

Supplemental Data

A Clinical Scoring System for Selection of Patients for PTEN Mutation Testing Is Proposed on the Basis of a Prospective Study of 3042 Probands

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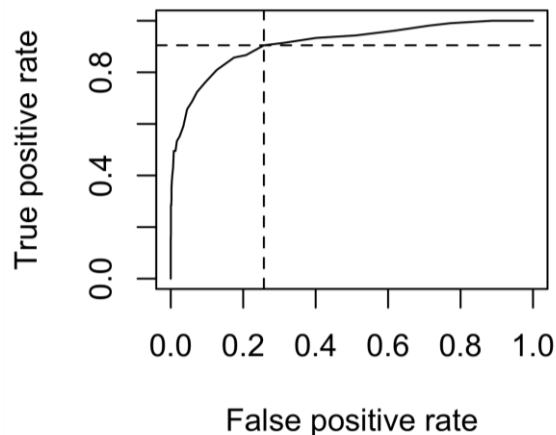


Figure S1. ROC Curve Showing the Performance of the Cleveland Clinic (CC) Score in the CC Data Set

The curve demonstrates true positive rate (sensitivity) varying with false positive rate (1-specificity) at various thresholds of CC score. The interrupted lines represent corresponding values at a threshold CC score of 10.

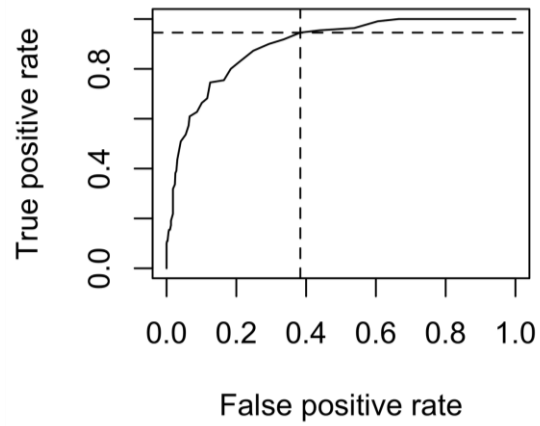


Figure S2. ROC Curve Showing the Performance of the Cleveland Clinic (CC) Score in the Independent Ohio State University Data Set

The curve demonstrates true positive rate (sensitivity) varying with false positive rate (1-specificity) at various thresholds of CC score. The interrupted lines represent corresponding values at a threshold CC score of 10.

Table S1. *PTEN* Primers Used in Light Scanner (LS) and Sequencing (Seq)

Name	Sequence 5' -> 3'	Annealing Temp (°C)
PTEN LS E1F PTEN LS E1R	GCAGCTTCTGCCATCTC GCATCCGTCTACTCCCAC	60
PTEN LS E2F PTEN LS E2R	AGTATTCTTTTAGTTTGATTGCTGC CTAAATGAAAACACAACATGAATATAAAC A	60
PTEN LS E3F PTEN LS E3R	ATGTTAGCTCATTTTTGTTAATGGTG CAAGCAGATAACTTTCACTTAATAGTTG	60
PTEN LS E4F PTEN LS E4R	TTTTTCTTCCTAAGTGCAAAGATAAC CAGTAAGATACAGTCTATCGGGT	60
PTEN LS E5F PTEN LS E5R PTEN LS E5SeqF	ACCTACTTGTTAATTAATAAATTCAAGAGT T ATCCAGGAAGAGGAAAGGAAA TGCAACATTTCTAAAGTTACCTACTTG	60 For sequencing
PTEN LS E6F PTEN LS E6R	CCCAGTTACCATAGCAATTTAGTGA TAGATATGGTTAAGAAAAGTGTCCAATA C	60
PTEN LS E7F PTEN LS E7R	CAGTTTGACAGTTAAAGGCATTTT AATATAGCTTTTAATCTGTCCTTATTTTG G	61
PTEN LS E8.1F PTEN LS E8.1R	TTTGTTGACTTTTTGCAAATGTTTAACAT A ATTTCTTGATCACATAGACTTCCA	61
PTEN LS E8.2F PTEN LS E8.2R	GTAATACATTCTTCATACCAGGACC GCTGTA CTCTAGAATTAACACAC	61
PTEN LS E9F PTEN LS E9R PTEN LS E9SeqF PTEN LS E9SeqR	AAGATGAGTCATATTTGTGGGTT TTTCAGTTTATTCAAGTTTATTTTCATGG AGATGAGTCATATTTGTGGGTTTT AAAGGTCCATTTTCAGTTTATTCAA	61 For sequencing For sequencing
PTEN LS SNP E8.2F PTEN LS SNP E8.2R PTEN LS SNP E8.2 Probe PTEN-Promoter1F PTEN-Promoter1R	GCAAATAAAGACAAAGCCAACCGA AGCTGTACTCCTAGAATTAACACACAT C CATACAAGTCACCAACCCCCAC-block GCGTGGTCACCTGGTCCTTT GCTGCTCACAGGCGCTGA	55 For Intronic 8 SNP 62 62

Table S2. Pathogenic Mutations Identified in 290 Probands
(HG19 BUILD, NM_000314.4)

	Age	Variant Nucleotide	Variant Function	Variant Protein
1	49.7	c.592_601del10	Deletion	p.(Met198Lysfs*20)
2	59.6	c.302delT	Deletion	p.(Ile101Thrfs*12)
3	48.5	c.48_49insA	Insertion	p.(Gln17Thrfs*27)
4	20.9	c.640C>T	Truncated protein	p.(Gln214*)
5	7.93	?_801+?del	LargeDel	
6	46.3	c.686C>G	Truncated protein	p.(Ser229*)
7	60.2	c.388C>T	Truncated protein	p.(Arg130*)
8	56.6	c.388C>T	Truncated protein	p.(Arg130*)
9	63.7	c.395G>A	Amino acid change	p.(Gly132Asp)
10	37.9	c.634+2T>C	Interference with splice junction	
11	18	c.461delT	Deletion	p.(Phe154Serfs*5)
12	56.9	c.406T>C	Amino acid change	p.(Cys136Arg)
13	35.8	c.635-1G>C	Interference with splice junction	
14	40.2	c.103A>G	Amino acid change	p.(Met35Val)
15	56.5	c.388C>T	Truncated protein	p.(Arg130*)
16	58.2	c.-1195del12	Promoter	
17	51.1	c.470_471insG	Insertion	p.(Val158Serfs*22)
18	8.54	c.188_189insAA	Insertion	p.(Asn63Lysfs*37)
19	53.5	c.210-1G>A	Interference with splice junction	
20	20.7	c.284C>T	Amino acid change	p.(Pro95Leu)
21	10.8	c.416T>G	Truncated protein	p.(Leu139*)
22	20	c.800delA	Deletion	p.(Lys267Argfs*9)
23	43.8	c.144C>A	Amino acid change	p.(Asn48Lys)
24	32.5	c.287C>G	Amino acid change	p.(Pro96Arg)
25	62.5	c.-1170C>T	Promoter	
26	8.25	c.1-?_79+?del	LargeDel	
27	31.7	c.1003C>T	Truncated protein	p.(Arg335*)
28	6.2	c.1003C>T	Truncated protein	p.(Arg335*)
29	51.1	c.884delT	Deletion	p.(Leu295Hisfs*12)
30	8.77	c.406T>C	Amino acid change	p.(Cys136Arg)
31	48.5	c.1027-2A>C	Interference with splice junction	
32	43.8	c.389G>A	Amino acid change	p.(Arg130Gln)
33	9.1	c.370T>C	Amino acid change	p.(Cys124Arg)
34	7.28	?_1212+?del	LargeDel	
35	61.4	c.253+1G>A	Interference with splice junction	
36	63.1	c.203A>G	Amino acid change	p.(Tyr68Cys)
37	31.3	c.491delA	Deletion	p.(Lys164Argfs*3)
38	53.6	c.1003C>T	Truncated protein	p.(Arg335*)
39	47.7	c.388C>G	Amino acid change	p.(Arg130Gly)
40	23.5	c.202T>C	Amino acid change	p.(Tyr68His)
41	70.3	c.1019delA	Deletion	p.(Asn340Ilefs*4)
42	11	c.697C>T	Truncated protein	p.(Arg233*)
43	78.3	c.633C>A	Truncated protein	p.(Cys211*)

44	45.7	c.542delT	Deletion	p.(Leu181Argfs*2)
45	51.2	c.388C>T	Truncated protein	p.(Arg130*)
46	61.1	c.493G>C	Amino acid change	p.(Gly165Arg)
47	18.7	c.407G>A	Amino acid change	p.(Cys136Tyr)
48	34.2	c.870delA	Deletion	p.(Glu291Lysfs*16)
49	47.1	c.-1170C>T	Promoter	
50	36.7	c.1003C>T	Truncated protein	p.(Arg335*)
51	8.31	c.697C>T	Truncated protein	p.(Arg233*)
52	52.5	c.18_19delAG	Deletion	p.(Glu7Aspfs*3)
53	77.6	c.359C>A	Amino acid change	p.(Ala120Glu)
54	8.7	c.127G>A	Amino acid change	p.(Glu43Lys)
55	7.02	c.44ins16	Insertion	
56	52.5	c.1066A>G	Amino acid change	p.(Asn356Asp)
57	63	c.613A>G	Amino acid change	p.(Met205Val)
58	68.6	c.388C>T	Truncated protein	p.(Arg130*)
59	38.9	c.1026+1G>C	Interference with splice junction	
60	58	c.150delT	Deletion	p.(Ile50Metfs*4)
61	14.5	c.697C>T	Truncated protein	p.(Arg233*)
62	50.4	?_164+?del	LargeDel	
63	45	-834C>T	Promoter	
64	27.4	c.388C>T	Truncated protein	p.(Arg130*)
65	9.25	c.1003C>T	Truncated protein	p.(Arg335*)
66	73.7	c.1003C>T	Truncated protein	p.(Arg335*)
67	5.78	c.18_19delAG	Deletion	p.(Glu7Aspfs*3)
68	64.6	c.209+1G>T	Interference with splice junction	
69	51.1	c.1061C>A	Amino acid change	p.(Pro354Gln)
70	6.33	?_164+?del	LargeDel	
71	51.8	c.720C>A	Truncated protein	p.(Tyr240*)
72	38.9	c.1003C>T	Truncated protein	p.(Arg335*)
73	56	c.328C>T	Truncated protein	p.(Gln110*)
74	16.9	c.892C>T	Truncated protein	p.(Gln298*)
75	7.78	c.491delA	Deletion	p.(Lys164Argfs*3)
76	45.8	c.633C>A	Truncated protein	p.(Cys211*)
77	58.2	c.1033C>G	Amino acid change	p.(Leu345Val)
78	40.6	c.406T>C	Amino acid change	p.(Cys136Arg)
79	52.2	c.182A>G	Amino acid change	p.(His61Arg)
80	15.7	c.1003C>T	Truncated protein	p.(Arg335*)
81	23.1	AAinsGA	InDel	p.(Asn323Metfs*21)
82	51.6	c.407_423del17	Deletion	p.(Cys136Serfs*38)
83	27.5	c.987_990del4	Deletion	p.(Asn329Lysfs*14)
84	22	c.586_587insT	Insertion	p.(His196Leufs*6)
85	30	c.406_407insA	Insertion	p.(Cys136*)
86	65.2	c.517C>T	Amino acid change	p.(Arg173Cys)
87	13.9	c.26delT	Deletion	p.(Ser10Alafs*14)
88	47.5	c.49C>T	Truncated protein	p.(Gln17*)
89	42.3	c.324delT	Deletion	p.(Asp109Thrfs*4)
90	30.5	c.41_43del3	Deletion	p.(Arg15del)

91	58.7	c.697C>T	Truncated protein	p.(Arg233*)
92	42.8	c.476G>C	Amino acid change	p.(Arg159Thr)
93	44	c.734_737del4	Deletion	p.(Gln245Argfs*10)
94	46.8	c.1003C>T	Truncated protein	p.(Arg335*)
95	59.8	c.388C>T	Truncated protein	p.(Arg130*)
96	12.3	c.1003C>T	Truncated protein	p.(Arg335*)
97	59.4	c.277C>T	Amino acid change	p.(His93Tyr)
98	56.3	c.388C>T	Truncated protein	p.(Arg130*)
99	19.4	c.512_513insA	Insertion	p.(Arg172Glu fs*8)
100	37.8	c.376G>C	Amino acid change	p.(Ala126Pro)
101	25.7	c.76A>C	Amino acid change	p.(Thr26Pro)
		c.609_611delT		
102	33.7	CCinsATAAAT	InDel	p.(Pro204*)
103	51.9	c.72_73ins3	Insertion	
104	59.1	c.-1195del12	Promoter	
105	54.6	c.542T>C	Amino acid change	p.(Leu181Pro)
106	39.6	c.181C>T	Amino acid change	p.(His61Tyr)
107	59	c.-834C>T	Promoter	
108	47.1	c.388C>T	Truncated protein	p.(Arg130*)
109	47.7	c.195_196insT	Insertion	p.(Lys66*)
110	42.8	c.389G>A	Amino acid change	p.(Arg130Gln)
111	48.5	c.389G>A	Amino acid change	p.(Arg130Gln)
112	4.7	c.302T>C	Amino acid change	p.(Ile101Thr)
113	48.8	c.235G>A	Amino acid change	p.(Ala79Thr)
114	3.64	c.737C>T	Amino acid change	p.(Pro246Leu)
115	46.1	c.697C>T	Truncated protein	p.(Arg233*)
116	24.5	c.382A>G	Amino acid change	p.(Lys128Glu)
117	5.14	c.486C>G	Amino acid change	p.(Asp162Glu)
118	9.62	c.75G>T	Amino acid change	p.(Leu25Phe)
119	57.7	c.71A>G	Amino acid change	p.(Asp24Gly)
120	45.4	c.475A>G	Amino acid change	p.(Arg159Gly)
121	7.69	c.204C>A	Truncated protein	p.(Tyr68*)
122	52.6	c.883_884insT	Insertion	p.(Cys296Met fs*2)
123	48.8	c.334_335insG	Insertion	p.(Leu112Arg fs*3)
124	65.4	c.70G>C	Amino acid change	p.(Asp24His)
125	21.8	c.1026+1G>A	Interference with splice junction	
126	72.8	c.389G>A	Amino acid change	p.(Arg130Gln)
127	42.8	c.388C>T	Truncated protein	p.(Arg130*)
128	22.5	c.509G>T	Amino acid change	p.(Ser170Ile)
129	4.98	c.562T>C	Amino acid change	p.(Tyr188His)
130	62.9	c.697C>T	Truncated protein	p.(Arg233*)
131	35.5	c.761A>C	Amino acid change	p.(Lys254Thr)
132	3.01	c.697C>T	Truncated protein	p.(Arg233*)
133	67.2	c.464A>G	Amino acid change	p.(Tyr155Cys)
134	29	c.635-1G>C	Interference with splice junction	
		c.177_179delA		
135	42	AA	Deletion	p.(Lys60del)
136	44.3	c.1026+1G>C	Interference with splice junction	
137	8.5	c.448G>T	Truncated protein	p.(Glu150*)
138	4.9	c.164+1-	Interference with splice junction	

		5delGTAAG c.872_873insAA		
139	11.4	AA	Insertion	p.(Asn292Lysfs*7)
140	12.6	c.513_514insA	Insertion	p.(Arg172Lysfs*8)
141	12.4	c.101C>A	Amino acid change	p.(Ala34Asp)
142	8.8	c.389G>A	Amino acid change	p.(Arg130Gln)
143	6.3	c.389G>A	Amino acid change	p.(Arg130Gln)
144	12.9	c.388C>T	Truncated protein	p.(Arg130*)
145	46.5	c.210-1G>A	Interference with splice junction	
146	47.8	c.1003C>T	Truncated protein	p.(Arg335*)
147	7.5	c.250A>T	Truncated protein	p.(Arg84*)
148	29.7	c.1003C>T	Truncated protein	p.(Arg335*)
149	55.4	c.513_514insA c.892_893insAA	Insertion	p.(Arg172Lysfs*8)
150	67.4	AA	Insertion	p.(Glu299Lysfs*5)
151	53	c.801+1G>A	Interference with splice junction	
152	29.5	c.801+1G>A	Interference with splice junction	
153	51.5	c.1-?_439+?del	LargeDel	
154	47.8	c.1-?_439+?del	LargeDel	
155	8.9	c.800delA	Deletion	p.(Lys267Argfs*9)
156	6.6	c.94_96delATT	Deletion	p.(Ile33del)
157	49.1	c.388C>G	Amino acid change	p.(Arg130Gly)
158	9	c.278A>G	Amino acid change	p.(His93Arg)
159	10.8	c.155A>G	Amino acid change	p.(Asp52Gly)
160	21.7	c.210-1G>A	Interference with splice junction	
161	42.1	c.315delT	Deletion	p.(Cys105Trpfs*8)
162	32.9	c.633C>A	Truncated protein	p.(Cys211*)
163	18.7	c.388C>T	Truncated protein	p.(Arg130*)
164	44	c.388C>T	Truncated protein	p.(Arg130*)
165	47	c.493G>C	Amino acid change	p.(Gly165Arg)
166	51.3	c.1003C>T c.459_461delTT	Truncated protein	p.(Arg335*)
167	43.6	T c.459_461delTT	Deletion	p.(Phe154del)
168	16	T	Deletion	p.(Phe154del)
169	12.1	c.104T>C	Amino acid change	p.(Met35Thr)
170	21.9	c.389G>A	Amino acid change	p.(Arg130Gln)
171	9.8	c.1003C>T	Truncated protein	p.(Arg335*)
172	41	c.176C>A	Truncated protein	p.(Ser59*)
173	41.7	c.1027-2A>C	Interference with splice junction	
174	35.4	c.445C>T c.1-	Truncated protein	p.(Gln149*)
175	14.2	?_1212+?del	LargeDel	
176	61.9	c.388C>T	Truncated protein	p.(Arg130*)
177	51.5	c.1033C>G c.155_157delAT	Amino acid change	p.(Leu345Val)
178	43	G c.955_958delA	Deletion	p.(Asp52del)
179	18.9	CTT	Deletion	p.(Thr319*)
180	60.1	c.389G>A	Amino acid change	p.(Arg130Gln)

181	16.9	c.316G>T	Truncated protein	p.(Glu106*)
182	15.3	c.316G>T	Truncated protein	p.(Glu106*)
183	44.1	c.316G>T	Truncated protein	p.(Glu106*)
184	49.3	c.957delT c.155_157delAT	Deletion	p.(Leu320*)
185	16.9	G	Deletion	p.(Asp52del)
186	45.7	c.634+1G>T	Interference with splice junction	
187	14	c.209+1G>A	Interference with splice junction	
188	48.2	c.1026+1G>A	Interference with splice junction	
189	4.1	c.388C>T	Truncated protein	p.(Arg130*)
190	53	c.527A>G	Amino acid change	p.(Tyr176Cys)
191	11.9	c.99delT c.69_74delAGA	Deletion	p.(Ile33Metfs*21)
192	59.7	CTT	Deletion	p.(Asp24_Leu25del)
193	31.2	c.634+2T>C	Interference with splice junction	
194	3.2	c.262T>C	Amino acid change	p.(Tyr88His)
195	47	c.741_742insA c.548_551delA	Insertion	p.(Pro248Thrfs*5)
196	55.4	GAA	Deletion	p.(Lys183Ilefs*15)
197	38.4	c.209+1G>A	Interference with splice junction	
198	18.3	c.640C>T	Truncated protein	p.(Gln214*)
199	9.2	c.895G>T	Truncated protein	p.(Glu299*)
200	46.5	c.463T>C	Amino acid change	p.(Tyr155His)
201	12	c.508A>C	Amino acid change	p.(Ser170Arg)
202	43	c.593_602del10 c.414_416delTT	Deletion	p.(Met198Lysfs*20)
203	56.7	T	Deletion	p.(Tyr138*)
204	47.4	c.959delT	Deletion	p.(Leu320*)
205	6.2	c.406T>C	Amino acid change	p.(Cys136Arg)
206	75.6	c.-853C>G	Promoter	
207	63.1	c.1003C>T	Truncated protein	p.(Arg335*)
208	5.4	c.697C>T	Truncated protein	p.(Arg233*)
209	36.1	c.395G>T	Amino acid change	p.(Gly132Val)
210	37.3	c.395G>T	Amino acid change	p.(Gly132Val)
211	39	c.48T>A	Truncated protein	p.(Tyr16*)
212	48.5	c.388C>T	Truncated protein	p.(Arg130*)
213	32	c.49C>T	Truncated protein	p.(Gln17*)
214	2.9	c.334C>G	Amino acid change	p.(Leu112Val)
215	45.2	c.1003C>T	Truncated protein	p.(Arg335*)
216	62.3	c.633C>A	Truncated protein	p.(Cys211*)
217	9.6	c.388C>T	Truncated protein	p.(Arg130*)
218	38.9	c.540C>G	Truncated protein	p.(Tyr180*)
219	49.6	c.-1177C>T	Promoter	
220	16.8	c.202T>C	Amino acid change	p.(Tyr68His)
221	38.1	c.389G>A	Amino acid change	p.(Arg130Gln)
222	39.7	c.18_22del5	Deletion	p.(Lys6Asnfs*3)
223	30.3	c.395G>T	Amino acid change	p.(Gly132Val)
224	4.2	c.755A>G	Amino acid change	p.(Asp252Gly)
225	57.3	c.1003C>T	Truncated protein	p.(Arg335*)
226	5.9	c.204C>A	Truncated protein	p.(Tyr68*)

227	46.8	c.389G>A c.706_708delG	Amino acid change	p.(Arg130Gln)
228	39.4	ACinsTTGT	InDel	p.(Asp236Leufs*2)
229	35.7	c.445_446insA	Insertion	p.(Glu150Argfs*30)
230	53.3	c.165-1G>A	Interference with splice junction	
231	49.8	c.49C>T	Truncated protein	p.(Gln17*)
232	38.9	c.742_743insA	Insertion	p.(Pro248Hisfs*5)
233	56.4	c.95T>A	Amino acid change	p.(Ile32Asn)
234	53.6	c.1003C>T	Truncated protein	p.(Arg335*)
235	15.7	c.388C>T	Truncated protein	p.(Arg130*)
236	5.1	c.722T>C c.956_959delC	Amino acid change	p.(Phe241Ser)
237	40.4	TTT c.597_599delG	Deletion	p.(Thr319Lysfs*24) p.(Met199_Phe200delinsIle)
238	14.9	TT	Deletion)
239	3.6	c.455T>C	Amino acid change	p.(Leu152Pro)
240	61.3	c.733C>T	Truncated protein	p.(Gln245*)
241	3.4	c.549_550insA	Insertion	p.(Asn184Lysfs*6)
242	44.1	c.407G>A	Amino acid change	p.(Cys136Tyr)
243	3.3	c.209+1insT	Interference with splice junction	
244	42.4	c.445_446insA	Insertion	p.(Glu150Argfs*30)
245	34.8	c.633C>A	Truncated protein	p.(Cys211*)
246	28.3	c.49C>T	Truncated protein	p.(Gln17*)
247	27.6	c.1026+1G>A	Interference with splice junction	
248	1.7	c.1026+1G>A	Interference with splice junction	
249	32.1	c.1003C>T	Truncated protein	p.(Arg335*)
250	48.9	c.655C>T	Truncated protein	p.(Gln219*)
251	50.5	c.388C>T	Truncated protein	p.(Arg130*)
252	46.7	c.388C>T	Truncated protein	p.(Arg130*)
253	32.1	c.405_406insA	Insertion	p.(Cys136Metfs*44)
254	3	c.127G>A	Amino acid change	p.(Glu43Lys)
255	52.8	c.404T>A	Amino acid change	p.(Ile135Lys)
256	57.3	c.872_873insA	Insertion	p.(Asn292Lysfs*6)
257	36.8	c.71A>G	Amino acid change	p.(Asp24Gly)
258	6.7	c.71A>G	Amino acid change	p.(Asp24Gly)
259	23.4	c.655C>T	Truncated protein	p.(Gln219*)
260	46.9	c.697C>T	Truncated protein	p.(Arg233*)
261	49.7	c.165-2A>G	Interference with splice junction	
262	31.1	c.195C>G	Truncated protein	p.(Tyr65*)
263	2.3	c.372T>G	Amino acid change	p.(Cys124Trp)
264	29.7	c.48T>A	Truncated protein	p.(Tyr16*)
265	19	c.76A>C	Amino acid change	p.(Thr26Pro)
266	49.5	c.388C>T	Truncated protein	p.(Arg130*)
267	42.9	c.846_855del10	Deletion	p.(Glu284Glnfs*4)
268	45.2	c.407G>A	Amino acid change	p.(Cys136Tyr)
269	54.5	c.545T>C	Truncated protein	p.(Leu182Ser)
270	49.8	c.1003C>T	Truncated protein	p.(Arg335*)
271	49.9	c.388C>T	Truncated protein	p.(Arg130*)
272	22.9	c.801+1G>T	Interference with splice junction	
273	37.4	c.1003C>T	Truncated protein	p.(Arg335*)

274	45.4	c.389G>T	Amino acid change	p.(Arg130Leu)
275	53.5	c.697C>T	Truncated protein	p.(Arg233*)
276	32.7	c.335T>C	Amino acid change	p.(Leu112Pro)
277	45.1	c.72_73insAGA	Insertion	p.(Asp24_Leu25insArg)
278	4.1	c.697C>T	Truncated protein	p.(Arg233*)
279	28.8	c.389G>T	Amino acid change	p.(Arg130Leu)
280	4.2	c.332G>A	Truncated protein	p.(Trp111*)
		c.741_742insC		
281	43.6	G	Insertion	p.(Pro248Argfs*9)
282	73.1	c.1026+1G>A	Interference with splice junction	
283	33.1	c.502delA	Deletion	p.(Ile168Phefs*15)
284	51.3	c.96delT	Deletion	p.(Ile33Leufs*21)
285	8.9	c.1003C>T	Truncated protein	p.(Arg335*)
286	25.8	c.287C>G	Amino acid change	p.(Pro96Arg)
287	5.2	c.640C>T	Truncated protein	p.(Gln214*)
288	20.1	c.211delT	Deletion	p.(Cys71Valfs*28)
289	39.6	c.686C>G	Truncated protein	p.(Ser229*)
290	26.7	c.371G>C	Amino acid change	p.(Cys124Ser)