SUPPLEMENTARY APPENDIX

Dynamics of mutations in patients with essential thrombocythemia treated with imetelstat

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Supplementary Appendix

Table S1. Patient baseline demographics.

Patients (N=18)	Median (Range) or N (%)
Age (years) >60 years	59.5 (21-83) 8 (44%)
History of thrombosis	5 (28%)
Sex Female Male	 10 (56%) 8 (44%)
Race Black/African American White	 2 (11%) 16 (89%)
Years since initial diagnosis	7.2 (0.3-24.9)
Platelet count (x10^3/μL)	788 (521-1359)*
Hematocrit	37.3 (26.4-45.4)
WBC count (x10^3/μL)	7.8 (3.0-14.6)
Splenomegaly	1 (5.6%)
Bone marrow reticulin fibrosis (grade 1+ or 2+)	6 (33%)
Bone marrow megakaryocyte hyperplasia	14 (78%)
Bone marrow cellularity Hypocellular Normocellular Hypercellular Missing	 2 (14%) 9 (64%) 3 (21%) 4
Prior hydroxyurea for ET	17 (94%)
Prior anagrelide for ET	13 (72%)
Prior interferon for ET	4 (22%)
Prior hydroxyurea and anagrelide for ET	12 (67%)
Prior hydroxyurea, anagrelide, and interferon for ET	3 (17%)
More than one prior therapy (anagrelide ± interferon)	13 (72%)
Resistant to at least one prior therapy	9 (50%)
Intolerant of at least one prior therapy	14 (78%)

*****One patient did not meet inclusion criteria for minimum baseline platelet count, but investigator felt cytoreductive therapy was clinically indicated.
Table adapted from Baerlocher *et al*., N Engl J Med 2015; 373:920-928, DOI: 10.1056/NEJMoa1503479

Table S2. Additional mutations at study entry.

Gene	Amino Acid Exchange	Position on Chromosome	Genomic Position (hg19)	Refe-rence	Variant	VAF* at study entry %	VAF* at best molecular response %	Patient UPN
ASXL1	p.Tyr591Ter	chr 20	31022287	-	A	30	4	17
ASXL1	p.Gly646Trpfs*12	chr 20	31022449	-	G	33	18	9
CBL	c.1432-1G>A	chr 11	119155678	G	A	5	<3	9
DNMT3A	p.Ala644Thr	chr 2	25466773	C	T	3	<3	14
DNMT3A	p.Arg688His	chr 2	25464450	C	T	4	6	14
DNMT3A	p.Gly722Asp	chr 2	25463517	C	T	16	35	18
DNMT3A	p.Tyr735Cys	chr 2	25463289	T	C	7	<3	4
DNMT3A	p.Met880Val	chr 2	25457249	T	C	45	6	10
DNMT3A	c.2597+1G>A	chr 2	25458575	C	T	8	7	14
EZH2	p.Asp293Ala	chr 7	148523575	T	G	10	<3	4
SF3B1	p.Lys666Arg	chr 2	198267360	T	C	9	8	4
SF3B1	p.Lys700Glu	chr 2	198266834	T	C	45	42	7
TET2	p.Arg1465Ter	chr 4	106193931	C	T	7	3	4
TET2	p.Tyr1608Leufs*6	chr 4	106196489	-	T	33	16	2
TET2	p.Met1772Cysfs*48	chr 4	106196981	A	-	42	4	10
TP53	p.His179Arg	chr 17	7578394	T	C	5	8	4
TP53	p.Arg249Lys	chr 17	7577535	C	T	3	<3	1
U2AF1	p.Gln157Pro	chr 21	44514777	T	G	28	4	9

VAF, variant allele frequency. *LOD at 2%