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The 3D Nuclear Organization of the Telomeres and Genomic Instability

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One of the main cause of genomic instability and cellular apoptosis is telomere dysfunction. Telomeres are specialized nucleo-protein complexes at the ends of chromosomes, which are composed of long tracts of double stranded TTAGGG repeats, bounded to highly repetitive DNA sequences. Besides its role for the maintenance of genomic stability, telomeres also provide a mechanism for "counting" cell divisions, and thus signal for replicative senescence. In this scenario, an important enzyme, telomerase, and several telomere binding factors with structural and regulatory action, participate in the control of telomere length and capping. The proper telomere capping preserves chromosomal integrity and prevents genomic instability.

With the advent of DNA sequence probes, the use of fluorescence microscopy applied to clinical and basic research has dramatically increased our knowledge about the genome organization, and has enable us to detect almost any genomic region inside the nucleus with high sensitivity. In this way, the three-dimensional (3D) nuclear organization of telomeres has permited the distinction between normal and tumor cells: nuclei of the latter tend to be disorganized with the presence of telomeric aggregate. Thus, defining the structural organization of the interfase nucleus is important to our understanding of the 3D genome organization in the interphase nucleus. This approach can be performed by fluorescence in situ hybridization (FISH), in combinatiom with deconvolution analysis. The parameters measured include: the number of telomeres, sizes of telomeres, nuclear distribution of telomeres, and the presence of telomeric aggregates.

Currently, the use of 3D telomere profiling analysis has enable the identification of telomere changes during tumor initiation and progression of distinct types of neoplasias. Moreover, this approach has permitted, the identification of cellular subpopulations correlated with diferent times of tumor progression. Using the same 3D imaging strategy, it has provided evidence that genomic instability is a result of these nuclear changes and these changes can be monitored during cancer progression, in patients samples.

Regarding specific genetic diseases, telomere loss has been also observed in Down syndrome, Fanconi anemia, Ataxia telangiectasia, Nijmegen breakage syndrome, Werner syndrome, Dyskeratosis congenita and Bloom syndrome. This fact could provide the signal for cells to enter in senescence, leading to the formation genome rearrangements and chromosomal abnormalities. In conclusion, 3D telomere profiling represents an important tool able to measure the degrre of genomic instability, and subsequentelly may help to define an specific diagnostic strategy.

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