XY sex reversal in a Quarter Horse mare
Brianne Simonsen, Bruce W. Christensen
Veterinary Clinical Sciences, College of Veterinary Medicine, Iowa State University, Ames, IA

Abnormalities of sexual differentiation in horses are commonly reported, including monosomy X and sex reversal cases.1-9 These manifest as irresolvable infertility issues with the animal having either ambiguous or female external genitalia and gonads that may not match the karyotype or are hypoplastic.2,3 A ten year-old Quarter Horse mare presented for breeding management. The client reported that the mare had never been bred and had erratic heat cycles. The external genitalia appeared normal. Transrectal palpation detected a cervix with low tone, small uterine horns and small, firm structures in the areas of the ovaries, which measured 1.2 by 1.6 cm and 1.3 by 1.8 cm on the left and right respectively, using transrectal ultrasonographic evaluation. An intersex condition with gonadal dysgenesis or a prior ovariectomy were considered as potential differential diagnoses. Scar tissue was not present on the flanks nor was it apparent through a vaginal speculum examination, discounting the likelihood of a prior flank laparotomy or colpotomy. Karyotyping revealed that nine cells had a chromosome count of 63,X and thirteen cells had a count of 64, XY. PCR testing with several Y chromosome markers confirmed the presence of a Y chromosome. The SRY gene PCR was negative. Other PCR tests for areas on the Y chromosome outside the SRY gene were positive. FISH was positive for a Y chromosome. Interphase nuclei X and Y probes revealed no XO cells, discounting the previous finding of 63, X cells. It was determined that the mare had a straight XY sex reversal with partial deletion of the SRY gene. The SRY gene initiates the genetic cascade resulting in testicular differentiation and male internal and external reproductive organs.1,10,11 Genes encoding for ovarian formation would be activated but an X dosage deficiency would prevent normal ovarian development and function.10

Keywords: Intersex, sex reversal, gonadal dysgenesis, genetic abnormality, infertility

References