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Genetic Check-Up
Health & Reproduction

Animal : **Chiësvar's blue bittersweet**

Identification No. :	**528 140 000 806 343**	Owner :	**Iris MADERN**
Breed :	**Australian Shepherd**	Sample No. :	**E00824257** (sampled on 07/01/2023)
Gender :	**Female**	Result code :	**A00023489**
Birth date :	**14/09/2020**	Sampler :	**C.g BREEDYL** (Veterinarian - Order No. : 100125)
Pedigree :			Sample **authenticated**
Result validated on :	20/01/2023	Document issued on :	20/01/2023

DISEASES	GENE Mutation	Expression mode	RESULT	EXPRESSION	TRANSMISSION
Collie Eye Anomaly (Choroidal Hypoplasia) (CEA)	NHEJ1 c.588+462_588+8260del7799	Autosomal recessive	**Normal homozygous**	✓	✓
Spinocerebellar Ataxia	PNPLA8 c.1169_1170dupTT	Autosomal recessive	**Normal homozygous**	✓	✓
Hereditary Cataract	HSF4 g.85286582delC	Autosomal codominant	**Heterozygous**	!	!
Degenerative Myelopathy	SOD1 c.118G>A	Autosomal recessive	**Normal homozygous**	✓	✓
Multidrug Sensitivity (MDR1)	MDR1 c.227_230delATAG	Autosomal codominant	**Heterozygous**	!	!
Achromatopsia	CNGB3 complete deletion	Autosomal recessive	**Normal homozygous**	✓	✓
Progressive Retinal Atrophy (PRA-prcd)	PRCD c.5G>A	Autosomal recessive	**Normal homozygous**	✓	✓
Neuronal Ceroid Lipofuscinosis					
NCL6	CLN6 c.829T>C	Autosomal recessive	**Heterozygous**	✓	!
NCL8	CLN8 c.585G>A	Autosomal recessive	**Normal homozygous**	✓	✓
Cobalamin Malabsorption	CUBN c.8392delC	Autosomal recessive	**Normal homozygous**	✓	✓
Hyperuricosuria	SLC2A9 c.616G>T	Autosomal recessive	**Normal homozygous**	✓	✓
von Willebrand Disease (vWD1)	VWF c.7142C>T	Autosomal recessive	**Normal homozygous**	✓	✓
Multifocal Retinopathy (CMR1)	BEST1 c.73C>T	Autosomal recessive	**Normal homozygous**	✓	✓

INTERPRETATION OF THE RESULT
Normal homozygous : the animal carries 2 normal copies of the gene.
Heterozygous : the animal carries a normal copy and a defective copy of the gene.
Mutated homozygous : the animal carries 2 defective copies of the gene.

	EXPRESSION	TRANSMISSION
✓	The animal will not develop the form of the disease associated to the tested mutation.	The animal does not transmit the tested mutation.
!	The animal will develop the disease without being able to predict the age of onset or severity of symptoms.	The animal will transmit the tested mutation to all or part of its offspring. Reproduction is to be avoided or adapted according to the disease and the associated frequency.

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MORPHOLOGICAL TRAITS

	GENE Mutation	Expression mode	RESULT
A Locus - Agouti			
a	ASIP c.286C>T	Autosomal recessive	a^w/a^w
A^y	ASIP c.244G>T/248G>A	Autosomal dominant	a^w/a^w
B Locus - Brown			**Brown or Carrier of brown (b/b ou B/b)**
b^a	TYRP1 c.555T>G	Autosomal recessive	**B/B**
b^c	TYRP1 c.121T>A	Autosomal recessive	**B/b^c**
b^d	TYRP1 c.1033_1035del	Autosomal recessive	**B/b^d**
b^e	TYRP1 c.1025T>G	Autosomal recessive	**B/B**
b^s	TYRP1 c.991C>T	Autosomal recessive	**B/B**
E Locus - Extension			**Melanistic mask (E^m/E)**
e	MC1R c.916C>T	Autosomal recessive	**E/E**
e^2	MC1R g.63695679C>G	Autosomal recessive	**E/E**
e^3	MC1R c.816_817delCT	Autosomal recessive	**E/E**
E^m	MC1R c.790A>G	Autosomal dominant	**E^m/E**
K Locus - Dominant Black (K^B)	CBD103 c.231_233del	Autosomal dominant	**Not expressed (k^y/k^y)**
M Locus - Merle	SILV SINE insertion	Autosomal dominant	**Merle heterozygous (M/m)**
Curly or Wavy Coat (c^1)	KRT71 c.451C>T	Autosomal dominant	**Non-carrier (C/C)**
Polydactyly	LMBR1 DC-2	Autosomal dominant	**Non-carrier of polydactyly**
Tail Length	T c.189C>G	Autosomal dominant	**Homozygous for long tail**
Shedding	MC5R g.24430748C>T	Autosomal codominant	**High shedding**

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DISEASES

	GENE Mutation	Expression mode	RESULT	EXPRESSION	TRANSMISSION
Congenital Macrothrombocytopenia	TUBB1 c.745G>A	Autosomal dominant	**Normal homozygous**	✓	✓

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