Abstract: P2205

Title: SHOULD TRANSFERRIN SATURATION >45% OR HIGH SERUM TRANSFERRIN LEVEL BE THE ONLY REQUIREMENT FOR HH GENE MUTATION DETECTION?

Abstract Type: e-Poster Presentation

Topic: Iron metabolism, deficiency and overload

Background:

Hemochromatosis is the abnormal accumulation of iron in parenchymal organs, leading to organ toxicity. The diagnosis of hemochromatosis is based on clinical features of the disease. Most patients are asymptomatic and are diagnosed when elevated serum iron levels are noted on a routine chemistry screening panel or when screening is performed because a relative is diagnosed with hemochromatosis.

Aims:

In this study we reviewed reasons for mutation screening and patient characteristics in patients diagnosed with hereditary hemochromatosis (HH) gene mutations (HFE and other mutations).

Methods:

We retrospectively scanned the clinical and laboratory data of patients whose HH gene mutation was detected by polymerase chain reaction (PCR) in our center between January 2019 and January 2024, and by next generation sequencing (NGS) method in the last 3 years, in our electronic record system.

Results:

Of the total 108 pts, 74 were male (%68,5), 32 were female (%29,5). Median age was 52 yrs (18-77) (males 18-76, median 47,3, females 40-77, median 64,3). Age of diagnosis was significantly lower in males than females (p=0.012). Of the fifty three pts with polycythemia, 7 diagnosed polycythemia vera (with Jak-2 mutation), 46 were secondary polycythemia. Other dignoses were as follows, 8 essential thrombocythemia (%7,4), 7 diabetes mellitus (%6,5), 4 chronic hepatitis (2 of them chronic etilism), 4 malign lyphoproliferative disease , 4 solid tumor (%3,7 each), 2 acute leukemia in remission 2 connective tissue disease, 2 inflamatory bowel disease,, 2 acromegali, 2 beta thalasemia trait, 2 obesity, 2 family history of HH mutation (%1,9 each), otoimmune hemolytic anemia in remission, ITP, prolactinoma one of each (%0,9 each). Table 1 shows mutation distribution in polycythemia, normal Hb levels, and anemic patients. Transferrin saturation % (TS), serum ferritin (SF) levels (decreased, normal, increased) with mutation distribution are shown on Table 2. Anemia/polycytemia and TS% / SF levels had no relation with mutations distribution (p=0.1 and p=0.2)

Table1. HH gene mutations in patients who have normal, low or high hemoglobin levels.

HH gene mutations	**Patients with anemia (no:40)**	**Patients with Hgb level within normal range (no: 11)**	**Patients with polycithemia (no:53)**	**Total (no:104)***
C82Y heterozygous	3	1	2	6
C82Y homozygous	1	0	0	1
C82Y/c617-49g>a	0	0	1	1
C82Y/H63D	0	0	2	2
H10Q heterozygous	1	0	0	1

HH gene mutations	Patients with anemia (no:40)	Patients with Hgb level within normal range (no: 11)	Patients with polycithemia (no:53)	Total (no:104)*
H63D heterozygous	31	9	47	8
H63D homozygous	4	0	0	4
H63D/T217I	0	1	0	1
R23H heterozygous	0	0	1	1

* Four patients from another centers who have lost data

Table.2 HH gene mutations in patients who normal, low or high TS % and serum ferritin level.

Serum ferritin level	HH gene mutation	TS% >%45 (no:55)	%40>TS%<%45 (no:7)	TS%<%40 (n:41)	Total no:103
>N	C82Y heterozygous	0	0	1	1
	C82Y homozygous	1	0	0	1
	C82Y/H63D	1	0	0	1
	H10Q heterozygous	0	1	0	1
	H63D heterozygous	10	2	9	21
	H63D homozygous	1	0	0	1
	H63D/T217I	1	0	0	1
	Total	**14**	**3**	**10**	**27**
Within normal range	C82Y heterozygous	2	0	3	5
	C82Y/c617-49g>a	1	0	0	1
	C82Y/H63D	1	0	0	1
	H63D heterozygous	35	4	24	63
	H63D homozygous	2	0	1	31
	R23H heterozygous	0	0	1	
	Total	**41**	**4**	**29**	**74**
<N	H63D heterozygous	0	0	2	2
	Total			**2**	**2**

* Five patients who have lost data

Summary/Conclusion:

This group of pts consisted of chronic myeloproliferative disease patients and patients referred for polycytemia etiology diagnosis. It is concluded that HH gene mutations (HFE and others mutations) analysis had important impact for diagnosis. We concluded that HH gene mutation examinations can be performed in patients with normal TS and serum ferritin levels or in patients with anemia, in cases of clinical suspicion of HH and taking into account comorbid diseases.

Keywords: Hemochromatosis