Lecture 9.
Types of structural chromosomal abnormalities

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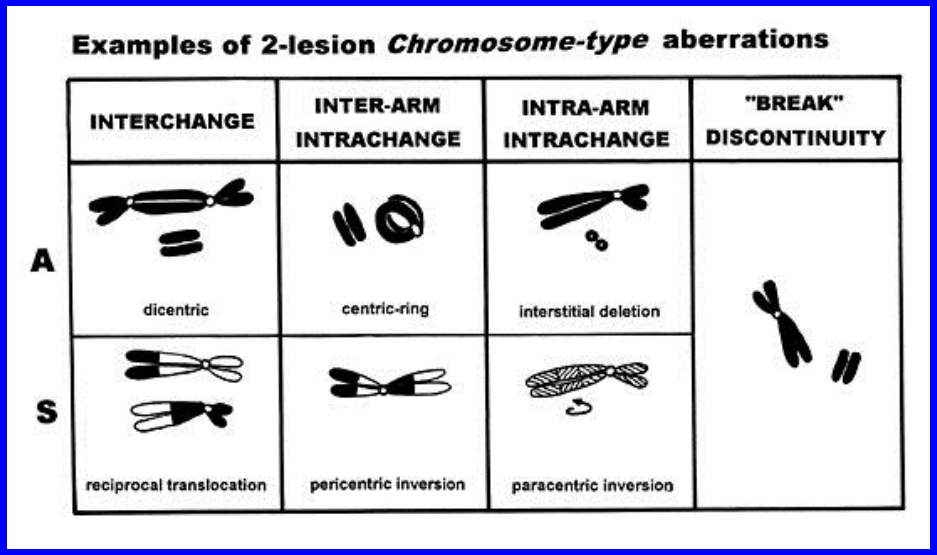
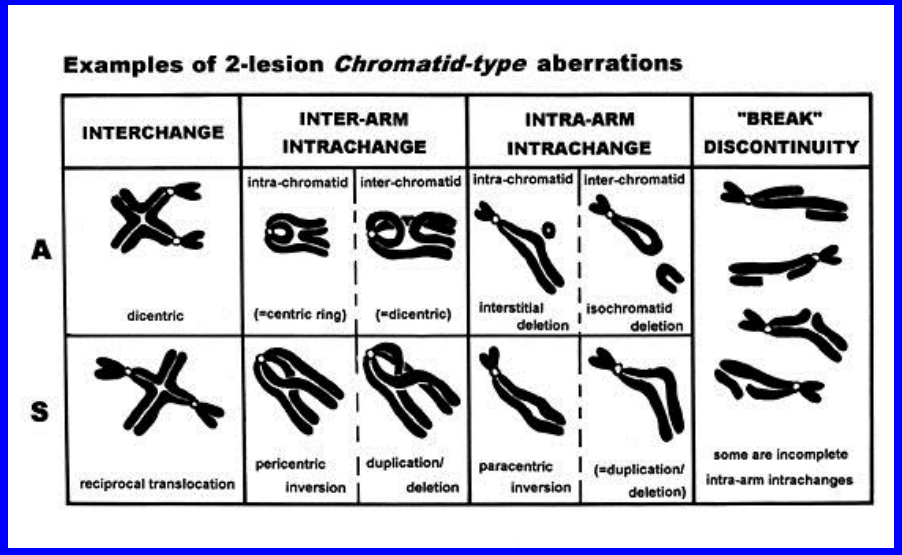
CLASSIFICATION OF PRIMARY CHANGES

Since the chromosome we see and score at metaphase has two (sister-) chromatids, it is convenient (and conventional) to divide all aberrations into two broad types:

- ✓ ***Chromosome-type*** where the breaks and re-joins always affect both sister-chromatids at any one locus, occur in the presynthetic phase (G1) of the cell cycle.
- ✓ ***Chromatid-type*** where the breaks and re-joins affect only one of the sister-chromatids at any one locus. This type of structural aberration occurs in chromosomes represented by one or two chromatids (late S-phase, G1-phase and prophase of mitosis)





CLASSIFICATION OF PRIMARY CHANGES

Nearly all the aberrations to result from the interaction ("re-joining") of two breaks, so we can further classify them on the basis of where these breaks are situated in relation to the chromosome arms.

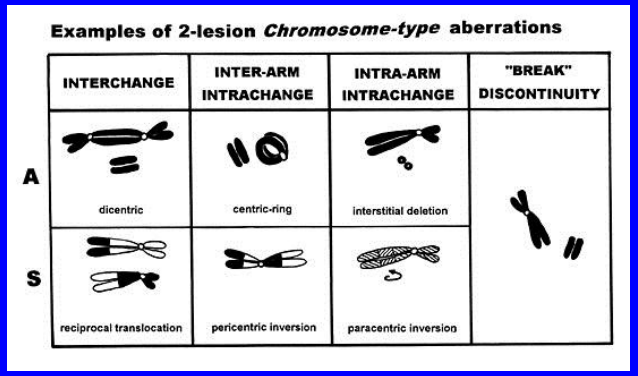
- If the breaks are situated in the arms of different (non-homologous or homologous) chromosomes we have the category of ***interchanges***.

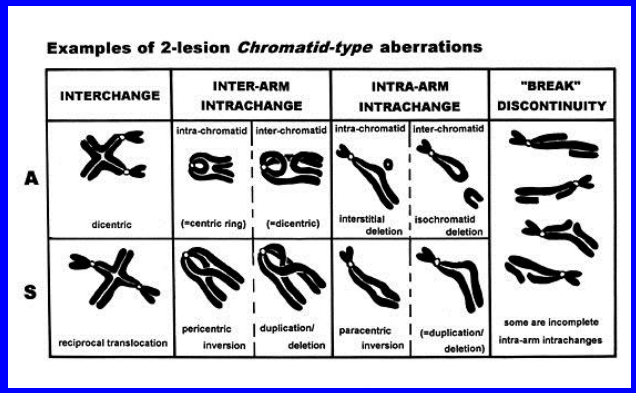
- If the breaks are in the opposite arms of the same chromosome, we have the category of ***inter-arm intrachanges***.

- If the two breaks are both in the same arm of a chromosome, we have the category of ***intra-arm intrachanges***.

These three categories are often referred to collectively as ***exchanges***.

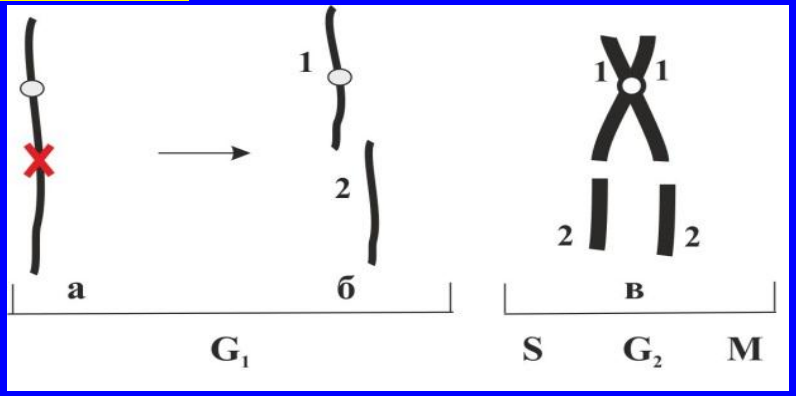
- Some aberrations to arise from a single, open break in just one arm. This category we term "***breaks***" or "***discontinuities***".

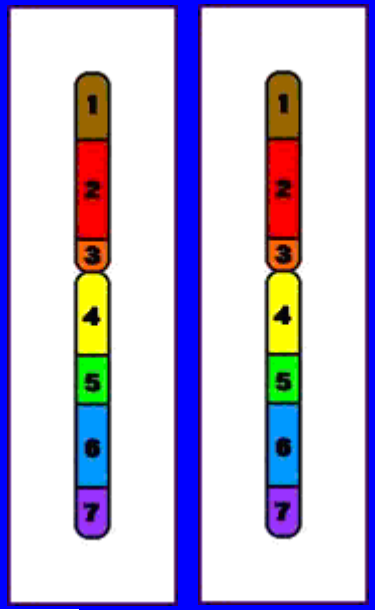




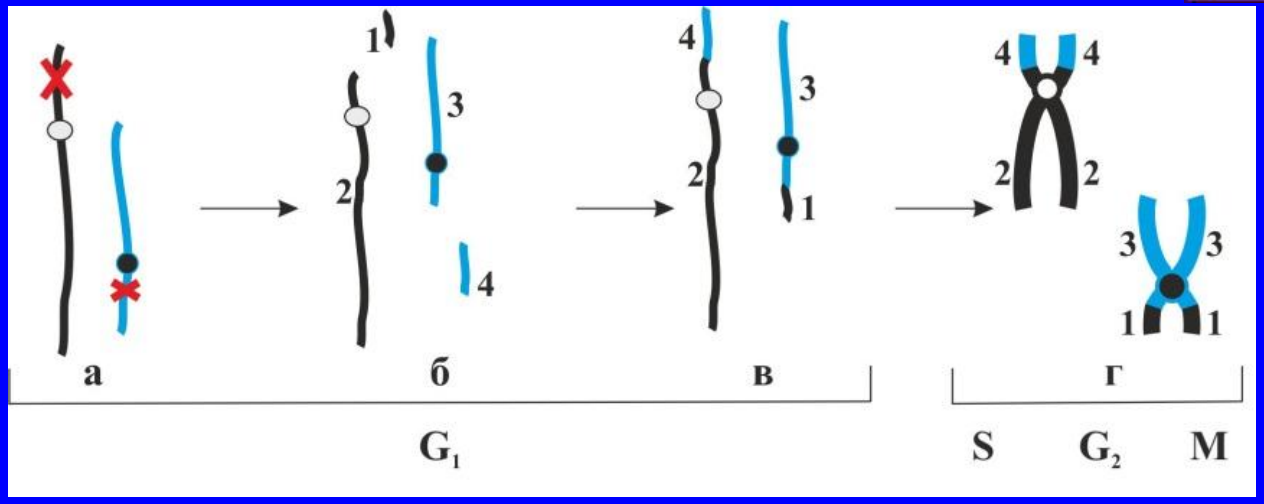
Chromosome-type Aberrations

Paired terminal deletions



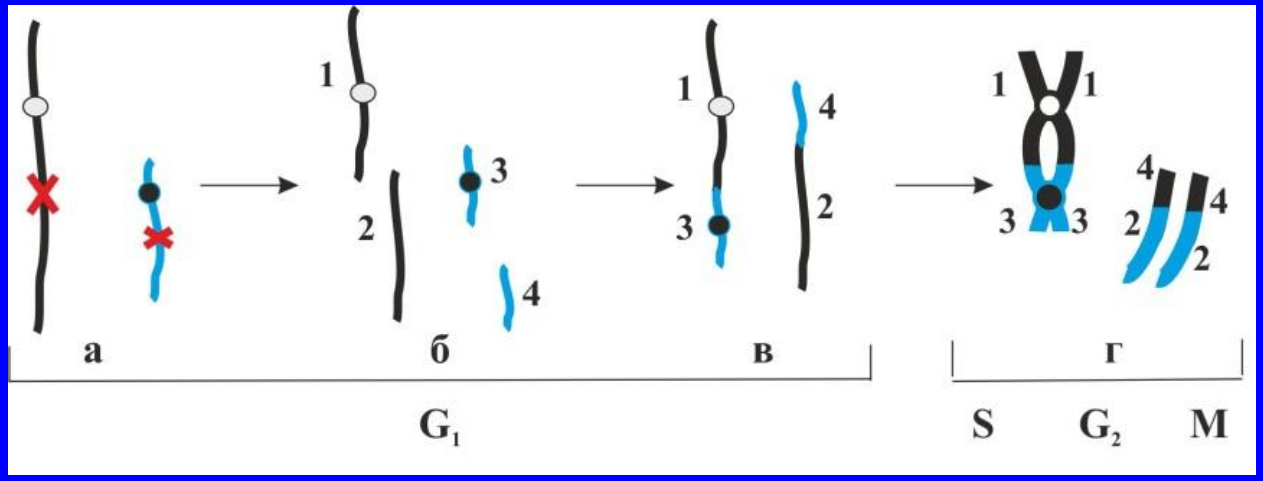


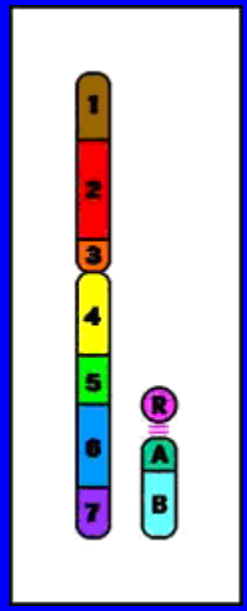
Symmetric chromosomal translocations

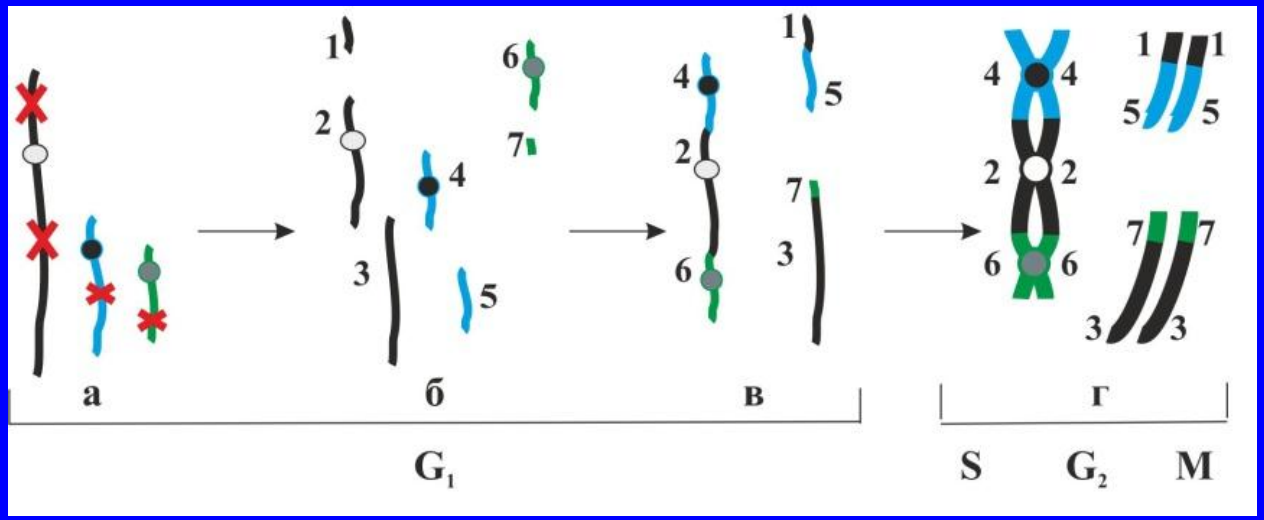


Chromosome-type Aberrations

Asymmetric chromosomal translocations: dicentrics and polycentrics.

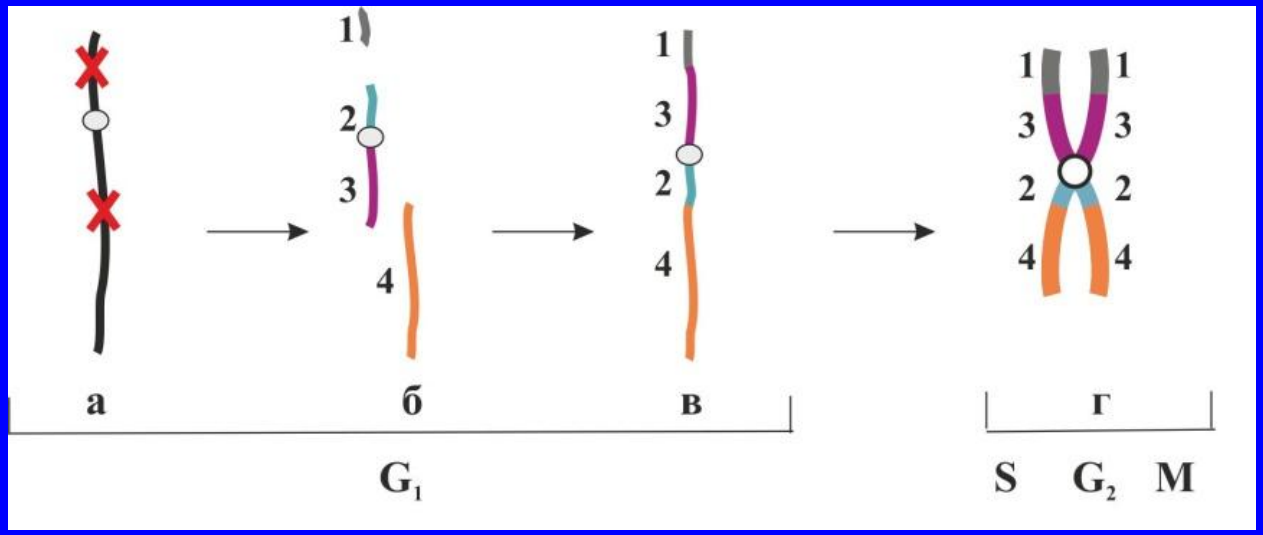


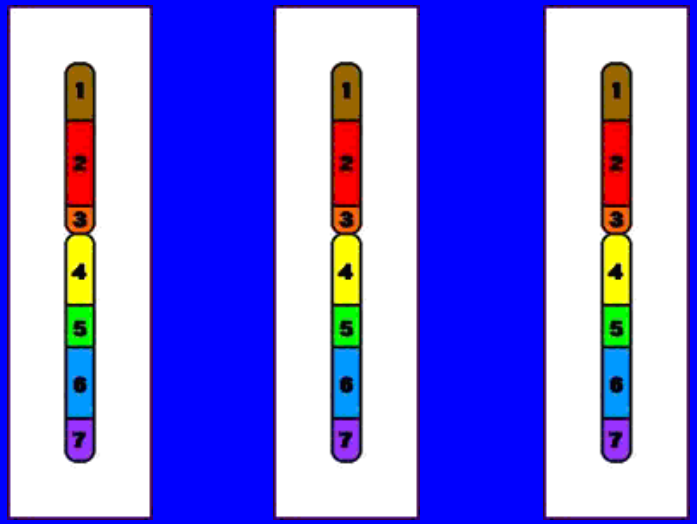




Chromosome-type Aberrations

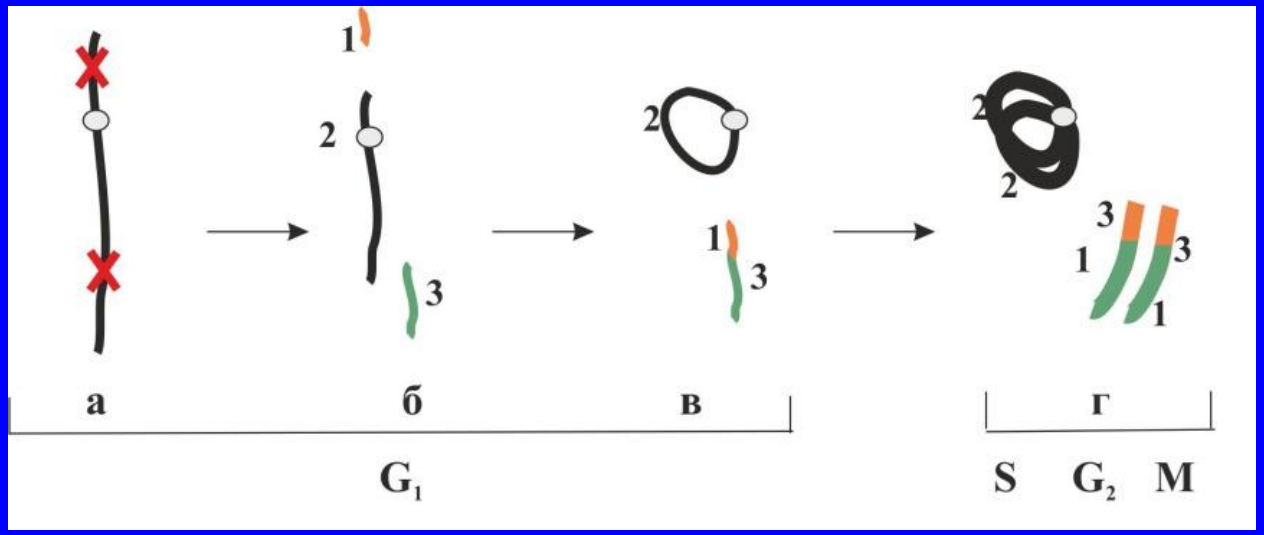
Inversion

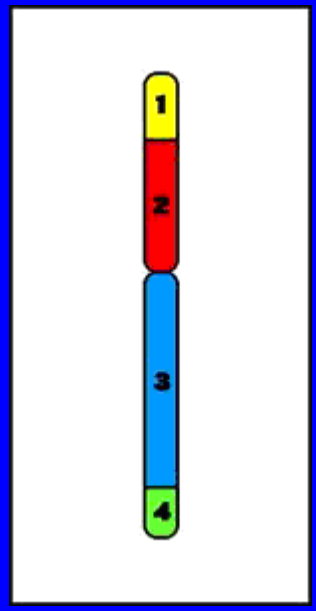


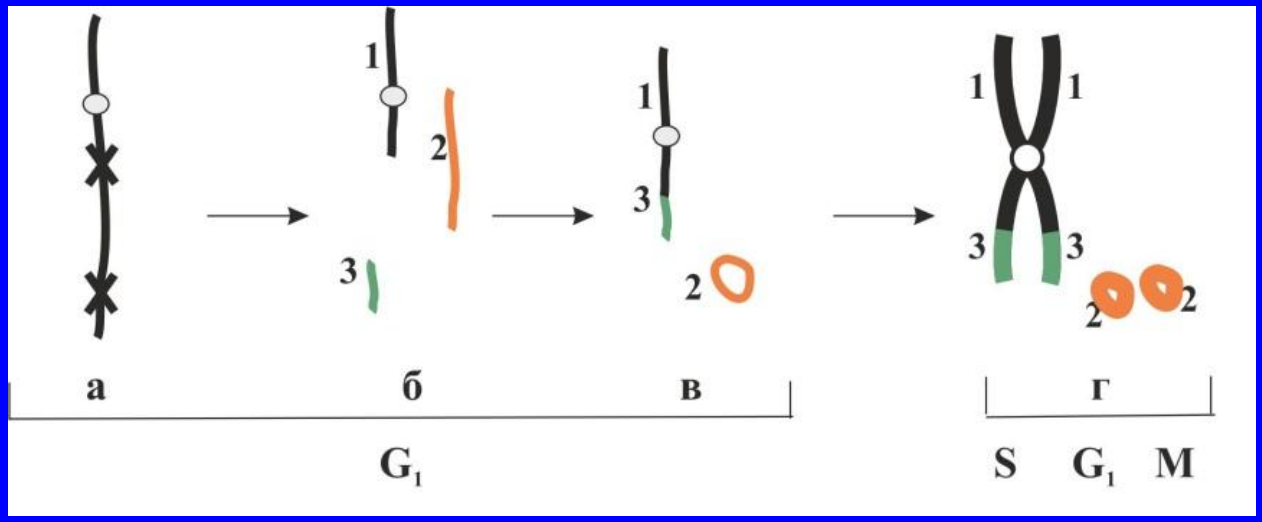


Chromosome-type Aberrations

Ring chromosome: centric ring, acentric ring

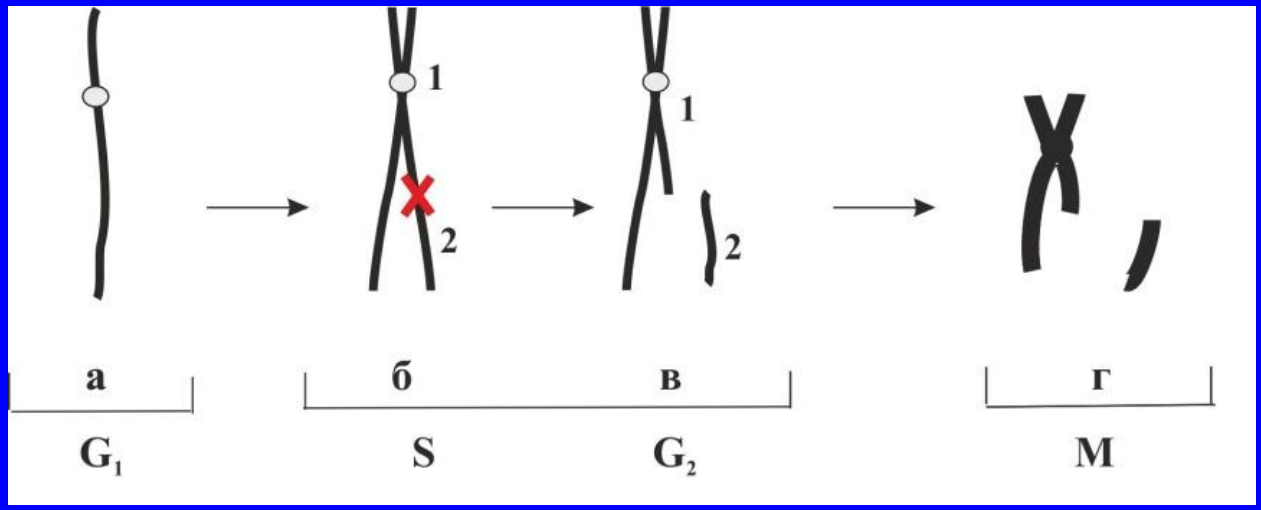




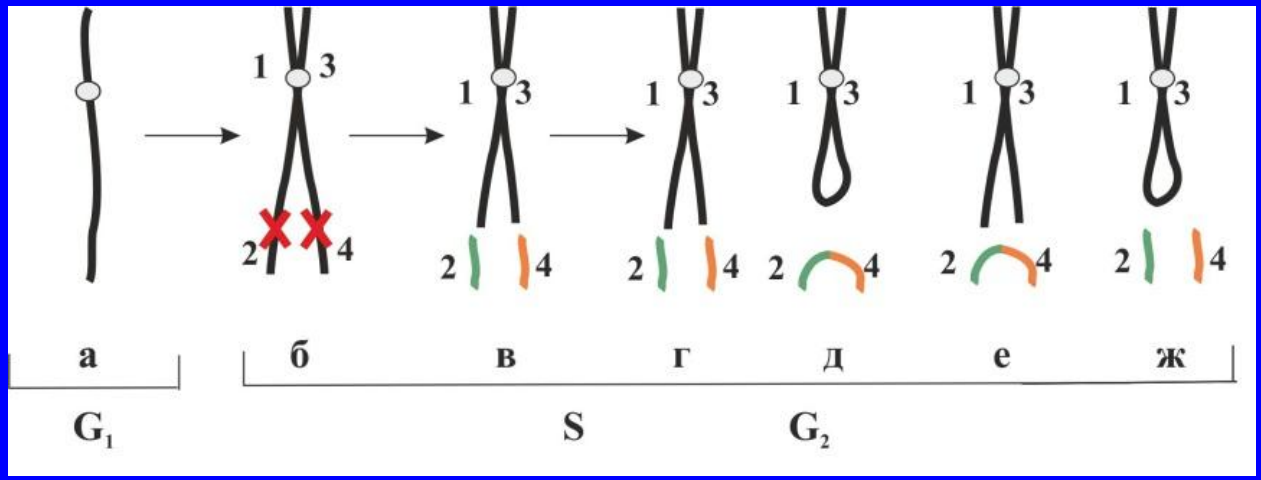


Chromatid-type Aberrations

Chromatid terminal deletions

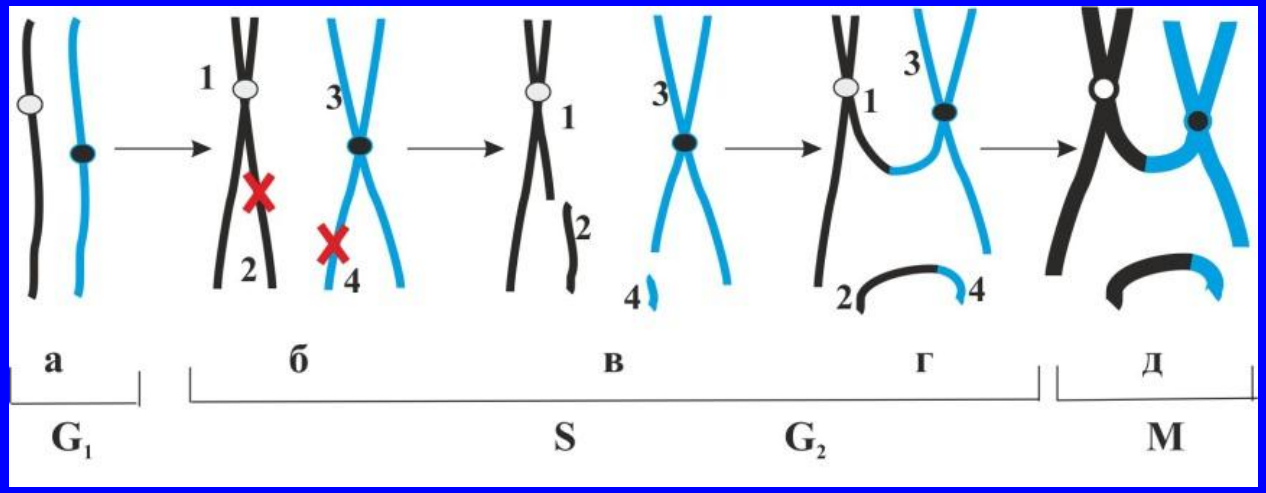


Isochromatid deletions

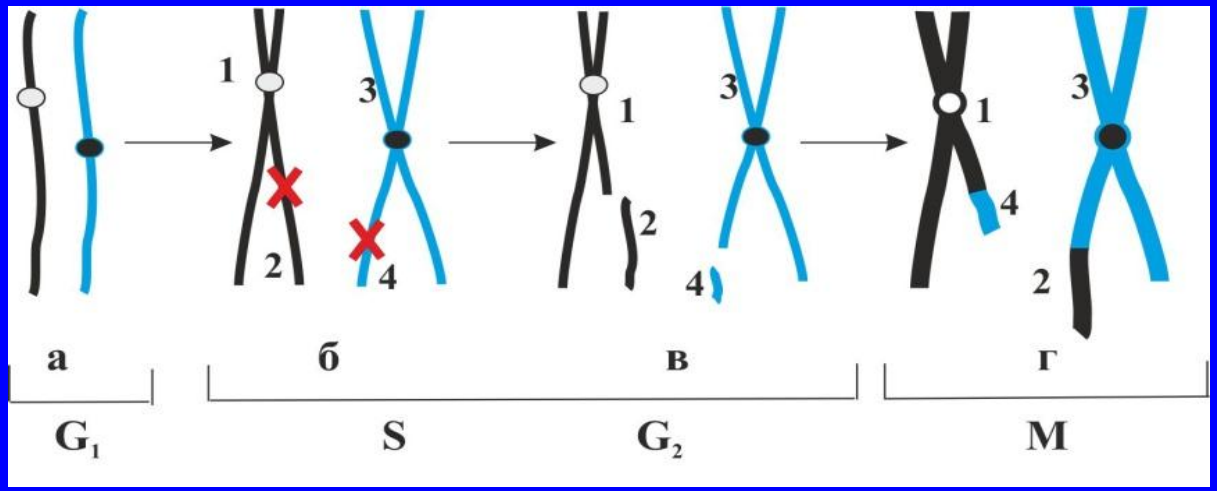


Chromatid-type Aberrations

Asymmetric chromatid translocation (chromatid dicentric)

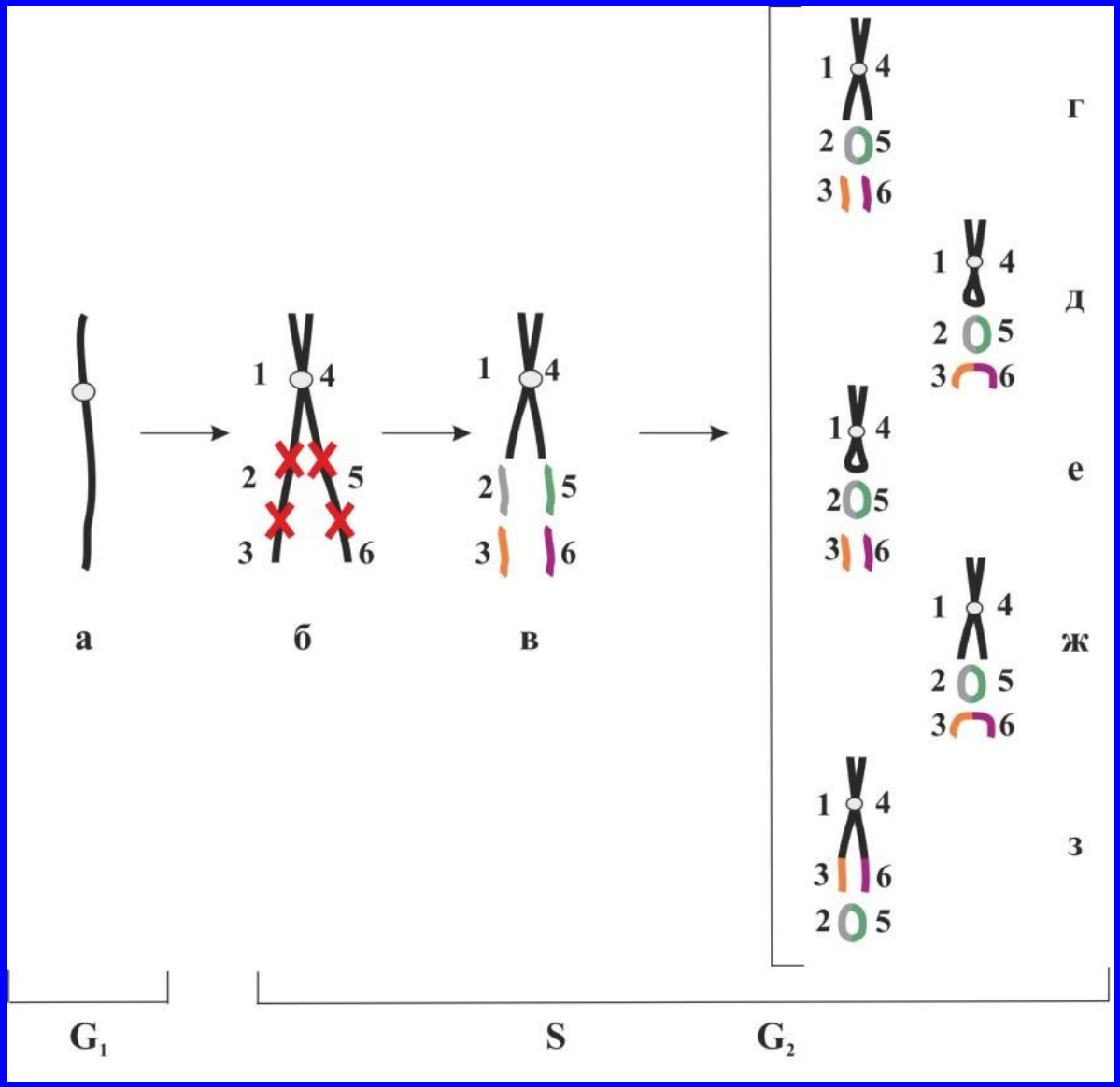


Symmetric chromatid exchanges (translocation)



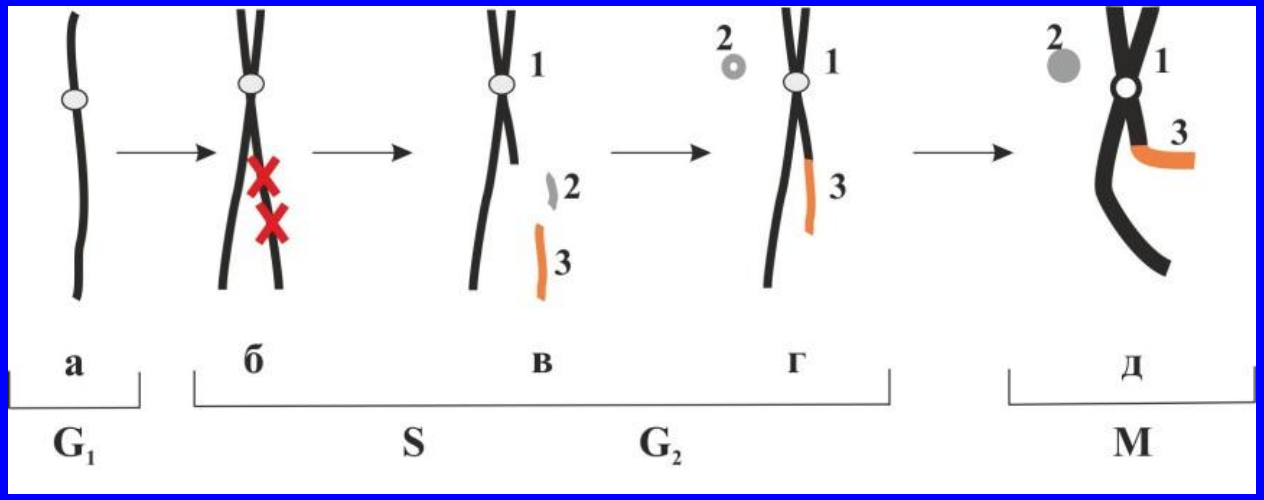
Chromatid-type Aberrations

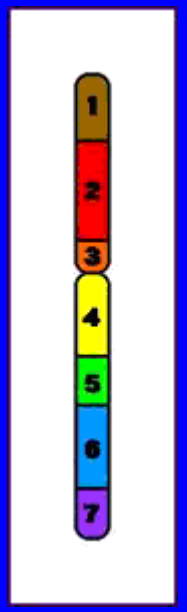
Isochromatic interstitial rings



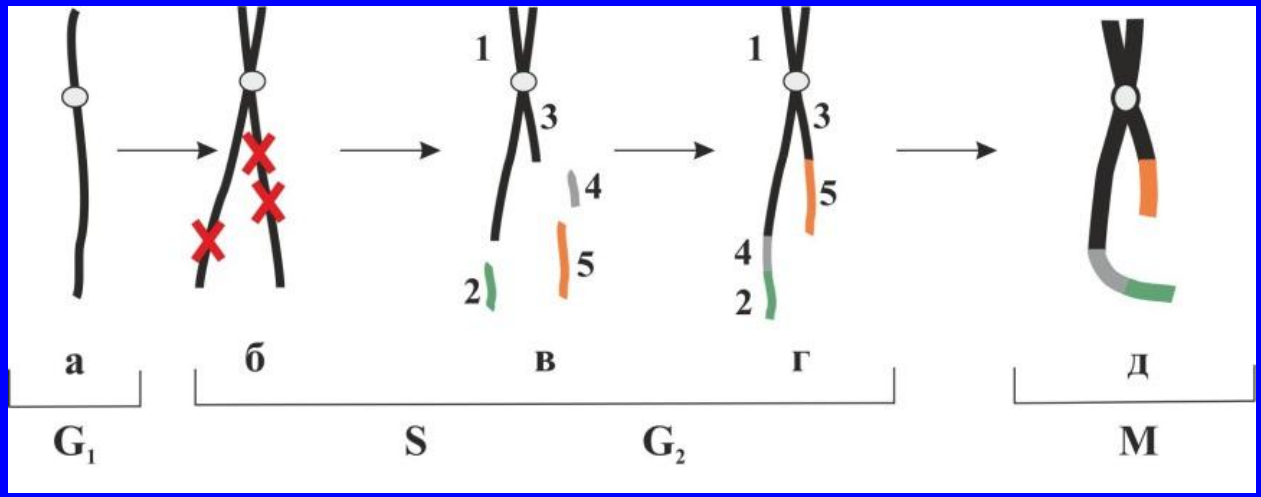
Chromatid-type Aberrations

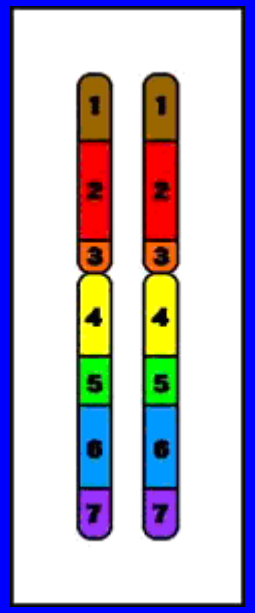
Ring interstitial chromatid deletion





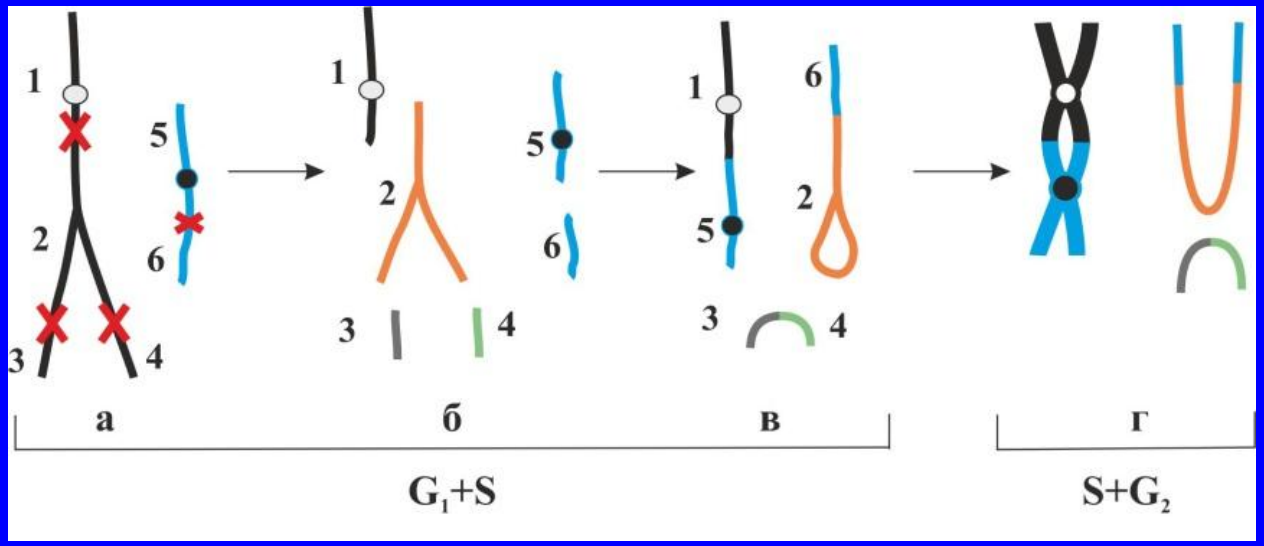
Chromatid interstitial duplication-deletion





Chromatid-type Aberrations

Chromosomal chromatid rearrangements



Thank you for attention!