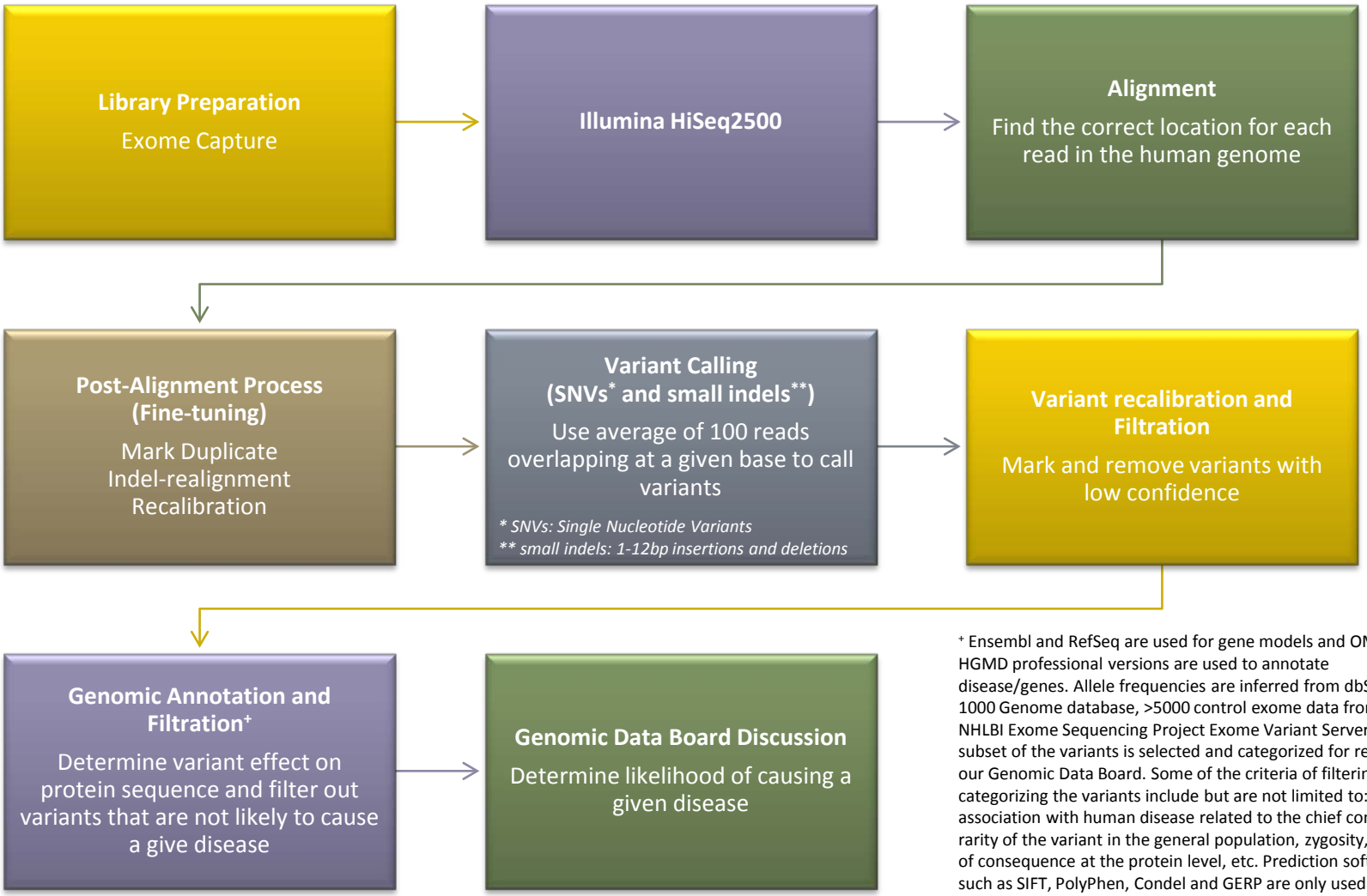
UCLA Clinical Exome Sequencing Workflow



+ Ensembl and RefSeq are used for gene models and OMIM and HGMD professional versions are used to annotate disease/genes. Allele frequencies are inferred from dbSNP135, 1000 Genome database, >5000 control exome data from the NHLBI Exome Sequencing Project Exome Variant Server. A subset of the variants is selected and categorized for review by our Genomic Data Board. Some of the criteria of filtering and categorizing the variants include but are not limited to: association with human disease related to the chief complaint, rarity of the variant in the general population, zygosity, degree of consequence at the protein level, etc. Prediction software such as SIFT, PolyPhen, Condel and GERP are only used for references and not to filter out variants.