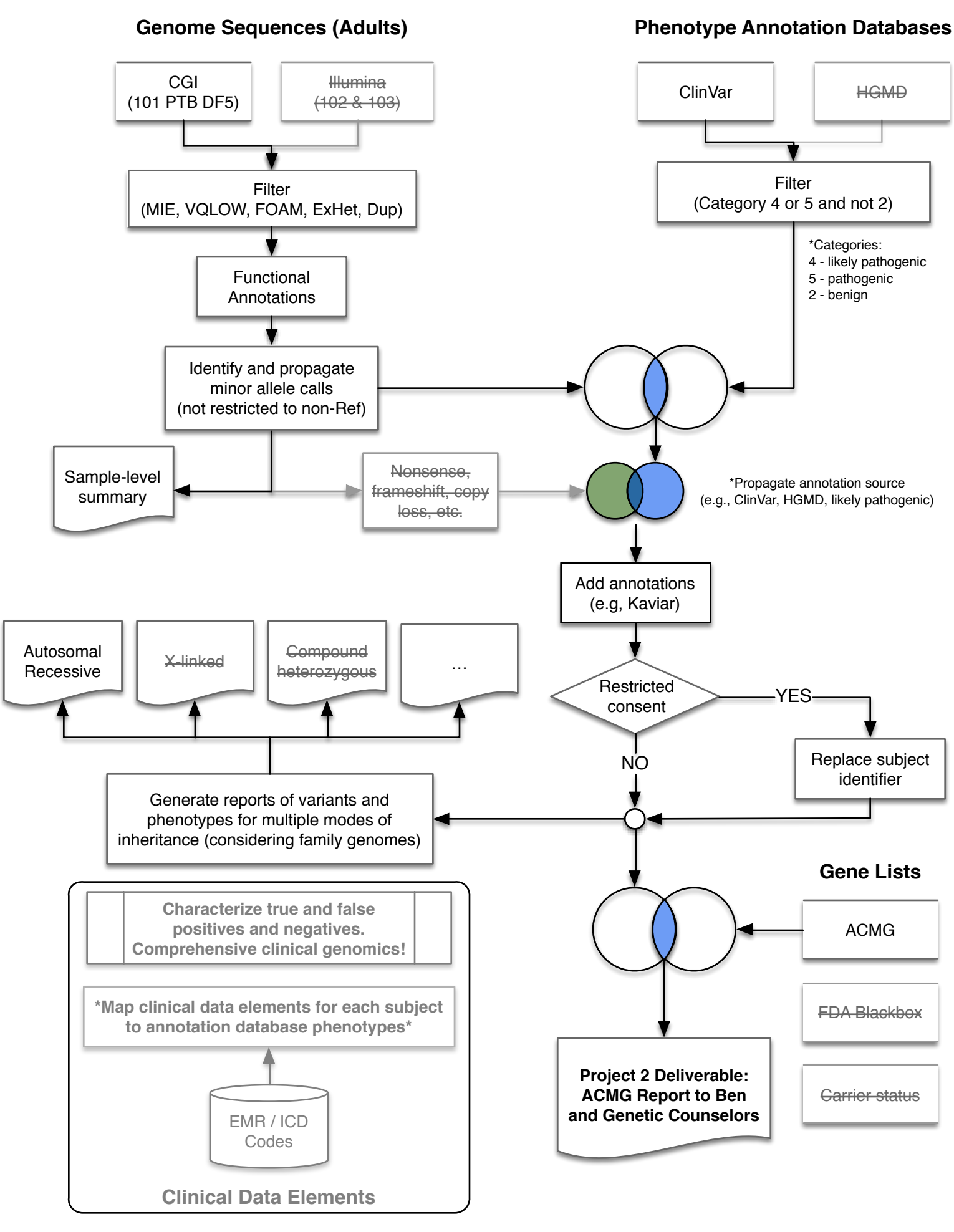
Genome Sequences (Adults)
Phenotype Annotation Databases
CGI
(101 PTB DF5)
~~Illumina~~
~~(102 & 103)~~
ClinVar
~~HGMD~~
Filter
(MIE, VQLOW, FOAM, ExHet, Dup)
Filter
(Category 4 or 5 and not 2)
*Categories:
4 - likely pathogenic
5 - pathogenic
2 - benign
Functional
Annotations
Identify and propagate
minor allele calls
(not restricted to non-Ref)
Sample-level
summary
~~Nonsense,~~
~~frameshift, copy~~
~~loss, etc.~~
*Propagate annotation source
(e.g., ClinVar, HGMD, likely pathogenic)
Add annotations
(e.g, Kaviar)
Autosomal
Recessive
~~X-linked~~
~~Compound~~
~~heterozygous~~
…
Restricted
consent
YES
NO
Replace subject
identifier
Generate reports of variants and
phenotypes for multiple modes of
inheritance (considering family genomes)
Gene Lists
ACMG
~~FDA Blackbox~~
~~Carrier status~~
Project 2 Deliverable:
ACMG Report to Ben
and Genetic Counselors
Characterize true and false
positives and negatives.
Comprehensive clinical genomics!
*Map clinical data elements for each subject
to annotation database phenotypes*
EMR / ICD
Codes
Clinical Data Elements