

AUTOMATED REPORTING

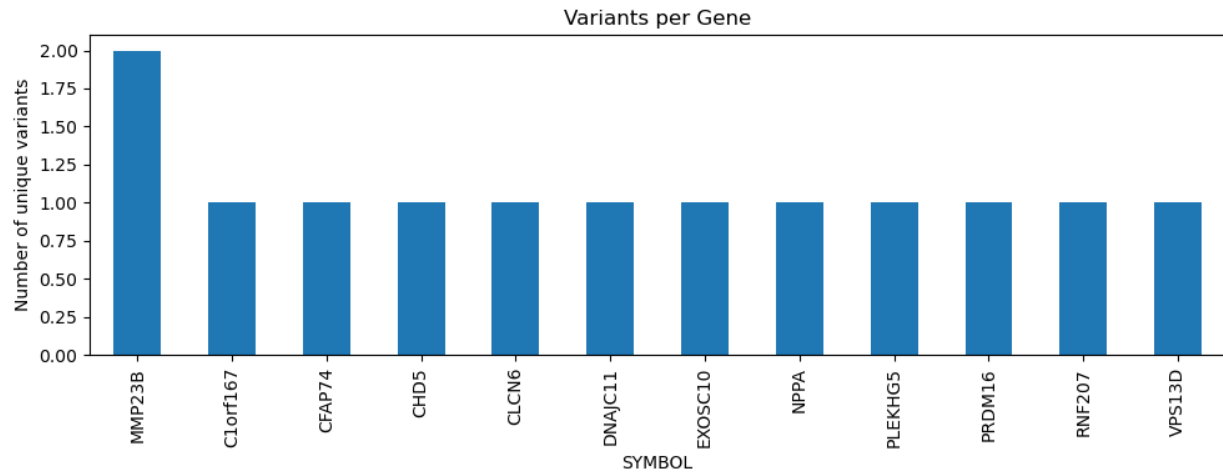


Fig 1: Variants per gene

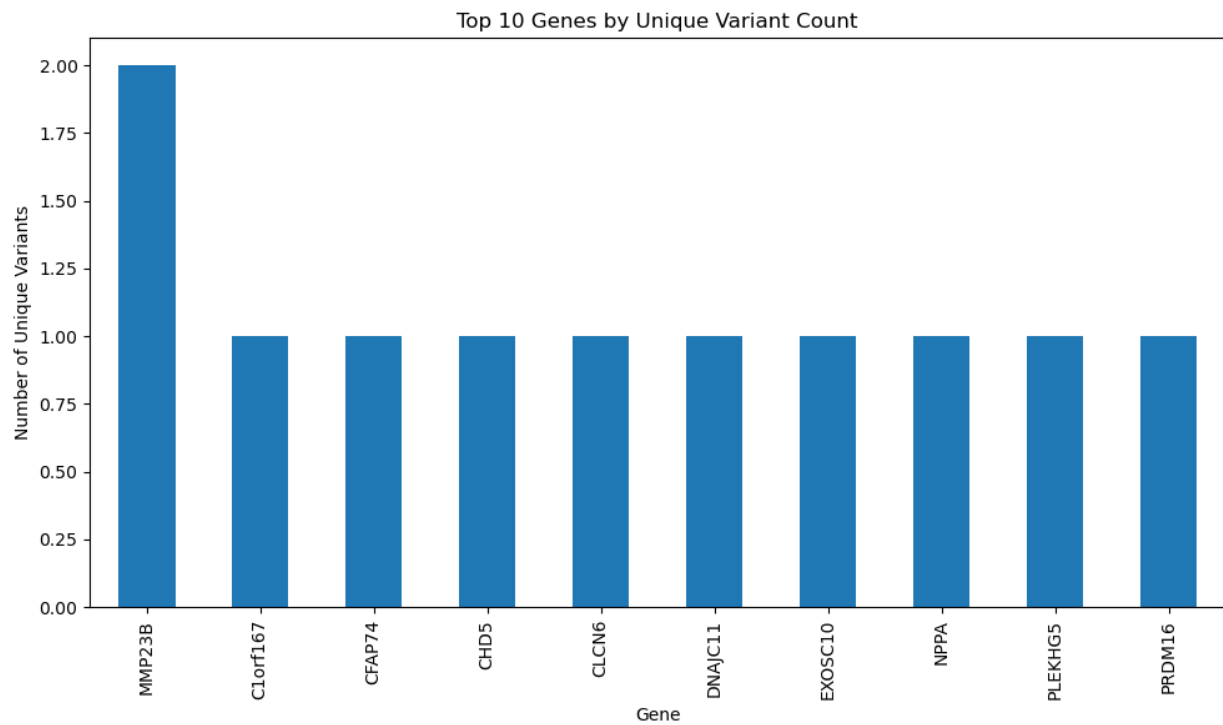


Fig 2: Top genes by Variant count

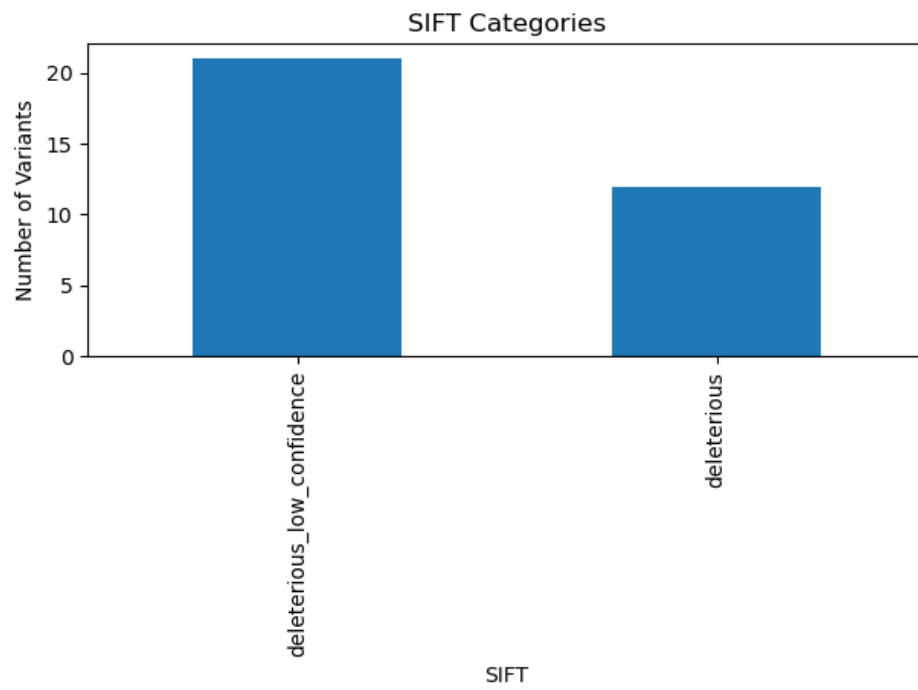


Fig 3: SIFT prediction categories

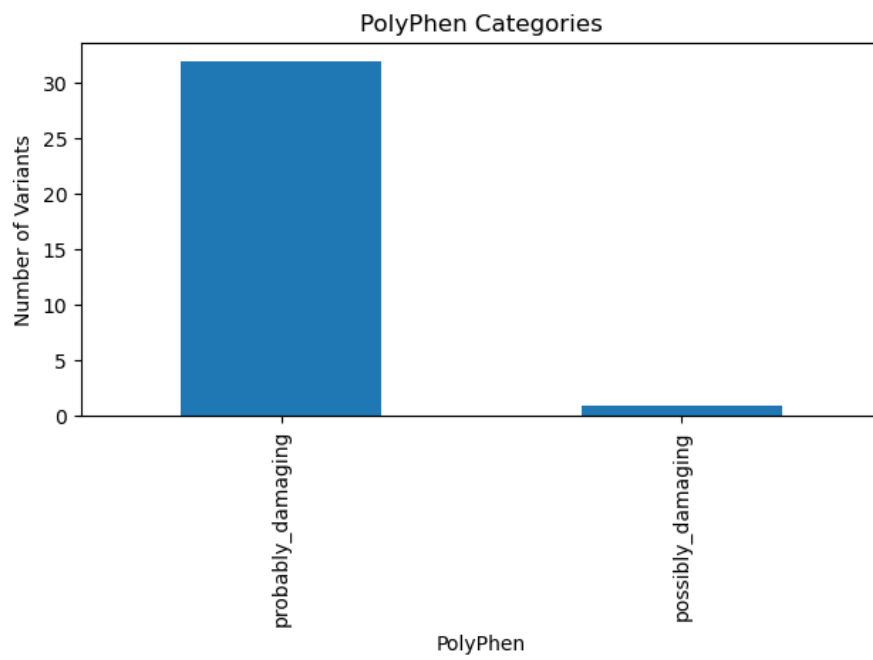


Fig 4: PolyPhen Prediction categories

Table 1: SRR18395025 Finalised variants

Variant	SYMBOL	Gene	Consequence	IMPACT	CLIN_SIG	SIFT	PolyPhen	AF	htg_varia	Location	Allele
1:1633004-1633004:C	MMP23B	ENSG00000189409	missense_variant	MODERATE	-	deleterious(0.02)	possibly_damaging(0.862)	-	-	1:1633004-1633004	C
1:1633004-1633004:C	MMP23B	ENSG00000189409	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.998)	-	-	1:1633004-1633004	C
1:1633007-1633007:C	MMP23B	ENSG00000189409	missense_variant	MODERATE	-	deleterious(0)	probably_damaging(0.951)	-	-	1:1633007-1633007	C
1:1633007-1633007:C	MMP23B	ENSG00000189409	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.998)	-	-	1:1633007-1633007	C
1:1922303-1922303:T	CFAP74	ENSG00000142609	missense_variant	MODERATE	-	deleterious(0)	probably_damaging(0.946)	-	-	1:1922303-1922303	T
1:1922303-1922303:T	CFAP74	ENSG00000142609	missense_variant	MODERATE	-	deleterious(0)	probably_damaging(0.928)	-	-	1:1922303-1922303	T
1:3417960-3417960:A	PRDM16	ENSG00000142611	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.992)	-	-	1:3417960-3417960	A
1:6142439-6142439:T	CHD5	ENSG00000116254	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.995)	-	-	1:6142439-6142439	T
1:6212408-6212408:A	RNF207	ENSG00000158286	missense_variant	MODERATE	-	deleterious(0)	probably_damaging(0.997)	-	-	1:6212408-6212408	A
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.993)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.997)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.993)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.993)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.993)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.997)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.993)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.993)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.997)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.993)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.997)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.993)	-	-	1:6473294-6473294	G
1:6473294-6473294:G	PLEKHG5	ENSG00000171680	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.993)	-	-	1:6473294-6473294	G
1:6645867-6645867:A	DNAJC11	ENSG00000007923	missense_variant	MODERATE	-	deleterious(0.02)	probably_damaging(0.974)	-	-	1:6645867-6645867	A
1:6645867-6645867:A	DNAJC11	ENSG00000007923	missense_variant	MODERATE	-	deleterious(0.02)	probably_damaging(0.97)	-	-	1:6645867-6645867	A
1:6645867-6645867:A	DNAJC11	ENSG00000007923	missense_variant	MODERATE	-	deleterious(0.02)	probably_damaging(0.999)	-	-	1:6645867-6645867	A
1:11087835-11087835:C	EXOSC10	ENSG00000171824	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(1)	-	-	1:11087835-11087835	C
1:11087835-11087835:C	EXOSC10	ENSG00000171824	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(1)	-	-	1:11087835-11087835	C
1:11766424-11766424:T	C1orf167	ENSG00000215910	missense_variant	MODERATE	-	deleterious(0.03)	probably_damaging(0.98)	-	-	1:11766424-11766424	T
1:11766424-11766424:T	C1orf167	ENSG00000215910	missense_variant	MODERATE	-	deleterious(0.02)	probably_damaging(0.995)	-	-	1:11766424-11766424	T
1:11838646-11838646:C	CLCN6	ENSG00000011021	missense_variant	MODERATE	-	deleterious(0)	probably_damaging(0.993)	-	-	1:11838646-11838646	C
1:11847436-11847436:C	NPPA	ENSG00000175206	missense_variant	MODERATE	-	deleterious(0)	probably_damaging(0.992)	-	-	1:11847436-11847436	C
1:12261972-12261972:G	VPS13D	ENSG00000048707	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.996)	-	-	1:12261972-12261972	G
1:12261972-12261972:G	VPS13D	ENSG00000048707	missense_variant	MODERATE	-	sterious_low_confidenc	probably_damaging(0.996)	-	-	1:12261972-12261972	G