

Supplementary table 1:
DHPLC analysis of PTK2 genomic sequence

Ex/Int	Variant	Aminoacid consequence	Splice site predictor	note
6	c.491-66 T>C		No consequence	Found in 2 patients
8	c.594-20 C>T		No consequence	Found in 6 patients
22	c.1947 T>C	Pro 649 Pro	No consequence	Found in 1 patient
28	c.2563-49 G>T		No consequence	Found in 2 patients
29	c.2652 T>C	His 884 His	No consequence	Found in 1 patient

Supplementary table 2:
dHPLC analysis of THOC2 genomic sequence

Ex/Int	Variant	Aminoacid consequence	Splice site predictor	note
2	c.71-91 T>G	none	No consequence	Found in 1 patients
22	c.2317-47 A>G	none	No consequence	Found in 6 patients
23	c.2482-49 G>A	none	No consequence	Found in 4 patients

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dHPLC analysis of THOC2 genomic sequence

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