Generation of possible single-nucleotide variants with a given effect on protein-coding sequence

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Aim: Create a tool to generate pathogenic and benign SNPs for OMIM genes by substitution of 1 nucleotide codon resulting in the same amino acid substitution.

Results

The script work accuracy was validated on coordinates of reference SNPs by Integrative Genomics Viewer, IGV; the script works correctly.

- Generated pathogenic and benign SNPs were not previously described in OMIM and ClinVar DBs (Tab.1)
- Generated pathogenic and benign SNPs could increase the accuracy of molecular genetic diagnosis of diseases associated with OMIM genes

generated SNPs	benign	likely benign	pathogenic	likely pathogenic
number	815	1609	876	994
with gnomAD freq	14%	17%	11%	10%
gnomAD freq mean	0.035	0.05	0.005	0.0001
std	0.13	0.13	0.007	0.0002
max	0.62	0.71	0.023	0.002

Workflow: