Mosaic

Chromosomal mosaicism is the presence of two (or more) cell lines with **different** karyotypes, originating **from a single zygote.** The mosaic must be distinguished from the so-called **chimera,** which contains two cell lines with different karyotypes, but they come from two zygotes. In humans, we know **the 46,XX/46,XY chimera,** a very rare genetic constitution that can arise from the **fertilization of an egg and a zygote,** each sperm with a different gonosome and subsequent fusion of both zygotes, or by parthenogenetic activation of the egg and fertilization of the two resulting haploid cells with sperm with a different gonosome, event fertilization of one haploid cell and diploid of the second. We distinguish only the type of chimera (XX/XY) associated with **true hermaphroditism,** probably most chimeras escape detection.

autosome trisomies (trisomy 21, 13, 18) occur less often in mosaicis. **trisomy 8** is also known , which occurs **only in mosaic form** in live births . Mosaic always arises **postzygotic,** i.e. nondisjunction during mitotic division or loss of a chromosome during division of a trisomic or normal zygote. The ratio of cell lineages then depends on which division the nondisjunction or loss of the chromosome occurred in and on how viable the abnormal cells are. E.g. postzygotic nondisjunction or loss of a chromosome affecting the autosomes would lead to the extinction of the monosomic line (monosomy of the autosomes is lethal even at the cellular level). Mosaicism resulting from the loss of one chromosome from a trisomic zygote results in a **mosaic of normal and trisomic** cell lines.

Early loss of a chromosome from a trisomic zygote can also lead to so-called **uniparental disomy** (UPD). UPD is the presence of two homologous chromosomes of the same parental origin, and the most common mechanism of UPD is the **loss of a chromosome** from the trisomic zygote, namely the chromosome that is represented only once in terms of parental origin in trisomy.

Mosaicism limited to the placenta

Mosaicism may be limited to the placenta . The literature reports that a relatively high percentage (1-2%) of first-trimester embryos are mosaics of trisomic and disomic (normal) lines, which was found when examining chorionic villus cells. Because mosaicism is not confirmed in most fetuses, such mosaicism is called confined placental mosaicism (CPM =confined placental mosaicism). However, some of these fetuses or neonates may show uniparental disomy.

Pseudomosaicism

During prenatal cytogenetic examination we can encounter so-called **pseudomosaicism.** Unlike true mosaicism, which is actually present in the cells of an individual, pseudomosaicism **arises when cells are cultivated in tissue culture.**

Lyonization

Inactivation of the X chromosome or **lyonization** occurs in the early stages of development (approximately at the stage of an embryo consisting of 100-200 cells) if the karyotype contains more than one X chromosome (most often in the case of a normal female karyotype 46,XX; however, it also occurs in male individuals sex with Klinefelter syndrome - karyotype 47,XXY and in other pathological karyotypes with more than one X chromosome so that in the final state there is only one active X chromosome in the cell). **Inactivation of the X chromosome is random** in each cell of the embryo , but also **permanent,** because all other cells arising from the division of this cell will already have the same inactivated chromosome, whether of maternal or paternal origin. The inactivated X chromosome in this way represents a deposit of highly condensed **chromatin,** visible as the so-called **Barr body** or **sex chromatin.** Individuals with monosomy 45,X, as well as 46,XY males, do not have a Barr body. The inactivation process is controlled by a regulatory region known as the **X-inactivation center (XIC).** In this region there is, among other things, the gene for non-coding RNA **XIST** (X inactive specific transcript (non-protein coding) ; Xq13.2; OMIM: *314670 (https://omim.org/entry/314670)) and several of its regulators including the **TSIX** gene (TSIX transcript, XIST antisense RNA ; Xq13.2; OMIM: *300181 (https://omim.org/entry/300181)). It is the RNA product of the XIST gene that induces changes in the conformation of the X chromosome, which ultimately lead to its inactivation.

Genes stored in the pseudoautosomal region of the X chromosome are not inactivated.

The inactivation of the X chromosome is also called **the Lyonization process** in honor of the British geneticist **Mary Frances Lyon** (1925-2014), who first described this process in 1961.

Links

Related Articles

- Chromosomal abnormalities
  - Numerical chromosomal abnormalities
  - Structural chromosomal aberrations
- Uniparental disomy

References

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- PRITCHARD, Dorian J. – KORF, Bruce R.. *Základy lékařské genetiky.* 1. edition. Praha : Galén, 2007. 182 pp. ISBN 978-80-7262-449-2.