



## FOCUS ARTICLE: TEST LAUNCHED FOR FOAL IMMUNODEFICIENCY SYNDROME

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Foal immunodeficiency syndrome (FIS), formally known as Fell Pony syndrome, was first recognised in UK Fell Pony foals in 1998, described as a syndrome of anaemia and immunodeficiency. Since, FIS cases in the Fell Pony have been reported across Europe and United States of America. Until recently the disease had only been reported in the Fell Pony, but in June 2008, the same condition was confirmed in a Dale Pony foal.

The disease manifests at 2-6 weeks after birth, the foals being apparently normal when born. The first clinical signs of this disease include diarrhoea, nasal discharge, poor growth, pale gums and decreased appetite. A progressive profound anaemia is a notable early feature of this disease (PCV<20%). Lymphocyte sub-population analysis show normal circulating T-lymphocyte numbers and severely reduced B-lymphocytes numbers.



Figure 4: Affected Fell Pony foal

Attempts at symptomatic treatment have been made; these include rehydration, antibiotics, vitamin and mineral (Se) supplements, analgesia, blood transfusions, erythropoietin injections. Despite extensive treatment and supportive therapy, foals die or are euthanased on the basis of lethargy, severe anaemia and persistent infections before sixteen weeks of age. To date there have been no validated cases where foals have survived this disease, FIS has a 100% mortality rate.

Breeders noted a familial pattern, with repeat matings producing both affected and unaffected foals. This together with examination of the stud book, which reveals a small effective breeding population and genetic bottlenecks in recent years, strongly suggests that FIS is an inherited disease caused by an autosomal recessive mutation. Research into the genetics of FIS, funded by The Horse Trust, has been performed at the Animal Health Trust, in collaboration with the University of Liverpool. Recent advances in equine genetics enabled this project to advance rapidly and in November 2009, a single point mutation was identified on chromosome 26 that segregated with the disease. The mutation was screened in 23 obligate carriers, including the sire and dam of an FIS affected Dales foal, all were heterozygous for the mutation. Thirty-one samples from affected animals were screened, including the affected Dales foal and all were homozygous for the mutation. Ninety-six samples from breeds which are unlikely to have interbred with the Fell or Dales, including Thoroughbred, Appaloosa, Arab and Sport Horses, were screened for the mutation, all were homozygous for the wild-type allele.

FIS carrier prevalence in the Dales and Fell population has been estimated by analysing a random anonymous selection of samples submitted to the Animal Health Trust for parentage verification. Two-hundred and fourteen Fell Pony samples were screened, of which eighty-two were carriers of the mutation (38%). Eighty-seven Dales Pony samples were screened, of which sixteen carried the FIS mutation (18%).

This breakthrough has led to the development of a test which can be used to screen the DNA of individual



ponies for the presence or absence of the mutation. The test, which is available from the Animal Health Trust, will enable breeders to make informed breeding decisions.

By identifying carriers, subsequent carrier-carrier matings can be avoided, since it is pairings of this type only which lead to FIS. We would urge any breeders of Fell or Dales ponies to utilise the test and also encourage using the test for any mixed breed ponies with Fell Pony or Dales Pony ancestry.

Since the launch of the FIS test, forty-one Fell Pony samples have been submitted for testing, twenty-two were carriers (54%) of FIS. Twenty-eight Dales Pony samples have been submitted for testing, three were carriers (11%).

Previously there has been no definitive test for an FIS foal so diagnosis was made on clinical signs and clinical history. In 2010, the test will act as a diagnostic aid to confirm suspected syndrome foals. Foal samples submitted will be processed as urgent and the result available in three working days from receipt of sample.

Our breeding advice is that all animals should remain actively breeding to avoid the loss of desirable breed traits and prevent a loss of genetic diversity in the population. Carriers can still be bred to tested clear ponies. On average, 50% of the offspring will be clear and 50% will be carriers; there can be no affected animals produced from a carrier-clear mating. Foals which will be used for breeding themselves can be DNA tested to determine whether they are clear or a carrier. In time, with careful breeding, the disease gene may eventually be eradicated from the population.

For further information on FIS testing please contact the Fell Pony Society, the Dales Pony Society or refer to the Animal Health Trust website at [http://www.aht.org.uk/genetics\\_tests.html#equine](http://www.aht.org.uk/genetics_tests.html#equine)

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