GENOMIA
genetic laboratory

Result certificate #256387

Screening for hereditary diseases and appearance traits of the dog

Customer: Klient, Street, 123 City, Czech Republic
Sample:
Sample: 123456
Date received: 20.02.2024
Sample type: buccal swab

Information provided by the customer
Name: Max
Breed: Labrador Retriever
Microchip: 123 456 789 123 456
Date of birth: 8.3.2021
Sex: male
Date of sampling: 19.02.2024
The identity of the animal has been checked by Mgr. Markéta Dajbychová, Genomia

Appearance

Name	Abbr.	Gene	Mutation	Copies	Result
Agouti locus, Recessive Black	a allele	ASIP	c.286C>T	0	no effect
Agouti locus, Fawn	A^y allele	ASIP	c.246G>T, c.250G>A	0	no effect
Locus K, Dominant Black	K^B allele	CBD103	c.67_69delGGA	2	may affect
Screw tail, Robinow-like syndrom		DVL2	c.2051del	0	no effect
Coat length	allele M3	FGF5	c.556_571del	0	no effect
Coat length	allele M4	FGF5	c.559_560dupGG	0	no effect
Coat length	allele M5	FGF5	c.578C>T	0	no effect
Coat length	allele M1	FGF5	c.284G>T	0	no effect
Locus cocoa		HPS3	c.2420G>A	0	no effect
Panda White Spotting (discovered in German Shepherds)		KIT	c.140_141insA	0	no effect
Curly Coat	c1 allele	KRT71	c.451C>T	0	clear
Locus EM, Melanistic mask	EM allele	MC1R	c.790A>G	0	no effect
Locus E, Recessive Red	e1 allele	MC1R	c.916C>T	0	no effect
Locus E, Recessive Red	e2 allele	MC1R	c.-432G>C	0	no effect
Locus E, Recessive Red	e3 allele	MC1R	c.816_817del	0	no effect
Locus E, grizzle and domino	LocEG	MC1R	c.233G>T	0	no effect
Locus E, sable (discovered in Cocker Spaniels)	allele EH	MC1R	c.250G>A	0	no effect
Locus I, phaeomelanin dilution	Locus I	MFSD12	c.151C>T	1	carrier
Locus D, eumelanine dilution	d1 allele	MLPH	c.-22G>A	0	no effect
Locus D, eumelanine dilution	d2 allele	MLPH	c.705G>C	0	no effect
Harlequin coat pattern	h allele	PSMB7	c.146T>G	0	no effect
Hairlessness (discovered in American Hairless Terriers)		SGK3	c.287_290delTTAG	0	no effect
Short Tail		T	c.189C>G	0	clear
Locus B, chocolade	b^d allele	TYRP1	c.1033_1035del	0	no effect

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Locus B, chocolade	b^s allele	TYRP1	c.991C>T	2	brown
Locus B, chocolade	b^c allele	TYRP1	c.121T>A	0	no effect
Locus B, chocolade (discovered in Australian Shepherds)	b^{aus} allele	TYRP1	c.555T>G	0	no effect

Autosomal dominant disorders

Name	Abbr.	Gene	Mutation	Copies	Result
Ichthyosis (discovered in German Shepherds)	ICTA	ASPRV1	c.1052T>C	0	clear
Craniomandibular Osteopathy (discovered in Terriers)	CMO	COL1A1	c.1332C>T	0	clear
Osteogenesis Imperfecta (discovered in Golden Retrievers)		COL1A1	c.1145G>C	0	clear
Osteogenesis Imperfecta (discovered in Beagle)	OI	COL1A2	c.3656_3859delinsTGTCATTGG	0	clear
Centronuclear Myopathy (discovered in Border Collies)	CNM	DNM2	c.1393C>T	0	clear
Renal Cystadenocarcinoma and Nodular Dermatofibrosis (discovered in German Shepherds)	RCND	FLCN	c.764A>G	0	clear
Alexander disease (discovered in Labrador Retrievers)		GFAP	c.719G>A	0	clear
Polyneuropathy (discovered in Leonbergers)	LPN2	GJA9	c.1107_1108delAG	0	clear
Muscular Hypertrophy, Double Muscling (discovered in Whippets)		MSTN	c.939_940delTG	0	clear
Periodic Fever Syndrome (discovered in Shar-Pei)	SPAID	MTBP	c.2623G>A	0	clear
May-Hegglin anomaly (discovered in Pugs)	MHA	MYH9	c.5521G>A	0	clear
Polycystic kidney disease (discovered in English Bull Terriers)	BTPKD	PKD1	c.9559G>A	0	clear
Dominant Progressive Retinal Atrophy (Discovered in English Mastiffs and Bullmastiffs)	AD-PRA	RHO	c.11C>G	0	clear
Malignant Hyperthermia (discovered in many breeds)	MH	RYR1	c.1643T>C	0	clear
Cystinuria Type II-A (discovered in Australian Cattle Dogs and Koolies)		SLC3A1	c.1098_1103del	0	clear
Cystinuria Type II-B (discovered in Miniature Pinschers)		SLC7A9	c.964G>A	0	clear

Autosomal recessive disorders

Name	Abbr.	Gene	Mutation	Copies	Result
Stargardt disease 1 (discovered in Labrador Retrievers)	STGD	ABCA4	c.4176dup	1	carrier
Ichthyosis 2 (discovered in Golden Retrievers)	ICTA2	ABHD5	c.1006_1019del	0	clear
Amelogenesis imperfecta (discovered in Akitas)	AI	ACP4	c.1189dupG	0	clear
Primary open angle glaucoma (discovered in Norwegian Elkhounds)	POAG	ADAMTS10	c.1159G>A	0	clear
Primary open angle glaucoma (discovered in Beagles)	POAG	ADAMTS10	c.1981G>A	0	clear
Primary Lens Luxation (discovered in many breeds)	PLL	ADAMTS17	c.1473+1G>A	0	clear
Primary open angle glaucoma and the primary lens luxation (discovered in Shar Peis)	POAG-PLL	ADAMTS17	c.3069_3074del	0	clear
Cleft lip with or without cleft palate (dicovered in Nova Scotia Duck Tolling retrievers)	CLPS (CL/P)	ADAMTS20	c.1358_1359del	0	clear
Musladin-Lueke Syndrome (discovered in Beagles)	MLS	ADAMTSL2	c.661C>T	0	clear
Glycogen Storage Disease Type IIIa (discovered in Curly Coated Retrievers)	GSD IIIa	AGL	c.4223del	0	clear

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Inflammatory pulmonary disease (discovered in Border Collies)	IPD	AKNA	c.2717_2720delACAG	0	clear
Succinic semialdehyde dehydrogenase deficiency (discovered in Salukis)	SSADHD	ALDH5A1	c.866G>A	0	clear
Hypophosphatasia (discovered in Karelian bear dogs)		ALPL	c.1301T>G	0	clear
Persistent Mullerian duct syndrome (discovered in Schnauzer Miniature)	PMDS	AMHR2	c.262C>T	0	clear
Respiratory distress syndrome (discovered in Dalmatians)	ARDS	ANLN	c.31C>T	0	clear
Scott Syndrome (discovered in German Shepherds)	CSS	ANO6	c.1934+1G>A	0	clear
Polyneuropathy (discovered in Leonbergers and Saint Bernards)	LPN1	ARHGEF10	c.1955_1958+6del	0	clear
Mucopolysaccharidosis VI (discovered in Miniature Pinschers)	MPS VI	ARSB	c.910G>A	0	clear
Neuronal ceroid lipofuscinosis 4A (discovered in American Staffordshire Terriers and American Pitbul Terriers)	NCL-A	ARSG	c.296G>A	0	clear
Neonatal encephalopathy with seizures (discovered in Standard Poodles)	NEWS	ATF2	c.152T>G	0	clear
Lysosomal Storage Diseases (discovered in Lagotto romagnolo)	LSDs	ATG4D	c.1288G>A	0	clear
Neuronal ceroid lipofuscinosis 12 (discovered in Tibetan Terriers)	NCL 12	ATP13A2	c.1623del	0	clear
Neuronal ceroid lipofuscinosis 12 (discovered in Australian Cattle Dogs)	NCL12	ATP13A2	c.1118C>T	0	clear
Bardet-Biedl syndrome 2 (discovered in Shetland Sheepdogs)	BBS2	BBS2	c.1222G>C	0	clear
Bardet-Biedl syndrome 4 (discovered in Puli)	BBS4	BBS4	c.58A>T	0	clear
Canine Multifocal Retinopathy 1 (discovered in Boerboel, Bull Mastiff, English Mastiff, Great Pyrenees)	CMR1	BEST1	c.73C>T	0	clear
Canine Multifocal Retinopathy 2 (discovered in Coton de Tulear)	CMR2	BEST1	c.482G>A	0	clear
Multifocal retinopathy 3 (discovered in Finnish Lapphund, Lapponian Herder)	CMR3	BEST1	c.1388del	0	clear
Inherited Myopathy (discovered in Great Danes)	IMGD	BIN1	c.786-2A>G	0	clear
Spinocerebellar ataxia, Late Onset Ataxia (discovered in Russell Terriers)	LOA	CAPN1	c.344G>A	0	clear
Mycobacterium Avium Complex (discovered in Miniature Schnauzers)	MAC	CARD9	deletion	0	clear
Hypocatalasia (discovered in Beagles and other breeds)		CAT	c.979G>A	0	clear
Early onset Progressive retinal atrophy (discovered in Portuguese water dogs)	EOPRA	CCDC66	c.2262_c.2263insA	0	clear
Deafness (discovered in Beaucerons)		CDH23	c.700C>T	0	clear
Congenital myasthenic syndrome (discovered in Old Danish Pointing Dogs)	CMS	CHAT	c.85G>A	0	clear
Myasthenic syndrome (discovered in Heideterriers)	CMS	CHRNE	c.1436_1437insG	0	clear
Myasthenic syndrome (discovered in Russell Terriers)	CMS	CHRNE	c.636_637insC	0	clear
Myotonia Congenita (Discovered in the Miniature Schnauzer)	MC	CLCN1	c.803C>T	0	clear
Congenital Myotonia (discovered in Australian Cattle Dogs and Border Collies)	MC	CLCN1	c.2647_2648insA	0	clear
Neuronal ceroid lipofuscinosis type 5 (discovered in Border Collies)	NCL5	CLN5	c.619C>T	0	clear

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Neuronal ceroid lipofuscinosis type 6 (discovered in Australian Shepherds)	NCL6	CLN6	c.829T>C	0	clear
Neuronal ceroid lipofuscinosis 8 (discovered in English Setter)	NCL8	CLN8	c.491T>C	0	clear
Progressive retinal atrophy (discovered in Shetland Sheepdogs)	PRA	CNGA1	c.1752_1755del	0	clear
Achromatopsia (discovered in German Shepherds)		CNGA3	c.1270C>T	0	clear
Achromatopsia (discovered in Labrador Retrievers)		CNGA3	c.1931_1933del	0	clear
Progressive retinal atrophy (discovered in Papillons)	Pap-PRA1	CNGB1	c.2387_2389delinsC TAGCTAC	0	clear
Achromatopsia-3 (discovered in German Shorthaired Pointers)		CNGB3	c.784G>A	0	clear
Laryngeal paralysis and polyneuropathy (discovered in Labrador Retrievers, Leonbergers, St. Bernards)	LPPN3	CNTNAP1	c.2810G>A	0	clear
Familial Nephropathy (discovered in the English Cocker Spaniel)	FN	COL4A4	c.115A>T	0	clear
Hereditary Nephropathy (discovered in English Springer Spaniels)	ARHN	COL4A4	c.2713C>T	0	clear
Muscular dystrophy (discovered in Landseers)	MDL	COL6A1	c.289G>T	0	clear
Epidermolysis bullosa (discovered in Golden Retrievers)		COL7A1	c.5716G>A	0	clear
Recessive dystrophic epidermolysis bullosa (discovered in Central Asian Shepherds)	RDEB	COL7A1	c.4579C>T	0	clear
Neuronal ceroid lipofuscinosis 10 (discovered in American Bulldogs)	NCL10	CTSD	c.597G>A	0	clear
Imerslund-Grasbeck Syndrome, Intestinal Malabsorption of Cobalamin (discovered in Beagles)	IGS	CUBN	c.786del	0	clear
Imerslund-Grasbeck Syndrome, Intestinal Malabsorption of Cobalamin (discovered in Border Collies)	IGS	CUBN	c.8392del	0	clear
Imerslund-Grasbeck Syndrome, Intestinal Malabsorption of Cobalamin (discovered in Komondor)	IGS	CUBN	c.8746+1G>A	0	clear
Myoclonic Epilepsy (discovered in Rhodesian ridgebacks)	JME	DIRAS1	c.564_567del	0	clear
Exercise-Induced Collapse (discovered in Labrador Retrievers)	EIC	DNM1	c.767G>T	0	clear
Amelogenesis imperfecta (discovered in Italian Greyhound)	AI	ENAM	c.1991_1995delTTT CC	0	clear
Factor VII deficiency (discovered in many breeds)	FVII def	F7	c.407G>A	0	clear
Dental Hypomineralization (discovered in Border Collies)	RS	FAM20C	c.899C>T	0	clear
Palmoplantar hyperkeratosis (discovered in Irish Terriers and Kromfohrländers)	HFH	FAM83G	c.155G>C	0	clear
Leukocyte Adhesion Deficiency III (discovered in German Shepherds)	LAD3	FERMT3	c.1349_1350insAAG ACGGCTGCC	0	clear
Hypomyelination of the central nervous system (discovered in Weimaraners)		FNIP2	c.1078del	0	clear
Glycogen storage disease Ia (discovered in Maltese)	GSDIa	G6PC	c.363G>C	0	clear
Glycogen storage disease II, Pompe disease (discovered in Lapphunds)	GSDII	GAA	c.2237G>A	0	clear
Globoid cell leukodystrophy, Krabbe disease (discovered in West Highland White Terriers and Cairn Terriers)	GLD	GALC	c.473A>C	0	clear
Acral mutilation syndrome (discovered in English Springer Spaniels and other breeds)	AMS	GDNF	g.70875561C>T	0	clear
Gangliosidosis 1 (discovered in Alaskan Husky)	GM1	GLB1	c.1688_1706dup	0	clear
Gangliosidosis 1 (discovered in Portuguese water dogs)	GM1	GLB1	c.179G>A	0	clear

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Gangliosidosis type 1 (discovered in Shiba-Inu)	GM1	GLB1	c.1649delC	0	clear
Progressive retinal atrophy (discovered in German Spitz)	PRA	GUCY2D	c.1598_1599insT	0	clear
Mucopolysaccharidosis VII (discovered in German Shepherds)	MPS VII	GUSB	c.497G>A	0	clear
Mucopolysaccharidosis VII (discovered in Brazilian Terriers)	MPS VII	GUSB	c.866C>T	0	clear
Ataxia (discovered in Norwegian Elkhound)		HACE1	c.1001del	0	clear
Narcolepsy (discovered in Dachshunds)		HCRTR2	c.160G>A	0	clear
Narcolepsy (discovered in Labrador Retrievers)		HCRTR2	c.1105+5G>A	0	clear
Spondylocostal Dysostosis, Comma Defect (discovered in Schnauzer Miniature)	SCD	HES7	c.126delG	0	clear
Gangliosidosis type 1 (discovered in Japanese Chin dogs)	GM2	HEXA	c.967G>A	0	clear
Gangliosidosis II, Sandhoff disease (discovered in Toy Poodles)	GM2	HEXB	c.391del	0	clear
Gangliosidosis 2 (discovered in Shiba-Inu)	GM2	HEXB	c.618_620delCCT	0	clear
Progressive retinal atrophy 1 (discovered in Miniature Schnauzers)	PRA1	HIVEP3	g.1432293G>A	0	clear
Progressive retinal atrophy (discovered in Lapponian Herder)	PRA	IFT122	c.3176G>A	0	clear
Diffuse cystic renal dysplasia and hepatic fibrosis (discovered in Norwich Terrier)	HRFCD	INPP5E	c.1572+5G>A	0	clear
Chondrodysplazia (discovered in Karelian Bear Dogs and Norwegian Elkhounds Grey)		ITGA10	c.2083C>T	0	clear
Glanzmann thrombastenia (discovered in Scottish Deerhound)		ITGA2B	c.1192G>C	0	clear
Glanzmann thrombastenia (discovered in Great Pyrenees)		ITGA2B	c.1360_1373dup	0	clear
Canine leukocyte adhesion deficiency	CLAD	ITGB2	c.107G>C	0	clear
Cerebellar ataxia (discovered in Norwegian Buhunds)		KCNIP4	c.436T>C	0	clear
Spongy Degeneration with Cerebellar Ataxia subtype 1 (discovered in Belgian shepherds)	SDCA1	KCNJ10	c.986T>C	0	clear
Spinocerebellar Ataxia with Myokymia and/or Seizures (discovered in Russell Terriers and Smooth-Haired Fox Terriers)	SCA	KCNJ10	c.627C>G	0	clear
Prekallikrein deficiency (discovered in Shih-Tzu)		KLKB1	c.988T>A	0	clear
Hyperkeratosis (discovered in Norfolk Terriers)		KRT10	c.1125+1G>T	0	clear
L-2-Hydroxyglutaric aciduria (discovered in the Staffordshire Bull Terrier)	L2HGA	L2HGDH	c.1298_1300delinsCTT	0	clear
Benign familial juvenile epilepsy (discovered in u Lagotto Romagnolo))	JEP	LGI2	c.1558A>T	0	clear
Congenital myasthenic syndrome (discovered in Golden Retrievers)	CMS	LOC608697	c.880G>A	0	clear
Congenital myasthenic syndrome (discovered in Labrador Retrievers)	CMS	LOC608697	c.1010T>C	0	clear
Deafness (discovered in Rottweilers)		LOXHD1	c.5747G>C	0	clear
Congenital Stationary Night Blindness (discovered in Beagles)	CSNB	LRIT3	c.763del	0	clear
MDR1 Medication Sensitivity (discovered in many breeds)	MDR1	MDR1 (ABCB1)	c.228_231del	0	clear
Mitochondrial fission encephalopathy, (discovered in Bull Mastiffs)	MFE	MFF	c.471_475delinsCGCTCT	0	clear
Neuronal ceroid lipofuscinosis type 7 (discovered in Chihuahuas and Chinese Crested Dogs)	NCL7	MFSD8	c.846del	0	clear

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Dental-skeletal-retinal anomaly (discovered in Cane Corso)	DSRA	MIA3	c.3822+3_3822+4del	0	clear
Ventricular arrhythmias and sudden death (discovered in Rhodesian Ridgebacks)		MICOS13	c.325G>A	0	clear
Lethal Acrodermatitis (discovered in Bull Terriers)	LAD	MKLN1	c.400+3A>C	0	clear
Xanthinuria type II (discovered in Cavalier King Charles Spaniels and English Cocker Spaniels)	XU	MOCOS	c.383del	0	clear
Xanthinuria type II (discovered in Dachshunds)	XU	MOCOS	c.137T>C	0	clear
Xanthinuria type II (discovered in Manchester Terriers)	XU	MOCOS	c.232G>T	0	clear
Deafness (discovered in Doberman Pinschers)		MYO7A	c.3719G>A	0	clear
Leukoencephalomyelopathy, (discovered in Great Danes and Rottweilers)	LEMP	NAPEPLD	c.345_346insC	0	clear
Early onset progressive polyneuropathy (discovered in Greyhound)		NDRG1	c.1080_1089del10	0	clear
Early onset progressive polyneuropathy (discovered in Alaskan Malamute)	AMPn	NDRG1	c.293G>T	0	clear
Progressive retinal atrophy (discovered in Giant Schnauzers)	PRA	NECAP1	c.544G>A	0	clear
Congenital ichthyosis (discovered in American Bulldogs)		NIPAL4	c.744delC	0	clear
Primary ciliary dyskinesia (discovered in Alaskan Malamutes)	PCD	NME5	c.43delA	0	clear
Goniodysgenesis and glaucoma (discovered in Border Collies)		OLFML3	c.590G>A	0	clear
Bleeding disorder (discovered in Greater Swiss Mountain Dogs)		P2RY12	c.516_518del	0	clear
Lundehund syndrom (discovered in Lundehunds)	LS	P3H2	c.1849G>C	0	clear
Skeletal dysplasia 3, Dwarfism (discovered in Vizslas)	SD3	PCYT1A	c.673T>C	0	clear
Rod-cone dysplasia 3 (discovered in Cardigan Welsh Corgi, Chinese Crested, Pomeranian)	PRA-rcd3	PDE6A	c.1847del	0	clear
Cone-rod dystrophy, (discovered in American Staffordshire Terriers and American Pit Bull Terriers)	CRD1	PDE6B	c.2404_2406del	0	clear
Rod-cone dysplasia 1a (discovered in Sloughi)	PRA-rcd1	PDE6B	c.2448_2449insTGAAGTCC	0	clear
Rod-cone dysplasia 1 (discovered in Irish Setter)	PRA-rcd1	PDE6B	c.2421G>A	0	clear
Pyruvate dehydrogenase phosphatase 1 deficiency (discovered in Clumber and Sussex Spaniels)	PDP1	PDP1	c.829C>T	0	clear
Phosphofructokinase deficiency (discovered in American Cocker Spaniel, English Springer Spaniel, Whippet)	PFK	PFKM	c.2228G>A	0	clear
Glycogen storage disease VII (discovered in German Spaniels)	GSD VII	PFKM	c.550C>T	0	clear
Paroxysmal dyskinesia (discovered in Soft Coated Wheaten Terriers)		PIGN	c.398C>T	0	clear
Juvenile Brain Disease, Juvenile Encephalopathy (discovered in Russell Terriers)	JBD	PITRM1	c.175_180del	0	clear
Pyruvate kinase deficiency of erythrocyte (discovered in Labrador Retrievers)	PK deficit	PKLR	c.799C>T	0	clear
Pyruvate kinase deficiency of erythrocyte (discovered in Pugs)	PK deficit	PKLR	c.848T>C	0	clear
Pyruvate kinase deficiency (discovered in Beagles)	PK deficit	PKLR	c.994G>A	0	clear
Pyruvate kinase deficiency of erythrocyte (discovered in Basenji)	PK deficit	PKLR	c.433del	0	clear
Pyruvate kinase deficiency of erythrocyte (discovered in West Highland White Terriers)	PK deficit	PKLR	c.1333_1338dup	0	clear

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Neuroaxonal Dystrophy (discovered in Papillons)	NAD	PLA2G6	c.1579G>A	0	clear
Ichthyosis 1 (discovered in Golden Retriever)	ICTA1	PNPLA1	c.1445_1447delinsTACTACTA	0	clear
Neuronal ceroid lipofuscinosis 1 (discovered in Dachshunds)	NCL1	PPT1	c.736_737insC	0	clear
Progressive Rod Cone Degeneration (discovered in many breeds)	PRA-prcd	PRCD	c.5G>A	0	clear
Severe combined immunodeficiency disease (discovered in Russell Terriers)	SCID	PRKDC	c.10849G>T	0	clear
Deafness (discovered in Doberman Pinschers)		PTPRQ	c.9230_9231insA	0	clear
Ataxia, cerebellar, juvenile to adolescent (discovered in Gordon Setters and Old English Sheepdogs)	HA	RAB24	c.113A>C	0	clear
Juvenile Laryngeal Paralysis and Polyneuropathy (discovered in Rottweilers and Black Russian Terriers)	JLPP	RAB3GAP1	c.743del	0	clear
Severe combined immunodeficiency disease (discovered in Frisian Water Dogs)	T-B-NK+ SCID	RAG1	c.2893G>T	0	clear
Thrombopathia (discovered in Basset Hounds)		RASGRP1	c.509_511del	0	clear
Thrombopathie (discovered in American Eskimo Spitz)		RASGRP1	c.452dup	0	clear
Thrombopathy (discovered in Landseers)		RASGRP2	c.982C>T	0	clear
Dilated cardiomyopathy (discovered in Schnauzers)	DCM	RBM20	22 bp deletion	0	clear
Congenital Stationary Night Blindness (discovered in Briards)	CSNB	RPE65	c.460_463del	0	clear
Neuronal degeneration (discovered in Great Pyrenees dogs)	SACS	SACS	c.12731_12734del	0	clear
Progressive retinal atrophy (discovered in Basenji)	Bas-PRA	SAG	c.1216T>C	0	clear
Charcot-Marie-Tooth disease (discovered in Miniature Schnauzers)	CMT	SBF2	c.2363+1G>T	0	clear
Van den Ende-Gupta syndrome (discovered in Fox Terriers Wire)	VDEGS	SCARF2	c.1873_1874del	0	clear
Spinocerebellar ataxia (discovered in Alpine Dachsbracke)		SCN8A	c.4898G>T	0	clear
Progressive Early-Onset Cerebellar ataxia (discovered in Finnish Hound)		SEL1L	c.1972T>C	0	clear
Canine Multiple Systems Degeneration (discovered in the Chinese Crested Dog)	CMSD	SERAC1	c.128+1_128+4delGTAA	0	clear
Canine Multiple System Degeneration (discovered in Kerry Blue Terriers)	CMSD	SERAC1	c.1482G>A	0	clear
Osteogenesis Imperfecta (discovered in Dachshund)	OI	SERPINH1	c.977T>C	0	clear
Mucopolysaccharidosis IIIA (discovered in Dachshunds)	MPS IIIA	SGSH	c.740_742delCCA	0	clear
Cerebellar degeneration-myositis complex (discovered in Nova Scotia Duck Tolling retrievers)	CDMC	SLC25A12	c.1337C>T	0	clear
Cerebellar degeneration-myositis complex (discovered in Dutch Shepherds)	CDMC	SLC25A12	c.1046T>C	0	clear
Ichthyosis (discovered in Great Danes)		SLC27A4	c.1250G>A	0	clear
Hyperuricosuria (discovered in many breeds)	HUU	SLC2A9	c.563G>T	0	clear
Cystinuria Type I-A		SLC3A1	c.586C>T	0	clear
Cystinuria I-A (discovered in Labrador Retrievers)		SLC3A1	c.350del	0	clear
Hyperekplexia, Startle disease (discovered in Galgo)		SLC6A5	c.1379_1380delCT	0	clear
Neonatal cerebellar cortical degeneration (discoverd in Hungarian Viszla)	NCCD	SNX14	c.26531G>A	0	clear
Degenerative Myelopathy (discovered in many breeds)	DM	SOD1	c.118G>A	0	clear

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Neonatal cerebellar cortical degeneration (discovered in Beagles)	NCCD	SPTBN2	c.5855_5862del	0	clear
Primary ciliary dyskinesia (discovered in Australian Shepherds)	PCD	STK36	c.2868-1G>A	0	clear
Nasal parakeratosis (discovered in Labrador Retrievers)	HNPK	SUV39H2	c.972T>G	0	clear
Neuroaxonal dystrophy (discovered in Spanish water dogs and Lagotto Romagnolo)	NAD	TECPR2	c.4009C>T	0	clear
Dystonia-ataxia syndrome (discovered in Weimaraners)		TNR	c.831dup	0	clear
Congenital hypothyroidism (discovered in French Bulldogs)	CHG	TPO	c.2242+2T>C	0	clear
Congenital hypothyroidism (discovered in Rat Terriers and Toy Fox Terriers)	CHG	TPO	c.331C>T	0	clear
Congenital hypothyroidism (discovered in Spanish Water Dogs)	CHG	TPO	c.39_40insG	0	clear
Progressive retinal atrophy 2 (discovered in Golden Retrievers)	GR-PRA2	TTC8	c.669delA	0	clear
Macrothrombocytopenia (discovered in many breeds)	MCT	TUBB1	c.745G>A	0	clear
Macrothrombocytopenia (discovered in Cairn Terriers and Norfolk Terriers)	MCT	TUBB1	c.5G>A	0	clear
Exfoliative cutaneous lupus erythematosus (discovered in German Shorthaired Pointers and Vizslas)	ECLE	UNC93B1	c.1438C>A	0	clear
Vitamin D-deficiency rickets type II (discovered in Pomeranians)		VDR	c.462del	0	clear
Cerebellar hypoplasia, Dandy-Walker-like malformation (discovered in Eurasians)	DWLM	VLDLR	c.1713del	0	clear
Neuroaxonal Dystrophy (discovered in Rottweilers)	NAD	VPS11	c.2504A>G	0	clear
Trapped Neutrophil Syndrome (discovered in Border Collies)	TNS	VPS13B	c.2893_2896del	0	clear
Von Willebrand disease II (discovered in Chinese Crested Dog and German Pointers)	VWD typ II	VWF	c.1657T>G	0	clear
Von Willebrand disease III (discovered in Shetland Sehhpdog)	vWD typ III	VWF	c.738del	0	clear
Von Willebrand disease I (discovered in many breeds)	VWD typ I	VWF	c.7437G>A	1	carrier
Von Willebrand disease II (discovered in many breeds)	VWD typ II	VWF	c.4937A>G	0	clear
Von Willebrand disease III (discovered in Scottish Terriers)	VWD typ III	VWF	c.255del	0	clear
Von Willebrand disease III (discovered in in Kooikerhondje)	VWD typ III	VWF	c.2186+1G>A	0	clear
Cardiomyopathy and juvenile mortality (discovered in Belgian Shepherds)	CJM	YARS2	c.1054G>A	0	clear

Association genetic tests

Name	Abbr.	Gene	Mutation	Copies	Result
Gallbladder mucoceles (discovered in Shetland Sheepdogs)	GBM	ABCB4	c.1660_1661insG	0	clear
Upper airway syndrome (discovered in Norwich Terriers)	UAS	ADAMTS3	c.2786G>A	0	clear
Brachycephaly		BMP3	c.1344C>A	0	clear
Dilated Cardiomyopathy (discovered in Welsh Springer Spaniels)	DCM	PLN	c.26G>A	0	clear
Obesity (discovered in Labrador and Flat-Coated Retrievers)		POMC	c.561_575del	0	clear
Cystinuria (discovered in Bulldogs)		SLC3A1	c.2092A>G	0	clear
Cystinuria (discovered in Bulldogs)		SLC3A1	c.574A>G	0	clear

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Cystinuria (discovered in Bulldogs)		SLC7A9	c.649G>A	0	clear

X-linked hereditary disorders

Name	Abbr.	Gene	Mutation	Copies	Result
X-linked hereditary nephropathy (discovered in Samoyed)	HN	COL4A5	c.3079G>T	0	clear
Muscular Dystrophy (discovered in Golden retrievers)	GRMD	DMD	c.531-2A>G	0	clear
Duchenne muscular dystrophy (discovered in Kavalier King Charles Spaniels)	DMD	DMD	c.7294+5G>T	0	clear
Duchenne muscular dystrophy (discovered in Kavalier King Charles Spaniels)	DMD	DMD	c.6057_6063del	0	clear
Muscular dystrophy, Duchenne type (discovered in Border Collies)	DMD	DMD	c.2841delT	0	clear
Muscular dystrophy, Duchenne type (discovered in Labradoodles)	DMD	DMD	c.2668C>T	0	clear
Muscular dystrophy, Duchenne type (discovered in Norfolk Terriers)	DMD	DMD	c.3084delG	0	clear
Anhidrotic Ectodermal Dysplasia (discovered in German Shepherds)		EDA	c.910-1G>A	0	clear
Haemophilia B (discovered in Rhodesian Ridgebacks)		F9	c.731G>A	0	clear
Severe combined immunodeficiency disease (discovered in Welsh Corgi)	XSCID	IL2RG	c.583 _584insC	0	clear
Myotubular myopathy 1 (discovered in Labrador Retrievers)	MTM1	MTM1	c.465C>A	0	clear
Myotubular myopathy 1 (discovered in Rottweilers)	MTM1	MTM1	c.1151A>C	0	clear
Shaking puppy syndrom, Tremor (discovered in English Springer Spaniels)	SPS	PLP1	c.110A>C	0	clear
X-Linked Progressive Retinal Atrophy 1 (discovered in Siberian Husky and Samoyed)	XL-PRA	RPGR	c.3416_3420del	0	clear

Explanation

Interpretation of the results can be found on the website https://www.genomia.cz/en/veterinarni/psi/ on the pages of the respective examinations.

The mutations listed are annotated according to the CanFam3.1 reference sequence.

Recessive inheritance: the trait (disease) becomes apparent if the individual inherits it from both parents (2 copies); carriers of the trait (disease) are asymptomatic but pass the causal mutation on to the next generation (1 copy).

Dominant inheritance: it is sufficient for an individual to inherit the trait (disease) from one parent (1 copy).

X-linked recessive inheritance: in males, 1 copy of the mutated gene is enough to cause the disease; in females, 2 copies of the mutated gene are needed to cause the disease.

The results of the association genetic tests indicate the predisposition to the disease. This is not a detection of a causal mutation of the disease.

Method: SOPAgriseq_canine, ngs

Date of issue: 08.03.2024
Date of testing: 20.02.2024 - 08.03.2024
Approved by: Mgr. Markéta Dajbychová, Deputy Laboratory Manager

verified and certified document GENOMIA genetic laboratory

Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
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