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Fragile regions of the human genome: Possible origins and implications for complex diseases

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Chromosomal fragile regions (rearrangement hotspots) are loci or regions susceptible to spontaneous or induced occurrence of gaps, breaks and rearrangements. In the human genome, more than 100 different fragile sites have been described with a potential role in human disease, including mental retardation, neurodegeneration, cancer, chromosome truncation syndromes and reproduction. Although pathogenic deletions and duplications have been described, their origins and molecular consequences remain obscure. Recent results have demonstrated the high frequency of micro inversions (<1 kb) in human evolution, and its effect on regulation of gene expression. Chromosomal evolution involves multiple changes in structural and numerical levels and these changes, which are related to the variation of the gene number and their location, can be tracked by the identification of syntenic blocks (SB). Some studies have identified evolutionary breakpoint regions (EBRs) and fragile sites at specific locations in the human X chromosome and other chromosomes. However, mapping these regions to date has involved using low-to-moderate resolution techniques. Such scenario might be related to underestimating their total number and giving an inaccurate location. Using different algorithms is possible to identify hotspot regions in the human genome. Recent research has found that human hotspot regions were enriched by repeated elements (LINE-1 and Alu), segmental duplications or gene duplications, high GC content, and CpG island density which may have led to genome rearrangement events. There are chromosomal deletions and duplications databases associated with several human diseases as cancer and other, supporting the current line of evidence and suggesting that a common mechanism can underlie the generation of constitutional and somatic rearrangements. After comparing these data with the new fragile regions postulated, the great overlap in their breakpoints which corroborates this hypothesis. Identifying new EBRs would thus contribute toward understanding chromosome evolution and breakpoint reuse-related processes, as well as identifying potential predisposing factors concerning complex human disease.

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