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The Intersection of Thalassemia Minor and Chronic Myeloproliferative Disorders: Clinical and Laboratory Implications

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INTRODUCTION

Thalassemia is a hereditary anemia caused by defects in hemoglobin production. Forms of thalassemia that generally present with mild anemia are called thalassemia minor. Chronic Myeloproliferative Disorders (CMPD) are clonal hematopoietic stem cell-derived disorders characterized by uncontrolled proliferation of differentiated myeloid cells. The coexistence of thalassemia and CMPD is extremely rare.

AIM

This study aims to explore the rare co-occurrence of thalassemia minor and CMPD, focusing on how thalassemia minor can obscure the diagnostic markers of CMPD, including hemoglobin levels, and the subsequent effects on clinical and laboratory outcomes.

METHOD

The investigation involved 32 patients diagnosed with thalassemia minor alongside CMPD at Cukurova University Faculty of Medicine's Department of Hematology between January 2003 and January 2024.

The diagnostic criteria utilized were the "2020 ELN Criteria" for CML and the "WHO Classification of Myeloid Neoplasms and Acute Leukemia, 2016 Revision Criteria" for BCR-ABL negative CMPDs.

RESULTS

Our findings highlight a spectrum of CMPD manifestations among the patients, with a significant portion displaying symptoms and signs such as hepatomegaly and splenomegaly.

A pivotal aspect of this study was the identification of the JAK2 V617F mutation in the majority of patients, underscoring the prevalence of CMPD despite the potential masking effect of thalassemia minor on certain hematologic markers.

Variable	All patients (N=32)	Patients with CML (n=5)	Patients with PV (n=9)	Patients with ET (n=11)	Patients with MF (n=5)
Hgb (g/dL), median (min-max)	13,70 (8,30-17,30)	11,80 (9,40-13,80)	15,3 (14,10-17,30)	12,60 (8,30-15,10)	10,30 (9,80-17,30)
Plt (x10³/µL), median (min-max)	590,5 (144-1.564)	262 (242-1.385)	337 (144-629)	797 (534-1.564)	453 (160-1.058)
WBC (/µL), median (min-max)	12.050 (4.500-24.400)	7.100 (4.500-15.920)	11.700 (7.700-18.530)	11.300 (7.400-19.300)	12.900 (4.700-24.300)
Constitutional symptoms (n=28; %87,5)	12 (37,5)	1 (20)	3 (33,3)	3 (27,3)	4 (80)
Hepatomegaly (n=29; %90,6)	17 (53,1)	1 (20)	6 (66,7)	5 (45,5)	4 (80)
Splenomegaly (n=29; %90,6)	19 (59,4)	0 (0)	7 (77,8)	6 (54,5)	5 (100)
JAK2 mutation status (n=26; %81,3)	24 (75)	2 (6,3)	8 (88,9)	9 (81,8)	4 (80)
CALR mutation status (n=10; %31,3)	3 (9,4)	0 (0)	0 (0)	1 (9,1)	2 (40)
MPL mutation status (n=4; %12,5)	0 (0)	0 (0)	0 (0)	0 (0)	0 (0)
BCR-ABL* (n=20; %62,5)	5 (15,6)	5 (100)	0 (0)	0 (0)	0 (0)
Triple-negative and BCR-ABL	3 (9,4)	0 (0)	1 (11,1)	1 (9,1)	0 (0)

All patients n=32		n (%)
Sex	Female	19 (59.4)
	Male	13 (40.6)
Diagnosis	ET	11 (34.4)
	PV	9 (28.1)
	CML	5 (15.6)
	PMF	1 (3.1)
	PPV-MF	1 (3.1)
	PET-MF	3 (9.4)
	PV+ET	2 (6.3)
JAK2 V617F mutation status	Positive	24 (92.3)
	Negative	2 (7.7)
Comorbidity	Yes	20 (62.5)
	No	12 (37.5)
Diabetes Mellitus		2 (6.3)
Hypertension		17 (53.1)
Coronary Artery Disease		9 (28.1)
Heart Failure		2 (6.3)
Symptoms	Symptomatic	12 (42.9)
	Asymptomatic	16 (57.1)
Hepatomegaly	Yes	17 (58.6)
	No	12 (41.4)
Splenomegaly	Yes	19 (65.5)
	No	10 (34.5)

CONCLUSIONS

The research illuminates the intricate relationship between thalassemia minor and CMPD, emphasizing the need for careful diagnostic consideration to ensure accurate identification and management of these patients.

This study contributes valuable insights into the complexities of diagnosing and treating individuals with concurrent thalassemia minor and CMPD, urging a nuanced approach to hematologic evaluation.

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