Cytogenetic Profiles and Chromosomal Abnormalities in Patients with Anaemia and Concomitant Cytopenias: A Cohort Study of 260 Patients

Ana Franjević, Ivana Franić Šimić, Iva Semren, Sanja Davidović-Mrsić

University Hospital Centre Zagreb, Department of Clinical Diagnostics, Department of Cytogenetics

INTRODUCTION

Background: Cytogenetic abnormalities play a critical role in the pathogenesis, diagnosis, and prognosis of haematological disorders presenting with peripheral cytopenias. While isolated anaemia is a frequent clinical finding and can be of various origin, its presentation alongside additional cytopenias may signal underlying bone marrow failure syndromes or clonal haematological malignancies.

Purpose: To evaluate the prevalence and spectrum of chromosomal abnormalities in a cohort of patients presenting with anaemia, with or without accompanying peripheral cytopenia(s).

Methods: We conducted a retrospective analysis of 260 patients presenting with anaemia alone or combined with other cytopenias, pooled from a single-centre database between January 2025 and March 2026. All patients underwent comprehensive clinical evaluation and bone marrow cytogenetic analysis via conventional karyotyping and/or fluorescence in situ hybridization (FISH) to screen and identify possible clonal chromosomal abnormalities.

RESULTS

Cohort Distribution: The study population (N = 260) was categorized into two primary clinical phenotypes: isolated anaemia (N = 181, 69.6%) and anaemia with concomitant cytopenias (bicytopenia or pancytopenia; N = 79, 30.4%).

Cytogenetic Findings: Among patients with isolated anaemia, 8% exhibited heterogeneous chromosomal abnormalities, showing an equal distribution of structural and numerical aberrations, with loss of chromosome Y (LOY) predominating in the male subgroup. Only 4% of patients with multilineage cytopenias displayed clonal abnormalities; these were less heterogeneous, predominantly structural in nature, and of myeloid significance (specifically 5q deletion and 7q deletion) (Figure 2).

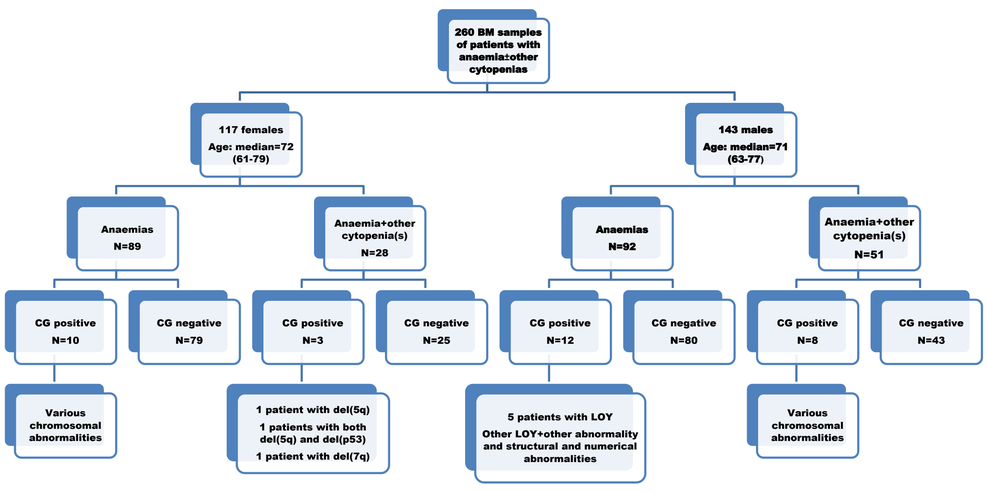

Figure 1. Flowchart of patients included in the study.

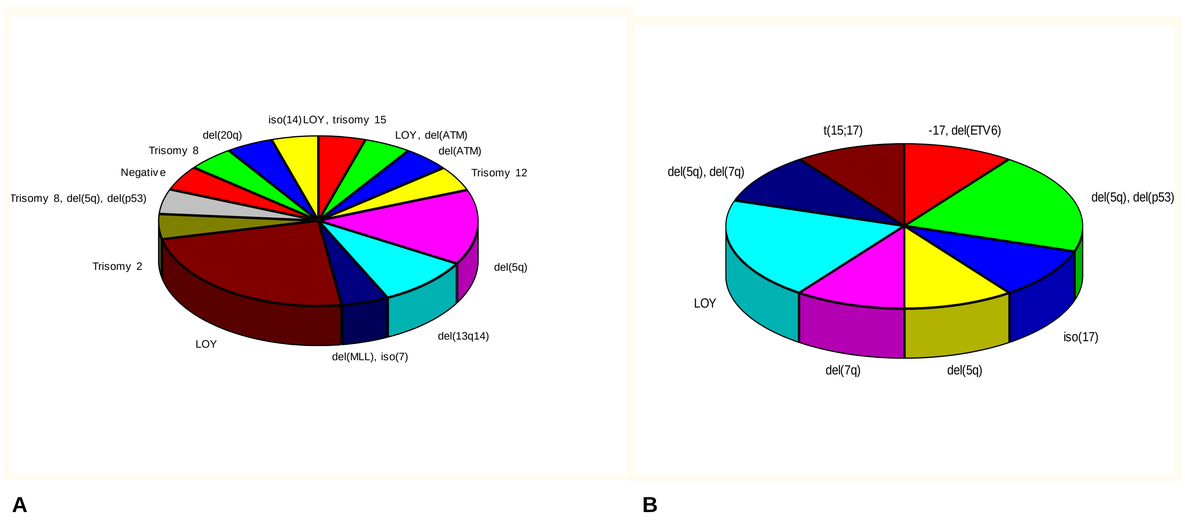

Figure 2. A: Chromosomal abnormalities in patients with anaemia; N=181.
B: Chromosomal abnormalities in patients with anaemia and concomitant cytopenias; N=79.

CONCLUSION

Cytogenetic abnormalities in anaemia alone appear to be more diverse that in group with the additional cytopenias. Predominantly there is LOY, that is common in aging males, but also more frequent in men with hematological disorders than without. There is general understanding that finding of ≥75% of metaphases with LOY, can be disease-related clonality, so those patients are regularly additionally checked on FISH. Other abnormalities are diverse, both structural and numerical.

In the group with concomitant cytopenias, abnormalites are predominantly myeloid in nature (alongside LOY in males), so cytogenetics was the early warning for follow-up and additional diagnostics.

Although the overall prevalence of chromosomal abnormalities in this cohort is low, cytogenetic evaluation remains a valuable diagnostic tool to identify high-risk subgroups.

Early cytogenetic screening is still necessary to guide to accurate diagnosis, risk stratification, and targeted therapeutic planning in clinical haematology.

REFERENCES

Wiktor A, Rybicki B, Piao ZS, Shurafa M, Barthel B, *et al.* Clinical significance of Y chromosome loss in hematologic disease. Genes, Chromosomes and Cancer. 2000; 27. 11-16.
Xie Z, Nanaa A, Saliba AN, et al. Treatment outcome of clonal cytopenias of undetermined significance: a single-institution retrospective study (https://pubmed.ncbi.nlm.nih.gov/33649321/). Blood Cancer J. 2021;11(3):43.
Zhang Y, Schlegelberger B. Cytogenetics in haematology. Best Pract Res Clin Haematol. 2024;37(2):101553.
Zhang LJ, Shin ES, Yu ZX, Li SB. Molecular genetic evidence of Y chromosome loss in male patients with hematological disorders. Chin Med J (Engl). 2007; 20;120(22):2002-5.

AKNOWLEDGEMENTS

The authors thank all the lab staff for technical assistance, sample processing, and valuable insights during this research.