Gruppo di Citogenetica

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Mix of two chromosomal aberrations in a newborn calf 2n=60,XX, t(11;25)(q11;q14-21)

A. Iannuzzi[1], A. Perucatti[1], A. Pauciullo[1], V. Genualdo[1], D. Incarnato[1], L. Pucciarelli[1], L. De Lorenzi[2], G. Varricchio[3], D. Matassino[3] , P. Parma[2], L. Iannuzzi[1] (alessandra.iannuzzi@cnr.it)

[1]Laboratory of Animal Cytogenetics and Gene Mapping, National Research Council (CNR), ISPAAM, Naples, Italy; [2]Dept. of Animal Science, Agricultural Faculty of Sciences, Milan, Italy; [3]ConSDABI, Sub-national Focal Point of FAO (Mediterranean Biodiversity), Benevento, Italy.

A newborn calf of the Agerolese breed underwent cytogenetic investigation because presented hyperflexion forelimbs, red eyes and inability to stand up. Anamnesis revealed the mother, phenotypically normal, was carrier of a t(11;25)(q11,q14-21). The newborn died after a few weeks and no internal alterations were found by veterinarian after the post mortem examination. The mother presented, after a cytogenetic investigation, a reciprocal translocation between chromosome 11 and 25 and the presence of two ders: der11 and der25, for the position of corresponding centromere. On the other hand, the veal revealed a different chromosomal aberration in comparison to her mother. In fact, after R- banded karyotype, the calf showed both chromosomes 25, one chromosome 11 and one der (der25). FISH analysis was performed with the same BAC clones used to detect the translocation in the mother: BAC142G06 mapped on the proximal region of both BTA25 and der25; BAC513H08 mapped to BTA 25q22dist; BAC533C11 mapped to the proximal region of BTA11 and der25. Finally, we confirmed both the localization of the breakpoints on band q11 (centromere) of chromosome 11 and q14–21 of chromosome 25, and the loss of the der11. In this way, it is showed a different cytogenetic aberration in the veal: a partial trisomy of chromosome 25 and a partial monosomy of chromosome 11. We have been studying a correlation between this aberration and some gene involved comparing it with corresponding human clinical cases.
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