

Experience of the Cytogenetics Laboratory Over One Year – Biology in Healthcare

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INTRODUCTION

Karyotyping is an important clinical cytogenetic method that enables detection of numerical and structural chromosomal abnormalities and provides valuable diagnostic information, particularly in reproductive medicine.

OBJECTIVE

To present the experience of the cytogenetics laboratory in routine diagnostics in healthcare and to highlight the role of biology and laboratory expertise throughout the diagnostic process – from sample processing and chromosome analysis to interpretation and reporting of cytogenetic findings.

MATERIALS AND METHODS

A retrospective analysis included data from 252 patients whose peripheral blood was received for karyotyping between April 2023 and December 2024.

Patient age and sex, indication for testing, karyotyping results, and turnaround time from sample receipt to report issuance were analyzed.

Cytogenetic analysis included:

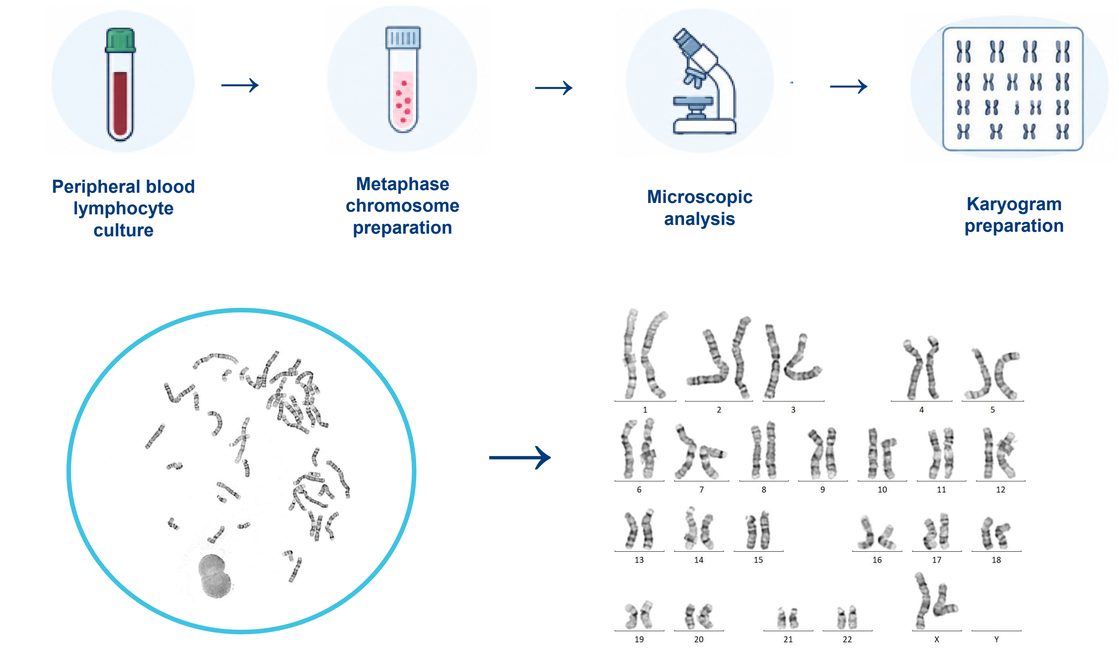


Figure 1. A metaphase spread image and the corresponding karyogram.
Normal female karyotype (46,XX).

KARYOTYPING IN PRACTICE

Karyotyping provides a genome-wide view at the cellular level and detects numerical and structural chromosomal changes. Despite advances in molecular methods, it remains indispensable in routine diagnostics and is especially important in the assessment of chromosomal abnormalities associated with reproductive outcomes.

RESULTS

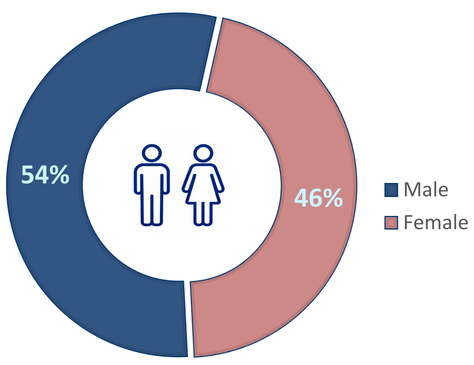


Figure 2. Gender distribution of participants.
Among the 252 patients included in the analysis, 137 (54.4%) were male and 115 (45.6%) were female.

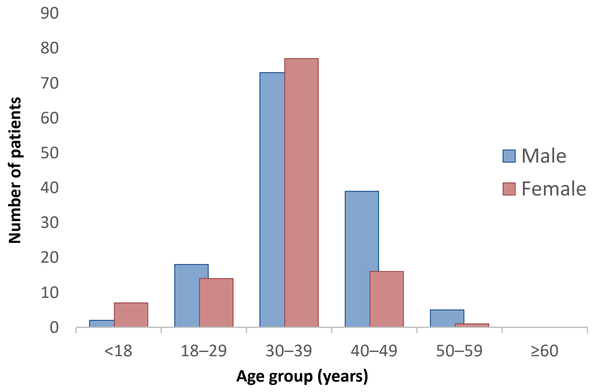


Figure 3. Age and sex distribution of patients.
Patients aged 30–39 years were the most represented age group in both sexes. The mean age was higher among males than females (37.1 vs. 33.6 years), with a greater proportion of males in the older age groups.

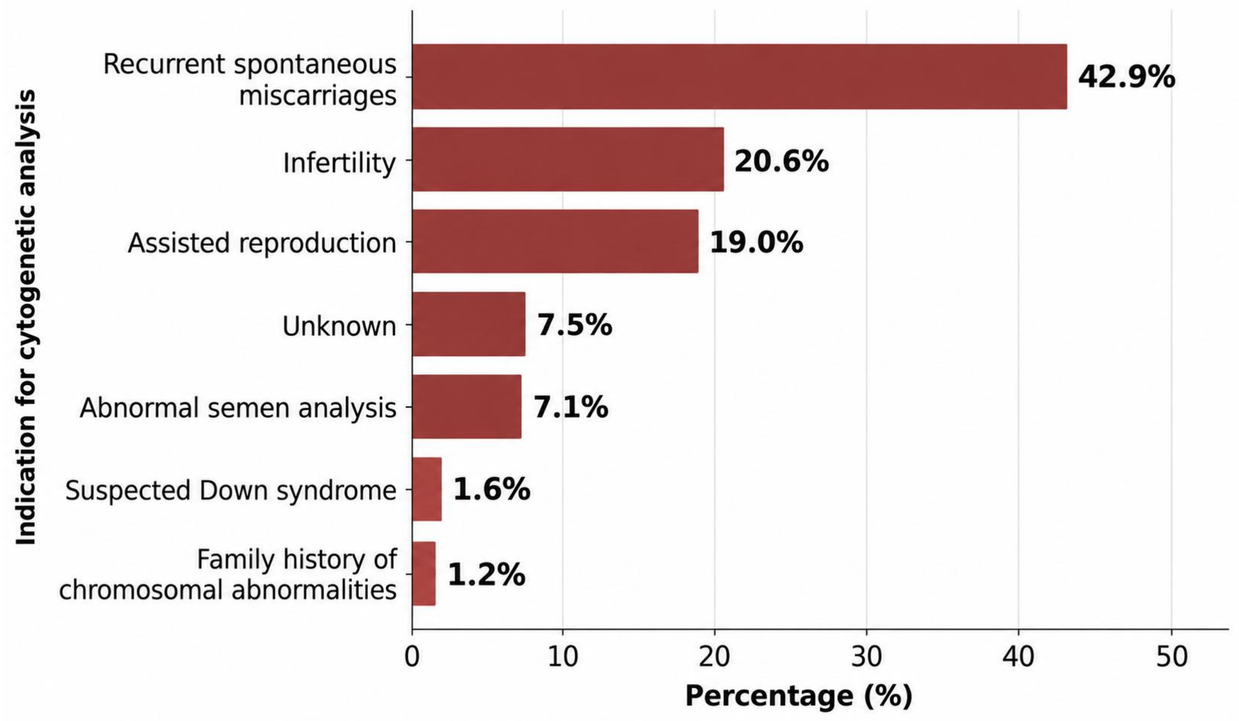


Figure 4. Indications for karyotyping.
Recurrent pregnancy loss was the most common indication for karyotyping, accounting for 42.9% of 252 patients, followed by infertility (20.6%) and assisted reproduction (19.0%). Abnormal semen analysis accounted for 7.1% of indications, while other indications were less frequently represented.

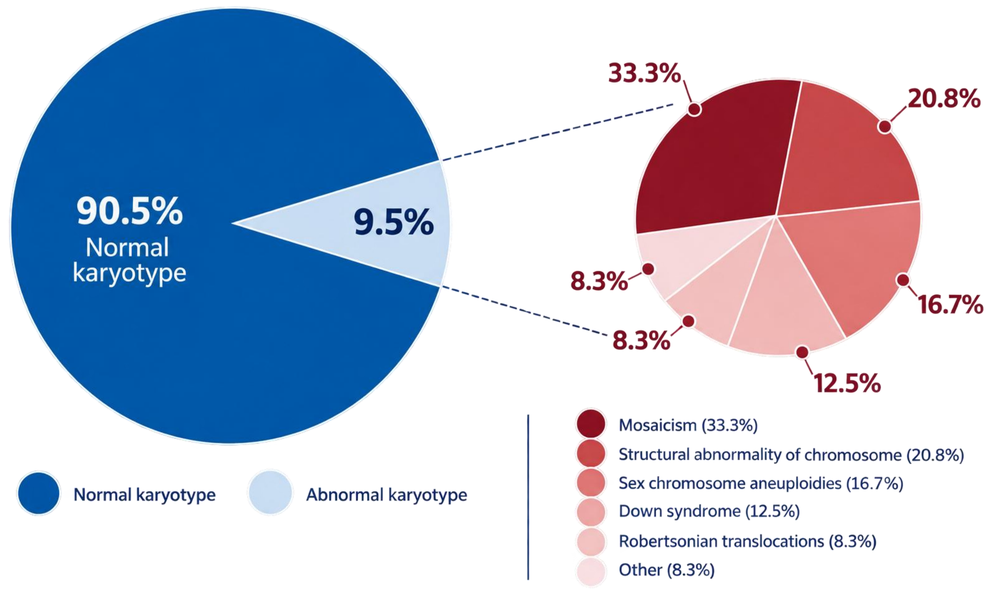


Figure 5. Karyotype findings.
The majority of patients had a normal karyotype (90.5%), while chromosomal abnormalities were identified in 9.5% of cases. Among the abnormal karyotypes, mosaic karyotypes were the most frequently observed (33.3%), followed by structural chromosomal abnormalities (20.8%).

BIOLOGY IN HEALTHCARE

The expertise of biologists is essential throughout the cytogenetic diagnostic process. From lymphocyte culture and metaphase preparation to microscopic chromosome analysis and interpretation, biologists and laboratory professionals ensure that each step is technically valid and clinically meaningful. Their expertise connects laboratory findings with patient care and supports accurate reporting in reproductive medicine.

CONCLUSION

Karyotyping remains a valuable diagnostic method in clinical cytogenetics, complementing biochemical and molecular diagnostic approaches. This is especially important in the evaluation of patients with reproductive disorders. In this one-year experience, most patients had a normal karyotype, while chromosomal abnormalities were identified in 9.5% of cases.

Reliable cytogenetic diagnosis depends on a standardized laboratory workflow, careful chromosome analysis and appropriate interpretation of findings. The coordinated work of biologists, laboratory professionals and clinicians is essential for generating high-quality results and translating cytogenetic findings into meaningful diagnostic information for patient care.

REFERENCES
1. Silva M, et al. European guidelines for constitutional cytogenomic analysis. Eur J Hum Genet. 2019;27:1–16.
2. ESHRE Guideline Group. Recurrent Pregnancy Loss: Guideline Update 2022. ESHRE; 2023.
3. World Health Organization. WHO Laboratory Manual for the Examination and Processing of Human Semen. 6th ed. WHO; 2021.

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