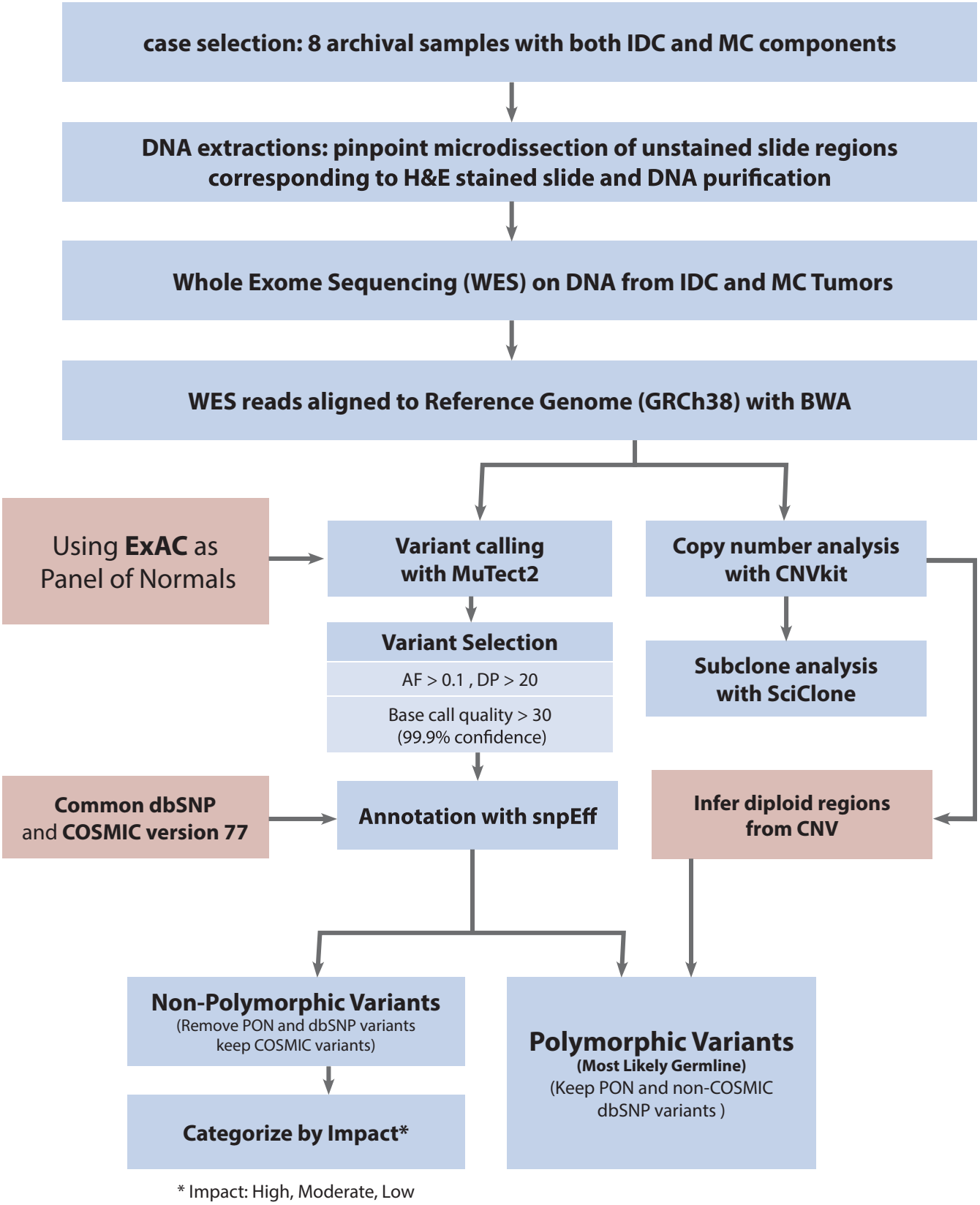
case selection: 8 archival samples with both IDC and MC components
DNA extractions: pinpoint microdissection of unstained slide regions corresponding to H&E stained slide and DNA purification
Whole Exome Sequencing (WES) on DNA from IDC and MC Tumors
WES reads aligned to Reference Genome (GRCh38) with BWA
Using ExAC as Panel of Normals
Variant calling with MuTect2
Copy number analysis with CNVkit
Variant Selection
AF > 0.1 , DP > 20
Base call quality > 30 (99.9% confidence)
Subclone analysis with SciClone
Common dbSNP and COSMIC version 77
Annotation with snpEff
Infer diploid regions from CNV
Non-Polymorphic Variants
(Remove PON and dbSNP variants keep COSMIC variants)
Categorize by Impact*
Polymorphic Variants
(Most Likely Germline)
(Keep PON and non-COSMIC dbSNP variants)
* Impact: High, Moderate, Low