GENOMIA
genetic laboratory

Result certificate #012345

Detection of c.2032G>A mutation in PLOD1 gene causing Warmblood Fragile Foal Syndrome

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.

Result: Mutation was not detected (N/N)

Legend: N/N = wild-type genotype. N/P = carrier of the mutation. P/P = mutated genotype (individual will be most probably affected with the disease). (N = negative, P = positive)

Explanation

Presence or absence of c.2032G>A mutation in PLOD1 gene causing Warmblood Fragile Foal Syndrome (WFFS) was tested. WFFS is an inherited disorder of connective tissue that affects the Warmblood and other relative breeds. The result of mutation is an extremely thin and fragile to tearing skin that is only loosely attached to subcutaneous tissue. Due to this mutation the skin is tearing and develops lacerations, seromas, heamatomas, ulcers and scars. Frequent skin lesions occur on legs, head and abdomen.

Mutation that causes WFFS is inherited as an autosomal recessive trait. That means the disease affects horses with P/P genotype only. The horses with N/P genotype are considered carriers of the disease (heterozygotes). In offspring of two heterozygous animals following genotype distribution can be expected: 25 % N/N (healthy non-carriers), 25 % P/P (affected), and 50 % N/P (healthy carriers).

Method: SOP175-WFFS, real-time PCR-ASA, accredited method

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

verified and certified document GENOMIA genetic laboratory

Genomia is accredited in compliance with ISO/IEC 17025:2018 under #1549
Genomia s.r.o, Republikánská 6, 31200 Plzeň, Czech Republic
www.genomia.cz, laborator@genomia.cz, tel: +420 373 749 999

ilac-MRA L 1549