

Patient	Eye	Age at Dx (mos)	results of clinical testing for <i>RB1</i> mutation in leukocytes	time of AH and blood sampling	AJCC TNM classification of eye	IIRC classification of eye	tumor tissue available due to enucleation	SCNA in AH?	SCNA in blood?	peak fragment size in blood (bp) without library primers	peak fragment size in AH (bp) without library primers
Patient 1	OD	4	c.1666C>T	at diagnosis	cT1B	B	no	no	no	180	162
	OS	—	—	at diagnosis	cT2B	D	no	no	no	180	157
Patient 2	OS	22	not present	at diagnosis	cT2B	D	no	yes	no	180	149
Patient 3	OD	8	not present	at diagnosis	cT2B	D	no	yes	no	178	158
Patient 4	OD	16	c.1389+2T>C	at diagnosis	cT2B	D	no	yes	no	187	149
Patient 5	OS	10	not present	at diagnosis	cT3D	D	no	yes	no	182	166
Patient 6	OS	5	not present	at diagnosis	cT2A	C	no	no	no	178	153
Patient 7	OD	6	not present	during treatment	cT1B	B	no	yes	no	186	167
Patient 8	OD	26	c.2520+1G>A	during treatment	cT2A	C	no	yes	no	184	165
	OS	—	—	during treatment	cT2B	D	no	yes	no	184	146
Patient 9	OD	3	familial RB	during treatment	cT2B	D	no	yes	no	181	157
Patient 10	OD	4	not present	during treatment	cT2B	D	no	yes	no	186	164
Patient 11	OS	13	not present	during treatment	cT2B	D	no	yes	no	186	158
Patient 12	OD	9	c.2527delG	during treatment	cT3B	E	no	no	no	180	159
	OS	—	—	during treatment	cT3B	E	no	no	no	180	121
Patient 13	OD	2	c.1421+12_1421+3 2del21bp	during treatment	cT2B	D	no	no	no	184	165
Patient 14	OD	45	not present	during treatment	cT2B	D	no	no	no	179	165
Patient 15	OD	4	c.865A>T	during treatment	cT2B	D	no	no	no	178	155
Patient 16	OS	13	c.1494t>G	during treatment	cT3C	E	no	no	no	188	157
Patient 17	OD	2	c.2425delC	during treatment	cT3C	E	yes	yes	no	185	151

Table 1 (supplemental): Patient and Sample Demographics

AH=aqueous humor

RB=retinoblastoma

RB1=retinoblastoma tumor suppressor gene

AJCC TNM=American Joint Committee on Cancer (Tumor Node Metastasis), 8th edition

IIRC: International Intraocular Retinoblastoma Classification

SCNA=somatic copy number alteration

bp=base pairs

Patient 17

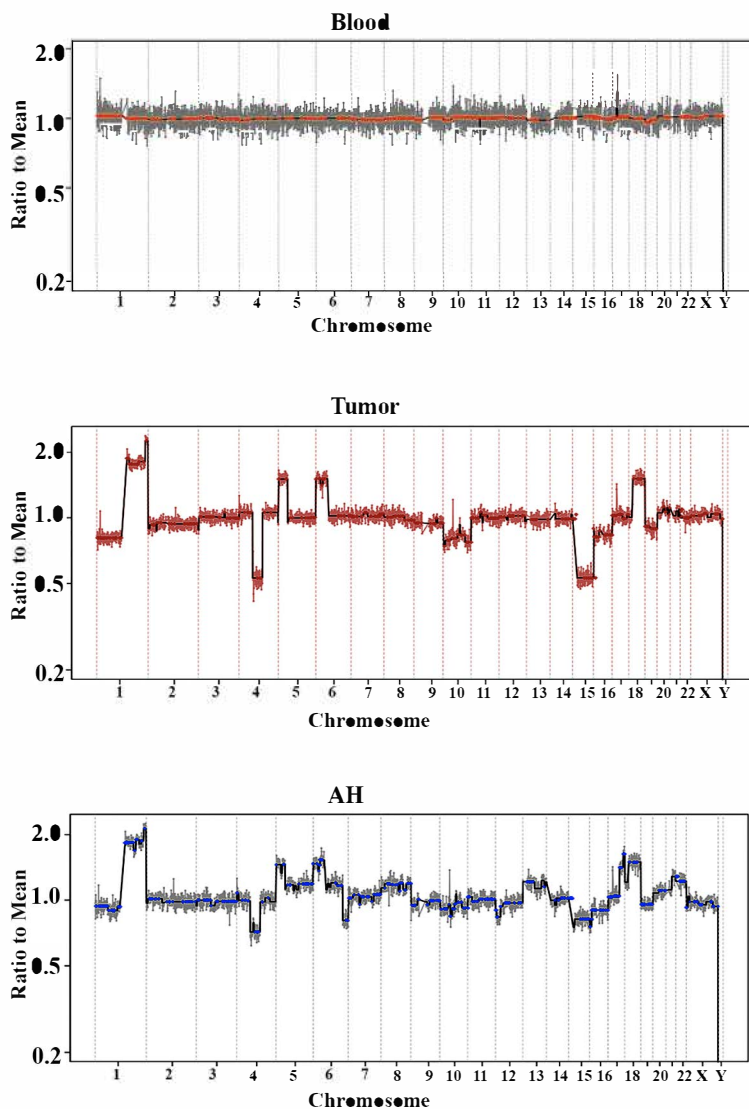


Figure 2 (supplemental): The genomic profile from the AH is concordant with the profile from the tumor
Direct evaluation of the tumor (secondary to enucleation) was done for Patient 17. The chromosomal alterations seen in the cfDNA from the AH are concordant and consistent with those from the tumor; the cfDNA in the blood did not demonstrate any similar alterations.