Array comparative genomic hybridization and sequencing of 23 genes in 80 patients with myelofibrosis at chronic or acute phase

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Supplemental Table 1. Clinical and biological characteristics of 104 MF samples studied by aCGH and DNA sequencing

A. Clinical and biological characteristics of 68 chronic MPNs

Samples	Sex/Age	Diagnosis	PB/ BM	IPSS	DIPSS	DIPSS plus	Time from diagnosis to sampling (m)	Karyotype	Leukocyte count (x 10^9/L)	Hemoglobin level (g/dL)	Hematocrit (%)	Platelet count (x 10^9/L)	RCT need	Ci blast cells	Symptoms	JAK2 V617F %	Previous Therapies	aCGH
HD-0577	F/57	post-PV MF	PB	NA	NA	NA	<1	46,XX[20]	27.5	14.7	47	255	no	no	satisfactory	80-90	HU	X
HD-0758	F/80	post-PV MF	PB	NA	NA	NA	<1	ND	10.4	9.7	29.9	132	no	yes	infectious health	50	ND	X
HD-1302	M/69	post-PV MF	PB	NA	NA	NA	<1	46,XY[20]	14.3	12.8	39.5	297	no	yes	no functional symptoms	80-90	HU	X
HD-1401_0842	M/67	post-PV MF	PB	NA	NA	NA	<1	46,XY,del(8)(q21q23)[7]/46,XY[19]	31.1	13.4	41.2	714	no	yes	weight loss	50-60	HU, antiplatelet therapy	X
HD-1426	M/82	post-PV MF	PB	NA	NA	NA	60	46,XY[20]	20.7	15	14.3	361	no	yes	asthenia and fatigue	80-90	X, PA, antiplatelet therapy	no
HD-1427_1656	F/71	post-PV MF	PB	NA	NA	NA	45	46,XX,t(7;14)(q11;q24)[2]/46,XX[18]	16.6	15.7	51.9	363	no	no	night sweats	80-90	M	X
HD-1480	F/64	post-PV MF	PB	NA	NA	NA	31	46,XX[20]	15.2	11.8	36.2	631	no	no	no functional symptoms	75	HU, X, antiplatelet therapy	X
HD-1537	F/70	post-PV MF	PB	NA	NA	NA	21	46,XX[20]	3.1	10.7	33.1	165	no	yes	no functioNAl symptoms	80	HU, EPO	X
HD-1559_1649	M/63	post-PV MF	BM	NA	NA	NA	<1	45,XY,-7[9]/47,XY,+9[3]/46,XY[8]	2	9.8	ND	39	no	no	asthenia and fatigue	30-50	antiplatelet therapy	X
HD-1570bis	F/55	post-PV MF	PB	NA	NA	NA	11	46,XX[20]	3.7	11	ND	85	no	no	satisfactory	80-90	none	X
HD-1602	F/72	post-PV MF	PB	NA	NA	NA	<1	46,XX,der(15)t(1;15)(p11;q12)[11]/46,XX[9]	24.6	12.4	ND	600	no	no	night sweats	80-90	HU, P, X	X
HD-1664	F/60	post-PV MF	PB	NA	NA	NA	14	46,XX,del(20)(q11q13)[3]/46,XX[17]	2.5	10.1	ND	81	no	yes	satisfactory	2	none	X
HD-1691_0789	M/69	post-PV MF	BM	NA	NA	NA	36	45,XY,-7,del(20)(q11q13)[20]/46,XY[2]	13.3	13.7	ND	60	no	no	night sweats	mutated	anti-JAK1/2	X
HD-1813_1836	M/77	post-PV MF	PB	NA	NA	NA	<1	45,XY,-7[6]/45,sl,del(12)(p12p13)[3]/ 45,sdl1,add(3)(q2?1)[13]	4.6	11.5	34.7	58	yes	yes	ND	2-5	none	X
HD-1824_0589	F/70	post-PV MF	PB	NA	NA	NA	117	46,XX,del(20)(q11q13)[8]/48,XX,+8,+9[3]/48,idem, der(9)t(1;9)(q21;q12q)[2]/46,XX[7]	2.5	8.8	26.9	71	no	yes	satisfactory	50-70	none	X
HD-0554	F/79	post-ET MF	PB	NA	NA	NA	<1	46,XX[20]	8.3	10.2	ND	540	no	no	asthenia and fatigue	30-50	P, HU, EPO, A	X
HD-0598	F/71	post-ET MF	PB	NA	NA	NA	<1	46,XX[20]	6.4	10.7	31.4	758	no	no	asthenia and fatigue	wt	HU, P, X	X
HD-0599	M/80	post-ET MF	PB	NA	NA	NA	3	ND	4.1	9.2	27.1	386	yes	no	no functional symptoms	30-40	HU, EPO	no
HD-0601	M/76	post-ET MF	PB	NA	NA	NA	<1	46,XY[20]	11	11	33.5	407	no	yes	asthenia and fatigue	60-70	HU, EPO, antiplatelet therapy, X	X
HD-0614_1112_1352	M/76	post-ET MF	PB	NA	NA	NA	139	46,XY[22]	4.5	11.2	35	269	no	yes	satisfactory	wt	EPO, antiplatelet	X

																	therapy	
HD-0725_1380	M/74	post-ET MF	PB	NA	NA	NA	11	46,XY[20]	35.4	12.6	37.6	315	no	yes	satisfactory	40	HU	X
HD-0983	M/32	post-ET MF	PB	NA	NA	NA	<1	ND	15.2	12.2	36.8	419	no	yes	satisfactory	wt	X	X
HD-1264	M/72	post-ET MF	BM	NA	NA	NA	<1	45,XY,der(7;18)(q10;q10)[20]	4	12	35	150	no	yes	satisfactory	wt	B, antiplatelet therapy	X
HD-1309_1741	F/36	post-ET MF	PB	NA	NA	NA	73	46,XX[20]	12.2	9.9	ND	750	no	yes	weight loss	wt	HU, A, P, T	X
HD-1360_1605	M/67	post-ET MF	PB	NA	NA	NA	12	ND	5.8	11	32.8	392	no	yes	no functional symptoms	30-50	HU, X, antiplatelet therapy	X
HD-1376	M/76	post-ET MF	PB	NA	NA	NA	9	46,XY[20]	27.2	7.3	21.9	49	no	yes	satisfactory	50-70	HU, antiplatelet therapy	X
HD-1462	F/63	post-ET MF	PB	NA	NA	NA	<1	46,XX[20]	13.2	11.5	ND	351	no	no	none	75	antiplatelet therapy	no
HD-1569	F/54	post-ET MF	PB	NA	NA	NA	47	46,XX[20]	31.8	12.5	37.7	317	no	yes	asthenia and fatigue	50-60	C, PA	no
HD-1616_0551	M/71	post-ET MF	PB	NA	NA	NA	<1	47,XY,+9[5]/46,XY[15]	11.4	11.4	34.2	376	no	no	ND	50-70	C, EPO	X
HD-0497_1307	F/82	PMF	PB	inter-2	inter-2	inter-2	160	46,XX,del(20)(q11q13)[6]/46,XX[3]	2.4	7.6	21.2	205	yes	no	satisfactory	20-30	T	X
HD-0586	M/62	PMF	PB	inter-2	inter-2	high	31	45,XY,-13[18]/46,XY[2]	3.2	7.8	23.8	83	yes	yes	satisfactory	wt	HU, T	X
HD-0607_1130	M/51	PMF	PB	low	inter-1	inter-1	70	46,XY[20]	7.6	13.3	39	471	no	yes	satisfactory	wt	HU, antiplatelet therapy	X
HD-0616	M/54	PMF	PB	low	low	low	11	46,XY[4]	14.3	17.8	52	257	no	no	satisfactory	35-40	none	X
HD-0648	F/60	PMF	PB	ND	inter-1	inter-1	129	46,XX[20]	20.3	11.5	35	112	no	yes	satisfactory	80-90	HU, EPO	X
HD-0679_1291	M/31	PMF	PB	low	low	low	<1	46,XY[20]	5.3	15.3	44.9	208	no	no	satisfactory	5-10	none	X
HD-0683	M/62	PMF	PB	inter-2	inter-2	inter-2	<1	ND	1.9	8.5	24.8	101	yes	yes	ND	10	none	X
HD-0689	F/74	PMF	PB	ND	inter-2	inter-2	298	ND	6	7.6	22.2	167	yes	yes	asthenia aND fatigue	80-90	HU, T	X
HD-0717_1244	M/65	PMF	PB	inter-2	inter-1	inter-1	70	46,XY[20]	9.3	14.5	44.3	191	no	yes	satisfactory	wt	none	X
HD-0719	M/85	PMF	PB	inter-1	inter-1	inter-1	<1	46,XY[20]	120	8.6	26	479	no	yes	fever	wt	none	X
HD-0728	F/54	PMF	PB	inter-2	low	low	<1	46,XX,t(6;17)(p21-22;q11)[10]/46,XX[13]	35.1	13.2	ND	691	no	no	satisfactory	70-80	none	X
HD-0759	F/63	PMF	PB	inter-2	inter-2	high	3	46,XX,del(5)(q13q33),del(20)(q11q13)[19]/46,XX[1]	4.1	8	23	77	yes	yes	asthenia and fatigue	50-75	EPO	X
HD-0777	M/86	PMF	PB	high	inter-2	inter-2	<1	ND	54.2	7.7	25.2	761	yes	no	asthenia and fatigue	wt	A, antiplatelet therapy	X
HD-0862	M/83	PMF	PB	high	high	high	4	46,XY[20]	4.6	9.7	29.7	302	yes	yes	weight loss	12-30	EPO	X
HD-1047	M/65	PMF	PB	inter-2	inter-2	inter-2	<1	46,XY,del(20)(q11q13)[3]/46,idem,add(9)(p2?3)[17]	26.8	13.6	40.8	467	no	yes	satisfactory	50-75	none	X
HD-1095	M/64	PMF	PB	ND	ND	ND	<1	ND	ND	ND	ND	ND	ND	ND	ND	50-70	ND	X
HD-1098	M/75	PMF	PB	ND	ND	ND	<1	ND	9.3	13.4	40.4	143	ND	ND	ND	wt	ND	X
HD-1100	M/73	PMF	PB	high	high	high	80	46,XY,del(13)(q13q22-31)[6]/46,sl,?del(17)(p12-13)[5]/44,sl,der(1)inv(1)(p32~34q23)?dup(1)(q31q32),-7,-16,?der(17)t(16;17)(p11-12;p12-13)[6]/44,sdl1,?del(17)(p12-13)[5] +der(17)t(16;17)(p11-12;p12-13)[6]/44,sdl1,?del(17)(p12-13)[5]	11.7	10.6	32.2	645	yes	yes	no functioNAl symptoms	31-50	HU, antiplatelet therapy	X
HD-1111	F/84	PMF	PB	inter-2	inter-1	inter-1	101	46,XX[20]	20.7	15	14.3	361	no	yes	asthenia and fatigue	80-90	HU, P, T, C	no
HD-1123	F/80	PMF	PB	inter-2	inter-2	inter-2	<1	46,XX[20]	27.3	10.2	32	1188	no	no	asthenia and fatigue	50-70	none	X
HD-1138_0540_1398	M/61	PMF	PB	inter-2	inter-1	inter-1	82	ND	27	12.0	36.2	378	no	yes	satisfactory	wt	none	X

HD-1207	M/64	PMF	PB	inter-1	inter-2	high	28	46,XY,ider(20)(q10)del(20)(q11q13)[20]	2.6	7.2	20.2	5	yes	yes	asthenia and fatigue	wt	T	X
HD-1208	M/74	PMF	PB	inter-2	high	high	3	48,XY,+8,+19[19]/46,XY[1]	8.6	8.7	25.6	101	yes	yes	fever	2	C	X
HD-1227	F/70	PMF	PB	low	inter-1	inter-2	82	48, XX +8,+9[7]/46,XX[13]	5.6	14.2	43.7	60	no	yes	asthenia and fatigue	2-5	EPO	X
HD-1265_0927_1461_1853	M/72	PMF	PB	high	inter-1	inter-1	13	ND	7.1	11.6	34.8	147	no	no	satisfactory	30-50	HU	X
HD-1283	M/75	PMF	PB	inter-2	inter-2	inter-2	84	ND	5	9.7	25	890	yes	no	satisfactory	wt	C, EPO, T, HU	X
HD-1289	M/75	PMF	PB	inter-1	inter-2	high	79	ND	11.4	12.5	37.1	88	no	yes	night sweats	mutated	HU, antiplatelet therapy	X
HD-1300_0528_1611	M/59	PMF	PB	inter-2	inter-2	inter-2	47	46, XY, del 20(q11q13)[4]/46,XY[16]	41.1	8.8	27.3	98	no	ND	satisfactory	80-90	graft therapy	X
HD-1362	F/67	PMF	PB	ND	inter-2	inter-2	195	46,XX,dup(1)(q44q12),del(13)(q13q14)[20]	1.3	9.8	29.7	123	yes	yes	satisfactory	80-90	E	X
HD-1412_1495	M/52	PMF	PB	ND	inter-1	inter-1	33	46,XY[2]	12	13.2	41.7	77	no	yes	satisfactory	wt	EPO, HU	X
HD-1422	F/58	PMF	PB	low	inter-1	inter-1	79	ND	12.8	12.2	ND	351	no	yes	satisfactory	wt	EPO	X
HD-1439	M/71	PMF	PB	low	inter-2	inter-2	89	46,XY,del(7)(q22q36)[2]/46,XY[21]	48.8	14.2	47	271	no	yes	satisfactory	50-75	HU	X
HD-1524bis	F/70	PMF	PB	inter-2	high	high	18	ND	76	5.8	18	26	yes	yes	weight loss	wt	HU, C	X
HD-1538	F/77	PMF	PB	high	inter-2	inter-2	<1	ND	3.4	9.7	28.8	366	no	yes	satisfactory	30-40	none	X
HD-1572bis	F/66	PMF	PB	inter-2	inter-1	inter-1	6	46,XX,t(12;17)(q12;q21)[15]/46,XX[5]	7.4	11.6	ND	228	no	yes	satisfactory	wt	HU	X
HD-1587	F/60	PMF	PB	inter-2	inter-2	inter-2	87	ND	3.5	9.2	ND	266	yes	yes	asthenia and fatigue	wt	C, EPO	X
HD-1606	F/40	PMF	BM	low	low	low	108	46,XX[20]	6.5	13.6	41.1	274	no	no	satisfactory	12-30	HU, antiplatelet therapy	X
HD-1617	F/82	PMF	PB	inter-1	inter-1	inter-2	18	46,XX[20]	6.5	12	35.8	36	yes	yes	satisfactory	wt	C	X
HD-1654	M/65	PMF	PB	low	inter-2	high	54	46,XY,inv(11)(q12q22)[c20]	4.2	7.6	22.2	38	yes	yes	asthenia and fatigue	wt	T	X

B. Clinical and biological characteristics of 19 matched pairs of MF

Samples	**Sex/ Age**	**Diagnosis**	**PB/ BM**	**IPSS**	**DIPSS**	**DIPSSplus**	**Time from diagnosis to sampling (m)**	**Karyotype**	**Leukocyte count (x 10^9/L)**	**Hemoglobin level (g/dL)**	**Hematocrit (%)**	**Platelet count (x 10^9/L)**	**RCT need**	**Ci blast cells**	**Symptoms**	**JAK2 V617F %**	**Previous Therapies**	**aCGH**
HD-0842_1401	M/64	PV	PB	NA	NA	NA	NA	ND	6.3	21.3	66.4	269	no	no	satisfactory	50-60	HU, antiplatelet therapy	X
HD-1401_0842	M/67	post-PV MF	PB	NA	NA	NA	<1	46,XY,del(8)(q21q23)[7]/46,XY[19]	31.1	13.4	41.2	714	no	yes	weight loss	50-60	HU, antiplatelet therapy	X

HD-0551_1616	M/66	ET	PB	NA	NA	NA	NA	ND	8.3	13.6	39.9	399	no	no	satisfactory	15-30	HU, antiplatelet therapy	X
HD-1616_0551	M/71	post-ET MF	PB	NA	NA	NA	<1	47,XY,+9[5]/46,XY[15]	11.4	11.4	34.2	373	no	no	ND	50-70	C, EPO	X
HD-1412_1495	M/52	PMF	PB	high	inter-1	inter-1	34	46,XY[2]	12	13.2	41.7	77	no	yes	satisfactory	wt	EPO, HU	X
HD-1495_1412	M/59	PMF	PB	inter-2	inter-2	NA	36	46,XY[2]	2.7	9.3	28.4	49	ND	yes	infectious disease	wt	EPO	X
HD-0725_1380	M/74	post-ET MF	PB	NA	NA	NA	11	46,XY[20]	19.2	12.6	37.6	321	no	yes	satisfactory	40	HU	X
HD-1380_0725	M/76	post-ET MF	PB	NA	NA	NA	40	46,XY[20]	35.4	12.6	37.6	315	no	yes	satisfactory	50-70	antiplatelet therapy	X
HD-0497_1307	F/82	PMF	PB	inter-2	inter-2	inter-2	160	46,XX,del(20)(q11q13)[6]/46,XX[3]	2.4	7.6	21.2	205	yes	no	satisfactory	20-30	T	X
HD-1307_0497	M/86	PMF	PB	NA	NA	NA	204	ND	3.2	11.8	34.7	64	yes	ND	asthenia and fatigue	ND	none	X
HD-0607_1130	M/51	PMF	PB	low	inter-1	inter-1	70	46,XY[20]	7.6	13.3	39	471	no	yes	satisfactory	wt	HU, antiplatelet therapy	X
HD-1130_0607	M/53	PMF	PB	low	inter-1	inter-1	102	46,XY[20]	9.5	14.5	42.7	474	no	yes	satisfactory	wt	HU, antiplatelet therapy	X
HD-0679_1291	M/31	PMF	PB	low	low	low	<1	46,XY[20]	5.3	15.3	44.9	208	no	no	satisfactory	5-10	none	X
HD-1291_0679	M/33	PMF	PB	low	low	low	26	ND	5	15.8	46.2	171	no	no	satisfactory	5-10	antiplatelet therapy	X
HD-0717_1244	M/65	PMF	PB	inter-2	inter-1	inter-1	71	46,XY[20]	9.3	14.5	44.3	191	no	yes	satisfactory	wt	none	X
HD-1244_0717	M/67	PMF	PB	inter-2	inter-1	inter-1	94	ND	6.5	13.3	40.9	115	no	yes	satisfactory	wt	none	X
HD-0589_1824	F/65	post-PV MF	PB	NA	NA	NA	55	48,XX,+8,+9[3]/46,XX,del(20)(q11q13)[2]/46,XX[15]	5.8	13.4	41.4	188	no	no	no functional symptoms	50-70	none	X
HD-1824_0589	F/70	post-PV MF	PB	NA	NA	NA	117	46,XX,del(20)(q11q13)[8]/48,XX,+8,+9[3]/48,idem, der(9)t(1;9)(q21;q12q)[2]/46,XX[7]	2.5	8.8	26.9	71	no	yes	satisfactory	50-70	none	X
HD-0789_1691	M/66	post-PV MF	PB	NA	NA	NA	<1	46,XY,del(20)(q11q13)[4]/46,XY[16]	20.3	13.7	45.7	360	ND	ND	ND	80-90	HU	X
HD-1691_0789	M/69	post-PV MF	BM	NA	NA	NA	36	45,XY,-7,del(20)(q11q13)[20]/46,XY[2]	13.3	13.7	ND	60	ND	ND	night sweats	ND	anti-JAK1/2	X
HD-1427_1656	F/71	PMF	PB	inter-2	inter-1	inter-1	45	46,XX,t(7;14)(q11;q24)[2]/46,XX[18]	16.6	15.7	51.9	363	no	no	weight loss	80-90	M	X
HD-1656_1427	F/72	PMF	PB	high	inter-2	inter-2	54	46,XX[21]	8.2	14	41.7	310	no	yes	night sweats	ND	antiplatelet therapy	X
HD-1360_1605	M/44	post-ET MF	PB	NA	NA	NA	12	ND	5.8	11	32.8	392	no	yes	no functional symptoms	30-50	X, antiplatelet therapy	X
HD-1605_1360	M/45	post-ET MF	BM	NA	NA	NA	20	ND	7.5	12	ND	547	no	ND	no functional symptoms	30-50	PA, antiplatelet therapy	X
HD-0614_1112_1352	M/76	post-ET MF	PB	NA	NA	NA	140	46,XY[22]	4.5	11.2	35	269	no	yes	satisfactory	wt	EPO, antiplatelet therapy	X
HD-1112_0614_1352	M/77	post-ET MF	PB	NA	NA	NA	170	ND	4.3	7.8	22.8	78	yes	yes	asthenia and fatigue	wt	EPO, antiplatelet	X

																	therapy	
HD-1352_0614_1112	M/78	post-ET MF	PB	NA	NA	NA	178	46,XY,del(7)(q22q36)[2]/46,XY,add(4)(q21-22),-12,-13,+der(?)t(?;4)(?;q21-22),+mar[3]/44,XY,add(4),der(6)t(4;6)(q21-22;p2?3),-7,-12,-15,-16,+2mar[3]	3.5	ND	23.4	159	yes	yes	asthenia aND fatigue	wt	EPO, antiplatelet therapy	X
HD-0540_1138_1398	M/58	PMF	PB	inter-1	ND	ND	46	46,XY[20]	11.5	12.3	ND	334	no	ND	satisfactory	wt	none	X
HD-1138_0540_1398	M/61	PMF	PB	inter-2	inter-1	inter-1	82	ND	27	12.0	36.2	378	no	yes	satisfactory	wt	none	X
HD-1398_0540_1138	M/62	PMF	PB	inter-2	inter-1	inter-1	91	ND	27.3	12.6	38	315	no	yes	satisfactory	wt	none	X
HD-0927_1265_1461_1853	M/72	PMF	PB	high	high	inter-2	<1	ND	42	11.7	36	658	no	yes	weight loss	40-50	none	X
HD-1265_0927_1461_1853	M/72	PMF	PB	high	inter-1	inter-1	13	ND	7.1	11.6	34.8	147	no	no	satisfactory	30-50	HU	X
HD-1461_0927_1265_1853	M/73	PMF	PB	high	inter-1	inter-1	20	ND	9.7	10.8	ND	328	no	no	satisfactory	30-50	HU	X
HD-1853_0927_1265_1461	M/75	BP-PMF	PB	NA	NA	NA	1	46,XY, +8[16]/46,XY[6]	5.1	10.2	32.6	168	yes	yes	asthenia and fatigue	ND	HU	X
HD-1559_1649	M/63	post-PV MF	BM	NA	NA	NA	<1	45,XY,-7[9]/47,XY,+9[3]/46,XY[8]	2.2	8.2	ND	25	ND	no	satisfactory	30-50	antiplatelet therapy	X
HD-1649_1559	M/63	BP post-PV MF	PB	NA	NA	NA	4	45,XY,-7[20]	73	10	ND	27	yes	yes	asthenia and fatigue	ND	C, T	X
HD-1813_1836	M/77	post-PV MF	PB	NA	NA	NA	<1	45,XY,-7[6]/45,sl,del(12)(p12p13)[3]/45,sdl1,add(3)(q2?1)[13]	4.6	11.5	34.7	58	yes	yes	ND	2-5	none	X
HD-1836_1813	M/77	BP post-PV MF	PB	NA	NA	NA	1	45,XY,-7[6]/45,sl,del(12)(p12p13)[3]/45,sdl1,add(3)(q2?1)[13]	1.5	8.4	24.2	13	yes	yes	ND	2-5	none	X
HD-0528_1300_1611	M/56	PMF	PB	inter-2	inter-1	inter-1	<1	46,XY[20]	29.4	13.2	38.6	295	no	yes	satisfactory	75	none	X
HD-1300_0528_1611	M/59	PMF	PB	inter-2	inter-2	inter-2	47	46, XY, del 20(q11q13)[4]/46,XY[16]	41.1	8.8	27.3	98	no	ND	satisfactory	80-90	graft therapy	X
HD-1611_0528_1300	M/60	BP-PMF	PB	NA	NA	NA	59	46,XY,del(20)(q11q13)[12]/46,XY,inv(3)(q21q26)[4]/46,idem,del(6)(q2?1q2?6)[3]/46,XY[1]	6.5	12	35.8	36	no	yes	asthenia aND fatigue	ND	M	X
HD-1309_1741	F/36	post-ET MF	PB	NA	NA	NA	73	46,XX[20]	16.2	9.9	ND	750	ND	yes	asthenia and fatigue	wt	none	X
HD-1741_1309	F/37	BP post-ET MF	PB	NA	NA	NA	87	46,XX,-3,der(5)t(3;5)(q13;q21),del(7)(q21q36),t(9;17)(q11;q11),+21[20]	77.3	9.6	ND	469	no	yes	asthenia and fatigue	wt	HU	X

C. Clinical and biological characteristics of 17 blast phase MF

Samples	Sex/ Age	Diagnosis	PB / BM	Time from diagnosis to sampling (m)	Karyotype	Leukocyte count (x 10^9/L)	Hemoglobin level (g/dL)	Hematocrit (%)	Platelet count (x 10^9/L)	JAK2 V617F %	Previous Therapies	aCGH
HD-0492	M/67	BP post-PV MF	PB	20	47,XY,+8[17]/48,idem,+9[3]	17.0	10.0	36.0	44	80-100	HU	X
HD-1517	M/89	BP post-PV MF	BM	ND	46,XY[20]	15.1	10.4	30.5	94	wt	ND	X
HD-1649_1559	M/63	BP post-PV MF	PB	4	45,XY,-7[20]	73	10	ND	27	ND	C, T	X
HD-1836_1813	M/77	BP post-PV MF	PB	<1	ND	1.5	8.4	24.2	13	2-5	none	X
HD-0535	M/68	BP post-ET MF	PB	153	46,XY,t(3;12)(q26-27;q13-14)[20]	10.3	11.5	ND	661	wt	HU	X
HD-0536	M/53	BP post-ET MF	PB	<1	46,XY,add(15)(q25-26)[5]/46XY[19]	5.0	13.1	39.2	535	wt	none	X
HD-0646	F/61	BP post-ET MF	PB	15	46,XX,inv(3)(q21q26)[3]/46,idem,der(16)t(1;16)(q11;q11)[17]	5.2	1.7	33.0	315	5-10	HU	X
HD-0856	F/57	BP post-ET MF	PB	15	46,XX,del(7)(q22q36),del(8)(q22q24)[18]/47,idem,+8[2]	7.7	9.2	29.0	1052	wt	HU	X
HD-1167	F/68	BP post-ET MF	PB	12	49,XX,add(3)(p14),del(5)(q31q35),+8,+8,+9,?del(11)(p11p13),-15,add(16)(p12),+mar[20]	15	8	23.7	117	wt	HU, A	X
HD-1387	M/60	BP post-ET MF	PB	15	ND	ND	ND	ND	ND	ND	ND	X
HD-1741_1309	F/37	BP post-ET MF	PB	87	46,XX,-3,der(5)t(3;5)(q13;q21),del(7)(q21q36),t(9;17)(q11;q11),+21[20]	77.3	9.6	ND	469	wt	HU	X
HD-0655	M/76	BP-PMF	PB	26	46,XY,inv(16)(p13q22)[20]	6.6	8.1	24.0	25	30-50	none	X
HD-1611_0528_1300	M/60	BP-PMF	PB	59	46,XY,del(20)(q11q13)[12]/46,XY,inv(3)(q21q26)[4]/46,idem,del(6)(q2?1q2?6)[3]/46,XY[1]	6.5	12	35.8	36	ND	M	X
HD-0635	M/76	BP-PMF	PB	26	46,XY,inv(16)(p13q22)[20]	6.6	8.1	24.0	25	30-50	none	X
HD-1847	M/79	BP-PMF	PB	<1	ND	24.4	11.6	36.2	164	mutated	HU	X
HD-1853_0927_1265_1461	M/75	BP-PMF	PB	<1	46,XY, +8[16]/46,XY[6]	5.1	10.2	32.6	168	ND	HU	X
HD-1137	M/82	BP post-MPN MF	PB	3	44,XY,add(1)(q4?2),add(3)(p14),del(5)(q23q34),der(9)t(9;10)(q23;q22),-10,?dic(15;20)(q12;p12),add(17)(p11)[19]/46,XY,del(12)(p11p13)[1]	106	9.4	30	46	wt	EPO, C	X

MF samples are classified according to the WHO criteria.

aCGH, array comparative genomic hybridization; A, anagrelid; B, busulfan; BM, bone-marrow; C, corticoid; Ci blasts, circulating blasts; DIPSS, dynamic international prognostic scoring system; DIPSSplus, dynamic international prognostic scoring system plus; E, eposin; F, female; HU, hydroxyurea; IPSS, international prognostic scoring system; m, month; M, male; ME, melphalan; MF, myelofibrosis; NA, not applicable; ND, not determined; P, pipobroman; PA, peginterferon alfa-2a; PB, peripheral blood; PMF, primary myelofibrosis; post-ET MF, post-essential thrombocythemia MF; post-PV MF, post-polycythemia vera MF; RCT need, red cell transfusion need; T, thalidomide; wt, wild-type; y, years. For PMF, IPSS, DIPSS and DIPPSplus scores depend on age (>65 years), on hemoglobin (< 10 g/dL), on leukocyte count (> 25.10^9/L), on circulating blasts (> or = 1%), on platelet count (< 100.10^9/L), on presence of constitutional symproms (weight loss, night sweats, fever). For DIPPSplus score, karyotype and transfusion status must be added to DIPSS. IPSS score was calculated at MF diagnosis whereas DIPPS and DIPPSplus score were calculated at sampling.

Supplemental Table 2. Sequencing primers

A List of primers used for PCR and sequencing

Genes	Forward primers	Sequence 5' to 3'	Reverse primers	Sequence 5' to 3'	annealing (°C)
ASXL1	ASXL1_EX12_F1	AGGTCAGATCACCCAGTCAGTT	ASXL1_EX12_R1	TAGCCCATCTGTGAGTCCAACTGT	55
	ASXL1_EX12_F2	AGAGGACCTGCCTTCTCTGAGAAA	ASXL1_EX12_R2	TTCGATGGGATGGGTATCCAATGC	55
	ASXL1_EX12_F3	ACTTGAAAACCAAGGCTCTCGT	ASXL1_EX12_R3	GCAACCATCCCATCTGTCCTTGTA	55
	ASXL1_EX12_F4	GGTGGACAAGGATGAGAAACCCAA	ASXL1_EX12_R4	TGTCCTGTGACATAGCACGGACTT	55
	ASXL1_EX12_F5	TGGATTCCAAAGAGCAGTTCTCTTC	ASXL1_EX12_R5	CATGACAAAGGGCATCCCTTCCAA	55
	ASXL1_EX12_F6	ACAGGAAAGCTACTGGGCATAGTC	ASXL1_EX12_R1	CAAGAGTGCTCCTGCCTAAAGAGT	55
BMI1	BMI1-F2-3	TGATTACTAGATGATCTCCATTCTTG	BMI1-R2-3	AAATAAAGAGGGTTGCCTTCAG	55
	BMI1_F4-5-6-7	TTGAAAGGCACACTTCTTTTG	BMI1_R4-5-6-7	CTAAAGGGCAACACAATCCC	61
	BMI1_F8-9	GGTCACCTCCAATTTTGTTTG	BMI1_R8-9	TGTATTTCAATGGAAGTGGACC	55
	BMI1_F10	GAGAAGGGTAAGTAGCATATCTGTTG	BMI1_R10	AACAAACTATGGCCCAATGC	55
CBL	CBL_F8	ACCCAGACTAGATGCTTTCTG	CBL_R8	AGGCCACCCCTTGTATCAGT	58
	CBL_F9	TTCAGATGCATCTGTTACTATCT	CBL_R9	AGTGTTTTACGGCTTTAGAAGACA	58
DNMT3A	DNMT3A_F15	TGTAAAACGACGGCCAGTTTTCCATTCCAGGTAGCACACC	DNMT3A_R15	CAGGAAACAGCTATGACCAGGCTCCTAGACCCACACACC	60
	DNMT3A_F16	TGTAAAACGACGGCCAGTAGGGTGTGTGGGTCTAGGAGC	DNMT3A_R16	CAGGAAACAGCTATGACCGCTGTGAAGCTAACCATCATTTCG	60
	DNMT3A_F17	TGTAAAACGACGGCCAGTGACTTGGGCCTACAGCTGACC	DNMT3A_R17	CAGGAAACAGCTATGACCAAATGAAAGGAGGCAAGGGC	60
	DNMT3A_F18	TGTAAAACGACGGCCAGTATAGGACAGTGGTGTGGCTCG	DNMT3A_R18	CAGGAAACAGCTATGACCTTCTTCCTGTCTGCCTCTGTCC	60
	DNMT3A_F19	TGTAAAACGACGGCCAGTGACAGCTATTCCCGATGACCC	DNMT3A_R19	CAGGAAACAGCTATGACCTGCAGATGAGACAGGATGAAGC	60
	DNMT3A_F20	TGTAAAACGACGGCCAGTGCCGGCGCTGTTTCATGC	DNMT3A_R20	CAGGAAACAGCTATGACCCCACTATGGGTCATCCCACCTGC	60
	DNMT3A_F21	TGTAAAACGACGGCCAGTCCTTCCCGCTGTTATCCAGG	DNMT3A_R21	CAGGAAACAGCTATGACCCATCCTGCCCTTCCTTCTCC	60
	DNMT3A_F22	TGTAAAACGACGGCCAGTTGGCATATTTGGTAGACGCATGAC	DNMT3A_R22	CAGGAAACAGCTATGACCTGGGAAATGCTTGATAAAACCCAC	60
	DNMT3A_F23	TGTAAAACGACGGCCAGTGTGTGGTTAGACGGCTTCCG	DNMT3A_R23	CAGGAAACAGCTATGACCCCCATGTCCCTTACACACACG	60
EZH2	EZH2_F2	GGTGATCATATTCAGGCTGG	EZH2_F2	AAACTTATTGAACTTAGGAGGGG	60
	EZH2_F3	GACACCCTGAGGTCAATGAT	EZH2_F3	ATCATTGACCTCAGGGTGTC	60
	EZH2_R4	GGCTACAGCTTAAGGTTGTCCT	EZH2_R4	CTGTCTTGATTCACCTTGACAAT	60
	EZH2_R5	AAATCTGGAGAACTGGGTAAAGAC	EZH2_R5	TCATGCCCTATATGCTTCATAAAC	60
	EZH2_R6	AGGCTATGCCTGTTTTGTCC	EZH2_R6	AAAAGAGAAAGAAGAAACTAAGCCC	60
	EZH2_R7	CTGACTGGCATTCCACAGAC	EZH2_R7	AAGTGTAGTGGCTCATCCGC	60
	EZH2_R8	CATCAAAAGTAACACATGGAAACC	EZH2_R8	TTGTAATAAATGATAGCACTCTCCAAG	60
	EZH2_R9	TCCATTAATTGACTTTTCCAGTG	EZH2_R9	ACCTCCACCAAAGTGCAAAG	60
	EZH2_R10	TTCTCTTCCATCAAAATGAGTTTTAG	EZH2_R10	TCCTCACAACACGAACTTTCAC	60
	EZH2_R11	GAGTTGTCCTCATCTTTTCGC	EZH2_R11	CCAAGAATTTTCTTTGTTTGGAC	60
	EZH2_F12	AAGAATGGTTTGCCTAAATAAGAC	EZH2_F12	CTTGCCTGCAGTGTCTATC	60

	EZH2_F13	TCTTGGCTTTAACGCATTCC	EZH2_F13	CAAATTGGTTTAACATACAGAAGGC	60
	EZH2_F14	TGATCGTTTCCATCTCCCTG	EZH2_F14	AGGGAGTGCTCCCATGTTC	60
	EZH2_F15	GAGAGTCAGTGAGATGCCCAG	EZH2_F15	TTTGCCCCAGCTAAATCATC	60
	EZH2_F16	TTTTTGATGATGTGATTGTGTTTT	EZH2_F16	TGGCAATTCATTTCCAATCA	60
	EZH2_F17	TTCTGTCAGGCTTGATCACC	EZH2_F17	CTCGTTTCTGAACACTCGGC	60
	EZH2_F18-19	AGGCAAACCCTGAAGAACTG	EZH2_F18-19	TTCCAATTCTCACGTCAAAGGTA	60
	EZH2_F20	CCGTCTTCATGCTCACTGAC	EZH2_F20	AAAAACCCTCCTTTGTCCAGA	60
IDH1	IDH_F4	GGATGCTGCAGAAGCTATAA	IDH_R4	CATGCAAAATCACATTATTGCC	53.6
IDH2	IDH_F4	CTGTGTTGTTGCTTGGGGTT	IDH_R4	GGGGTGAAGACCATTTTGAA	56.9
JAK2	JAK2_F12	CTCCTCTTTGGAGCAATTCA	JAK2_R12	GAGAACTTGGGAGTTGCATA	55
KRAS	KRAS_F2	GCCATTTGTCCGTCATCTTTGGAG	KRAS_R2	TGCATGGCATTAGCAAAGACTCA	55
	KRAS_F3	AGGAAGGAAAATTTGGTGTAGTGGA	KRAS_R3	AGAAGCAATGCCCTCTCAAGAGAC	54
	KRAS_F4a	CTTGCACATGGCTTTCCCAGTA	KRAS_R4a	AGTGGTTGCCACCTTGTTACCT	54
	KRAS_F4b	CCTGTACACATGAAGCCATCGT	KRAS_R4b	CTAACAGTCTGCATGGAGCAGGAA	54
LNK	LNK_F2a	CACCCACGTGTCTTTCAGC	LNK_R2a	TCCAGGGCAGGAACTTCT	62
	LNK_F2b	CCCTGCTCCTTCCAGCAC	LNK_R2b	CTGGAAAGCCATCACACCTC	63
	LNK_F3-4	GGGACTCCTGGGGAGACTAT	LNK_R3-4	TGCATCTCTGCTTCTGTGCT	62
	LNK_F5-6	ACCACCTTTGCTGCTACCAC	LNK_R5-6	TGTCCCTCAGGACCCTGAA	62
	LNK_F7	AACTTTCAGGGGATAGCCAAG	LNK_R7	GATACCTGTACCCCTGGGTCT	62
	LNK_F8	TCTGTGTCCTGTCAGCACTTG	LNK_R8	TCTGGAAGGAAGGAAAGATCA	61
MPL	MPL_F10	CCGAAGTCTGACCCTTTTTG	MPL_R10	ACAGAGCGAACCAAGAATGC	60
NF1	NF1_F1	TAGTGGGGAGAGCGACCA	NF1_R1	TCCCCTCACCTACTCTGTCC	60
	NF1_F2	ATCATATATTGGTGTTTGAGATGC	NF1_R2	ACAGTTACTTGTGCTCACTGAATC	60
	NF1_F3	TGTGTGTTGATTGGTAGCAGAA	NF1_R3	GGACTGTCCTCTTGGTCCAC	60
	NF1_F4	TTTTGTTCTGTGTGTGTGTTTGA	NF1_R4	AAAACCTCATTTCCATTAACTTTTT	60
	NF1_F5	GAGATCCTCCTCCCTTAGCC	NF1_R5	GCCACCCTTGAGAGATCAAA	60
	NF1_F6-7	TTAGGTGTCTTTACCTTTCATTGC	NF1_R6-7	CAGTATTTCCTATTTGACACCAGTTG	55
	NF1_F8	CAGAATGCATTTGTGTAGTTGC	NF1_R8	AAGTCCATCAAACAAAGAAACC	60
	NF1_F9	AAATTATGAAATTGAAAACCACAAA	NF1_R9	TCAGTCCATTTAGGCTGATGAA	55
	NF1_F9_seq	CAAATATAAATTATGCATT			
	NF1_F10-11	GTGTGGGTAATGTGTTGATGTT	NF1_R10-11	ACGCAAAGAAAAGAAAGAAAAA	55
	NF1_F10-11_seq	TGTGTTGATGTTATTACATG			
	NF1_F12	GAAATCATGGTGTGTGTTTGC	NF1_R12	TGAAGGACCCATTCAATTCTC	60
	NF1_F13	TGGATAGCTATTATCCTGAGTCTTATG	NF1_R13	GCTAAACCCAATTAACTTAGTGTGA	60
	NF1_F13_seq	CTGAGTCTTATGTCTGATACC			
	NF1_F14	TCTTCCTCCTTCTAATCTCTCTCG	NF1_R14	GTTACAATCTTTCCTTCAAGAACAT	55
	NF1_F15	TGTTTGAGTGAGTCTTCTCTTTGTC	NF1_R15	CATAAAACCTTTGGAAGTGTAAGTTTT	60
	NF1_F16	TGCATTAGGTTATTGATGATGC	NF1_R16	TTGGAATGGTAATGGTGAGAGA	60
	NF1_F17	CCTCTTGGTTGTCAGTGCTTC	NF1_R17	TCATTCAGAAAACAAACAGAGCA	60
	NF1_F18	TTTATACATAAAATTACCCAAGTTGC	NF1_R18	TCAGTCTGCCTCAAAGCACA	60
	NF1_F19-20	TTTGGGTGGAGCTTATCAGG	NF1_R19-20	TGTTTACTTTACTGAGCGACTCTTG	60

	NF1_F21	TTGGATAAAGCATAATTTGTCAAGTC	NF1_R21	TTAAATAATCTGAAAGAAAAAGGCTTA	55
	NF1_F22-23	GGTCTGTCTTCTGGGCATTG	NF1_R22-23	TCCTTTCTACCAATAACCGCATA	60
	NF1_F24	AAGGTGTGTGTGTGGCTTCA	NF1_R24	GTTATAACTCTCACAGTAAAACCCACT	60
	NF1_F25	GAGGGGAAGTGAAAGAACTTGA	NF1_R25	GGGGGCTTTATTTGCTTTTT	60
	NF1_F26	CCATTCACACCATGCACATA	NF1_R26	TGTGGACAAACAGATGCAAA	60
	NF1_F27	TGGCATGTAAGAGAAGCAAAAA	NF1_R27	TGCCAACCACTTCCCTACAG	60
	NF1_F28-29	AACTTTGGGTTTACATTTTTGCTACT	NF1_R28-29	GCCTTGCTTCATGCAGTGT	60
	NF1_F30	GTCTACACGTTGCACTTGGC	NF1_R30	AGACAATTCCAACAATTCTCAATG	60
	NF1_F31	TCCATTTGTGTTACATTTTATGGTG	NF1_R31	CACAAATTGCCTTTGGGATAA	60
	NF1_F32-33	GCAAAGTTTGACCTTTGAACTCT	NF1_R32-33	TGGATTTATGTGAAACCGAAAA	60
	NF1_F34	TCTGGGTGTATCTGGTGTTGA	NF1_R34	TCTAAATTTAAACGGAGAGTGTTCA	60
	NF1_F35	CCTGAGGTCTTTTTGGTGCT	NF1_R35	ACATCTTTCCTCATAGTCTTCTATCAT	60
	NF1_F36	CTCAGTAGACAACATAAAGCCTCA	NF1_R36	CCCTGCTATATGCTCCCAAG	60
	NF1_F37	TGAATCCAGACTTTGAAGAATTG	NF1_R37	ACCTTGACATCCCAAAGTGC	60
	NF1_F38	GGTTGGTTTCTGGAGCCTTTT	NF1_R38	AGCAACAAACCCCAAATCAA	60
	NF1_F39	AGCCTCACAGTGCTCTTATGG	NF1_R39	GGTGGATTTCTCAATGTGGC	60
	NF1_F40	CAGGCCTGATTCTAGGTAATAGTC	NF1_R40	GGGTGAGTGTCTACATGCTTTC	60
	NF1_F41-42	TTGATTAGGCTGTTCCAATGAA	NF1_R41-42	GCAACTTGGTGTTAGAGCACA	60
	NF1_F43	GAGGTTTGATTTAGGGAACATGA	NF1_R43	CCAATAACACAGTCCATGCAA	55
	NF1_F44-45	TTGCATGGACTGTGTTATTGG	NF1_R44-45	TTTCATTGACCTCAAATTTAAACG	55
	NF1_F46	TTCCTGAATTCATTCCGAGATT	NF1_R46	TTCAACACTGATACCCAAAATGA	55
	NF1_F47	TTCTCAGTCCAGCTAACAGTGTC	NF1_R47	TTTATGAGCTGGTAAGGAAATATACTC	60
	NF1_F48	TCAAATTGAAAGGATTACTTATCTTG	NF1_R48	TGCATTTAAAGTAAGACATAAGGGC	60
	NF1_F49	ATGGCTATTCTTGGAAAGTAGGA	NF1_R49	TGTGAACTTTCTGCTCTGCC	60
	NF1_F50	TGCACATTTAACAGGTACTATGCTC	NF1_R50	CCATGTTACCTTCTAGATTACTCTGC	60
	NF1_F51	TGATTGCTGTTGTTAGGAAATAGG	NF1_R51	AAATGGCATCAAAAACTTTGC	60
	NF1_F52-53	CCAGGGATGTATTAGAGCTTTC	NF1_R52-53	GCTGAATTGTTATCTGTTTGGG	60
	NF1_F54	CAGGCGCTGCTTCTTACTG	NF1_R54	CCTCTTTGCAACCAGTGC	60
	NF1_F55-56	GCACATTATTCTGGGGAATG	NF1_R55-56	TGAACAGAAACCTTCATTTTGC	60
	NF1_F57	GTCTTGTGCATGGCTTTCAG	NF1_R57	TCAACCTTGGAATATCATGTGG	60
	NF1_F58	TGATTGTTTCCTAGAATGTGTCC	NF1_R58	TGCAGCATTACAACATGGG	60
NRAS	N-RAS_F1	TGTAGATGTGGCTCGCCAAT	N-RAS_R1	TGATCCGACAAGTGAGAGAC	55
	N-RAS_F2	GGCAATAGCATTGCATTCCC	N-RAS_R2	TCCCTAGTGTGGTAACCTCA	55
PPP1R16B	PPP16R1B_F2	CACAGGCCACACCATGAG	PPP16R1B_R2	GCCCACACGTCTACTGCAC	66.5
	PPP16R1B_F3	GGGAGTTGTTACTACATTCTTCATAGG	PPP16R1B_R3	CTCATTCCCTCCTCCCTGAG	57
	PPP16R1B_F4	CTGGATTCAAACAGGCCTTC	PPP16R1B_R4	AGAAAGAAGGAAGTTGGAAAAAGA	57
	PPP16R1B_F5	ACAGTAGGATTGCAGGCGAG	PPP16R1B_R5	GCCGATGCGTAGCTAATCTG	57
	PPP16R1B_F6	TGATCTATGGGCATGTCTGC	PPP16R1B_R6	TTCTTGCGGAGGTCCAAG	57
	PPP16R1B_F7	GGATGCTGTGGGAGTTGG	PPP16R1B_R7	GAGCTATATTCAACACCTTAAGCCT	57
	PPP16R1B_F8	TGGGTCACATCCTAAGAGGC	PPP16R1B_R8	CCATCTATCTATCTAGACACCAATCCA	57
	PPP16R1B_F9-10	CATGCCCTGAAAGATGCTTG	PPP16R1B_R9-10	GTTTAAGGGCAGTGCCTGGC	57

	PPP16R1B_F11	TGTACGGAAATTCATAGATTGGG	PPP16R1B_R11	AAGCAGAGCCCACCTCTCC	65
PTPN11	PTPN11_F3	CGACGTGGAAGATGAGATCTGA	PTPN11_R3	CAGTCACAAGCCTTTGGAGTCAG	60
	PTPN11_F13	CAACACTGTAGCCATTGCAACA	PTPN11_R13	CGTATCCAAGAGGCCTAGCAAG	60
RCOR1	RCOR1_F3	TTTCAGCAGACTCTCGCCTC	RCOR1_R3	CAACAGAGCCAGAATCCATC	63
	RCOR1_F4	GCAGACCATAGGGTGTACTCTG	RCOR1_R4	GAAGAAACACTGCTACTCAATACAAAG	55
	RCOR1_F5	GCTGGTCTCATTTCCATTTTC	RCOR1_R5	CAAGTATGCTGCAAAAGAGAGTTAG	55
	RCOR1_F6	CCGGCCTTAAACATTTGATT	RCOR1_R6	CTAAGAGCCCCAGATACACA	55
	RCOR1_F7	AAATCAATTTATCGTTTGTATTCGG	RCOR1_R7	TGCTGCTTTCCACATAGCAC	55
	RCOR1_F8	TGTTTTGGTGCCATGTTAAGG	RCOR1_R8	ATGCTCCTTTGAGATTTGCG	55
	RCOR1_F9-10	TCTCAGAATTCAGCTACAACAGATG	RCOR1_R9-10	AAGCAAATGCGTCAACCTTC	55
	RCOR1_F11	TCTTATCTGGATAAAGAGGTAAGTGTC	RCOR1_R11	TTTAGAAACTGGAGTATCAAGCG	55
	RCOR1_F12	TTGCTGGCTACCTTCTCTTTTC	RCOR1_R12	CACCTAGCCATCTGCATCAC	55
SF3B1	SF3B1_F15-16	TGTGAACATATTCTGCAGTTTGG	SF3B1_R15-16	AAACAAATCAAACAGTATTCGTGTAAC	56.9
SRSF2	SRSF2_F1-2	AGACGCCATTTCCCCAG	SRSF2_R1-2	TTCGAGAAGTACGGGCG	55
SOCS2	SOCS2_F2	CGTTTTGGGATTCGCACTGACTTC	SOCS2_R2	TATTCTTTGCTTCCCACCTCGACC	55
	SOCS2_F3	CCGGTAAACTTTCGCTCACA	SOCS2_F3	CCAGAGCACTCAAGTTTTGGTTC	53
SUZ12	SUZ12_F10	TTAAGGCAAATCCACATTGAC	SUZ12_R10	AAAGCACAAGACCTAACTCTGC	55
	SUZ12_F11	TCCTGCGATCATCAGTTGAG	SUZ12_R11	AGTGGTGGCATGTGCTTG	60
	SUZ12_F12	TTTGACAGTGTCTGCCTTGG	SUZ12_R12	TTGGTTGAGGATTTTGTGAGTC	55
	SUZ12_F12_seq	TGGGGTACAAGAGATTTCTTAACAC			
	SUZ12_F13	AAATTGGAGGGATATTTAGGATAGG	SUZ12_R13	TCATCTGGCTCCTTTGTTCC	55
	SUZ12_F14	TCCTAGTCGTGAGGTTAGATGG	SUZ12_R14	ATTTTAAAACCTGTACATTTCCG	55
	SUZ12_F15	GAAATGTTGCCACTTTGCTG	SUZ12_R15	CATTGGAATAGAATAAAATTTGGG	55
	SUZ12_F16	TGGTTATCACTGTTGCATTTGAC	SUZ12_R16	CATAATTCTTAGAGGATGAATTCCC	55
TET2	TET2_F3-1	TGAACTTCCCACATTAGCTGGT	TET2_R3-1	GAAACTGTAGCACCATTAGGCATT	variable
	TET2_F3-1_seq	GATAGAAATAAACACATTTT			variable
	TET2_F3-2	CAAAAGGCTAATGGAGAAAGACGTA	TET2_R3-2	GCAGAAAAGGAATCCTTAGTGAACA	variable
	TET2_F3-3	GCCAGTAAACTAGCTGCAATGCTAA	TET2_R3-3	TGCCTCATTACGTTTTAGATGGG	variable
	TET2_F3-4	GACCAATGTCAGAACACCTCAA	TET2_R3-4	TTGATTTTGAATACTGATTTTCACCA	variable
	TET2_F3-5	TTGCAACATAAGCCTCATAAACAG	TET2_R3-5	ATTGGCCTGTGCATCTGACTAT	variable
	TET2_F3-6	GCAACTTGCTCAGCAAAGGTACT	TET2_R3-6	TGCTGCCAGACTCAAGATTTAAAA	variable
	TET2_F4	ATACTACATATAATACATTCTAATTCCCTCACTG	TET2_R4	TGTTTACTGCTTTGTGTGTGAAGG	variable
	TET2_F5	CATTTCTCAGGATGTGGTCATAGAAT	TET2_R5	CCCAATTCTCAGGGTCAGATTTA	variable
	TET2_F6	AGACTTATGTATCTTTCATCTAGCTCTGG	TET2_R6	ACTCTCTTCCTTTCAACCAAAGATT	variable
	TET2_F7	ATGCCACAGCTTAATACAGAGTTAGAT	TET2_R7	TGTCATATTGTTCACTTCATCTAAGCTAAT	variable
	TET2_F8	GATGCTTTATTTAGTAATAAAGGCACCA	TET2_R8	TTCAACAATTAAGAGGAAAAGTTAGAATAATATTT	variable
	TET2_F9	TGTCATTCCATTTTGTTTCTGGATA	TET2_R9	AAATTACCCAGTCTTGCATATGTCTT	variable
	TET2_F10	CTGGATCAACTAGGCCACCAAC	TET2_R10	CCAAAATTAACAATGTTCATTTTACAATAAGAG	variable
	TET2_F11-1	GCTCTTATCTTTGCTTAATGGGTGT	TET2_R11-1	TGTACATTTGGTCTAATGGTACAACTG	variable
	TET2_F11-2	AATGGAAACCTATCAGTGGACAAC	TET2_R11-2	TATATATCTGTTGTAAGGCCCTGTGA	variable
TP53	TP53_F5-6	TTCCTCTTCCTACAGTACTC	TP53_R5-6	AGTTGCAAACCAGACCTCAG	55

	TP53_F7-8	AGGTTGGCTCTGACTGTACC	TP53_R7-8	CTTGTCCTGCTTGCTTACCTC	55
TRPS1	TRPS1_F2	TGCAGATGTACAGATCAGCTCTC	TRPS1_R2	AGCCACCCTCAAGTTATTATCTC	62
	TRPS1_F3.1	AAAGAATAAGTAATAGGGAGGGTAAAC	TRPS1_R3.1	TTGACAATTGGCTTGACCAC	64
	TRPS1_F3.2	GACCCTCAAGATATGGCCTG	TRPS1_R3.2	GGCTACCTGTCTGGTACTGGG	64
	TRPS1_F4.1	GCCTCTCAAAGAGTAGCTGTTG	TRPS1_R4.1	TGATAATGACGGAGAAGTGGC	64
	TRPS1_F4.2	AGTCAGGCGGCCTTAATCC	TRPS1_R4.2	CTCTCAACAATTCCCGGTTC	64
	TRPS1_F5	GTTAAGAATATTGCATTTCTTTCTACC	TRPS1_R5	GAGACAGATCATTAAGTTTCACTTCAC	62
	TRPS1_F6	TGACCTCATTATGGGCAGTG	TRPS1_R6	ACTGCAAGCCAGGGAATGG	57
	TRPS1_F7.1	AGCAGAAAGCCTATCCCTGC	TRPS1_R7.1	GATTCCCAGGAACGGAGA	57
	TRPS1_F7.2	ATTATTCACCACCAGGCAGC	TRPS1_R7.2	TGTTGGATAAGGCAGGCTCT	66.5

B

PCR program of *TET2*

Time	Temperature (°C)	cycles number
15''	94	1
20''	94	2
20''	56	
depending amplicon lenght	72	
20''	94	2
20''	54	
depending amplicon	72	
20''	94	2
20''	52	
depending amplicon lenght	72	
20''	94	37
20''	50	
depending amplicon lenght	72	
10'	72	1

', minutes; '', secondes; F, forward; R, reverse.

Supplementary Table S3. Summary of all detected CNAs in 34 primary and secondary myelofibrosis

Diagnosis	Samples	Karyotype	CNA nb	Deletion/ Gain	Chr band	Max Range (bp)	Max Size (bp)	Type of CNA	Nb of genes	Log2 ratio	Genes
post-PV MF	HD-0758	ND	4	D	1p36.11	chr1:25570060-25663551	93 491	2	7	-0.5	*SYF2, C1orf63, RHD, RHCE, TMEM57, LDLRAP1*
				G	1q44	chr1:247419685-249224147	1 804 462	2	>50	+0.5	*BCL9, ETV3, MNDA*
				G	2p25.3	chr2:435421-754937	319 516	2	5	+0.5	*TPO*
				G	9p	chr9:163131-47212417	47 049 287	1	>50	+0.5	*JAK2, MLLT3*
post-PV MF	HD-1401_0842	46,XY,del(8)(q21q23)[7] /46,XY[19]	1	D	8q22.3-8q24.13	chr8:104231983-127127107	22 895 124	1	>30	-0.4	*CSMD3, RAD21, MYC*
post-PV MF	HD-1427_1656	46,XX,t(7;14)(q11;q24)[2] /46,XX[18]	3	D	1p35.1-1p34.3	chr1:33696472-35645300	1 948 828	2	>10	-0.9	*CSMD2, PHC2*
				G	6q22.31	chr6:123527545-124319262	791 717	2	3	+0.5	*CLVS2, TRDN, NKAIN2*
				D	17q11.2	chr17:29071112-30451370	1 380 258	2	5	-0.2	*NF1, SUZ12*
post-PV MF	HD-1537	46,XX[20]	1	D	17q11.2	chr17:29187397-30244809	1 057 412	2	5	-0.2	*NF1*
post-PV MF	HD-1559_1649	45,XY,-7[9]/ 7,XY,+9[3]/46,XY[8]	3	D	7	chr7:45130-159118566	159 073 436	1	total	-0.2	*EZH2, HOXA9, ZNFN1A1, EGFR, MLL3, IKZF1*
				G	9p	chr9:163131-47212417	47049287	1	>50	+0.5	*JAK2, MLLT3*
				D	12p13.31-12p12.3	chr12:7695666-15370548	7 674 882	1	>50	-0.2	*ETV6, CDKN1B*
post-PV MF	HD-1602	46,XX,der(15)t(1;15)(p11;q12)[11]/46,XX[9]	1	G	1q	chr1:143646652-249224147	105 577 495	1	total	+0.2	*HIF1-β, PBX1, RGS2, CR1*
post-PV MF	HD-1664	46,XX,del(20)(q11q13)[3] /46,XX[17]	1	D	20q11-20q13	-	-	1	-	-	*CDR1, CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
post-PV MF	HD-1691_0789	45,XY,-7,del(20)(q11q13)[20]/46,XY[2]	2	D	7	chr7:45130-159118566	159 073 436	1	>100	-0.2	*EZH2, HOXA9, ZNFN1A1, EGFR, MLL3, IKZF1*
				D	20q12-20q13.31	chr20:40216156-54565054	143 48 898	1	>100	-0.2	*CDR2, PTPRT, SRSF6, L3MBTL1*
post-PV MF	HD-1813_1836	45,XY,-7[6]/45,sl,del(12)(p12p13)[3]/ 45,sdl1,add(3)(q2?1)[13]	3	D	7	chr:45130-159118566	159 073 436	1	total	-0.8	*EZH2, HOXA9, ZNFN1A1, EGFR, MLL3, IKZF1*
				D	12p13.2-12p12.3	chr:11743077-16162282	4 419 205	2	>10	-0.8	*ETV6, CDKN1B*
				D	17q11	chr:28926042-30195057	1 269 015	2	>5	-0.8	*NF1,SUZ12*
post-PV MF	HD-1824_0589	46,XX,del(20)(q11q13)[8]/48,XX,+8,+9 [3]/48,idem,der(9)t(1;9)(q21;q12q)[2]/ 46,XX[7]	1	D	20q11.22-20q13.2	chr20:31891717-53143776	21 252 059	1	>50	-0.1	*CDR1, CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
post-ET MF	HD-1264	45,XY,der(7;18)(q10;q10)[20]	2	D	7p22.3-7p11.2	chr7:45130-57278548	57 233 418	1	>100	-0.4	*ETV1, HDAC9, CBX3, IKZF1, JAZF1, IL6, ZNFN1A1, EGFR*
				D	18p11.32-18p11.21	chr18:14316-14130869	14 116 553	1	>50	-0.4	*L3MBTL4, PTPN2*
post-ET MF	HD-1616_0551	47,XY,+9[5]/46,XY[15]	1	D	11q22.3-11q23.3	chr11:106783511-114591737	7 808 226	1	>30	-0.8	*ANKK1, ARGHEF12, ETS1*
PMF	HD-0497_1307	46,XX,del(20)(q11q13)[6] /46,XX[3]	1	D	20q11-20q13.32	chr20:31373532-57649807	26 276 275	1	>50	-0.8	*CDR1, CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
PMF	HD-0586	45,XY,-13[18]/46,XY[2]	2	D	13q12.3-13q34	chr13:30330695-115105806	84 775 111	1	>100	-0.5	*CDX2*
				D	18p11.32-18p11.21	chr18:14316-14796024	14 781 708	1	>50	-0.5	*L3MBTL4, PTPN2*
PMF	HD-0648	46,XX[20]	1	D	14q11-14q12	chr14:20445941-29023459	8 577 518	1	>50	-1	*BCL2L2*
PMF	HD-0689	ND	3	D	4q24	chr4:105992420-106249015	256 595	2	2	-0.3	*TET2*
				D	14q11.2	chr14:23489983-25294985	1 805 002	2	>10	-0.3	*BCL2L2*
				D	17q11.2	chr17:29124299-30227273	1 102 974	2	3	-0.3	*NF1*
PMF	HD-0719	46,XY[20]	1	D	9q34.13-9q34.2	chr9:134453373-135867067	1 413 694	2	10	-1	*SETX*
PMF	HD-0728	46,XX,t(6;17)(p21-22;q11)[10]/46,XX[13]	7	D	6p22.3-6p22.1	chr6:16186392-27130840	43 317 232	1	>30	-0.9	*CDKL1*
				D	6p25.3-6p25.2	chr6:1490872-2821073	1 330 201	2	10	-0.9	*GMDS*
				D	14q32.31-14q32.33	chr14:102027999-106559103	4 531 104	1	>15	-0.9	*RCOR1*
				D	17p13.3-17p13.1	chr17:29169-8883422	8 854 253	1	>30	-0.9	*YWHAE, PAFAH1B1*
				D	17q11.2-17q21.33	chr17:28805330-48948882	20 143 552	1	>50	-0.9	*NF1, SUZ12*
				D	17q25.1-17q25.3	chr17:71663856-81124227	9 460 371	1	>10	-0.9	*SOCS7*
				D	22q11.1-22q13.33	chr22:16124218-51219009	35 094 791	1	>100	-0.2 to -0.9	*NF2, SOX10, SH3BP1*
PMF	HD-0759	46,XX,del(5)(q13q33),del(20) (q11q13)[19]/46,XX[1]	2	D	5q14.3-5q35.1	chr5:91751929-168088896	76 336 967	1	>50	-0.8	*CDC23, MADH5, IRF1, NPM1*
				D	20q11.21-20q13.2	chr20:30784003-50789021	20 005 018	1	>50	-0.8	*ASXL1, DNMT3B, CDR1, CDR2, TOP1, SRSF6, L3MBTL1*
PMF	HD-0777	nd	1	G	18q23	chr18:76690903-77153667	462 764	2	3	+1	*SALL3, ATP9B, NFATC1*
PMF	HD-1047	46,XY,del(20)(q11q13)[3]/46,idemadd(9)(p2?3)[17]	2	G	9p24.3-9p13.3	chr9:163131-34316631	34 153 500	1	>50	+0.5	*JAK2, MLLT3*
				D	20q11.23-20q13.13	chr20:35029598-49772864	14 743 266	1	>50	-1	*CDR1, CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
PMF	HD-1095	ND	4	D	6q23.2-6q23.3	chr6:135507017-137366363	1 859 346	2	10	-0.8	*TNFAIP3*
				D	7p22.1	chr7:5227285-5794960	567 675	2	>5	-0.8	*FBXL18*
				D	10p12.31-10p12.2	chr10:21234380-23388380	2 154 000	2	>10	-0.8	*BMI1,COMMD3,COMMD3-BMI1*
				D	12q22	chr12:93329167-94430427	1 101 260	2	5	-0.8	*SOCS2*
PMF	HD-1100	46,XY,del(13)(q13q22-31)[6]/46,sl,?del(17)(p12-13)[5]/44,sl,der(1)inv(1)(p32~34q23)?dup(1)(q31q32),-7,-16,?der(17)t(16;17)(p11-12;p12-13)[6]/44,sdl1,?del(17)(p12-13)[5] +der(17)t(16;17)(p11-12;p12-13)[6]/44,sdl1,?del(17)(p12-13)[5]	3	D	1p13.3-1p11.2	chr1:110645371-120693961	10 048 590	1	>30	-0.8	*NOTCH2, CD53*
				D	13q12.3-13q21.33	chr13:31686948-71733123	40 046 175	1	>50	-0.9	*RB1, CDX2*
				D	17q22-17q23.1	chr17:56125121-57161042	1 035 921	2	>10	-0.9	*RAD51C*
PMF	HD-1207	46,XY,ider(20)(q10) del(20)(q11q13)[20]	2	D	20p13-20p11.1	chr20:60747-25937994	25 877 247	1	>100	-0.5	*MACROD2*
				D	20q11.21-20q13.33	chr20:31930935-60222113	28 291 178	1	>100	-0.5	*CDR1, CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
PMF	HD-1265_0927	ND	1	D	20q11.21-20q13.32	chr20:38348387-53396700	15 048 313	1	>100	-0.2	*CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
PMF	HD-1283	ND	1	D	20q11.21-20q13.32	chr20:31706596-57020553	25 313 957	1	>100	-0.9	*CDR1, CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
PMF	HD-1289	ND	1	D	20q11.23-20q13.2	chr20:37244953-52543027	15 298 074	1	>100	-0.9	*CDR1, CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
PMF	HD-1300_0528_1611	46, XY, del20(q11q13)[4]/46,XY[16]	1	D	20q11.22-20q13.12	chr20:33873527-47688996	13 815 469	1	>100	-0.2	*CDR1, CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
PMF	HD-1362	46,XX,dup(1)(q44q12), del(13)(q13q14)[20]	6	G	1q21.2-1q44	chr1:149041013-248808428	99 767 415	1	>100	+0.5	*SPTA1*
				D	8q23.3	chr8:116508962-117333595	824 633	2	1	-0.8	*TRPS1*
				D	13q14.2-13q21.1	chr13:47729733-57537767	9 808 034	1	>30	-0.9	*RB1*
				D	13q31.1-13q31.2	chr13:87547986-88697857	1 149 871	2	1	-0.9	*SLITRK5*
				D	18q21.2-18q21.33	chr18:52787524-60012588	7 225 064	1	>20	-0.8	*TCF4*
				D	20q11.21-20q12	chr20:31984856-38055012	6 070 156	1	>50	-0.4	*CDR1*
PMF	HD-1422	ND	1	D	17q23.1-17q23.2	chr17:57404219-58144817	740 598	2	>10	-0.8	*miR-21, TMEM49, TUBD1, RPS6KB1*
PMF	HD-1524	ND	1	D	11q14.1-11q24.2	chr11:81041293-127474614	46 433 321	1	>100	-1	*EED, ARGHEF12, ETS1*
PMF	HD-1538	ND	4	D	1p32.2-1p32.3	chr1:54184496-55619136	1 434 640	2	>20	-0.9	*GLIS1, DHC24, USP24, TTC4, TTC22*
				D	20q11.21-20q12	chr20:31219192-37630533	6 411 341	1	>50	-0.8	*CDR1*
				D	20q13.12-20q13.13	chr20:43752761-48061828	4 309 067	1	>20	-0.8	*CDR2*
				D	22q12.1	chr22:28674993-29966075	1 291 082	2	>10	-0.8	*NF2*
PMF	HD-1587	ND	3	D	6q21	chr6:107735547-110341820	2 606 273	2	>10	-0.2	*FOXO3*
				D	6q22.31-6q23.3	chr6:125619860-138310979	12 691 119	1	>30	-0.2	*MYB*
				D	20q11.22-20q13.13	chr20:33909838-47688996	13 779 158	1	>50	-0.2	*CDR1, CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
PMF	HD-1654	46,XY,inv(11)(q12q22)[20]	1	D	7q22.1	chr7:101066625-102101201	1034576	2	>5	-0.4	*CUX1, SH2B2*

bp, base pairs; CDR, commonly deleted region; Chr, chromosome; CNA, copy number aberrations; D, deletion; G, gain; MF, myelofibrosis; nb, number; nd, not determined; PMF, primary myelofibrosis; post-ET MF, post-essential thrombocythemia MF; post-PV MF, post-polycythemia vera MF.

Supplemental Table S4. List of genes located in the two 20q commonly deleted regions (CDR1 and CDR2).

CDR1	Functions
LOC388796	uncharacterized
SNORA71A	small nucleolar RNA, H/ACA box 71A
SNORA71B	small nucleolar RNA, H/ACA box 71B
SNORA71C	small nucleolar RNA, H/ACA box 71C
SNORA71D	small nucleolar RNA, H/ACA box 71D
SNHG11	member of the non-protein-coding multiple snoRNA host gene family, two snoRNAs are derived from the introns of this host gene
SNORA30	snoRNAs are derived from the introns of SNHG11
SNORA60	snoRNAs are derived from the introns of SNHG11
RALGAPB	Ral GTPase activating protein, beta subunit (non-catalytic)
MIR548O2	microRNA
RPS3P2	ribosomal protein S3 pseudogene 2
ADIG/SMAF1	adipocyte differentiation
ARHGAP40	Rho GTPase activating protein 40
LOC391247	INS complex subunit 2 (Psf2 homolog) pseudogene
SLC32A1	member of amino acid/polyamine transporter family II
ACTR5	in yeast, DNA double strand break, chromatin remodelling
PPP1R16B	synthesis of the encoded protein is inhibited by transforming growth factor beta-1 mRNAoverexpressed in hematopoietic cells
FAM83D	family with sequence similarity 83, member D
DHX35	member of the DEAD box protein family are believed to be involved in embryogenesis, spermatogenesis, and cellular growth and division
NPM1P19	nucleophosmin 1 (nucleolar phosphoprotein B23, numatrin) pseudogene 19
LOC339568	uncharacterized

CDR2	Functions
YWHAB	interaction with RAF1 and CDC25 phosphatases, suggesting that it may play a role in linking mitogenic signaling and the cell cycle machinery
PABPC1L	poly(A) binding protein, cytoplasmic 1-like
TOMM34	chaperone-like activity, binding the mature portion of unfolded proteins and aiding their import into mitochondria
STK4	cytoplasmic kinase that is structurally similar to the yeast Ste20p kinase, which acts upstream of the stress-induced mitogen-activated protein kinase cascade
KCNS1	belongs to the S subfamily of the potassium channel family
WFDC5	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
WFDC12	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
PI3	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
SEMG1	predominant protein in semen
SEMG2	predominant protein in semen
SLPI	protects epithelial tissues from serine proteases
MATN4	member of von Willebrand factor A domain containing protein family

RBPJL	recombination signal binding protein for immunoglobulin kappa J region-like
SDC4	a syndecan-4/CXCR4 complex expressed on human primary lymphocytes and macrophages and HeLa cell line binds the CXC chemokine stromal cell-derived factor-1 (SDF-1)
SYS1	nonsense-mediated mRNA decay
SYS1-DBNDD2	nonsense-mediated mRNA decay
TP53TG5	TP53 target 5
PIGT	chosphatidylinositol glycan anchor biosynthesis, class T
AK127953	uncharacterized
WFDC2	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
WFDC6	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
SPINLW1-WFDC6	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
WFDC8	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
WFDC9	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
WFDC11	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
WFDC10B	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
WFDC13	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
WFDC3	member of the WAP-type four-disulfide core (WFDC) domain family , contains eight cysteines forming four disulfide bonds at the core of the protein, and functions as a protease inhibitor
DNTTIP1	deoxynucleotidyltransferase, terminal, interacting protein 1
UBE2C	destruction of mitotic cyclins and for cell cycle progression
TNNC2	regulation of striated muscle contraction
SNX21	members of this family contain a phox (PX) domain, which is a phosphoinositide binding domain, and are involved in intracellular trafficking
ACOT8	peroxisomal thioesterase that appears to be involved more in the oxidation of fatty acids rather than in their formatio
ZSWIM3	zinc finger, SWIM-type containing 3
ZSWIM1	zinc finger, SWIM-type containing 1
C20orf165	uncharacterized
NEURL2	non-protein coding RNA due to the presence of an upstream open reading frame (uORF)
CTSA	glycoprotein which associates with lysosomal enzymes beta-galactosidase and neuraminidase to form a complex of high molecular weight multimers
PLTP	lipid transfer proteins
AK097925	uncharacterized
PCIF1	PDX1 C-terminal inhibiting factor 1
ZNF335	transcriptional activation by ligand-bound nuclear hormone receptors
MMP9	proteins of the matrix metalloproteinase
SLC12A5	an integral membrane K-Cl cotransporter
NCOA5	coregulator for the alpha and beta estrogen receptors and the orphan nuclear receptor NR1D2
CD40	member of the TNF-receptor superfamily
CDH22	cadherin 22, type 2
SLC35C2	solute carrier family 35, member C2
ELMO2	phagocytosis of apoptotic cells and in cell migration
ZNF334	zinc finger protein 334
C20orf123	uncharacterized
SLC13A3	Solute carrier family 13 (sodium-dependent dicarboxylate transporter), member 3
TP53RK	TP53 regulating kinase

SLC2A10	regulation of glucose homeostasis
BC065739	uncharacterized
EYA2	role in eye development
ZMYND8	receptor for activated C-kinase (RACK) protein
NCOA3	uclear receptor coactivator that interacts with nuclear hormone receptors to enhance their transcriptional activator functions
SULF2	cell signaling
AK129540	uncharacterized
LOC284749	uncharacterized
PREX1	guanine nucleotide exchange factor for the RHO family of small GTP-binding proteins (RACs)
ARFGEF2	play an important role in intracellular vesicular trafficking
CSE1L	poptosis and in cell proliferation
STAU1	member of the family of double-stranded RNA (dsRNA)-binding proteins involved in the transport and/or localization of mRNAs to different subcellular compartments and/or organelles
DDX27	member of the DEAD box protein family are believed to be involved in embryogenesis, spermatogenesis, and cellular growth and division
ZNFX1	zinc finger, NFX1-type containing 1
ZFAS1	potential marker for breast cancer
SNORD12	small nucleolar RNA, C/D box 12
SNORD12B	small nucleolar RNA, C/D box 12B
SNORD12C	small nucleolar RNA, C/D box 12C
KCNB1	potassium voltage-gated channel
PTGIS	member of the cytochrome P450 superfamily of enzymes
B4GALT5	membrane-bound glycoproteins

Supplemental Table S5.

Gene mutations of 68 MF cases with TET2, IDH1/2, DNMT3A, ASXL1, EZH2, SUZ12, BMI1, RCOR1, JAK2, MPL, CBL, SOCS2, LNK, NF1, KRAS, NRAS, PTPN11, PPP1R16B, TRPS1, SF3B1, SRSF2 and TP53 mutations.

Diagnosis	Samples	*TET2* (E3-11)	*IDH1/2* (E4)	*DNMT3A* (E15-23)	*ASXL1* (E12)	*EZH2* (E2-20)	*SUZ12* (E10-16)	*BMI1* (E1-10)	*RCOR1* (E3-12)	*JAK2* (E12 ;14)	*MPL* (E10)	*CBL* (E8-9)	*SOCS2* (E1-2)	*LNK* (E2-8)	*NF1* (E1-58)	*KRAS* (E2-4)	*NRAS* (E1-2)	*PTPN11* (E3;13)	*PPP1R16B* (2-11)	*TRPS1* (E2-7)	*SF3B1* (E15-16)	*SRSF2* (E2)	*TP* (E5
post-PV MF	HD-0577	wt	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	wt	wt	wt	wt	w
post-PV MF	HD-0758	wt	wt	wt	**c.1774C>T;p.Q592X**	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	ND	wt	wt	wt	ND	ND	wt	wt	w
post-PV MF	HD-1302	wt	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	ND	ND	wt	wt	w
post-PV MF	HD-1401_0842	wt	wt	wt	**c.2324T>A;p.L775X**$^{N}_{C}$	**c.446T>G;p.L149R**$^{N}_{C}$	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	ND	ND	ND	wt	w
post-PV MF	HD-1426	**c.4462A>T p.K1488X**	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	**c.639C>A;p.S213R**	ND	wt	wt	wt	ND	ND	wt	ND	w
post-PV MF	HD-1427_1656	wt	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
post-PV MF	HD-1480	wt	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	wt	wt	wt	wt	w
post-PV MF	HD-1537	wt	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
post-PV MF	HD-1559_1649	wt	wt	**c.2711C>T;p.P904L**	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	wt	wt	wt	wt	w
post-PV MF	HD-1570bis	wt	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	ND	ND	ND	wt	wt	w
post-PV MF	HD-1602	**c.1526C>A;p.S509X and c.3594_3594+1G>C**	wt	wt	**c.1934dupG;p.G646WfsX12**	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	ND	ND	ND	wt	wt	w
post-PV MF	HD-1664	ND	ND	ND	ND	ND	ND	ND	ND	**p.V617F**	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	wt	N
post-PV MF	HD-1691_0789	wt	wt	wt	**c.1934dupG;p.G646WfsX12**	ND	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	ND	ND	ND	wt	ND	w
post-PV MF	HD-1813_1836	ND	ND	ND	ND	ND	ND	ND	ND	**p.V617F**	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	wt	N
post-PV MF	HD-1824_0589	wt	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	ND	w
post-ET MF	HD-0554	wt	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	**c.2098A>G;p.K700E**	wt	w
post-ET MF	HD-0598	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
post-ET MF	HD-0599	wt	wt	wt	wt	ND	wt	wt	wt	**p.V617F**	wt	wt	wt	**c.1336A>G;p.I446V**	wt	wt	wt	ND	wt	wt	wt	wt	w

post-ET MF	HD-0601	**c.5693C>T;p.S1898F**	wt	wt	**c.1888-1909del;p.H630PfsX66**	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	w
post-ET MF	HD-0614_1112_1352	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
post-ET MF	HD-0725_1380	**c.3820C>T;p.Q1274X^{NC} aND c.4960C>T;p.Q1654X**	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
post-ET MF	HD-0983	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
post-ET MF	HD-1264	wt	wt	wt	wt	wt	wt	ND	wt	wt	wt	wt	ND	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
post-ET MF	HD-1309_1741	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	wt	ND	wt	ND	wt	wt	wt	ND	ND	ND	wt	w
post-ET MF	HD-1360_1605	wt	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	wt	wt	wt	wt	w
post-ET MF	HD-1376	**c.5618T>C;p.I1873T aND c.5326delT;p.S1776LfsX44**	wt	wt	**c.1934dupG;p.G646WfsX12**	**c.2192A>G;p.Y731C**	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	wt	wt	wt	wt	w
post-ET MF	HD-1462	wt	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	ND	ND	wt	wt	w
post-ET MF	HD-1569	wt	wt	wt	wt	ND	wt	ND	ND	**p.V617F**	ND	wt	ND	wt	ND	ND	wt	ND	ND	ND	ND	wt	w
post-ET MF	HD-1616_0551	wt	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	wt	wt	wt	wt	w
PMF	HD-0497_1307	**c.1996_1997delGA;p.D666PfsX14^{NC}**	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-0586	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	ND	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-0607_1130	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-0616	wt	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-0648	wt	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	w
PMF	HD-0679_1291	wt	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	w
PMF	HD-0683	wt	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	w
PMF	HD-0689	wt	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-0717_1244	**c.1972delC;p.H658TfsX42**	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-0719	wt	wt	**c.2644G>A;p.R882S**	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	**c.35G>A;p.G12D**	wt	ND	ND	wt	**c.284_307del;p.P95R**	w
PMF	HD-0728	wt	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-0759	wt	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	wt	ND	wt	wt	**c.66 >T;p 22**

PMF	HD-0777	**c.5602C>T;p.H1868Y**	wt	wt	wt	**c.2050C>T;p.R684C**	wt	wt	wt	wt	wt	wt	wt	wt	ND	wt	wt	**c.1471C>T;p.P491S**	ND	ND	wt	wt	w
PMF	HD-0862	wt	wt	wt	**c.1934dupG;p.G646WfsX12**	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	ND	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-1047	wt	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-1095	wt	wt	wt	wt	wt	wt	wt	ND	**p.V617F**	wt	wt	wt	wt	ND	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-1098	wt	wt	wt	wt	ND	wt	wt	ND	wt	wt	wt	wt	wt	ND	wt	wt	ND	wt	wt	wt	ND	w
PMF	HD-1100	wt	wt	wt	**c.1934dupG;p.G646WfsX12**	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	**c.73 >A; 24**
PMF	HD-1111	wt	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	w
PMF	HD-1123	wt	wt	wt	**c.1934dupG;p.G646WfsX12**	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	**c.3790G>A;p.E1264K^{NC}**	wt	wt	wt	ND	ND	wt	**c.283C>G;p.P95A**	w
PMF	HD-1138_0540_1398	wt	wt	wt	**c.1934dupG;p.G646WfsX12**	wt	wt	wt	wt	wt	**c.1543T>A;p.W515R^{NC}**	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-1207	wt	wt	wt	wt	wt	wt	ND	wt	wt	wt	wt	ND	wt	wt	wt	wt	wt	ND	ND	**c.2098A>G;p.K700E**	wt	w
PMF	HD-1208	wt	wt	wt	wt	**c.2050C>T;p.R684C**	wt	ND	wt	**p.V617F**	wt	wt	ND	**c.764_765delinsG;p.S255X aND c.1566dupC;E523RfsX23**	ND	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-1227	wt	wt	wt	wt	wt	wt	ND	wt	**p.V617F**	wt	wt	ND	wt	**c.8499T>G;p.D2833K^{NC}**	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-1265_0927_1461_1853	wt	wt	wt	**c.1934dupG;p.G646WfsX12**	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	**c.284C>A;p.P95H**	w
PMF	HD-1283	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	wt	ND	wt	ND	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-1289	**c.481_482insT;p.K161IfsX10^{NC}**	wt	wt	**c.1934dupG;p.G646WfsX12^{NC}**	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	**c.1508G>A;p.G503E^{NC}**	ND	ND	wt	wt	w
PMF	HD-1300_0528_1611	**c.4062delA;p.A1355HfsX8^{NC} aND c.1648C>T; p.Arg550X^{NC}**	wt	wt	wt	wt	wt	wt	wt	**p.V617F**	wt	wt	wt	wt	wt	wt	wt	wt	ND	ND	wt	**c.284C>A;p.P95H**	w
PMF	HD-1362	wt	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-1412_1495	wt	wt	wt	wt	wt	wt	ND	ND	wt	wt	wt	ND	wt	ND	wt	wt	wt	wt	wt	**c.2111T>G;p.I704S**	wt	w
PMF	HD-1422	wt	wt	wt	**c.1934dupG;p.G646WfsX12**	wt	wt	ND	ND	wt	wt	wt	ND	wt	ND	wt	wt	wt	wt	wt	wt	wt	w
PMF	HD-1439	ND	wt	wt	**c.2077C>T;p.R693X**	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-1524bis	wt	wt	wt	**c.1934dupG;p.G646WfsX12**	**c.2036T>A;p.V679E**	wt	ND	ND	wt	wt	**c.1301T>C;p.F434S**	ND	wt	ND	wt	wt	wt	ND	ND	wt	wt	w

PMF	HD-1538	wt	wt	wt	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	wt	wt	wt	wt	w
PMF	HD-1572bis	wt	wt	wt	**c.1934dupG;p.G646WfsX12 aND c.2418_2423delinsG;p.V807CfsX13**	wt	wt	ND	ND	wt	wt	wt	ND	wt	ND	wt	ND	ND	ND	ND	ND	wt	w
PMF	HD-1587	ND	wt	ND	wt	wt	wt	ND	ND	wt	wt	wt	ND	ND	ND	ND	ND	wt	ND	ND	ND	wt	N
PMF	HD-1606	wt	wt	**c.2431_2433del;p.D811K**	wt	wt	wt	ND	ND	**p.V617F**	wt	wt	ND	wt	ND	wt	wt	wt	wt	wt	wt	wt	w
PMF	HD-1617	wt	wt	wt	**c.1860insGGCG p.L622CfsX14**	wt	wt	ND	ND	wt	wt	wt	ND	wt	ND	wt	wt	wt	ND	ND	wt	wt	w
PMF	HD-1654	ND	ND	ND	ND	ND	ND	ND	ND	wt	wt	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	wt	N

References for nucleotide and protein sequences
TET2, NM_001127208.1, NP_001120680.1; IDH1, NM_005896.2, NP_005887.2; IDH2, NM_002168.2, NP_002159.2; DNMT3A, NM_022552.3, NP_072046.2; ASXL1, NM_015338.5 NP_056153.2; EZH2, NM_004456.4, NP_004447.2; SUZ12, NM_015355.2, NP_056170.2; BMI1, NM_005180.8, NP_005171.4;
RCOR1, NM_015156.3, NP_055971.2; JAK2, NM_004972.3, NP_004963.1; MPL, NM_005373.2, NP_005364.1; CBL, NM_005188.2, NP_005179.2; SOCS2, NM_001270470.1, NP_001257399.1; NF1, NM_001042492.2, NP_001035957.1; LNK, NM_005475.2, NP_005466.1; KRAS, NM_004985.3, NP_004976.2 and NM_033360.2, NP_203524.1; NRAS, NM_002524.4, NP_002515.1; PTPN11, NM_002834.3, NP_002825.3; PPP1R16B, NM_015568.2, NP_056383.1; TRPS1, NM_014112.2, NP_054831.2; SF3B1, NM_012433.2, NP_036565.2; SRSF2, NM_003016.4, NP_003007.2 TP53, NM_000546.5, NP_000537.3.

MF, myelofibrosis; NC, not constitutional mutation; ND, not determined; PMF, primary myelofibrosis; Post-ET MF, post-essential thrombocythemia MF; post-PV MF, post-polycythemia vera MF; wt, wild-type.

Supplementary Table S6. Features of MF cases with gene mutations

Genes		*JAK2*			*LNK*			*PTPN11*			Signaling (*CBL, JAK2, LNK, MPL, NF1, NRAS, PTPN11*)			*ASXL1*		
Status		mut	wt	p	mut	wt	p	mut	wt	p	mut	wt	p	mut	wt	p
MF	n	47	21		3	62		2	61		49	8		17	49	
Sex	M	24	13	0.44	2	34	0.41	1	33	0.08	26	7	0.15	8	28	0.47
	F	23	8		1	28		1	28		24	1		9	21	
MF	primary	24	16	**0.06**	1	37	0.61	2	36	0.51	27	4	1	11	29	0.77
	secondary	23	5		3	35		0	25		23	4		6	21	
Diag	PMF	24	16	**0.009**	1	37	0.66	2	36	0.51	27	4	0.07	11	28	0.53
	post-PV MF	14	0		1	12		0	13		14	0		4	9	
	post-ET MF	9	5		1	13		0	12		9	4		2	12	
Age (y) median (range)		70 (30-89)	65 (32-86)	0.51	80 (73-82)	66 (30-89)	**0.05**	80.5 (75-86)	66 (30-89)	0.06	69.5 (30-89)	64.5 (32-75)	0.21	71 (57-83)	65 (30-89)	**0.02**
Leukocyte count (x 10^9/L); median (range)		11.5 (1.3-48.8)	7.4 (2.6-120)	0.22	8.6 (4.1-20.7)	11.4 (1.3-120)	0.74	32.8 (11.4-54.2)	11.2 (1.3-120)	0.26	13 (1.3-120)	7 (2.6-15.2)	0.11	12.8 (4.6-76)	8.45 (1.3-120)	**0.01**
Hemoglobin count (g/dL); median (range)		11.5 (7.6-17.8)	11.5 (5.8-14.5)	0.28	9.2 (8.7-15)	11.8 (5.8-17.8)	0.61	10.1 (7.7-12.5)	11.7 (5.8-17.8)	0.47	11.5 (5.8-17.8)	12.1 (7.2-14.5)	0.92	12 (5.8-14.2)	11.5 (7.2-17.8)	0.98
Hematocrit count (%); median (range)		33.3 (14.3-52)	34.8 (18-44.3)	0.60	25.6 (14.3-27.1)	35.8 (14.3-52)	**0.04**	31.2 (25.2-37.1)	35 (14.3-52)	0.69	33.5 (14.3-52)	35 (20.2-44.3)	0.87	34.65 (18-47)	35 (14.3-52)	0.79
Platelet count (x 10^9/L); median (range)		300 (39-1188)	266 (5-890)	0.77	361 (101-386)	302 (5-1188)	1	425 (88-761)	300 (5-1188)	0.78	315 (26-1188)	301.5 (5-758)	1	334 (26-1188)	284.5 (5-890)	0.47

Genes		*EZH2*			*TET2*			Epigenetic (*ASXL1, EZH2, DNMT3A, TET2*)			*SRSF2*			*SF3B1*			Splicing (*SRSF2, SF3B1*		
Status		mut	wt	p	mut	wt	p	mut	wt	p	mut	wt	p	mut	wt	p	mut	wt	p
MF	n	5	58		9	55		27	36		4	62		3	58		7	53	
Sex	M	3	31	0.78	6	30	0.50	15	19	0.83	3	27	0.12	1	34	0.39	4	25	0.6
	F	2	27		3	25		12	17		0	23		2	24		2	20	
MF	primary	3	35	1	5	32	1	18	20	0.44	3	30	0.28	2	35	1	5	28	0.4
	secondary	2	23		4	23		9	16		0	20		1	23		1	17	
Diag	PMF	3	35	1	5	32	0.98	18	20	0.38	3	30	0.38	2	35	0.62	5	28	0.4
	post-PV MF	1	12		2	11		6	7		0	10		0	12		0	9	
	post-ET MF	1	11		2	12		3	9		0	10		1	11		1	8	
Age (y) median (range)		73 (66-86)	66 (30-89)	0.09	75 (55-86)	66 (30-89)	**0.03**	73 (39-86)	64 (30-89)	**0.002**	75 (55-85)	66 (30-89)	**0.27**	64 (59-79)	70 (30-89)	0.83	75 (59-85)	69 (30-86)	0.2
Leukocyte count (x 10^9/L); median (range)		35.4 (8.6-76)	11.0 (1.3-120)	**0.009**	20.7 (2.4-54.2)	9.85 (1.3-120)	0.12	12.8 (2-120)	6.4 (1.3-35.1)	**0.004**	35.7 (27.3-120)	9.3 (1.3-76)	**0.006**	8.3 (2.6-12)	11.4 (1.3-120)	0.37	19.65 (2.6-120)	9.85 (1.3-76)	0.2
Hemoglobin count (g/dL); median (range)		8.7 (5.8-13.4)	11.7 (7.2-17.8)	0.18	12.5 (7.6-15)	11.55 (5.8-17.8)	0.56	12 (5.8-15)	11.5 (7.2-17.8)	0.83	10.9 (8.6-13.2)	11.5 (5.8-17.8)	0.71	10.2 (7.2-13.2)	11.8 (5.8-17.8)	0.34	10.2 (7.2-13.2)	11.65 (5.8-15.7)	0.2
Hematocrit count (%); median (range)		25.4 (18-41.2)	35 (14.3-52)	0.19	35.3 (14.3-44.3)	35 (14.3-52)	0.59	33.5 (14.3-47)	35 (14.3-52)	0.62	34 (26.0-38.6)	35 (14.3-51.9)	0.89	30.95 (20.2-41.7)	35 (14.3-52)	0.77	32 (20.2-41.7)	35 (18-51.9)	0.5
Platelet count (x 10^9/L); median (range)		315 (26-761)	297 (5-1188)	0.87	315 (88-761)	308.5 (5-1188)	0.61	315 (26-1188)	269 (5-890)	0.68	568.5 (295-1188)	272 (5-890)	**0.04**	77 (5-540)	302 (26-1188)	0.25	509.5 (5-1188)	296 (26-890)	0.4

Diag, diagnosis; DIPSS, dynamic international prognostic scoring system ; F, female ; Inter, intermediate ; IPSS, international prognostic scoring system ; M, male ; MF, myelofibrosis ; mut, mutated ; PMF, primary myelofibrosis ; post-ET MF, post-essential thrombocythemia MF ; post-PV MF, post-polycythemia vera MF ; wt, wild-type ; y, years. For PMF, IPSS, DIPSS and DIPPSplus scores depend on age (>65 years), on hemoglobin (< 10 g/dL), on leukocyte count (> 25.10^9/L), on circulating blasts (> or = 1%), on platelet count (< 100.10^9/L), on presence of constitutional symptoms (weight loss, night sweats, fever). For DIPPSplus score, karyotype and transfusion status must be added to DIPSS. IPSS score was calculated at MF diagnosis whereas DIPPS and DIPPSplus score were calculated at sampling.

Supplementary Table S7. Molecular data of 19 matched pairs of MF

A.Evolution to MF

Diagnosis	Samples	M	Add	Karyotype	aCGH	12p
PV	HD-0842_1401	24	yes	ND	No CNA	
post-PV MF	HD-1401_0842			46,XY,del(8)(q21q23)[7]/46,XY[19]	del 8q22.3-8q24.13(**-0.4**)	
ET	HD-0551_1616	61	yes	ND	del 11q22.3-11q23.3(**-0.1**)	
post-ET MF	HD-1616_0551			47,XY,+9[5]/46,XY[15]	gain 9 (+0.1), del 11q22.3-11q23.3(**-0.8**)	

B.MF disease course

Diagnosis	Samples	M	Add	Karyotype	aCGH	12p
PMF	HD-1412_1495	2	no	ND	No CNA	
PMF	HD-1495_1412			46,XY[2]	No CNA	
post-ET MF	HD-0725_1380	28	no	46,XY[20]	No CNA	
post-ET MF	HD-1380_0725			46,XY[20]	No CNA	
PMF	HD-0497_1307	44	no	46,XX,del(20)(q11q13)[6]/46,XX[3]	del 20q11-q13.32(**-0.8**)	
PMF	HD-1307_0497			ND	del 20q11-q13.32(**-1**)	
PMF	HD-0607_1130	32	no	46,XY[20]	No CNA	
PMF	HD-1130_0607			46,XY[20]	No CNA	
PMF	HD-0679_1291	26	no	46,XY[20]	No CNA	
PMF	HD-1291_0679			ND	No CNA	
PMF	HD-0717_1244	23	no	46,XY[20]	No CNA	
PMF	HD-1244_0717			ND	No CNA	
post-PV MF	HD-0589_1824	62	no	48,XX,+8,+9[3]/46,XX,del(20)(q11q13)[2]/46,XX[15]	No CNA	
post-PV MF	HD-1824_0589			46,XX,del(20)(q11q13)[8]/48,XX,+8,+9[3]/48,idem, der(9)t(1;9)(q21;q12)[2]/46,XX[7]	del 20q11.22-q13.2(**-0.1**)	
post-PV MF	HD-1427_1656	9	no	46,XX,t(7;14)(q11;q24)[2]/46,XX[18]	del 1p35.1-1p34.3(-0.8), gain 6q22.31(+0.5), del 17q11.2(-0.2)	
post-PV MF	HD-1656_1427			46,XX[20]	del 1p35.1-1p34.3(**-1**), gain 6q22.31(+0.5), del 17q11.2(**-0.5**)	
post-PV MF	HD-0789_1691	37	yes	46, XY, del(20)(q11q13)[4]/46,XY[16]	No CNA	
post-PV MF	HD-1691_0789			45,XY,-7,del(20)(q11q13)[20]/46,XY[2]	del 7(**-0.2**), del 20q12-q13.31(**-0.2**)	
post-ET MF	HD-1360_1605	8	yes	ND	No CNA	
post-ET MF	HD-1605_1360			ND	del 12p13.2 (**-0.9**)	**del**
post-ET MF	HD-0614_1112_1352	29	yes	46,XY[22]	No CNA	
post-ET MF	HD-1112_0614_1352			ND	No CNA	
post-ET MF	HD-1352_0614_1112	9		46,XY,del(7)(q22q36)[2] /46,XY,add(4)(q21-22),-12,-13,+der(?)t(?;4)(?;q21-22),+mar[3]/44,XY,add(4),der(6)t(4;6)(q21-22;p2?3),-7,-12,-15,-16,+2mar[3]	del 4p15.31(**-0.4**), del 4q24 (**-0.4**), de l6p22.1-p25.3(**-0.2**), del 7p12.1-7p22.3(**-0.2**), del 12q23.1(**-0.2**), del 13q14.3(**-0.2**)	
PMF	HD-0540_1138_1398	36	yes	46,XY[20]	No CNA	
PMF	HD-1138_0540_1398	9		ND	No CNA	
PMF	HD-1398_0540_1138			ND	No CNA	

C. Blast phase MF transformation

Diagnosis	Samples	M	Add	Karyotype	aCGH	12p
post-PV MF	HD-1559_1649	4	no	45,XY,-7[9]/47,XY,+9[3]/46,XY[8]	dell 7(**-0.2**), gain 9p(**+0.5**), del 12p12.3-p13.31(**-0.9**)	**del**
BP post-PV MF	HD-1649_1559			45,XY,-7[20]	del 7(**-0.2**), del 12p12.3-p13.31(**-0.2**)	**del**
post-PV MF	HD-1813_1836	1	no	45,XY,-7[6]/45,sl,del(12)(p12p13)[3] /45,sdl1,add(3)(q2?1)[13]	del 7 (-0.8), del 12p (-0.8), del 17q11 (-0.8)	**del**
BP post-PV MF	HD-1836_1813			45,XY,-7[6]/45,sl,del(12)(p12p13)[3] /45,sdl1,add(3)(q2?1)[13]	ND	**del**
PMF	HD-0927_1265_1461_1853		yes	ND	No CNA	
PMF	HD-1265_0927_1461_1853	13		ND	del 20q11-q13.32(-0.2)	
PMF	HD-1461_0927_1265_1853	8		ND	del 20q11-q13.32(-0.1)	
BP PMF	HD-1853_0927_1265_1461	15		46,XY,+8[16]/46,XY[6]	trisomy 8 (+0.2)	ND
PMF	HD-0528_1300_1611	47	yes	46, XY[20]	No CNA	
PMF	HD-1300_0528_1611			46, XY, del20(q11q13)[4]/46,XY[16]	No CNA	
BP PMF	HD-1611_1300_0528	12		46,XY,del(20)(q11q13)[12]/46,XY,inv(3)(q21q26)[4]/ 46,idem,del(6)(q2?1q2?6)[3]/46,XY[1]	del 6q12-q24.1(**-0,2**), del 20q11.22-q13.12(**-0,2**)	
post-ET MF	HD-1309_1741	14	yes	46,XX[20]	No CNA	
BP post-ET MF	HD-1741_1309			46,XX,-3,der(5)t(3;5)(q13;q21), del(7)(q21q36),t(9;17)(q11;q11),+21[11]	del 3p25.2-p26.3 (**-0.8**), del3 p22.1-p25.1(**-0.8**), del 3p12.1-p21.2(**-0.8**), del 5q14.1-q35.2(**-0.8**), del 7q21.11-q35(**-0.8**), del 9p12-p13.3(**-0.8**), del 12p12.3-p13.2(**-0.8**), del 17p12-p13.1(**-0.8**), gain 21(**+0.5**)	**del**

For each CGH data, log2 ratio of the gain or loss is in parenthesis. The number of crosses corresponds to the number of mutations identified. Add , additional alterations (CNAs and mutations); BP, blast phase; CNA, copy number aberrations; del, deletion; M, months; MF, myelofibrosis; ND, not determined; PMF, primary MF; post-ET MF, post- essential thrombocythemia MF; post-PV MF, post-polycythemia vera MF

Supplemental Table S8. Summary of all detected CNAs in 14 blast phase MF

Diagnosis	Samples	Karyotype	CNA nb	Deletion/Gain	Chr baND	Max range (bp)	Max size (bp)	Type of CNA	Nb of genes	Log2 ratio	Genes
Blast phase post-PV MF	HD-0492	47 XY,+8[17]/48,idem,+9[3]	1	G	8p23.3-8q24.3	chr8:73810-146294098	146 220 288	1	>200	+0.6	
Blast phase post-PV MF	HD-1517	46,XY[20]	1	D	21q22.11-21q22.12	chr21:34863258-36788958	1 925 700	2	>10	-0.2	*RUNX1*
Blast phase post-PV MF	HD-1649_1559	45,XY,-7[20]	2	D	7	chr7:45130-159118566	159 073 436	1	>250	-1	*EZH2, HOXA9, ZNFN1A1, EGFR, MLL3, IKZF1*
				D	12p12.3-12p13.31	chr12:7695666-15359104	7 663 438	1	>50	-1	*ETV6, CDKN1B, p27*
Blast phase post-PV MF	HD-1691_0789	45,XY,-7,del(20)(q11q13)[20]/46,XY[2]	2	D	7	chr7:45130-159118566	159 073 436	1	>200	-0.2	*EZH2, HOXA9, ZNFN1A1, EGFR, MLL3, IKZF1*
				D	20q12-20q13.2	chr20:40216156-54565054	14 348 898	1	>100	-0.2	*CDR1, CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
Blast phase post-PV MF	HD-1836_1813	45,XY,-7[6]/45,sl,del(12)(p12p13)[3]/45,sdl1,add(3)(q2?1)[13]	3	D	7	chr:45130-159118566	159 073 436	1	total	-0.8	*EZH2, HOXA9, ZNFN1A1, EGFR, MLL3, IKZF1*
				D	12p13.2-12p12.3	chr:11743077-16162282	4 419 205	2	>10	-0.8	*ETV6, CDKN1B*
				D	17q11	chr:28926042-30195057	1 269 015	2	>5	-0.8	*NF1,SUZ12*
Blast phase post-ET MF	HD-0535	46,XY,t(3;12)(q26-27;q13-14)[20]	5	G	2p25.3-2p14	chr2:30341-66555302	66 524 961	1	>100	+0.4	
				D	2p13.2-2p13.1	chr2:73007094-73763847	756 753	2	9	-0.4	*FBX041*
				D	3q29	chr3:193435136-194860954	1 425 818	2	8	-0.9	*HES1*
				D	6p25.3-6p21.32	chr6:152634-32624845	32 472 211	1	>50	-0.6	*PBX2, DEK*
				D	17q11.1-17q21.33	chr17:25291809-48544433	23 252 624	1	>10	-0.9	*NF1, SUZ12*
Blast phase post-ET MF	HD-0856	46,XX,del(7)(q22q36),del(8)(q22q24)[18]/47,idem,+8[2]	3	G	3q26.2-3q29	chr3:168672810-197895228	29 222 418	1	>50	+0.5	*EVI1*
				D	7q21.2-7q36	chr7:92389957-159118566	66 728 609	1	>50	-0.8	*EZH2, CAV1, MLL3*
				D	8q23.3-q24.22	chr8:113604482-137231948	23 627 466	1	>50	-0.8	*RAD21, MYC*
Blast phase post-ET MF	HD-1167	49,XX,add(3)(p14),del(5)(q31q35),+8,+8,+9,?del(11)(p11p13),-15,add(16)(p12),+mar[20]	13	G	8p23.3-8q24.3	chr8:73810-146294098	146 220 288	1	>200	+1	
				G	9p24.3-9q34.3	chr9:163131-141122114	140 958 983	1	>200	+0.5	*JAK2*
				G	13q31.1-13q34	chr13:83076377-115064121	31 987 744	1	>50	+0.5	
				G	17q23.3-q24.2	chr17:62111720-65955949	3 844 229	2	>20	+0.5	
				D	3p22.3-3p11	chr3:32434406-90309067	57 874 661	1	>30	-1	
				G	4p16.3-4p11	chr4:1589458-49581867	47992409	1	>100	+0.5	
				D	5q11.2	chr5:55877912-56107654	229 742	2	8	-1	
				D	5q11.2	chr5:56201487-57921887	1 720 400	2	5	-1	
				D	5q12.3-5q13.2	chr5:66508627-70587018	4 078 391	1	>10	-1	
				D	5q23.3-5q35.3	chr5:130551687-180712263	50 160 576	1	>100	-1	
				D	11p15.5-11p11.2	chr11:192372-47869001	47 676 629	1	>100	-1	
				D	15q21.3	chr15:56395780-57217890	822 110	2	6	-0.8	*TCF12*
				D	18p11.21	chr18:11649170-12999918	1 350 748	2	>10	-0.8	*PTPN2*
Blast phase post-ET MF	HD-1387	ND	6	G	1q21.1-1q44	chr1:143532171-249224147	105 691 976	1	>100	-0,5	
				G	3q21.3-3q29	chr3:128592775-197895228	69 302 453	1	>100	+0,2	*GATA2, SHOX2*
				D	7q11.21-7q36.3	chr7:61831840-159118566	97 286 726	1	>100	-0,2	*EZH2, CAV1, MLL3*
				G	9p24.3-9p11.2	chr9:163131-47212417	47 049 286	1	>100	+0,5	*JAK2, MLLT3*
				G	9q12-9q34.3	chr9:65632517-141122114	75 489 597	1	>100	+0.2	
				D	11q14.2-11q23.3	chr11:85800206-117441022	31 640 816	1	>50	-0,8	*EED, ARGHEF12, ETS1*
Blast phase post-ET MF	HD-1741_1309	46,XX,-3,der(5)t(3;5)(q13;q21),del(7)(q21q36),t(9;17)(q11;q11),+21[11]	10	D	3p25.2-3p26.3	chr3:64066-11023254	10 959 188	1	>20	-0.8	*RAD18, EDEM1*
				D	3p26.1-3p25.3	chr3:7300772-11023254	37 22 482	1	>30	-0.8	
				D	3p22.1-3p25.1	chr5:79057767-180712263	101 654 496	1	>30	-0.8	*TGFBR2, FBXL2*
				D	3p12.1-3p21.2	chr3:50719155-88428270	37 709 115	1	>100	-0.8	*DOCK3, FHIT*
				D	5q14.1-3q35.2	chr5:79057767-180712263	101 654 496	1	>200	-0.8	
				D	7q21.11-7q35	chr7:80855869-149015317	68 159 448	1	>200	-0.8	
				D	9p12-9p13.3	chr9:32955146-39254388	6 299 242	1	>50	-0.8	*SNOR12B, PAX5*
				D	12p12.3-12p13.2	chr12:10336209-13445554	3 109 345	2	>20	-0.8	*ETV6, CDKN1B, p27*
				D	17p12-17p13.1	chr17:3792543-12133847	8 341 304	1	>50	-0.8	
				G	21	chr21:10874759-48090317	37 215 558	1	>200	+0.5	*RUNX1*
Blast phase PMF	HD-1611_0528_1300	46,XY,del(20)(q11q13)[12]/46,XY,inv(3)(q21q26)[4]/46,idem,del(6)(q2?1q2?6)[3]/46,XY[1]	2	D	6q12-6q24.1	chr6:66438112-115733789	49 295 677	1	>200	-0.2	*TNFAIP3*
				D	20q11.22-20q13.12	chr20:33894460-47723303	13 828 843	1	>100	-0.2	*CDR1, CDR2, TOP1, PTPRT, SRSF6, L3MBTL1*
Blast phase PMF	HD-1847	ND	2	D	12p13.2	chr12:11743077-14796474	3 053 397	2	>10	-0.8	*ETV6, CDKN1B*
				D	13q14.11-13q14.3	chr13:41303677-53419331	12 115 654	1	>20	-0.5	*RB1*
Blast phase PMF	HD-1853_0927_1265_1461	46, XY, +8[16]/46,XY[6]	1	G	8	chr8:73810-146294098	146 220 288	1	>250	+0.2	
Blast phase post-MPN MF	HD-1137	44,XY,add(1)(q4?2),add(3)(p14),del(5)(q23q34),der(9)t(9;10)(q23;q22),-10,?dic(15;20)(q12;p12),add(17)(p11)[19]/46,XY,del(12)(p11p13)[1]	16	D	3p22.1-3p14.2	chr3:41897844-62216929	20 319 085	1	>50	-0.4	*FHIT*
				D	5q15-5q21.3	chr5:96081976-106522034	10 440 058	1	>10	-0.4	
				D	5q22.2-5q23.2	chr5:111541767-123042265	11 500 498	1	>10	-0.4	
				D	5q23.2-5q32	chr5:125057622-147179938	22 122 316	1	>50	-0.4	
				D	5q34-5q35.3	chr5:167579682-180712263	13 132 581	1	>50	-0.4	
				D	9q33.3-9q34.3	chr9:127413137-140513193	13 100 056	1	>20	-0.4	
				D	10q22.3-10q23.33	chr10:77474006-94633158	17 159 152	1	>20	-0.4	
				D	10q23.33-10q24.32	chr10:95591726-103584723	7 992 997	1	>20	-0.4	
				D	10q24.32-10q25	chr10:104190680-109625441	5 434 761	1	>10	-0.4	
				D	12p11.23-12p13.31	chr12:10357913-27227777	16 869 864	1	>20	-0.4	*ETV6, CDKN1B, p27*
				D	15q13.3-15q21.1	chr15:32928004-47656834	14 728 830	1	>50	-0.4	
				D	17p13.3-17p11.2	chr17:29169-18562710	18 533 541	1	>50	-0.4	
				D	20p11.23-20p12.1	chr20:11982565-18353006	6 370 441	1	>20	-0.4	*MACROD2*
				D	20p13-20p12.3	chr20:60747-6769229	6 708 482	1	>20	-0.4	
				D	20p12.2-20p11.23	chr20:11982565-18353006	6 370 441	1	>20	-0.4	
				G	21q22.13-21q22.3	chr21:39096247-48090317	8 994 070	1	>50	+0.2	

bp, base pairs; CDR, commonly deleted region; Chr, chromosome; CNA, copy number aberrations; D, deletion; G, gain; MF, myelofibrosis; nb, number; ND, not determined; MPN, myeloproliferative neoplasm; PMF, primary myelofibrosis; post-ET MF, post-essential thrombocythemia MF; post-PV MF, post-polycythemia vera MF.

Supplemental Table S9. Gene mutations of 17 blast phase MF cases with TET2, IDH1/2, DNMT3A, ASXL1, EZH2, SUZ12, BMI1, RCOR1, JAK2, MPL, CBL, SOCS2, LNK, NF1, KRAS, NRAS, PTPN11, PPP1R16B, TRPS1, SF3B1, SRSF2 and TP53 mutations.

Diagnosis	Samples	*TET2* (E3-11)	*IDH1/2* (E4)	*DNMT3A* (E15-23)	*ASXL1* (E12)	*EZH2* (E2-20)	*SUZ12* (E10-16)	*BMI1* (E1-10)	*RCOR1* (3-12)	*JAK2* (E12;14)	*MPL* (E10)	*CBL* (E8-9)	*SOCS2* (E1-2)	*LNK* (E2-8)	*NF1* (E1-58)	*KRAS* (E2-4)	*NRAS* (E1-2)	*PTPN11* (E3;13)	*PPP1R16B* (2-11)	*TRPS1* (E2-7)	*SF3B1* (E15-16)	*SRSF2* (E2)	*TP53* (E5-8)
BP post-PV MF	HD-0492	wt	wt	wt	wt	ND	wt	wt	wt	**80-100**	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt	wt
BP post-PV MF	HD-1517	ND	ND	ND	**c.1934dupG;p.G646WfsX12**	**c.2077 A>T p.N692Y**	ND	ND	ND	ND	wt	ND	ND	ND	ND	ND	ND	wt	wt	wt	ND	wt	wt
BP post-PV MF	HD-1649_1559	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	wt	ND
BP post-PV MF	HD-1836_1813	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
BP post-ET MF	HD-0535	wt	wt	wt	**c.2285dupT; p.L762FfsX12**	ND	wt	wt	wt	wt	wt	wt	wt	ND	wt	ND	ND	ND	ND	ND	wt	ND	**c.868C>T p.R290C**
BP post-ET MF	HD-0536	wt	wt	wt	**c.1934dupG; p.G646WfsX12**	**c.1712 G>A p.C571Y**	wt	wt	ND	wt	wt	wt	wt	ND	ND	ND	ND	wt	ND	ND	wt	wt	wt
BP post-ET MF	HD-0646	wt	wt	wt	wt	wt	wt	wt	ND	**5-10**	wt	wt	wt	ND	ND	ND	ND	wt	wt	wt	c.**2098 A>G; p. K700E**	wt	wt
BP post-ET MF	HD-0856	wt	wt	wt	wt	wt	wt	wt	ND	wt	wt	wt	wt	ND	ND	ND	ND	wt	wt	wt	wt	wt	wt
BP post-ET MF	HD-1167	wt	wt	wt	wt	wt	wt	ND	wt	wt	wt	wt	ND	wt	wt	wt	wt	wt	wt	wt	wt	wt	**c.722C>T p.S241F**
BP post-ET MF	HD-1387	ND	ND	ND	wt	wt	ND	ND	ND	ND	ND	ND	ND	**c.494 C>G p.T165S** **c.544 T>C p.F182L**	ND	ND	ND	wt	**c.1606 A>C p.K473Q**	wt	ND	wt	wt
BP post-ET MF	HD-1741_1309	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	wt	wt
BP-PMF	HD-0655	wt	wt	wt	wt	wt	wt	wt	ND	**30-50**	wt	wt	wt	ND	ND	ND	ND	wt	wt	wt	wt	ND	wt
BP-PMF	HD-1611_0528_1300	**c.4062delA; p.A1355HfsX8^{NC}** **c.1648 C>T p.R550X**	wt	wt	wt	wt	wt	ND	ND	ND	wt	wt	ND	wt	ND	wt	wt	ND	wt	wt	wt	**c.284 C>A p.P95H**	wt
BP-PMF	HD-0635	wt	wt	wt	wt	ND	wt	ND	ND	**30-50**	wt	wt	ND	ND	ND	ND	ND	ND	ND	ND	wt	wt	wt
BP-PMF	HD-1847	ND	ND	ND	ND	ND	ND	ND	ND	mutated	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
BP-PMF	HD-11853_0927_1265_1461	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND	ND
BP post-MPN MF	HD-1137	wt	wt	**c.2644G>A;p.A882S**	wt	wt	wt	ND	wt	wt	wt	wt	ND	wt	wt	wt	wt	wt	ND	ND	wt	wt	**intron 5+1G>A**

References for nucleotide and protein sequences
TET2, NM_001127208.1, NP_001120680.1; IDH1, NM_005896.2, NP_005887.2; IDH2, NM_002168.2, NP_002159.2; DNMT3A, NM_022552.3, NP_072046.2; ASXL1, NM_015338.5, NP_056153.2; EZH2, NM_004456.4, NP_004447.2; SUZ12, NM_015355.2, NP_056170.2; BMI1, NM_005180.8, NP_005171.4;
RCOR1, NM_015156.3, NP_055971.2; JAK2, NM_004972.3, NP_004963.1; MPL, NM_005373.2, NP_005364.1; CBL, NM_005188.2, NP_005179.2; SOCS2, NM_001270470.1, NP_001257399.1; NF1, NM_001042492.2, NP_001035957.1; LNK, NM_005475.2, NP_005466.1; KRAS, NM_004985.3, NP_004976.2 and NM_033360.2, NP_203524.1; NRAS, NM_002524.4, NP_002515.1; PTPN11, NM_002834.3, NP_002825.3; PPP1R16B, NM_015568.2, NP_056383.1; TRPS1, NM_014112.2, NP_054831.2; SF3B1, NM_012433.2, NP_036565.2; SRSF2, NM_003016.4, NP_003007.2 TP53, NM_000546.5, NP_000537.3.

BP, Blast phase; MF, myelofibrosis; NC, not constitutional mutation; ND, not determined; PMF, primary myelofibrosis; Post-ET MF, post-essential thrombocythemia MF; post-PV MF, post-polycythemia vera MF; wt, wild-type.

Supplemental Table S10. Prognostic impact of CNAs and gene mutations (>5%) on Time To Acute Transformation (TTAT) and Overall Survival (OS)

Univariate (log-rang tests) and multivariate (Cox models) analyses

		TTAT-UNIVARIATE				**OS-UNIVARIATE**				**OS-MULTIVARIATE**		
	Alterations	**n at risk**	**5 years TTAT %**	**95% CI**	**p**	**n at risk**	**5 years OS %**	**95% CI**	**p**	**HR**	**95% CI**	**p**
CNAs	no	3	95	0.86-1.00	0.58	11	64	0.45-0.9	0.89			
	yes	5	80	0.64-1.00		12	63	0.45-0.86				
del(20q), del(17q), del(12p)	no	1	96	0.88-1.00	**0.03**	13	72	0.56-0.9	0.72			
	yes	6	75	0.57-1.00		9	48	0.26-1.00				
Number of mutations	0-1 mutation	2	100	0.79-1.00	**0.04**	7	80	0.65-0.75	**0.001**			
	> 1 mutation	5	72	0.52-1.00		16	44	0.3-0.75				
Epigenetic-associated genes mutated	no	2	100	0.75-1.00	0.10	8	72	0.55-0.97	**0.03**			
	yes	5	74	0.55-1.00		13	58	0.45-0.88				
ASXL1 mutated	no	4	92	0.81-1.00	0.21	13	71	0.57-0.9	**0.02**	2.4	0.94-6.27	0.07
	yes	3	77	0.54-1.00		9	48	0.33-0.96				
EZH2 mutated	no	6	91	0.81-1.00	**0.01**	19	69	0.59-0.88	**0.003**	6.8	1.11-413.0	**0.04**
	yes	1	0	0.13-1.00		2	0	NA				
TET2 mutated	no	6	92	0.83-1.00	0.76	18	59	0.47-0.82	0.59			
	yes	1	80	0.52-1.00		3	86	0.63-1.00				
DNMT3A mutated	no	6	90	0.8-1.00	0.21	20	66	0.52-0.84	0.17			
	yes	1	50	0.13-1.00		2	33	0.07-1.00				
Splicing-associated gene mutated	no	4	95	0.87-1.00	0.07	16	69	0.54-0.87	**0.004**			
	yes	2	0	0.43-1.00		5	21	0.5-1				
SF3B1 mutated	no	6	87	0.76-1.00	0.65	19	66	0.56-0.86	0.17			
	yes	0	NA	NA		2	0	NA				
SRSF2 mutated	no	6	94	0.88-1.00	**0.0002**	21	65	0.50-0.84	**0.01**	6.2	1.66-23.00	**0.007**
	yes	2	0	0.13-1.00		3	0	NA				
Signaling-associated gene mutated	no	0	NA	NA	0.20	1	NA	NA	0.08			
	yes	6	84	0.69-1.00		19	58	0.47-0.79				
JAK2 mutated	no	2	94	0.84-1.00	0.54	NA	NA	NA	NA			
	yes	6	83	0.69-1.00		NA	NA	NA	NA			
LNK mutated	no	7	88	0.77-1.00	0.67	NA	NA	NA	NA			
	yes	0	NA	NA		NA	NA	NA	NA			
NF1 mutated	no	3	86	0.7-1.00	0.21	NA	NA	NA	NA			
	yes	1	0	NA		NA	NA	NA	NA			

Supplemental Figures

Supplemental Figure 1. Examples of aCGH profiles. A: aCGH profile of chromosome 1 in case HD-1427 showing a deletion including *CSMD2*. **B**: aCGH profile of chromosome 8 in case HD-1362 showing a deletion including *TRPS1.* **C**: aCGH profile of chromosome 14 in case HD-0728 showing a deletion including *RCOR1.* **D**: aCGH profile of chromosome 18 in case HD-0777 showing a gain including *SALL3*.

Supplemental Figure 2. Deduced localization of mutations in our myelofibrosis series (n=68). ASXL1, ASXH: additional sex combs homology domain; PHD: plant homeodomain; NR box: nuclear receptor box. TET2, BOX1 and BOX2: conserved regions 1134-1444, 1842-1921. EZH2, SANT: putative DNA binding domain in the SWI-SNF and ADA complexes, the transcriptional co-repressor N-CoR and TFIIIB; C-rich: cysteine-rich domain; SET domain: protein lysine methyltransferase enzymes. DNMT3A, PHD: plant homeodomain; PRC2: polycomb repressive complex 2; PWWP: proline tryptophan tryptophan proline. CBL, TKB: tyrosine kinase binding; RF: ring finger; PPP: proline rich region; LZ: leucine zipper; UBA: ubiquitin associated. NF1, Sec: sec14-like; PH: pleckstrin homology. LNK, Pro/DD: proline-rich dimerization domain; PH: pleckstrin homology; SH2: src homology 2. PTPN11, N-SH2 and C-SH2: amino SRC homology 2; PTP: protein tyrosine phosphatase. SRSF2, RRM: RNA recognition motif; RS: arginine/serine domain. SF3B1, HD: heat domains. Triangles represent nonsense mutations, circles frameshift mutations and squares missense mutations.

Supplemental Figure 3. Histogram comparing alterations found in patients studied at diagnosis and during disease course using aCGH (n=63) and sequencing (n=68). CNA: copy number aberration.

Supplemental Figure 4. Examples of aCGH profiles in blast phase MF. A: aCGH profile of chromosome 12 in 7 samples (HD-1559_1649, HD-1649_1559, HD-1137, HD-1741_1309, HD-1813_1836, HD-1813_1836 and HD-1847) showing recurrent deletions in 12p13 including *ETV6, CDKN1B/p27*. **B**: aCGH profile of chromosome 15 in HD-1167 showing a deletion including *TCF12*. **C**: aCGH profile of chromosome 18 in HD-1167 showing a deletion including *PTPN2*. **D**: aCGH profile of chromosome 21 in HD-1517 showing a deletion including *RUNX1*.

Supplemental Figure 5. Schematic representation of ways to primary or secondary myelofibrosis from a JAK2 mutated or non-JAK2 clone as deduced from the mutation analysis.

Supplemental Figure 6. Ideogram of the long arm of chromosome 20 comparing deleted regions in myeloid malignancies. Commonly-deleted regions (CDRs) determined in several studies including the present one are shown by

														therapy	
HD-0725_1380	M/74	post-ET MF	PB	NA	NA	NA	11	46,XY[20]	35.4	12.6	37.6	315	no	yes	satisfactory	40	HU	X
HD-0983	M/32	post-ET MF	PB	NA	NA	NA	<1	ND	15.2	12.2	36.8	419	no	yes	satisfactory	wt	X	X
HD-1264	M/72	post-ET MF	BM	NA	NA	NA	<1	45,XY,der(7;18)(q10;q10)[20]	4	12	35	150	no	yes	satisfactory	wt	B, antiplatelet therapy	X
HD-1309_1741	F/36	post-ET MF	PB	NA	NA	NA	73	46,XX[20]	12.2	9.9	ND	750	no	yes	weight loss	wt	HU, A, P, T	X
HD-1360_1605	M/67	post-ET MF	PB	NA	NA	NA	12	ND	5.8	11	32.8	392	no	yes	no functional symptoms	30-50	HU, X, antiplatelet therapy	X
HD-1376	M/76	post-ET MF	PB	NA	NA	NA	9	46,XY[20]	27.2	7.3	21.9	49	no	yes	satisfactory	50-70	HU, antiplatelet therapy	X
HD-1462	F/63	post-ET MF	PB	NA	NA	NA	<1	46,XX[20]	13.2	11.5	ND	351	no	no	none	75	antiplatelet therapy	no
HD-1569	F/54	post-ET MF	PB	NA	NA	NA	47	46,XX[20]	31.8	12.5	37.7	317	no	yes	asthenia and fatigue	50-60	C, PA	no
HD-1616_0551	M/71	post-ET MF	PB	NA	NA	NA	<1	47,XY,+9[5]/46,XY[15]	11.4	11.4	34.2	376	no	no	ND	50-70	C, EPO	X
HD-0497_1307	F/82	PMF	PB	inter-2	inter-2	inter-2	160	46,XX,del(20)(q11q13)[6]/46,XX[3]	2.4	7.6	21.2	205	yes	no	satisfactory	20-30	T	X
HD-0586	M/62	PMF	PB	inter-2	inter-2	high	31	45,XY,-13[18]/46,XY[2]	3.2	7.8	23.8	83	yes	yes	satisfactory	wt	HU, T	X
HD-0607_1130	M/51	PMF	PB	low	inter-1	inter-1	70	46,XY[20]	7.6	13.3	39	471	no	yes	satisfactory	wt	HU, antiplatelet therapy	X
HD-0616	M/54	PMF	PB	low	low	low	11	46,XY[4]	14.3	17.8	52	257	no	no	satisfactory	35-40	none	X
HD-0648	F/60	PMF	PB	ND	inter-1	inter-1	129	46,XX[20]	20.3	11.5	35	112	no	yes	satisfactory	80-90	HU, EPO	X
HD-0679_1291	M/31	PMF	PB	low	low	low	<1	46,XY[20]	5.3	15.3	44.9	208	no	no	satisfactory	5-10	none	X
HD-0683	M/62	PMF	PB	inter-2	inter-2	inter-2	<1	ND	1.9	8.5	24.8	101	yes	yes	ND	10	none	X
HD-0689	F/74	PMF	PB	ND	inter-2	inter-2	298	ND	6	7.6	22.2	167	yes	yes	asthenia aND fatigue	80-90	HU, T	X
HD-0717_1244	M/65	PMF	PB	inter-2	inter-1	inter-1	70	46,XY[20]	9.3	14.5	44.3	191	no	yes	satisfactory	wt	none	X
HD-0719	M/85	PMF	PB	inter-1	inter-1	inter-1	<1	46,XY[20]	120	8.6	26	479	no	yes	fever	wt	none	X
HD-0728	F/54	PMF	PB	inter-2	low	low	<1	46,XX,t(6;17)(p21-22;q11)[10]/46,XX[13]	35.1	13.2	ND	691	no	no	satisfactory	70-80	none	X
HD-0759	F/63	PMF	PB	inter-2	inter-2	high	3	46,XX,del(5)(q13q33),del(20)(q11q13)[19]/46,XX[1]	4.1	8	23	77	yes	yes	asthenia and fatigue	50-75	EPO	X
HD-0777	M/86	PMF	PB	high	inter-2	inter-2	<1	ND	54.2	7.7	25.2	761	yes	no	asthenia and fatigue	wt	A, antiplatelet therapy	X
HD-0862	M/83	PMF	PB	high	high	high	4	46,XY[20]	4.6	9.7	29.7	302	yes	yes	weight loss	12-30	EPO	X
HD-1047	M/65	PMF	PB	inter-2	inter-2	inter-2	<1	46,XY,del(20)(q11q13)[3]/46,idem,add(9)(p2?3)[17]	26.8	13.6	40.8	467	no	yes	satisfactory	50-75	none	X
HD-1095	M/64	PMF	PB	ND	ND	ND	<1	ND	ND	ND	ND	ND	ND	ND	ND	50-70	ND	X
HD-1098	M/75	PMF	PB	ND	ND	ND	<1	ND	9.3	13.4	40.4	143	ND	ND	ND	wt	ND	X
HD-1100	M/73	PMF	PB	high	high	high	80	46,XY,del(13)(q13q22-31)[6]/46,sl,?del(17)(p12-13)[5]/44,sl,der(1)inv(1)(p32~34q23)?dup(1)(q31q32),-7,-16,?der(17)t(16;17)(p11-12;p12-13)[6]/44,sdl1,?del(17)(p12-13)[5] +der(17)t(16;17)(p11-12;p12-13)[6]/44,sdl1,?del(17)(p12-13)[5]	11.7	10.6	32.2	645	yes	yes	no functioNAl symptoms	31-50	HU, antiplatelet therapy	X
HD-1111	F/84	PMF	PB	inter-2	inter-1	inter-1	101	46,XX[20]	20.7	15	14.3	361	no	yes	asthenia and fatigue	80-90	HU, P, T, C	no
HD-1123	F/80	PMF	PB	inter-2	inter-2	inter-2	<1	46,XX[20]	27.3	10.2	32	1188	no	no	asthenia and fatigue	50-70	none	X
HD-1138_0540_1398	M/61	PMF	PB	inter-2	inter-1	inter-1	82	ND	27	12.0	36.2	378	no	yes	satisfactory	wt	none	X

HD-1207	M/64	PMF	PB	inter-1	inter-2	high	28	46,XY,ider(20)(q10)del(20)(q11q13)[20]	2.6	7.2	20.2	5	yes	yes	asthenia and fatigue	wt	T	X
HD-1208	M/74	PMF	PB	inter-2	high	high	3	48,XY,+8,+19[19]/46,XY[1]	8.6	8.7	25.6	101	yes	yes	fever	2	C	X
HD-1227	F/70	PMF	PB	low	inter-1	inter-2	82	48, XX +8,+9[7]/46,XX[13]	5.6	14.2	43.7	60	no	yes	asthenia and fatigue	2-5	EPO	X
HD-1265_0927_1461_1853	M/72	PMF	PB	high	inter-1	inter-1	13	ND	7.1	11.6	34.8	147	no	no	satisfactory	30-50	HU	X
HD-1283	M/75	PMF	PB	inter-2	inter-2	inter-2	84	ND	5	9.7	25	890	yes	no	satisfactory	wt	C, EPO, T, HU	X
HD-1289	M/75	PMF	PB	inter-1	inter-2	high	79	ND	11.4	12.5	37.1	88	no	yes	night sweats	mutated	HU, antiplatelet therapy	X
HD-1300_0528_1611	M/59	PMF	PB	inter-2	inter-2	inter-2	47	46, XY, del 20(q11q13)[4]/46,XY[16]	41.1	8.8	27.3	98	no	ND	satisfactory	80-90	graft therapy	X
HD-1362	F/67	PMF	PB	ND	inter-2	inter-2	195	46,XX,dup(1)(q44q12),del(13)(q13q14)[20]	1.3	9.8	29.7	123	yes	yes	satisfactory	80-90	E	X
HD-1412_1495	M/52	PMF	PB	ND	inter-1	inter-1	33	46,XY[2]	12	13.2	41.7	77	no	yes	satisfactory	wt	EPO, HU	X
HD-1422	F/58	PMF	PB	low	inter-1	inter-1	79	ND	12.8	12.2	ND	351	no	yes	satisfactory	wt	EPO	X
HD-1439	M/71	PMF	PB	low	inter-2	inter-2	89	46,XY,del(7)(q22q36)[2]/46,XY[21]	48.8	14.2	47	271	no	yes	satisfactory	50-75	HU	X
HD-1524bis	F/70	PMF	PB	inter-2	high	high	18	ND	76	5.8	18	26	yes	yes	weight loss	wt	HU, C	X
HD-1538	F/77	PMF	PB	high	inter-2	inter-2	<1	ND	3.4	9.7	28.8	366	no	yes	satisfactory	30-40	none	X
HD-1572bis	F/66	PMF	PB	inter-2	inter-1	inter-1	6	46,XX,t(12;17)(q12;q21)[15]/46,XX[5]	7.4	11.6	ND	228	no	yes	satisfactory	wt	HU	X
HD-1587	F/60	PMF	PB	inter-2	inter-2	inter-2	87	ND	3.5	9.2	ND	266	yes	yes	asthenia and fatigue	wt	C, EPO	X
HD-1606	F/40	PMF	BM	low	low	low	108	46,XX[20]	6.5	13.6	41.1	274	no	no	satisfactory	12-30	HU, antiplatelet therapy	X
HD-1617	F/82	PMF	PB	inter-1	inter-1	inter-2	18	46,XX[20]	6.5	12	35.8	36	yes	yes	satisfactory	wt	C	X
HD-1654	M/65	PMF	PB	low	inter-2	high	54	46,XY,inv(11)(q12q22)[c20]	4.2	7.6	22.2	38	yes	yes	asthenia and fatigue	wt	T	X

B. Clinical and biological characteristics of 19 matched pairs of MF

Samples	Sex/ Age	Diagnosis	PB/ BM	IPSS	DIPSS	DIPSSplus	Time from diagnosis to sampling (m)	Karyotype	Leukocyte count (x 10^9/L)	Hemoglobin level (g/dL)	Hematocrit (%)	Platelet count (x 10^9/L)	RCT need	Ci blast cells	Symptoms	JAK2 V617F %	Previous Therapies	aCGH
HD-0842_1401	M/64	PV	PB	NA	NA	NA	NA	ND	6.3	21.3	66.4	269	no	no	satisfactory	50-60	HU, antiplatelet therapy	X
HD-1401_0842	M/67	post-PV MF	PB	NA	NA	NA	<1	46,XY,del(8)(q21q23)[7]/46,XY[19]	31.1	13.4	41.2	714	no	yes	weight loss	50-60	HU, antiplatelet therapy	X

HD-0551_1616	M/66	ET	PB	NA	NA	NA	NA	ND	8.3	13.6	39.9	399	no	no	satisfactory	15-30	HU, antiplatelet therapy	X
HD-1616_0551	M/71	post-ET MF	PB	NA	NA	NA	<1	47,XY,+9[5]/46,XY[15]	11.4	11.4	34.2	373	no	no	ND	50-70	C, EPO	X
HD-1412_1495	M/52	PMF	PB	high	inter-1	inter-1	34	46,XY[2]	12	13.2	41.7	77	no	yes	satisfactory	wt	EPO, HU	X
HD-1495_1412	M/59	PMF	PB	inter-2	inter-2	NA	36	46,XY[2]	2.7	9.3	28.4	49	ND	yes	infectious disease	wt	EPO	X
HD-0725_1380	M/74	post-ET MF	PB	NA	NA	NA	11	46,XY[20]	19.2	12.6	37.6	321	no	yes	satisfactory	40	HU	X
HD-1380_0725	M/76	post-ET MF	PB	NA	NA	NA	40	46,XY[20]	35.4	12.6	37.6	315	no	yes	satisfactory	50-70	antiplatelet therapy	X
HD-0497_1307	F/82	PMF	PB	inter-2	inter-2	inter-2	160	46,XX,del(20)(q11q13)[6]/46,XX[3]	2.4	7.6	21.2	205	yes	no	satisfactory	20-30	T	X
HD-1307_0497	M/86	PMF	PB	NA	NA	NA	204	ND	3.2	11.8	34.7	64	yes	ND	asthenia and fatigue	ND	none	X
HD-0607_1130	M/51	PMF	PB	low	inter-1	inter-1	70	46,XY[20]	7.6	13.3	39	471	no	yes	satisfactory	wt	HU, antiplatelet therapy	X
HD-1130_0607	M/53	PMF	PB	low	inter-1	inter-1	102	46,XY[20]	9.5	14.5	42.7	474	no	yes	satisfactory	wt	HU, antiplatelet therapy	X
HD-0679_1291	M/31	PMF	PB	low	low	low	<1	46,XY[20]	5.3	15.3	44.9	208	no	no	satisfactory	5-10	none	X
HD-1291_0679	M/33	PMF	PB	low	low	low	26	ND	5	15.8	46.2	171	no	no	satisfactory	5-10	antiplatelet therapy	X
HD-0717_1244	M/65	PMF	PB	inter-2	inter-1	inter-1	71	46,XY[20]	9.3	14.5	44.3	191	no	yes	satisfactory	wt	none	X
HD-1244_0717	M/67	PMF	PB	inter-2	inter-1	inter-1	94	ND	6.5	13.3	40.9	115	no	yes	satisfactory	wt	none	X
HD-0589_1824	F/65	post-PV MF	PB	NA	NA	NA	55	48,XX,+8,+9[3]/46,XX,del(20)(q11q13)[2]/46,XX[15]	5.8	13.4	41.4	188	no	no	no functional symptoms	50-70	none	X
HD-1824_0589	F/70	post-PV MF	PB	NA	NA	NA	117	46,XX,del(20)(q11q13)[8]/48,XX,+8,+9[3]/48,idem, der(9)t(1;9)(q21;q12q)[2]/46,XX[7]	2.5	8.8	26.9	71	no	yes	satisfactory	50-70	none	X
HD-0789_1691	M/66	post-PV MF	PB	NA	NA	NA	<1	46,XY,del(20)(q11q13)[4]/46,XY[16]	20.3	13.7	45.7	360	ND	ND	ND	80-90	HU	X
HD-1691_0789	M/69	post-PV MF	BM	NA	NA	NA	36	45,XY,-7,del(20)(q11q13)[20]/46,XY[2]	13.3	13.7	ND	60	ND	ND	night sweats	ND	anti-JAK1/2	X
HD-1427_1656	F/71	PMF	PB	inter-2	inter-1	inter-1	45	46,XX,t(7;14)(q11;q24)[2]/46,XX[18]	16.6	15.7	51.9	363	no	no	weight loss	80-90	M	X
HD-1656_1427	F/72	PMF	PB	high	inter-2	inter-2	54	46,XX[21]	8.2	14	41.7	310	no	yes	night sweats	ND	antiplatelet therapy	X
HD-1360_1605	M/44	post-ET MF	PB	NA	NA	NA	12	ND	5.8	11	32.8	392	no	yes	no functional symptoms	30-50	X, antiplatelet therapy	X
HD-1605_1360	M/45	post-ET MF	BM	NA	NA	NA	20	ND	7.5	12	ND	547	no	ND	no functional symptoms	30-50	PA, antiplatelet therapy	X
HD-0614_1112_1352	M/76	post-ET MF	PB	NA	NA	NA	140	46,XY[22]	4.5	11.2	35	269	no	yes	satisfactory	wt	EPO, antiplatelet therapy	X
HD-1112_0614_1352	M/77	post-ET MF	PB	NA	NA	NA	170	ND	4.3	7.8	22.8	78	yes	yes	asthenia and fatigue	wt	EPO, antiplatelet	X

																	therapy	
HD-1352_0614_1112	M/78	post-ET MF	PB	NA	NA	NA	178	46,XY,del(7)(q22q36)[2]/46,XY,add(4)(q21-22),-12,-13,+der(?)t(?;4)(?;q21-22),+mar[3]/44,XY,add(4),der(6)t(4;6)(q21-22;p2?3),-7,-12,-15,-16,+2mar[3]	3.5	ND	23.4	159	yes	yes	asthenia aND fatigue	wt	EPO, antiplatelet therapy	X
HD-0540_1138_1398	M/58	PMF	PB	inter-1	ND	ND	46	46,XY[20]	11.5	12.3	ND	334	no	ND	satisfactory	wt	none	X
HD-1138_0540_1398	M/61	PMF	PB	inter-2	inter-1	inter-1	82	ND	27	12.0	36.2	378	no	yes	satisfactory	wt	none	X
HD-1398_0540_1138	M/62	PMF	PB	inter-2	inter-1	inter-1	91	ND	27.3	12.6	38	315	no	yes	satisfactory	wt	none	X
HD-0927_1265_1461_1853	M/72	PMF	PB	high	high	inter-2	<1	ND	42	11.7	36	658	no	yes	weight loss	40-50	none	X
HD-1265_0927_1461_1853	M/72	PMF	PB	high	inter-1	inter-1	13	ND	7.1	11.6	34.8	147	no	no	satisfactory	30-50	HU	X
HD-1461_0927_1265_1853	M/73	PMF	PB	high	inter-1	inter-1	20	ND	9.7	10.8	ND	328	no	no	satisfactory	30-50	HU	X
HD-1853_0927_1265_1461	M/75	BP-PMF	PB	NA	NA	NA	1	46,XY, +8[16]/46,XY[6]	5.1	10.2	32.6	168	yes	yes	asthenia and fatigue	ND	HU	X
HD-1559_1649	M/63	post-PV MF	BM	NA	NA	NA	<1	45,XY,-7[9]/47,XY,+9[3]/46,XY[8]	2.2	8.2	ND	25	ND	no	satisfactory	30-50	antiplatelet therapy	X
HD-1649_1559	M/63	BP post-PV MF	PB	NA	NA	NA	4	45,XY,-7[20]	73	10	ND	27	yes	yes	asthenia and fatigue	ND	C, T	X
HD-1813_1836	M/77	post-PV MF	PB	NA	NA	NA	<1	45,XY,-7[6]/45,sl,del(12)(p12p13)[3]/ 45,sdl1,add(3)(q2?1)[13]	4.6	11.5	34.7	58	yes	yes	ND	2-5	none	X
HD-1836_1813	M/77	BP post-PV MF	PB	NA	NA	NA	1	45,XY,-7[6]/45,sl,del(12)(p12p13)[3]/ 45,sdl1,add(3)(q2?1)[13]	1.5	8.4	24.2	13	yes	yes	ND	2-5	none	X
HD-0528_1300_1611	M/56	PMF	PB	inter-2	inter-1	inter-1	<1	46,XY[20]	29.4	13.2	38.6	295	no	yes	satisfactory	75	none	X
HD-1300_0528_1611	M/59	PMF	PB	inter-2	inter-2	inter-2	47	46, XY, del 20(q11q13)[4]/46,XY[16]	41.1	8.8	27.3	98	no	ND	satisfactory	80-90	graft therapy	X
HD-1611_0528_1300	M/60	BP-PMF	PB	NA	NA	NA	59	46,XY,del(20)(q11q13)[12]/ 46,XY,inv(3)(q21q26)[4]/ 46,idem,del(6)(q2?1q2?6)[3]/46,XY[1]	6.5	12	35.8	36	no	yes	asthenia aND fatigue	ND	M	X
HD-1309_1741	F/36	post-ET MF	PB	NA	NA	NA	73	46,XX[20]	16.2	9.9	ND	750	ND	yes	asthenia and fatigue	wt	none	X
HD-1741_1309	F/37	BP post-ET MF	PB	NA	NA	NA	87	46,XX,-3,der(5)t(3;5)(q13;q21),del(7)(q21q36),t(9;17)(q11;q11),+21[20]	77.3	9.6	ND	469	no	yes	asthenia and fatigue	wt	HU	X

C. Clinical and biological characteristics of 17 blast phase MF

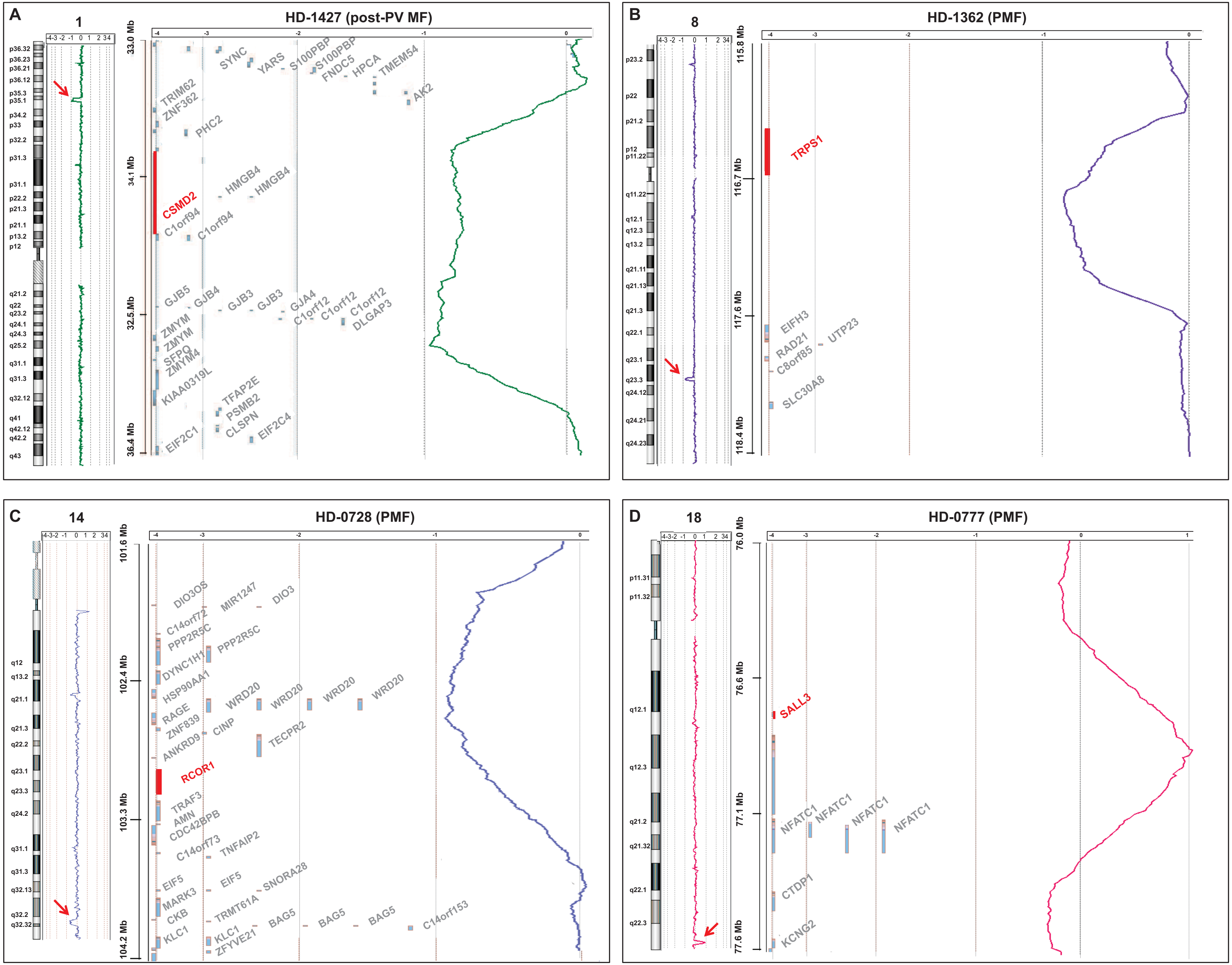
Samples	Sex/ Age	Diagnosis	PB / BM	Time from diagnosis to sampling (m)	Karyotype	Leukocyte count (x 10^9/L)	Hemoglobin level (g/dL)	Hematocrit (%)	Platelet count (x 10^9/L)	JAK2 V617F %	Previous Therapies	aCGH
HD-0492	M/67	BP post-PV MF	PB	20	47,XY,+8[17]/48,idem,+9[3]	17.0	10.0	36.0	44	80-100	HU	X
HD-1517	M/89	BP post-PV MF	BM	ND	46,XY[20]	15.1	10.4	30.5	94	wt	ND	X
HD-1649_1559	M/63	BP post-PV MF	PB	4	45,XY,-7[20]	73	10	ND	27	ND	C, T	X
HD-1836_1813	M/77	BP post-PV MF	PB	<1	ND	1.5	8.4	24.2	13	2-5	none	X
HD-0535	M/68	BP post-ET MF	PB	153	46,XY,t(3;12)(q26-27;q13-14)[20]	10.3	11.5	ND	661	wt	HU	X
HD-0536	M/53	BP post-ET MF	PB	<1	46,XY,add(15)(q25-26)[5]/46XY[19]	5.0	13.1	39.2	535	wt	none	X
HD-0646	F/61	BP post-ET MF	PB	15	46,XX,inv(3)(q21q26)[3]/46,idem,der(16)t(1;16)(q11;q11)[17]	5.2	1.7	33.0	315	5-10	HU	X
HD-0856	F/57	BP post-ET MF	PB	15	46,XX,del(7)(q22q36),del(8)(q22q24)[18]/47,idem,+8[2]	7.7	9.2	29.0	1052	wt	HU	X
HD-1167	F/68	BP post-ET MF	PB	12	49,XX,add(3)(p14),del(5)(q31q35),+8,+8,+9,?del(11)(p11p13),-15,add(16)(p12),+mar[20]	15	8	23.7	117	wt	HU, A	X
HD-1387	M/60	BP post-ET MF	PB	15	ND	ND	ND	ND	ND	ND	ND	X
HD-1741_1309	F/37	BP post-ET MF	PB	87	46,XX,-3,der(5)t(3;5)(q13;q21),del(7)(q21q36),t(9;17)(q11;q11),+21[20]	77.3	9.6	ND	469	wt	HU	X
HD-0655	M/76	BP-PMF	PB	26	46,XY,inv(16)(p13q22)[20]	6.6	8.1	24.0	25	30-50	none	X
HD-1611_0528_1300	M/60	BP-PMF	PB	59	46,XY,del(20)(q11q13)[12]/46,XY,inv(3)(q21q26)[4]/46,idem,del(6)(q2?1q2?6)[3]/46,XY[1]	6.5	12	35.8	36	ND	M	X
HD-0635	M/76	BP-PMF	PB	26	46,XY,inv(16)(p13q22)[20]	6.6	8.1	24.0	25	30-50	none	X
HD-1847	M/79	BP-PMF	PB	<1	ND	24.4	11.6	36.2	164	mutated	HU	X
HD-1853_0927_1265_1461	M/75	BP-PMF	PB	<1	46,XY, +8[16]/46,XY[6]	5.1	10.2	32.6	168	ND	HU	X
HD-1137	M/82	BP post-MPN MF	PB	3	44,XY,add(1)(q4?2),add(3)(p14),del(5)(q23q34),der(9)t(9;10)(q23;q22),-10,?dic(15;20)(q12;p12),add(17)(p11)[19]/46,XY,del(12)(p11p13)[1]	106	9.4	30	46	wt	EPO, C	X

MF samples are classified according to the WHO criteria.

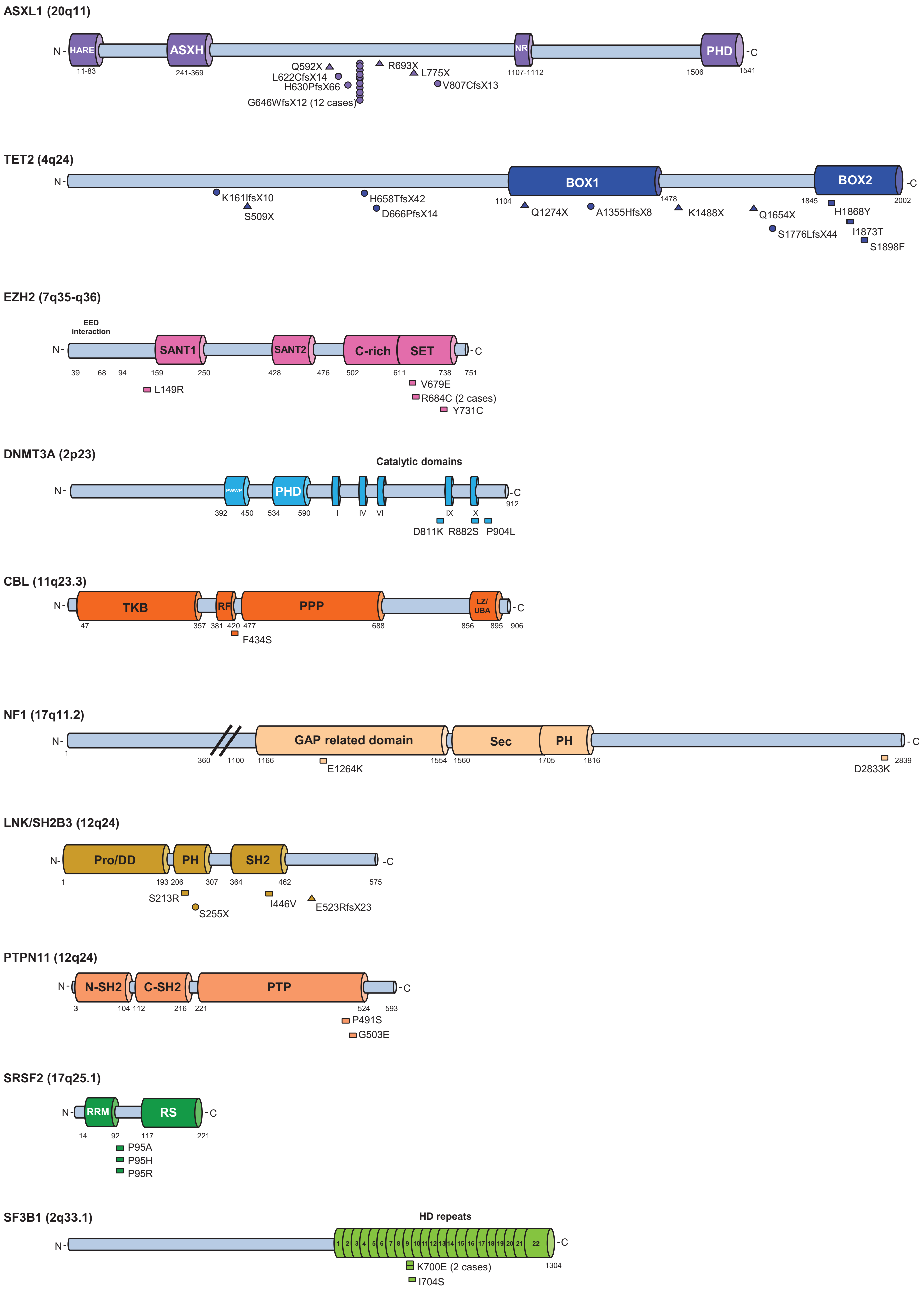
aCGH, array comparative genomic hybridization; A, anagrelid; B, busulfan; BM, bone-marrow; C, corticoid; Ci blasts, circulating blasts; DIPSS, dynamic international prognostic scoring system; DIPSSplus, dynamic international prognostic scoring system plus; E, eposin; F, female; HU, hydroxyurea; IPSS, international prognostic scoring system; m, month; M, male; ME, melphalan; MF, myelofibrosis; NA, not applicable; ND, not determined; P, pipobroman; PA, peginterferon alfa-2a; PB, peripheral blood; PMF, primary myelofibrosis; post-ET MF, post-essential thrombocythemia MF; post-PV MF, post-polycythemia vera MF; RCT need, red cell transfusion need; T, thalidomide; wt, wild-type; y, years. For PMF, IPSS, DIPSS and DIPPSplus scores depend on age (>65 years), on hemoglobin (< 10 g/dL), on leukocyte count (> 25.10^9/L), on circulating blasts (> or = 1%), on platelet count (< 100.10^9/L), on presence of constitutional symproms (weight loss, night sweats, fever). For DIPPSplus score, karyotype and transfusion status must be added to DIPSS. IPSS score was calculated at MF diagnosis whereas DIPPS and DIPPSplus score were calculated at sampling.

colored rectangles, with position in Mb. The colored lines indicate the deleted region(s) in each patient. aCGH: array comparative genomic hybridization, AML: acute myeloid leukemia, aSNP: array single nucleotide polymorphism, CEL: chronic eosinophilic leukemia, CMML: chronic myelomonocytic leukemia, ET: essential thrombocythemia, FISH: fluorescence in situ hybridization, MDS: myelodysplastic syndrome, MPN: myeloproliferative neoplasm, MF: myelofibrosis, sAML, secondary acute myeloid leukemia, PMF: primary myelofibrosis, PV: polycythemia vera.

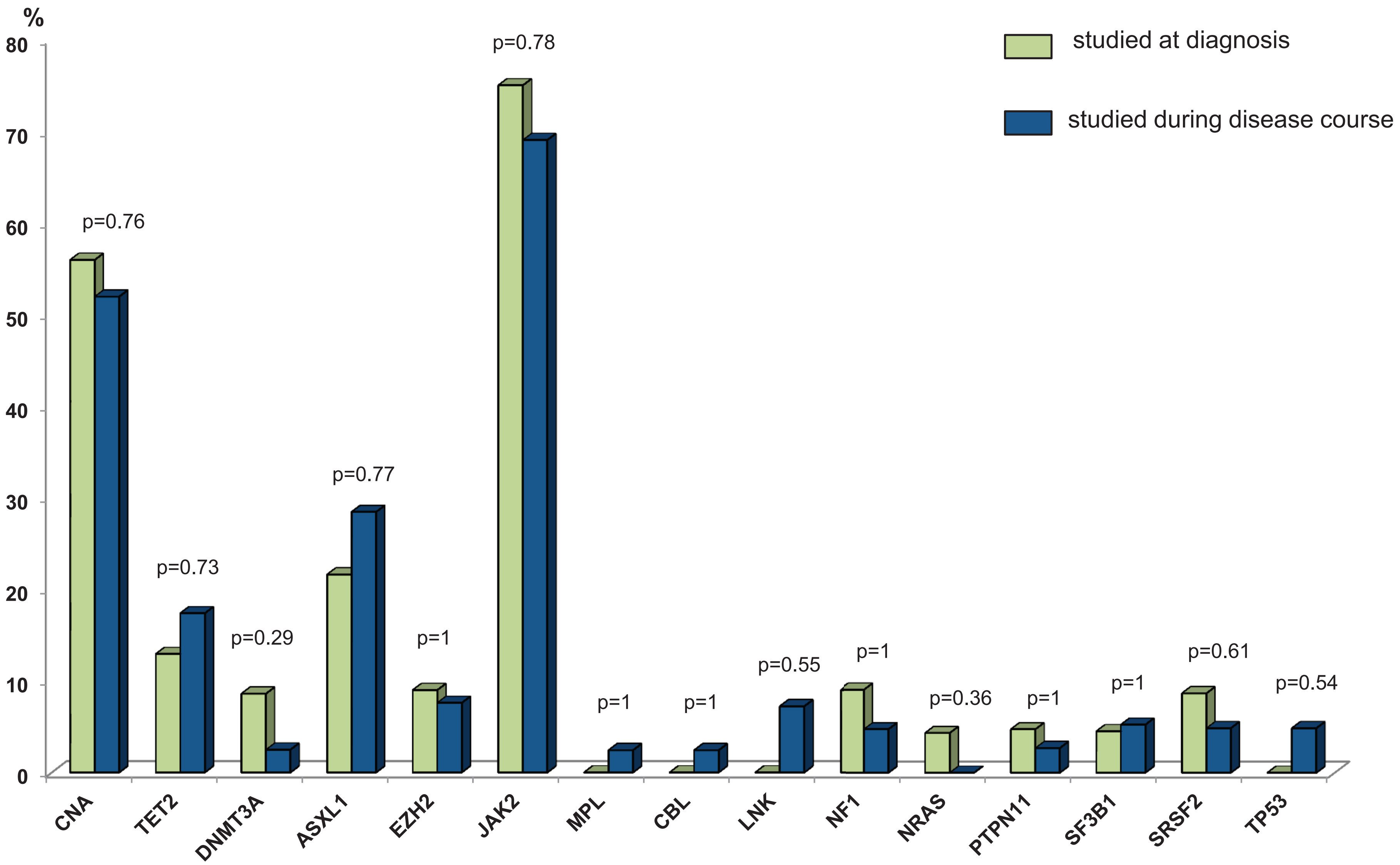
Supplemental Figure 1



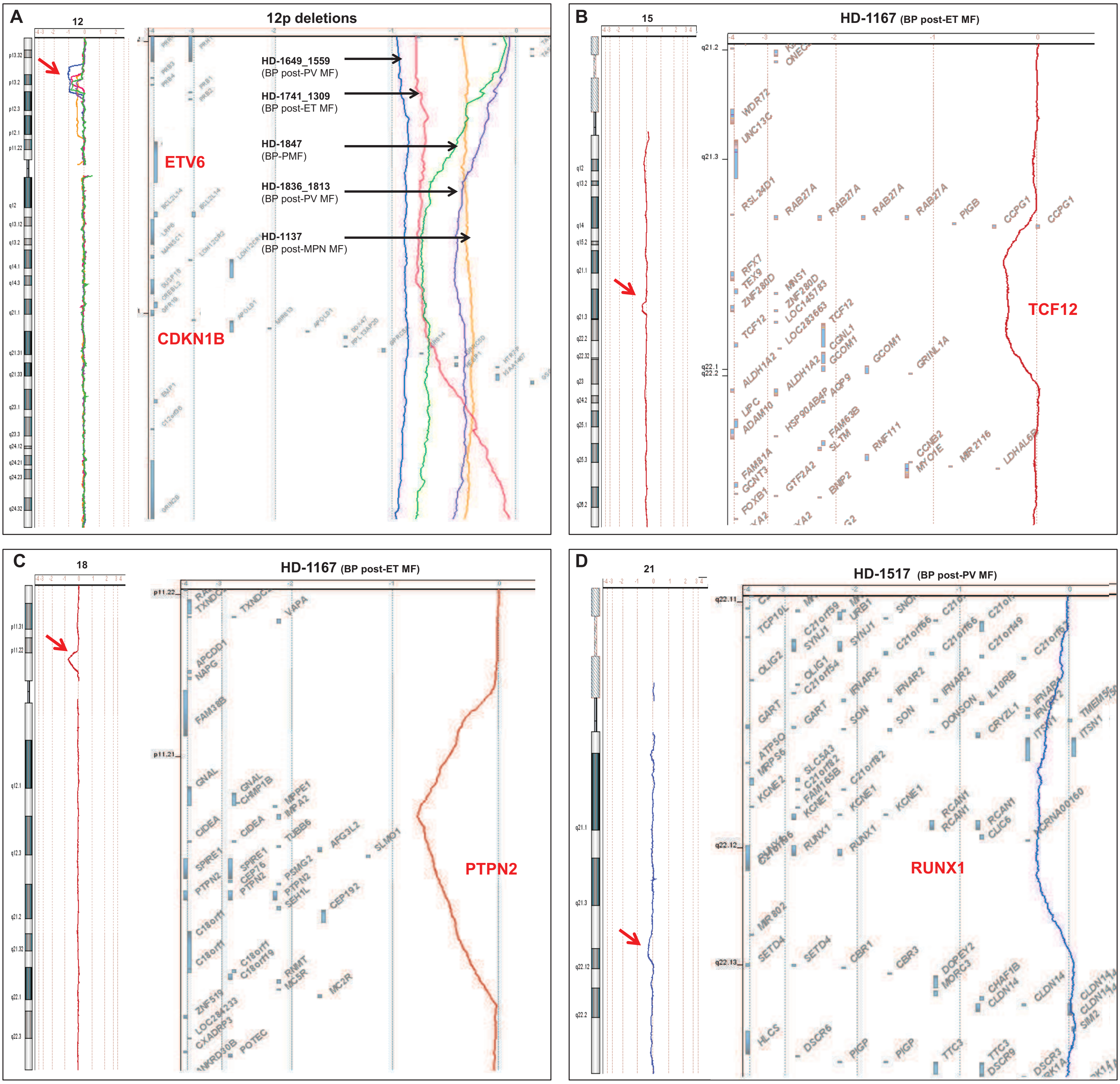
Supplemental Figure 2



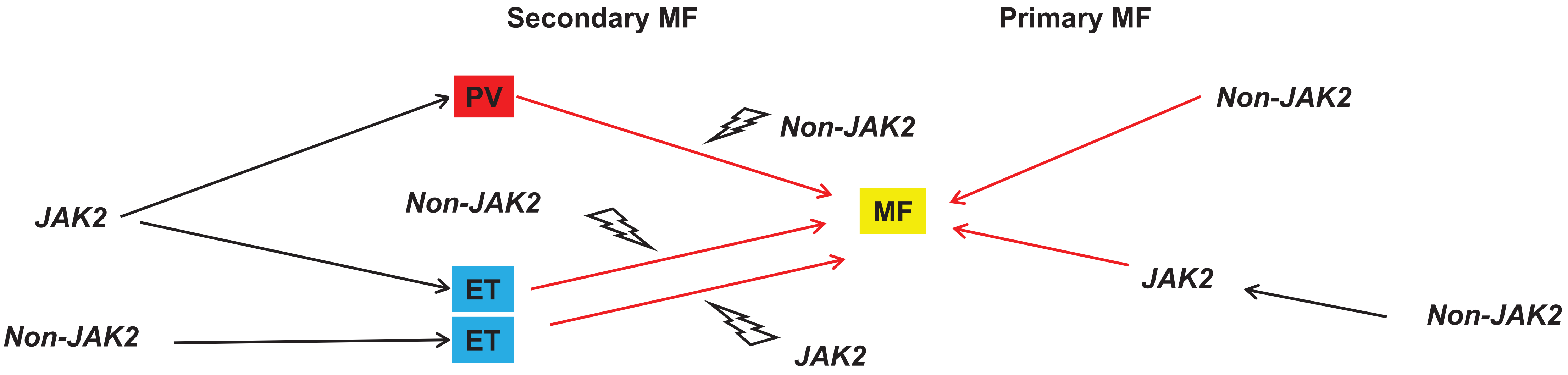
Supplemental Figure 3



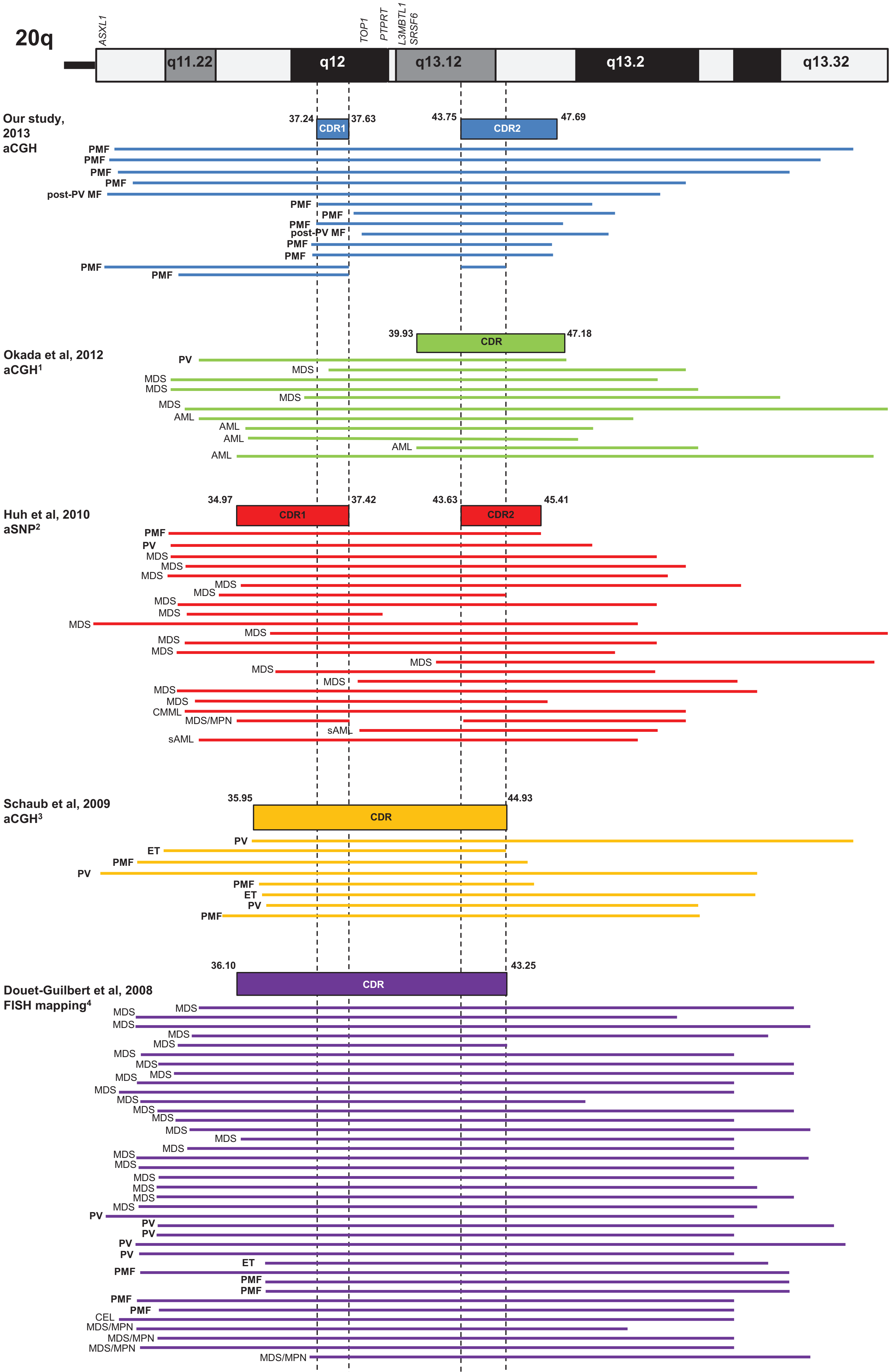
Supplementary Figure 4



Supplemental Figure 5



Supplemental Figure 6



Supplemental References

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