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New Artificial Intelligence Based Computer-Aided Chromosome Analysis and Karyotyping a Pilot Evaluation

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Background & Introduction

Over the past decade, digital imaging has helped streamline the chromosome analysis and karyotyping process by providing automated tools to identify metaphases for analysis, separate chromosomes, and classify them in a karyogram. Although efficient in reducing turnaround time as compared to fully manual processes, results of these digital technologies still require many manual adjustments.

In order to further decrease time to results, laboratories are increasingly adopting computer-aided systems which further automate the multiple steps involved with the preparation of karyograms for review.

This pilot study evaluates the accuracy of artificial intelligence (AI)-based algorithms for the analysis and karyotyping of both peripheral blood and bone marrow specimens while scanning.

Methods

Following standard preparation and staining, G-banded slides were scanned and analyzed using the HiBand system (Applied Spectral Imaging). Metaphases, automatically identified at 10X magnification, were imaged at 100X and twenty cells were automatically selected for each case based on a predefined quality score.

A newly developed AI-based computer-aided karyotyping application was used to automatically analyze and karyotype selected metaphases while scanning. Resulting karyograms were reviewed by certified Cytogenetic professionals to assess the number of correctly segmented, classified and oriented chromosomes as well as the number of manual adjustments required to correct each automatically karyotyped metaphase.

Results

Two normal cases, one peripheral blood from AWFBH and one bone marrow from AON, were included in this pilot evaluation. Twenty representative metaphases were reviewed for each specimen type (total of 40), comprising 917 peripheral blood chromosomes (3 random losses) and 916 bone marrow chromosomes (4 random losses). Peripheral blood and bone marrow metaphases represented an average of 11.5±3.2 (range 5-17) and 10.3±5.8 (range 3-26) overlaps, touching chromosomes or more complex clusters respectively. Extra-long peripheral blood chromosomes (>650 bands) and bone marrow chromosomes with split chromatid were not included in this pilot evaluation.

The system automatically segmented 871 out of the 917 blood chromosomes (95%) of which all (100%) were correctly placed in the karyogram. The system also automatically segmented 824 out of the 916 bone marrow chromosomes (90%) of which 96% were correctly placed in the karyogram. Orientation was accurate for 98% of the correctly segmented and classified blood and bone marrow chromosomes.

Peripheral blood specimens required an average of 2.1±2.1 (range 0-6) manual adjustments, 1.8±1.7 (range 0-5) for segmentation correction and 0.3±0.6 (range 0-2) for classification adjustment. Bone marrow specimens required an average of 7.4±2.8 (range 3-13) manual adjustments, 4.4±2.8 (range 1-10) for segmentation correction and 3.1±2.2 (range 0-8) for classification adjustment.

Conclusions

This pilot evaluation indicates that the new AI-based karyotyping application correctly segmented and classified 95% of peripheral blood and 86% of bone marrow chromosomes in representative metaphases containing an average of 11 overlaps, touching chromosomes or more complex clusters.

This high accuracy, resulting in an average of 5 corrections per metaphase (2 for peripheral blood and 7 for bone marrow), is anticipated to significantly reduce the time spent on preparing karyograms, allowing Cytogenetics professionals to focus more time on higher-value tasks.

This pilot evaluation should be further expanded to assess the time spent correcting karyograms automatically generated by the AI-based solution as compared to karyograms generated by current techniques.

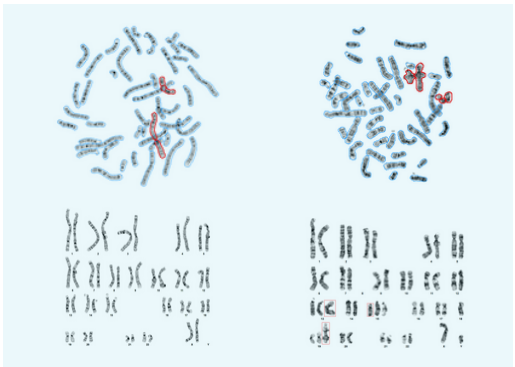

Figure 1
Representative examples of automatically segmented metaphases (top) and automatically placed chromosomes in karyograms following manual segmentation adjustments (left: peripheral blood, right: bone marrow, errors are highlighted in red).

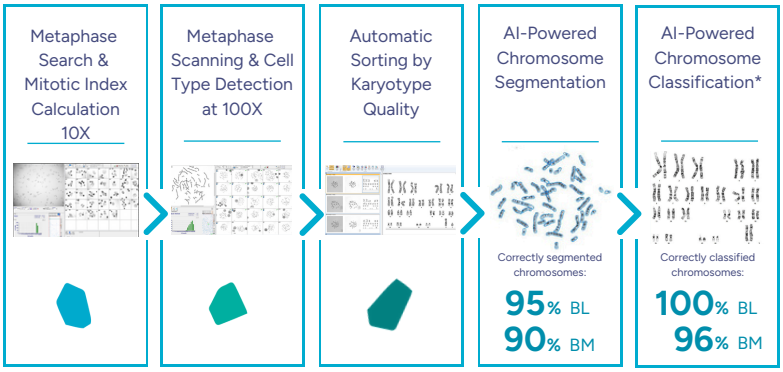

Figure 2
Fully automated karyotyping workflow supporting karyotype review and approval while scanning
* Following manual adjustments to correct automatic segmentation

Disclosures
A.F.T., J.B., L.S.R., K.A.G., R.K.O and J.D. have no disclosure. Y.G., M.K., C.S. are employees of Applied Spectral Imaging.