

Additional File 5. Mutations selected for Sanger verification.

Gene Name	Chromosome	Position	Reference allele	Alternative allele	Type of mutation	ConsScore	PolyPhen	SIFT	CADD-Score	SampleID
<i>IDH2</i>	15	90628543	A	C	SNV	7	possibly_damaging	deleterious	11.61	Grade2-1, Grade2-2
<i>ADAMTSL3</i>	15	84657544	TGGAAAGT	T	DEL	7	NA	NA	17.6	Grade2-1, Grade2-2, Grade2-3, Grade2-4
<i>SMYD1</i>	2	88387407	T	G	SNV	7	benign	deleterious	18.33	Grade2-2
<i>EPHB3</i>	3	184297360	G	T	SNV	7	probably_damaging	deleterious	19.15	Grade1
<i>SLC35B1</i>	17	47783669	A	C	SNV	7	benign	tolerated	21.9	Grade2-2
<i>ECEL1</i>	2	233348878	A	G	SNV	7	probably_damaging	tolerated	24.1	Grade2-4
<i>BRD8</i>	5	137503737	C	A	SNV	7	probably_damaging	tolerated	24.2	Grade1
<i>SASH1</i>	6	148792578	T	A	SNV	7	probably_damaging	deleterious	25	Grade2-2
<i>IRF2</i>	4	185340674	A	C	SNV	7	benign	deleterious	25.2	Grade2-1
<i>CAPN5</i>	11	76804840	G	A	SNV	7	possibly_damaging	tolerated	25.9	Grade2-1, Grade2-2, Grade2-3, Grade2-4
<i>SLC4A3</i>	2	220504339	C	CA	INS	7	NA	NA	37	Grade2-1