Copy number variation

Copy number variation (CNV) is a phenomenon in which sections of the genome are repeated and the number of repeats in the genome varies between individuals in the human population.

Copy number variation is a type of structural variation: specifically, it is a type of duplication or deletion event that affects a considerable number of base pairs.

However, note that although modern genomics research is mostly focused on human genomes, copy number variations also occur in a variety of other organisms including E. coli and S. cerevisiae.

Recent research indicates that approximately two thirds of the entire human genome is composed of repeats and 4.8–9.5 % of the human genome can be classified as copy number variations.

In mammals, copy number variations play an important role in generating necessary variation in the population as well as disease phenotype.

The prevalence of copy number variants (CNV), which is emerging as a mechanism of tumorigenesis in sporadic pituitary neuroendocrine tumors in general, is also unclear in prolactinomas. To characterize the genetic events underpinning sporadic prolactinomas, De Sousa et al. performed whole exome sequencing of paired tumor and germline DNA from 12 prolactinoma patients. De Sousa et al. observed recurrent large-scale CNV, most commonly in the form of copy number gains. We also identified sequence variants of interest in 15 genes. This included the DRD2, PRL, TMEM67, and MLH3 genes with plausible links to prolactinoma formation. Of the 15 genes of interest, CNV was seen at the gene locus in the corresponding tumor in 10 cases, and pituitary expression of eight genes was in the top 10% of tissues. However, none of our shortlisted somatic variants appeared to be classical driver mutations as no variant was found in more than one tumor. Future directions of research include mechanistic studies to investigate how CNV may contribute to prolactinoma formation, larger studies of relevant prolactinoma subsets according to clinical characteristics, and additional genetic investigations for aberrations not captured by whole exome sequencing [1)].

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De Sousa SMC, Wang PPS, Santoreneos S, Shen A, Yates CJ, Babic M, Eshraghi L, Feng J, Koszyca B, Roberts-Thomson S, Schreiber AW, Torpy DJ, Scott HS. The Genomic Landscape of Sporadic Prolactinomas. Endocr Pathol. 2019 Aug 31. doi: 10.1007/s12022-019-09587-0. [Epub ahead of print] PubMed PMID: 31473917.