Preliminary Experience With AI-Based Karyotyping In Peripheral Blood Specimens

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Background and Introduction

- Cytogenetics laboratories are challenged by the **reduction of highly experienced workforce** with expertise in chromosome analysis and karyotyping.
- To address this drawback, **artificial intelligence (AI)** has been recently integrated into karyotyping software to reduce turnaround time.
- Using AI karyotyping, time to results has been reported to be reduced by 50% to an average of **32 minutes for both peripheral blood** and **bone marrow cases** (1-5).
- The goal of the present evaluation was to assess the turnaround time in an **independent laboratory** and determine possible **impact of band resolution**.

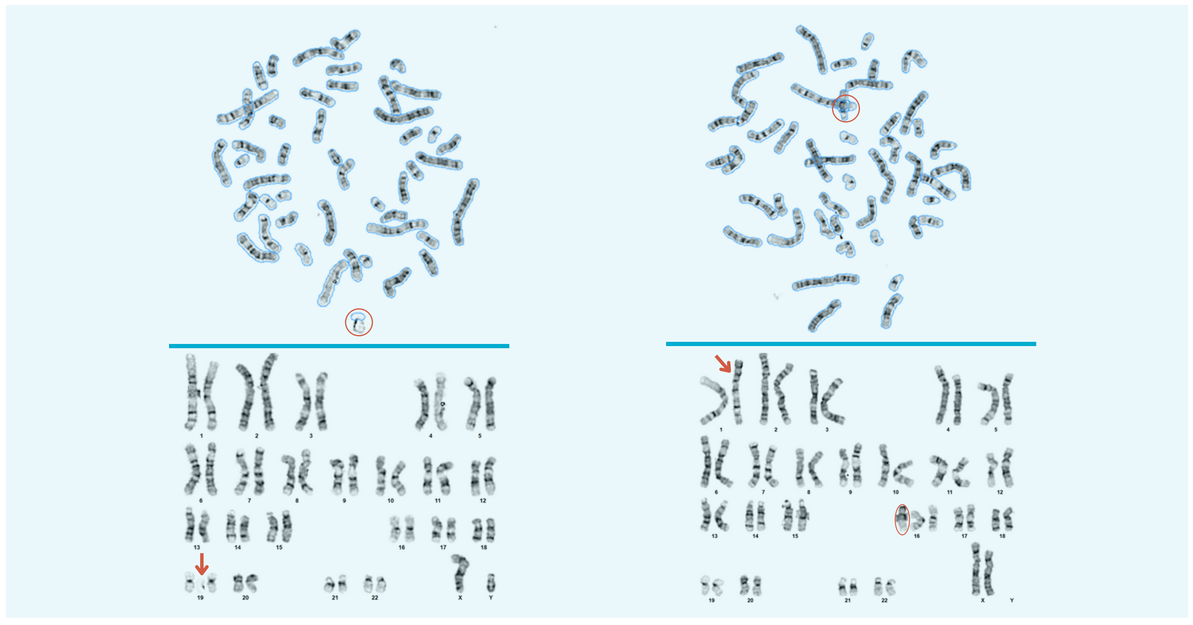

Figure 1
Representative examples of automatically segmented metaphases (top) and automatically placed chromosomes in karyograms (bottom).
Left: metaphase with estimated lower resolution. Right: metaphase with estimated higher resolution.

Methods

- **G-banded slides of peripheral blood** samples were prepared by standard methods and scanned using the HiBand system (Applied Spectral Imaging).
- For each slide, **thirty metaphases**, automatically selected by the system at 10X, were captured at 100X.
- Computer-aided **AI-powered karyograms were generated** based on a predefined range of detected objects (35-49).
- Cells were classified according to their band resolution **(high: >550 and low: <500)**.
- Three cytogeneticists with **different degree of experience with the system** reviewed the suggested karyograms.
- **Time required to correct each karyogram** was recorded.
- Statistical significance was assessed using the **Mann-Whitney U test**. A p-value < 0.05 was considered significant.

Results

- **Thirty G-banded blood cases** were included in this analysis for a total of **62 slides**.
- Thirty metaphases were scanned at high magnification for each slide, resulting in a total of **1,860 metaphases**.
- Computer-aided karyograms were automatically generated for 1,749 of the scanned metaphases **(94%)**.
- **Hundred-fifty cells** (5 per case) were selected by the users for analysis. Among these cells, **68 had high band resolution** (45%) and **82 had low band resolution** (55%).
- Across all users, the average time spent on each case was **24±9 minutes** (range 9-35 minutes).
- In average across all users, **no significant difference** was encountered between time spent on metaphases with high and low band resolution.
- However, for the user most experienced with the system, a shorter average analysis time was reported for **cells with lower band resolution** compared to cells with higher band resolution (2.2±0.7 vs 2.7±0.8, p=0.01).

Conclusions

- The turnaround time observed in this evaluation is **comparable to earlier reports** using AI-based chromosome analysis on peripheral blood specimens.
- In average, band resolution was **not found to significantly impact the time spent** on each metaphase.
- However, a follow-up investigation including users with different levels of experience is warranted in order to validate this **preliminary observation**.

References

1. Rosenblum L et al. Genes 2025, 16(6):685
2. Taghiyev A et al. Genetics in Medicine Open 2024, 2(1):101575
3. Burnside R et al. Genetics in Medicine Open 2025, 3(2):103053
4. Burnside R et al. Genetics in Medicine Open 2025, 3(1):101918
5. Steele C et al. Genetics in Medicine Open 2025, 3(2):103033

Disclosures

JB, JH, HP, AC, AV and MM have no disclosure.
EZ and YG are employees of Applied Spectral Imaging.