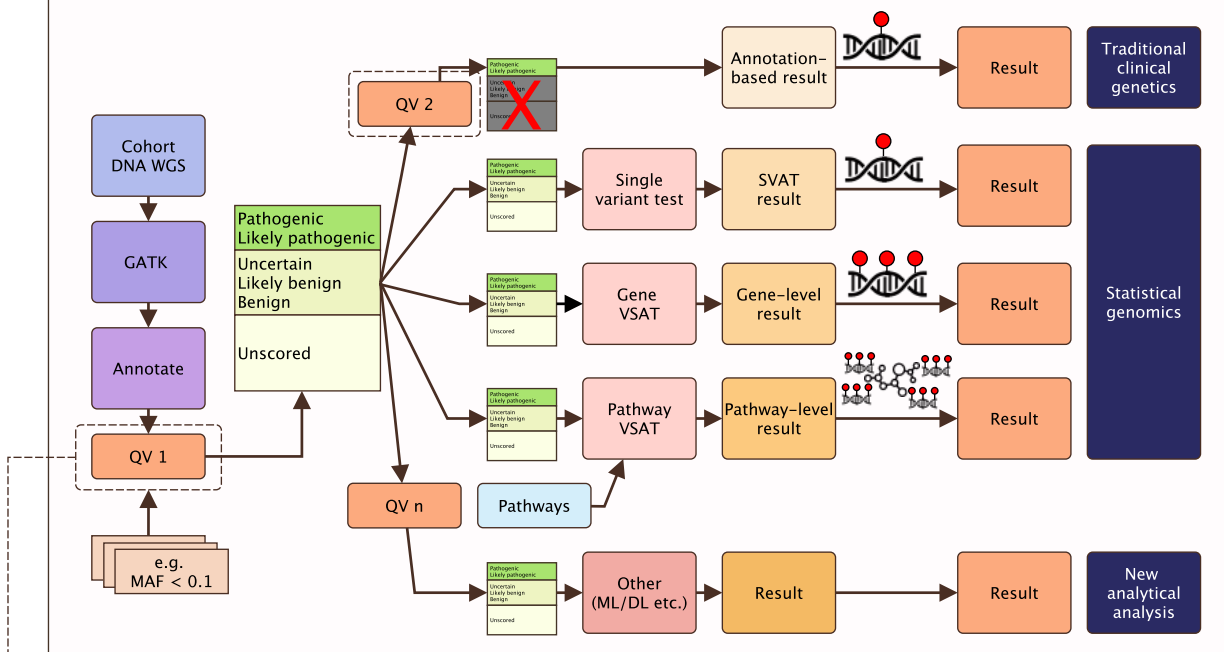
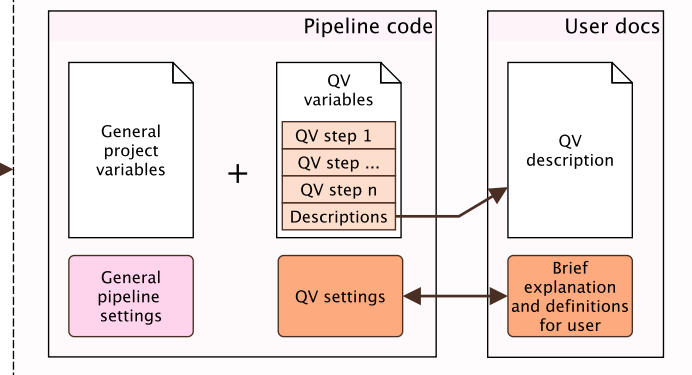


A

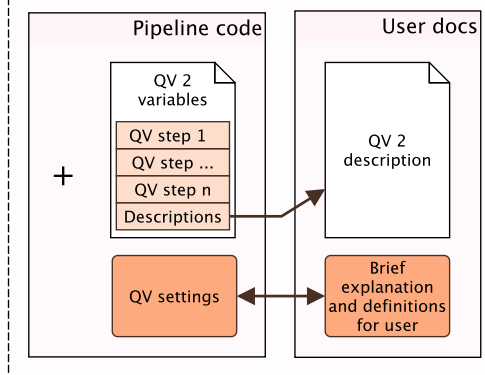
Pipeline: DNA SNV INDEL v1



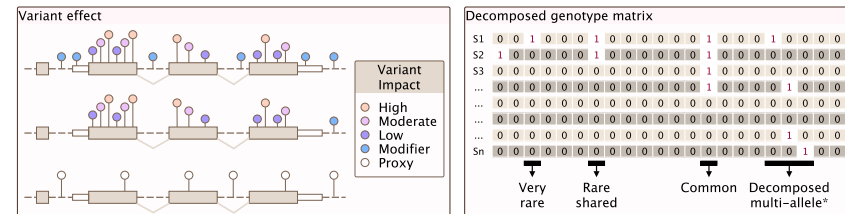
QV set in pipeline code (i.e. QV 1)



Additional QV sets (i.e. QV 2)



B



Variant	Set	Sample	Genotype	Age	Sex	Cohort AF	CADD phred	REVEL	ClinVar	FATHMM	OMIM	PANTHER	Gene ontology	...	GnomAD AF
1	A	S1	0	18	0	.3	25.90	.902	PL	.7	179615	11539	0002331	...	3E-05
1	A	...	0	21	1	.3	25.90	.902	PL	.7	179615	11539	0002331	...	3E-05
1	A	Sn	1	45	0	.3	25.90	.902	PL	.7	179615	11539	0002331	...	3E-05
2	A	S1	1	18	0	.01	29.3	.783	P	.9	179615	11539	0002331	...	7E-06
2	A	...	0	21	1	.01	29.3	.783	P	.9	179615	11539	0002331	...	7E-06
2	A	Sn	0	45	0	.01	29.3	.783	P	.9	179615	11539	0002331	...	7E-06
3	B	S1	1	18	0	.5	25.9	.888	PL	NA	147660	10960	0002331	...	3E-05
3	B	...	0	21	1	.5	25.9	.888	PL	NA	147660	10960	0002331	...	3E-05
3	B	Sn	0	45	0	.5	25.9	.888	PL	NA	147660	10960	0002331	...	3E-05
4	B	S1	0	18	0	.02	12.1	NA	NA	NA	147660	10960	0002331	...	3E-05
4	B	...	0	21	1	.02	12.1	NA	NA	NA	147660	10960	0002331	...	3E-05
4	B	Sn	0	45	0	.02	12.1	NA	NA	NA	147660	10960	0002331	...	3E-05

Set level, Sample level, Variant level, Gene level, Ontology level

C

Comparison of ACMG criteria counts on disease cohort
QV acmg_criteria.yaml vs Manually encoded