The equine Adam lived fairly recently: Close relationships among modern stallions

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In mammals, an individual's sex is determined by the chromosomes it inherits from its parents. Two X chromosomes lead to a female, whereas one X and one Y lead to a male. Y chromosomes are only passed from fathers to sons, so each Y chromosome represents the male genealogy of the animal in question. In contrast, mitochondria are passed on by mothers to all their offspring. This means that an analysis of the genetic material or DNA of mitochondria can give information on the female ancestry. For the modern horse, it is well known that mitochondrial DNA is extremely diverse and this has been interpreted to mean that many ancestral female horses have passed their DNA on to modern horse breeds.

Until recently, though, essentially no sequence diversity had been detected on the Y chromosome of the domestic horse. Not only does the lack of sequence markers on the Y chromosome make it impossible to trace male lineages with confidence, it also represents a scientific paradox. How can a species with so many female lines have so few male lines? The issue has now been addressed by Barbara Wallner and colleagues at the Institute of Animal Breeding and Genetics, University of Veterinary Medicine, Vienna (Vetmeduni Vienna).

Wallner initially selected seventeen horses from a range of European breeds. She pooled their DNAs and used modern sequencing technology to examine the level of diversity on a 200 kb portion of the Y chromosome she had previously sequenced. The Y chromosomes were found to be highly similar: only five positions turned out to be variable.

As Wallner says, "the results confirmed what we had previously suspected: that the Y chromosomes of modern breeds of horse show far less variability than those of other domestic animals."

The five variable positions, or polymorphisms, were nevertheless sufficient to enable the researchers to derive a type of "family tree" for the various breeds of modern horse they investigated. An examination of over 600 stallions from 58 (largely European) breeds showed that the animals could be grouped into six basic lines or haplotypes. The ancestral haplotype is distributed across almost all breeds and geographical regions. A second haplotype also occurs at high frequencies across a broad range of breeds, although not in northern European breeds or in horses from the Iberian Peninsula. A third haplotype is present in almost all English Thoroughbreds and in many warm-blooded breeds. The final three haplotypes are only found in local northern European breeds: one in Icelandic horses, one in Norwegian Fjord horses and one in Shetland ponies.

The pedigree of horses is very tightly controlled, with studbooks in many cases going as far back as the 18th century. Combining the results of the genetic analysis with pedigree data enabled the scientists to trace the paternal roots of many of the current male lines. Wallner feels that, "the results were intriguing, for example in the way the distribution of one haplotype reflects the widespread movement of stallions from the Middle East to Central and Western Europe in the past 200 years. Another haplotype results from a mutation that occurred in the famous English Thoroughbred stallion 'Eclipse' or in his son or grandson. It is amazing to see how much influence this line has had on modern sport horses: almost all English Thoroughbreds and nearly half the modern sport horse breeds carry the Eclipse haplotype."

The Vetmeduni Vienna scientists have confirmed the low diversity of the horse Y chromosome, which contrasts sharply with range of

mitochondrial DNA haplotypes observed in modern horses. The difference is presumably due to the strong variation in male reproductive success. Wild horses have a polygynous breeding pattern, while the intensive breeding practices in domestic horses mean that single stallions can effectively pass on their DNA to entire generations. The senior author on the paper, Gottfried Brem, comments that, "most modern breeds were established in the last two centuries, during which time the horse has undergone a transition from working and military use towards leisure and sports. This has largely been achieved through the use in breeding of a few selected males. The restricted genetic diversity of the modern horse Y chromosome is a reflection of what has survived the species' dynamic history."

More information: The paper "Identification of genetic variation on the horse Y chromosome and the tracing of male founder lineages in modern breeds" by Barbara Wallner, Claus Vogl, Priyank Shukla, Joerg P Burgstaller, Thomas Druml and Gottfried Brem has just been published online in *PLOS ONE*.
dx.plos.org/10.1371/journal.pone.0060015

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