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Genetic Check-Up
Health & Reproduction

Animal : **Bullbrothers Hott As Hell**

Identification No. :	**900 133 000 583 270**	Owner :	**Matteo MANTUANO**
Breed :	**Staffordshire Bull Terrier**	Sample No. :	**E00847325** (sampled on 24/05/2023)
Gender :	**Male**	Result code :	**A00038092**
Birth date:	**04/11/2021**	Sampler :	**Hélène HENNEBOT** (Veterinarian - Order No. : 25614)
Pedigree :			Sample **authenticated**
Result validated on :	16/06/2023	Document issued on :	16/06/2023

DISEASES	GENE Mutation	Expression mode	RESULT	EXPRESSION	TRANSMISSION
L-2-Hydroxyglutaric Aciduria	L2HGDH c.1297T>C et c.1299C>T	Autosomal recessive	**Normal homozygous**	✓	✓
Hereditary Cataract	HSF4 g.85286582insC	Autosomal recessive	**Normal homozygous**	✓	✓
Hyperuricosuria	SLC2A9 c.616G>T	Autosomal recessive	**Normal homozygous**	✓	✓
Multidrug Sensitivity (MDR1)	MDR1 c.227_230delATAG	Autosomal codominant	**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^y/A^y
B Locus - Brown			**Non-carrier of brown (B/B)**
b^a	TYRP1 c.555T>G	Autosomal recessive	**B/B**
b^c	TYRP1 c.121T>A	Autosomal recessive	**B/B**
b^d	TYRP1 c.1033_1035del	Autosomal recessive	**B/B**
b^e	TYRP1 c.1025T>G	Autosomal recessive	**B/B**
b^s	TYRP1 c.991C>T	Autosomal recessive	**B/B**
D Locus - Dilution			**Non-carrier of dilution (D/D)**
d	MLPH c.-22G>A	Autosomal recessive	**D/D**
d^2	MLPH c.705G>C	Autosomal recessive	**D/D**
d^3	MLPH c.667_668insC	Autosomal recessive	**D/D**
E Locus - Extension			**Melanistic mask (E^m/E^m)**
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^m
K Locus - Dominant Black (K^B)	CBD103 c.231_233del	Autosomal dominant	**Not expressed (k^y/k^y)**
Polydactyly	LMBR1 DC-2	Autosomal dominant	**Non-carrier of polydactyly**
Shedding	MC5R g.24430748C>T	Autosomal codominant	**Low shedding**

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DISEASES	GENE Mutation	Expression mode	RESULT	EXPRESSION	TRANSMISSION
Congenital Macrothrombocytopenia	TUBB1 c.745G>A	Autosomal dominant	**Normal homozygous**	✓	✓
Degenerative Myelopathy	SOD1 c.118G>A	Autosomal recessive	**Normal homozygous**	✓	✓

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