

Supplemental Table S2. Histopathological features, biallelic inactivation status of *FH*, and somatic *MED12* exon 2 mutation status of 34 uterine leiomyomas from 14 HLRCC patients.

Family name	Patient	Tissue material	Mitotic figures / 10 HPF	Atypia	Cellularity	Orangeophilic nucleoli	Germline <i>FH</i> mutation	<i>FH</i> second hit	Somatic <i>MED12</i> exon 2	Germline <i>MED12</i> exon 2***
FAM-2 (B)	3	FFPE	5	0	+	-	c.671_672delAG, p.E224fs	-	c.131G>A, p.G44D	wt
		FFPE	NA	NA	NA	NA	c.671_672delAG, p.E224fs	-	wt	wt
		FFPE	7	1	+	-	c.671_672delAG, p.E224fs	LOH	wt	wt
	7	fresh frozen	NA	NA	NA	NA	c.671_672delAG, p.E224fs	-	c.113-160del48, p.A38_G53del	wt
		fresh frozen	0-1	0	-	-	c.671_672delAG, p.E224fs	LOH*	wt	wt
		fresh frozen	0-1	0	-	-	c.671_672delAG, p.E224fs	protein deficient*	wt	wt
		fresh frozen	0-1	0	+	-	c.671_672delAG, p.E224fs	-	wt	wt
		fresh frozen	NA	NA	NA	NA	c.671_672delAG, p.E224fs	LOH*	wt	wt
fresh frozen	0-1	0	+	-	c.671_672delAG, p.E224fs	LOH*	wt	wt		
FAM-3 (C)	5	FFPE	0-1	0	-	-	c.1027C>T, p.R343X	LOH**	wt	-
		FFPE	0-1	0	-	-	c.1027C>T, p.R343X	LOH**	wt	-
		FFPE	0-1	0	+	(+)	c.1027C>T, p.R343X	-	wt	-
	8	FFPE	0-1	1	+	++	c.1027C>T, p.R343X	LOH	wt	-
		FFPE	NA	NA	NA	NA	c.1027C>T, p.R343X	LOH	wt	-
FAM-4 (D)	10	FFPE	0-1	1	++	+	c.587A>G, p.H196R	-	wt	-
		FFPE	0-1	1	++	++	c.587A>G, p.H196R	LOH	wt	-
FAM-5 (E)	1	FFPE	0-1	0	+	-	c.587A>G, p.H196R	- **	wt	wt
		FFPE	0-1	0	-	+	c.587A>G, p.H196R	- **	wt	wt
		FFPE	0-1	0	+	+	c.587A>G, p.H196R	- **	wt	wt
		FFPE	0-1	0	-	-	c.587A>G, p.H196R	- **	c.130G>A, p.G44S	wt
FAM-1 (M)	1	FFPE	0	0	-	-	c.671_672delAG, p.E224fs	LOH	wt	-
	2	FFPE	NA	NA	NA	NA	c.671_672delAG, p.E224fs	LOH	wt	-
	4	FFPE	0-1	1	++	++	c.671_672delAG, p.E224fs	-	wt	-
	17	FFPE	NA	NA	NA	NA	c.671_672delAG, p.E224fs	LOH**	wt	-
	18	FFPE	NA	NA	NA	NA	c.671_672delAG, p.E224fs	LOH**	wt	-
	26	FFPE	0-1	1	++	++	c.671_672delAG, p.E224fs	LOH	wt	-
	27	FFPE	NA	NA	NA	NA	c.671_672delAG, p.E224fs	-	wt	-
		FFPE	0-1	0	+	-	c.671_672delAG, p.E224fs	-	wt	-
FAM-9 (N)	7	fresh frozen	0-1	0	-	-	c.1027C>T, p.R343X	LOH**	wt	-
		fresh frozen	NA	NA	NA	NA	c.1027C>T, p.R343X	LOH**	wt	-
		fresh frozen	0-1	0	+	-	c.1027C>T, p.R343X	c.959C>A, p.A320D**	wt	-
		fresh frozen	0-1	0	+	-	c.1027C>T, p.R343X	c.959C>A, p.A320D**	wt	-
		fresh frozen	0-1	0	-	-	c.1027C>T, p.R343X	c.302G>C, p.R101P**	wt	-
		fresh frozen	0-1	0	+	-	c.1027C>T, p.R343X	c.554A>T, p.Q185L**	wt	-

FFPE; formalin-fixed paraffin-embedded, HPF; high power field, NA; not available (tumor too small or block not available), LOH; loss of heterozygosity, *From Vanharanta et al. 2006, **Unpublished data, ***Determined when a tumor with a *MED12* exon 2 mutation was identified