THE CLINICAL, PROGNOSTIC, AND THERAPEUTIC IMPLICATIONS OF MUTATIONAL PROFILES IN ESSENTIAL THROMBOCYTHEMIA: INSIGHTS FROM A RETROSPECTIVE STUDY AT A LEADING SINGLE CENTER

N. Ben Sayed[1,2], R. Aidli[1], A. Ben Amor[1], A. Rahal[1], N. Sassi[1], M. Guermazi[1], Z. Kmira[1], H. Regaieg[1] and Y. Ben Youssef[1]

1. Hematology Department, Hospital Farhat Hached, Sousse, Tunisia
2. Ibn Jazzar Medical College, Sousse, Tunisia

INTRODUCTION

Essential thrombocythemia (ET) is a myeloproliferative neoplasm (MPN) characterized by significant phenotypic heterogeneity and a variable risk of life-threatening thrombotic events.

While driver mutations such as JAK2 V617F are established diagnostic markers, their role in predicting event-free survival (EFS) and long-term outcomes across diverse clinical cohorts remains a critical area of investigation.

AIM

To characterize the clinicobiological profile of ET and to evaluate the efficacy of the revised International Prognostic Score of Thrombosis for Essential Thrombocythemia (IPSET-thrombosis) score and mutational status in predicting patient outcomes.

METHOD

Retrospective study of 56 patients diagnosed with ET (WHO 2016 criteria) at Farhat Hached University Hospital, Sousse, Tunisia, from 2016 to 2021.

- Statistical analysis performed using SPSS, version 23.
- Kaplan-Meier method used for survival estimation.
- Log-rank test used to compare survival curves.

CONCLUSIONS

The JAK2 V617F mutation is a primary driver of clinical complications and reduced event-free survival (EFS) in ET.

The strong prognostic correlation of the revised IPSET-thrombosis score validates its utility as a critical tool for identifying high-risk patients who require intensive therapeutic monitoring to mitigate thrombotic risks.

RESULTS

The study included 56 patients with a mean age of 60.4 years and a female predominance (male/female ratio, 0.75); diagnosis was incidental in 82% of cases.

Molecular analysis identified the JAK2 V617F mutation in 60.7% of patients, while 39.3% were JAK2-negative.

According to the revised IPSET-thrombosis score, 46.4% of patients were classified as high risk, 12.5% as intermediate, and 41.1% as low or very low risk.

The JAK2 V617F mutation was significantly associated with thrombotic events (P=0.027), affecting 30.6% of the overall population.

The 6-year overall survival rate was 85%, but EFS was strongly influenced by mutational status: 6-year EFS was 100% in JAK2-negative patients versus 60% in JAK2 V617F carriers.

Similarly, 6-year EFS dropped from 100% in the low-risk group to 0% in the high-risk group by revised IPSET-thrombosis score (P=0.007).

Hydroxyurea was the primary cytoreductive agent, achieving a complete response in 49% of cases.

AT A GLANCE

56
patients with ET

60.7%
JAK2 V617F positive

46.4%
high risk (IPSET-thrombosis)

85%
6-year overall survival

100% vs 60%
6-y EFS, JAK2-neg vs JAK2+

0%
6-y EFS in high-risk IPSET group

Demographics & Presentation	Molecular Profile	Risk Stratification (IPSET)	Treatment	Outcomes
Patients: n=56	JAK2 V617F positive: 60.7%	High risk: 46.4%	Hydroxyurea: primary agent	6-y overall survival: 85%
Mean age: 60.4y	JAK2 negative: 39.3%	Intermediate risk: 12.5%	Complete response: 49%	6-y EFS, JAK2-negative: 100%
Male:Female ratio: 0.75	Thrombotic events: 30.6%	Low / very low risk: 41.1%		6-y EFS, JAK2 V617F+: 60%
Incidental diagnosis: 82%	JAK2+ vs thrombosis: P=0.027			6-y EFS, low-risk IPSET: 100%
				6-y EFS, high-risk IPSET: 0%
				IPSET risk vs EFS: P=0.007

JAK2 status, IPSET risk, and outcomes at a glance.

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CONTACT INFORMATION

Nesrine Ben Sayed – Hematology Department, Hospital Farhat Hached, Sousse, Tunisia – [add corresponding author email]

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