

Annotate variants in genes

pLoF

AlphaMissense

5 annotation algorithms:
SIFT, PolyPhen2 (HIV), PolyPhen2 (HVAR),
LRT and MutationTaster

4 masks: pLoF, pLoF+AlphaMissense,
pLoF+Missense(5/5), pLoF+Missense(1/5)

3 AAF: 1%, 0.1%, singletons

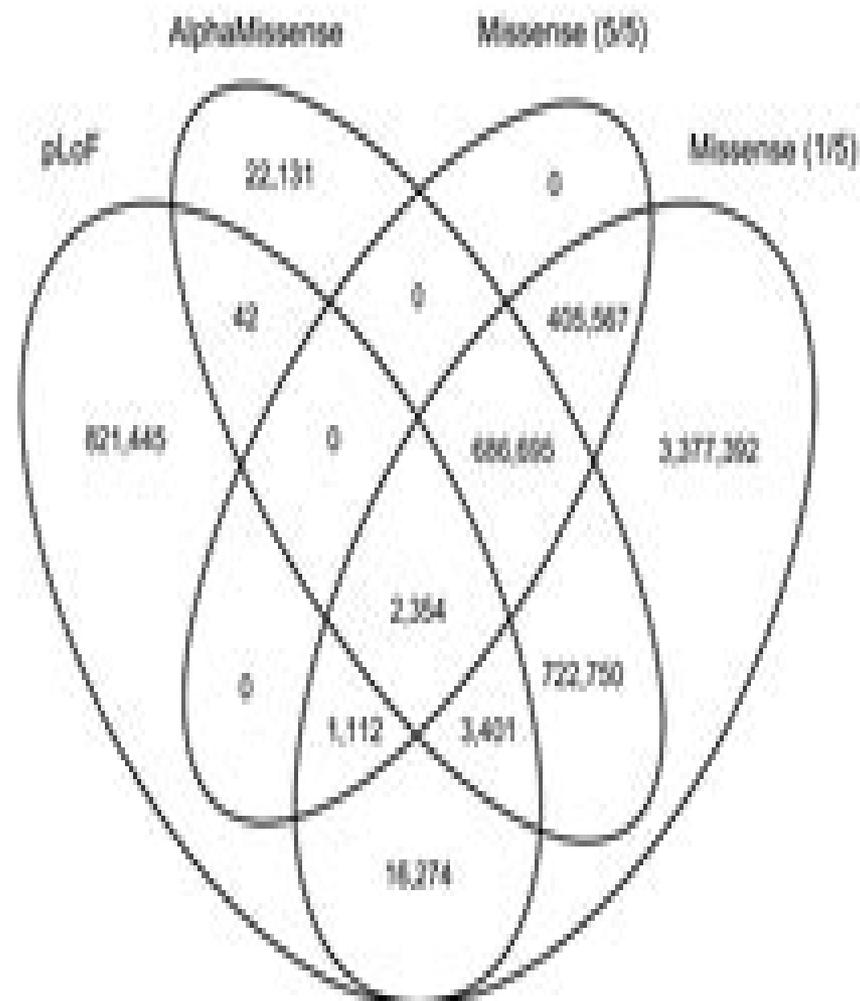
1 AAF: 1%

Burden test

SKAT, SKAT-O

Gene p-value

B



C

	pLoF	pLoF+ AlphaMissense	pLoF+ Missense (5/5)	pLoF+ Missense (5/5)
Tested genes	18,574	18,758	18,715	18,768

Advances In Human Genetics 3

Margaret E. Hertzig, Ellen A. Farber



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