Acquired chromosomal aberrations

Acquired chromosomal aberrations (ACA) arise during an individual's life as a result of mutagenic environmental influences or as a result of a malfunction of repair mechanisms.

Cytogenetic analysis of ACA

The analysis of ACA in peripheral blood lymphocytes is used, for example, as a group biological test for **monitoring of exposure to factors in the working environment**. Monitoring of ACA in tissue cultures *in vitro* is used to test genotoxicity of substances.

Principle of the ACA analysis method in peripheral blood lymphocytes

After a short-term (48 h) cultivation of peripheral blood lymphocytes, the cultures are treated with colchicine, a mitotic poison that stops cell division in the metaphase stage, and the cell suspension is processed by the cytogenetic method (hypotonia and repeated fixation). If we culture peripheral blood lymphocytes, phytohaemagglutinin, a substance that stimulates lymphocytes to divide, is added to the culture. After processing, the prepared preparations are stained with Giemsa dye.

Evaluation of preparations

By default, 100 (in the case of a group test) or up to 300 mitos (in the case of an individual test) are evaluated. An optical microscope (magnification 1000×) is used for evaluation. In the evaluated mitoses, the occurrence and number of:

- breaks – violation of the integrity of one or both chromatids, if the width of the break is greater than the width of the given chromatid.
- fragments – a part of a chromosome without a centromere;
- minutes, or double minute – a part of the chromatid with a diameter smaller than the width of the chromatid
- chromatid exchanges;
- chromosome rearrangements: translocations, ring chromosomes and dicentric chromosomes.

The following are not included in the total number of aberrant cells, but are also monitored:

- numerical changes (polyploidy or aneuploidy)
- endoreduplication
- gaps – violation of the integrity of one or both chromatids, if the width of the break is equal to or smaller than the width of the given chromatid.

While dicentric or ring chromosomes and translocations (aberrations of chromosome type) are typical aberrations caused by ionizing radiation, chromatid breaks and chromatid exchanges are typical after exposure to chemicals.

Since the frequency of dicentric chromosomes closely follows the radiation dose, cytogenetic examination and determination of the number of dicenters are used for biological dosimetry.

Translocations are difficult to recognize on classically stained slides, so most of them escape detection. Some chromosomal aberrations are unstable, they are lost during repeated cell division (dicentery, ring, fragments without centromeres...), so it is necessary to examine acquired chromosomal aberrations only in the first mitoses, i.e. after 48 hours of cultivation.

Interpretation of results

In the group test, he answers:

- less than 2% of aberrated cells (AB.C.) spontaneous frequency;
- 2-4% AB.B. increased exposure;
- more than 4% high exposure to genotoxic substances.

In an individual assessment, **frequency of 5 or more % AB.C.** is considered risky. Repeated finding of such frequency AB.B. means for the affected individual an increased risk of cancer, the risk of accelerated cell loss and an increased risk of congenital defects in the offspring.

A significantly increased % of aberrant cells is also found in patients with so-called syndromes associated with increased chromosome fragility (Fanconi anemia, Bloom's syndrome, xeroderma pigmentosum, ataxia telangiectasia).

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