GENOMIA
genetic laboratory

Result certificate #012345

Screening for hereditary diseases and appearance traits of the horse

Customer: Jan Novák, Dlouhá 1, 30000 Plzeň, Czech Republic
Sample:
Sample: 21-12351
Date received: 01.02.2021
Sample type: horsehair

Information provided by the customer
Name: Black And White DEMO
Breed: Český teplokrevník
Date of birth: 25.11.2016
Reg.number : DE-123-456-789-012
Tattoo: 123456789012345
Sex: male
Date of sampling: 01.02.2021
The identity of the animal has been checked by MVDr. Veselý Josef.

SAMPLE

Health summary

clear: 28 | no effect: 2 | may affect: 1

Appearance

Name	Abbr.	Gene	Mutation	Copies	Result
Coat colour, agouti	a	ASIP	c.187_197del	1	carrier
Megacolon (overo)	frame overo	EDNRB	c.353_354delinsAG	0	clear
Coat colour, dominant white (sabino)	SB1	KIT	g.79544206A>T	0	no effect
Coat colour, dominant white	W18	KIT	c.1346+1G>A	0	no effect
Coat colour, dominant white	W20	KIT	c.2045G>A	2	white spots
Curly coat		KRT25	c.266G>A	0	no effect
Coat colour, extension	e	MC1R	c.248C>T	2	chesnut
Coat colour, extension	e^a	MC1R-1-MC1R-2	c.250G>A	0	no effect
Coat colour, white spotting	macchiato	MITF	c.629A>G	0	no effect
Coat colour, white spotting	SW1	MITF	g.20117302Tdelins11	0	no effect
Coat colour, white spotting	SW3	MITF	c.837_841del	0	no effect
Coat colour, white spotting	MITF^244Glu	MITF	c.1031G>A	0	no effect
Coat colour, silver		PMEL	c.1849C>T	0	no effect
Coat colour, cream dilution	C^Cr	SLC45A2	c.601G>A	1	may affect
Coat colour, cream dilution	C^prl	SLC45A2	c.1129G>A	0	no effect
Coat colour, cream dilution	C^sno	SLC45A2-649	c.449G>A	0	no effect

Autosomal dominant disorders

Name	Abbr.	Gene	Mutation	Copies	Result

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Polysaccharide storage myopathy/PSSM1/Exertional rhabdomyolysis	PSSM1	GYS1	c.926G>A	0	clear
Myositis, immune-mediated	IMM	MYH1	c.959A>G	0	clear
Auditory-pigmentary syndrome	SW2	PAX3	c.209G>A	0	clear
Auditory-pigmentary syndrome, PAX3-related	SW4	PAX3-1	c.95C>G	0	clear
Malignant hyperthermia	MH	RYR1	c.7363C>G	0	clear
Hyperkalemic Periodic Paralysis	HYPP	SCN4A	c.4248C>G	0	clear

Autosomal recessive disorders

Name	Abbr.	Gene	Mutation	Copies	Result
Dwarfism	D4	ACAN	c.7633_7653del	0	clear
Dwarfism	D1	ACAN	c.245del	0	clear
Dwarfism	D2	ACAN	c.1270G>A	0	clear
Dwarfism	D3	ACAN	c.1513G>C	0	clear
Hydrocephalus		B3GALNT2	c.1423C>T	0	clear
Dwarfism, B4GALT7-related		B4GALT7	c.50G>A	0	clear
Cerebellar abiotrophy	NCCD	CA /MUTH	c.284G>A	0	clear
Myotonia		CLCN1	c.1775A>C	0	clear
Ocular squamous cell carcinoma	SCC	DDB2	c.1013C>T	0	clear
Glanzmann thrombasthenia		ITGA2B	g.19247983_19247992del	0	clear
Epidermolysis bullosa, junctionalis	JEB	LAMC2	c.1372dup	0	clear
Dilute coat color with neurological defects	LFS	MYO5A	c.4249del	0	clear
kyphoscoliotic Ehlers-Danlos syndrome (kEDS), PLOD1-related	WFFS	PLOD1	c.2032G>A	0	clear
Ehlers-Danlos syndrome, generic	HERDA	PPIB / HERDA	c.115G>A	0	clear
Severe combined immunodeficiency disease, autosomal	SCID	PRKDC	c.9478_9482del	0	clear
Hypoparathyroidism, RAPGEF5-related		RAPGEF5	c.2624C>A	0	clear
Hoof wall separation syndrome	HWSD	SERPINB11	c.504_505insC	0	clear
Immunodeficiency syndrome, SLC5A3-related	FIS	SLC5A3	c.1352C>T	0	clear

Association genetic tests

Name	Abbr.	Gene	Mutation	Copies	Result
Gaitedness, DMRT3-related		DMRT3	c.902C>A	0	no effect
Racing distance		MSTN	18:g.66608679T>C	0	no effect
Leopard complex pattern modifier	PATN1	RFWD3	24352525T>G	1	may affect

X-linked hereditary disorders

Name	Abbr.	Gene	Mutation	Copies	Result
Androgen insensitivity syndrome	AIS	AR	c.1630_1654del	0	clear
Androgen insensitivity syndrome	AIS	AR	c.1A>G	0	clear
Androgen insensitivity syndrome	AIS	AR	c.2132C>T	0	clear
Incontinentia pigmenti	IP	IKBKG	c.184C>T	0	clear

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Explanation

Interpretation of the results can be found on the website https://www.genomia.cz/en/veterinarni/horse/ on the pages of the respective examinations.

The mutations listed are annotated according to the EquCab3.0 reference sequence.

Recessive inheritance: the trait (disease) becomes apparent if the individual inherits it from both parents (2 copies); carriers of the trait (disease) are asymptomatic but pass the causal mutation on to the next generation (1 copy).

Dominant inheritance: it is sufficient for an individual to inherit the trait (disease) from one parent (1 copy).

X-linked recessive inheritance: in males, 1 copy of the mutated gene is enough to cause the disease; in females, 2 copies of the mutated gene are needed to cause the disease.

The results of the association genetic tests indicate the predisposition to the disease. This is not a detection of a causal mutation of the disease.

Method: SOP188-MPS-equine, MPS

Date of issue: 06.02.2021
Date of testing: 01.02.2021 - 06.02.2021
Approved by: Mgr. Martina Šafrová, Laboratory Manager

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verified and certified document GENOMIA genetic laboratory

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