# New Q-Band Artificial Intelligence - Based Chromosome Analysis and Karyotyping
## a Pilot Evaluation

Liz Pardo, BSc, MSc, PhD(c) [1], Ivan Acosta, BSc[1], Claudia Acosta, BSc [1], Carolina Villamizar, BSc [1],
Cristina Steele, CG (ASCP), MBA [2], Einat Zindany, BSc [2], Yael Glickman, PhD, MBA [2]

1 Cytogenomics Laboratory - Genomics Sciences Unit, COLCAN-ANNAR; 2 Applied Spectral Imaging, Carlsbad, CA, USA

## Introduction

- The **Q-banding karyotyping method** is broadly used to identify individual chromosomes and their structural anomalies. In this technique, Quinacrine stain causes the AT-rich regions of the chromosomes to fluoresce more brightly than the GC-rich regions, therefore creating Q-bands.

- Recent advancements in artificial intelligence (AI) have been integrated into **G-banded karyotyping platforms,** significantly reducing the number of manual interventions required to correct a metaphase when compared to conventional digital imaging systems that rely on traditional image processing technologies.

- Multicenter studies performed on G-banded specimens have reported **91% correct chromosome segmentation** and **97% correct chromosome placement** when using new AI capabilities. (1-5)

- The present pilot evaluation reports preliminary assessment of a new **AI-based Q-banded karyotyping** application.
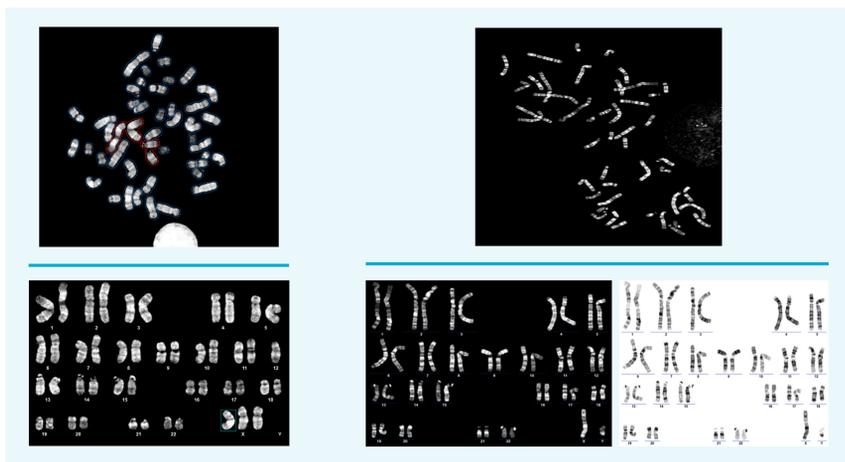


**Figure 1**
Representative examples of automatically segmented metaphases (top) and automatically placed chromosomes in karyograms following manual segmentation adjustments (bottom)
Left: bone marrow. Right: peripheral blood. Errors are highlighted).

## Methods

- Standard cytogenetic cell cultures were processed and spread onto microscope slides for staining. **Q-banding slides were scanned and analyzed** using the **HiBand system** (Applied Spectral Imaging).

- Metaphases, automatically identified by the system at 10X magnification, were captured at 100X. A newly developed AI-based computer-aided karyotyping application was used to automatically analyze selected cells.

- System-suggested aided karyograms were generated based on a predefined range of detected objects (35-49).

- Aided karyograms were compared with the karyotype table prepared by certified cytogenetics professionals to assess the number of correctly segmented and classified chromosomes.

- The number of manual adjustments required to correct the automatically suggested karyograms were then evaluated.

## Results

- **Ten Q-band peripheral blood and ten Q-band bone marrow metaphases** with an average of 9±6 overlaps and/or touching chromosomes were included in this pilot evaluation.

- Aided karyograms were generated for **85% of the metaphases**, representing a total of 782 chromosomes.

- The system **automatically segmented 94%** of the blood chromosomes (390 out of 413) and **79%** of bone marrow chromosomes (292 out of 369).

- Following segmentation correction, 408 blood chromosomes **(99%)** and 351 bone marrow chromosomes **(95%)** were **correctly placed** in the karyotype table.

- For both cell types, 98% of the correctly placed chromosomes were also correctly oriented.

- In average, **3±2 manual adjustments** were necessary to correct both segmentation and classification of peripheral blood metaphases, and **10±6 manual adjustments** were necessary to correct bone marrow metaphases.
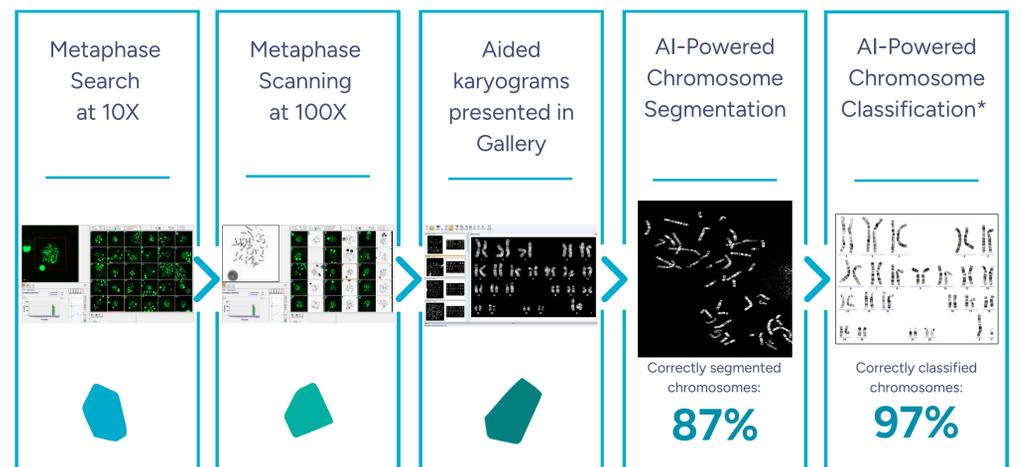


**Figure 2**
Representation of the Q-banded karyotyping workflow
* Following manual adjustments to correct automatic segmentation

## Discussion and Conclusions

- This pilot evaluation is the first report of a new AI-powered software for the analysis of Q-banded stained slides.

- The AI software **correctly segmented 87%** of peripheral blood or bone marrow chromosomes in metaphases containing an average of 9 overlaps and/or touching chromosomes.

- In addition, **97% of the chromosomes were correctly placed** in the karyotype table following segmentation correction.

- These results are **aligned with accuracy reported for G-banded specimens**.

- This pilot evaluation should be expanded to further confirm these preliminary results.

**References**
1. Taghiyev A et al. Genetics in Medicine Open 2024, 2(1):101575
2. Burnside R et al. Genetics in Medicine Open 2025, 3(2):103053
3. Burnside R et al. Genetics in Medicine Open 2025, 3(1):101918
4. Steele C et al. Genetics in Medicine Open 2025, 3(2):103033
5. Rosenblum L et al. Genes 2025, 16(6):285