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Analysis Certificate

H323 CombiBreed Labrador Retriever

Customer Info

Name	:	DANIEL MASIÁ CEBRIAN
Adress	:	Calle Algaira 5-7
Zip Code / City	:	46340 REQUENA
Customer no.	:	139046

Animal Info

Name	:	POLUX
Animal ID	:	939000002697745
Breed	:	Labrador retriever
Gender	:	Male
Date of Birth	:	16.10.2023
VHL ID	:	H644146

Sample Info

Order number	:	ES36909
Sample type	:	Swab
Certificate number	:	H113912
Test date	:	8.1.2025

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DNA is our core

Health Conditions

An explanation of these results is accessible in our Online Results Portal, which can be found in your account on the Combibreed Webshop. Within this portal, you will also discover comprehensive details for each test, including the breed relevance associated with each DNA test.

Breed Relevant Test Results

Code	Test Name	Gene	Mode of Inheritance	Result
H672	**Exercise Induced Collapse, EIC**	**DNM1**	**Autosomal Recessive**	**Carrier**
H387	Achromatopsia 2 (Day Blindness) – Labrador Retriever	CNGA3	Autosomal Recessive	Normal
H389	Alexander Disease	GFAP	Autosomal Dominant	Normal
H749	Centronuclear Myopathy (CNM) - Labrador Retriever	HACD1/PTPLA	Autosomal Recessive	Normal
H339	Congenital Myasthenic Syndrome (CMS) - Labrador Retriever	COLQ	Autosomal Recessive	Normal
H643	Cystinuria (Type I – A) – Labrador Retriever	SLC3A1	Autosomal Recessive	Normal
H673	Degenerative Myelopathy Exon 2 (DM Exon 2)	SOD1	Autosomal Recessive	Normal
H283	Dyserythropoietic Anemia and Myopathy Syndrome (DAMS) – Labrador Retriever	EHBP1L1	Autosomal Recessive	Normal
H686	Ehlers-Danlos Syndrome Type 1 – Labrador Retriever	COL5A1	Autosomal Dominant	Normal
H286	Haemophilia A – Labrador Retriever	F8	X-Linked Recessive	Normal
H675	HNPK (External Lab)	SUV39H2	Autosomal Recessive	Normal
H811	Hyperuricemia (HUU)	SLC2A9	Autosomal Recessive	Normal
H625	Inflammatory Linear Verrucous Epidermal Nevi (ILVEN) – Labrador Retriever	NSDHL	X-Linked Semi-domiant	Normal
H693	Laryngeal paralysis and polyneuropathy, CNTNAP1-related	CNTNAP1	Autosomal Recessive	Normal
H317	Macular Corneal Dystrophy	LOC489707	Autosomal Recessive	Normal
H746	Malignant Hyperthermia (MH) - All breeds	RYR1	Autosomal Dominant	Normal
H824	Modifier of Copper Toxicosis, ATP7A-related	ATP7A	X-Linked Recessive	Normal
H662	Modifier of Copper Toxicosis, RETN-related	RETN	Unknown	Normal
H714	Muscular Dystrophy (MD) – Labrador Retriever 1	COL6A3	Autosomal Recessive	Normal
H706	Muscular Dystrophy (MD) – Labrador Retriever 2	COL6A3	Autosomal Dominant	Normal
H688	Muscular Dystrophy-Dystroglycanopathy (MDD) – Labrador Retriever	LARGE1	Autosomal Recessive	Normal
H690	Myotonia Congenita – Labrador Retriever	CLCN1	Autosomal Recessive	Normal
H698	Narcolepsy – Labrador Retriever	HCRTR2	Autosomal Recessive	Normal
H895	Obesity	POMC	Unknown	Normal
H794	Oculoskeletal Dysplasia 1 (OSD1) / Retinal Dysplasia – Labrador Retriever	COL9A3	Autosomal Recessive	Normal
H473	Progressive Retinal Atrophy (GR-PRA2) – Golden Retriever	TTC8	Autosomal Recessive	Normal

Code	Test Name	Gene	Mode of Inheritance	Result
H704	Progressive Retinal Atrophy (prcd-PRA)	PRCD	Autosomal Recessive	Normal
H741	Pyruvate Kinase Deficiency (PKDef) – Labrador Retriever	PKLR	Autosomal Recessive	Normal
H510	Skeletal Dysplasia 2 (SD2) – Labrador Retriever	COL11A2	Autosomal Recessive	Normal
H907	Stargardt disease 1	ABCA4	Autosomal Recessive	Normal
H825	Wilson Disease, ATP7B-related	ATP7B	Unknown	Normal
H427	X-linked Myotubular Myopathy (XLMTM) – Labrador Retriever	MTM1	X-Linked Recessive	Normal

Trait Characteristics

Coat colour and patterns are caused by the interaction of several genetic traits. For in-depth insights into these genetic traits, you can visit the online portal with test results or access the knowledgebase via the CombiBreed webshop.

Coat Colours

Code	Test Name	Gene	Mode of Inheritance	Result	Phenotype
H847	Coat Colour D-Locus 1 - Dog	MLPH	Autosomal Recessive	D/D	No effect
H734	Coat Colour E-Locus - e1 (red/yellow)	MC1R	Autosomal Recessive	E/E	No effect
H632	Coat Colour E-Locus - e2 (Australian Cattle Dog cream colour variant)	MC1R	Autosomal Recessive	E/E	No effect
H783	Coat Colour E-Locus - e3 (Husky pale yellow/white variant)	MC1R	Autosomal Recessive	E/E	No effect
H819	Coat Colour K-Locus (Dominant Black)	CBD103	Autosomal Dominant	KB/KB	Fully black coat likely

Coat Patterns

Code	Test Name	Gene	Mode of Inheritance	Result	Phenotype
H820	Coat Colour A-Locus (Agouti) - Dog	ASIP	Autosomal Recessive	at/at	Black-and-tan coat
H784	Coat Colour E-Locus - Eg (Grizzle)	MC1R	Autosomal Dominant	E/E	No effect
H818	Coat Colour E-Locus - Em (Melanistic Mask)	MC1R	Autosomal Dominant	N/N	No effect

Other Coat Features

Code	Test Name	Gene	Mode of Inheritance	Result	Phenotype
H765	Hair Length - 1	FGF5	Autosomal Recessive	S/S	Short coat
H885	Hair Length - 2	FGF5	Autosomal Recessive	S/S	Short coat
H664	Hair Length - 3	FGF5	Autosomal Recessive	S/S	Short coat
H665	Hair Length - 4	FGF5	Autosomal Recessive	S/S	Short coat
H666	Hair Length - 5	FGF5	Autosomal Recessive	S/S	Short coat

On behalf of VHLGenetics B.V.,
A. de Lange MBA

Modes of Inheritance

Autosomal Co-Dominant: A mode of inheritance where the affected and normal alleles are expressed equally, leading to an intermediate phenotype when both alleles are present in carriers.

Autosomal Dominant: A single copy of a dominant allele from one parent is sufficient to express the disease/trait. Individuals with at least one dominant allele will exhibit the trait.

Autosomal Incompletely Dominant: A genetic inheritance pattern that functions as normal Autosomal Dominant. However, carriers are not guaranteed to express the trait.

Autosomal Incompletely Recessive: A genetic inheritance pattern that functions as normal Autosomal Recessive. However, affected individuals are not guaranteed to express the disease/trait.

Autosomal Recessive: Two copies of a recessive allele must be present for the trait to be expressed. If an individual has two recessive alleles, the disease/trait will be expressed. If they have one recessive allele, they are a carrier but do not exhibit the trait.

Autosomal Recessive Lethal: A genetic inheritance pattern where an individual must inherit two copies of the recessive allele to express a lethal trait, typically resulting in spontaneous abortion, stillbirth or early death.

Autosomal Semi-Dominant: A mode of inheritance where the phenotype is dependent on the number of copies present. Individuals with a single copy of the affected allele express a version of the trait specific to carriers. Individuals with two copies express the version specific to affected.

Mitochondrial: Genes located in the mitochondria, outside the cell nucleus, are inherited from the mother. Both sons and daughters can inherit these genes, but only daughters pass them on to their offspring.

Multifactorial: Disease/trait is influenced by multiple genetic and/or environmental factors, and may be difficult to predict.

Resistance/Susceptibility: The genetic predisposition of an individual or organism to either resist or be susceptible to a particular condition, disease, or treatment.

Risk factor: A risk factor in genetics refers to a specific genetic variation, trait, or condition that increases the likelihood of an individual developing a particular disease or health issue.

Unknown: Refers to cases where the mode of inheritance associated with the mutation is not yet fully identified or understood.

X-Linked Dominant: Dominant alleles located on the X chromosome result in the expression of the disease or trait. In females, a single copy of the allele is sufficient. In males, who have only one X chromosome, the presence of the dominant allele leads to the trait's expression.

X-Linked Recessive: Recessive alleles on the X chromosome cause the disease/trait to be expressed in males, who have only one X chromosome with the allele. Females need two copies of the recessive allele to exhibit the disease/trait.

X-Linked Semi-domiant: A mode of inheritance where the phenotype is dependent on the number of copies present. Females with a single copy of the affected allele express a version of the trait specific to carriers. Females with two copies, and males carrying the allele, express the version specific to affected animals.

Y-Linked: Genes on the Y chromosome are passed exclusively from father to son. Traits determined by Y-linked genes are inherited in a straightforward manner down the paternal lineage.