COMPARISON OF CLINICAL AND LABORATORY DATA, INCLUDING JAK-2 46/1 HAPLOTYPE, BETWEEN PATIENTS WITH IDIOPATHIC ERYTHROCYTOSIS AND POLYCYTHEMIA VERA.

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(Abstract release date: May 18, 2017) EHA Learning Center. Napolitano M. May 18, 2017; 182753
Label: Myeloproliferative neoplasms - Clinical

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Abstract:PB2039

Type: Publication Only

Background

Idiopathic erythrocytosis(IE) is a relatively rare finding characterized by an increased red blood cell mass without an identifiable cause. Diagnosis of IE is based on the exclusion of primary and secondary erythrocytosis including JAK2-wild-type polycythemia Vera (PV).

Aims

In the current study, we report clinical features and laboratory data able to discriminate IE from PV, at diagnosis

Methods

We have here analyzed clinical and laboratory parameters, including Jak-2 46/1 haplotype, from patients with a confirmed diagnosis of IE and PV, followed from January 2010 to December 2016. Data were statistically analyzed, nominal variables were compared with X_2 test and continuous variables with the Mann-Whitney test.

Results

Overall, 40 patients with IE and 93 patients with PV were included in the current analysis (Table 1). Splenomegaly and itch were reported only in one patient with IE. History of thrombosis and cardiovascular events was positive in one case with IE. Jak-2 (V617F) and exon 12 mutations were negative in all patients with IE, while Jak-2 46/1 haplotype was found at heterozygous state in 18 patients and at homozygous state in 2 patients with IE.

	PV	IE	p
Patients N.	93	40	
MALE N. (%)	59 (63,44%	38 (95%)	0.0001
FEMALE N (%)	34 (36.56%)	2 (5 %)	0.0001
MEDIAN AGE AT DIAGNOSIS, YEARS	66 (23 - 95)	58 (17 - 83)	0.007
SPLENOMEGALY N° (%)	38 (40,86%)	1 (2,5%)	0.000001
ITCH	36 (38,70%)	1 (2,5 %)	0.00000 1
MEDIAN WBC COUNT $X10_9$	9,3 (4,535,2)	8.1 (4,2-14,3)	0.03
MEDIAN HB g/dl	17,5 (15,1-21,0)	17,4 (16,1-19.1)	0.9
MEDIAN HT %	53,3 (48.4 - 64.3)	52,4 (48.2 - 55.3)	0.1
MEDIAN PLTS COUNT X 10_9	435.0 (270-1013)	216 (178.339)	0.001
V617 F OF JAK2 POSITIVE N. (%)	86 (92,47)	0	
JAK2 EXON 12 MUTATION N. (%)	2 (2,15%)	0	
HAPLOTYPE 46/1 OF JAK2, ETHEROZIGOUS	42 (45,16%)	18 (45,0%)	0,98
HAPLOTYPE 46/1 OF JAK 2, HOMOZIGOUS	25 (26,88%)	2 (5 %)	0.008
PATIENTS WITH CARDIOVASCULAR EVENTS OR THROMBOSIS N %	32 (34,4%)	1 (2,5%)	0.000008

Conclusion

In the current study, we highlight peculiar clinical and laboratory findings of IE, in comparison with Polycythemia Vera. As shown by available studies, Hb and HCT level do not easily discriminate between the two categories of patients while gene panels may be useful to improve diagnostic accuracy of IE. We have here first observed the presence of Jak-2 46/1 haplotype in approximately half patients with IE, even in absence of JAk-2 mutations; the homozygous status was statistically different among PV and IE patients. The role of such association deserves further specific studies.

Session topic:16. Myeloproliferative neoplasms - Clinical

Keyword(s): Polycythemia vera, Haplotype analysis, erythrocytosis