



19/2/2015

MyDogDNA PASS

8700 2321 8864 691

Echo de'Chien Bellatrix, Pyrenean Mountain Dog

Registered name: Echo de'Chien Bellatrix

Nickname: Bella

Registration ID: EST-00068/12

Microchip: 981098102852559

Breed: Pyrenean Mountain Dog

Gender: Female

Owner: Marion-Silvia Diener

Country: Estonia

Testing date: 18/2/2015

DNA identification profile:
Identified with standard ISAG
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
Canine Multifocal Retinopathy 1 (cmr1), Mastiff-related breeds mutation	Eye disorders	Autosomal Recessive	Clear
Glanzmann Thrombasthenia (GT), Type I; mutation originally found in Pyrenean Mountain Dog	Blood disorders	Autosomal Recessive	Clear
Malignant Hyperthermia (MH)	Pharmacogenetics	Autosomal Dominant	Clear

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 Genoscooper Laboratories,

SIGNATURE

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



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Test results - Traits

Trait	Genotype	Description
Colour Locus A	ay/ay	The dog is homozygous for ay-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 heterozygous for e allele and 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	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 / "Fluffy" in Welsh Corgi	T/T	The dog carries two copies of the genetic variant typically associated with a long-haired coat. Dogs with this genotype typically have long coat.
Curly coat	C/C	The dog is genetically non-curly.
Ear erectness (pricked ears versus floppy ears), variant chr10:11072007	C/C	Your dog is homozygous for (carries two copies of) a genetic variant typically associated with floppy ears. This genotype is common in breeds like English Springer Spaniel, Leonberger, Saluki, and Dachshunds. Interestingly, the C-allele of this variant is the ancestral allele frequent in wolf.
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)).

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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 Dog	X-linked 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
Congenital hypothyroidism; mutation originally found in Toy Fox- and Rat Terrier	Autosomal Recessive	Clear
Hypothyroidism; mutation originally found in Tenterfield Terrier	Autosomal Recessive	Clear

Eye disorders

Disorder	Mode of inheritance	Result
Achromatopsia or Cone Degeneration (CD); mutation originally found in German Shorthaired Pointer	Autosomal Recessive	Clear
Autosomal Dominant Progressive Retinal Atrophy (ADPRA)	Autosomal Dominant	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-rod Dystrophy (cord1-PRA / crd4)	Autosomal Recessive	Clear
Cone-rod dystrophy (crd SWD); mutation originally found in Standard Wire-haired Dachshund	Autosomal Recessive	Clear
Generalized Progressive Retinal Atrophy; mutation originally found in Schapendoes	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; mutation originally found in Beagle	Autosomal Recessive	Clear
Rod-Cone Dysplasia 1 (rcd1); mutation originally found in Irish Setter	Autosomal Recessive	Clear
Rod-Cone Dysplasia 1a (rcd1a); mutation originally found in Sloughi	Autosomal Recessive	Clear
Rod-Cone Dysplasia 3 (rcd3)	Autosomal Recessive	Clear
X-Linked Progressive Retinal Atrophy 1 (XLPRA1)	X-linked Recessive	Clear



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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 IIIa (GSDIIIa)	Autosomal Recessive	Clear
Hypocatalasia or Acatlasemia	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
Mucopolysaccharidosis Type VII (MPSVII); mutation originally found in Brazilian Terrier	Autosomal Recessive	Clear
Pyruvate Dehydrogenase Deficiency	Autosomal Recessive	Clear



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
Hyperekplexia or Startle Disease	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
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



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



Neuromuscular disorders

Disorder

Alpha Fucosidosis
 Episodic falling (EF)
 GM1 Gangliosidosis; mutation originally found in Alaskan Husky
 GM1 Gangliosidosis; mutation originally found in Portuguese Water Dog
 GM1 Gangliosidosis; mutation originally found in Shiba Dog
 GM2 Gangliosidosis; mutation originally found in Toy Poodle
 Globoid Cell Leukodystrophy (GLD) or Krabbe's disease, Terrier mutation

Mode of inheritance

Autosomal Recessive
 Autosomal Recessive
 Autosomal Recessive
 Autosomal Recessive
 Autosomal Recessive
 Autosomal Recessive
 Autosomal Recessive

Result

Clear
 Clear
 Clear
 Clear
 Clear
 Clear
 Clear

Skeletal disorders

Disorder

Chondrodysplasia (dwarfism); mutation originally found in Norwegian Elkhound and Karelian Bear Dog
 Craniomandibular Osteopathy (CMO)
 Osteogenesis imperfecta (OI) or Brittle Bone Disease; mutation originally found in Dachshund
 Skeletal Dysplasia 2 (SD2)

Mode of inheritance

Autosomal Recessive
 Autosomal Dominant
 Autosomal Recessive
 Autosomal Recessive

Result

Clear
 Clear
 Clear
 Clear



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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
Musladin-Lueke syndrome (MLS)	Autosomal Recessive	Clear

Other disorders

Disorder	Mode of inheritance	Result
Congenital Keratoconjunctivitis Sicca and Ichthyosiform Dermatitis (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
Persistent 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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