

Laboklin GmbH & Co. KG, Steubenstraße 4, 97688 Bad Kissingen

Mr.
Andre Melby
Duaasen 34
2100 Skarnes
Norwegen

Report No.:	2503-W-746359
Date of arrival:	20.03.2025
Date of report:	01.04.2025
Testing started:	20.03.2025
Testing completed:	01.04.2025
Status of the report:	Final report

Species:	Dog
Breed:	Miniature American Shepherd
Gender:	Female
Name:	Mocca - Qubera Recinto's Bitcoin
Stud book No.:	NO48072/23
Chip No.:	578098100814751
Date of birth / Age:	13.06.2023
Type of sample:	EDTA-Blood
Date sample was taken:	12.03.2025
Owner / Animal-ID:	Melby, Andre
IT No. / Report-ID:	---

Breeding club discounts were granted for discountable services!



In genetic testing, we analyse the genetic variants associated with hereditary diseases or genetic traits. The results of these genetic tests always show both alleles of the animal for the variant that has been tested. The symbol "N" indicates the presence of the wild-type allele, while the variant alleles are designated according to the associated diseases (in the example referred to as 'mut').

Possible results:

- N/N: The genetic variant associated with the disease is absent.
- N/mut: The tested animal carries one copy of the analysed variant.
- mut/mut: The tested animal carries two copies of the analysed variant.

It is important to note that solely relying on this genetic information cannot provide definitive insight into whether, when, or to what extent a disease may manifest. For certain diseases, the severity of the condition is influenced by additional factors, some of which are not detectable through genetic testing. Variable penetrance, which involves varying degrees of severity, also frequently plays a role. In cases of recessive hereditary diseases, the disease usually only manifests when an individual possesses two copies of the investigated variant. In contrast, for dominant hereditary diseases, the presence of a single copy of the variant already influences the likelihood of disease occurrence. The annotation numbers **r** (autosomal recessive), **d** (autosomal dominant), and **Xr** (X chromosomal recessive) indicate the respective mode of inheritance.

Not every noticeable result necessarily has health consequences for the animal or its offspring. In cases where a dog is heterozygous (a carrier) for a monogenic autosomal recessive disease, the detected findings have no impact on the animal's health and, when bred with a clear dog, pose no risk to the offspring.

The following applies to non-breed-specific results:

So far, no correlation between the tested variant and the associated clinical symptoms has been scientifically proven in the breed of this animal.

For more comprehensive information regarding specific hereditary diseases, please refer to our website.

BREED SPECIFIC VARIANTS

Noticeable results	Genotype	Gene	Variant
Brachyuria ^d	N/ST	TBXT	G-C
Unremarkable results	Genotype	Gene	Variant
Canine Multi-Focal Retinopathy CMR1 ^r	N/N	BEST1	G-A
Chondrodysplasia (CDPA) ^d	N/N	FGF4/ CFA18	COMPLEX
Chondrodystrophy (CDDY and IVDD risk) ^d	N/N	FGF4/ CFA12	COMPLEX
Collie Eye Anomaly (CEA) ^{r,1}	N/N	NHEJ1	COMPLEX
Degenerative Myelopathy ^{r,2,4}	N/N (exon 2)	SOD1	G-A
Hereditary Ataxia (HA) - Australian Shepherd ^r	N/N	PNPLA8	INS
Hyperuricosuria ^r	N/N	SLC2A9	G-T
Malignant Hyperthermia (MH) ^d	N/N	RYR1	A-G
MDR1 gene variant ^r	N/N (+/+)	ABCB1	DEL
Neuroaxonal Dystrophy (NAD) - Miniature American Shepherd ^r	N/N	RNF170	DEL
Neuronal Ceroid Lipofuscinosis CLN6 ^r	N/N	CLN6	A-G

BREED SPECIFIC VARIANTS

Unremarkable results	Genotype	Gene	Variant
Neuronal Ceroid Lipofuscinosis CLN8 - Australian Shepherd ^r	N/N	CLN8	G-A
Progressive Retinal Atrophy (prcd-PRA) ^{r,1}	N/N (A)	PRCD	C-T
von Willebrand disease type I (vWD1) ^{d,3}	N/N	VWF	G-A

BREED NON-SPECIFIC VARIANTS (NO CORRELATION DETECTED IN YOUR BREED)

Noticeable results	Genotype	Gene	Variant
Degenerative myelopathy risk modifier (DMRM) ^{d,4}	DMWM/DMWM	SP110	C-T
Dilated cardiomyopathy (DCM3) - Doberman ⁴	N/DCM3	---	G-A
Myxomatous Mitral Valve Disease (MMVD) ^{r,4}	N/MMVD	NEBL	G-A
Progressive Retinal Atrophy (crd3-PRA) associated SNP- Glen of Imaal Terrier ^{r,5}	crd3/crd3	associated SNP	C-T
Unremarkable results	Genotype	Gene	Variant
Acatalasemia ^r	N/N	CAT	C-T
Achromatopsia - German Shepherd Dog ^r	N/N	CNGA3	G-A
Achromatopsia - Labrador Retriever ^r	N/N	CNGA3	DEL
Acral Mutilation Syndrome (AMS) ^r	N/N	GNDF	C-T
Acute respiratory distress syndrome (ARDS) ^r	N/N	ANLN	C-T
Afibrinogenemia (AFG)	N/N	FGA	DEL
Alaskan Husky Encephalopathy (AHE) ^r	N/N	SLC19A3	COMPLEX
Alaskan Malamute Polyneuropathy (AMPN) ^r	N/N	NDRG1	C-A
Alexander Disease ^d	N/N	GFAP	G-A
Amelogenesis imperfecta (AI) - Akita ^r	N/N	ACP4	INS
Amelogenesis imperfecta (AI) - Italian Sighthound ^r	N/N	ENAM	DEL
Amelogenesis imperfecta (AI) - Parson Russell Terrier ^r	N/N	ENAM	C-T
Amelogenesis imperfecta (AI) - Samoyed ^r	N/N	SLC24A4	INS
Anhidrotic ectodermal dysplasia (EDA) - German Shepherd ^r	N/N	EDA	G-A
Bunny Hopping Syndrome (BHS1) ^r	N/N	EFNB3	INS
C3 Deficiency ^r	N/N	C3	DEL
Canine Leukocytes Deficiency (CLAD) ^r	N/N	ITGB2	C-G
Canine Multi-Focal Retinopathy CMR2 ^r	N/N	BEST1	C-T
Canine Multi-Focal Retinopathy CMR3 ^r	N/N	BEST1	DEL
Canine Multiple System Degeneration - Chinese Crested Dog ^r	N/N	SERAC1	DEL
Canine Multiple System Degeneration - Kerry Blue Terrier ^r	N/N	SERAC1	C-T
Cardiomyopathy with juvenile mortality (CJM) ^r	N/N	YARS2	G-A
Cerebellar ataxia (CA1) - Belgian Shepherd Dog ^r	N/N	RALGAPA1	COMPLEX
Cerebellar ataxia (CA1) - Pyrenean Mountain Dog ^r	N/N	SACS	DEL
Cerebellar degeneration and myositis complex (CDMC) ^r	N/N	SLC25A12	G-A
Cerebellar hypoplasia (CH) ^r	N/N	RELN	DEL
Cerebral dysfunction ^r	N/N	SLC6A3	G-A
Charcot-Marie-Tooth Neuropathy (CMT) ^r	N/N	SBF2	C-A
Chondrodysplasia - Chinook, Karelian Bear Dog, Norwegian Elkhound ^r	N/N	ITGA10	G-A
CNS atrophy with cerebellar ataxia (CACA) ^r	N/N	SELENOP	COMPLEX
Color dilution and neurological defects (CDN) ^r	N/N	MYO5A	INS
Comma Defect (Spondylocostal Dysostosis) ^r	N/N	HES7	DEL

BREED NON-SPECIFIC VARIANTS (NO CORRELATION DETECTED IN YOUR BREED)

Unremarkable results	Genotype	Gene	Variant
Cone Degeneration (CD) ^r	N/N	CNGB3	C-T
Congenital Hypothyroidism with goiter (CHG) - Fox, Rat Terrier ^r	N/N	TPO	C-T
Congenital Hypothyroidism with goiter (CHG) - French Bulldog ^r	N/N	TPO	T-C
Congenital Hypothyroidism with goiter (CHG) - Tenterfield Terrier ^r	N/N	TPO	C-T
Congenital myasthenic syndrome (CMS) - Golden Retriever ^r	N/N	LOC608697	G-A
Congenital myasthenic syndrome (CMS) - Labrador Retriever ^r	N/N	LOC608697	T-C
Congenital myasthenic syndrome (CMS) - Old Danish Pointing Dog ^r	N/N	CHAT	G-A
Congenital myasthenic syndrome (CMS) - Russell Terrier ^r	N/N	CHRNE	INS
Copper toxicosis - modifier ^{2,3}	X(N)/X(N) or X(N)/Y	ATP7A	C-T
Copper toxicosis - risk factor ^{d,2,3,4}	N/N	ATP7B	G-A
Craniomandibular Osteopathy (CMO) ^{d,3}	N/N (CMO-0)	SLC37A2	G-A
Cystinuria type IA - Labrador Retriever ^r	N/N	SLC3A1	DEL
Cystinuria type IA - Newfoundland ^r	N/N	SLC3A1	C-T
Cystinuria type IB ^d	N/N	SLC7A9	G-A
Cystinuria type IIA ^d	N/N	SLC3A1	DEL
Cystinuria type III ^{r,5}	N/N	SLC3A1	A-G
Delayed postoperative haemorrhage (DEPOH) - Deerhound, Greyhound ^{d,3,4}	N/N	SERPINF2	C-T
Dental-skeletal-retinal anomaly (DSRA) ^r	N/N	MIA3	DEL
Digital Hyperkeratosis (DH) - Dogue de Bordeaux ^r	N/N	KRT16	COMPLEX
Digital Hyperkeratosis (DH) - Irish Terrier, Kromfohlrländer ^r	N/N	FAM83G	G-C
Dilated cardiomyopathy (DCM) - Giant Schnauzer, Standard Schnauzer ^r	N/N	RBM20	DEL
Dilated cardiomyopathy (DCM) - Manchester Terrier ^{r,4}	N/N	ABCC9	G-A
Dilated cardiomyopathy (DCM) - Nova Scotia Duck Tolling Retriever ^{r,4}	N/N	LMNA	DEL
Dilated cardiomyopathy (DCM) - Welsh Springer Spaniel ^{3,4}	N/N	PLN	G-A
Dilated cardiomyopathy (DCM1) - Dobermann ⁴	N/N	PDK4	DEL
Dilated cardiomyopathy (DCM2) - Dobermann ⁴	N/N	TTN	C-T
Dilated cardiomyopathy (DCM4) - Dobermann ⁴	N/N	---	G-A
Disproportionate dwarfism - Dogo Argentino ^r	N/N	PRKG2	C-A
Disproportionate dwarfism - Magyar Vizsla ^r	N/N	PCYT1A	A-G
Dwarfism (growth-hormone deficiency) - Chihuahua ^r	N/N	GH1	DEL
Dwarfism (Skeletal Dysplasia 2) ^r	N/N	COL11A2	C-G
Dyserythropoietic anemia and myopathy (DAMS) - Labrador Retriever ^r	N/N	EHBP1L1	G-A
Dystrophic Epidermolysis bullosa (DEB) - Central Asian Shepherd ^r	N/N	COL7A1	C-T
Dystrophic Epidermolysis bullosa (DEB) - Golden Retriever ^r	N/N	COL7A1	G-A
Ehlers-Danlos-Syndrome - Catahoula Leopard Dog ^r	N/N	ADAMTS2	G-A
Ehlers-Danlos-Syndrome - Doberman ^r	N/N	ADAMTS2	C-T
Epidermolytic hyperkeratosis ^r	N/N	KRT10	G-T

BREED NON-SPECIFIC VARIANTS (NO CORRELATION DETECTED IN YOUR BREED)

Unremarkable results	Genotype	Gene	Variant
Episodic Falling ^{r,2}	N/N	BCAN	COMPLEX
Exercise Induced Collapse (EIC) ^{r,2}	N/N	DNM1	C-A
Exfoliative Cutaneous Lupus Erythematosus (ECLE) ^r	N/N	UNC93B1	C-A
Factor VII Deficiency ^r	N/N	F7	G-A
Familial Nephropathy (FN) - English Cocker Spaniel, Welsh Springer Spaniel ^{r,1}	N/N	COL4A4	T-A
Familial Nephropathy (FN) - English Springer Spaniel ^r	N/N	COL4A4	G-A
Familial Nephropathy (FN) - Samoyed ^{Xr}	female X(N)/X(N) male X(N)/Y	COL4A5	G-T
Familial thyroid follicular cell carcinoma risk factor 1 - German Longhair ⁴	N/N	TPO	G-A
Familial thyroid follicular cell carcinoma risk factor 2 - German Longhair ⁴	N/N	TPO	C-T
Fanconi-Syndrom	N/N	FAN1	COMPLEX
Finnish Hound Ataxia ^r	N/N	SEL1L	A-G
Gallbladder Mucocoeles (GBM) ^{d,3}	N/N	ABCB4	INS
Glanzmann's Thrombasthenia (GT) ^r	N/N	ITGA2B	DEL
Glaucoma and Goniodysgenesis (GG) ^r	N/N	OLFML3	G-A
Globoid Cell Leukodystrophy - Terrier ^r	N/N	GALC	T-G
Glycogen Storage Disease GSD Ia - Maltese ^r	N/N	G6PC	C-G
Glycogene storage disease (GSD IIIa) ^r	N/N	AGL	DEL
GM1-Gangliosidosis - Husky ^r	N/N	GLB1	INS
GM1-Gangliosidosis - Portuguese Water Dog ^r	N/N	GLB1	G-A
GM1-Gangliosidosis - Shiba ^r	N/N	GLB1	DEL
GM2-Gangliosidosis - Japanese Chin ^r	N/N	HEXA	C-T
GM2-Gangliosidosis - Miniature Poodle ^r	N/N	HEXB	G-T
GM2-Gangliosidosis - Shiba Inu ^r	N/N	HEXB	DEL
Haemophilia A - Boxer ^{Xr}	female X(N)/X(N) male X(N)/Y	F8	G-C
Haemophilia A - German Shepherd Dog ^{Xr}	female X(N)/X(N) male X(N)/Y	F8	C-T
Haemophilia A - Labrador Retriever ^{Xr}	f X(N)/X(N) m X(N)/Y	F8	DEL
Haemophilia A - Old English Sheepdog ^{Xr}	female X(N)/X(N) male X(N)/Y	F8	G-A
Haemophilia B - American Akita ^{Xr,2}	female X(N)/X(N) male X(N)/Y	F9	T-C
Haemophilia B - Hovawart ^{Xr,2}	female X(N)/X(N) male X(N)/Y	F9	DEL
Haemophilia B - Lhasa Apso ^{Xr,2}	female X(N)/X(N) male X(N)/Y	F9	COMPLEX
Haemophilia B - Rhodesian Ridgeback ^{Xr,2}	female X(N)/X(N) male X(N)/Y	F9	G-A
Haemorrhagic diathesis (Scott Syndrom) ^r	N/N	ANO6	C-T
Hereditary Ataxia (HA) - Malinois ^r	N/N	SLC12A6	COMPLEX
Hereditary Ataxia (HA) - Norwegian Buhund ^r	N/N	KCNIP4	T-C
Hereditary Ataxia (HA) - Norwegian Elkhound ^r	N/N	HACE1	DEL
Hereditary Ataxia (HA) - Old English Sheepdog, Gordon Setter ^r	N/N	RAB24	A-C
Hereditary Cataract - Wire-Haired Pointing Griffon	N/N	FYCO1	DEL
Korthals ^r			
Hereditary deafness (DINGS1) ^r	N/N	PTPRQ	INS
Hereditary deafness (DINGS2) - Doberman ^r	N/N	MYO7A	C-T

BREED NON-SPECIFIC VARIANTS (NO CORRELATION DETECTED IN YOUR BREED)

Unremarkable results	Genotype	Gene	Variant
Hereditary deafness/Hearing loss (EOAD) - Beauceron ^r	N/N	CDH23	C-T
Hereditary deafness/Hearing loss (EOAD) - Rhodesian Ridgeback ^r	N/N	EPS8L2	DEL
Hereditary deafness/Hearing loss (EOAD) - Rottweiler ^r	N/N	LOXHD1	G-C
Hereditary myopathy (CNM) - German Hunting Terrier ^r	N/N	ACADVL	C-A
Hereditary myopathy (CNM) - Great Dane ^r	N/N	BIN1	A-G
Hereditary nasal parakeratosis (HNPK) - Greyhound ^r	N/N	SUV39H2	DEL
Hereditary nasal parakeratosis (HNPK) - Labrador Retriever ^{r,2}	N/N	SUV39H2	A-C
Hereditary Neuropathy ^r	N/N	NDRG1	DEL
Hereditary vitamin D-dependent rickets (HVDDR) ^r	N/N	VDR	DEL
Hypomyelination - English Springer Spaniel ^r	N/N	PLP1	A-C
Hypomyelination - Weimaraner ^r	N/N	FNIP2	DEL
Hypophosphatasia (HPP) ^r	N/N	ALPL	A-C
Ichthyosis - American Bulldog ^r	N/N	NIPAL4	DEL
Ichthyosis - Chihuahua ^r	N/N	SDR9C7	G-A
Ichthyosis - German Shepherd ^d	N/N	ASPRV1	A-G
Ichthyosis - Golden Retriever ^r	N/N	PNPLA1	INS
Ichthyosis type 2 - Golden Retriever	N/N	ABHD5	DEL
Imerslund-Gräsbeck-Syndrome (IGS) - Beagle ^r	N/N	CUBN	DEL
Imerslund-Gräsbeck-Syndrome (IGS) - Border Collie ^r	N/N	CUBN	DEL
Imerslund-Gräsbeck-Syndrome (IGS) - Komondor ^r	N/N	CUBN	G-A
Inflammatory Myopathy (IM) - Dutch Shepherd Dog ^r	N/N	SLC25A12	A-G
Inflammatory pulmonary disease (IPD) ^r	N/N	AKNA	DEL
Junctional epidermolysis bullosa (JEB) ^r	N/N	LAMA3	G-A
Juvenile Brain disease (JBD) ^r	N/N	PITRM1	DEL
Juvenile Epilepsy (JE) - Lagotto Romagnolo ^r	N/N	LGI2	A-T
Juvenile Laryngeal Paralysis & Polyneuropathy (JLPP) ^r	N/N	RAB3GAP1	DEL
Juvenile Myoclonic Epilepsy (JME) ^r	N/N	DIRAS1	DEL
L-2-Hydroxyglutaric Aciduria (L2HGA) - Staffordshire Bull Terrier ^r	N/N	L2HGDH	COMPLEX
Lagotto Storage Disease (LSD) ^r	N/N	ATG4D	C-T
Laryngeal paralysis with polyneuropathy type 3 (LPPN3) ^r	N/N	CNTNAP1	C-T
Late onset Ataxia (LOA) ^r	N/N	CAPN1	C-T
Leonberger Polyneuropathy (LPN1) ^r	N/N	ARHGEF10	DEL
Leonberger Polyneuropathy (LPN2) ^{d,3}	N/N	GJA9	DEL
Lethal acrodermatitis ^r	N/N	MKLN1	T-G
Lethal lung disease (LAMP3) ^r	N/N	LAMP3	C-T
Leukocyte Adhesion deficiency III (LAD-III) ^r	N/N	FERMT3	INS
Leukoencephalomyelopathy (LEMP) - Leonberger ^r	N/N	NAPEPLD	G-C
Leukoencephalopathy (LEP) ^r	N/N	TSEN54	C-T
Limb-Girdle Muscular Dystrophy (LGMD) ^r	N/N	SGCA	G-A
Lundehund syndrome ^r	N/N	P3H2	C-G
Lysosomal storage disease (LSD) ^r	N/N	MAN2B1	A-G
Macrothrombocytopenia type A ^d	N/N	TUBB1	G-A
Macrothrombocytopenia type B ^r	N/N	TUBB1	G-A
Macular corneal dystrophy (MCD)	N/N	LOC489707	C-A
Maxillary canine tooth mesioversion (MCM) ^{d,4}	N/N	FTSJ3	T-C
May-Hegglin Anomaly (MHA) ^d	N/N	MYH9	G-A
MCAD deficiency ^r	N/N	ACADM	COMPLEX

BREED NON-SPECIFIC VARIANTS (NO CORRELATION DETECTED IN YOUR BREED)

Unremarkable results	Genotype	Gene	Variant
Methaemoglobinaemia (MetHg) ^r	N/N	CYB5R3	A-C
Microphthalmia (RBP4) ^r	N/N	RBP4	DEL
Mitochondrial fission encephalopathy (MFE) ^r	N/N	MFF	COMPLEX
Mucopolysaccharidosis (MPS) type IIIa - Dachshund (Dackel) ^r	N/N	SGSH	DEL
Mucopolysaccharidosis (MPS) type IIIa - New Zealand Huntaway ^r	N/N	SGSH	INS
Mucopolysaccharidosis (MPS) type VII - Brazilian Terrier ^r	N/N	GUSB	G-A
Mucopolysaccharidosis (MPS) type VII - German Shepherd Dog ^r	N/N	GUSB	C-T
Mucopolysaccharidosis Type VI - Miniature Pinscher	N/N	ARSB	G-A
Mucopolysaccharidosis type VI ^r	N/N	ARSB	C-T
Muscular Dystrophy - American Staffordshire Terrier ^r	N/N	COL6A3	DEL
Muscular Dystrophy - Cavalier King Charles Spaniel ^{Xr}	female X(N)/X(N) male X(N)/Y	DMD	C-A
Muscular Dystrophy - Golden Retriever ^{Xr}	female X(N)/X(N) male X(N)/Y	DMD	T-C
Muscular Dystrophy - Landseer ^r	N/N	COL6A1	G-T
Muscular Dystrophy - Norfolk Terrier ^{Xr}	female X(N)/X(N) male X(N)/Y	DMD	DEL
Muscular Dystrophy 2 - Cavalier King Charles Spaniel	f X(N)/X(N) m X(N)/Y	DMD	DEL
Musladin-Lueke Syndrome (MLS) ^r	N/N	ADAMTSL2	C-T
Mycobacterium avium complex sensitivity (MAC) ^r	N/N	CARD9	DEL
Myostatin variant ('Bully') - Whippet ^r	N/N	MSTN	DEL
Myotonia congenita - Labrador Retriever ^r	N/N	CLCN1	T-A
Myotonia congenita - Miniature Schnauzer ^r	N/N	CLCN1	G-A
Narcolepsy - Dachshund (Dackel) ^r	N/N	HCRTR2	G-A
Narcolepsy - Labrador Retriever ^r	N/N	HCRTR2	G-A
Necrotizing Meningoencephalitis (PDE) ^{r,4}	N/N	DLA-DPB1	DEL
Necrotizing myelopathy (HNM) ^r	N/N	IBA57	G-A
Nemalin myopathy ^r	N/N	NEB	G-T
Neonatal Cerebellar Abiotrophy (NCCD) - Beagle ^r	N/N	SPTBN2	DEL
Neonatal Cerebellar Abiotrophy (NCCD) - Hungarian Greyhound (Magyar agár) ^r	N/N	SNX14	C-T
Neonatal Encephalopathy ^r	N/N	ATF2	A-C
Neuroaxonal Dystrophy (NAD) - Lagotto Romagnolo, Spanish Water Dog ^r	N/N	TECPR2	C-T
Neuroaxonal Dystrophy (NAD) - Papillon ^r	N/N	PLA2G6	G-A
Neuroaxonal Dystrophy (NAD) - Rottweiler ^r	N/N	VPS11	T-C
Neuronal Ceroid Lipofuscinosis - American Staffordshire Terrier ^{r,2}	N/N	ARSG	G-A
Neuronal Ceroid Lipofuscinosis CLN1 - Dachshund (Dackel)	N/N	PPT1	INS
Neuronal Ceroid Lipofuscinosis CLN1 - Italian Cane Corso ^r	N/N	PPT1	G-A
Neuronal Ceroid Lipofuscinosis CLN10 - American Bulldog ^r	N/N	CTSD	C-T
Neuronal Ceroid Lipofuscinosis CLN12 - Tibetan Terrier ^r	N/N	ATP13A2	DEL

BREED NON-SPECIFIC VARIANTS (NO CORRELATION DETECTED IN YOUR BREED)

Unremarkable results	Genotype	Gene	Variant
Neuronal Ceroid Lipofuscinosis CLN12 Adult Onset - Australian Cattle Dog	N/N	ATP13A2	C-T
Neuronal Ceroid Lipofuscinosis CLN2 - Dachshund ^r	N/N	TPP1	DEL
Neuronal Ceroid Lipofuscinosis CLN5 - Border Collie, Australian Cattle Dog ^r	N/N	CLN5	C-T
Neuronal Ceroid Lipofuscinosis CLN5 - Golden Retriever ^r	N/N	CLN5	DEL
Neuronal Ceroid Lipofuscinosis CLN7 - Chihuahua, Chinese Crested Dog ^r	N/N	MFSD8	DEL
Neuronal Ceroid Lipofuscinosis CLN8 - Setter ^r	N/N	CLN8	T-C
Night Blindness (CSNB) ^r	N/N	RPE65	DEL
Obesity	N/N	POMC	DEL
Osteochondrodysplasia (OCD) ^r	N/N	SLC13A1	COMPLEX
Osteogenesis imperfecta - Beagle ^d	N/N	COL1A2	COMPLEX
Osteogenesis imperfecta - Dachshund (Dackel) ^r	N/N	SERPINH1	A-G
Osteogenesis imperfecta - Golden Retriever ^d	N/N	COL1A1	C-G
Paroxysmal Dyskinesia (PxD) ^r	N/N	PIGN	C-T
Paroxysmal Exercise-Induced Dyskinesia (PED) - Shetland Sheepdog (Sheltie) ^r	N/N	PCK2	G-A
Paroxysmal Exercise-Induced Dyskinesia (PED) - Weimaraner ^r	N/N	TNR	INS
Persistent Müllerian Duct Syndrome (PMDS) ^r	N/N	AMHR2	G-A
Phosphofructokinase Deficiency (PFKD) - American Cocker Spaniel, English Springer Spaniel, Whippet ^r	N/N	PFKM	C-T
Phosphofructokinase Deficiency (PFKD) - German Spaniel ^r	N/N	PFKM	G-A
Pituitary Dwarfism - Karelian Bear Dog, Lapponian Herder ^r	N/N	POU1F1	C-A
Polycystic Kidney Disease (PKD) ^d	N/N	PKD1	G-A
Polydactyly ^d	N/N	SHH	C-T
Pompe Disease (Glycogen storage disease II) ^r	N/N	GAA	C-T
Postoperative haemorrhage (P2Y12) - Great Swiss Mountain Dog ^d	N/N	P2RY12	DEL
Prekallikrein Deficiency ^r	N/N	KLKB1	A-T
Primary Ciliary Dyskinesia - Alaskan Malamute ^r	N/N	NME5	DEL
Primary Ciliary Dyskinesia - Old English Sheepdog ^r	N/N	CCDC39	G-A
Primary Lens Luxation (PLL) ^r	N/N	ADAMTS17	G-A
Primary open angle glaucoma (POAG) - Basset Fauve de Bretagne ^r	N/N	ADAMTS17	G-A
Primary open angle glaucoma (POAG) - Beagle ^r	N/N	ADAMTS10	C-T
Primary open angle glaucoma (POAG) - Norwegian Elkhound ^r	N/N	ADAMTS10	C-T
Primary open angle glaucoma and lens luxation (POAG/PLL) ^r	N/N	ADAMTS17	DEL
Progressive Retinal Atrophy (Bas-PRA1) - Basenji ^r	N/N	SAG	T-C
Progressive Retinal Atrophy (BBS2-PRA) - Shetland Sheepdog ^r	N/N	BBS2	G-C
Progressive Retinal Atrophy (BBS4-PRA) - Puli ^r	N/N	BBS4	A-T
Progressive Retinal Atrophy (CNGA1-PRA) - Shetland Sheepdog ^r	N/N	CNGA1	DEL
Progressive Retinal Atrophy (crd-PRA) - Dachshund ^r	N/N	NPHP4	COMPLEX

BREED NON-SPECIFIC VARIANTS (NO CORRELATION DETECTED IN YOUR BREED)

Unremarkable results	Genotype	Gene	Variant
Progressive Retinal Atrophy (crd1-PRA) - American Staffordshire Terrier ^r	N/N	PDE6B	DEL
Progressive Retinal Atrophy (dominant PRA) ^d	N/N	RHO	G-C
Progressive Retinal Atrophy (generalized PRA) - Schapendoes ^r	N/N	CCDC66	INS
Progressive Retinal Atrophy (GR-PRA1) - Golden Retriever ^r	N/N	SLC4A3	DEL
Progressive Retinal Atrophy (GR_PRA2) - Golden Retriever ^r	N/N	TTC8	DEL
Progressive Retinal Atrophy (GUCY2D-PRA) - German Spitz ^r	N/N	GUCY2D	INS
Progressive Retinal Atrophy (IFT122-PRA) - Lapponian Herder ^r	N/N	IFT122	C-T
Progressive Retinal Atrophy (JPH2-PRA) - Shih Tzu	N/N	JPH2	A-C
Progressive Retinal Atrophy (MERTK-PRA) - Swedish Vallhund ^r	N/N	MERTK	COMPLEX
Progressive Retinal Atrophy (NECAP1-PRA) - Giant Schnauzer ^r	N/N	NECAP1	G-A
Progressive Retinal Atrophy (Pap-PRA1) - Papillon ^r	N/N	CNGB1	INS
Progressive Retinal Atrophy (rcd1-PRA) - Irish Red Setter, Irish Red and White Setter ^r	N/N	PDE6B	C-T
Progressive Retinal Atrophy (rcd1a) - Sloughi ^r	N/N	PDE6B	INS
Progressive Retinal Atrophy (rcd3-PRA) - Welsh Corgi Cardigan, Pomeranian, Chinese Crested ^r	N/N	PDE6A	DEL
Progressive Retinal Atrophy (Type B1-PRA, HIVEP3) - Miniature Schnauzer ^r	N/N	HIVEP3	G-A
Progressive Retinal Atrophy early-onset (eo-PRA) - Portuguese Water Dog ^r	N/N	CCDC66	INS
Progressive Retinal Atrophy early-onset (eo-PRA) - Spanish Water Dog ^r	N/N	PDE6B	DEL
Protein Losing Nephropathy (PLN) ^r	N/N	KIRREL2, NPHS1	G-C, G-A
Pyruvat Dehydrogenase Phosphatase 1 Deficiency ^r	N/N	PDP1	C-T
Pyruvatkinase-Deficiency (PK) - Basenji ^r	N/N	PKLR	DEL
Pyruvatkinase-Deficiency (PK) - Beagle ^r	N/N	PKLR	G-A
Pyruvatkinase-Deficiency (PK) - Labrador Retriever ^r	N/N	PKLR	C-T
Pyruvatkinase-Deficiency (PK) - Pug ^r	N/N	PKLR	T-C
Raine Syndrome ^r	N/N	FAM20C	G-A
Renal Cystadenocarcinoma and Nodular Dermatofibrosis (RCND) ^d	N/N	FLCN	A-G
Renal dysplasia and hepatic fibrosis (RDHN) ^r	N/N	INPP5E	G-A
Retinal dysplasia (OSD) - Northern Inuit, Tamaskan ^r	N/N	COL9A3	C-T
Robinow-like syndrome (DVL2) ^{r,3,4}	N/N	DVL2	DEL
Sensory Neuropathy (SN) ^r	N/N	FAM134B	COMPLEX
Severe Combined Immunodeficiency (SCID) - Frisian Water Dog ^r	N/N	RAG1	C-A
Severe Combined Immunodeficiency (SCID) - Russell Terrier ^r	N/N	PRKDC	C-A
Shar Pei Autoinflammatory Disease (SPAID) ^{d,3}	N/N	MTBP	G-A
Spinal Dysraphism ^r	N/N	NKX2-8	COMPLEX
Spinocerebellar Ataxia (SCA) - Alpine Dachsbracke ^r	N/N	SCN8A	C-A
Spinocerebellar Ataxia (SCA) - Terrier ^r	N/N	KCNJ10	C-G

BREED NON-SPECIFIC VARIANTS (NO CORRELATION DETECTED IN YOUR BREED)

Unremarkable results	Genotype	Gene	Variant
Spongi Degeneration with Cerebellar Ataxia (SDCA1) ^r	N/N	KCNJ10	T-C
Startle Disease - Spanish Greyhound ^{r,2}	N/N	SLC6A5	DEL
STGD-PRA (Stargardt disease) ^r	N/N	ABCA4	INS
Succinic semialdehyde dehydrogenase deficiency (SSADHD) ^r	N/N	ALDH5A1	G-A
Thrombopathia - Basset Hound ^r	N/N	RASGRP2	DEL
Trapped Neutrophil Syndrome (TNS) ^r	N/N	VPS13B	DEL
Upper Airway Syndrome (UAS) ^r	N/N	ADAMTS3	C-T
van den Ende-Gupta syndrome (VDEGS) ^r	N/N	SCARF2	DEL
Ventricular arrhythmia (IVA) ^{3,4}	N/N	MICOS13	G-A
von Willebrand disease type II (vWD2) - German Wire-haired Pointing Dog ^r	N/N	VWF	T-G
von Willebrand disease type III (vWD3) - Kooikerhondje ^r	N/N	VWF	G-A
von Willebrand disease type III (vWD3) - Scottish Terrier ^r	N/N	VWF	DEL
von Willebrand disease type III (vWD3) - Shetland Sheepdog (Sheltie) ^r	N/N	VWF	DEL
X-chromosomal severe immunodeficiency - Basset Hound ^{Xr}	female X(N)/X(N) male X(N)/Y	IL2RG	DEL
X-chromosomal severe immunodeficiency - Welsh Corgi ^{Xr}	female X(N)/X(N) male X(N)/Y	IL2RG	DEL
X-linked Myopathy (XL-MTM) - Labrador Retriever ^{Xr}	female X(N)/X(N) male X(N)/Y	MTM1	C-A
X-linked Myopathy (XL-MTM) - Rottweiler ^{Xr}	female X(N)/X(N) male X(N)/Y	MTM1	A-C
Xanthinuria type II - Cavalier King Charles Spaniel, English Cocker Spaniel ^r	N/N	MOCOS	DEL
Xanthinuria type II - Dachshund (Dackel) ^r	N/N	MOCOS	A-G
Xanthinuria type II - English Toy Terrier, Manchester Terrier ^r	N/N	MOCOS	C-A

Not evaluable

L-2-Hydroxyglutaric Aciduria (L2HGA) - Yorkshire Terrier ^r
Paradoxical pseudomyotonia (PP) ^r
Primary open angle glaucoma (POAG) - Basset Hound ^r
Progressive Retinal Atrophy (crd2-PRA) - American Pitbull Terrier ^r

COAT COLORS & COAT CHARACTERISTICS

Genetic test	Genotype	Allelic series
A-locus (ASIP-Haplotype)	BB1/BB1	DY>SY>AG>BS/BB>a
B-locus variant b4 ^r	N/N (B/B)	N(B)>b4
B-locus variant bc ^r	N/N (B/B)	N(B)>bc
B-locus variant bd ^r	N/N (B/B)	N(B)>bd
B-locus variant be ^r	N/N (B/B)	N(B)>be
B-locus variant bh ^r	N/N (B/B)	N(B)>bh
B-locus variant bs ^r	N/bs (B/bs)	N(B)>bs
C-locus variant caL ^r	N/N (C/C)	N(C)>caL
C-locus variant OCA2 ^r	N/N (C/C)	N(C)>oca2

COAT COLORS & COAT CHARACTERISTICS

Genetic test	Genotype	Allelic series
C-locus variant OCA4 (Bullmastif) ^r	N/N (C/C)	N(C)>oca4
C-locus variant OCA4 (Dobermann) ^r	N/N (C/C)	N(C)>oca4
Coat length variant C587T ^r	L/L	L>l
Coat length variant del16 ^r	L/L	L>l
Coat length variant dupGG ^r	L/L	L>l
Coat length variant G284T ^r	l/l	L>l
Coat length variant T8193A ^r	L/L	L>l
Cocoa ^r	N/N	N>cocoa
Curly Coat variant C1 ^d	NC/NC	C1>NC
D-locus variant d1 ^r	N/N (D/D)	N(D)>d1
D-locus variant d2 ^r	N/N (D/D)	N(D)>d2
Double Coat variant 1 ^d	D/D	A>D
Double Coat variant 2 ^d	A/D	A>D
E-locus variant e1 ^r	N/N (E/E)	N(E)>e1
E-locus variant e3 ^r	N/N (E/E)	N(E)>e3
E-locus variant eA ^r	N/N (E/E)	N(E)>eA
E-locus variant eG ^r	N/N (E/E)	N(E)>eg
E-locus variant eH ^r	N/N (E/E)	N(E)>eh
E-locus variant EM ^d	N/N (E/E)	EM>N(E)
H-locus (Harlequin) ^d	h/h	H>h
Hairlessness (Deerhound) ^r	N/N	N>H
I-locus (pheomelanin intensity) ^r	N/i (I/i)	N(I)>i
K-locus	ky/ky	Kb>ky
Panda white spotting (German Shepherd) ^d	N/N	P>N
S-locus	N/N	
Saddle Tan - Basset Hound, Welsh Corgi ^d	St/bt	St>bt
T-locus (Ticking, Roan, Mottle, Spotted) ^d	N/N	TR>N

Not evaluable

Furnishing ^d

Improper Coat ^r

The current results are only valid for the sample submitted to our laboratory. The sender is responsible for the correct information regarding the sample material. The laboratory can not be made liable. Furthermore, any obligation for compensation is limited to the value of the tests performed.

There is a possibility that other mutations may have caused the disease/phenotype. The analysis was performed according to the latest knowledge and technology.

The laboratory is accredited for the performed tests according to DIN EN ISO/IEC 17025:2018. (except partner lab tests).

Annotation numbers

Detailed information on the annotation numbers can be found here:

<https://shop.labogen.com/annotations-info-dog>



Explanations on coat colour genetics

Help for interpreting the genetic variants can be found here:

<https://shop.labogen.com/coat-colour-genetics-dog>



These results are based on the sample material submitted to our laboratory.

This was suitable if not stated otherwise. The submitter is responsible for the accuracy of the information regarding the sample. This report can only be transmitted in toto and unchanged. Doing otherwise requires written permission from Laboklin GmbH & Co. KG.

LABOKLIN is an officially accredited laboratory according to DIN EN ISO/IEC 17025:2018, DAkkS No. D-PL-13186-01-01 D-PL-13186-1-02 and D-PL-13186-01-03. The accreditation applies to all test procedures listed in the accreditation certificate.

Fr. MSc Laura Hübner
Abt. Molekularbiologie

***** END of report *****



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