

CombiBreed Breeding Healthy Pets - Dog (Belgium) - H169 - Included tests

Test code	Name	Gene	Mutation
H904	Achromatopsia 3 (Day Blindness)	CNGB3	complete deletion
H918	Achromatopsia 3 (Day Blindness) - Pointer type	CNGB3	c.784G>A
H336	Acral Mutilation Syndrome (AMS)	GDNF	g.70875561C>T
H680	Amelogenesis Imperfecta (AI) - Akita	ACPT	c.1189dupG
H351	Amelogenesis Imperfecta (AI) - Greyhound	ENAM	c.1991_1995delITTTCC
H679	Amelogenesis Imperfecta (AI) - Russel Terrier	ENAM	c.716C>T
H889	Bilateral Deafness and Vestibular Dysfunction (MYO7A, DINGS2) - Doberman	MYO7A	c.3719G>A
H493	Bully Whippet - Whippet Double Muscling	MSTN	c.939_940delITG
H165	Canine Multiple System Degeneration (CMSD) - Chinese Crested	SERAC1	c.182+1_182+4del
H392	Canine Scott Syndrome (CSS)	ANO6	c.1934+1G>A
H807	Cardiomyopathy and juvenile mortality (CJM) - Belgian Shepherd	YARS2	c.1054G>A
H749	Centronuclear Myopathy (CNM) - Labrador Retriever	HACD1	c.203_204ins[N[236];CACACAAAGGTTT]
H331	Cerebellar Ataxia (SDCA1) - Belgian Shepherd	KCNJ10	c.986T>C
H341	Cerebellar Ataxia (SDCA2) - Belgian Shepherd	ATP1B2	c.130_131ins[LT796559.1:g.50_276]
H284	Cerebellar Degeneration-Myositis Complex (CDMC) - NSDTR	SLC25A12	c.1337C>T
H992	Cerebellar Hypoplasia (CH) - White Swiss Shepherd	RELN	c.2839delG
H709	CLAD (Canine Leukocyte Adhesion Deficiency) Type I - Irish Setter	ITGB2	c.107G>C
H484	CLAD (Canine Leukocyte Adhesion Deficiency) Type III - German Shepherd	FERMT3	c.1349_1350insAAGACGGCTGCC
H871	CMR1 (Canine Multifocal Retinopathy)	BEST1	c.73C>T
H730	CMR2 (Canine Multifocal Retinopathy)	BEST1	c.482G>A
H306	CMR3 (Canine Multifocal Retinopathy) 1	BEST1	c.1466G>T
H307	CMR3 (Canine Multifocal Retinopathy) 2	BEST1	c.1388delC
H950	CNS Atrophy with Cerebellar Ataxia (CACA) - Belgian Shepherd	SELENOP	c.-6582_*516del
H630	Coat Colour Merle	PMEL	SINE insertion (200-280 bp)
H705	Collie Eye Anomaly CEA, CH	NHEJ1	c.588+462_588+8260del7799bp
H446	Congenital Dysrhomonogenic Hypothyroidism with Goiter (CDH)	SLC5A5	c.1172-1G>A
H416	Congenital Hypothyroidism with Goiter (CHG) - Spanish Water Dog	TPO	c.39_40insG
H391	Congenital Myasthenic Syndrome (CMS) - Heideterrier	CHRNE	c.1508dup
H339	Congenital Myasthenic Syndrome (CMS) - Labrador Retriever	COLQ	c.1010T>C
H728	Congenital Stationary Night Blindness (CSNB) - Briard	RPE65	c.460_463del
H312	Craniomandibular Osteopathy (CMO) - Terrier Type	SLC37A2	c.1332C>T
H151	Craniomandibular Osteopathy (CMO) - Weimaraner	SLC35D1	c.1021_1024delTCAG
H355	Dandy-Walker-Like Malformation (DWLM) / Cerebellar Hypoplasia (CH) - Eurasier	VLDLR	c.1713delC
H308	Degenerative Myelopathy Exon 1 (DM Exon 1) - Bernese Mountain Dog	SOD1	c.52A>T
H673	Degenerative Myelopathy Exon 2 (DM Exon 2)	SOD1	c.118G>A
H959	Demyelinating Polyneuropathy (DP)	SBF2	c.2363+1G>T
H327	Dental Hypomineralization	FAM20C	c.899C>T
H974	Dental-Skeletal-Retinal Anomaly (DSRA)	MIA3	c.3822+3_3822+4del
H149	Disproportionate Dwarfism - Dalmatian	PRKG2	c.1601T>G
H913	Dry Eye Curly Coat Syndrome	FAM83H	c.977delC
H497	Dystrophic Epidermolysis Bullosa (RDEB) - Golden Retriever	COL7A1	c.5716G>A
H914	Early Onset Progressive Polyneuropathy (EOPP)	NGRD1	c.1080_1089del
H185	Ehlers-Danlos Syndrome (EDS, TNXB-related) - Dog (variant 1)	TNXB	c.2012G>A
H189	Ehlers-Danlos Syndrome (EDS, TNXB-related) - Dog (variant 2)	TNXB	c.2900G>A
H672	Exercise Induced Collapse, EIC	DNM1	c.767G>T
H980	Exfoliative Cutaneous Lupus Erythematosus	UNC93B1	438C>A
H435	Factor VII deficiency	F7	c.407G>A
H863	Fanconi syndrome - FS	FAN1	:c.2954_3090+181del
H676	FN, Familial Nephropathy - (English) Cocker Spaniel	COL4A4	c.115A>T
H361	Gangliosidosis 2	HEXA	c.967G>A
H702	Gangliosidosis (GM1) - Shiba Inu	GLB1	c.1649delC
H297	Gangliosidosis (GM2, Sandhoff Disease) - Shiba Inu	HEXB	c.618_620delCCT
H447	Glanzmann's Thrombasthenia (GT)2-Dog	ITGA2B	c.1360_1373dup
H737	Globoid Cell Leukodystrophy (GCL, Krabbes disease) - Terrier Type	GALC	c.473A>C
H415	Glycogen Storage Disease Ia (GSD1a) - Maltese	G6PC	c.450G>C
H347	Glycogen Storage Disease II (GSD2, Pompe) - Dog	GAA	c.2237G>A
H717	Glycogen Storage Disease VII (GSD7) / Phosphofructokinase Deficiency (PFK Def)	PFKM	c.2228G>A
H752	Gray Collie Syndrome (Cyclic Neutropenia)	AP3B1	c.2401_2402insA
H653	Hereditary Ataxia (RAB24-related) - Old English Sheepdog and Gordon Setter	RAB24	c.113A>C
H781	Hereditary Ataxia (SCA) - Australian Shepherd	PNPLA8	c.1169_1170dup
H699	Hereditary Cataract (HC, HSF4-related)	HSF4	c.971_972insC
H809	Hereditary Cataract (HC, HSF4-related) - Australian Shepherd	HSF4	c.971del
H182	Hereditary Deafness (EAOD) - Beauceron	CDH23	c.700C>T
H492	Hereditary Footpad Hyperkeratosis, FAM83G-related	FAM83G	c.155G>C
H444	Hereditary Footpad Hyperkeratosis, KRT16-related (FNEPPK1)	KRT16	c.[1147_1148delinsCGGA;1165del]
H460	Hereditary Nasal Parakeratosis (HNPK) - Greyhound	SUV39H2	c.996+3_996+6delAAGT
H988	Hereditary Necrotizing Myelopathy (HNM)	IBA57	c.439C>T
H364	Hypocalata	CAT	c.979G>A
H365	Hypomyelination / Shaking Puppy Syndrome (SPS) - Weimaraner	FNIP2	c.880delA
H378	Ichthyosis - American Bulldog Type	NIPAL4	g.52737379delC
H873	Ichthyosis - Golden Retriever Type 1	PNPLA1	c.1445_1447delinsTACTACTA
H782	Ichthyosis - Golden Retriever Type 2	ABHD5	c.1006_1019del
H368	Inherited Myopathy of Great Danes (IMGD) / Centronuclear Myopathy (CNM, HMLR)	BIN1	c.786-2A>G

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H366	Intestinal Cobalamin Malabsorption (ICM, IGS) – Beagle	CUBN	c.786delC
H367	Intestinal Cobalamin Malabsorption (ICM, IGS) – Border Collie	CUBN	c.8392delC
H861	Junctional Epidermolysis Bullosa (JEB, LAMA3-related) - German Pointer	LAMA3	c.1514C>T
H779	Juvenile Brain Disease (JBD) – Juvenile Encephalopathy	PITRM1	c.175_180del
H486	Juvenile Epilepsy (JE/BFJE) – Lagotto Romagnolo	LG12	c.1552A>T
H329	Juvenile Laryngeal Paralysis Polyneuropathy (JLPP)	RAB3GAP1	c.743delC
H335	Juvenile Myoclonic Epilepsy (JME) – Rhodesian Ridgeback	DIRAS1	c.564_567delAGAC
H724	L-2-Hydroxyglutaric Aciduria (L2HGA) - Staffordshire Bull Terrier	L2HGDH	c.1298_1300delinsCTT
H627	Lagotto Storage Disease (LSD)	ATG4D	c.1288G>A
H693	Laryngeal paralysis and polyneuropathy, CNTNAP1-related	CNTNAP1	c.2810G>A
H328	Late Onset Ataxia (LOA)	CAPN1	c.344G>A
H463	Lethal Acrodermatitis (LAD)	MKLN1	c.400+3A>C
H961	Lethal Lung Disease (LAMP3) – Airedale Terrier	LAMP3	XP_848889.2:p.(E387K)
H417	Leukoencephalomyelopathy – LEMP	NAPEPLD	c.345_346insC
H892	Leukoencephalomyelopathy – LEMP (Leonberger)	NAPEPLD	c.538G>C
H629	MDR1 Multi Drug Resistance – Dog	ABCB1	c.295_298delAGAT
H631	Microphthalmia (RBP4-related) – Irish Soft Coated Wheaten Terrier	RBP4	c.282_284del
H285	Mitochondrial Fission Encephalopathy (MFE) – Bullmastiff	MFF	c.471_475delinsCGCTCT
H424	Musladin-Lueke Syndroom (MLS)	ADAMTSL2	c.661C>T
H282	Mycobacterium Avium Complex (MAC) – Schnauzer	CARD9	c.493-495delAAG
H498	Myotonia Congenita – Australian Cattle Dog and Border Collie	CLCN1	c.2665insA
H410	Neonatal Cerebellar Ataxia – Coton de Tulear and Havanese	GRM1	c.2331_2332ins62bp
H812	Neonatal Encephalopathy (NEWS)	ATF2	c.152T>G
H289	Neuroaxonal Dystrophy (NAD) – Miniature American Shepherd	RNF170	c.367delG
H890	Neuroaxonal Dystrophy (NAD) – Papillon	PLA2G6	c.1579G>A
H888	Neuroaxonal Dystrophy (NAD) – Rottweiler	VPS11	c.2504A>G
H462	Neuroaxonal Dystrophy (NAD) – Spanish Water Dog	TECPR2	c.4009C>T
H987	Neuronal Ceroid Lipofuscinosis 12 (NCL12) – Australian Cattle Dog	ATP13A2	c.1118C>T
H458	Neuronal Ceroid Lipofuscinosis 12 (NCL12) – Tibetan Terrier	ATP13A2	c.1620delG
H804	Neuronal Ceroid Lipofuscinosis 4A (NCL4A) – Cerebellar Ataxia	ARSG	c.296G>A
H721	Neuronal Ceroid Lipofuscinosis 5 (NCL5)	CLN5	c.619C>T
H380	Neuronal Ceroid Lipofuscinosis 5 (NCL5) – Golden Retriever	CLN5	c.934_935del
H330	Neuronal Ceroid Lipofuscinosis 6 (NCL6) – Australian Shepherd	CLN6	c.829T>C
H978	Neuronal Ceroid Lipofuscinosis 7 (NCL7)	MFSD8	c.843delT
H958	Neuronal Ceroid Lipofuscinosis 8 (NCL8) – Saluki	CLN8	c.349dupT
H652	Neuronal Ceroid Lipofuscinosis 8-1 (NCL8-1) – English Setter	CLN8	c.491T>C
H337	Neuronal Ceroid Lipofuscinosis 8-2 (NCL8-2)	CLN8	c.585G>A
H965	Nonsyndromic Hearing Loss – Rottweiler	LOXHD1	c.5747G>C
H426	Oculoskeletale dysplasie 2 (OSD2)	COL9A2	g.5652893C>T
H898	Osteochondrodysplasia (OC)	SLC13A1	c.99+3353_*56671del
H431	Osteogenesis Imperfecta (OI) – Dachshund	SERPINH1	c.977T>C
H450	P2RY12 Receptor Platelet Disorder	P2RY12	c.516_518del
H634	Paroxysmal Dyskinesia (PED) – Irish Soft Coated Wheaten Terrier	PIGN	c.398C>T
H984	Paradoxical Pseudomyotonia	SLC7A10	c.126C>A
H996	Paroxysmal Exercise-Induced Dyskinesia (PED) – Weimaraner	TNR	c.831dupC
H963	Persistent Mullerian Duct Syndrome (PMDS)	AMHR2	c.262C>T
H872	Pituitary Dwarfism – Shepherd Type	LHX3	del[GTGTTTT] in intron 5
H495	Polyneuropathy (AMPN) – Alaskan Malamute	NDRG1	c.293G>T
H379	Polyneuropathy (LPN1)	ARHGEF10	c.1955_1958+6delCACGGTGAGC
H342	Polyneuropathy (LPN2) – Leonberger	GJA9	c.1107_1108delIAG
H439	Prekallikrein deficiency	KLKB1	c.988T>A
H790	Primary Ciliary Dyskinesia (PCD) – Alaskan Malamute	NME5	c.43delA
H414	Primary Ciliary Dyskinesia (PCD) – Old English Sheepdog	CCDC39	c.286C>T
H849	Primary Lens Luxation – PLL	ADAMTSL17	c.1473+1G>A
H757	Primary Open Angle Glaucoma (POAG) – Basset Fauve de Bretagne	ADAMTSL17	c.1552G>A
H496	Primary Open Angle Glaucoma (POAG) - Beagle	ADAMTSL10	c.1981G>A
H457	Primary Open Angle Glaucoma (POAG) / Primary Lens Luxation (PLL) – Shar-Pei	ADAMTSL17	c.3069_3074del
H371	Progressive Retinal Atrophy (Bas-PRA) – Basenji	SAG	c.1216T>C
H866	Progressive Retinal Atrophy (BBS2-PRA) – Shetland Sheepdog	BBS2	c.1222G>C
H357	Progressive Retinal Atrophy (crd1-PRA) – American Staffordshire Terrier	PDE6B	c.2404_2406del
H358	Progressive Retinal Atrophy (crd2-PRA) – American Pit Bull Terrier	IQCB1	c.952_53insC
H766	Progressive Retinal Atrophy (crd4-PRA/crd1)	RPGRIPI1	c.142_143ins
H739	Progressive Retinal Atrophy (Dominant PRA) – Mastiff	RHO	c.11C>G
H868	Progressive Retinal Atrophy (GR-PRA1) – Golden Retriever	SLC4A3	c.2601_2602insC
H473	Progressive Retinal Atrophy (GR-PRA2) – Golden Retriever	TTC8	c.669delA
H962	Progressive Retinal Atrophy (IFT122-PRA) - Lapponian Herder	IFT122	c.3176G>A
H166	Progressive Retinal Atrophy (MERTK-PRA)	MERTK	LINE1
H348	Progressive Retinal Atrophy (PAP-PRA1)	CNGB1	c.2685delA2687_2688insTAGCTA and c.2387_2389delinsCTAGCTAC
H280	Progressive Retinal Atrophy (PCYT2-Deficiency) – Saarloos Wolfdog	PCYT2	c.4A>G
H682	Progressive Retinal Atrophy (PRA, NECAP1-related) – Schnauzer	NECAP1	c.544G>A
H373	Progressive Retinal Atrophy (PRA3) – Tibetan Type	FAM161A	SINE insertie
H704	Progressive Retinal Atrophy (prcd-PRA)	PRCD	c.5G>A
H769	Progressive Retinal Atrophy (rcd1a-PRA) – Sloughi	PDE6B	c.2448_2449insTGAAGTCC
H768	Progressive Retinal Atrophy (rcd1-PRA) – Irish Setter	PDE6B	c.2421G>A
H770	Progressive Retinal Atrophy (rcd3-PRA)	PDE6A	c.1940delA
H511	Progressive Retinal Atrophy (rcd4-PRA)	PCARE	c.3149_3150insC
H455	Pyruvate Kinase Deficiency (PKDef) - Beagle	PKLR	c.994G>A
H971	Pyruvate Kinase Deficiency (PKDef) – West Highland White Terrier	PKLR	c.1333_1338dup

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H388	Sensory Neuropathy - Border Collie	FAM134B	g.80439639_86910352inv
H990	Skeletal Dysplasia (SD3) - Vizsla	PCYT1A	c.673T>C
H157	Spastic Ataxia (SACS-related) - Great Pyrenees	SACS	c.12731_12734del
H377	Spinal Dysraphism (SD) / Neural Tube Defects (NTD)	NKX2-8	c.449delinsTT
H303	Spinocerebellar Ataxia (SCA) - Terrier Type	KCNJ10	c.627C>G
H907	Stargardt disease 1	ABCA4	c.4176insC
H442	Thrombopathia - Basset Hound	RASGRP2	c.509_511del
H787	Trapped Neutrophil Syndrome (TNS)	VPS13B	c.2893_2896del
H911	Unilateral Deafness and Vestibular Dysfunction (PTPRQ, DINGS1) - Doberman	PTPRQ	c.9230_9231insA
H187	Van den Ende-Gupta Syndrome (VDEGS)	SCARF2	c.1873_1874del
H167	Von-Willebrand Disease Type 3 - Shetland Sheepdog	VWF	c.738del
H677	Von-Willebrand Disease Type 1	VWF	c.7437G>A
H345	Von-Willebrand Disease Type 2-2	VWF	c.1657T>G
H744	Von-Willebrand Disease Type 3 - Scottish Terrier	VWF	c.255delC
H916	Coefficient of Inbreeding		
H917	Heterozygosity		