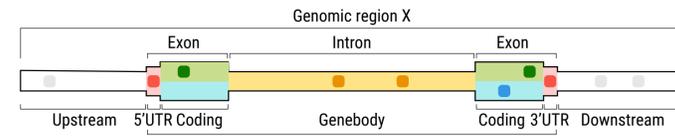


ACTG-Haplotype Notation

Gene | Transcript Separator

H B B : **a 1** **c 1** **t 1** **g 1**



Variant

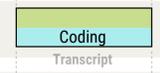
- amino-acid effect
- genebody effect
- coding effect
- transcript effect
- upstream or downstream effect

Haplotype levels

Amino acid sequence level
(Missense, Stop, Frameshift)



Coding sequence (CDS) level
(Nonsynonymous)



Transcript level
(3'UTR or 5'UTR in exon region)



Genebody level
(Intron)

