

Category	Gene(s)	Diseases, coat colours & characteristics	Abbreviation	DogCheck V4.0
Diversity		Calculation of the genetic inbreeding coefficient (IK)		x
Diversity		Calculation of genetic diversity (heterozygosity)		x
Platform		Access to the digital breeder platform myFERAGEN		x
Platform		DogMatching - Finding genetically matching breeding partners		x
Coat colours	ASIP	A-Lokus Agouti (Black and Tan)	at-Loc	x
Coat colours	ASIP	A-locus Agouti (recessive black)	a-Loc	x
Coat colours	ASIP	A-Lokus Agouti (Sable)	ay-Loc	x
Coat colours	ASIP	A-Lokus Agouti (Sable)	ay-Loc	x
Disease	GDNF	Acral mutilation syndrome	AMS	x
Coat colours	SLC45A2	Albinism, oculocutaneous (small breeds)	OCA4	x
Disease	GFAP	Alexander Krankheit (Labrador Retriever)	AxD	x
Disease	ENAM	Amelogenesis Imperfecta (Italian wind chime)	AI	x
Disease	ENAM	Amelogenesis Imperfecta (Parson Russell Terrier)	AI	x
Disease	SIX6	Congenital eye malformation (Golden Retriever)	CEM	x
Disease	SELENOP, SEPP1	Ataxia, CNS atrophy with cerebellar ataxia (Belgian Malinois)	CaCa	x
Disease	MTBP	Autoinflammatory disease (Shar-Pei)	SPAID	x
Coat colours	TYRP1	B-Lokus ba (Brown - Australian Shepherd)	ba-Loc	x
Coat colours	TYRP1	B-locus bc (brown)	bc-Loc	x
Coat colours	TYRP1	B-Lokus bd (brown)	bd-Loc	x
Coat colours	TYRP1	B-Lokus bs (brown)	bs-Loc	x
Disease	LGI2	Benign Familial Juvenile Epilepsy (Lagotto Romagnolo)	BFJE	x
Disease	GP9	Bernard-Soulier syndrome (Cocker Spaniel)	BSS	x
Features	BMP3	Brachycephaly, snout length, skull shape		x
Disease	TMEM16F/ ANO6	Canine Scott Syndrome (German Shepherd Dog)	CSS	x
Disease	RFLN	Cerebellar hypoplasia	CH	x
Disease	FGF4	Chondrodysplasia	CDPA	x
Disease	ITGA10	Chondrodysplasia (Karelian Bear Dog)	CDPA	x
Disease	FGF4	Chondrodystrophy and predisposition to herniated discs	CDDY-IVDD	x
Coat colours	HPS3	Co-Locus Cocoa Brown (French Bulldog)	Co-Loc	x
Disease	NHEJ1	Collie eye anomaly	CEA	*xP
Disease	CNGB3	Cone degeneration	CD	x
Disease	CNGA3	Cone Degeneration (German Shepherd Dog)	CD	x
Disease	CNGA3	Cone Degeneration (Labrador Retriever)	CD	x
Disease	SLC37A2	Craniomandibular osteopathy	CMO	x
Coat characteristics	KRT71	Cu1 locus, curls	Cu1-Loc	x
Disease	SLC3A1	Cystinuria (Australian Cattle Dog)	Cyst-2a	x
Disease	SLC3A1	Cystinuria (Newfoundland)	Cyst-1a	x
Disease	SLC7A9	Cystinuria (Miniature Pinscher)	Cyst-2	x
Disease	SLC3A1	Cystinuria risk factor type 3 variant 1 (bulldog)	Cyst3-1	x
Disease	SLC3A1	Cystinuria risk factor type 3 variant 2 (bulldog)	Cyst3-2	x
Coat colours	MLPH	D-Lokus d1 colour thinner (standard variant)	d1-Loc	x
Coat colours	MLPH	D-Lokus d2 Colour thinner	d2-Loc	x

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Disease	VLDLR	Dandy-Walker-Like Syndrome (Eurasier)	DWLM	x
Disease	SOD1	Degenerative myelopathy (Bernese Mountain Dog - exon 1)	DM - SOD1B	x
Disease	SOD1	Degenerative myelopathy (classic variant - exon 2)	DM - SOD1A	*xP
Disease	SP110	Degenerative Myelopathy, Early-Onset Modifier (Pembroke Welsh Corgi)	DM modifier	x
Disease	FAM20c	Dental hypomineralisation (Border Collie)		x
Disease	INPP5E	Diffuse cystic renal dysplasia and hepatic fibrosis (Norwich Terrier)		x
Disease	FAM83H	Dry Eye Curly Coat Syndrome (Cavalier King Charles Spaniel)	CKCSID	x
Coat colours	MC1R	E locus e1 Recessive red	e-Loc	x
Coat colours	MC1R	E-Lokus e3 Recessive red (Husky)	e3-Loc	x
Coat colours	MC1R	E-Lokus Ea Ancient Domino	Ea-Loc	x
Coat colours	MC1R	E-Lokus Eg Grizzle Domino	Eg-Loc	x
Coat colours	MC1R	E-Lokus Eh Cocker Sable	Eh-Loc	x
Coat colours	MC1R	E-Lokus Em melanistic mask	Em-Loc	x
Disease	TNXB	Ehlers-Danlos syndrome variant 1 (poodle)	EDS	x
Disease	TNXB	Ehlers-Danlos syndrome variant 2	EDS	x
Disease	PKP1	Ectodermal dysplasia (Chesapeake Bay Retriever)	ED-SFS	x
Disease	DFA	Ectodermal dysplasia (dachshund)	XHED	x
Disease	DFA	Ectodermal dysplasia, X-linked (German shepherd type)	XED	x
Disease	SPTB	Elliptocytosis (Labrador Retriever)	HE	x
Disease	LAMB3	Epidermolysis bullosa, junctionalis (Australian Shepherd)	JEB	x
Disease	KRT10	Epidermolytic hyperkeratosis (Norfolk Terrier)		x
Disease	DNM1	Exercise Induced Collapse	EIC	*xP
Disease	F7	Factor VII deficiency	F7	x
Disease	COL4A4	Familial nephropathy (Cocker Spaniel)	FN	x
Disease	COL4A4	Familial nephropathy (Springer Spaniel)	FN	x
Disease	FAN1	Fanconi syndrome (Basenji)		x
Disease	ABCB4	Gallbladder mucocele		x
Disease	GLB1	Gangliosidosis GM1 (Shiba Inu)	GM1	x
Disease	HEXB	Gangliosidosis GM2 (poodle)	GM2	x
Disease	HEXA	Gangliosidosis GM2 type 1b (Japan Chin)	GM2-1b	x
Disease	ADAMTS20	Cleft palate and syndactyly (Nova Scotia Duck Tolling Retriever)	CLPS	x
Features	AMEL	Sex marker amelogenin	Sex	x
Disease	ITGA2B	Glanzmann Thrombastenie (Otterhound)	GT	x
Disease	OLFML3	Glaucoma and goniodysgenesis (Border Collie)	GG/PCAG	x
Disease	GALC	Globoid cell leukodystrophy (terrier)	GLD	x
Disease	G6PC	Glycogen storage disease Ia (Maltese)	GSD Ia	x
Disease	AGL	Glycogen storage disease II (Lapponian Herder)	GSD II	x
Disease	AGL	Glycogen storage disease IIIa (Curly Coated Retriever)	GSD IIIa	x
Disease	PFKM	Glycogen storage disease VII (English Springer Spaniel)	GSD VII	x
Disease	PFKM	Glycogen storage disease VII (quail dog)	GSD VII	x
Coat colours	PSMB7	H-Lokus Harlequin (Great Dane)	H-Loc	x
Disease	RAB24	Hereditary ataxia (Gordon Setter, Old English Sheepdog)	HA	x
Disease	FAM83G	Hereditary footpad hyperkeratosis (Irish Terrier & Kromfohländer)	HFH	x

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Disease	SUV39H2	Hereditary nasal parakeratosis (greyhound)	HNPk	x
Disease	SUV39H2	Hereditary nasal parakeratosis (Labrador Retriever)	HNPk	*xP
Disease	COL4A5	Hereditary XL nephritis (Samoyede)	XLHN	x
Disease	HSF4	Hereditary cataract	HC1	x
Disease	HSF4	Hereditary cataract (Australian Shepherd type)	HC2	x
Disease	SLC2A9	Hyperuricosuria	HUU	x
Disease	FNIP2	Hypomyelination (Weimaraner)	HYM	x
Disease	TPO	Hypothyroidism (French Bulldog)	CHG	x
Disease	TPO	Hypothyroidism (Toy Fox Terrier)	CHG	x
Other	EPAS1	Hypoxia (altitude adaptation)	Hypoxia	x
Disease	F8	Haemophilia A (German Boxer)	Hem A	x
Disease	F8	Haemophilia A (German Shepherd Type 1)	Hem A	x
Disease	F8	Haemophilia A (German shepherd dog, type 2)	Hem A	x
Coat colours	MFS12	I-locus intensity (pheomelanin colour dilution)	I-Loc	x
Coat characteristics	RSPO2	IC-Lokus Impropper Coat (Furnishing/Rauhaar)	IC-Loc	x
Disease	NIPAL4	Ichthyosis (American Bulldog)	I	x
Disease	SLC27A4	Ichthyosis (Great Dane)	I	x
Disease	PNPLA1	Ichthyosis 1 (Golden Retriever)	ICH-GR1	x
Disease	CUBN	Immerslund-Gräsbeck syndrome (Beagle)	IGS Beagle	x
Disease	CUBN	Immerslund-Gräsbeck syndrome (Border Collie)	IGS-BC	x
Disease	CUBN	Immerslund-Gräsbeck syndrome (Komondor)	IGS-KOM	x
Disease	SLC25A12	Inflammatory myopathy (Dutch shepherd dog)		x
Disease	AKNA	Inflammatory Pulmonary Disease (Long-haired Collie)	IPD	x
Disease	DIRAS1	Juvenile myoclonic epilepsy (Rhodesian Ridgeback)	JME	x
Coat colours	CBD103	K-Lokus Dominant black	K-Loc	x
Disease	RBM20	Cardiomyopathy, dilated (Schnauzer)	DCM	x
Disease	TTN	Cardiomyopathy, dilated, risk variant 2 (Dobermann)	DCM	x
Disease	YARS2	Cardiomyopathy, juvenile mortality (Belgian Malinois)	CJM	x
Disease	CAT	Catalase deficiency (Beagle)		x
Disease	CNTNAP1	Laryngeal paralysis and polyneuropathy (Leonberger)	LPPN3	x
Disease	C3	Complement 3 deficiency (Epagneul Breton)	C3 Def	x
Disease	CYB5R3	Congenital methaemoglobinaemia (dwarf spitz)		x
Disease	LRIT3	Congenital stationary night blindness (Beagle)	CSNB	x
Disease	RPE65	Congenital stationary night blindness (Briard)	CSNB	x
Disease	CHRNE	Congenital myasthenic syndrome (Jack Russell Terrier)	CMS	x
Disease	COLQ, LOC608697	Congenital myasthenic syndrome (Labrador Retriever)	CMS	x
Disease	CHAT	Congenital myasthenic syndrome (Old Danish Pointer type)	CMS	x
Other	ATP7A	Copper toxicosis-protective modifier (Labrador Retriever)		x
Features	FSTS3	Body size and dental anomalies 1 (Shetland Sheepdog)	Dental	x
Features	GH1	Body size and dental anomalies 2 (Shetland Sheepdog)	Body Size	x
Features	GHR	Body size-GHR type1	Body Size	x
Features	GHR	Body size-GHR type2	Body Size	x

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Features	HMGA2	Body size-HMGA2	Body Size	x
Features	IGF1	Body size-IGF1	Body Size	x
Features	IGFR1	Body size-IGFR1	Body Size	x
Features	STC2	Body size-STC2	Body Size	x
Disease	L2HGDH	L-2-hydroxyglutaric aciduria (Staffordshire Bull Terrier)	L-2-HGA	x
Coat characteristics	FGF5	L1-Lokus longhair (common variant)	L1-Loc	x
Coat characteristics	FGF5	L2-Lokus Longhair (Akita)	L2-Loc	x
Coat characteristics	FGF5	L4-Lokus Longhair (Afghan, French Bulldog)	L4-Loc	x
Disease	ATG4D	Lagotto storage disease (Lagotto Romagnolo)	LS	x
Disease	TSEN54	Leukodystrophy (middle schnauzer)	LD	x
Disease	NAPEPLD	Leukoencephalomyelopathy (Leonberger)	LEMP	x
Disease	ITGB2	Leukocyte adhesion deficiency type I (Irish setter)	CLAD-I	x
Disease	FERMT3	Leukocyte adhesion deficiency type III (German shepherd dog)	CLAD-III	x
Disease	P3H2	Lundehund syndrome (Norwegian Lundehund)	LS	x
Disease	TUBB1	Macrothrombocytopenia (Cavalier King Charles Spaniel)	MTCP	x
Disease	TUBB1	Macrothrombocytopenia (Jack Russell Terrier)	MTCP	x
Disease	MYH9	May-Hegglin anomaly	MHA	x
Disease	SGSH	Mucopolysaccharidosis IIIA (Dachshund)	MPS IIIA	x
Disease	GUSB	Mucopolysaccharidosis VII (German Shepherd Dog)	MPS VII	x
Disease	IDUA	Mucopolysaccharidosis I (Plott Hound)	MPS I	x
Disease	SGSH	Mucopolysaccharidosis IIIA (New Zealand Huntaway)	MPS IIIA	x
Disease	ABCB1	Multidrug resistance 1	MDR1	x
Disease	BEST1	Multifocal retinopathy 1	CMR1	x
Disease	BEST1	Multifocal retinopathy 2 (Coton de Tulear)	CMR2	x
Disease	SERAC1	Multiple system degeneration (Chinese Crested)	CMSD	x
Disease	SERAC1	Multiple system degeneration (Kerry Blue Terrier)	CMSD	x
Disease	COL6A1	Muscular dystrophy (Landseer)	MD	x
Disease	COL6A3	Muscular dystrophy 1 (Labrador Retriever)	MD	x
Disease	COL6A3	Muscular dystrophy 2 (Labrador Retriever)	MD	x
Disease	DMD	Duchenne muscular dystrophy (Golden Retriever)	DMD	x
Disease	DMD	Duchenne muscular dystrophy (Norfolk terrier)	DMD	x
Disease	DMD	Duchenne muscular dystrophy 1 (Cavalier King Charles Spaniel)	DMD	x
Disease	DMD	Duchenne muscular dystrophy 2 (Cavalier King Charles Spaniel)	DMD	x
Disease	ADAMTSL2	Musladin-Lueke syndrome (Beagle)	MLS	x
Disease	MSTN	Myostatin deficiency (Whippet)		x
Disease	CLCN1	Myotonia Congenita (Australian Cattle Dog)	MC	x
Disease	CLCN1	Myotonia Congenita (Schnauzer)	MC	x
Disease	MTM1	Myotubular myopathy (Labrador Retriever)	MTM-XL	x
Disease	MTM1	Myotubular myopathy (Rottweiler)	MTM-XL	x
Disease	HCRTR2	Narcolepsy (Dachshund)	NARC	x
Disease	HCRTR2	Narcolepsy (Labrador Retriever)	NARC	x
Disease	ATF2	Neonatal encephalopathy with seizures (poodle)	NEWS	x

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Disease	SPTBN2	Neonatal cortical cerebellar degeneration	NCCD	x
Disease	PLA2G6	Neuroaxonal dystrophy (Papillon)	NAD	x
Disease	VPS11	Neuroaxonal dystrophy (Rottweiler)	NAD	x
Disease	TECPR2	Neuroaxonal dystrophy (Spanish water dog)	NAD	x
Disease	PPT1	Neuronal ceroid lipofuscinosis 1	NCL-1	x
Disease	CTSD	Neuronal ceroid lipofuscinosis 10 (American Bulldog)	NCL-10	x
Disease	TPP1	Neuronal ceroid lipofuscinosis 2	NCL-2	x
Disease	ARSG	Neuronal ceroid lipofuscinosis 4A (American Staffordshire Terrier)	NCL-4A	x
Disease	CLN5	Neuronal ceroid lipofuscinosis 5 (Golden Retriever)	NCL-5	x
Disease	CLN5	Neuronal ceroid lipofuscinosis 5 (herding dogs)	NCL-5	x
Disease	CLN6	Neuronal ceroid lipofuscinosis 6	NCL-6	x
Disease	MFSD8	Neuronal ceroid lipofuscinosis 7 (Chihuahua, Chinese crested dog)	NCL-7	x
Disease	CNL8	Neuronal ceroid lipofuscinosis 8 (Australian Shepherd)	NCL-8	x
Disease	CLN8	Neuronal ceroid lipofuscinosis 8 (Saluki)	NCL-8	x
Disease	CNL8	Neuronal ceroid lipofuscinosis 8 (setter)	NCL-8	x
Disease	FLCN	Renal cell carcinoma and nodular dermatofibrosis	RCND	x
Disease	SLC13A1	Osteochondrodysplasia (miniature poodle)	OCD	x
Disease	COL1A2	Osteogenesis Imperfecta (Beagle)	OI	x
Disease	SERPINH1	Osteogenesis Imperfecta (Dachshund)	OI	x
Disease	COL1A1	Osteogenesis Imperfecta Type 3 (Golden Retriever)	OI	x
Disease	P2Y12	P2RY12 receptor defect (Greater Swiss Mountain Dog)	P2Y12	x
Disease	PIGN	Paroxysmal dyskinesia	PxD	x
Disease	AMHR2	Persistent Müllergang syndrome (Schnauzer)	PMDS	x
Disease	NDRG1	Polyneuropathy (greyhound)		x
Disease	GJA9	Polyneuropathy 2 (Leonberger)	LPN2	x
Disease	RAB3GAP1	Polyneuropathy with eye abnormalities and neuronal vacuoles (Rottweiler)	POANV/JLPP	x
Disease	PKD1	Polycystic kidney disease (Bull Terrier)	PKD	x
Disease	KLKB1	Prekallikrein deficiency (Hairless Terrier, Shih-Tzu)		x
Disease	AGXT	Primary hyperoxaluria	PH1	x
Disease	ADAMTS17	Primary lens luxation	PLL	x
Disease	NME5	Primary ciliary dyskinesia (Alaskan Malamute)	PCD	x
Disease	CCDC39	Primary ciliary dyskinesia (bobtail)	PCD	x
Disease	ADAMTS17	Primary open-angle glaucoma (Basset Fauve de Bretagne)	POAG	x
Disease	ADAMTS10	Primary open-angle glaucoma (Beagle)	POAG	x
Disease	PDE6B	Progressive retinal atrophy (Irish Setter)	PRA	x
Disease	NECAP1	Progressive retinal atrophy (Giant Schnauzer)	PRA Schnauzer	x
Disease	CNGB1	Progressive retinal atrophy 1 (Papillon)	PRA-Pap1	x
Disease	PDE6B	Progressive retinal atrophy, cone-rod dystrophy 1 (American Pit Bull Terrier)	PRA-crd1	x
Disease	Rho	Progressive retinal atrophy, autosomal dominant (Mastiff)	PRA-AD	x
Disease	RPGRIP1	Progressive retinal atrophy, cone-rod dystrophy 4/crd1	PRA-crd1/crd4	x
Disease	PDE6B	Progressive retinal atrophy, early onset (Spanish water dog)	PRA-EO	x
Disease	IFT122	Progressive Retinal Atrophy, Late Onset (Lapponian Herder)	PRA	x
Disease	PRCD	Progressive retinal atrophy, progressive rod-cone degeneration	PRA-PRCD	*xP

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Disease	PDE6A	Progressive retinal atrophy, Rod-cone dysplasia 3 (Cardigan Welsh Corgi)	PRA-rcd3	x
Disease	C2orf71	Progressive retinal atrophy, rod-cone dysplasia 4	PRA-rcd4	x
Disease	RPGR	Progressive retinal atrophy, X-linked 1 (Husky)	XLPR1	x
Disease	KIRREL2	Protein Losing Nephropathy 1	PLN1	x
Disease	PDP1	Pyruvate dehydrogenase deficiency	PDH	x
Disease	PKLR	Pyruvate kinase deficiency (Basenji type)	PKDef	x
Disease	PKLR	Pyruvate kinase deficiency (Beagle type)	PKDef	x
Disease	PKLR	Pyruvate kinase deficiency (Labrador Retriever)	PKDef	x
Disease	PKLR	Pyruvate kinase deficiency (pug)	PKDef	x
Disease	PRKDC	Severe combined immunodeficiency (terrier)	SCID	x
Disease	RAG1	Severe combined immunodeficiency (Wetterhoun)	SCID	x
Disease	IL2RG	Severe combined immunodeficiency, x-linked (Basset)	XSCID	x
Disease	IL2RG	Severe combined immunodeficiency, x-linked (corgi)	XSCID	x
Coat characteristics	MC5R	SD locus (shedding, hair)	SD-Loc	x
Disease	COL11A2	Skeletal dysplasia 2	SD2	x
Coat colours	MITF	Sp1-Lokus Piebald (white piebald, Parti)	Sp1-Loc	x
Coat colours	MITF	Sp2-Lokus Piebald (white piebald, Parti)	Sp2-Loc	x
Disease	CAPN1	Spinocerebellar ataxia (late onset ataxia)	LOA/SCA	x
Disease	HES7	Spondylocostal dysostosis	SCD	x
Disease	KCNJ10	Cancellous degeneration with cerebellar ataxia 1 (Belgian Malinois)	SDCA1	x
Disease	KCNJ10	Cancellous degeneration with cerebellar ataxia 1 (Jack Russell Terrier)	SDCA1	x
Disease	ATP1B2	Cancellous degeneration with cerebellar ataxia 2 (Belgian Malinois)	SDCA2	x
Disease	ABCA4	Stargardt disease, Stargardt's disease (Labrador Retriever)	STGD	x
Features	non-coding region	Prick ears		x
Disease	ALDH5A1	Succinate-semi-aldehyde dehydrogenase deficiency (Saluki)	SSADHD	x
Features	T-box transcription factor T	T-Lokus stub rod (bobtail, natural short rod)	T-Loc	x
Disease	CDH23	Deafness (Beauceron)	EOAD	x
Disease	EPS8L2	Deafness (Rhodesian Ridgeback)	EOAD	x
Disease	LOXHD1	Deafness (Rottweiler)	Deafness	x
Disease	RASGRP1	Thrombopathy (Basset)		x
Disease	RASGRP1	Thrombopathy (Newfoundland)		x
Disease	VPS13B	Trapped Neutrophil Syndrome (Border Collie)	TNS	x
Disease	APRT	Urolithiasis		x
Disease	SCARF2	Van Den Ende-Gupta syndrome	VDEGS	x
Disease	MICOS13, C19orf70	Ventricular arrhythmia (Rhodesian Ridgeback)	IVA	x
Disease	SERPINF2	Delayed postoperative bleeding tendency (DEPOH)	DEPOH	x
Disease	VDR	Vitamin D-dependent rickets	HVDRR	x
Disease	VWF	Von Willebrand disease I	vWD I	x
Disease	VWF	Von Willebrand Disease II (German Shorthair)	vWD II	x
Disease	VWF	Von Willebrand disease III (Kooikerhondje)	vWD III	x

NEW

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Disease	VWF	Von Willebrand disease III (Shetland Sheepdog)	vWD III	x
Disease	BIN1	Centronuclear myopathy (Great Dane)	CNM	x
Disease	SEL1L	Cerebellar ataxia (Finnish Hound)	CA	x
Disease	SNX14	Cerebellar cortical degeneration (Magyar Vizsla)		x
Features	GH1	Dwarfism, growth hormone deficiency (Chihuahua)	Body Size	x