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A CLINICAL CYTOGENETICAL STUDY OF  
THE HORSE

A thesis presented in partial  
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ABSTRACT

Chromosome preparations were derived from short-term cultures of blood lymphocytes and from fibroblast-type cell cultures of subcutaneous fascia obtained from 44 clinically normal horses. The quality of mitotic cells compared favourably with similar studies in other species. Some characteristic morphological features were found which aid in the recognition of certain chromosomes, but the identification of many homologous pairs by conventional techniques was difficult. The normal karyotype of the horse ( $2n = 64$ ) was characterized following Giemsa staining and C-banding. The latter procedure was shown to be a valuable aid for the identification of specific chromosomes, in particular the sex chromosomes. After centromere staining, the X chromosome of the horse is characterized by the presence of prominent C-bands on its long arms, while the Y chromosome appears as an intensely-stained block.

Analysis of mitotic chromosomes prepared from ten phenotypically normal but infertile mares revealed, in one animal, the consistent lack of one sex chromosome in the four tissues studied. Centromere staining and sex chromatin studies confirmed that this mare had the karyotype  $63X0$ . Examination of the genitalia of this XO mare disclosed the presence of apparently normal reproductive organs except that macroscopic follicles were absent from both ovaries. Histologically, a number of presumptive atretic Graafian follicles and a discrete area of apparently functional luteal tissue were discovered in the right ovary. The significance of these findings are discussed and compared with

the features of the analogous syndromes associated with the XO anomaly in other species.

It is suggested that disorders of the sex chromosomes may occur commonly in horses and may contribute significantly towards the low fertility of the thoroughbred mare. Attention is drawn to the important practical applications of cytogenetic studies in the investigation of infertility in broodmares, and in the certification for sale of young horses intended for use as breeding stock.

Cytogenetic studies of spontaneous abortions in women have established that a large proportion of pregnancy wastage in humans is due to chromosome aberrations. The counterparts of many of the chromosome errors initially described in humans have now been reported among domestic animals. In particular, the XO anomaly seems remarkably common among infertile mares. These findings have indicated that chromosomal abnormalities may play a significant role in the aetiology of spontaneous abortion and embryonic loss in domestic animals. To investigate an hypothesis that chromosome anomalies are an important cause of prenatal loss in the mare, an attempt was made to analyze, cytogenetically, a series of equine abortions. From a range of tissues obtained from 26 aborted foals, cell cultures were prepared but failed to grow, and chromosome analysis was therefore not possible for any of these specimens. It seems that the equine foetus is retained in the uterus of the mare for a considerable period after the foetus has died and usually consists of partially-autolyzed non-vital tissues at the time of its expulsion.

Consequently, a study was made of the metaphase

chromosomes prepared from 22 equine embryos after their surgical removal from mares' uteri. A normal karyotype was found in each specimen. Although inconclusive on their own, the results of this study form an important contribution towards cumulative studies of embryonic loss in the horse. The current findings are discussed in the light of similar studies of induced abortions in women and embryonic loss in animals, and the potential for further investigation in this field is emphasized.

From the limited number of cytogenetic studies in domestic animals, it appears that chromosome analyses, particularly with the aid of more refined techniques such as centromere staining, may be of considerable value in investigations of infertility and embryonic loss in the horse.

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