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Structural chromosomal mosaicism due to partial monosomy (3q-) in a Murrah buffalo (*Bubalus Bubalis*) bull

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Abstract
The coexistence of two or more genetically distinct cell populations derived from a single zygote is mosaicism which may be because of numerical or structural alteration in autosomes or sex chromosomes or both at any stage of development, from two-cell stage onward. Present study reveal a case of mosaicism due to structural alteration in one of the chromosome number 3. Peripheral blood was collected from a phenotypically normal and fertile Murrah buffalo bull aged between 40 to 42 months for routine cytogenetic investigation. A conventional whole blood culture was set up for chromosomal preparation. The Giemsa stained and GTG banding exhibited one field normal with 50, XY chromosomal complement out of 50 fields whereas other fields exhibited partial monosomy (3q-) due to terminal deletion in chromosome number 3. Bull was appeared to be normal with normal libido and normal semen quality. The bull is under collection. However, it is advisable that the bull having mosaicism should be checked for the fertility before use in artificial Insemination and breeding programmes.

Keywords: Mosaicism, zygote, chromosome, monosomy, culture, GTG banding

1. Introduction
Mosaicism occurs due to mitotic non-disjunction during development. The coexistence of two or more genetically distinct cell populations derived originally from a single zygote is known as mosaicism. Mosaicism may arise at any stage of development, from the two-cell stage onward, or in any tissue which actively proliferates thereafter. Mosaicism could be developed due to sex chromosomes or autosomes and both. The phenomenon is commonly observed in many species of animals and plants, but the physiological effects of mosaicism are largely unknown. Most mosaic alterations remain undetectable with current analytical approaches, although the presence of such alterations is increasingly implicated as causative for disease. In humans, when chromosomal mosaicism arises during development, pregnancy outcome depends on which tissue, and how much of that tissue is abnormal. If a majority of the cells are abnormal then human development is likely to be abnormal. If only a small fraction of some cells are involved, it may likely to have little effect on growth and development. Abnormalities in chromosome structure generally have no phenotypic expression but are very often associated with reproductive disorders [1]. However, the association between chromosomal abnormalities and reduced fertility in domestic animals is well recorded and has been studied for decades [2]. Chromosomal aberrations may occur due to numerical or structural rearrangements usually without causing phenotypic abnormalities on carrier animals [3]. The magnitude and significance of occurrence of chromosome anomalies are also little understood. Although reduced fertility occurs mostly due to a combination of several factors, in some instances, it can be traced to a single genetic factor. For example, with regard to chromosome anomalies, bulls with Y-autosome reciprocal translocations [4], sex chromosome mosaicism in Holstein cow, unusual XX translocaion in buffalo, chimerism in Holstein bull etc., [5, 6, 7, 8] show an apparent normal phenotype but found with oligozoospermia or azoospermia in male or failure to oogenesis in female. Various chromosomal abnormalities have been reported in India, which varies from no effect or minor to major effects mainly on fertility [9]. The present studies exhibit interesting case of mosaicism owing to imbalance of chromosome No. 3 (3q-) in a fertile Murrah bull.

2. Materials and Methods
Peripheral blood was collected from a phenotypically normal and fertile Murrah buffalo bull aged between 40 to 42 months. Briefly, whole blood was cultured in RPMI-1640 (Himedia) medium supplemented with antibiotics, 15% fetal calf serum and 1% pokeweed mitogen17. The whole blood cultures were incubated at 37°C for 69 hours. To increase the relative frequency of prometaphase chromosomes, ethidium bromide (Sigma) @10 µg/ml was added to the culture, 2 hrs prior to harvesting. Similarly, to arrest somatic cell division at metaphase stage, Colchicine (Sigma) @ 2 µg/ml was added at 1 h prior to harvesting. The cells were separated by centrifugation at 1200 rpm for 5 minutes followed by hypotonic treatment with 0.075 M KCl for 30 minutes at 37°C and fixed in 3:1 ratio of methanol and acetic acid glacial. Finally, cell suspension was dropped on slides and air dried. Conventionally staining by Giemsa and routine GTG banding with little modification were performed on chromosome slides [10]. Around 30 Giemsa stained and G-banded metaphase plates were screened per animal for chromosomal analysis using Olympus microscope attached to image Analyzer system (CytoVision).

3. Results and Discussion
Out of 50 metaphase fields, one field exhibited normal 50, XY chromosomal complement (fig-1 & 2), whereas other fields exhibited partial monosomy (3q-) due to terminal

deletion in chromosome number 3 (fig-3).

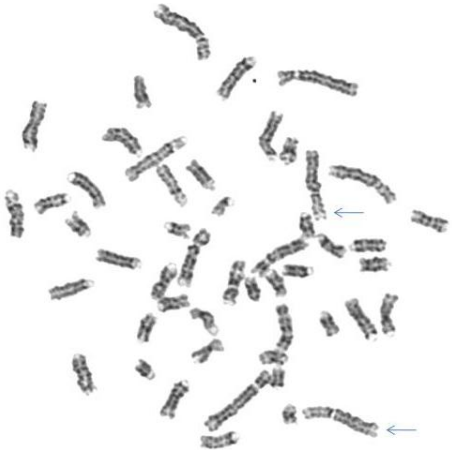

Fig 1: Metaphase plate showing normal chromosomes. Arrows indicate normal length of chromosome pair 3

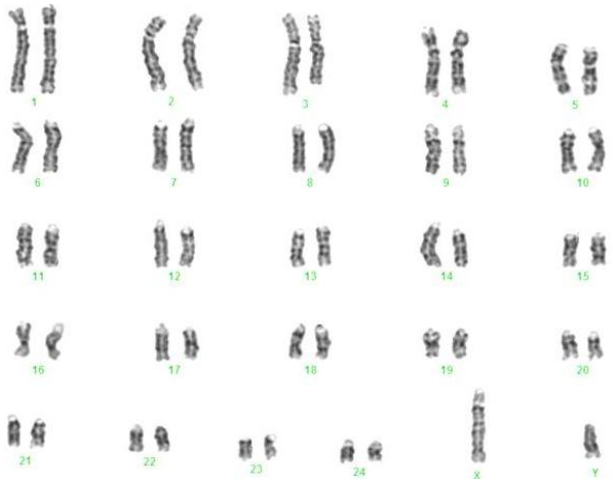

Fig 2: karyotype of metaphase plate shown in figure 1

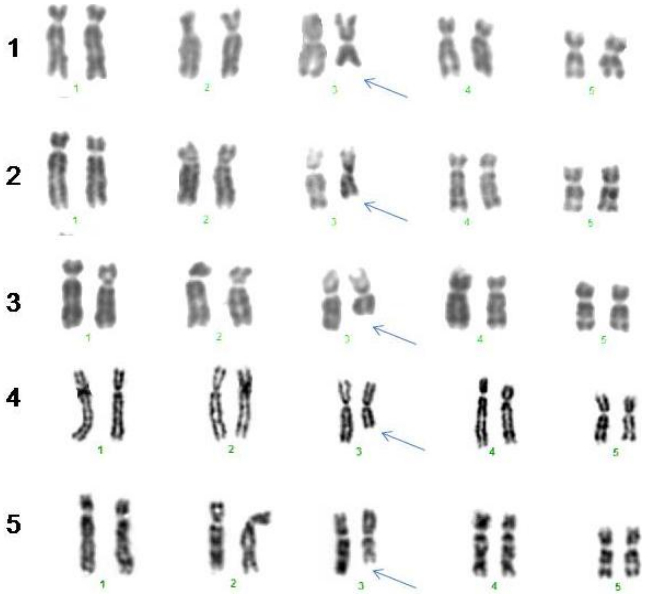

Fig 3: Five lines of the chromosoems reveal first five pairs of chromosomes taken from different metaphase plates of Murrah buffalo bulls. Arrows indicate variation in the length of one of the chromosome pair 3

Bull was appeared to be normal with normal libido and normal semen quality. The bull is under collection. Abnormalities in chromosome structure generally have no phenotypic expression but are very often associated with reproductive disorders [1]. Numerical mosaicism is very common with effect on fertility [5, 11, 12] or without effect on fertility as animals could not reach puberty at the time of cytogenetic investigation [13, 14]. Mosaicism owing to structural chromosomal aberration is rare aberration [15, 16, 10]. Mosaicism may be due to any reasons, in dairy animals is with varying degree of their impact on fertility. However, many reports on human chromosomes support the theory of structural mosaicism. Shackelford *et al*. [17] presented a rare case of mosaicism for structural abnormalities in chromosome 12 in a six month old patient with phenotypic features of Pallister Killian Syndrome. Chromosome analysis of cultured peripheral blood lymphocytes revealed an additional ring chromosome in 13% of the metaphases. Both parents had a normal karyotype, demonstrating the de novo origin of this ring chromosome [18]. The mosaicism for the 15q13.3 deletion explained the milder phenotype observed in two monozygotic twins [19] etc. Classical G-band analysis is limited in detecting structural mosaicism in general. Recent advances in whole genome microarrays have improved the detection of mosaicism even at low level in human. Similarly, bovine microarray chips are being developed to detect microdeletion, duplication, etc.

4. Conclusion

However, it is advisable that the bull having mosaicism should be checked for the fertility before use in artificial Insemination and breeding programmes.

5. References

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