

Diagnostic Impact of Karyotype Resolution in the Investigation of Balanced Rearrangements in Infertility and Miscarriage

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INTRODUCTION

Constitutional karyotyping remains a fundamental tool in the investigation of infertility and recurrent pregnancy loss, enabling the detection of numerical and structural chromosomal abnormalities. Through the evaluation of G-banded metaphase chromosomes, classified by size, centromeric position, and banding pattern, balanced rearrangements that may underlie reproductive failure can be identified. The diagnostic yield of this technique is closely related to the resolution employed, with higher banding levels increasing the likelihood of detecting subtle chromosomal alterations.

OBJECTIVE

To demonstrate a case study showing a chromosomal alteration visible only at a resolution of 500/550 bands compared to the resolution of 400 bands.

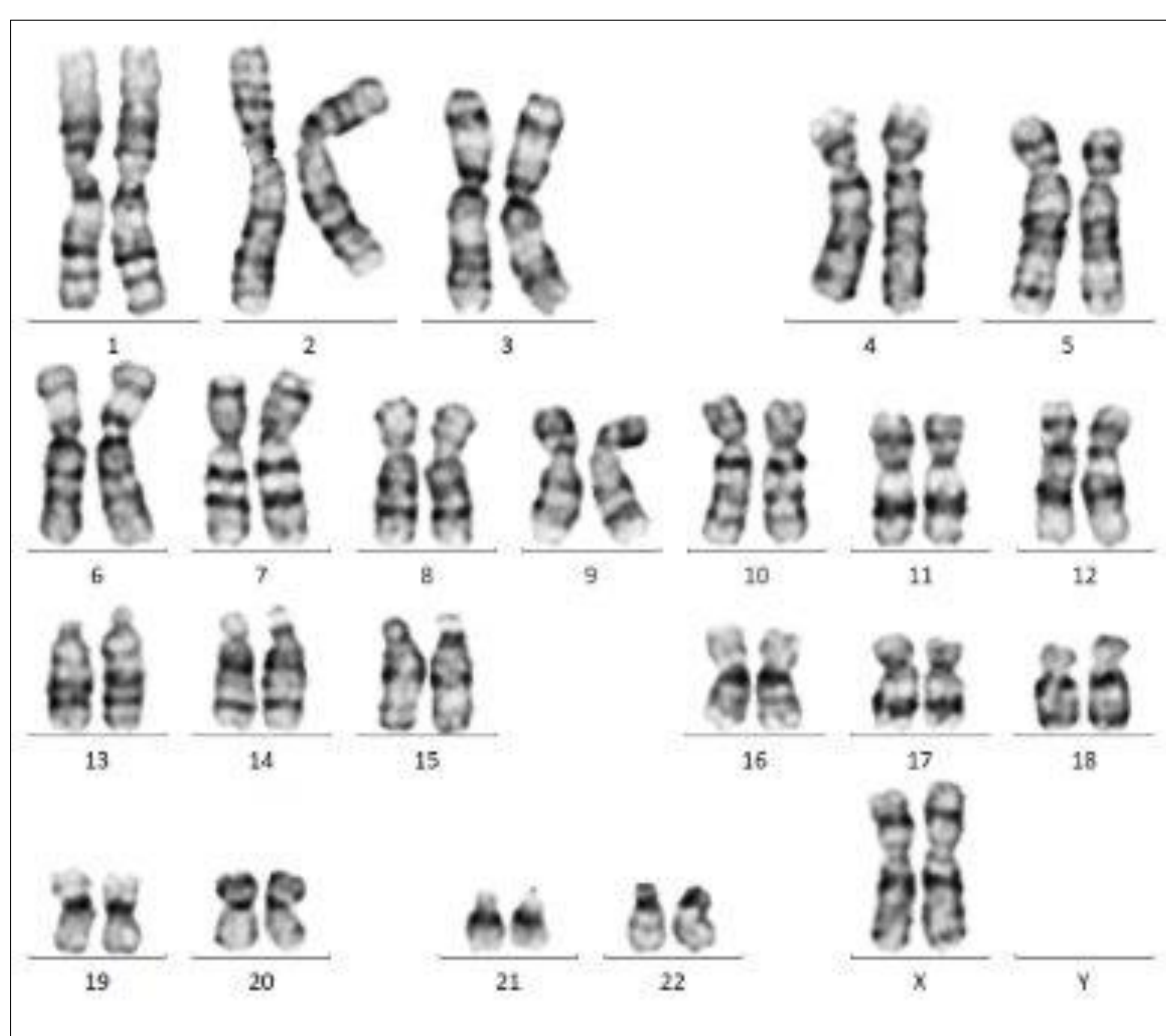
METHODS

A 36-year-old female patient with a history of infertility and miscarriage underwent constitutional karyotyping in our laboratory as part of the diagnostic workflow. The cell culture protocol included the use of a DNA intercalating agent to obtain well-spread metaphases, achieving a resolution between 400 and 550 bands.

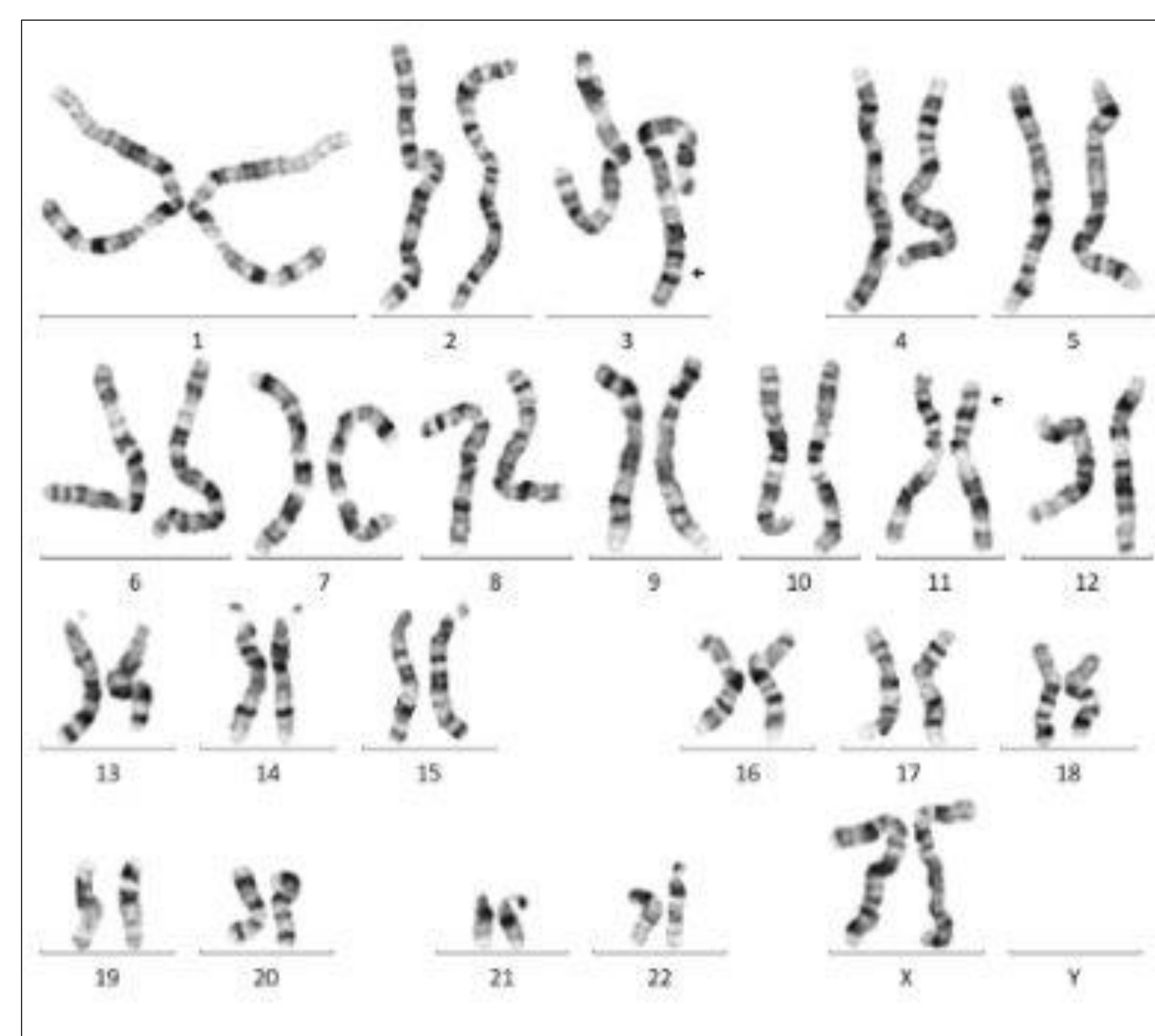
RESULTS/DISCUSSION

Due to its higher resolution, the 500/550-band karyotype allowed the detection of a balanced translocation: $t(3;11)(q26.2;p14)$. Analysis at 400-band resolution did not permit clear visualization of this rearrangement, highlighting the limitations of lower-resolution karyotyping in detecting subtle chromosomal alterations.

The analysis of metaphases at 500/550-band resolution enabled the identification of a clinically relevant chromosomal abnormality in the context of infertility and miscarriage, reinforcing the importance of adequate resolution in cytogenetic diagnostics. We emphasize the need for selecting appropriate resolution to ensure accurate diagnosis, avoid inconclusive results, and guide optimal clinical management based on precise cytogenetic findings.



400 RESOLUTION



500/550 RESOLUTION