



8700 2322 2457 318  
Dog1, Cirneco dell'Etna

**Call Name:** Dog1

**Owner:** Diana Elisova

**Microchip:** 967000009623795

**Country:** Italy

**Breed:** Cirneco dell'Etna

**Testing date:** 2016/5/6

**Gender:** Female

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

## 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	e/E	The dog is likely to express the coat color defined by the K and A loci. The dog carries recessive red.
Color Locus B - Brown	bc/bs	The dog is likely to have brown coat.
Color Locus K - Dominant Black	KB/ky    kbr/ky	The dog is genetically dominant black or brindle.
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.

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## Test results - Traits - page 2

Trait	Genotype	Description
Furnishings / Improper Coat in Portuguese Water Dogs (marker test)	GG/CC	The dog is not genetically likely to express furnishings.
Color Pattern - Saddle Tan	-/-	The dog may have saddle tan pattern if it has also tan point genotype at the A locus.
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.
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	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.
Curly coat	C/C	The dog is genetically non-curly.
Coat length /	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	A/A	Your dog is homozygous for a genetic variant typically found in tiny-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
Canine Leucocyte Adhesion Deficiency (CLAD), type III	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
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 SNP 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 (5 mutations)	Autosomal Recessive	Clear
Thrombopathia (3 mutations)	Autosomal Recessive	Clear
Trapped Neutrophil Syndrome, (TNS)	Autosomal Recessive	Clear
Von Willebrand's Disease (vWD) Type II	Autosomal Recessive	Clear



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

### Ocular Disorders

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 (2 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
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
Glaucoma in the Norwegian Elkhound	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
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 1, (XLPRA1)	X-linked Recessive	Clear
X-Linked Progressive Retinal Atrophy 2, (XLPRA2)	X-linked Recessive	Clear



## 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
Autosomal Recessive Severe Combined Immunodeficiency, (ARSCID)	Autosomal Recessive	Clear
Complement 3 (C3) Deficiency	Autosomal Recessive	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 and Cystinuria Type II-B (2 mutations)	Autosomal Dominant	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
Renal Cystadenocarcinoma and Nodular Dermatofibrosis, (RCND)	Autosomal Dominant	Clear
X-Linked Hereditary Nephropathy, (XLHN)	X-linked Recessive	Clear



## 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 Acatalasemia	Autosomal Recessive	Clear
Intestinal Cobalamin Malabsorption or Imerslund-Gräsbeck Syndrome, (IGS) (2 mutations)	Autosomal Recessive	Clear
Mucopolysaccharidosis Type 3A, (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

### 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)	X-linked Recessive	Clear
Muscular Hypertrophy (Double Muscling)	Autosomal Recessive	Clear
Myotonia Congenita (2 mutations)	Autosomal Recessive	Clear
X-Linked Myotubular Myopathy	X-linked Recessive	Clear





## 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
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
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 in Weimaraners	Autosomal Recessive	Clear
L-2-Hydroxyglutaric aciduria, (L2HGA); mutation 1 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
Neuronal Ceroid Lipofuscinosis 8, (NCL8) and NCL8 rare variant (2 mutations)	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	No call
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



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

### Neuromuscular Disorders

Disorder	Mode of Inheritance	Result
Congenital Myasthenic Syndrome, (CMS)	Autosomal Recessive	Clear
Episodic Falling, (EF)	Autosomal Recessive	Clear
GM1 Gangliosidosis (3 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

### 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
Hereditary Vitamin D-Resistant Rickets, (HVDRR)	Autosomal Recessive	Clear
Oculoskeletal Dysplasia 2 or Dwarfism-Retinal Dysplasia 2	Autosomal Recessive	Clear
Osteochondrodysplasia in Miniature Poodles	Autosomal Recessive	Clear
Osteogenesis Imperfecta, (OI) found in the Beagle		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 8

### Dermal Disorders

Disorder	Mode of Inheritance	Result
Dystrophic Epidermolysis Bullosa	Autosomal Recessive	Clear
Epidermolytic Hyperkeratosis	Autosomal Recessive	Clear
Hereditary Footpad Hyperkeratosis, (HFH)	Autosomal Recessive	Clear
Lamellar Ichthyosis, (LI)	Autosomal Recessive	Clear
Musladin-Lueke syndrome, (MLS)	Autosomal Recessive	Clear
X-Linked Ectodermal Dysplasia, (XHED)	X-linked Recessive	Clear

### Other Disorders

Disorder	Mode of Inheritance	Result
Amelogenesis Imperfecta, (AI)	Autosomal Recessive	Clear
Cleft palate; DLX6 gene mutation originally found in Nova Scotia Duck Tolling Retriever	Autosomal Recessive	Clear
Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis, (CKCSID)	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.

## Genoscooper Laboratories - Legal Notice

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