Myeloid/lymphoid neoplasms with eosinophilia/basophilia and *ETV6-ABL1* fusion: cell-of-origin and response to tyrosine kinase inhibition

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Table S1: *ETV6* breakpoints in myeloid neoplasms with *ETV6-ABL1* fusion

Gene 1	Gene 2	Position 1	Position 2	Transcript 1	Exon No. 1	Transcript Strand 1	Transcript 2	Exon No. 2	Transcript Strand 2	Fusion Junction sequence	Frameshift class
ETV6	*ABL1*	Chr 12: 12022903	Chr 9: 133729451	NM_001987	5	+	NM_005157	2	+	GCCCATTGGGAGAATAGCAG/*AAGCCCTTCAGCGGCCAGTA*	In frame

Table S2: Mutational Profiling of myeloid neoplasms with *ETV6-ABL1* fusion

Case No.	Genes	Transcript ID	Exon No.	AA Change	cDNA Change	VAF (%)	Biological effect	Clinical Implications
1	*ARID2*	NM_152641	15	S731Vfs*27	2190delT	8	Likely loss of function	Likely oncogenic
	TP53	NM_000546	3	V31I	91G>A	47	NA	Variant of uncertain significance
2	*ARID2*	NM_152641	15	N697Kfs*10	2090dupA	36	Likely loss of function	Likely oncogenic
	CDKN1B	NM_004064	1	P133Qfs*12	398delC	39	Likely loss of function	Likely oncogenic
5	SETD2	NM_014159	17	S2382Lfs*47	7143dupC	3	Likely loss of function	Likely oncogenic
6	*CCT6B*	NM_006584	NA	NA	615-2A>G	NA	NA	Likely benign

Table S3 Clinical features of myeloid/lymphoid neoplasms with *ETV6-ABL1* fusion

Reference	Age at Dx(years)	Gender	Initial CBC	Initial Dx	Eosinophilia (% in PB)	absolute EO (K/mcL in PB)	Basophils (% in PB)	Progression	Splenomagaly	TKI	Other treatment	Survival
1	32	M	27/NA/454	CML	9	2.4	3		no	No		Alive (CR >3Y)
2	59	M	27/6.6/344	CML	15	4.1	5		yes	No	HU-Corticosteriod and Thioguanine	Died (13M)
3	53	M	22/13.1/378	CML	11	2.4	0		yes	No	HU/IFN,splenectomy	Alive (5Y)
4	44	F	37/12.3/370	CML	9	3.3	1		no	No	HU/IFN	Alive (3M)
5	38	M	Within normal	CML-BC	NA	NA	NA	AML	yes	Imatinib	HU-chemotherapy(FLAG)	Died (6M)
6	79	M	41.6/14/202	aCML	6	2.5	0	AML	NA	Imatinib	NA	Died(13M)
7	36	M	238/9.6/88	CML	13	30.9	4	AML	yes	Imatinib	chemotherapy	Died(8M)
8	72	M	6/9.8/32	CML	yes	NA	NA	B-ALL	NA	Imatinib		Died (2M)
9	26	F	40/ NA/NA	MPN	yes (marrow 20-30)	NA	NA	B-ALL	NA	Imatinib	chemotherapy	died (25M)
10	57	M	25/NA/NA	aCML	10	1.9	NA		NA	no	IFN	Alive (15Y)
11	36	M	55/NA/NA	aCML	3	1.7	2		yes	Imatinib	HU	Alive(5Y)
12	46	F	NA	aCML	yes	2.5	yes		NA	Yes	No	Yes (22M FU)
13	24	F	99/11.3/261	CML	4	4.0	1		yes	Imatinib		Alive (10M)
14	54	M	22/12.6/294	CML	yes	NA	NA		No	NA	NA	NA
15	65	F	22/11.1/63	MPN	yes (marrow)	NA	no		NA	No	chemotherapy	Died (12M)
16	61	F	92/11.4/400	MPN	12	11.0	10		yes	Imatinib, nilotinib		Alive (CR17m, relapse then CR11M)
17	31	M	51/11.3/438	MPN	11	5.6	NA	T-ALL (in LN)	NA	dasatinib	Chemotherapy-Allo-SCT	Died (11M)
18	59	M	25/NA/NA	MPN	yes	NA	NA		NA	Imatinib	chemotherapy	died(64M)
19	68	M	52/NA/NA	MPN	NA	NA	NA		NA	Nilo		Alive (CR>41M)
20	77	M	15/13.2/246	MPN	3.5	0.5	4		no	na	na	na
21	47	M	50/NA/NA	MPN	yes	NA	NA		NA	Imatinib, Dasatinib	None	Alive(9Y)
22	48	M	8/6.5/20	RAEB-2/AML	yes (marrow)	NA	1		NA	No	IA regimen chemotherapy	Died (6M)
23	29	M	40/12.6/139	AML	42	16.8	2		yes	No	GIMEMA protocol LAM99, then haploidentical SCT	Alive (20M)
24	81	M	na	AML	NA	NA	NA		NA	No		Died (3M)

25	40	F	83/NA/NA	AML	Yes	NA	NA	NA	No	chemotherapy	Died (4M)
	Median age: 48	M:F 18:7	Median 40/11.4/249		Median 10.5	Median 2.9	Median 2.5				Median: 13M

Table S4 Cytogenetic and molecular features of myeloid neoplasms with *ETV6-ABL1* fusion

Reference	Karyotype	FISH/ETV6	FISH/ABL1	Fusion Type (*ETV6-ABL1* A/B or *ETV6-FLT3*)
1	46,XY,t(12;14)(p12;q11-13)	5'ETV6 on 9q34	ABL1 3'/5' on 9q34	B
2	46,XY,del(6)(p21),?t(9;12) (q34;p12)	5'ETV6 on 9q34	ABL1 3'/5' on 9q34	B
3	46,XY	NA	ABL1 3'/5' on 9q34	B
4	46,XX,t(9;12)(q34;p13)	5'-3'ETV6 on 9q34	ABL1 3'/5' on 9q34	N/A
5	49, XY,+11, t(9;12)(q34;p1?), +der(12)t(9;12), +19, der(22)t(1;22)(q21;q11)	NA	3'ABL1 on 12p	B, A
6	46,XY	5'-3'ETV6 on 12p	3'ABL1 on 12p	NA
7	45,XY-7,t(9;12)(q34;q13)	NA	3'ABL1 on 12p	B
8	46,XY,t(12;17) (p11.2;p11.2)	5'ETV6 on 17p	3'ABL1 on 17p	B
9	46, XX, der(9)t(9;12)(q34;p13), del(10)(q24), del(11)(q23), der(12)t(10;12)(q24;p13)[8]/46, XX[2].		ETV6/ABL1	B
10	46,XY	NA	3'ABL1 on 12p	NA
11	46,XY, t(9;12)(q34;p13)	NA	ETV6/ABL1(cytogenetics)	NA
12	46,XX,t(9;12)(q34;p13)	5'ETV6 on 9q34	ABL1 3'/5' on 9q34	B
13	46,XX	5'ETV6 on 9q34	ABL1 3'/5' on 9q34	B, A
14	46,XY,t(9;12)(q34;p13)		9q34	B
15	46,XX ,t(5;9)(q13;q34)	NA	3'ABL1 on 12p	B, A
16	46,XX	5'-3'ETV6 on 12p	3'ABL1 on 12p	NA
17	46,XY,der(9)t(9;12)(q34;p13),del(12)(p13)[1]/46,sl,t(7;14)(p13;q11.2)[18]/47,sdl1,+19[1]	positive	positive	B, A
18	46,XY,del(6), t(9;12)(q34;p13)	NA	3'ABL1 on 12p	B, A
19	46,XY,t(5;12;16)	abnormal	abnormal	B, A
20	46,XY,t(9;12)(q34;p13)	NA	NA	na
21	46,XY,add(9)(q34),der(12)ins(12;9)(p13;q34q34),t(21;22)(q21;q11.2)[20]	NA	ABL1X3	NA
22	46,XY,t(9;12)(q34;p13)	NA	3'ABL1 on 12p	B
23	46,XY,t(8;12)(p21;p13)	5'ETV6 on 8p21	3'ABL1 on 8p21	A
24	45, add(X)(p11), Y, 23, der(3) t(3;15)(q12;q15), t(4;7)(q21;q22), 25, t(9;12;14)(q34;p13;q22), del(12)(p11p13), der(15) del(15)(q11q15), t(3;15)(q12; q15), der(18) t(3;18)(q11;q1?2), 1mar	NA	NA	B
25	46,XY	5'-3'ETV6 on 12p	3'ABL1 on 12p	B

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