



KARYOTYPING FAQ

Why karyotype a horse?

The genes of the horse are found in cell nuclei on linear structures called chromosomes. Every species has a characteristic number and array of chromosomes which are necessary for the precise control of development, metabolic function and reproduction. In the domestic horse, each parent contributes 32 chromosomes via sperm or egg to form a normal equine zygote with 64 chromosomes. Included in the array of chromosomes are those which determine sex; the mare contributes a chromosome called an X and the stallion either an X or a Y. Thus the normal karyotype for a mare is 64, XX and a stallion 64, XY.

Occasionally chromosomal material is lost or rearranged during the formation of gametes or during cell division of the early embryo. Usually such changes are so detrimental to normal development and the maintenance of life that either the resulting gametes fail to form a zygote or growth stops in early embryonic stages and the pregnancy is terminated by miscarriage or resorption. Rarely, if the chromosomal rearrangement or loss is relatively minor, the resulting embryo completes development, but subsequently may have abnormal patterns of growth, maturation and reproduction.

A karyotyping test for rarely encountered animals with chromosomal problems can in the long run save breeders time and money by identifying horses that are poor performance and breeding risks.

Primary infertility in mares

The most commonly encountered chromosomal abnormality in horses, XO Gonadal Dysgenesis, was first described 1975. Mares with this disease lack one of the pair of sex chromosomes resulting in a 63,X karyotype. The mare is presented as an infertile female, usually small in size for age and breed, with small, inactive ovaries. The condition is similar to a well-documented condition in human females known as Turner syndrome. The prognosis for fertility is extremely poor. The origin of the defect is probably an accident in development, not an inherited problem. The syndrome has been diagnosed in diverse horse breeds throughout the world including Thoroughbred, Arabian, Welsh Pony, Tennessee Walking Horse, Standardbred, American Saddlebred, Paso Fino, Belgian, Quarter Horse, Appaloosa, as well as in grade horses.

The second most common finding in infertile mares with inactive gonads is the karyotype of a male horse (64,XY)(XY Gonadal Dysgenesis, XY Sex Reversal, Testicular Feminization). Such mares are of normal or even large size and may have successful show and performance careers prior to being sent to stud. The problem may have a genetic origin. It has been encountered in Arabians, Thoroughbreds, Quarter Horses, Appaloosas, Morgans, Standardbreds and a pony of unknown pedigree. Fertility has been reported in XY mares, but is extremely infrequent.

Reduced fertility

Reduced fertility in humans, pigs and cattle has been associated with chromosomal rearrangements called balanced reciprocal translocations. Recently the first example has been described in a mare and it is anticipated that others will be identified as the power of karyotyping tests to diagnose the basis of certain kinds of infertility becomes more widely known.

Small, unthrifty foals

Extremely rarely encountered are young horses with abnormalities of the autosomes, not the sex chromosomes. The four cases described have been identified because of small size, unusual behavior and poor conformation, not for fertility problems. Each has had an extra chromosome, but the specific chromosome has been different, so the descriptions of the affected animals have varied. In a general way, these cases correspond to such human syndromes as Downs, associated with mental retardation and failure to thrive.

Blood samples used for tests

Karyotyping can be performed from peripheral blood lymphocytes isolated from a blood sample which has been sent to the laboratory by rapid courier. Lymphocytes are cultured for 3 days under conditions which stimulate them to divide, then harvested, stained and photographed. The procedure generally takes from 3-5 weeks to complete.

Banding techniques may be applied to the sample to confirm the diagnosis of a detected abnormality. Three types of banding (CBG, GTG or RBG) may be used to increase resolution, but will prolong the sample processing time.

References

Bowling, A.T., L. Millon & J.P. Hughes (1987) An update of chromosomal abnormalities in mares. J. Reprod. Fert., Suppl. 35: 149-155.

Horse GTG Idiogram (<https://www.vgl.ucdavis.edu/services/idiogram.php>)

Order Test

On occasion, staff at the Veterinary Genetics Laboratory will recommend Karyotyping for certain horses. In such cases, the owner will be contacted by Registry staff. Should you wish to order a test, please submit a Karyotyping Submission Form to the Veterinary Genetics Laboratory.