



12 SNVs with significant effects

Gene	Chr	Position	Ref	Alt	AA Change	Protein domain	Polyphen 2 score	Polyphen2 result	SIFT score	SIFT prediction
<i>FCN1</i>	25	36,824,050	G	T	Pro54Gln	CLD	1	probably damaging	0	deleterious
<i>MBL1</i>	1	88,935,539	G	A	Cys237Tyr	CRD	1	probably damaging	0.04	deleterious
<i>MASP1</i>	19	25,180,346	C	T	Ala137Thr	CUB domain	1	probably damaging	0	deleterious
<i>MBL1</i>	1	88,933,562	G	C	Gly82Ala	CLD	0.999	probably damaging	0	deleterious
<i>FCN3</i>	2	28,415,514	G	T	Gln123His	FBG	0.998	probably damaging	0	deleterious
<i>MASP1</i>	19	25,154,003	A	G	Ile463Thr	Peptidase S1 domain	0.926	possibly damaging	0.05	deleterious
<i>MBL1</i>	1	88,933,006	A	C	Thr36Pro	N-terminal domain	0.86	possibly damaging	0.05	tolerated
<i>FCN1</i>	25	36,824,033	C	T	Val60Met	CLD	0.859	possibly damaging	0.06	tolerated
<i>FCN1</i>	25	36,820,482	C	G	Asp149His	FBG	0.651	possibly damaging	0.18	tolerated
<i>FCN1</i>	25	36,824,026	G	A	Pro62Leu	CLD	0.539	possibly damaging	0.04	deleterious
<i>SFTPD</i>	1	88,950,438	G	A	Pro191Leu	CLD	0.483	possibly damaging	0.23	tolerated
<i>FCN1</i>	25	36,819,956	G	T	Ala174Glu	FBG	0.465	possibly damaging	0.04	deleterious
<i>COLEC10</i>	9	62,298,342	C	T	Thr193Met	CRD	0.284	benign	0.04	deleterious
<i>MASP2</i>	2	40,427,714	G	A	Lys90Arg	CUB domain	0.057	benign	0	deleterious





SNVs with significant effects

Gene	Chr	Position	Ref	Alt	Protein change	Protein domain	Polyphen2 Prediction	Polyphen 2 Score	SIFT Prediction	SIFT Score
<i>FCN1</i>	11	106,827,968	C	T	Arg142Cys	FBG	probably damaging	0.986	tolerated	0.06
<i>MBL2</i>	26	6,344,919	C	A	Pro42Gln	CLD	probably damaging	0.974	deleterious	0
<i>SFTPD</i>	28	35,820,078	C	T	Pro132Ser	CLD	probably damaging	0.958	deleterious	0.05
<i>CGN</i>	28	35,598,640	G	A	Arg173His	CLD	possibly damaging	0.72	tolerated	0.16
<i>CL46</i>	28	35,675,371	C	T	Pro185Leu	CLD	possibly damaging	0.672	tolerated	0.32
<i>CL43</i>	28	35,718,034	A	G	Thr117Ala	CLD	possibly damaging	0.659	tolerated	0.9
<i>CL43</i>	28	35,718,807	G	T	Gln185His	neck region	possibly damaging	0.634	deleterious	0.01
<i>MASP2</i>	16	43,463,621	G	A	Gly102Ser	CUB domain	benign	0.191	deleterious	0.01
<i>FCN1</i>	11	106,828,710	C	T	Thr193Met	FBG	benign	0.042	deleterious	0.03
<i>CGN</i>	28	35,602,463	A	C	Glu302Asp	CRD	benign	0.036	deleterious	0.04
<i>SFTPD</i>	28	35,824,136	C	G	Ala288Gly	CRD	benign	0.002	deleterious	0.02

12 SNVs with significant effects

Gene	Chr	Position	Ref	Alt	AA Change	Protein domain	Polyphen 2 score	Polyphen2 result	SIFT score	SIFT prediction
<i>FCN1</i>	25	36,824,050	G	T	Pro54Gln	CLD	1	probably damaging	0	deleterious
<i>MBL1</i>	1	88,935,539	G	A	Cys237Tyr	CRD	1	probably damaging	0.04	deleterious
<i>MASP1</i>	19	25,180,346	C	T	Ala137Thr	CUB domain	1	probably damaging	0	deleterious
<i>MBL1</i>	1	88,933,562	G	C	Gly82Ala	CLD	0.999	probably damaging	0	deleterious
<i>FCN3</i>	2	28,415,514	G	T	Gln123His	FBG	0.998	probably damaging	0	deleterious
<i>MASP1</i>	19	25,154,003	A	G	Ile463Thr	Peptidase S1 domain	0.926	possibly damaging	0.05	deleterious
<i>MBL1</i>	1	88,933,006	A	C	Thr36Pro	N-terminal domain	0.86	possibly damaging	0.05	tolerated
<i>FCN1</i>	25	36,824,033	C	T	Val60Met	CLD	0.859	possibly damaging	0.06	tolerated
<i>FCN1</i>	25	36,820,482	C	G	Asp149His	FBG	0.651	possibly damaging	0.18	tolerated
<i>FCN1</i>	25	36,824,026	G	A	Pro62Leu	CLD	0.539	possibly damaging	0.04	deleterious
<i>SFTPD</i>	1	88,950,438	G	A	Pro191Leu	CLD	0.483	possibly damaging	0.23	tolerated
<i>FCN1</i>	25	36,819,956	G	T	Ala174Glu	FBG	0.465	possibly damaging	0.04	deleterious
<i>COLEC10</i>	9	62,298,342	C	T	Thr193Met	CRD	0.284	benign	0.04	deleterious
<i>MASP2</i>	2	40,427,714	G	A	Lys90Arg	CUB domain	0.057	benign	0	deleterious