



## POLYSACCHARIDE STORAGE MYOPATHY TYPE 1 (PSSM1) IN NEW FOREST PONIES

The New Forest Pony Breeding and Cattle Society was notified that a New Forest pony had tested positive n/P1 (heterozygous) for the PSSM1-carrying gene in the summer of 2017. The sire tested n/P1, and testing of all New Forest stallions, at the Society's expense, began immediately. This statement sets out the position in spring 2018, before the start of the 2018 breeding season.

### **PSSM – What is it?**

Polysaccharide Storage Myopathy (PSSM1) is a dominant autosomal hereditary condition that can cause a genetic form of tying-up, with muscle damage and inability to move. In some horses, symptoms may begin by 2-3 years of age, while others remain sub-clinical. In many cases horse that have tested positive have had no history of 'tying-up' or other symptoms associated with PSSM.

It is reported that the genetic mutation responsible for type 1 PSSM has been found in more than 20 breeds of horses world-wide. Some horses make and store abnormal muscle glycogen and cannot tolerate dietary starches and sugars. Some horses with PSSM can be maintained with low-starch and low-sugar rations and precise exercise protocols.

### **Clinical Signs of PSSM**

(This section is taken from the Michigan State University website).

Horses with PSSM1 can have signs typically associated with tying-up. These signs are most commonly muscle stiffness, sweating, and reluctance to move in conjunction with increased serum creatine kinase (CK) activity. The signs are most often seen in horses when they are put into initial training or after a lay-up period when they receive little active turn-out. Episodes usually begin after very light exercise such as 10-20 minutes of walking and trotting. Horses with PSSM1 can exhibit symptoms without exercise.

During an episode, horses seem lazy, have a shifting lameness, tense up their abdomen, and develop tremors in their flank area. When horses stop moving they may stretch out as if to urinate. They are painful, stiff, sweat profusely, and have firm hard muscles, particularly over their hindquarters. Some horses will try pawing and rolling immediately after exercise. Most horses with PSSM1 have a history of numerous episodes of muscle stiffness at the commencement of training; however, mildly affected horses may have only one or two episodes/year. Rarely, episodes of muscle pain and stiffness can be quite severe, resulting in a horse being unable to stand and being uncomfortable even when lying down. The urine in such horses is often coffee coloured, due to muscle proteins being released into the bloodstream and passed into the urine. This is a serious situation, as it can damage the horse's kidneys if they become dehydrated.

Very young foals with PSSM1 occasionally show signs of severe muscle pain and weakness. This occurs more often if they have a concurrent infection such as pneumonia or diarrhoea.

### **How PSSM is inherited**

The genetic test verifies the presence of the dominant PSSM mutation. Because it is dominant, a horse having only one copy (heterozygous) is affected, and has a 50% chance of passing the PSSM allele to its offspring. This 'dominant' effect has major implications for breeders – there are no 'carriers' – a horse has got the condition, or it hasn't got it.

### **How the test results are presented**

The genetic DNA test verifies the presence of the dominant PSSM mutation and presents results as one of the following:

#### **P1/P1 Affected**

Positive for the dominant PSSM gene mutation, indicates the animal carries two inherited copies. Homozygous PSSM horses are genetically bound to pass the gene to 100% of their progeny when bred meaning all foals will have at least one copy of the dominant PSSM gene mutation.

#### **n/P1 Affected**

Both the normal and PSSM alleles were detected. Horse tested heterozygous for PSSM. The horse is affected with the PSSM genetic disorder and there is a 50% chance that this horse will pass a PSSM allele to its offspring.

### **n/n Clear**

Horse tested negative for PSSM and does not carry the PSSM gene mutation. The horse will not pass on the defective gene to its offspring.

### **Submitting a sample for testing**

Mane hair – pulled – with follicles, in a labelled Ziploc bag. The Society has a £5 discount. If you submit through the Society, it currently costs £25. The results will come back to the Society, and may be used by the Society.

You can submit your own samples to Animal Genetics UK in Cornwall for £30. We hope you will voluntarily release the results to the Society.

### **What New Forest ponies have been found to be affected?**

So far, two separate lines have been identified.

#### **Manor Boy of Howen n/P1**

Affected stallion sons – Blackwell Jonathan n/P1 (deceased), Blackwell Romany n/P1 (gelding).

Other sons – Whycome Pluto, Sprattsdown Del Boy (both deceased), Gatebridge Rag Time, Blackwell Flashman (both geldings, yet to be tested)

#### **Crabbswood Zanussi n/P1** (his sire Silverlea Mighty Don has tested n/n)

Palace Farmer – deceased due to accident summer 2017, before testing began.

Furzey Lodge Zennica – n/P1, de-licensed for breeding from 2018

Sons of Zennica:

Rushmoor Huntley n/P1, de-licensed for breeding from 2018

Lucky Lane Rollo n/n and Brookshill Mustang n/n are by Zennica but have tested clear and can continue to be used for breeding.

Hatchett Sunseeker, Blakeswater Winky (both geldings, yet to be tested)

We cannot exclude the possibility that further ‘lines’ carrying the PSSM gene will be found.

### **New Forest Stallions**

All New Forest stallions licensed for use in 2018 have tested n/n, so are free of the PSSM1-carrying gene, they cannot pass it on, and are safe to use for breeding. All colts coming forward for inspection from 2018 onwards will be tested for PSSM1 at the Society’s expense (to be reviewed after 5 years) and will only be licensed if they test clear.

### **Owners of female breeding stock from the affected lines**

Breeders will need to review the pedigrees of all mares and identify those that may be affected. Each breeder will have their own breeding plan with their own aims, goals and objectives, so each breeder will have to take their own individual decisions as to how to deal with possibly affected mares. The only way forward is through responsible testing and each breeder will have to decide on which of their mares to test, and when, according to when they are likely to breed next, and in accordance with your plans for that mare.

With a genetic test available, breeders can avoid breeding affected foals. Voluntary disclosure of a pony’s tested status, whether they are used for breeding, or being offered for sale, should become the norm, rather than the exception. All stallions used in the future will be PSSM-free. Through responsible testing and careful breeding, breeders can eliminate the possibility of any affected foal ever being born.

### **Summary**

There are hundreds of genetic conditions which occur in horses of all breeds, and New Forest is certainly not the only breed to have problems with inherited disorders. There is no doubt that more will be found in the future as new genetic tests become available.

It is very important to understand that this is nobody’s fault. Through science, and the available genetic test we can all learn to deal with the facts as they are today. The gene has been identified, the gene test is available, and this can be used as a tool by responsible breeders, so that affected foals are not produced.

**The mantra must be  
‘to prevent affected foals, test before you breed’**

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**NFPBCS, Deepslade House, Ringwood Road, Bransgore, Hants BH23 8AA  
Tel: 01425 672775 email: [info@newforestpony.com](mailto:info@newforestpony.com)  
[www.newforestpony.com](http://www.newforestpony.com)**