Abstract

Chromoanagenesis is a catch-all term of recently described catastrophic events that generate complex karyotypes. These events are divided according to the characteristic features and are termed chromothripsis, chromoplexis and chromoanasynthesis. Chromothripsis represents a disintegration of chromosomes or their parts into hundreds of small fragments. Those chromosome fragments are then incorrectly reassembled. Chromoplexis rearrangements are not very different from chromothripsis rearrangements. The main difference is a lower number of breakpoints and the distribution of aberrations in the whole genome. The erroneous replication processes occur during chromoanasynthesis.

There are several mechanisms responsible for breakdowns of a DNA molecule. In the case of chromothripsis, micronucleus formation is probably the most important mechanism. During chromoplexis, transcriptional stress plays a major role. Replication stress is associated with chromoanasynthesis rearrangements. The result of all these processes are highly rearranged chromosomes with numerous losses or gains of genetic material.

This work summarizes the current knowledge of the mechanisms that are mentioned above and the genesis of complex aberrations. At the same time, it represents the connection between complex karyotype and clonal cells development. Cytogenomic methods that are used to detect chromosomal abnormalities including their characteristics are also stated.