

## 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 within the flexible scope of accreditation

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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