Abstract

Mosaicism is represented by two or more chromosomally different cell lines in an individual. Mosaics are most often caused by chromosome malsegregation during mitosis, resulting in the gain or loss of chromosomes, known as aneuploidy, but structural aberrations can also occur in mosaic form. The problem is the limitation of detection with standart cytogenetic methods. The present study was carried out to compare the efficiency of FISH, array CGH and cytogenetic techniques in detection of mosaicism.

In the practical part the results of 45 patients with mosaicisms of aneuplody of gonosomes (26 patients) and mosaicisms of autosomes (19 patients) were compared. The data show that we have different peripheral blood karyotype and FISH results in 23 of 37 patients (62%). There was a case of failure of detection of the mosaicism on the karyotype and the FISH method revealed a abnormal cell lines with a percentage of less than 5%. The array CGH method confirmed the karyotype and FISH results in 10 out of 12 patients (83%) in peripheral blood tests.

The work also dealt with artificially made mosaics. From the results, it is obvious that the FISH method has a more accurate percentage of mosaic capture compared to the karyotype.

The results indicate that using the techniques in parallel allow in clinical practice to achieve high sensitivity and specificity for the correct evaluation of the cytogenetic basis of abnormal clinical features in mosaic patients.

Keywords

Mosaicism, chromosome aberrations, karyotyping, FISH, array CGH, diagnostics