



Registered name: Chiquinha do BR Terra de Vera Owner: Mauro Sérgio Carvalho

Cruz Prizibela

Nickname: Chiquinha Country: Brazil

Registration ID: RG/PRG/10/04038 **Testing date:** 30/4/2014

Microchip: none

Breed: Terrier Brazileiro

DNA identification profile:

Gender: Female

Identified with standard ISAG

markers



Test results - Known disorders in the breed

| Disorder | Туре | Mode of inheritance | Result |
|---|---------------------|---------------------|--------|
| Mucopolysaccharidosis Type VII (MPSVII); mutation originally found in Brazilian Terrier | Metabolic disorders | Autosomal Recessive | Clear |
| Malignant Hyperthermia (MH) | Pharmacogenetics | Autosomal Dominant | Clear |

Test results - New potential disorders in the breed

| Disorder | Туре | Mode of inheritance | Result |
|--|-----------|---------------------|---------|
| Canine Multifocal Retinopathy 1 (cmr1), Mastiff-related breeds | Eye | Autosomal Recessive | Carrier |
| mutation | disorders | | |

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,

SIGNATURE

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





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Test results - Traits

| Trait | Genotype | Description |
|---|-------------------------|--|
| Colour Locus A | at/at | The dog is homozygous for at-allele. |
| Colour Locus B | B/B B/bd bd/bd | The dog does not carry any of the tested b alleles. |
| Colour Locus E | E/E | The dog is homozygous for E allele. |
| Colour Locus H | h/h | The dog is homozygous for h allele. |
| Colour Locus K | ky/ky | The dog is homozygous for ky allele. |
| Furnishings / Improper Coat in Portuguese Water Dogs (marker test) | GG/CC | The dog is not genetically likely to express furnishings. |
| Body mass, insulin-like growth factor 1 (IGF1) gene variant | A/A | The dog is homozygous for the genetic variant typically associated with small body mass. This genotype is common e.g. in Yorkshire Terrier, Chihuahua and Chinese Crested Dogs. |
| Coat length | G/G | The dog carries two copies of the genetic variant typically associated with a short-haired coat. |
| Curly coat | C/C | The dog is genetically non-curly. |
| 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. |
| 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. |
| 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). |
| 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)). |

On behalf of Genoscoper Laboratories,

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Test results - Additional disorders found in other breeds - page 1/6

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, Gly379Glu mutation | X-linked Recessive | Clear |
| Factor IX Deficiency or Haemophilia B; mutation originally found in Lhasa Apso | X-linked Recessive | Clear |
| Factor VII Deficiency | Autosomal Recessive | Clear |
| Factor VIII deficiency or Haemophilia A; mutation originally found in German Shepherd | 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; mutation originally found in Beagle | Autosomal Recessive | Clear |
| Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Labrador Retriever | Autosomal Recessive | Clear |
| Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in Pug | Autosomal Recessive | Clear |
| Pyruvate Kinase Deficiency of Erythrocyte; mutation originally found in West Highland White Terrier | Autosomal Recessive | Clear |
| Trapped Neutrophil Syndrome (TNS) | Autosomal Recessive | Clear |

Cardiological disorders

| Disorder | Mode of inheritance | Result |
|--|---------------------|--------|
| Dilated Cardiomyopathy; mutation originally found in Doberman Pinscher (USA) | Autosomal Dominant | Clear |



Test results - Additional disorders found in other breeds - page 2/6

Endocrine disorders

| Disorder | Mode of inheritance | Result |
|---|---------------------|--------|
| Hypothyroidism; mutation originally found in Tenterfield Terrier | Autosomal Recessive | Clear |
| Hypothyroidism; mutation originally found in Toy Fox- and Rat Terrier | Autosomal Recessive | Clear |

Eye disorders

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Test results - Additional disorders found in other breeds - page 3/6

Immunological disorders

| Disorder | Mode of inheritance | Result |
|--|---------------------|--------|
| ARSCID (Autosomal Recessive Severe Combined Immunodeficiency) | Autosomal Recessive | Clear |
| C3 deficiency | Autosomal Recessive | Clear |
| X-linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Basset Hound | X-linked Recessive | Clear |
| X-linked Severe Combined Immunodeficiency (XSCID); mutation originally found in Cardigan Welsh Corgi | X-linked Recessive | Clear |

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 |

Metabolic disorders

| Disorder | Mode of inheritance | Result |
|---|---------------------|--------|
| Glycogen Storage Disease, Type Ia (GSDIa) | Autosomal Recessive | Clear |
| Glycogen Storage Disease, type II or Pompe's disease | Autosomal Recessive | Clear |
| Glycogen Storage Disease, type Illa (GSDIlla) | Autosomal Recessive | Clear |
| Hypocatalasia or Acatalasemia | Autosomal Recessive | Clear |
| Mucopolysaccharidosis Type IIIA (MPSIIIA); mutation originally found in Dachshund | Autosomal Recessive | Clear |
| Mucopolysaccharidosis Type VI (MPSVI); mutation originally found in Poodle | Autosomal Recessive | Clear |
| Pyruvate Dehydrogenase Deficiency | Autosomal Recessive | Clear |



Chiquinha do BR Terra de Vera Cruz, Terrier Brazileiro

Test results - Additional disorders found in other breeds - page 4/6

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 |

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 |
| L-2-Hydroxyglutaric aciduria (L2HGA); mutation 1 originally found in Staffordshire Bull Terrier | Autosomal Recessive | Clear |
| L-2-Hydroxyglutaric aciduria (L2HGA); mutation 2 originally found in Staffordshire Bull Terrier | Autosomal Recessive | Clear |
| L-2-hydroxyglutaric Aciduria (L2HGA), Yorkshire Terrier mutation | Autosomal Recessive | Clear |
| Neonatal Encephalopathy with Seizures (NEWS) | Autosomal Recessive | Clear |
| Neuronal Ceroid Lipofuscinosis 1 (NCL1) | Autosomal Recessive | Clear |
| Neuronal Ceroid Lipofuscinosis 10 (NCL10) | Autosomal Recessive | Clear |
| Neuronal Ceroid Lipofuscinosis 2 (NCL2) | Autosomal Recessive | Clear |
| Neuronal Ceroid Lipofuscinosis 6 (NCL6) | Autosomal Recessive | Clear |
| Neuronal Ceroid Lipofuscinosis, type 12, mutation originally found in Tibetan terrier | Autosomal Recessive | Clear |
| Polyneuropathy; mutation originally found in Alaskan Malamute | Autosomal Recessive | Clear |
| Polyneuropathy; mutation originally found in Greyhound | Autosomal Recessive | Clear |
| Progressive early-onset cerebellar ataxia; mutation originally found in Finnish Hound | Autosomal Recessive | Clear |



Test results - Additional disorders found in other breeds - page 5/6

Neuromuscular disorders

| Disorder | Mode of inheritance | Result |
|--|---------------------|--------|
| Alpha Fucosidosis | Autosomal Recessive | Clear |
| Episodic falling (EF) | Autosomal Recessive | Clear |
| GM1 Gangliosidosis; mutation originally found in Alaskan Husky | Autosomal Recessive | Clear |
| GM1 Gangliosidosis; mutation originally found in Portuguese Water Dog | Autosomal Recessive | Clear |
| GM1 Gangliosidosis; mutation originally found in Shiba Dog | 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 |
| Globoid Cell Leukodystrophy (GLD) or Krabbe's disease; mutation originally found in Irish Setter | Autosomal Recessive | Clear |
| Hyperekplexia or Startle Disease | 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 | Clear |
| Osteogenesis imperfecta (OI) or Brittle Bone Disease; mutation originally found in Dachshund | Autosomal Recessive | Clear |
| Skeletal Dysplasia 2 (SD2) | Autosomal Recessive | Clear |



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

Skin disorders

| Disorder | Mode of inheritance | Result |
|---|---------------------|--------|
| Ectodermal dysplasia or Skin Fragility Syndrome (ED-SFS) | Autosomal Recessive | Clear |
| Epidermolysis bullosa, dystrophic | Autosomal Recessive | Clear |
| Epidermolytic Hyperkeratosis or Ichthyosis in Norfolk Terrier | Autosomal Recessive | Clear |
| Lamellar Ichthyosis (LI) | 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 |
| Gallbladder Mucocele Formation | Autosomal Dominant | Clear |
| Narcolepsy; mutation originally found in Dobermann | Autosomal Recessive | Clear |
| Persistant Mullerian 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.

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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