

Genomic instability: the players and the rules of a risky game

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The human current genomic architectural configuration and functionality is the sum of the most diverse sequential molecular events occurred during evolution. The events with capability for shaping genomes are based on structural and quantitative chromosomal alterations of variable dimensions, from very small to large regions that may completely change the genome structure and the cellular function. Amongst these, chromosome fusions, fissions, inversions and insertions are, perhaps, the ones with a stronger impact. Variation may occur in many different genome fields, but there are some regions that seem to be hotspots to instability. The result of such events can be then transmitted either as neutral, associated with a selective advantage that will eventually conduct to adaptation and speciation or as a potential harmful variation if occurred in fundamental cells where the outcome can be devastating. Genomic instability can thus be viewed as the strength to promote evolution but also the fragility to cause disease. Among the players of this risky game, repetitive elements such as satellite DNA sequences (the Satellitome) and transposable elements (the Mobilome), stand out as fine candidates for genomic variation and instability due to their dynamics that can impact genome architecture, genes' structure and regulation. Often considered a part of the dark matter of genomes, these sequences are now starting to be deciphered and, as players, can also be the key to understand genome adaptation, as well as normal and abnormal cellular processes, a step towards specific diseases management and even global wellness improvement.