



## Test for Fragile Foal Syndrome Type 1

Please send a kit (\$35) for the following horse(s):

1) Registered name of Horse \_\_\_\_\_  
Reg # \_\_\_\_\_  
Breed \_\_\_\_\_ Birthdate: \_\_\_\_\_

2) Registered name of Horse \_\_\_\_\_  
Reg # \_\_\_\_\_  
Breed \_\_\_\_\_ Birthdate: \_\_\_\_\_

3) Registered name of Horse \_\_\_\_\_  
Reg # \_\_\_\_\_  
Breed \_\_\_\_\_ Birthdate: \_\_\_\_\_

4) Registered name of Horse \_\_\_\_\_  
Reg # \_\_\_\_\_  
Breed \_\_\_\_\_ Birthdate: \_\_\_\_\_

5) Registered name of Horse \_\_\_\_\_  
Reg # \_\_\_\_\_  
Breed \_\_\_\_\_ Birthdate: \_\_\_\_\_

- My horse already has a DNA sample on file with UC Davis (***must be a WPN DNA number originating at UC Davis. We cannot order this test with another registry's DNA sample, a converted blood sample or a file from Europe. Please send new sample if your horse does not meet this criteria.***)
- I am sending in a new DNA hair sample for testing directly to UC Davis (*Once ordered, the test will come to you via email from the laboratory. You will attach the hair and send directly to UC Davis*)

Owner Name \_\_\_\_\_  
Address \_\_\_\_\_  
City: \_\_\_\_\_ State/Prov: \_\_\_\_\_  
Zip/Postal Code: \_\_\_\_\_  
Phone \_\_\_\_\_ Fax \_\_\_\_\_  
Email \_\_\_\_\_

If kit should be emailed to a different email address:

Email kit to: \_\_\_\_\_

\_\_\_ Check \_\_\_ Card (Visa, MC, Discover, AmEx) \_\_\_ Please Invoice Me  
Credit card number \_\_\_\_\_ Exp. date \_\_\_\_\_  
Validation Code \_\_\_\_\_

Signature \_\_\_\_\_

KWPN of North America, KWPN-NA  
4037 Iron Works Parkway, Suite 140  
Lexington, KY 40511  
Phone: 859-225-5331 ♦ Fax: 859-554-0366 ♦ E-mail: drew@kwpn-na.org

## Fragile Foal Syndrome Type 1

Fragile Foal Syndrome Type 1 (FFS) is a fatal genetic defect of connective tissue characterized by hyperextensible, abnormally thin and fragile skin that cause extensive lesions of skin and mucous membrane (tissue that lines cavities and covers organs) throughout the body. Other signs of the disease include hyperextension of limb articulations, floppy ears, hydrops (accumulation of fluid in fetus), subcutaneous emphysema, hematomas and premature birth. Signs of the disease are present from birth. Newborn foals have to be euthanized because of poor prognosis for an untreatable condition.

FFS is caused by a mutation (c.2032G > A) in the *PLOD1* gene (*procollagen-lysine, 2-oxoglutarate 5-dioxygenase1*) that codes for an enzyme important for biosynthesis of collagen, which are complex molecules that provide strength and support to many body tissues. The mutation impairs normal function of the enzyme, which leads to development of the disease. In humans, mutations in this gene are associated with a similar defect known as Ehlers-Danlos Syndrome Type VI.

FFS is inherited as an autosomal recessive defect, which means that both males and females are equally affected and that two copies of the mutation (FFS/FFS) are needed to cause the disorder. Horses that carry one copy (N/FFS) of this recessive mutation are normal but can transmit the mutation to 50% of their offspring.

The Veterinary Genetics Laboratory offers a DNA test for FFS. The test benefits clinicians to help with diagnosis of FFS. The test benefits breeders and owners to identify carriers of FFS and to select mating pairs accordingly to avoid producing affected foals. Mating between two carriers has a 25% chance of producing affected foals.

**Testing recommendation:** The FFS mutation occurs in Warmblood horse populations that include Hanoverian, Selle Français, KWPN (Dutch), Oldenburg and Westphalian lineages. In addition, Holsteiner, Wurtemberger, Rhineland, Gelderlander, Zweibrucker and Bavarian Warmblood breeds are at risk for carrying this disorder. The incidence of carriers in Warmblood horses is estimated to be around 9-11%.

Note: Because of European patent restriction, the VGL accepts orders for FFS testing from any country, except those located in Europe.

Results are reported as:

N/N	No copies of the FFS mutation; animal is normal.
N/WFFS	1 copy of the FFS mutation; animal is normal but is a carrier. Horse can pass on the mutation to 50% of offspring.
FFS/FFS	2 copies of the FFS mutation; horse is affected.