

Gene										FATHMM		
ABCC8	Serial No.	Protein ID	Substitution	Prediction	Score	Warning HMM ID	HMM Description	HMM Pos.	HMM Prob. W.	HMM Prob. M.	HMM Weights D.	HMM Weights O.
	1	Q09428	L582V	DAMAGING	-2.87	ABC_membrane	ABC transporter transmembrane region	267	0.2894652569	0.0819218119	471	83
	2	Q09428	C435R	DAMAGING	-3.4	53060	ABC transporter transmembrane region	139	0.01018845564	0.014875347	382	36
	3	Q09428	H1023Y	DAMAGING	-3.4	53060	ABC transporter transmembrane region	27	0.01285143244	0.01976465574	382	36
	4	Q09428	R1182Q	DAMAGING	-2.52	ABC_membrane	ABC transporter transmembrane region	162	0.03925250559	0.02598593107	471	83
	5	Q09428	R1379C	DAMAGING	-3.4	ABC_tran	ABC transporter	19	0.09709843706	0.004470969516	547	57

Gene										FATHMM		
TCF7L2	Serial No.	Protein ID	Substitution	Prediction	Score	Warning HMM ID	HMM Description	HMM Pos.	HMM Prob. W.	HMM Prob. M.	HMM Weights D.	HMM Weights O.
	1	Q9NQB0	E322A	DAMAGING	-5.09	7989		322	0.205	0.074	88	3
	2	Q9NQB0	D16A	DAMAGING	-7.46	7989		16	0.835	0.012	88	3
	3	Q9NQB0	E17A	DAMAGING	-7.27	7989		17	0.812	0.014	88	3
	4	Q9NQB0	H19A	DAMAGING	-5.4	7989		19	0.329	0.038	88	3
	5	Q9NQB0	F21A	DAMAGING	-6.57	7989		21	0.693	0.079	88	3
	6	Q9NQB0	E24A	DAMAGING	-6.81	7989		24	0.743	0.019	88	3
	7	Q9NQB0	E26A	DAMAGING	-5.74	7989		26	0.464	0.025	88	3
	8	Q9NQB0	E28A	DAMAGING	-5.49	7989		28	0.375	0.039	88	3
	9	Q9NQB0	E29A	DAMAGING	-5.39	7989		29	0.328	0.04	88	3
	10	Q9NQB0	L48A	DAMAGING	-6.52	7989		48	0.687	0.021	88	3
	11	Q9NQB0	K320R	DAMAGING	-5.16	7989		320	0.235	0.065	88	3

Gene										FATHMM		
KCQN1	Serial No.	Protein ID	Substitution	Prediction	Score	Warning HMM ID	HMM Description	HMM Pos.	HMM Prob. W.	HMM Prob. M.	HMM Weights D.	HMM Weights O.
	1	P51787	S27A	DAMAGING	-5.64			27	0.139	0.084	375	8
	2	P51787	V324L	DAMAGING	-4.9	Ion_Trans	Ion Transport protein	176	0.119	0.121	1141	38
	3	P51787	T327V	DAMAGING	-4.78	Ion_trans	Ion Transport protein	179	0.0611	0.14	1141	38
	4	P51787	I328L	DAMAGING	-4.81	Ion_trans	Ion Transport protein	180	0.0694	0.13	1141	38
	5	P51787	F340C	DAMAGING	-5.08	Ion_trans	Ion Transport protein	193	0.12	0.0095	1141	38
	6	P51787	A590W	DAMAGING	-6.53	KCNQ_channel	KCNQ voltage K Chan	156	0.191	0.0027	75	1
	7	P51787	L602A	DAMAGING	-6.37	KCNQ_channel	KCNQ voltage K Chan	168	0.17	0.084	75	1
	8	P51787	I609A	DAMAGING	-6.49	KCNQ_channel	KCNQ voltage K Chan	175	0.193	0.0302	75	1

Prediction	Median Sequence	Score	Sequence
AFFECT	3.04	0	21
AFFECT	3.04	0.02	21
TOLERATED	3.04	0.33	21
TOLERATED	3.04	0.08	21
AFFECT	3.04	0	21

Prediction	Median Sequence	Score	Sequence
DAMAGING	3.51	0	23
DAMAGING	3.63	0	14
DAMAGING	3.63	0	14
DAMAGING	3.63	0	14
DAMAGING	3.63	0	14
DAMAGING	3.63	0	14
DAMAGING	3.63	0	14
DAMAGING	3.63	0	14
DAMAGING	3.63	0	14
DAMAGING	3.63	0	14
DAMAGING	3.63	0	14
DAMAGING	3.52	0	22

Prediction	Median Sequence	Score	Sequence
TOLERATED	4.32	0.06	4
TOLERATED	3.01	0.65	134
TOLERATED	3.01	0.33	134
TOLERATED	3.01	0.43	134
DAMAGING	3.01	0	134
DAMAGING	3.4	0	15
DAMAGING	3.4	0	15
DAMAGING	3.49	0.01	7

Description	Score	Sensitivity	Specificity
DAMAGING	0.995	0.68	0.97
BENIGN	0.311	0.9	0.89
BENIGN	0.274	0.91	0.88
BENIGN	0.098	0.93	0.85
DAMAGING	1	0	1

Description	Score	Sensitivity	Specificity
DAMAGING	0.997	0.71	0.97
DAMAGING	0.999	0.14	0.99
DAMAGING	0.999	0.14	0.99
DAMAGING	0.998	0.27	0.99
DAMAGING	0.998	0.27	0.99
DAMAGING	0.999	0.14	0.99
DAMAGING	0.996	0.55	0.98
DAMAGING	0.884	0.82	0.94
DAMAGING	0.969	0.77	0.95
DAMAGING	0.999	0.14	0.99
DAMAGING	0.991	0.71	0.97

Description	Score	Sensitivity	Specificity
DAMAGING	0.624	0.87	0.91
BENIGN	0.004	0.97	0.59
BENIGN	0.365	0.9	0.89
DAMAGING	0.78	0.85	0.93
DAMAGING	1	1	0
DAMAGING	0.997	0.41	0.98
BENIGN	0.059	0.94	0.84
DAMAGING	0.901	0.82	0.94