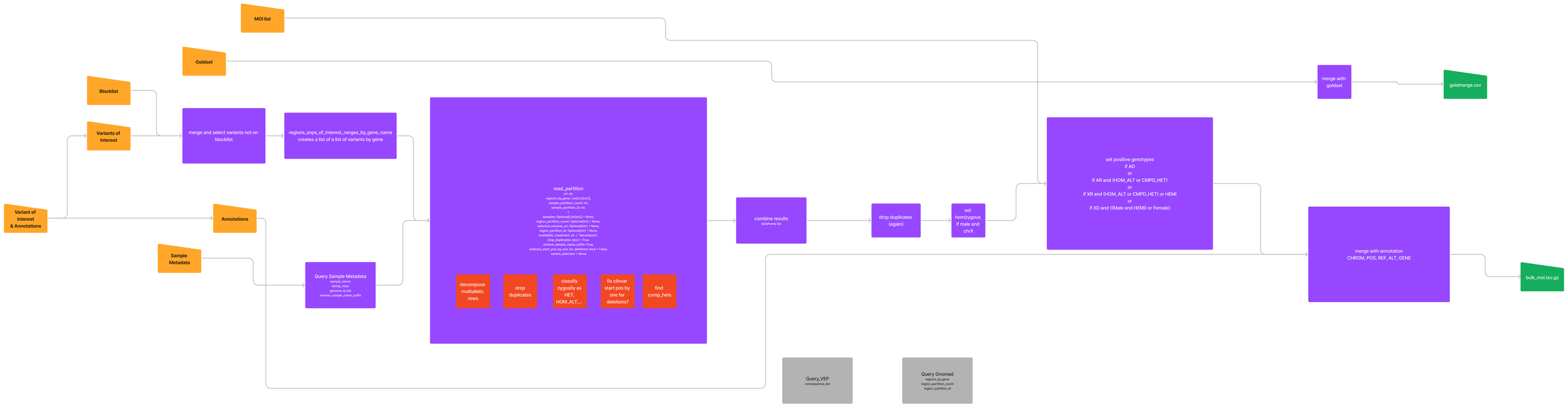
MOI list
Goldset
Blocklist
Variants of Interest
Variant of Interest & Annotations
Annotations
Sample Metadata
merge and select variants not on blocklist
regions_snps_of_interest_ranges_by_gene_name
creates a list of a list of variants by gene
Query Sample Metadata
sample_ssems
family_roles
genome_id_list
remove_sample_name_suffix
read_partition
set: etc
regions_by_gene: List[List[str]],
sample_partition_count: int,
sample_partition_id: int,
*,
samples: Optional[List[str]] = None,
region_partition_count: Optional[int] = None,
selected_variants_url: Optional[str] = None,
region_partition_id: Optional[int] = None,
multiallelic_treatment: str = 'decompose',
drop_duplicates: bool = True,
remove_sample_name_suffix=True,
subtract_start_pos_by_one_for_deletions: bool = False,
variant_selection = None
decompose multiallelic rows
drop duplicates
classify zygosity as HET, HOM_ALT,...
fix clinvar start pos by one for deletions?
find comp_hets
combine results
dataframe list
drop duplicates (again)
set hemizygous if male and chrX
set positive genotypes
if AD
or
if AR and (HOM_ALT or CMPD_HET)
or
if XR and (HOM_ALT or CMPD_HET) or HEMI
or
if XD and ((Male and HEMI) or Female)
merge with goldset
goldmerge.csv
merge with annotation
CHROM, POS, REF, ALT, GENE
bulk_moi.tsv.gz
Query_VEP
consequence_list
Query Gnomad
regions_by_gene
region_partition_count
region_partition_id