Dear all,

We would like to inform you of the following: The faculty of veterinarian medicine of the University of Utrecht has diagnosed two Dutch New Forest foals with myotony (literally: *muscle tension*).
This defect was not yet known amongst horses. Through DNA testing it was established that the parents of both foals carried an abnormal (i.e.'faulty') gene.
The origins of which cannot yet be fully determined. Further research is necessary. The University of Utrecht has informed the Dutch New Forest Pony Studbook of their findings on November 16, 2011 with a presentation to a delegation of the board, the studbook veterinarian and the relevant pony owners.

In consultation with the University of Utrecht, the board of the Dutch New Forest Pony Studbook society has taken the following action:

• to inform its members and the other New Forest Pony Studbooks through an interview with Mrs. I. Wijnberg, horse specialist internal medicine at the University of Utrecht, which is published in our magazine 'The Forester' (see Appendix for a translated version).
• If possible to investigate whether the gene is wider spread than the blood line that contains the current findings.
• If possible to find out whether the origin can be determined.

Based on the outcome of further research, we will take appropriate measures and if we have any news we will contact you as soon as possible. In the meantime, if you have any questions or concerns, please contact us via email at:

info@newforestpony.nl

Kind regards,

The Board of the Dutch New Forest Pony Studbook Society

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Appendix: interview with Mrs. I. Wijnberg (translated version). Quote from this interview is permitted only with reference to the source.

Interview with Inge Wijnberg Ph.D, Specialist in Internal Horse Diseases at the University Horse clinic, Faculty of Veterinary Medicine, University of Utrecht

Myotony diagnosed in New Forest ponies.

In 2009, a breeder noticed a very particular abnormality in one of his foals: when moving, the animal sometimes became completely rigid and after being chased in the meadow, the foal would fall over with stiffened legs. The breeder brought the foal in to be examined at the Faculty of Veterinary Medicine in Utrecht, where it was diagnosed with *myotony*.

Inge Wijnberg: "We first established that we were dealing with myotony. We knew that certain forms of myotony can be hereditary. That was why we were looking for other animals with comparable symptoms. And we ended up with goats. The pony's symptoms appeared very similar to a muscle ailment familiar in goats which is caused by a disruption in the transportation of chlorine. This defect was not yet known amongst horses for as far as we could establish, based on scientific literature."
A specialized research laboratory of the University of Bern (Switzerland) was then requested to perform hereditary testing. The question naturally is what were the results.
Inge: "In this testing it was established that a hereditary defect was indeed involved and that it was connected with this form of myotony. A change took place here. The altered information of a piece of DNA caused another protein than the normal one to be created. This also caused a change in function. The faulty protein (or maybe we should say the lack of the proper protein) leads to the clinical appearance of myotony. A similar defect in the DNA can develop in a spontaneous change, e.g. a change in hereditary material without finding any immediate cause."

In our studbook
The foal brought to Utrecht for the examination, was a New Forest foal. Once the diagnosis of *myotony* was known, DNA testing (as described above) could be carried out on the foal and its parents. This was possible thanks to the co-operation of the owner of the patient and a renowned breeder. It was further also possible to do a random survey amongst related NF ponies and non-related NF ponies, once again thanks to the co-operation of the breeder. It was thus established that it involved a form of congenital myotony.
Sometimes a foal receives the 'faulty' gene from both the stallion and the mare (homozygote). Thus the myotony symptoms become visible. A heterozygote variation was also found. Then the animal only receives it from one side and shows no symptoms, but it passes the "faulty" gene on to half of its descendants. Such an animal is called a "carrier" by geneticists or the people doing hereditary studies. These heterozygote ponies appeared to be present several times in the group of related animals. In other words: there were various "carriers" amongst the related animals. Further research must indicate whether, and to what extent, the mutation is distributed amongst the population.

A second patient has since been discovered. Both sufferers, patients in other words, were bred from one specific stallion line (Kantje's Ronaldo). Further hereditary / DNA research is necessary for more clarity: In the stallion itself, the parents of the stallion and if possible generations further back. Other stallion lines should also be investigated. The further back, the better the starting point can be established. Useful material could be blood, hair roots and sperm. With that material, a lot is technically possible. But: who can and wants to pay for that?

A forewarned man
Is it not far too early at this stage to disclose study information? Inge Wijnberg is quite clear: "The earlier the breeders know what is going on, the better. Better safe than sorry. This issue has significant consequences for animal welfare, and these foals are basically useless. That alone is already good enough reason why you can never start too early. I find the attitude of the relevant New Forest pony breeders extremely professional and I value their preparedness to co-operate very much. I know that this must after all be frustrating news for you as a breeder. It indicates the good intentions the breeders have for the breed and that they want to and dare to look further than just serving their own immediate interests."

Inge continues: "There are examples in other breeds of how a DNA change could quickly spread through a group of animals and cause a lot of problems. As is known, the stallion "Impressive" had a

major influence in the Quarter horse. Because of his particular muscle structure, he was very popular. Unfortunately he also passed on a muscle disorder to his descendants and that was a lot less pleasing. At the time when the problem was discovered, DNA research was still in its infancy, if not impossible. The genetic defect spread through the population at an enormous rate. Thus problems could still emerge today if this stallion appears in the blood line".
Maybe it is possible to prevent this in the mutation found now by means of a fast and adequate course of action.

Should the studbook have taken measures at an earlier point in time?
Inge: "No, obviously not. As long as you as a veterinary doctor or researcher have no signals that something is wrong, you cannot investigate anything. This was the first time that I was confronted by such a disorder. After all: the full DNA of the horse has only been known for a short time. It was published in the scientific journal *Science* in 2009. Therefore DNA research became relatively a lot simpler in recent years. Basically it is possible and also necessary from a veterinary perspective, to breed the defect out of the population by only breeding with "clean" parents".
For as far as it is known, there are no European regulations on hereditary disorders amongst horses and ponies. So how studbooks deal with it is mainly their responsibility and that of their members.

Gerti Nieuhoff

What is myotony?
With every movement the animal makes, muscles contract and relax. This appears as a flowing motion. Sometimes, muscles contract too much and they fail to relax immediately. There are diseases where there is a disruption in this balance of contacting and relaxing; *myotony* (literally: *muscle tension*) is an example of this.

Certain minerals are necessary for the functioning of a muscle, including potassium, lime (calcium) and chlorine. A disruption in the transportation of these minerals in the muscle can cause myotony. Hereditary muscle disorders occur in various forms, and amongst various animal species. With horses there is a form of myotony amongst for instance Quarter horses based on a disruption in the potassium system and with racing horses there is a muscle disorder known based on a defect in the calcium system. The disorder detected recently is found in the New Forest pony breed, and is also present in some dogs and goat breeds. Two forms are also known in humans: Becker's disease and Thomsen's disease. In this case it concerns an error in the chlorine transport route and the (clinical) effect is that an animal can easily become over stimulated.

With over stimulation, the muscles contract quickly and excessive, and they relax only slowly. During movement it seems for example that a leg becomes stiff, therefore the animal stops moving. It relaxes again gradually, after which it can start moving again. Sometimes the animal falls over completely because of stiffness. Another example: The pony starts whinnying but proceeds no further than a hoarse squeak because the muscles of the vocal chords are tightened excessively. After a while, the muscles relax and normal sound follows. A third example: When you push the eyelids of a patient aside (for example to view the mucous membranes) the eyeball retracts and you see the third eyelid. After some time it recovers because the muscles relax and the eyeball emerges again.

This article has been reviewed and approved by I. Wijnberg, Phd.