

All SNPs - Renin

#SNP	Seq pos	Provean	PPH2	PPH2 prob	PhD-SNP	PhD-SNP RI	FATHMM	FATHMM score	HUMA (Disease associated?)
G3E	3	0,152	benign	0	Neutral	5	TOLERATED	0,02	
R5G	5	-0,357	benign	0,002	Neutral	4	TOLERATED	0,09	
R6K	6	-0,367	benign	0	Neutral	7	TOLERATED	0,08	
M7L	7	-1,01	benign	0,001	Disease	2	TOLERATED	0,31	
P8A	8	-1,375	benign	0,003	Disease	1	TOLERATED	0,22	
R9H	9	-0,631	benign	0,008	Neutral	5	TOLERATED	0,08	
L16R	16	-1,851	possibly damaging	0,754	Disease	6	TOLERATED	-0,08	HYPERURICEMIC NEPHROPATHY
L24F	24	-0,968	benign	0,326	Neutral	1	TOLERATED	-0,07	
P25L	25	-3,037	benign	0,186	Disease	4	TOLERATED	0,15	
T28N	28	-0,793	possibly damaging	0,514	Neutral	5	TOLERATED	0,13	
T29N	29	0,259	benign	0,05	Neutral	7	TOLERATED	0,13	
K32T	32	-1,199	benign	0,053	Neutral	9	TOLERATED	0,45	
R33Q	33	-2,95	probably damaging	1	Disease	7	TOLERATED	0,1	
R33W	33	-5,675	probably damaging	1	Disease	8	TOLERATED	-0,04	
R43*	43	None	None	None	None	None			RENAL TUBULAR DYSGENESIS
E48D	48	-1,605	benign	0,364	Disease	3	TOLERATED	0,06	
R49*	49	None	None	None	None	None			RENAL TUBULAR DYSGENESIS
R49Q	49	-0,701	possibly damaging	0,806	Neutral	3	TOLERATED	1,02	
D52H	52	-1,409	benign	0,066	Neutral	2	TOLERATED	0,91	
R55T	55	-1,6	benign	0,409	Disease	1	TOLERATED	0,97	
G57S	57	-0,146	benign	0,004	Disease	2	TOLERATED	0,43	
E59K	59	-2,02	benign	0,021	Neutral	3	TOLERATED	0,44	
W60R	60	-2,435	benign	0	Neutral	3	TOLERATED	0,49	
R66M	66	-1,23	benign	0,032	Neutral	5	TOLERATED	0,32	
G70D	70	-1,986	benign	0	Disease	4	TOLERATED	0,38	
N71D	71	-3,045	benign	0	Neutral	5	TOLERATED	0,44	
T72N	72	0,744	benign	0	Neutral	5	TOLERATED	0,47	
T73N	73	-3,081	benign	0,081	Neutral	2	TOLERATED	0,41	
V76M	76	-1,191	possibly damaging	0,894	Disease	2	TOLERATED	0,27	
I77M	77	-1,1	benign	0,134	Disease	1	TOLERATED	0,37	
T79S	79	-2,884	possibly damaging	0,459	Neutral	8	TOLERATED	0,42	
M82K	82	-2,574	benign	0,01	Disease	6	TOLERATED	0,41	
T84N	84	-3,51	possibly damaging	0,459	Neutral	2	TOLERATED	0,31	
Y86C	86	-8,365	probably damaging	1	Disease	7	TOLERATED	-1,35	
Y87H	87	-4,34	probably damaging	1	Disease	4	TOLERATED	0,24	
E89K	89	-3,58	probably damaging	0,997	Disease	3	TOLERATED	0,33	
I90T	90	-4,618	probably damaging	1	Disease	5	TOLERATED	-0,21	
P96H	96	-4,923	probably damaging	1	Disease	6	TOLERATED	0,05	
V101I	101	-0,931	probably damaging	0,997	Neutral	1	TOLERATED	-0,17	
D104N	104	-4,653	probably damaging	1	Disease	6	DAMAGING	-1,83	RENAL TUBULAR DYSGENESIS
S108A	108	0,941	benign	0,085	Disease	1	TOLERATED	-0,09	
R119H	119	-0,296	possibly damaging	0,915	Neutral	7	TOLERATED	0,4	
R119C	119	-1,058	probably damaging	0,963	Neutral	7	TOLERATED	0,4	
T122A	122	-1,738	benign	0	Neutral	6	TOLERATED	0,41	
Y126C	126	-0,57	possibly damaging	0,506	Neutral	5	TOLERATED	0,39	
H127R	127	-7,29	probably damaging	0,984	Disease	5	TOLERATED	0,29	
L129F	129	-1,672	benign	0,269	Neutral	0	TOLERATED	0,32	
S133L	133	-4,375	benign	0,023	Disease	2	TOLERATED	0,18	
S135Y	135	-5,549	probably damaging	1	Disease	8	TOLERATED	-1,46	RENAL TUBULAR DYSGENESIS
S136P	136	-2,991	benign	0,054	Disease	7	TOLERATED	-0,19	
K139*	139	None	None	None	None	None			
N141S	141	-4,334	benign	0,035	Disease	2	TOLERATED	0,35	
E144Q	144	-1,037	benign	0,002	Neutral	7	TOLERATED	0,34	
T146P	146	-2,197	benign	0,001	Disease	7	TOLERATED	0,33	
R148C	148	-3,89	possibly damaging	0,852	Neutral	4	TOLERATED	0,32	
R148H	148	-0,157	benign	0	Neutral	5	TOLERATED	0,38	
Q160K	160	-3,211	benign	0,005	Disease	7	TOLERATED	0,38	
Q160R	160	-3,329	benign	0,003	Disease	2	TOLERATED	0,3	
D161G	161	-6,432	possibly damaging	0,854	Disease	5	TOLERATED	-0,92	
I162T	162	-1,852	benign	0	Neutral	8	TOLERATED	0,74	
T164N	164	-3,169	benign	0,005	Neutral	4	TOLERATED	0,23	
G167E	167	-3,792	probably damaging	0,986	Disease	5	TOLERATED	0,08	
T169M	169	-2,077	possibly damaging	0,87	Disease	6	TOLERATED	0,23	
T178M	178	-4,434	probably damaging	0,996	Neutral	4	TOLERATED	0,18	
M180T	180	-2,734	benign	0,062	Neutral	2	TOLERATED	0,45	
M186I	186	-0,053	benign	0	Neutral	4	TOLERATED	0,45	
A188V	188	-3,581	probably damaging	1	Disease	6	TOLERATED	0,22	
E189K	189	2,188	benign	0,005	Neutral	6	TOLERATED	0,48	
G195D	195	-6,716	probably damaging	1	Disease	7	DAMAGING	-3,3	
I199T	199	1,606	benign	0	Neutral	2	TOLERATED	0,51	
A202T	202	-2,822	probably damaging	1	Disease	6	TOLERATED	0,31	
P208S	208	-7,728	probably damaging	1	Disease	3	TOLERATED	0,24	

#SNP	Seq pos	Provean	PPH2	PPH2 prob	PhD-SNP	PhD-SNP RI	FATHMM	FATHMM score	HUMA (Disease associated?)
N212S	212	-2,526	benign	0,451	Disease	8	TOLERATED	0,32	
I214V	214	-0,601	benign	0,038	Neutral	9	TOLERATED	0,48	
S215Y	215	-4,842	possibly damaging	0,645	Disease	1	TOLERATED	0,23	
G217R	217	0,792	benign	0,077	Disease	2	TOLERATED	-0,09	
V218L	218	-1,833	benign	0	Neutral	6	TOLERATED	0,57	
R230K	230	-2,719	possibly damaging	0,576	Disease	4	TOLERATED	0,35	RENAL TUBULAR DYSGENESIS
D231N	231	0,018	benign	0	Neutral	1	TOLERATED	0,41	
D231V	231	-3,273	benign	0,058	Disease	7	TOLERATED	0,25	
D231E	231	-0,895	benign	0,022	Disease	2	TOLERATED	0,41	
E233K	233	-0,146	benign	0,001	Disease	4	TOLERATED	0,38	
Q236P	236	2,153	benign	0,002	Disease	1	TOLERATED	0,05	
S237L	237	0,704	benign	0,262	Disease	2	TOLERATED	0,33	
G240R	240	-6,699	probably damaging	1	Disease	8	TOLERATED	-0,57	
Y252H	252	-4,061	probably damaging	0,998	Disease	5	TOLERATED	0,08	
E253K	253	-0,29	benign	0,114	Neutral	0	TOLERATED	0,48	
F256L	256	-5,042	possibly damaging	0,719	Neutral	5	TOLERATED	0,68	
H257R	257	-5,386	probably damaging	0,991	Disease	3	TOLERATED	0,41	
H257Q	257	-5,123	probably damaging	0,971	Disease	1	TOLERATED	0,42	
Y258F	258	-3,395	possibly damaging	0,935	Disease	7	TOLERATED	0,25	
I262T	262	2,221	benign	0,007	Neutral	8	TOLERATED	0,56	
K263R	263	-0,452	benign	0	Neutral	3	TOLERATED	0,47	
M271T	271	-4,788	probably damaging	0,998	Disease	4	TOLERATED	0,29	
G273R	273	-6,483	probably damaging	1	Disease	3	TOLERATED	0,17	
V274L	274	-2,305	benign	0,36	Disease	2	TOLERATED	0,22	
L281F	281	-3,146	benign	0,067	Neutral	2	TOLERATED	0,22	
L282F	282	1,325	benign	0,009	Neutral	7	TOLERATED	0,44	
G286S	286	-5,686	probably damaging	1	Disease	9	TOLERATED	0,27	
T293N	293	-4,729	probably damaging	1	Disease	2	TOLERATED	-0,45	
T302I	302	-2,751	probably damaging	0,96	Neutral	4	TOLERATED	0,27	
E306K	306	0,588	benign	0	Disease	1	TOLERATED	0,38	
L308I	308	-1,523	benign	0,03	Neutral	4	TOLERATED	0,57	
L318R	318	0,957	benign	0	Disease	3	TOLERATED	0,44	
F319V	319	0,509	benign	0,009	Disease	3	TOLERATED	0,42	
D320H	320	-2,581	possibly damaging	0,916	Disease	1	TOLERATED	0,37	
Y321*	321	None	None	None	None	None			
V323M	323	-2,757	probably damaging	1	Disease	5	TOLERATED	0,02	
K324R	324	-0,901	benign	0,317	Neutral	7	TOLERATED	0,4	
N326D	326	0,498	benign	0	Neutral	5	TOLERATED	0,47	
D333N	333	-1,084	benign	0,384	Neutral	2	TOLERATED	0,39	
H337Y	337	-4,252	benign	0,04	Disease	1	TOLERATED	0,38	
G340V	340	-8,006	probably damaging	1	Disease	6	TOLERATED	-0,23	
T344M	344	-3,425	probably damaging	1	Disease	3	TOLERATED	0,27	
L345F	345	-3,82	probably damaging	1	Disease	6	TOLERATED	0,08	
F352I	352	1,384	benign	0,008	Neutral	2	TOLERATED	0,5	
E354G	354	-3,189	benign	0,15	Neutral	2	TOLERATED	0,3	
S358I	358	-3,823	possibly damaging	0,788	Neutral	1	TOLERATED	0,28	
L364P	364	-3,556	probably damaging	0,989	Disease	5	TOLERATED	0,1	
P372L	372	-8,028	probably damaging	0,983	Disease	6	TOLERATED	0,28	
P373L	373	-8,269	probably damaging	0,99	Disease	6	TOLERATED	0,28	
T375N	375	-2,552	benign	0,127	Neutral	7	TOLERATED	0,39	
T384A	384	-1,98	benign	0,448	Neutral	6	TOLERATED	0,42	
T384N	384	-1,948	benign	0,128	Disease	1	TOLERATED	0,39	
I386T	386	-4,668	probably damaging	0,971	Disease	4	TOLERATED	0,15	
R387Q	387	-1,243	probably damaging	0,987	Disease	5	TOLERATED	0,16	
R387*	387	None	None	None	None	None			
Y390H	390	-4,697	probably damaging	1	Neutral	2	TOLERATED	-0,18	
R395L	395	-6,282	probably damaging	0,986	Neutral	3	TOLERATED	0,54	
R396C	396	-5,15	possibly damaging	0,886	Neutral	3	TOLERATED	0,36	
N398D	398	-4,158	benign	0,356	Neutral	1	TOLERATED	0,36	
R399C	399	-7,247	probably damaging	1	Disease	6	TOLERATED	0,15	
R399H	399	-4,43	probably damaging	1	Disease	5	TOLERATED	0,17	
R406H	406	-1,982	probably damaging	1	Disease	5	TOLERATED	-0,11	
R406C	406	-2,183	probably damaging	1	Disease	7	TOLERATED	-0,15	

Damaging SNPs - Renin

#SNP	Seq pos	Provean	PPH2	PPH2 prob	PhD-SNP	PhD-SNP RI	FATHMM	FATHMM score	HUMA (Disease associated?)
L16R	16	-1,851	possibly damaging	0,754	Disease	6	TOLERATED	-0,08	<b>HYPERURICEMIC NEPHROPATHY</b>
R33Q	33	-2,95	probably damaging	1	Disease	7	TOLERATED	0,1	
R33W	33	-5,675	probably damaging	1	Disease	8	TOLERATED	-0,04	
R43*	43								<b>RENAL TUBULAR DYSGENESIS</b>
R49*	49								<b>RENAL TUBULAR DYSGENESIS</b>
Y86C	86	-8,365	probably damaging	1	Disease	7	TOLERATED	-1,35	
Y87H	87	-4,34	probably damaging	1	Disease	4	TOLERATED	0,24	
E89K	89	-3,58	probably damaging	0,997	Disease	3	TOLERATED	0,33	
I90T	90	-4,618	probably damaging	1	Disease	5	TOLERATED	-0,21	
P96H	96	-4,923	probably damaging	1	Disease	6	TOLERATED	0,05	
D104N	104	-4,653	probably damaging	1	Disease	6	<b>DAMAGING</b>	<b>-1,83</b>	<b>RENAL TUBULAR DYSGENESIS</b>
H127R	127	-7,29	probably damaging	0,984	Disease	5	TOLERATED	0,29	
S135Y	135	-5,549	probably damaging	1	Disease	8	TOLERATED	-1,46	<b>RENAL TUBULAR DYSGENESIS</b>
D161G	161	-6,432	possibly damaging	0,854	Disease	5	TOLERATED	-0,92	
G167E	167	-3,792	probably damaging	0,986	Disease	5	TOLERATED	0,08	
A188V	188	-3,581	probably damaging	1	Disease	6	TOLERATED	0,22	
G195D	195	-6,716	probably damaging	1	Disease	7	<b>DAMAGING</b>	<b>-3,3</b>	
A202T	202	-2,822	probably damaging	1	Disease	6	TOLERATED	0,31	
P208S	208	-7,728	probably damaging	1	Disease	3	TOLERATED	0,24	
S215Y	215	-4,842	possibly damaging	0,645	Disease	1	TOLERATED	0,23	
R230K	230	-2,719	possibly damaging	0,576	Disease	4	TOLERATED	0,35	<b>RENAL TUBULAR DYSGENESIS</b>
G240R	240	-6,699	probably damaging	1	Disease	8	TOLERATED	-0,57	
Y252H	252	-4,061	probably damaging	0,998	Disease	5	TOLERATED	0,08	
H257R	257	-5,386	probably damaging	0,991	Disease	3	TOLERATED	0,41	
H257Q	257	-5,123	probably damaging	0,971	Disease	1	TOLERATED	0,42	
Y258F	258	-3,395	possibly damaging	0,935	Disease	7	TOLERATED	0,25	
M271T	271	-4,788	probably damaging	0,998	Disease	4	TOLERATED	0,29	
G273R	273	-6,483	probably damaging	1	Disease	3	TOLERATED	0,17	
G286S	286	-5,686	probably damaging	1	Disease	9	TOLERATED	0,27	
T293N	293	-4,729	probably damaging	1	Disease	2	TOLERATED	-0,45	
D320H	320	-2,581	possibly damaging	0,916	Disease	1	TOLERATED	0,37	
V323M	323	-2,757	probably damaging	1	Disease	5	TOLERATED	0,02	
G340V	340	-8,006	probably damaging	1	Disease	6	TOLERATED	-0,23	
T344M	344	-3,425	probably damaging	1	Disease	3	TOLERATED	0,27	
L345F	345	-3,82	probably damaging	1	Disease	6	TOLERATED	0,08	
L364P	364	-3,556	probably damaging	0,989	Disease	5	TOLERATED	0,1	
P372L	372	-8,028	probably damaging	0,983	Disease	6	TOLERATED	0,28	
P373L	373	-8,269	probably damaging	0,99	Disease	6	TOLERATED	0,28	
I386T	386	-4,668	probably damaging	0,971	Disease	4	TOLERATED	0,15	
R399C	399	-7,247	probably damaging	1	Disease	6	TOLERATED	0,15	
R399H	399	-4,43	probably damaging	1	Disease	5	TOLERATED	0,17	

\* SNPs were predicted to be damaging if at least 3 out of the 4 tools agreed or they if they were associated with a disease in HUMA

Position	Position (PDB)	Residue
<b>82</b>	<b>9</b>	<b>MET</b>
85	12	GLN
102	29	VAL
<b>104</b>	<b>31</b>	<b>ASP</b>
106	33	GLY
<b>119</b>	<b>46</b>	<b>ARG</b>
120	47	LEU
121	48	TYR
<b>148</b>	<b>75</b>	<b>ARG</b>
149	76	TYR
150	77	SER
151	78	THR
153	80	THR
181	108	PRO
182	109	ALA
183	110	LEU
184	111	PRO
185	112	PHE
<b>186</b>	<b>113</b>	<b>MET</b>
187	114	LEU
<b>188</b>	<b>115</b>	<b>ALA</b>
190	117	PHE
193	120	VAL
203	130	ILE
290	217	LEU
292	219	ASP
295	222	ALA
296	223	SER
297	224	TYR
299	226	SER
<b>318</b>	<b>245</b>	<b>LEU</b>
<b>319</b>	<b>246</b>	<b>PHE</b>
<b>352</b>	<b>279</b>	<b>PHE</b>
363	290	THR
371	298	ILE
380	307	ALA

\* SNPs occur at positions highlighted in yellow

\*\* Damaging SNPs occur at positions highlighted in red

\*\*\* The "Position (PDB)" column refers to the numbering in the PDB file