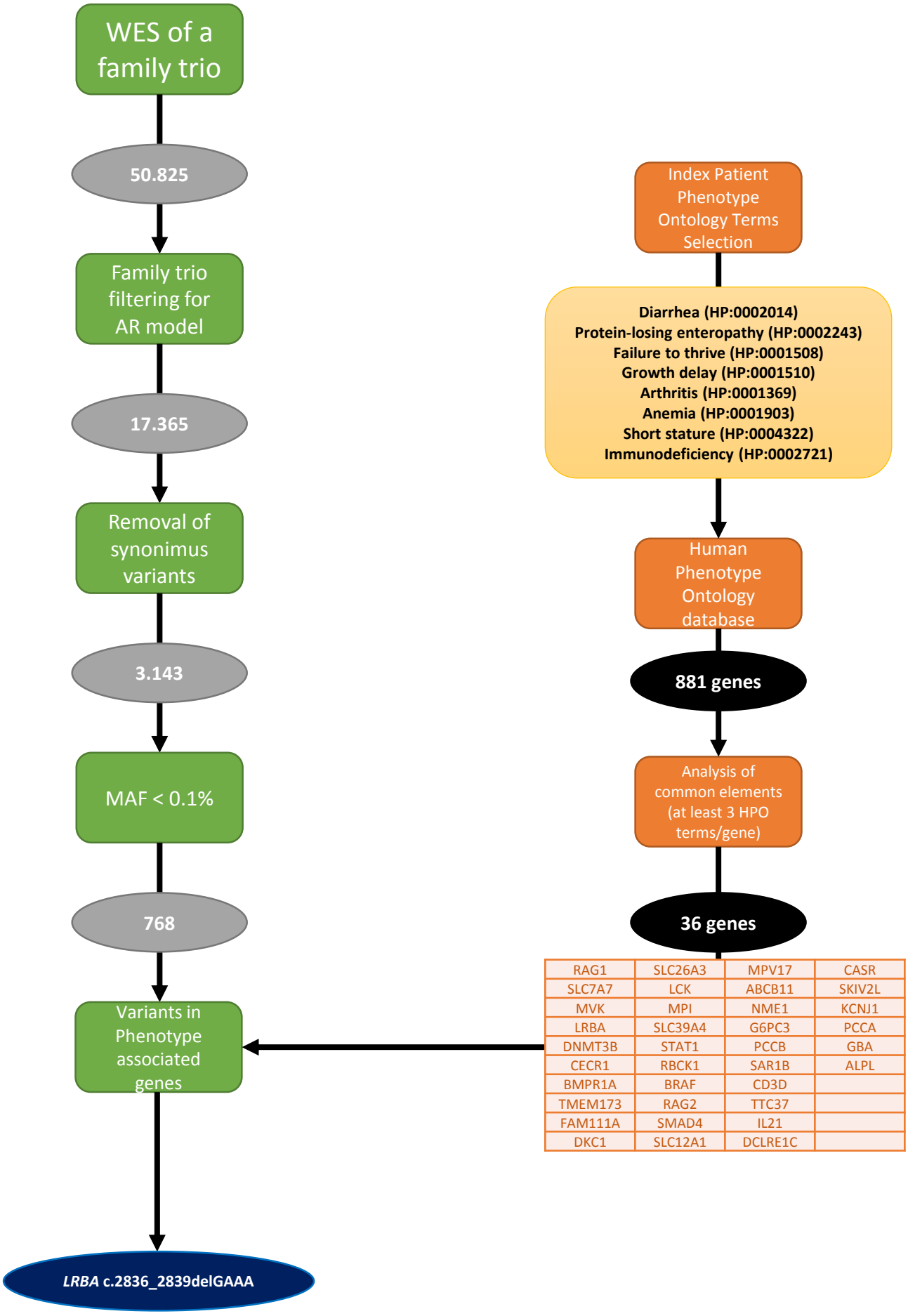
WES of a family trio
50.825
Family trio filtering for AR model
17.365
Removal of synonimus variants
3.143
MAF < 0.1%
768
Variants in Phenotype associated genes
LRBA c.2836_2839delGAAA
Index Patient Phenotype Ontology Terms Selection
Diarrhea (HP:0002014)
Protein-losing enteropathy (HP:0002243)
Failure to thrive (HP:0001508)
Growth delay (HP:0001510)
Arthritis (HP:0001369)
Anemia (HP:0001903)
Short stature (HP:0004322)
Immunodeficiency (HP:0002721)
Human Phenotype Ontology database
881 genes
Analysis of common elements (at least 3 HPO terms/gene)
36 genes
RAG1 SLC26A3 MPV17 CASR
SLC7A7 LCK ABCB11 SKIV2L
MVK MPI NME1 KCNJ1
LRBA SLC39A4 G6PC3 PCCA
DNMT3B STAT1 PCCB GBA
CECR1 RBCK1 SAR1B ALPL
BMPR1A BRAF CD3D
TMEM173 RAG2 TTC37
FAM111A SMAD4 IL21
DKC1 SLC12A1 DCLRE1C