

Somatic and gametic chromosomal aberrations

Chromosomal aberrations are numerical or structural deviations from the normal structure of the karyotype.

Somatic chromosomal aberrations

- *it is not passed on to offspring*
- **in the early stages of the development of the zygote** - a mosaic occurs, syndromes similar to gametic chromosomal aberrations occur
- postnatally, they usually mean the formation of **tumors**
- numerical and structural aberrations
- for example, the Philadelphia chromosome - t(9;22) in CML, translocation t(8;14) in Burkitt's lymphoma, etc.

Gametic chromosomal aberrations

- *it is passed on to offspring*
- **numerical**

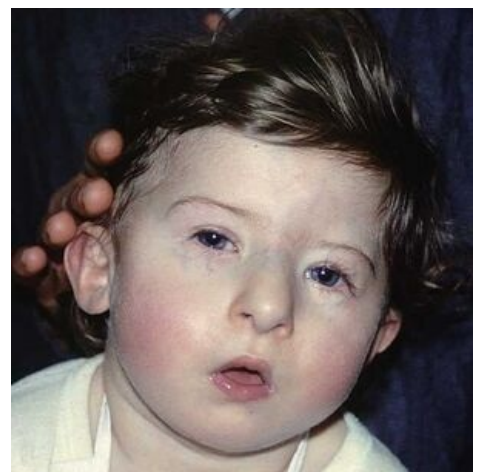
1. nondisjunction in meiosis I results in a gamete with 2 different (one from the father, one from the mother) or none of the pair of chromosomes in which the nondisjunction took place
2. nondisjunction in meiosis II results in a gamete with 2 identical (both from the father or the mother) or none of the pairs of chromosomes in which nondisjunction occurred
3. polyploidy - it is the multiplication of entire sets of chromosomes, genomic aberrations (hydatiform moles arise, incompatible with life)
4. aneuploidy - trisomy (Down syndrome, Patau syndrome, Edwards syndrome, Klinefelter syndrome, XXX syndrome, XYY syndrome) or monosomy (Turner's syndrome)

- **structural**

1. deletion (deletion syndromes: Cri du chat syndrome (5th chromosome), deletion form of Turner syndrome; microdeletion syndromes: Wolf-Hirschhorn syndrome (4th chromosome), Prader-Willi and Angelman syndrome (15th chromosome), DiGeorge syndrome, etc.) - terminal and interstitial deletion
2. duplication
3. insertion
4. inversion
5. ring chromosome
6. chromosome marker
7. dicentric chromosome
8. translocation - reciprocal and Robertsonian
9. isochromosome



Wolf-Hirschhorn syndrome



DiGeorge Syndrome

Links

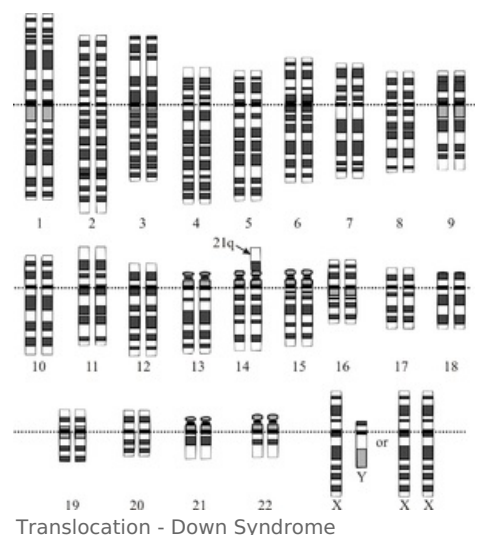
Related Articles

- Chromosomal abnormalities
 - Numerical chromosomal abnormalities
 - Structural chromosomal abnormalities
- Chromosomal aberrations in the etiology of neoplasia

References

- OTOVA, Berta, et al. *Medical biology and genetics I. vol.* 1st edition. Prague: Karolinum, 2008. 123 pp. ISBN 978-80-246-1594-3 .

Category: Genetics



Translocation - Down Syndrome