

VETERINARY ASPECT OF SEX CHROMATIN RESEARCH IN VARIOUS
MAMMALIAN SPECIES - A REVIEW

Tierärztlicher Aspekt der Geschlechtschromatinforschung in
verschiedenen Säugetierarten - ein Übersicht

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INDIA

The remarkable advances in the field of sex chromatin studies in human beings in recent years have furnished new insights into the diagnosis of many human disorders associated with the errors of sex development due to abnormal sex chromosomal complements which impair the reproductive efficiency and mental development. The prenatal diagnosis of sex of an expected baby, an old problem of mankind has also been brought beyond guesswork and superstition into scientific reality by sex chromatin technique. Last two decades have witnessed a significant development in the sex chromatin studies of various mammalian species. However, in various laboratory, domestic and farm animals reports are sporadic and the available information is limited. This paper attempts to summarise the application of sex chromatin studies in economically important animals for sex chromosomal abnormalities, developmental anomalies of reproductive system and reproductive inefficiency alongwith the role of sex chromatin markers in foetal sex prediction.

Sex chromosomal abnormalities

Although sex chromatin as an indicator of sex chromosomal anomalies has been utilised to a limited extent in various economically important mammalian species, however, the studies made revealed its usefulness and importance in uncovering the sex chromosomal abnormalities. Rauluszkiewicz and Senze (1971) studied the behaviour of sex chromatin in cryptorchid dogs and found the amount of sex chromatin (drumsticks) to be similar to that found in bitches indicating an XXY chromosome constitution probably arising from nondisjunction. Sex chromatin and peripheral blood culture analysis of a 10 months old castrated male pig with lymphosarcoma revealed a 39 XXY/40 XXXY karyotype and the aberration was noted to be same as seen in sex chromatin positive Klinefelter's syndrome in human males (Harvey, 1948). A three year old horse revealed 68% of the cell nuclei containing two sex chromatin bodies per cell which suggested the presence of three X chromosomes in the animal. The karyotype analysis confirmed 62 autosomes, 3 X chromosomes and one Y chromosome. This 2A+XXY chromosome formula, together with the histological findings on gonadal tissue led to the conclusion that the horse represented the clinical expression of Klinefelter's syndrome (Gluhovachi et al., 1970).

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Intersexuality/Freemartinism

Intersexuality occurs about as frequently in some of the farm animals as it does in humans. However, in other domestic and laboratory animals intersexuality has been noted to be uncommon. Intersexuality in most laboratory animals is hardly known and sex chromatin studies have not been done undoubtedly due to unidentifiable sex chromatin in most of their somatic tissues. The reports on sex chromatin patterns of intersexes of domestic animals are also scanty. Schultz (1962) used the sex chromatin test in the diagnosis of pseudohermaphroditism in the dog.

Intersexuality occurs occasionally in various farm animals especially in swine where it is possible that some are undetected freemartins. Four of the Cantwell et al's (1958) male pseudohermaphrodite pigs had sex chromatin positive nuclei similar to normal females. One sex chromatin positive intersex appeared to be a case of gonadal dysgenesis and two others were classified by them as true hermaphrodites. Examination of nervous tissues of these intersexes revealed them to be genetic females. Several other workers have also determined the genetic sex of pig and swine intersexes by sex chromatin test in order to establish the mode of inheritance of the abnormal condition (Miyake, 1972 and Okamoto, 1976). Intersexuality occurs in goats with an average of about 5-10 percent. On the basis of the occurrence of sex chromatin in the nerve cells, Bislanska (1960) noted 9 of the 11 intersex goats to be genetic females and 2 were genetic males. Lüere and Strück (1960) concluded that three intersex goats examined by sex chromatin test were all females. Ferguson-Smith (1966) proposed a hypothesis which might explain the genetic femaleness of majority of intersexes. He postulated that one of the X-chromosomes possessed testis determinants which had been transferred to it by means of crossing over from a paternally derived Y-chromosome during meiosis i.e. the X-chromosome was in reality an XY. The random inactivation of the normal X or the X with the Y bearing genes would give rise to unequal local dominance of the trait-determining gene during development and could account for the genetic femaleness along with many of the anatomic findings in true intersexes.

Freemartinism in cattle has long been a classical example of naturally occurring intersexuality in a common animal species. A freemartin is usually a sterile female born as a co-twin to a male. Fehleimer et al (1963) in the cytogenetic study of freemartins, postulated that the condition might be caused by sex chromosome mosaicism (XX/XY). Harvey (1976) suggested that the basic factor which produced freemartinism was that in about 90 percent of twinnings there was chorionic vascular anastomosis with a resultant common circulation in the twin foetuses - a fact proved by the presence of both male and female populations of white cells after chromosome analysis and also a mixed population of red cells. Sex chromatin and chromosomal studies of 5 intersex sheep which were known to be either co-twin, triplet or quadruplet to male lambs also established that permanent white blood cell chimerism occurred in these sheep, which verified that they were freemartins (Bruere and Macnab, 1968). Though the proportion of XY cells in male and female twins of bovines could vary widely (Marcus, 1974), however,

the presence of even one XY cell in a female or an XX in the male co-twin could indicate that the female would be sterile or likely to be a poor breeder. (Eldridge and Blazak, 1977). Bhatia (1981) performed cytological sexing on the peripheral blood neutrophils of several bovine freemartins which failed to exhibit true female sex diagnostic drumstick appendages.

From the foregoing account on various types of intersexuality in several mammalian species it is imperative that the sex chromatin test might be used as a rapid, practical, cheap and convenient method for the early diagnosis of some of the intersexual conditions of the pseudohermaphrodites and freemartins soon after their birth.

Reproductive performance

During past few years much emphasis has been laid on the cytogenetic assessment of human's reproductive efficiency by examining the sex chromatin patterns in their blood smears. However, the cytogenetic reports elucidating the relation of fertility/infertility with sex chromatin attributes in various mammalian species are limited. Rauluszkiewicz *et al.* (1971) carried out the cytogenetic examination of 64 female and 21 male minks by sex chromatin test. The infertile females possessed too few drumsticks whereas, males with poor libido and poor semen quality tended to be sex chromatin negative. Sex chromatin analysis of mares which showed definite signs of anoestrus and gonadal dysgenesis also revealed significantly low frequencies of drumsticks than the normal females (Miyake *et al.*, 1979). Sixty six cattle having gross reproductive inefficiency due to repeat breeding, anoestrus and late maturity were observed to exhibit a significantly low frequency of drumstick appendages in their blood smears by Bhatia (1981). Similar observations were reported by Prusinowska and Maria (1979) in cows with a history of still births. A 75% Charolais and a Simmental heifer which failed to show oestrus were reported to possess sex chromatin negative nuclei by Chapman *et al.* (1970) and Catinelli *et al.* (1978) respectively. In the light of above observations, it is suggested that the association of the sex chromatin with the reproductive efficiency is required to be studied thoroughly to establish it as a norm for selective breeding in mammalian species of economic importance.

Foetal sex prediction

Preliminary cytological sex determinations by sex chromatin test have been carried out on foetal membranes of various mammalian species. Moller and Neimann-Sorensen (1957) for the first time, detected the genetic sex of the foetus in-utero by the presence/absence of the sex chromatin body in the cellular debris of amniotic fluid of cattle. Gluhovachi *et al.* (1970) diagnosed the foetal sex with 91 percent accuracy by studying the sex chromatin appendages in the amniotic fluid samples of 100 pregnant cows. Bhatia (1981) accurately predicted the sex of 9 fetuses using smears of foetal fluid cells by sex chromatin analysis. The early detection of the foetal sex in cattle might contribute significantly in the establishment of a dairy herd in a relatively shorter period and in the detection of some cytogenetic defects in the unborn, if any, in advance (Bhatia and Shanker, 1979). The antenatal diagnosis of foetal sex in bovines might thus prove helpful in formulating programmes aimed for the betterment of the livestock.

SUMMARY

A review dealing with the use of sex chromatin in the investigation of sex chromosomal abnormalities, freemartinism/intersexuality, reproductive performance and foetal sex diagnosis in various laboratory, domestic and farm animals.

ZUSAMMENFASSUNG

Ein Überblick über Verwendung von Geschlechtschromatin in den Untersuchungen über Geschlechtschromosomaldeformität, Zwitterrindheit/Intersexualität, Fortpflanzungsleistung und Fötusgeschlechtsdiagnose in verschiedenen Laboratorium, Haustiern - und Heftieren.

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