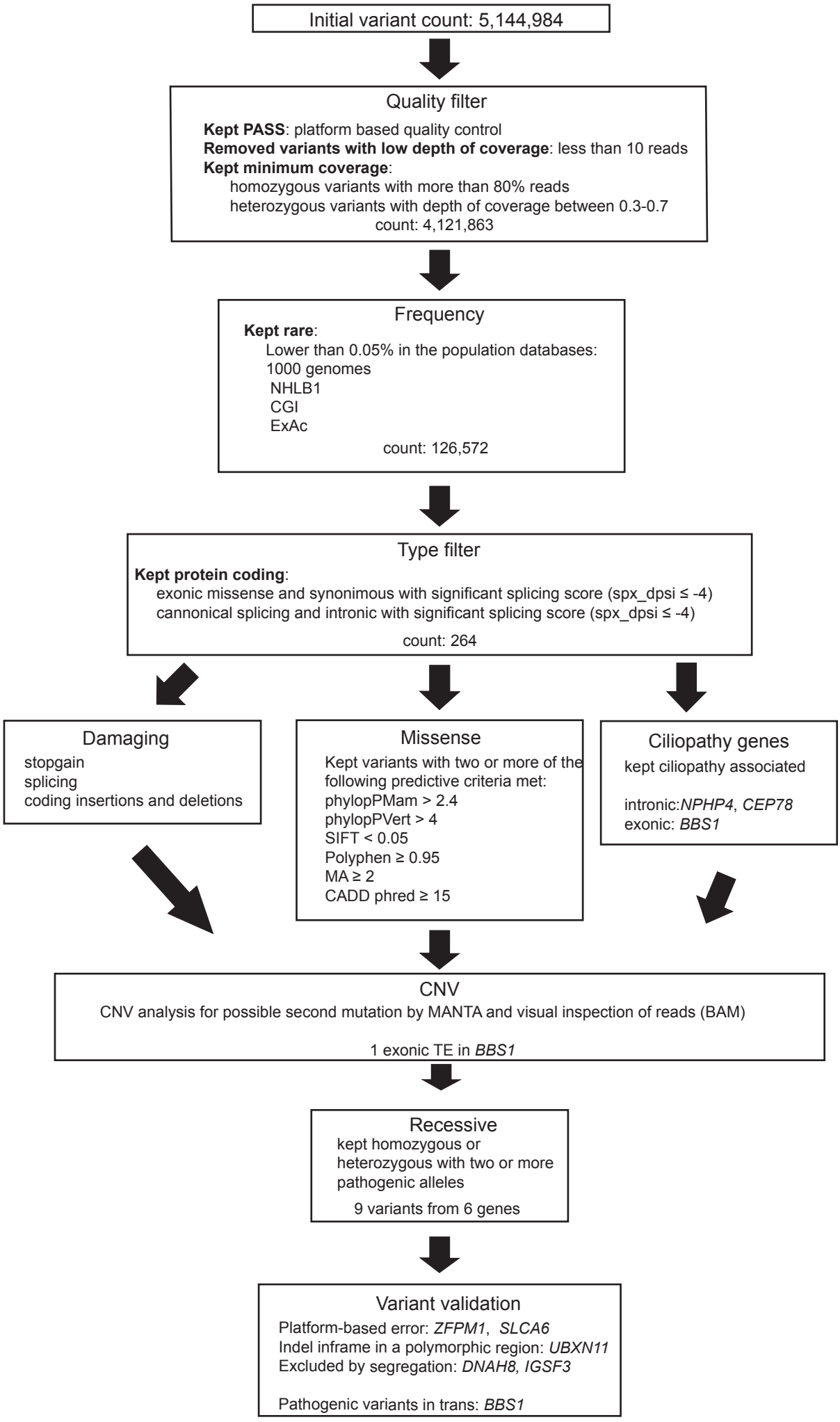
Initial variant count: 5,144,984
Quality filter
Kept PASS: platform based quality control
Removed variants with low depth of coverage: less than 10 reads
Kept minimum coverage:
homozygous variants with more than 80% reads
heterozygous variants with depth of coverage between 0.3-0.7
count: 4,121,863
Frequency
Kept rare:
Lower than 0.05% in the population databases:
1000 genomes
NHLB1
CGI
ExAc
count: 126,572
Type filter
Kept protein coding:
exonic missense and synonimous with significant splicing score (spx_dpsi ≤ -4)
cannonical splicing and intronic with significant splicing score (spx_dpsi ≤ -4)
count: 264
Damaging
stopgain
splicing
coding insertions and deletions
Missense
Kept variants with two or more of the following predictive criteria met:
phylopPMam > 2.4
phylopPVert > 4
SIFT < 0.05
Polyphen ≥ 0.95
MA ≥ 2
CADD phred ≥ 15
Ciliopathy genes
kept ciliopathy associated
intronic:NPHP4, CEP78
exonic: BBS1
CNV
CNV analysis for possible second mutation by MANTA and visual inspection of reads (BAM)
1 exonic TE in BBS1
Recessive
kept homozygous or heterozygous with two or more pathogenic alleles
9 variants from 6 genes
Variant validation
Platform-based error: ZFPM1, SLCA6
Indel inframe in a polymorphic region: UBXN11
Excluded by segregation: DNAH8, IGSF3
Pathogenic variants in trans: BBS1