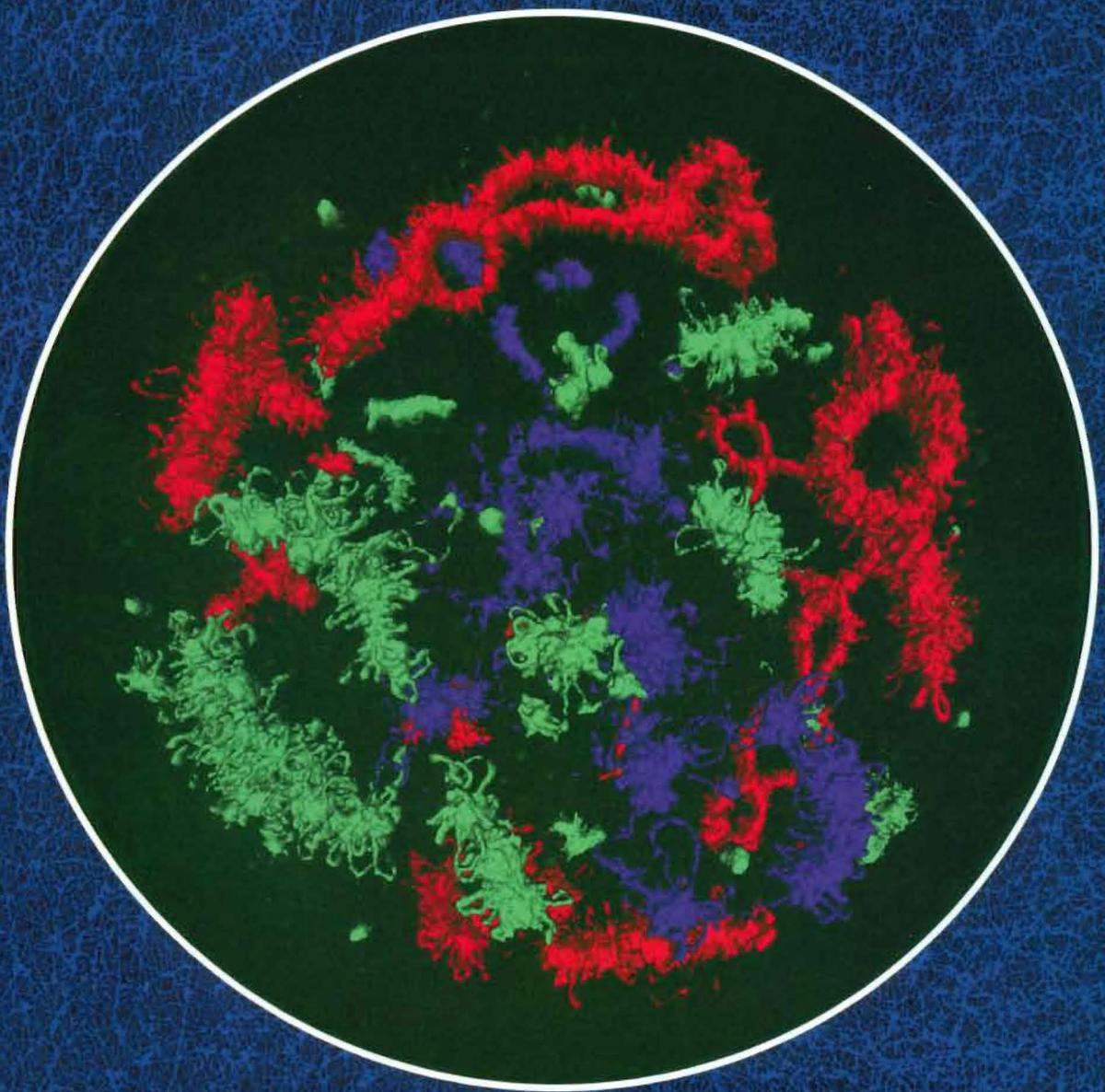


# CHROMOSOME RESEARCH

*The Biology of Chromatin and Chromosomes*



**A Tribute to Simon W.L. Chan, PhD (1974–2012)** 657

**Visualization of the spatial positioning of the *SNRPN*, *UBE3A*, and *GABRB3* genes in the normal human nucleus by three-color 3D fluorescence in situ hybridization**

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## ABSTRACTS

**20th International colloquium on animal cytogenetics and gene mapping** 779

## 20th International colloquium on animal cytogenetics and gene mapping

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Córdoba, Spain

### *Clinical Cytogenetics*

#### L1

##### **Clinical cytogenetics in domestic bovids: an update Iannuzzi L.**

Institute of Animal Production Systems in Mediterranean Environments (ISPAAM), National Research Council (CNR) of Italy, Laboratory of Animal Cytogenetics and Gene Mapping, Naples, Italy

The discovery of rob(1;29) in cattle and proof of its deleterious effects on carriers has opened the door to clinical cytogenetics applied to domestic animals, especially cattle and pigs. This important sector started by using only conventional chromosome staining, allowing the discovery of only numerical chromosome abnormalities and structural chromosome abnormalities which change the chromosome shape (i.e., centric fusions and pericentric inversions, as well as reciprocal translocations with evident derivative chromosomes). The advent of banding techniques and the availability of standard karyotypes for all domestic species have accelerated the progress made by clinical cytogenetics, allowing more precise identification of chromosomes involved in chromosome abnormalities. A further important step was made when FISH-mapping analyses were applied by using both chromosome painting and specific molecular probes (in particular BAC-clones). Indeed, both chromosomes and chromosome regions (gene order) involved in such chromosome

abnormalities were identified without any doubt, as occurred earlier when chromosome identification was uncertain, especially when poor chromosome banding techniques were used. Unfortunately, few labs are actually involved in clinical cytogenetics of domestic animals, probably due to the difficulties organizing this type of work at the national or regional level. Indeed, only sound collaboration among cytogenetic labs, breeders, breeder associations and veterinary practitioners can ensure the development of this important sector. In particular, all male breeders and females with reproductive problems (females which do not show any oestrus at fertile age or those showing reduced fertility due to delays in returning to oestrus or large inter-birth intervals) should be investigated with banded karyotypes. Indeed, most cytogenetic analyses are still performed using only conventional staining techniques for the most widely investigated species, namely cattle. In the present paper the most important chromosome abnormalities discovered in domestic bovids (i.e. cattle, river buffalo and sheep) and their effects on fertility are summarized and discussed.

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#### O1

##### **Molecular and cytogenetic characterization of an Xp-chromosome deletion in cattle De Lorenzi L.<sup>1</sup>, Rossi E.<sup>2</sup>, Genuardo V.<sup>3</sup>, Gimelli S.<sup>4</sup>, Lasagna E.<sup>5</sup>, Perucatti A.<sup>3</sup>, Iannuzzi L.<sup>3</sup>, Parma P.<sup>1</sup>**