

Genetic Basis of Osteochondritis Dissecans Probed

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For a Thoroughbred the road to the racetrack is sometimes a rocky one, wrought with physical challenges such as the musculoskeletal condition osteochondrosis (OC). But recently, English researchers have taken a step forward in understanding a potential genetic component of OC.

"Osteochondrosis is a condition in which the articular cartilage lining the ends of joints does not form properly in young horses," explains Laura Corbin, PhD, who completed her work with colleagues at The Roslin Institute and Royal (Dick) School of Veterinary Studies at the University of Edinburgh, Scotland.

OC occurs most commonly in the stifle, fetlock, and hock, and clinical signs include joint swelling (effusion), lameness, and pain, making OC an important cause of horse wastage. Current estimates suggest that approximately 25% of Thoroughbreds are affected.

"Certain environmental factors such as nutrition, growth, body size, conformation, and endocrine (hormonal) (issues) are thought to play a role in the development of OC," noted Corbin. "There is also evidence that there is a genetic component to OC, but the exact cause of the condition is not known."

It is generally thought that many genes contribute to OC in horses, and several groups have attempted to identify such genes. The [decoding of the equine genome](#) in 2007 opened a gateway for genetic research, and Corbin refused to miss the boat.

She and a research team from The Animal Health Trust, in Newmarket, U.K., collected blood samples from 169 horses diagnosed with osteochondritis dissecans (OCD), a form of OC in which the diseased cartilage in the affected joint has separated from the underlying bone. They also collected samples from 179 horses without the disease for comparison.

"We extracted the DNA from the blood samples and produced a genetic profile for each horse based on around 40,000 genetic variants called single-nucleotide polymorphisms or SNPs (pronounced snips)," Corbin explained. More than 1.5 million such SNPs are scattered throughout the equine genome.

Researchers then compared the DNA sequences of horses suffering from OCD to those without the disease. Of the 40,000 tested SNPs, a single SNP located on equine chromosome 3 was significantly associated with the presence of OCD.

"Whilst the SNP itself is unlikely to be contributing to the disease, it may be acting as a marker for a nearby gene which *is* influential," explained Corbin. "If we could find such a gene, it would help us to better understand OC and ultimately reduce its incidence in the future."

The research groups' results need to be validated using an independent sample of horses, at which point fine-mapping the region identified on equine chromosome 3 could help identify the gene involved in OC.

Since publishing this work, Corbin has been investigating the potential for creating a "biobank" (sponsored by the British Equestrian Federation). Such a bank would store genetic information from a large number of horses with well-characterized conditions, like OC, for scientists to better conduct genetic research (<http://www.roslin.ed.ac.uk/john-woolliams/equine-biobank-project/>). In the future, advanced breeding methods using genomic data, such as those already used in dairy cattle, could be implemented to help reduce the incidence of diseases like OC.

The study, "A genome-wide association study of osteochondritis dissecans in the Thoroughbred," was published in *Mammalian Genome*. The abstract is available [online](#).

Disclaimer: Seek the advice of a qualified veterinarian before proceeding with any diagnosis, treatment, or therapy.