A Nomenclature for Copy Number Variant Genes?

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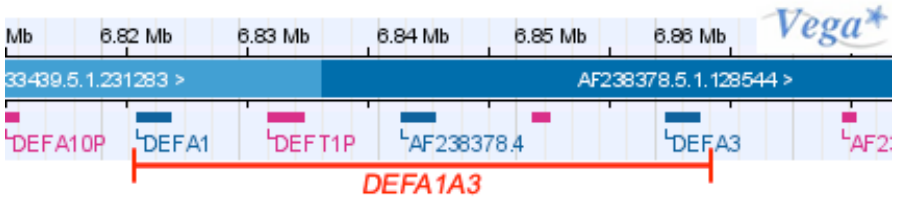
What the HGNC does now…

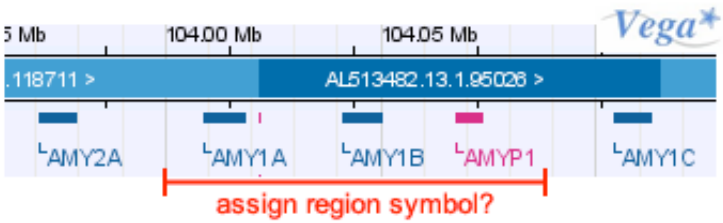
Currently the HGNC only assigns nomenclature to CNV genes that are present on the reference or an alternate genome assembly (including annotated alternate haplotypes).

On rare occasions we have assigned nomenclature to regions of extensive copy number variation when requested **e.g.** the region containing variable copy numbers of the *DEFA1, DEFT1P* and *DEFA3* genes has been assigned the symbol *DEFA1A3* "defensin, alpha 1 and alpha 3, variable copy number locus"



What we may consider doing…

Increasing the assignment of CNV region symbols for regions where the start and end points can be well established, if there is potential to aid scientific communication by doing so **e.g.** amylase genes. Individual haplotypes originally described as *AMY2B-AMY2A-*[*AMY1A-AMY1B-AMYP1*]n*-AMY1C*



What we avoid…

In some instances localised CNV is already being described using allele nomenclature. In such cases the HGNC would avoid creating additional nomenclature. e.g *CYP2D6*1XN* (N active *CYP2D6* genes) or the allele nomenclature for hybrid genes arising from rearrangement within in the *CYP2D* cluster

adapted from Gaedigk *et al*, 2010

Factors to consider in assigning CNV locus nomenclature

- The level of resolution at which the CNV has been described
- The frequency at which the CNV is observed
- Whether the CNV is contiguous or interspersed throughout the genome
- The requirements of researchers studying the gene/ region
- Minimising redundancy and overlap with existing or proposed allele nomenclature
- HGNC does not assign names to alleles or haplotypes

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