

Supplemental Data Table S5. Distribution of mtDNA minisatellite instability in the HV1 and HV2 regions of pediatric AML patients and controls

Location of Poly C tract	Reference sequence	Patterns in pediatric AML	N	%	Patterns in control	N	%	Comment
303 poly C (nt303-315 in HV2)	CCCCCCCCTCCCCCC (7CT6C)	7CT6C, 8CT6C 7CT6C, 8CT6C, 9CT6C 7CT6C, 8CT6C, 9CT6C, 10CT6C 7CT6C, 8CT6C, 9CT6C, 10CT6C, 11CT6C 7CT6C, 8CT6C, 9CT6C, 10CT6C, 11CT6C, 12CT6C 8CT6C, 9CT6C 8CT6C, 9CT6C, 10CT6C 8CT6C, 9CT6C, 10CT6C, 11CT6C 8CT6C, 9CT6C, 10CT6C, 11CT6C, 12CT6C 8CT6C, 9CT6C, 10CT6C, 11CT6C 8CT6C, 9CT6C, 10CT6C, 11CT6C, 12CT6C 9CT6C, 10CT6C, 11CT6C 12CT6C 12C, 13C, 15C, 16C, 17C, 18C 14C, 15C, 16C, 17C 14C, 15C, 16C, 17C, 18C, 19C 15C, 16C, 17C, 18C 15C, 16C, 17C, 18C, 19C	15 1 3 1 1 5 10 7 4 1 1 1 1 3 1	27.3 1.8 5.5 1.8 1.8 9.1 18.2 12.7 7.3 1.8 1.8 1.8 1.8 5.5 1.8	6CT6C, 7CT6C 7CT6C, 8CT6C 7CT6C, 8CT6C, 9CT6C, 10CT6C 8CT6C, 9CT6C 8CT6C, 9CT6C, 10CT6C, 11CT6C 8CT6C, 9CT6C, 10CT6C, 11CT6C, 12CT6C 10C, 11C, 12C, 13C, 14C, 15C 14C, 15C, 16C, 17C, 18C 15C, 16C, 17C, 18C 15C, 16C, 17C, 18C, 19C	1 15 7 6 10 10 1 1 1 1 1 2 1 1 1 1 55	1.8 27.3 12.7 10.9 18.2 18.2 1.8 1.8 1.8 1.8 1.8 3.6 1.8 1.8 1.8 100	Association of T310C
Total			55	100		55	100	
16,189 poly C (nt16,184-16, 193 in HV1)	CCCCCTCCCC (5CT4C)	4CT4C, 5CT4C 4CT4C, 5CT4C, 6CT4C, 7CT4C, 8CT4C, 9CT4C 4CT4C, 5CT4C, 6CT4C, 7CT4C 5CT4C 5CT4C, 6CT4C, 7CT4C, 8CT4C 6CT4C 4C, 5C, 6C, 7C, 8C 8C, 9C, 10C, 11C 8C, 9C, 10C, 11C, 12C, 13C, 14C 8C, 9C, 10C, 11C 8C, 9C, 10C, 11C, 12C 9C, 10C, 11C, 12C, 13C 9C, 10C, 11C, 12C 9C, 10C, 11C, 12C, 13C 10C, 11C, 12C, 13C 10C, 11C, 12C, 13C, 14C	5 2 1 25 1 2 1 1 1 1 1 1 1 1 1 1 1 1	9.1 3.6 1.8 45.5 1.8 3.6 1.8 1.8 1.8 1.8 1.8 1.8 1.8 1.8 1.8 1.8 1.8 1.8	4CT4C, 5CT4C 5CT4C 5CT4C, 6CT4C, 7CT4C, 8CT4C 5CT4C, 6CT4C, 7CT4C, 8CT4C, 9CT4C 8CT4C 8C, 9C, 10C, 11C, 12C, 13C 9C, 10C, 11C 9C, 10C, 11C, 12C, 13C, 14C 9C, 10C, 11C, 12C 10C, 11C 10C, 11C, 12C 10C, 11C, 12C, 13C 10C, 11C, 12C, 13C 10C, 11C, 12C, 13C, 14C	15 12 2 2 2 1 1 11 1 1 1 1 1 3 2 4 11 1 1 3	27.3 21.8 3.6 3.6 3.6 1.8 1.8 20 1.8 1.8 1.8 1.8 5.5 3.6 7.3 20 1.8 1.8 5.5	Association of T16189C
Total			55	100		55	100	
514 CA (nt514-523 in HV2)	CACACACACA (CA) 5repeats	(CA)4 (CA)5	19 36	34.5 65.5	(CA)4 (CA)5	19 36	34.5 65.5	
Total			55	100		55	100	