

Diagnosis and prognosis are supported by integrated assessment of next-generation sequencing in chronic myeloid malignancies. A real-life study

Sophie Vantyghem,¹ Pierre Peterlin,¹ Sylvain Thépot,^{2,3} Audrey Ménard,⁴ Viviane Dubruille,¹ Camille Debord,⁴ Thierry Guillaume,¹ Alice Garnier,¹ Amandine Le Bourgeois,¹ Soraya Wuilleme,⁴ Catherine Godon,⁴ Olivier Theisen,⁴ Marion Eveillard,^{3,4} Jacques Delaunay,⁵ Hervé Maisonneuve,⁶ Nadine Morineau,⁶ Bruno Villemagne,⁶ Stéphane Vigouroux,⁶ François Subiger,⁷ Elsa Lestang,⁸ Marion Loirat,⁸ Anne Parcelier,⁹ Pascal Godmer,⁹ Mélanie Mercier,⁹ Adrien Trebouet,¹⁰ Damien Luque Paz,^{3,11} Ronan Le Calloch,¹² Lenaig Le Clech,¹² Céline Bossard,¹³ Anne Moreau,¹³ Valérie Ugo,^{3,11} Mathilde Hunault,^{2,3} Philippe Moreau,^{1,3} Steven Le Gouill,^{1,3} Patrice Chevallier,^{1,3} Marie C. Béné^{3,4} and Yannick Le Bris^{3,4}

¹Hematology Clinic, Nantes University Hospital, Nantes; ²Hematology Clinic, Angers University Hospital, Angers; ³CRCINA, INSERM, CNRS, Université de Nantes, Université d'Angers, Pays de la Loire; ⁴Hematology Biology, Nantes University Hospital, Nantes; ⁵Le Confluent, Nantes; ⁶Hematology Clinic, Vendée Hospital Center, La Roche sur Yon; ⁷Biology Laboratory, Vendée Hospital Center, La Roche sur Yon; ⁸Hematology Clinic, Saint Nazaire Hospital, Saint Nazaire; ⁹Hematology Clinic, Bretagne Atlantique Hospital, Vannes; ¹⁰Hematology Clinic, Bretagne Sud Hospital, Lorient; ¹¹Hematology Biology, Angers University Hospital, Angers; ¹²Hematology Clinic, Quimper Hospital, Quimper and ¹³Pathology Department, Nantes University Hospital, Nantes, France

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Correspondence: YANNICK LE BRIS - yannick.lebris@chu-nantes.fr

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Supplemental methods and data

Supplemental Table 1 : Targeted genes panel

Gene	Chromosome	Cytoband	Transcript NCBI	Transcript ENSEMBL	Targeted Exons
ASXL1	20	q11.2	NM_015335.5	ENST00000176927	13-15
BCL6	X	Xp11.4	NM_001123385	ENST00000378444	Full CDS
BCL6L1	X	Xq25.1	NM_001047172	ENST00000181477	Full CDS
CBF1	11	11q23.3	NM_051988.2	ENST00000264033	4-17
CSP3R	1	1p34.3	NM_056909	ENST00000319103	8-9
DNMT3A	2	2p23.3	NM_175262.1	ENST00000264709	Full CDS
ETV6	12	12p13.2	NM_001897.4	ENST0000030273	Full CDS
ETP2	7	7q36.1	NM_004465.4	ENST00000303256	2-6
GATA2	3	3p21.3	NM_002358	ENST00000311106	2-6
IKZF1	2	2q34	NM_005886.2	ENST00000345146	4
IKZF2	15	15q26.1	NM_002186.2	ENST00000330662	2-6
JAK2	9	9p24.1	NM_004972.3	ENST00000381652	Full CDS
KDM6A	X	Xp11.3	NM_021140	ENST00000377965	Full CDS
KIT	4	4q12	NM_002022.2	ENST00000288155	9-11/17
KRAS	12	12p12.1	NM_004953.3	ENST0000011936	2-4
MPL	1	1p34.2	NM_000373.2	ENST00000374710	Full CDS
NPM1	5	5q35.1	NM_002520.6	ENST00000269920	11
NUP214	5	5q31.3	NM_001246208.1	ENST00000384644	Full CDS
NRAS	X	Xp11.3	NM_002524.4	ENST00000389535	2-3
PKNOX1	X	Xq25.2	NM_002611	ENST00000336950	Full CDS
PPFWD1	12	12q24.13	NM_002834	ENST00000316777	2-3
RAG2	8	8q24.11	NM_002019.2	ENST00000370738	1-13
RUNX1	21	21q22.12	NM_001754.4	ENST00000437180	3-9
SETBP1	18	18q12.3	NM_055656.2	ENST00000200330	4
SF3B1	2	2q31.1	NM_012433.2	ENST00000335508	13-18
SMDL4	X	Xp11.22	NM_004443.3	ENST00000322113	Full CDS
SMC3	10	10q25.2	NM_004443.3	ENST0000031804	Full CDS
SRSF2	17	17q12.1	NM_003016.4	ENST00000300465	1
STAT2	X	Xq25	NM_001247249	ENST00000218089	Full CDS
TEF2	4	4q24	NM_001127208	ENST00000380013	Full CDS
TNFSF10	6	6q23.3	NM_001270508	ENST00000237289	Full CDS
TNFSF17	17	17p13.1	NM_005454.1	ENST00000389356	Full CDS
UGAF1	21	21q22.3	NM_006758.2	ENST00000291552	2-5/6
ZRSR2	X	Xp22.2	NM_005068.3	ENST00000307771	Full CDS

CDS: coding DNA segments

Supplemental table 2 : Details of the results of cytogenetic analyses and NGS sequencing and their indications.

UPN	Pathology	Group A	Group B	Karyotype results	Genes	c.	t.	p.	NAF (%)	VS gene f	p.	p.	NAF (%)	
F1	MDS	1	0	46,XX[20]										
F2	MDS/MPN	0	1	46,XY[20]	ASXL1 RUNX1	c.1900_1922del c.314A>G		p.Glu45Asp+15 p.His105Arg	18% 26%					
F3	MDS	0	1	culture failure	TP53 JAK2 DNMT3A TEF2	c.464C>T c.1849G>T c.2855G>A c.5248delG		p.Pro132Ser p.Val817Phe p.Arg620His p.Asp1750Thr+13	2% 44% 4% 2%					
F4	MDS/MPN	0	1	47,XY,+8[246,X(1)8]	ASXL1 TEF2 UGAF1 TP53 CBF1 EP30 E2F8 JAK2 SETBP1	c.1809G>T c.1988_1994delinsGTTT c.479A>G c.745G>A c.1268G>T c.2098C>T c.2000C>T c.1849G>T c.5202G>A		p.Glu537* p.Phe620Leu+17 p.Gln137Arg p.Arg380His p.Arg607* p.Arg60Cys p.Arg64Cys p.Val817Phe p.Arg654Asn	44% 27% 44% 4% 2% 2% 1% 2%					
F5	MDS	0	1	46,XX[20]	TP53 SF3B1 ASXL1	c.711G>A c.1774G>A c.1934del		p.Met237Ile p.Glu52Lys p.Gly487Phe+12	34% 30% 27%					
F6	MDS	1	0	unrealized (thrombocytosis context)	DNMT3A MPL	c.1570G>A c.1544G>T		p.Cys581Tyr p.Tyr515Leu	47% 6%					
F7	MDS	0	1	46,XX[20]	JAK2	c.1849G>T		p.Val817Phe	43%					
F8	MDS/MPN	0	1	47,XY,+8[846,XY,+del(1)(p24-q3)(8)(45XY)[4]	SF3B1 JAK2	c.1866G>C c.1849G>T		p.Glu222Asp p.Val817Phe	26% 12%					
F9	MDS/MPN	1	0	46,XX[20]	DNMT3A	c.1544G>T		p.Val817Phe	50%	JAK2	c.3323A>G	p.Asn1108Ser	50%	
F10	MDS	0	1	46,XY[20]	SRSF2 TEF2 DNMT3A KMT6A	c.284C>T c.3873G>A c.2796_2798dup c.163A>G		p.Pro308His p.Tyr1291* p.Gln533_368del+18p p.Ser15Arg	26% 27% 29% 2%					
F11	MDS	1	1	unrealized (thrombocytosis context)							JAK2	c.3323A>G	p.Asn1108Ser	50%
F12	MDS	0	1	46,XY[20]	DNMT3A	c.1253G>A		p.Gly418Asp	15%		JAK2	c.3323A>G	p.Asn1108Ser	50%
F13	MDS	0	1	46,XY,del(20)(q11q13)(9)(45,XY[17])	JAK2 ASXL1 UGAF1 TEF2 MPL	c.1849G>T c.1934del c.1270del c.111 c.1771T>G		p.Val817Phe p.Gly487Phe+12 p.Gln137Arg p.Ser444Asp+3 p.Tyr581Asp	22% 11% 4% 2%					
F14	MDS	1	0	46,XY[20]										
F15	Cytopenia	1	0	46,XY[20]	ASXL1	c.1934del		p.Gly487Phe+12	18%					
F16	MDS	0	1	46,XY[20]	ASXL1 ASXL1	c.2407C>T c.1772del		p.Gln803* p.Tyr581*	23% 6%					
F17	MDS	0	1	unrealized (unassprable marrow)	TEF2	c.1660del		p.Thr556Asn+11	35%					
F18	MDS	0	1	46,XY[26]										
F19	MDS	0	1	46,XY[20]	TP53 SF3B1 DNMT3A SRSF2 MPL	c.817C>T c.844C>T c.1934del c.284C>T c.345C>G		p.Arg273Cys p.Arg287Tyr p.Arg1408His p.Phe65Leu p.Gly12Arg	6% 2% 21% 21% 4%					
F20	MDS	0	1	45,X,-X[18],46,XX[20]	TEF2 TEF2 ZRSR2 ZRSR2	c.4100C>T c.3594G>A c.985G>T c.985G>T		p.Pro136Leu p.7 p.Cys129Phe p.Arg269*	14% 11% 13% 2%					
F21	MDS	0	1	culture failure	TEF2	c.39561G>A		p.7	3%					
F22	MDS	0	1	46,XX[20]	IKZF1	c.394C>T		p.R132C	4%					
F23	AcuteMDS	0	1	46,XY[20]										
F24	AcuteMDS	1	1	46,XY[20]	TP53	c.668del		p.Pro223Leu+24	2%	SETBP1	c.2117A>G	p.Glu706Gly	45%	
F25	MDS	0	1	unrealized (unassprable marrow)	ASXL1 ASXL1	c.2583dup c.2555_2558delG		p.Phe822Leu+2 p.Asn648Asp+9	58% 12%					
F26	MDS/MPN	1	1	46,XX[20]	TEF2 TEF2	c.3863G>A c.2081_2082dup		p.Gly1288Asp p.Met695Leu+9	26% 26%					
F27	MDS/MPN	1	1	46,XY,del(20)(q11q13)[20]	ASXL1 SRSF2 TEF2 TEF2	c.2254dup c.290C>G c.670C>T c.1633C>T		p.Ala753Gly+22 p.Pro308His p.Gln341* p.Arg544*	6% 48% 46% 42%					
F28	MDS	0	1	46,XY[20]	TEF2 TEF2	c.4240_4244del c.4781C>G		p.Gln1414Ser+10 p.Ser1589*	52% 44%	SETBP1 ZRSR2	c.851T>C c.1482C>G	p.Leu284Pro p.Arg98Gly	83% 50%	
F29	MDS/MPN	0	1	46,XY[20]	TEF2 TEF2 SF3B1 MPL	c.978G>A c.688G>A c.208A>G c.352A>A		p.Tyr1259* p.Ser188Tyr p.Lys700Gln p.Gly13Asp	36% 40% 33% 2%					
F30	MDS	0	1	46,XX,der(1)(3)(p11q11)[99],46,XX[11]	JAK2	c.1849G>T		p.Val817Phe	85%					
F31	MDS	0	1	unrealized (unassprable marrow)	ZRSR2 CBF1	c.1545del c.1511G>A		p.Ser368Leu+7 p.Val64Tyr*	41% 15%					
F32	MDS/MPN	0	1	46,XY[20]	ASXL1 RUNX1 SRSF2 TEF2	c.1934del c.1167dup c.283C>G c.454G>T		p.Gly487Phe+12 p.Gln390Asp+210 p.Pro308His p.Arg1518*	18% 43% 37% 48% 27%					
F33	MDS	1	0	46,XX[20]							BCL2L1	c.1834G>A	p.Ala121Thr	
F34	MDS	1	0	unrealized (thrombocytosis context)	MPL SF3B1 SF3B1 SF3B1	c.1544G>C c.2342G>C c.1890G>C c.1866G>C		p.Tyr155Ser p.Asp791Gly p.Val65Asn p.Glu222Asp	7% 13% 14% 8%	RAD21	c.1289A>C	p.Gln430Pro	46%	
F35	MDS	1	0	unrealized (polycythemia vera suspicion)										
F36	Cytopenia	1	0	46,XX[20]										
F37	MDS	1	0	unrealized (thrombocytosis context)	MPL ASXL1 TEF2	c.1544G>T c.1772dup c.407G>T		p.Tyr515Leu p.Tyr581* p.Asp1560Cys	3% 36% 8%					
F38	MDS	0	1	46,XY[20]	TEF2 DNMT3A	c.1865C>T c.2448C>T		p.Gln647* p.Gln1816*	59% 34%	SRSF2	c.238_240dup	p.Leu80dup	85%	
F39	MDS	1	0	46,XY[20]										
F40	MDS	0	1	46,XY,(1-19)(p22,q24)(8)	ASXL1	c.1871delT		p.Arg23Phe+10	28%		BCL2L1	c.3548G>A	p.Arg1183Gln	100%
F41	MDS/MPN	1	0	46,XY[20]	ASXL1 SETBP1 SRSF2 TEF2 PPFWD1 MPL	c.2302C>T c.2088G>A c.284C>A c.3778del c.1056G>T c.352A>A		p.Gln768* p.Gln768* p.Pro308His p.His1055Asp+14 p.Ser502Leu p.Gly12Arg	47% 20% 53% 46% 4% 2%					
F42	MDS/MPN	0	1	46,XX[20]	ASXL1 TEF2 TEF2	c.2783del c.2913G>G c.5127T>A		p.Val922Leu+23 p.Arg1811Gly p.Cys1709*	12% 20% 32%	SETBP1 KDM6A	c.1276G>A c.3337G>A	p.Ala428Thr p.Val1113Ile	50% 51%	
F43	MDS	1	0	46,XX[20]										
F44	MDS	1	0	unrealized (thrombocytosis context)	MPL MPL	c.1495delCAspGT c.1544G>T		p.His491Val+46 p.Tyr515Leu	5% 4%					
F45	MDS	1	0	46,XX[20]										
F46	MDS	1	0	46,XX[20]										
F47	MDS	1	0	unrealized (polycythemia vera suspicion)										
F48	MDS	1	0	46,XY[20]	JAK2	c.1849G>T		p.Val817Phe	1%	NO7CH1 DNMT3A	c.5248G>A c.1555-3T>A	p.Val1750Met p.7	55% 8%	

