Reproductive findings in two trisomy X (65,XXX) Thoroughbred mares  
M. R. Schnobrich, a E. A. Bradecamp, a T. L. Lear b  
Rood and Riddle Equine Hospital; Maxwell H. Gluck Equine Research Center, Lexington, KY 

This abstract describes trisomy X (65,XXX) in two four-year old Thoroughbred mares. While human individuals with trisomy X appear to be fertile, this is not the case in horses. Horses with X trisomy exhibit normal external genitalia but gonadal dysgenesis. The first mare was presented for breeding soundness evaluation following two years of infertility. The mare was bred unsuccessfully multiple times the first year. The second year the mare was bred when two follicles >30 mm developed on the left ovary. An ovulation inducing agent was administered and ovulation was detected within 48 hrs. The mare was administered a post-breeding lavage, and long-acting progesterone five days after ovulation but no pregnancy was detected 14 days later. Referred for a breeding soundness evaluation in mid-April, the mare’s external genitalia appeared normal. Trans-rectal ultrasound revealed small, hypoplastic ovaries (<2 cm of ovarian stroma). The left ovary had a 20 mm follicle, the right ovary had a 35 mm follicle, and the uterus had moderate endometrial edema with no intra-luminal fluid. On vaginoscopy the cervix appeared small, pale pink and elevated off of the vaginal floor. Digital examination of the cervix revealed a shortened canal (3 cm long). Hysteroscopic examination revealed no abnormal findings. Small volume uterine lavage was performed and no bacterial growth was recovered on aerobic culture and there was no evidence of inflammation following cytologic evaluation. A uterine biopsy (category IIA) revealed mild lymphocytic infiltration. Based on the presence of small dysplastic ovaries, a small cervix and the history of infertility, a karyotype was performed. The karyotype revealed that the mare had three copies of the X chromosome (65,XXX) and no further breeding was performed. The second mare was presented for routine evaluation following one unsuccessful breeding that season. Reportedly the mare developed a 30 mm follicle and ovulated following breeding but no pregnancy was detected. The mare was evaluated in December and was normal on physical examination. The mare’s external genitalia appeared normal and transrectal ultrasonography revealed small ovaries (1.5 cm x 2 cm) with no palpable ovulation fossae and no follicular development. The uterus was flaccid and small. Evaluation of the cervix revealed a shortened (2.5 cm long) cervical canal with no tone. Due to suspicion of a chromosomal abnormality a karyotype was performed and revealed the mare’s karyotype to be 65,XXX. These cases describe the general phenotypic and karyotype findings of two confirmed 65,XXX mares and may be helpful in identifying future cases. The incidence of 65,XXX karyotype in the mare is rare. X chromosome monosomy (63,X) is the most frequently reported sex chromosome abnormality reported in horses followed by 64,XY,SRY negative sex-reversal. Both 63,X and 64,XY,SRY negative cases present with normal external genitalia but an infantile internal reproductive tract and no follicular development. In these two 65,XXX mares there was follicular development, highlighting a phenotype to be aware of when suspecting chromosomal abnormalities in horses.

Keywords: Trisomy X, karyotype, reproductive dysfunction, reproductive pathology, infertility