

COMPARISON OF FLUORESCENCE IN SITU HYBRIDIZATION AND CONVENTIONAL KARYOTYPING IN THE DIAGNOSIS OF CHRONIC MYELOID LEUKAEMIA AT AN ACADEMIC LABORATORY

Chauke P^{1,2}, Dlova AN^{2,3}

¹ Department of Chemical Pathology, Sefako Makgatho Health Sciences University, Ga-Rankuwa, South Africa.

² Dr George Mukhari Academic Laboratory, National Health Laboratory Service, Ga-Rankuwa, South Africa.

³ Department of Haematological Pathology, Sefako Makgatho Health Sciences University, Ga-Rankuwa, South Africa.

BACKGROUND AND OBJECTIVES: Chronic myeloid leukaemia (CML) is a myeloproliferative neoplasm that is characterised by the presence of the BCR::ABL1 fusion gene. A previous study that reported on the laboratory profile of patients with CML in our laboratory did not elaborate on additional chromosomal abnormalities found in these patients. These abnormalities could indicate disease progression. The objectives of the study were to compare fluorescence in situ hybridization (FISH) and conventional karyotyping (CK) methods in the diagnosis of CML patients and to document additional chromosomal abnormalities reported on conventional karyotyping.

METHOD: A cross-sectional descriptive study. The study included patients diagnosed with CML from the year 2020 - 2024 at Dr George Mukhari Academic Laboratory. Archival data was obtained from the National Health Laboratory Services laboratory information system as well as a sample logbook in the cytogenetics laboratory. STATA 16.1 software package was employed to analyse the data.

RESULTS: The study population was n = 60. FISH successfully diagnosed CML in 100% of cases (~78% bone marrow and ~22% peripheral blood samples). In contrast, CK diagnosed 22% of CML cases. The difference between these two methods in CML diagnosis was statistically significant for bone marrow samples ($p < 0.001$), in contrast to peripheral blood samples. CK detected additional chromosomal abnormalities such as trisomy 8, trisomy 19, inv(3) (q21q26), del(13)(q14), and variant three-way translocations, which are compatible with clonal evolution in CML.

CONCLUSION: This study demonstrated FISH diagnostic efficiency in CML diagnosis, although CK remains useful for detecting additional chromosomal aberrations and clonal evolution. We recommend that future studies cover a longer study period and to determine the association between the chromosomal abnormalities and disease progression or overall survival in our patients.

