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An inexpensive system for quantitative analysis of mosaic form of Turner/Klinefelter syndrome

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In our contribution, a prototype of a simple low-cost system for automated evaluation of mosaic form of chromosome aneuploidies will be introduced. The primary goal of this system is to improve the accuracy of evaluation of simple chromosome aberrations in mosaic form, which are related to the Turner (mos 45,X/46,XX) and Klinefelter syndrome (mos 47,XXY/46,XY).

Since the frequency of the aberrant nuclei is rather low in the mosaic form (up to approx. 5%), it is necessary to evaluate the counts of the fluorescence signals in a sufficient set of nuclei images (approx. 1000 interphase nuclei or more). From the point of view of image processing, this includes the standard operations of image preprocessing, segmentation, spot detection and parameter evaluation, followed by filtering based on object parameters. The prototype system is composed of hardware part, which provides automated image acquisition, and software part for image processing, image database access, statistical evaluation, visualization, etc. To large extent, the system is composed of the COTS components: the Euromex OX.3075 fluorescence microscope with a plan fluorite lens and an Edmund Optics motorized stage have been used for image acquisition. The control protocol for the stage and the illumination system have been developed inhouse by the CAMEA company, to ensure optimum performance. The software part of the system builds partly on the open-source software: the image processing module (preprocessing, segmentation, parameter evaluation) has been designed as an ImageJ plugin, the machine learning part, which is used to filter the objects (nuclei and signals) on the basis of parameters, is implemented using the WEKA library. To store the images, the MySQL database is used. Currently, we are also testing incorporation of the OMERO client-server software, which could serve both for storing the images and for visualization, as well as the backend system.

To test the system, samples of cultivated lymphocytes from patients with different forms of gonosomal aneuploidies have been used, as well as artificial mixed male and female samples with known ratio of cells with different karyotypes. The samples were hybridized with alpha-satellite FISH probes, which marked centromeres of the X chromosome in the individual cells (for Turner syndrome). A 40x and 60x magnification lens has been used in our experiments, resulting in images with approx. 20 nuclei. To obtain good quality images, exposition time of 500 ms was used and we have achieved processing time for one image below 2 s.

Currently, works are undergoing on integration of the components of the system into final prototype.

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