

Characterisation of sex chromosome aneuploidy in female cattle using genotype information

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Background

Aneuploidy is a genetic condition characterized by the loss (monosomy) or gain (trisomy) of one or more chromosomes. Aneuploidy affecting the sex chromosomes often leads to infertility. The objective of this study was to estimate the prevalence of sex chromosome aneuploidy in a large juvenile population using routinely available genotype intensity information.

Methods

Genotype and genotype intensity data was available on 145,476 female dairy and beef cattle below 15 months of age at the time of genotyping, with no recorded progeny. Genotype intensity data included the LogR Ratio (LRR), R-value (the sum of X and Y SNP probe intensities), and B-allele frequency (BAF) measurements. For the present study only Single Nucleotide Polymorphisms (SNPs) on the sex chromosome were analysed which included 9 Y-chromosome and 259-X chromosome SNPs. To determine aneuploidy, the mean X-chromosome LRR relative to the autosomal mean LRR for each female was calculated, as was the mean X-chromosome R-value relative to the autosomal mean R-value. Additionally, the number of SNPs falling within the expected heterozygous BAF range (0.45-0.55) on the X-chromosome was determined and the number of genotype calls on the Y-chromosome. The likelihood of an animal's LRR and R-value statistics was estimated using the probability density function, while the cumulative density function was utilized to estimate the likelihood of a female having a specific number of heterozygous BAF calls and Y-chromosome SNPs. Cytogenetic analysis will be completed on suspect cases for validation.

Results

This is the first study to estimate the frequency of X chromosome abnormalities in a large cattle population, and although not prevalent, preliminary findings suggest that X-chromosome monosomy and trisomy occurred at rates of 0.10% and 0.05%, respectively.

Additionally, Klinefelter syndrome (XXY) was estimated to occur in approximately 0.11% of females. Notably, none of these animals, despite breeding attempts, were successful in producing offspring.

Conclusion

Aneuploidy can be readily detected using genotype information at birth, aiding producers in the swift exclusion of these animals from breeding endeavors at an early stage.