



8700 2412 5318

Nari, Shiba

Call Name: Nari

Microchip: 191100000742246 HRV

Breed: Shiba

Gender: Female

Owner: Jelena Kučić

Country: Croatia

Testing date: 2016/9/26

DNA identification profile: Identified with standard ISAG 2006 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 |
|--|-------------------------|---------------------|--------|
| GM1 Gangliosidosis; mutation originally found in Shiba Dog | Neuromuscular Disorders | Autosomal Recessive | Clear |

On behalf of Genoscoper Laboratories,

SIGNATURE

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



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Test results - Traits - page 1

| Trait | Genotype | Description |
|---|----------------------------|--|
| Color Locus E - Extensions | E/E | The dog is likely to express the coat color defined by the K and A loci. |
| 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. |
| Furnishings / Improper Coat in Portuguese Water Dogs (marker test) | GG/CC | The dog is not genetically likely to express furnishings. |
| 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 - Traits - page 2

| 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). |
| Color Locus C - Albinism (caL-allele) | C/C | This dog does not carry the tested mutation for albinism. |
| 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. |
| Coat length, FGF5 gene variant | G/T | 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. |
| Curly coat | C/C | The dog is genetically non-curly. |
| 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 - 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 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 I | Autosomal Recessive | Clear |
| Von Willebrand's Disease (vWD) Type II | Autosomal Recessive | Clear |
| Von Willebrand's Disease (vWD) Type III (3 mutations) | Autosomal Recessive | Clear |



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

Ocular Disorders - page 1

| Disorder | Mode of Inheritance | Result |
|--|---|--------|
| Canine Multifocal Retinopathy 1, (CMR1); Mastiff-related breeds mutation | 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 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 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 |
| Early Retinal Degeneration, (erd); mutation originally found in Norwegian Elkhound | 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 |
| 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 |



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

Ocular Disorders - page 2

| Disorder | Mode of Inheritance | Result |
|---|---------------------|--------|
| 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 | Clear |

Cardiac Disorders

| Disorder | Mode of Inheritance | Result |
|------------------|---------------------|--------|
| Long QT Syndrome | Autosomal Dominant | Clear |

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 |
|---|---------------------|--------|
| 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 |



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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 and Cystinuria Type II-B (2 mutations) | Autosomal Dominant | Clear |
| Fanconi Syndrome | | 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 |
| 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 |



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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 (MDL), Ullrich-type; mutation originally found in Landseer | Autosomal Recessive | Clear |
| Muscular Hypertrophy (Double Muscling) | 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

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 |
| Cerebral Dysfunction; mutation originally found in Friesian Stabyhoun | Autosomal Recessive | Clear |
| Dandy-Walker-Like Malformation (DWLM); mutation originally found in Eurasier | 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; 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 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 |
| Neuronal Ceroid Lipofuscinosis 8, (NCL8) (2 mutations) | Autosomal Recessive | Clear |



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

Neurologic Disorders - page 2

| Disorder | Mode of Inheritance | Result |
|--|---------------------|--------|
| 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 Jack Russell Terrier | Autosomal Recessive | Clear |
| 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 |
| GM1 Gangliosidosis (2 mutations) | Autosomal Recessive | 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 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; DLX6 gene mutation originally found in Nova Scotia Duck Tolling Retriever | 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 |
| 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, (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 |



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

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/DH) | | 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 |
| Multidrug resistance 1 (MDR1 gene mutation) | Autosomal Recessive | Clear |



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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 Dermatitis, (CKCSID) | Autosomal Recessive | Clear |
| Dental Hypomineralization; 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 |



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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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