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Additional Chromosomal Abnormalities In Patints with Chronic Myeloid Leukemia- Data From A Tertiary Care Center In Pakistan

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Introduction

Alongside Philadelphia chromosome, additional chromosomal abnormalities (ACAs) have also been identified in CML patients. Different ACAs impact the prognosis and treatment of Chronic Myeloid Leukemia differently[1]. In Pakistan, the disease burden of CML is quite high, yet there is a lack of scientific evidence regarding ACAs. This study focuses on the prevalence of ACAs in CML and association of ACAs with important clinical and hematological parameters.

Aim

To observe the prevalence of various additional cytogenetic abnormalities in chronic myeloid leukemia and their association with important clinical and hematological parameters.

Material and Methods

- Design: Retrospective Cross-sectional study
- Setting: Department of Pathology and Laboratory Medicine, Aga Khan University, Karachi.
- Data duration: January 2018- December 2021
- We retrieved medical records of 2796 CML patients, who underwent bone marrow karyotyping, at our lab Frequency of various ACAs was noted. Their association with blood counts and clinical parameters (splenomegaly, disease phase, age, gender) was also statistically analyzed in SPSS.

Results

- Total 2796 patients had t (9;22) on karyotyping.
- ACAs were found in 88 (3.14%) patients.
- After reviewing bone marrow biopsy reports and clinical history provided, we excluded 12 cases because these were B cell acute lymphoblastic leukemia with t (9;22). So total 76 (2.71%) CML patients were identified with ACAs.
- M: F ratio = 1.3:1 with ages ranging from 10 to 75 years and the median age was 36.5 years..
- Most of the patients (71%) were in chronic phase, while 15.8% and 11.8% patients were in accelerated phase and blast crisis respectively.
- Major route ACAs were noted in 19.7% patients, Minor route ACAs in 21.1% patients, variant translocations in 47.4% patients and mixed in 11.8% patients.
- Overall, the most prevalent major route abnormality was extra Philadelphia chromosome noted in eighteen patients.
- Splenomegaly, phase of CML on bone marrow biopsy, and timing of ACA detection with reference to CML diagnosis was found to have statistically significant difference among different ACA groups.

Table I: Comparison of clinical parameters among patients with different ACA groups

Parameters		Major route	Minor route	Mix	Variant translocations	p- value
Gender	Male	11	10	6	17	
	Female	4	6	3	19	0.312
Age groups (in years)	<18	1	2	0	3	
	19-30	4	1	2	8	
	31-50	9	8	5	21	
	>50	1	5	2	4	0.608
Splenomegaly	Yes	7	4	6	24	
	No	8	12	3	12	0.034
Phase of CML on BM biopsy	CP	11	9	3	31	
	AP	3	2	3	4	
	BC	1	5	3	0	
	UA	0	0	0	1	0.016
Timing of ACA detection	At the time of CML diagnosis	9	9	4	33	
	Previously diagnosed with CML	6	7	5	3	0.011

Table II: Phase-wise comparison of hematological parameters among patients with different ACA groups

Parameters	Type of ACA	Overall		Chronic Phase		Accelerated Phase		Blast Crisis	
		Mean	p-value	Mean	p-value	Mean	p-value	Mean	p-value
Hb (g/dl)	Major route	8.6	0.598	9.1	0.634	7.8	0.378	6.3	0.255
	Minor route	9.0		9.5		9.9		7.9	
	Mixed	9.7		11.6		7.3		10.3	
	Variant translocations	9.2		9.4		7.5		-	
TLC ($x10^9$/L)	Major route	159.9	0.049	182.3	0.234	116.4	0.712	44	0.726
	Minor route	140.5		179.1		37.8		112	
	Mixed	160.6		186		120.6		175.3	
	Variant translocations	244		253.8		227.7		-	
Platelets ($x10^9$/L)	Major route	207.8	0.003	223.6	0.005	110	0.541	327	0.434
	Minor route	678		603.8		335.5		948.6	
	Mixed	427.8		142		999		142.6	
	Variant translocations	339.4		354.2		301.7		-	

Hb= Hemoglobin, TLC= Total Leucocyte Count, PB= Peripheral Blood, BM= Bone Marrow

Discussion

- ACAs detected in 2.7% of CML patients, lower than local and international reports[2].
- Patients were younger (median 36.5 yrs) with slight male predominance.
- ACA emergence increased with disease progression (highest in blast crisis).
- Extra Philadelphia chromosome was most common major ACA; variant translocations dominated overall.
- Splenomegaly frequent; platelet count differences significant.
- Findings highlight prognostic role of ACAs and provide important local data.

Conclusion

ACAs in CML are relatively uncommon (2.7%) in this population, but their type and timing correlate with disease progression. This study provides valuable local epidemiologic data and underscores the prognostic importance of ACAs for risk stratification in CML.

References

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