

Supplemental tables

Molecular characterization of AML with *RUNX1-RUNX1T1* at diagnosis and relapse reveals net loss of co-mutations

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Supplemental table 1:
Patients' characteristics at relapse

Molecular relapse only	n=13
Morphologic relapse*	n=4
White blood cell count ($10^9/l$)	
median	5.2
range	1.6-37
Hemoglobin (mg/dl)	
median	13
range	8-16
Platelet count ($10^9/l$)	
median	121
range	9-250
<i>RUNX1-RUNX1T1/ABL</i> ratio	
median	73
range	1.7-50
	0

*) morphologic review is not routinely performed at follow up due to the nature of the central review lab.

Supplemental table 2: sequencing quality and results

Case No	Gene	Result	DNA	AA	Type	DNA Input	DNA conc	reads total (coverage)	wild type reads	mutated reads	VAF	Technology	DNA Input	DNA conc	reads total (coverage)	wild type reads	mutated reads	VAF	Technology
Diagnosis													Relapse						
1	ASXL1	positive	c.1900_1922del	p.Glu635Argfs*15	frame-shift	1 µl	25 ng/µl	11981	6627	5354	45	NGS	2 µl	25 ng/µl	13269	7715	5554	42	NGS
	NRAS	positive	c.34_35delinsTA	p.Gly12Tyr	missense	1 µl	25 ng/µl	6033	4923	1110	18	NGS	1 µl	25 ng/µl	3190	3190	0	-	NGS
	KIT	positive	c.2446G>T	p.Asp816Tyr	missense	1 µl	25 ng/µl	1979	1979	0	-	NGS	2 µl	25 ng/µl	2166	1182	984	45	NGS
2	KRAS	positive	c.38G>A	p.Gly13Asp	missense	1 µl	25 ng/µl	858	796	62	7	NGS	1 µl	25 ng/µl	2217	2217	0	-	NGS
3	ASXL1	positive	c.1900_1922del	p.Glu635Argfs*15	frame-shift	1 µl	25 ng/µl	3925	3426	499	13	NGS	1 µl	25 ng/µl	-	-	-	-	Sanger*
	IDH2	positive	c.419G>A	p.Arg140Gln	missense	1 µl	25 ng/µl	2152	1283	869	40	NGS	1 µl	25 ng/µl	-	-	-	10	Sanger*
4	KIT	positive	c.2441_2442insAAGGGT	p.Arg815_Asp816insValArg	in-frame	1 µl	25 ng/µl	3000	2004	996	33	NGS	2 µl	25 ng/µl	1245	641	604	49	NGS
5	ASXL2	positive	c.1293_1294insTACT	p.Lys432Tyrfs*2	frame-shift	1 µl	25 ng/µl	7987	4210	3777	47	NGS	2 µl	25 ng/µl	12728	12579	149	1,2	NGS
	KIT	positive	c.2445_2446delinsGC	p.Asp816His	missense	1 µl	25 ng/µl	3177	2781	396	12	NGS	2 µl	25 ng/µl	2445	2445	0	-	NGS
	KIT	positive	c.2466T>G	p.Asn822Lys	missense	1 µl	25 ng/µl	5663	4271	1392	25	NGS	2 µl	25 ng/µl	5849	5695	154	2,63	NGS
	PHF6	positive	c.890G>T	p.Cys297Phe	missense	1 µl	25 ng/µl	1081	1081	0	-	NGS	2 µl	25 ng/µl	1089	1047	42	3,86	NGS
6	KIT	positive	c.2466T>A	p.Asn822Lys	missense	1 µl	25 ng/µl	5302	4122	1180	22	NGS	1 µl	25 ng/µl	865	865	0	-	NGS
	KIT	positive	c.2466T>G	p.Asn822Lys	missense	1 µl	25 ng/µl	5302	4745	557	11	NGS	1 µl	25 ng/µl	865	865	0	-	NGS
7	ASXL2	positive	c.2377_2441del	p.Pro793Serfs*12	frame-shift	1 µl	25 ng/µl	3439	3354	85	3	NGS	1 µl	25 ng/µl	2689	2689	0	-	NGS
	ASXL2	positive	c.2218_2219insAGGC	p.Thr740Lysfs*17	frame-shift	1 µl	25 ng/µl	9073	5277	3796	42	NGS	1 µl	25 ng/µl	2689	2689	0	-	NGS
8	ASXL1	positive	c.1900_1922del	p.Glu635Argfs*15	frame-shift	1 µl	25 ng/µl	5429	4931	498	9	NGS	2 µl	25 ng/µl	5440	3503	1937	36	NGS
	ASXL1	positive	c.1882_1884delins7	p.Arg628Glyfs*8	frame-shift	1 µl	25 ng/µl	3502	2711	791	23	NGS	1 µl	25 ng/µl	2644	2644	0	-	NGS
	CSF3R	positive	c.2465T>A	p.Leu822Gln	missense	1 µl	25 ng/µl	2183	1323	860	39	NGS	1 µl	25 ng/µl	2571	2571	0	-	NGS
	CSF3R	positive	c.1853C>T	p.Thr618Ile	missense	1 µl	25 ng/µl	3866	2715	1151	30	NGS	1 µl	25 ng/µl	537	537	0	-	NGS
	NRAS	positive	c.34G>A	p.Gly12Ser	missense	1 µl	25 ng/µl	3548	3380	168	5	NGS	1 µl	25 ng/µl	1924	1924	0	-	NGS
	TET2	positive	c.5609C>T	p.Ser1870Leu	missense	1 µl	25 ng/µl	1894	1894	0	-	NGS	2 µl	25 ng/µl	2260	742	1518	67	NGS
9	ASXL2	positive	c.1771_1774dupCGCC	p.Gln592Profs*17	frame-shift	1 µl	25 ng/µl	4148	3217	931	22	NGS	1 µl	25 ng/µl	4965	4115	850	17	NGS
	FLT3TKD	positive	c.2504A>C	p.Asp835Ala	missense	1 µl	25 ng/µl	4171	2712	1459	35	NGS	1 µl	25 ng/µl	4701	2760	1941	41	NGS
10	KIT	positive	c.2446G>T	p.Asp816Tyr	missense	1 µl	25 ng/µl	794	794	0	-	NGS	1 µl	25 ng/µl	1747	1321	426	24	NGS
11	ASXL1	positive	c.1957_1960delinsCGGCA	p.Gly653Argfs*6	frame-shift	1 µl	25 ng/µl	8375	7573	802	10	NGS	2 µl	25 ng/µl	908	863	45	5	NGS
	TET2	positive	c.4964C>T	p.Pro1655Leu	missense	1 µl	25 ng/µl	13011	6198	6813	52	NGS	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.	n.a.
12	ASXL1	positive	c.1934dup	p.Gly646Trpfs*12	frame-shift	1 µl	25 ng/µl	-	-	-	10	Sanger*	1 µl	25 ng/µl	2255	2255	0	-	NGS
13	ASXL2	positive	c.2208_2219delinsAGTGGC	p.Ser736Argfs*18	frame-shift	1 µl	25 ng/µl	5579	5287	292	5	NGS	1 µl	25 ng/µl	4362	4362	0	-	NGS
	RAD21	positive	c.1354C>T	p.Gln452*	nonsense	1 µl	25 ng/µl	6874	6306	568	8	NGS	1 µl	25 ng/µl	1113	1113	0	-	NGS
14	SRSF2	positive	c.287delC	p.Pro96Argfs*148	frame-shift	1 µl	25 ng/µl	667	658	9	1,35	NGS	1 µl	25 ng/µl	819	819	0	-	NGS
15	FLT3TKD	positive	TKD	-	-	2 µl	50 ng/µl	-	-	5	-	Melting curve*	2 µl	50 ng/µl	-	-	-	-	Melting curve*

*) missing data was completed using routine diagnostic information including Sanger sequencing and melting curve analysis for 3 patients.