| Hex Color | * | SO term | SO description | SO accession | Old Ensembl term |
|-----------|---|-----------------------------------|---|-------------------|---------------------------------|
| #02599C | | intron_variant | A transcript variant occurring within an intron | SO:0001627 | Intronic |
| #32CD32 | | non_coding_exon_variant | A sequence variant that changes non-coding exon sequence | SO:0001792 | Within non coding gene |
| #32CD32 | | nc_transcript_variant | A transcript variant of a non coding RNA | SO:0001619 | Within non coding gene |
| #458B00 | | coding_sequence_variant | A sequence variant that changes the coding sequence | SO:0001580 | Coding unknown |
| #458B00 | | mature_miRNA_variant | A transcript variant located with the sequence of the mature miRNA | SO:0001620 | Within mature miRNA |
| #636363 | | intergenic_variant | A sequence variant located in the intergenic region, between genes | SO:0001628 | Intergenic |
| | | | A sequence variant where at least one base in the terminator codon is changed, but | 50,0001567 | |
| #76EE00 | | stop_retained_variant | the terminator remains | SO:0001567 | Synonymous coding |
| #76EE00 | | synonymous_variant | A sequence variant where there is no resulting change to the encoded amino acid | SO:0001819 | Synonymous coding |
| #7AC5CD | | 5_prime_UTR_variant | A UTR variant of the 5' UTR | SO:0001623 | 5prime UTR |
| #7AC5CD | | 3_prime_UTR_variant | A UTR variant of the 3' UTR | SO:0001624 | 3prime UTR |
| #7F7F7F | | feature_elongation | A sequence variant that causes the extension of a genomic feature, with regard to the reference sequence | SO:0001907 | Feature elongation |
| #7F7F7F | | feature_truncation | A sequence variant that causes the reduction of a genomic feature, with regard to the reference sequence | SO:0001906 | Feature truncation |
| #A2B5CD | | upstream_gene_variant | A sequence variant located 5' of a gene | SO:0001631 | Upstream |
| #A2B5CD | | downstream_gene_variant | A sequence variant located 3' of a gene | SO:0001632 | Downstream |
| #A52A2A | | TFBS_ablation | A feature ablation whereby the deleted region includes a transcription factor binding site | SO:0001895 | Tfbs ablation |
| #A52A2A | | TFBS_amplification | A feature amplification of a region containing a transcription factor binding site | SO:0001892 | Tfbs amplification |
| #A52A2A | | TF_binding_site_variant | A sequence variant located within a transcription factor binding site | <u>SO:0001782</u> | Regulatory region |
| #A52A2A | | regulatory_region_ablation | A feature ablation whereby the deleted region includes a regulatory region | <u>SO:0001894</u> | Regulatory region ablation |
| #A52A2A | | regulatory_region_amplification | A feature amplification of a region containing a regulatory region | SO:0001891 | Regulatory region amplification |
| #A52A2A | | regulatory_region_variant | A sequence variant located within a regulatory region | SO:0001566 | Regulatory region |
| #FF0000 | | transcript_ablation | A feature ablation whereby the deleted region includes a transcript feature | <u>SO:0001893</u> | Transcript ablation |
| #FF0000 | | stop_gained | A sequence variant whereby at least one base of a codon is changed, resulting in a | SO:0001587 | Stop gained |
| | | | premature stop codon, leading to a shortened transcript | | |
| #FF0000 | | stop_lost | A sequence variant where at least one base of the terminator codon (stop) is changed, resulting in an elongated transcript | <u>SO:0001578</u> | Stop lost |
| #FF00FF | | incomplete_terminal_codon_variant | A sequence variant where at least one base of the final codon of an incompletely annotated transcript is changed | SO:0001626 | Partial codon |
| #FF4500 | | NMD_transcript_variant | A variant in a transcript that is the target of NMD | <u>SO:0001621</u> | NMD transcript |
| #FF69B4 | | frameshift_variant | A sequence variant which causes a disruption of the translational reading frame, because the number of nucleotides inserted or deleted is not a multiple of three | SO:0001589 | Frameshift coding |
| #FF69B4 | | transcript_amplification | A feature amplification of a region containing a transcript | SO:0001889 | Transcript amplification |
| #FF69B4 | | inframe_insertion | An inframe non synonymous variant that inserts bases into in the coding sequence | SO:0001821 | Non synonymous coding |
| #FF69B4 | | inframe_deletion | An inframe non synonymous variant that deletes bases from the coding sequence | SO:0001822 | Non synonymous coding |
| #FF7F50 | | splice_donor_variant | A splice variant that changes the 2 base region at the 5' end of an intron | SO:0001575 | Essential splice site |
| #FF7F50 | | splice_acceptor_variant | A splice variant that changes the 2 base region at the 3' end of an intron | SO:0001574 | Essential splice site |
| #FF7F50 | | splice_region_variant | A sequence variant in which a change has occurred within the region of the splice site, either within 1-3 bases of the exon or 3-8 bases of the intron | SO:0001630 | Splice site |
| #FFD700 | | initiator_codon_variant | A codon variant that changes at least one base of the first codon of a transcript | SO:0001582 | Non synonymous coding |
| #FFD700 | | missense_variant | A sequence variant, that changes one or more bases, resulting in a different amino acid sequence but where the length is preserved | <u>SO:0001583</u> | Non synonymous coding |