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Chromosome anomalies in domesticated animals and impaired fertility: A Review

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Abstract
The association between chromosomal abnormalities and reduced fertility in domestic animals is well-recorded and has been studied for decades. The viability of zygotes and embryos, meiosis, and gametogenesis are all directly impacted by chromosomal abnormalities. Balanced structural rearrangements can occasionally be passed on, resulting in issues with fertility in next generations. An essential stage in the identification of sterile or infertile animals is cytogenetic analysis. For more than 50 years, researchers have studied domestic animal chromosomes. These research results unequivocally show a connection between chromosomal abnormalities and decreased fertility. The discipline of cytogenetics has seen several stages of growth over the years. According to the number of discoveries and publications, the 1980s and 1990s were the height of animal clinical cytogenetics. During this time, chromosomal naming systems for the majority of domestic animals were created, and a large number of chromosome aberrations were documented. Accurate categorization of diseases of sex development (DSD) also depends on the examination of sex chromosomes. For a long time, chromosomal research mostly focused on cattle species, but more lately, there has been an increase in interest in such studies in companion animals. With the development of new molecular and cytogenetic tools and methods, chromosomal mutations may now be identified with greater accuracy. In addition to chromosomal banding, fluorescence in situ hybridization is one of them and is the industry standard. Recent developments in the cytogenetic diagnosis of cattle, pigs, horses, dogs, and cats are discussed in this review.

Keywords: Chromosomal abnormalities, infertility, disease of sex development, mutations

Introduction
Important concerns in animal breeding include the genetic basis of diseases of sex development (DSD) and decreased fertility (IF) coexisting with normal sex development. Since artificial insemination is a common method of reproduction in these species, it is crucial to identify the genes and chromosome mutations that are responsible. In several nations [Ducos *et al*., 2007] [7], a regular cytogenetic assessment of sires is carried out for such species. Since balanced chromosomal translocations or inversions can cause IF in phenotypically normal males and females, chromosomal studies are also advised in these cases. For DSD animals with ambiguous genitalia or aberrant sexual behaviours, the cytogenetic study is regarded as the initial diagnostic step.

Over 50 years ago, chromosome analyses of domesticated mammals were initiated. Ingemar Gustavsson, who discovered a central fusion (Robertsonian translocation) between chromosomes 1 and 29 in Swedish Red and White cattle, highlighted their significance in animal breeding [Gustavsson, 1969] [12]. Five years later, it was discovered that the carriers' fertility had been negatively impacted [Switonski M. 2014] [21]. Since this discovery, veterinary cytogenetics of livestock species has advanced incredibly quickly. With the exception of Holstein-Friesian cattle, it was discovered that 1/29 centric fusion occurs often in cow breeds [Long, 1991]. While reciprocal translocations predominated in pigs, X monosomy appeared to be the most frequent chromosome abnormality in horses [McGee and Lear, 2012)] [16].

Anomalies in the chromosomes

One of a species' defining characteristics is the presence of 2n diploid chromosomes. Because B chromosomes are present, some species (like the red fox and raccoon dog) have a variable number of chromosomes. Based on the arm ratio, which is the product of the lengths of the long and short arms of a chromosome, which is a quotient, chromosomes are categorised into four morphological types.

There are three primary kinds of chromosomal abnormalities, and each has a different impact. Early embryonic mortality is often caused by euploidies, autosomal aneuploidies, and significant imbalanced structural rearrangements. Sex chromosomal aneuploidies are an anomaly, as they often do not result in death but instead cause problems with sex development. Because unbalanced chromosome complements in embryos die, balanced chromosome rearrangements reduce fertility.

To identify sex development problems, the sex chromosome complement must be examined. The identification of the X and Y chromosomes in cattle and dogs is made easier with the use of traditional Giemsa-stained chromosomes.

In some, however, it is a difficult task. The morphology of both sex chromosomes in cats and horses resembles some autosomes, thus banding or FISH methods must be used to identify them. Only the Y chromosome in pigs can be distinguished with ease.

Classification of sex development diseases

Mammals' sex is determined via a process that involves hundreds of genes, sex chromosomes, and other factors [Pasterski., 2010] [17]. Few of these genes, such as SRY (sex determining region Y), which causes male gonad differentiation and is located on the Y chromosome, and AR (androgen receptor), which is involved in the development of Wolffian ducts and external male genitalia and has its locus on the X chromosome, are located on the sex chromosomes. The majority of these genes are located on autosomes. Interesting facts about the location of key ovarian differentiation genes on autosomes include the presence of RSPO1 (R-spondin 1), CTTNB1 (catenin beta 1), WNT-4 (wingless-type mouse mammary tumour virus integration site family, member 4) and FOXL2.

The sex chromosome DSD includes the lymphocyte chimerism (XX/XY) seen in freemartins, structural rearrangements involving the sex chromosomes, and sex chromosome aneuploidies. A significant anomaly in XX DSD animals is the existence of testicles or ovotestes, as well as the uterus with oviducts and duct deferens despite the absence of the SRY gene. This aberration has been identified in numerous species (goat, pig, horse, and dog). The XY DSD include both monogenic anomalies, such as androgen insensitivity syndrome (AIS) and persistent Mullerian duct syndrome (PMDS), as well as disorders whose aetiology is obscure or complicated, such as cryptorchidism and hypospadias.

In general, sex chromosome abnormalities are more frequent and have milder effect on viability than autosomal rearrangements. Phenotypes associated with sex chromosomal abnormalities are diverse and primarily involve various fertility issues and DSDs. In many cases, phenotypic changes range from limited to non-evident. However, sex chromosome abnormalities vary in the degree of prevalence between species, as well as in the severity of phenotypic consequences. Sex chromosome sequencing and functional studies are helping to clarify the underlying reasons for some of these differences. In the following paragraphs, sex chromosome abnormalities in domestic animals are discussed in light of these recent developments.

DSD sex chromosome

Following a clinical investigation, cytogenetic analysis aimed at identifying sex chromosomes should be the first step in the diagnosis of DSD. The diagnosis of DSDs brought on by chromosomal anomalies, such as the XX/XY chimerism found in freemartins or sex chromosome aneuploidies (X monosomy and trisomies: XXY, XXY, and XXX), is made easier by this technique. Sex chromosomal disorders (DSDs) are known to frequently result in infertility or sterility [Favetta, 2012] [22], but it should be noted that some of them are also linked to an increased risk of gonad malignancy [Krumrych].

Asexual sex chromosomes

X monosomy and XXY trisomy have the greatest detrimental impact on sex development, which can result in sterility. Across species, the prevalence of sex chromosome aneuploidies varies. The X monosomy is more prevalent in mares than in dogs and cats, highly uncommon in cattle, and very uncommon in pigs. In contrast, diagnosis of the XXY trisomy is fairly uncommon, with the exception of male tortoiseshell cats. Inadvertently, two more sex chromosomal aneuploidies (XXX and XYY) were reported. Because only males are cytogenetically analysed and their numbers vary greatly between species (very large in cattle, large in pigs, and rather small in dogs and cats), these approximations should be used with caution. Reduced fertility in balanced chromosomal aberration carriers.

Inheritable mutations called balanced structural rearrangements (central fusions, tandem fusions, reciprocal translocations, and inversions) impact carriers' fertility without obviously changing sex development. Reciprocal translocations, including sterility in males with the X-autosome or Y-autosome rearrangements, are linked to the most significant decline in fertility. In contrast, carriers of other balanced aberrations (central fusions, tandem fusions, and inversions) only experience a modest reduction in fertility.

Robertsonian translocations (central fusions)

Due to the finding of a widespread distribution of the 1/29 centric fusion in cattle, centric fusions are the chromosomal abnormalities in domestic mammals that have received the greatest research attention. Recent research using cutting-edge molecular methods has demonstrated that the 1/29 mutation is a complicated rearrangement including deletion, inversion, and centric fusion rather than a straightforward centric fusion.

Numerous cattle breeds have the 1/29 Robertsonian translocation, but Holstein-Friesian cattle, the most significant dairy breed, don't appear to have it. Its prevalence reaches 10% in certain beef breeds [Van Haeringen, 2014], whereas it is more common than 50% in some local breeds, such as those in Portugal. Additionally, various centric fusions involving different autosomes have been reported in cattle. Centric fusions have only very infrequently been observed in sheep, goats, pigs, and dogs; there are no similar reports for horses or cats.

Mutually beneficial relocations

Pigs most frequently have reciprocal translocations when their chromosomes are reshuffle. In 7,700 young boars that were

cytogenetically evaluated in France, it was estimated that these rearrangements were present on average at a frequency of 0.47% [Ducos, 2007] [24]. By 2012, this species had approximately 130 distinct translocations in total, and more examples have emerged since.

It was uncommon to find reciprocal translocations in other domesticated animals. Three translocations, including two reciprocal ones, were found between the autosomes of mares with early embryonic loss that occurred more than once: (1;21), t (16;22), and t (4;13). Szczerbal *et al.*, described an intriguing case in which a female cat (38, XX) with a disturbance of sex development, characterised by the existence of a rudimentary penis, was identified for the first time in domestic animals with a translocation between the X and Y chromosomes, t(X; Y) (p22;p12).

Various additional balanced chromosome rearrangements

Although chromosome inversions in cattle and pigs have been identified, little is known about how they affect the carriers' fertility. One inversion scenario, in particular, is of particular relevance. Inv (4) (p1.4; q2.3), a pericentric inversion of the pig chromosome 4, was found in multiple AI boars in France. Analysis of the male and female carriers' sperms and oocytes revealed that around 4% of them had imbalanced gametes.

Conclusion

Heritable, balanced chromosomal abnormalities are the primary cause of decreased fertility but do not appear as obvious phenotypic effects, according to a long history of cytogenetic studies of domestic animals, primarily on males used in artificial insemination. As a result, cytogenetically evaluating young guys who are potential candidates for use in artificial insemination stations is prevalent. It is significant to note that cytogenetic analysis is the initial step in the categorization of abnormalities of sex development. Ultimately, the utilisation of chromosomal studies is vital in the hunt for the reasons of infertility in both people and animals with aberrant sex development.

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