



- regulatory region
- TF binding site

- 5 prime UTR variant
- start retained variant
- start lost

- splice donor variant
- splice acceptor variant

- 3 prime UTR variant

- downstream gene variant

- intergenic variant
- upstream gene variant

- synonymous variant
- missense variant
- inframe insertion
- inframe deletion
- stop gained
- frameshift variant
- coding sequence variant

- splice region variant
- splice donor 5th base
- splice donor region
- splice polypyrimidine tract
- intron variant

- stop lost
- stop retained variant
- incomplete terminal codon variant

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|---------------------------|------------------------------------|
| transcript ablation | non coding transcript exon variant |
| transcript amplification | non coding transcript variant |
| protein altering variant | TFBS ablation / amplification |
| mature miRNA variant | feature elongation |
| NMD transcript variant | feature truncation |
| coding transcript variant | sequence variant |