailed phenotype.
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).

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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, Grey Collie Syndrome, (CN)	Autosomal Recessive	Clear
Canine Scott Syndrome, (CSS)	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 (4 mutations)	X-linked 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 (3 mutations)	Autosomal Recessive	Clear
Trapped Neutrophil Syndrome, (TNS)	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type II	Autosomal Recessive	Clear

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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 (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
Dominant Progressive Retinal Atrophy, (DPRA)	Autosomal Dominant	Clear
Golden Retriever Progressive Retinal Atrophy 1, (GR_PRA 1)	Autosomal Recessive	Clear
Golden Retriever Progressive Retinal Atrophy 2, (GR_PRA 2)	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
X-Linked Progressive Retinal Atrophy 2, (XLPRA2)	X-linked Recessive	Clear

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

Endocrine Disorders

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

Immunologic 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

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
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

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

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); mutation originally found in Dachshund	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); 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 (MDL), Ullrich-type; mutation originally found in Landseer	Autosomal Recessive	Clear
Muscular Hypertrophy (Double Muscling)	Autosomal Recessive	Clear
Myotonia Congenita; mutation originally found in Australian Cattle Dog	Autosomal Recessive	Clear
Myotubular Myopathy; mutation originally found in Rottweiler	X-linked Recessive	Clear

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

Neurologic 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
Degenerative Myelopathy, (DM)	Autosomal Recessive (Incomplete Penetrance)	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; 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)	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis 8, (NCL8) (2 mutations)	Autosomal Recessive	Clear
Neuronal Ceroid Lipofuscinosis, (NCL7); mutation originally found in Chinese Crested Dog and Chihuahua	Autosomal Recessive	Clear

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

Neurologic Disorders - page 2

Disorder	Mode of Inheritance	Result
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 Old Danish Pointing Dog	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

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

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; Cleft Lip and Palate with Syndactyly; 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
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

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

Dermal Disorders

Disorder	Mode of Inheritance	Result
Dystrophic Epidermolysis Bullosa (2 mutations)	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
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
Multi-Drug Resistance 1, (MDR1) or Ivermectin Sensitivity	Autosomal Dominant	Clear

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

Other Disorders

Disorder	Mode of Inheritance	Result
Amelogenesis Imperfecta, (AI)	Autosomal Recessive	Clear
Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatosis, (CKCSID)	Autosomal Recessive	Clear
Dental Hypomineralization; mutation originally found in Border Collie	Autosomal Recessive	Clear
Narcolepsy (2 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

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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.

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OPTIMAL SELECTION™ DNA TEST TERMS AND CONDITIONS

Optimal Selection™ Genetic Breeding Analysis is a proprietary process designed and intended to be used on purebred dogs solely to 1) Help quantify the genetic compatibility of potential breeding pairs and 2) To identify specific alleles or DNA mutations that are associated with certain inherited diseases or traits. No other purpose is authorized or permitted. It is not intended to diagnose diseases or predict behavior in any particular dog.

Upon receipt of your dog's DNA sample, Mars Veterinary will analyze your dog's DNA to determine chromosomal similarities and differences in the genetic profile of a potential sire and dam and provide a match analysis. Your dog's DNA will also be analyzed for the presence of specific alleles that are associated with inherited conditions identified as occurring in your dog's breed. Mars Veterinary's testing procedures are designed to provide reliable and accurate results, but are not guaranteed. By submitting your dog's sample(s) for Optimal Selection™ analysis it is understood that you agree that the sample(s), analysis, results and related information may be used confidentially by Mars in conjunction with other samples to increase the understanding of the breed's genetic structure, as well as for internal, research and development, or statistical purposes and may be shared with third parties for these purposes.

Samples may be disposed of or stored at Mars Veterinary's option and will not be returned. Please view the full Mars Privacy Policy here: http://www.mars.com/global/policies/privacy/pp-english.aspx It is also understood that future releases of the Optimal Selection™ test may refine results as more information is obtained regarding the breed structure and/or if new genetic markers are included.

Optimal Selection™ genetic assessments for individual dogs and potential mates will be available online to the person(s) who registered the sample. A dog's results, photo and other information may be shared by the owner with other individuals whom they choose or transferred to a new owner if the dog changes ownership. The content of such online services 1) may be altered due to changes, additions, or removals of a dog's information in the Optimal Selection™ database or due to changes in technical or other design of such services and 2) includes information about third parties and other Mars Veterinary clients' dogs, which Mars Veterinary is not responsible or liable for. Mars Veterinary has right to terminate access to online services one year from the purchase date, unless a longer period has been agreed upon.

You agree to Mars Veterinary instructions related to ordering process, payment, sampling and sample delivery. You also certify that the animal described in your order is the same animal whose sample is submitted for analysis, and that all information is accurate. You warrant that you are entitled to obtain and supply samples to Mars Veterinary.

In the unlikely event that it is not possible to provide an analysis (for example due to an insufficient DNA sample) or that an error in the analysis occurs, liability by Mars Veterinary or related companies and individuals is disclaimed and damages in any event are limited to the payment actually received by Mars Veterinary for the specified analysis at issue. Mars Veterinary's study of the complexities of the canine genome is ongoing with the goal of continuing to provide the most advanced and complete analysis possible.

Mars Veterinary reserves the right to use any third party of its choice to undertake the testing, analysis or laboratory services for the analysis.