P-684

Improved Karyotyping Efficiency with Artificial Intelligence: a Multicenter Evaluation of Peripheral Blood Karyograms

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Background & Introduction

- Cytogenetics laboratories are experiencing a reduction in the workforce and challenges replacing highly experienced technologists.
- To address these challenges and the **growing demand to optimize resource allocation,** we have incorporated **artificial intelligence (AI)** into the software for karyogram preparation.
- Integration of AI into the ASI HiBand system will **significantly reduce the turnaround time** required to achieve results compared to conventional digital imaging systems that rely on traditional image processing technologies.
- Following a small pilot evaluation reporting 92% correct segmentation and classification of cytogenetically normal peripheral blood and bone marrow chromosomes with the AI technology (1), **a recent multi-center study showed a 46% reduction in time technologists spend karyotyping** cytogenetically normal bone marrow samples with AI-based software compared to existing non-AI processes (2).
- This work expands on the previous study by comparing the two methods for karyotyping peripheral blood metaphases.

Methods

- G-banded slides of normal peripheral blood samples from ten patients (5 females and 5 males) were prepared by standard methods and scanned using the HiBand system (Applied Spectral Imaging).
- Metaphases were automatically identified at 10X magnification and images were captured at 100X. Twenty cells were selected for each case based on predefined criteria and established laboratory analysis protocols.
- Cases were analyzed twice by cytogenetics technologists, once with the current non-AI software (Figure 1) and a second time with the AI-based technology (Figure 2).
- Suggested karyograms automatically generated by the AI algorithms were compared to the karyotype tables prepared by the cytogeneticists. Segmentation and placement errors were computed together with the number of manual adjustments necessary to correct each metaphase.
- Time required to correct and analyze each case was recorded with both methods and compared. Statistical significance was assessed using the Wilcoxon Signed-Rank test. A p-value lower than 0.05 was considered significant.

References

(1) Taghiyev A, Best J, Rosenblum L et al. Genetics in Medicine Open 2024. 2(1):P671-101575
(2) Burnside R, Best J, Spudich L et al. American Cytogenomics Conference 2024

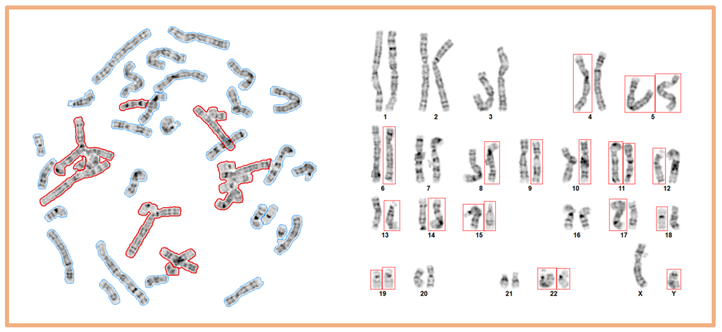

Figure 1

Representative example of the segmentation and classification of a peripheral blood metaphase using conventional non-AI method. Left: automatic segmentation. Right: automatic chromosome placement in the karyotype table following manual segmentation correction. Errors are highlighted in red.

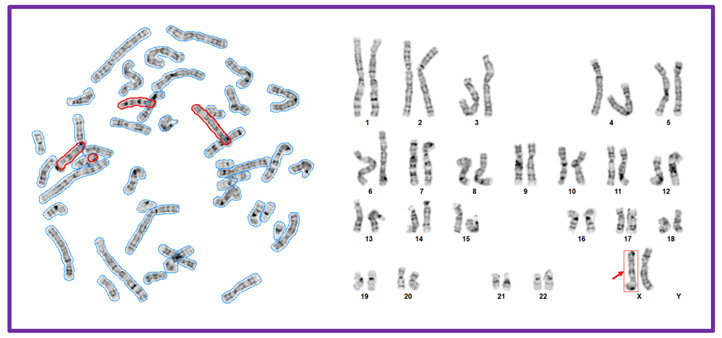

Figure 2

Representative example of the segmentation and classification of a peripheral blood metaphase using AI-based algorithms. Left: automatic segmentation. Right: automatic chromosome placement in the karyotype table following manual segmentation correction. Errors are highlighted in red.

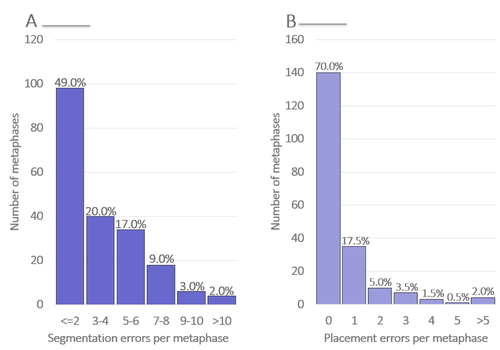

Figure 3

A: Number of segmentation errors per metaphase resulting from automatic AI-based segmentation.
B: Number of placement errors per metaphase resulting from automatic AI-based placement following manual segmentation corrections.

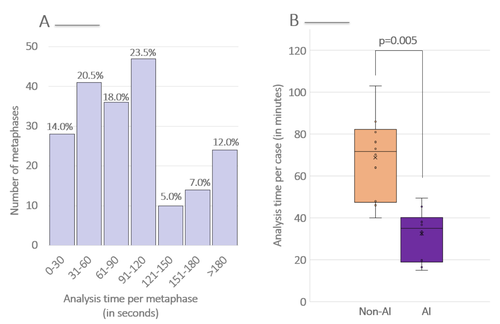

Figure 4

A: Time per metaphase required to analyze automatically generated karyotype tables, including segmentation, classification and orientation correction.
B: Box and whisker plot comparing the analysis time per case (20 metaphases) for non-AI versus AI-based karyotyping (p value is indicated).

Results

- Two hundred cytogenetically normal peripheral blood metaphases were included in this study, representing a total of 9,187 chromosomes (13 random chromosome losses, 0.1%).
- **Correct segmentation was achieved for 93%** of the analyzed chromosomes. The average number of segmentation errors per metaphase was 3.4±2.8 (range 0-14, median 3.0). Forty nine percent of the metaphases showed less than 2 segmentation errors (Figure 3A).
- **Correct placement in the karyogram was reported for 99%** of the chromosomes following manual segmentation correction. The average number of placement errors per metaphase was 0.6 (range 0-11, median 0.0).
- **No placement error was reported in 70%** of the metaphases (140 out of 200). Eighteen percent of the metaphases (35 out of 200) showed only one placement error (Figure 3B).
- The average number of manual adjustments required per karyogram was 3.6±2.8, with a range of 0-14 adjustments and a median of 3.0 adjustments per metaphase.
- The average analysis time per metaphase for both segmentation and placement was 97.4 seconds, with a standard deviation of 69.8 seconds (range 13-431 seconds, median 82 seconds) (Figure 4A).
- The **overall time required** to complete a peripheral blood case including 20 metaphases **was significantly reduced by 53%** (p=0.005), from 68.7±19.7 minutes with the current non-AI software version (range 40.0-103.0, median 71.6) to 32.5±11.9 minutes using AI-assisted karyotyping (range 14.9-49.4, median 35.0) (Figure 4B).

Conclusions

- The accuracy and performance observed for peripheral blood specimens was comparable to those reported for bone marrow cases.
- For both sample types, the **AI technology reduces by approximately half the time necessary to review and correct karyograms**, requiring an average of 3.6 corrections per metaphase.
- Accuracy in chromosome segmentation and classification is projected to further improve while algorithm models are exposed to data from additional sources.
- Moreover, **as AI technology progresses**, correct contouring and placement of abnormal chromosomes, including those with rearrangements, will **further increase efficiency** in the karyotyping process.

Disclosures

RDB, KP, JB, AFT, LS, AL, SC, RKO and LSR have no disclosure. MK, YG and CS are employees of Applied Spectral Imaging.