Supplementary Table S1: DNA sample source, sequencing method and mutation summary

PitB CASE	Germ-line DNA Sample Source	Germ-line DNA Sequencing Method	Germ-line *DICER1* Mutation	Tumour DNA Sample Source	Tumour DNA Sequencing Method	Somatic *DICER1* Mutation
CASE 1	Not Available	N/A	N/A	FFPE tissue	PCR Sequencing of RNase IIIa & IIIb domains	c.5437G>A[a]
CASE 2	DNA extracted from blood lymphocytes	*DICER1* Full gene PCR Sequencing	c.3277_3280delAACT	FFPE tissue	PCR Sequencing of RNase IIIa & IIIb domains	Negative
CASE 3	DNA extracted from blood lymphocytes	*DICER1* Full gene PCR Sequencing	c.2379T>G	Not Available	N/A	N/A
CASE 4	DNA extracted from saliva	*DICER1* Full gene PCR Sequencing	c.3535_3538delTCTT	FFPE tissue	PCR Sequencing of RNase IIIa & IIIb domains	c.5125G>T
CASE 5	DNA extracted from 3-5mL EDTA-preserved blood	*DICER1* Full gene PCR Sequencing	c.1525C>T	FFPE tissue	PCR Sequencing of RNase IIIa & IIIb domains	c.5425G>T
CASE 6	FFPE tissue	Fluidigm Access Array; Next-generation Sequencing	c.4309_4312delGACT	FFPE tissue	PCR Sequencing of RNase IIIa & IIIb domains	c.5125G>A
CASE 7	Not Available	N/A	N/A	Not Available	N/A	N/A
CASE 8	FFPE tissue	*DICER1* Full gene PCR Sequencing	Poor quality DNA	FFPE tissue	PCR Sequencing of RNase IIIa & IIIb domains	Poor quality DNA
CASE 9	DNA extracted from saliva	*DICER1* Full gene PCR Sequencing	c.2026C>T	FFPE tissue & fresh frozen	PCR Sequencing of RNase IIIa & IIIb domains	c.5439G>T
CASE 10	FFPE tissue	Fluidigm Access Array; Next-generation Sequencing	c.2026C>T	FFPE tissue	PCR Sequencing of RNase IIIa & IIIb domains	c.5438A>T
CASE 11	DNA extracted from blood lymphocytes	Fluidigm Access Array; Next-generation Sequencing	c.1284delGA	FFPE tissue	PCR Sequencing of RNase IIIa & IIIb domains	c.5437G>A
CASE 12	DNA extracted from blood lymphocytes	*DICER1* Full gene PCR Sequencing	c.5125G>C	FFPE tissue & fresh frozen	PCR Sequencing of RNase IIIa & IIIb domains	LOH
CASE 13	DNA extracted from blood lymphocytes	Fluidigm Access Array & MLPA	Negative	Extracted tumour DNA	PCR Sequencing of RNase IIIa & IIIb domains; STR marker genotyping	LOH

In summary: germ-line and somatic samples were acquired from ten cases; tumour only from one case; germ-line only from one case; and no tumour or germ-line sample from one case. **Abbreviations:** FFPE, formalin-fixed paraffin-embedded; LOH, loss of heterozygosity; NMD, nonsense-mediated decay; MLPA, Multiplex Ligation-based Probe Amplification assay; STR, short tandem repeat.

[a] Mutation not confirmed to be somatic in origin.