Missense mutations

- Polyphen2
  - Aligns homologues and identifies conserved AAs
  - 8 sequence based, 3 structure based features
  - Sens: 88 %
  - Spec: 62 %

- SIFT
  - Aligns homologues and identifies conserved AAs

  - Sens: 85 %
  - Spec: 69 %

12 SNVs with significant effects

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

Missense mutations

- Polyphen2
  - Aligns homologues and identifies conserved AAs
  - 8 sequence based, 3 structure based features
  - Sens: 88 %
  - Spec: 62 %

- SIFT
  - Aligns homologues and identifies conserved AAs

  - Sens: 85 %
  - Spec: 69 %