

Comparative Evaluation of an AI-Based Scanner-Agnostic Add-On Utility Versus Conventional Karyotyping

Janet Lukacs, MBA, CG(ASCP)MB¹, Elizabeth A Shinn, BA, CG(ASCP)¹, Cody A Felty, BS, MLS(ASCP)CG¹, Marian L Johnson, BSMT, CG(ASCP)¹, Michael B Lynn, BSMT, CG(ASCP)¹, Misty G Koch, BSMT, MLS(ASCP)CG¹, Melissa D Knowlton, BSMT, CG(ASCP)¹, Ohad Frenkel, BSc², Yael A Glickman, PhD, MBA², Cristina Steele, MBA, CG(ASCP)²

¹ Cytogenetics Laboratory-University of Arkansas for Medical Sciences, Little Rock, AR, USA; ² Applied Spectral Imaging, Carlsbad, CA, USA;

Introduction

- Over the past few years, **Artificial Intelligence (AI)** has become the standard in chromosome analysis and karyotyping.
- Multiple studies¹⁻³ have demonstrated that normal metaphases are analyzed in an average of **1.5 minutes** when using AI-powered computer-aided software, a 50% decrease in analysis time compared to traditional karyotyping methodologies.
- Laboratories which do not currently have access to AI-powered metaphase analysis software are seeking **scanner-agnostic add-on utilities** compatible with their installed scanning systems⁴, particularly if such systems were recently acquired and comply with the latest information technology and regulatory standards.
- The present study evaluates such a commercially available AI add-on utility, comparing the **metaphase analysis time** to the non-AI application installed on the scanning system.

Methods

- G-banded slides of bone marrow (BM) and peripheral blood (PB)** samples were prepared following standard laboratory protocols.
- Slides were scanned using the Cytolinsight GSL Scanning, Capture and Review System v8.0 (Leica Biosystems) to generate **metaphase images at 100x magnification**.
- Each metaphase was then analyzed on the Cytolinsight software, and the **time required to karyotype each cell was recorded**.
- The **exact same metaphase images** were then imported into the HiBand scanner-agnostic karyotyping software application v8.4.1 (Applied Spectral Imaging) through an automated, unsupervised procedure.
- During the import process, suggested **AI-based computer-aided karyograms** were automatically generated and displayed in the cell gallery, ready for review (Figure 1).
- The **analysis times needed to correct a metaphase** on the HiBand add-on utility were recorded and compared to the analysis time measured using the Cytolinsight GSL karyotyping application.
- Statistical significance** was assessed using the Wilcoxon Signed-Rank two-tailed test for paired samples. A p-value lower than 0.05 was considered significant.

References

- (1) Rosenblum LS et al, The emergence of artificial intelligence-guided karyotyping: a review and reflection. *Genes (Basel)* 2025; 16(6):685
- (2) Burnside RD et al, Multicenter evaluation of a new AI-Based Karyotyping software on bone marrow specimens, *Genetics in Medicine* 2025; 3(1):101918
- (3) Burnside RD et al, Improved karyotyping efficiency with artificial intelligence: A multicenter evaluation of peripheral blood karyogram. *Genetics in Medicine Open* 2025; 3(2):103053
- (4) Steele C et al, Enhancing interoperability to enable broader adoption of artificial intelligence in chromosome analysis and karyotyping: A pilot evaluation. *Genetics in Medicine Open* 2025; 3(2):103033

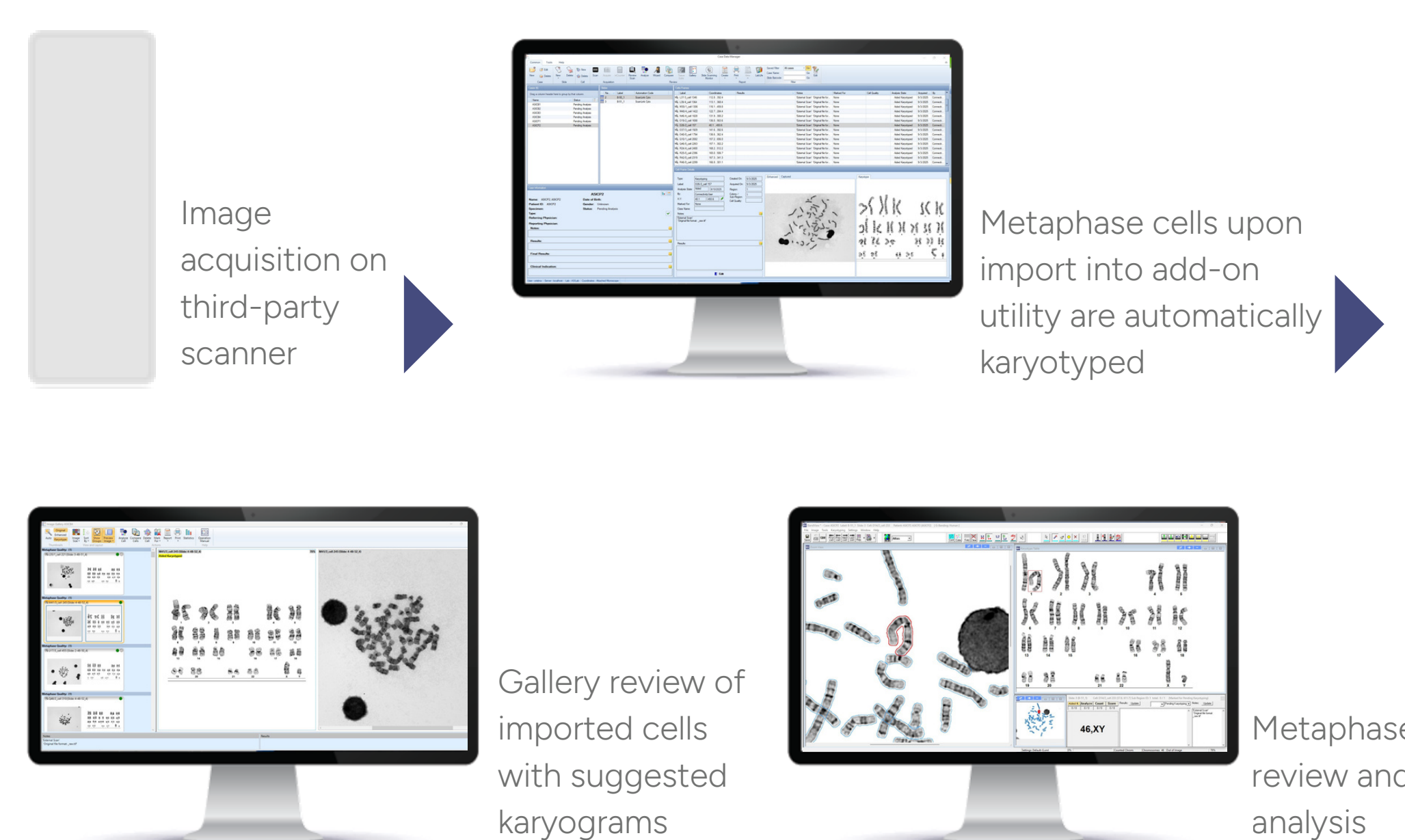


Figure 1
Workflow illustrating the integration of a non-AI third-party scanner with an add-on AI-based utility for automated karyotyping and downstream analysis.

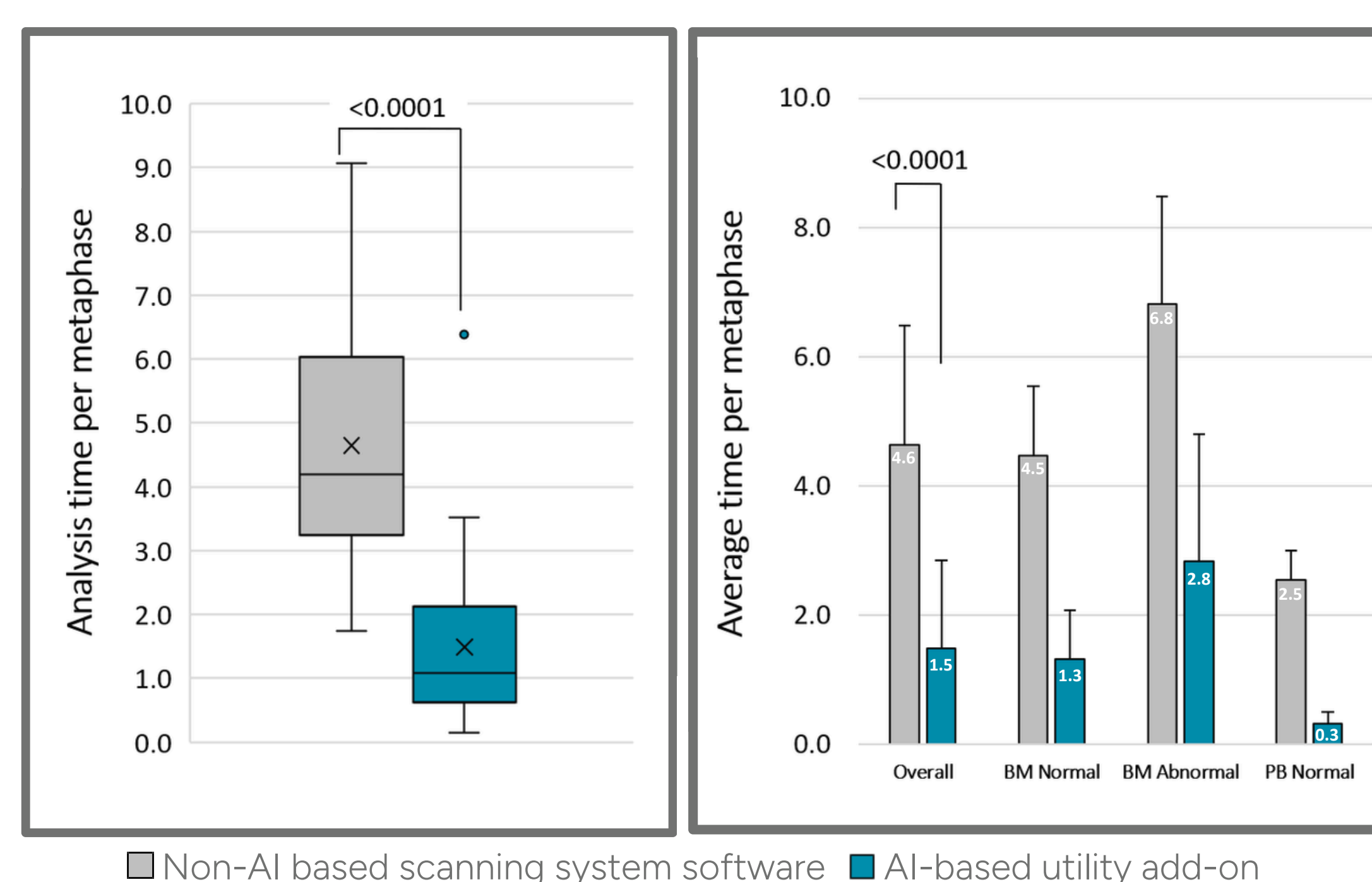


Figure 2
Left: Box and whisker plot comparing the analysis time with the non-AI scanning system software (grey) and the AI-based utility add-on (blue).
Right: Column plot comparing the average analysis time with the non-AI scanning system software (grey) and the AI-based utility add-on (blue) for all cells, normal bone marrows (BM), abnormal bone marrows and normal peripheral bloods (PB). P values are indicated on the graphs.

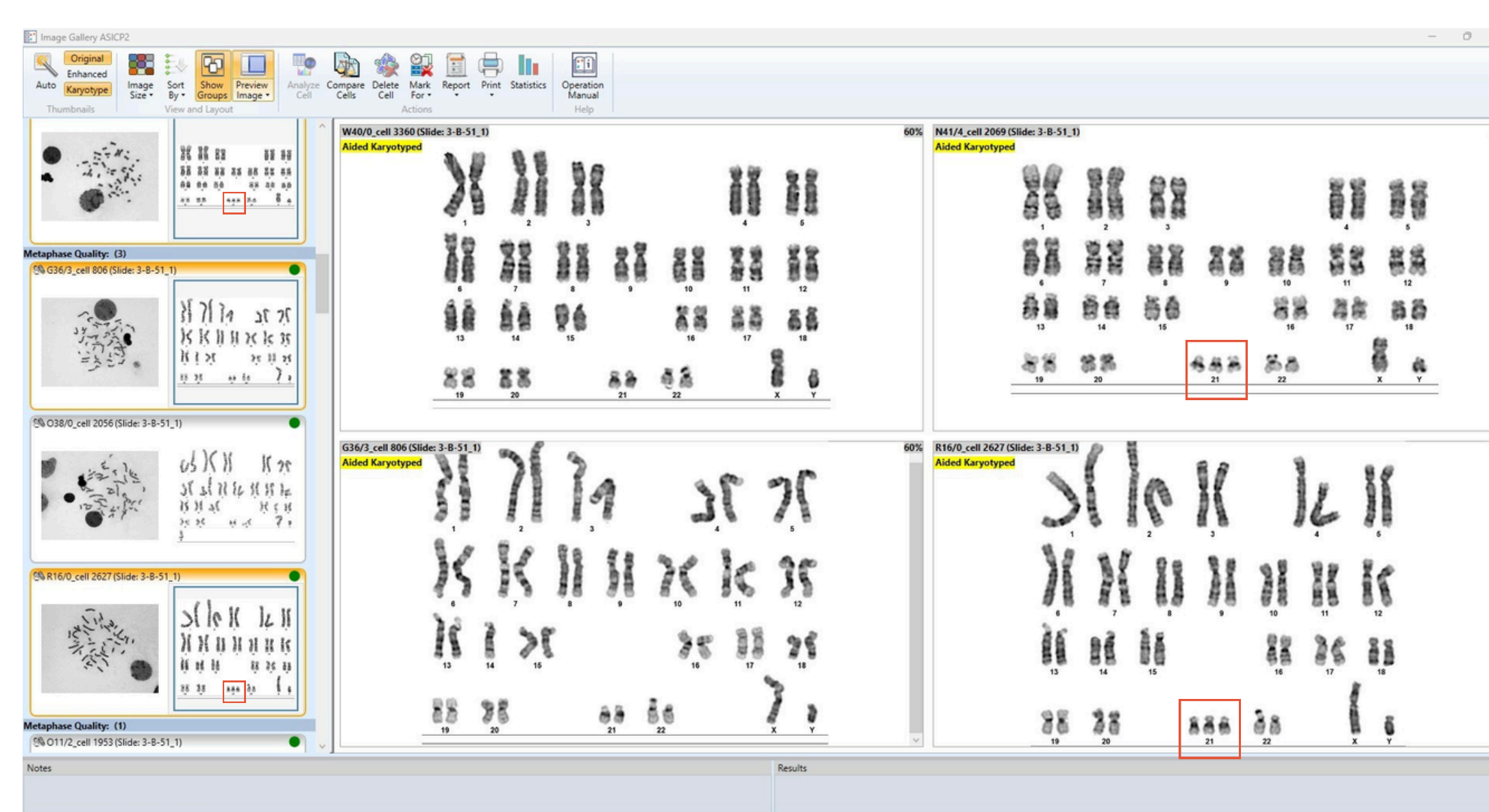


Figure 3
Image gallery of a mosaic Down syndrome case featuring cells and corresponding suggested karyograms automatically prepared by the AI-based utility, ready for review and analysis.

Results

- A total of five cases (**one peripheral blood and four bone marrows**) with five cells from each were included in this evaluation.
- Of the **25 metaphases analyzed**, the five PB cells from a mosaic Down syndrome case displayed normal karyotypes, while three BM cases were normal and two were abnormal.
- One of the abnormal BMs showed a **composite karyotype** of 59~62, <3n>,XXY,+Y,-2,-3,4,del(5)(q22q35),?t(6;22)(p21.3;q13),-7,-9,-12, del (13)(q12q22),-14,16,del(17)(p11.2),del(20)(q11.2q13.3) in five cells.
- The second abnormal BM was **mosaic**, with four normal cells and one showing 46,XY,del(7)(q22q34).
- The average karyotyping time for the 25 cells was **1.5±1.4 minutes** (range 0.1-6.4) when using the AI-powered HiBand add-on system compared to **4.6±1.8 minutes** (range 1.7-9.1) with the Cytolinsight GSL karyotyping application (p<0.0001) (Figure 2).
- Average chromosome analysis and karyotyping time for the **14 normal BM cells** was 1.3±0.8 minutes with the AI-based HiBand add-on compared to 4.5±1.0 minutes with the current non-AI Cytolinsight GSL software.
- Similarly, the average time to analyze the **6 abnormal BM metaphases** was 2.8±2.0 minutes with the AI-based add-on compared to 6.8±1.7 minutes with the current non-AI software.
- Finally, the average time to analyze the **5 normal PB metaphases** was 0.3±0.2 minutes with the AI-based add-on versus 2.5±0.5 minutes with the current non-AI karyotyping application.

- A **rapid review of the gallery** displaying all AI-based suggested karyograms for this PB case allowed immediate confirmation of **mosaic Down syndrome**, as evident in the additional cells not selected for detailed analysis in this evaluation (Figure 3).

Conclusions

- The evaluated AI-based, scanner-agnostic karyotyping add-on utility **significantly reduced metaphase analysis and karyotyping time** compared to the non-AI software integrated within the scanning system.
- The observed threefold decrease in analysis time across both normal and abnormal samples highlights the potential of AI-assisted tools to **enhance laboratory efficiency** without reducing accuracy.
- These findings support the **integration of AI-powered add-on applications into existing imaging platforms** as a cost-effective strategy to modernize cytogenetic laboratories and improve turnaround times.

Disclosures

JL, EAS, CAF, MLJ, MBL, MGK and MDK have no disclosure. OF, YAG and CS are employees of Applied Spectral Imaging.

