

**Supplementary table 2** Results of the new version of the p53 functional assay on 34 lymphoblastoid cell lines derived from control individuals or individuals with germline *TP53* variants and comparison with the primary version of the assay

<i>TP53</i> variant <sup>a</sup>	Clinical presentation <sup>b</sup>	Primary version of the p53 functional assay, (Zerdoumi <i>et al.</i> , 2017)		New version of the p53 functional assay					
		RT-QMPSF		RT-QMPSF		RT-MLPA		Mean values	
		p53 score	mRNA (%)	p53 score	mRNA (%)	p53 score	mRNA (%)	p53 score	mRNA (%)
<i>Individuals with wild-type TP53 genotype</i>									
Control 1	Unaffected	13.1	100	16.7	100	10.4	100	13.6	100
Control 2	Unaffected	11.0	79	11.1	84	10.5	105	10.8	95
Control 3	Unaffected	14.6	100	12.0	79	10.9	97	11.5	88
<i>Individuals with dominant-negative TP53 missense variants</i>									
c.524G>A, p.(Arg175His)	RMS (1)	<b>4.1</b>	86	<b>6.4</b>	107	<b>5.1</b>	102	<b>5.8</b>	105
c.535C>T, p.(His179Tyr)	Glioblastoma (14), Osteosarcoma (16)	<b>3.8</b>	78	<b>3.8</b>	97	<b>3.2</b>	95	<b>3.5</b>	96
c.638G>A, p.(Arg213Gln)	Choroïd plexus carcinoma (0.3)	<b>4.1</b>	98	<b>5.1</b>	101	<b>6.1</b>	95	<b>5.6</b>	98
c.736A>G, p.(Met246Val)	Bilat. breast cancer (25)	<b>2.7</b>	90	<b>2.9</b>	99	<b>3.2</b>	95	<b>3.1</b>	97
c.742C>T, p.(Arg248Trp)	Breast cancer (29, 34)	<b>3.2</b>	73	<b>2.4</b>	89	<b>3.1</b>	98	<b>2.8</b>	94
c.743G>A, p.(Arg248Gln)	Osteosarcoma (16), Breast cancer (28)	<b>2.2</b>	76	<b>2.1</b>	89	<b>1.9</b>	92	<b>2.0</b>	91
c.818G>A, p.(Arg273His)	Choriocarcinoma (17, 19)	<b>1.9</b>	104	<b>2.9</b>	75	<b>2.7</b>	92	<b>2.8</b>	84
c.844C>T, p.(Arg282Trp)	Unaffected	<b>4.6</b>	76	<b>5.7</b>	80	<b>5.3</b>	85	<b>5.5</b>	83
<i>Individuals with null TP53 variants</i>									
c.(?-202)_(*1207_?)del, p.0	Unaffected	<b>7.2</b>	57	<b>6.0</b>	38	<b>8.1</b>	59	<b>7.1</b>	49
c.(?-202)_(29+1_-28+1)del, p.?	Unaffected	<b>5.4</b>	53	<b>5.9</b>	51	<b>5.7</b>	65	<b>5.8</b>	58
c.216dup, p.(Val73Argfs*76)	Breast cancer (28)	<b>6.5</b>	44	<b>5.8</b>	35	<b>4.9</b>	55	<b>5.4</b>	45
c.323_329dup, p.(Leu111Phefs*40)	Breast cancer (27)	<b>6.9</b>	59	<b>5.3</b>	59	<b>6.6</b>	73	<b>6.0</b>	66
c.455del, p.(Pro152Argfs*18)	Breast cancer (26, 29)	<b>4.7</b>	63	<b>4.3</b>	54	<b>3.7</b>	76	<b>4.0</b>	65
c.491_494del, p.(Lys164Serfs*5)	Osteosarcoma (11)	<b>8.6</b>	50	<b>9.1</b>	51	<b>8.0</b>	68	<b>8.6</b>	60
c.632_641del, p.(Thr211Ilefs*33)	Bilat. breast cancer (25)	<b>5.7</b>	50	<b>5.4</b>	54	<b>4.4</b>	77	<b>4.9</b>	66
c.673-2A>G, p. ?	Unaffected	<b>5.5</b>	42	<b>5.6</b>	40	<b>4.2</b>	61	<b>4.9</b>	51
c.690del, p.(Thr231Profs*16)	Unaffected	<b>9.4</b>	52	<b>10.4</b>	53	<b>11.1</b>	68	<b>10.8</b>	61
c.820del p.(Val274Phefs*71)	ACC (11)	<b>7.0</b>	53	<b>7.1</b>	41	<b>6.2</b>	71	<b>6.7</b>	56

<i>Individuals with other TP53 missense variants</i>									
c.31G>C, p.(Glu11Gln)	Neuroblastoma (2)	<b>3.0</b>	120	<b>4.9</b>	75	<b>5.8</b>	97	<b>5.4</b>	86
c.323G>A, p.(Gly108Asp)	Unaffected	<b>4.2</b>	81	<b>5.2</b>	81	<b>4.5</b>	100	<b>4.9</b>	91
c.523C>G, p.(Arg175Gly)	Osteosarcoma (18), Breast cancer (27), Lung cancer (43)	<b>3.2</b>	101	<b>3.6</b>	83	<b>3.4</b>	82	<b>3.5</b>	83
c.577C>G, p.(His193Asp)	Liposarcoma (24), Breast cancer (26)	<b>3.7</b>	91	<b>4.0</b>	109	<b>4.1</b>	88	<b>4.1</b>	99
c.578A>C, p.(His193Pro)	Leiomyosarcoma (49), ACC (54), Melanoma (55)	<b>3.7</b>	109	<b>3.7</b>	104	<b>4.6</b>	104	<b>4.2</b>	104
c.646G>A, p.(Val216Met)	RMS (3), Sarcoma (17)	<b>4.3</b>	94	<b>4.3</b>	93	<b>4.4</b>	98	<b>4.4</b>	96
c.685T>C, p.(Cys229Arg)	Breast cancer (34)	<b>4.3</b>	82	<b>5.3</b>	110	<b>4.7</b>	99	<b>5.0</b>	105
c.761T>C, p.(Ile254Thr)	Unaffected	<b>2.8</b>	72	<b>2.6</b>	65	<b>3.6</b>	88	<b>3.1</b>	77
c.845G>C, p.(Arg282Pro)	Choroid plexus carcinoma (17), Sarcoma (24)	<b>4.7</b>	105	<b>3.7</b>	98	<b>3.3</b>	98	<b>3.5</b>	98
c.869G>A, p.(Arg290His)	Breast cancer (47)	<b>10.9</b>	85	<b>8.6</b>	91	<b>6.8</b>	106	<b>7.7</b>	99
c.904G>A, p.(Gly302Arg)	Breast cancer (41)	<b>10.0</b>	71	<b>8.1</b>	61	<b>6.1</b>	76	<b>7.1</b>	69
c.910A>G, p.(Thr304Ala)	Teratocarcinoma (28)	<b>8.3</b>	78	<b>7.1</b>	69	<b>6.5</b>	90	<b>6.8</b>	80
c.1010G>A, p.(Arg337His)	ACC (2)	<b>5.8</b>	81	<b>6.7</b>	84	<b>6.0</b>	80	<b>6.4</b>	82

<sup>a</sup>For each individual, the *TP53* variant is indicated: cDNA numbering is given with the first nucleotide corresponding to the A of the ATG translation initiation codon in the reference sequence (GenBank RefSeq-file accession number NM\_000546.5) and protein numbering with the initiation codon numbered as codon 1 (accession number NP\_000537.3).<sup>b</sup>Age at diagnosis is indicated in years, if not specified. ACC: Adrenocortical carcinoma; Bilat.: bilateral; RMS: rhabdomyosarcoma.