# Chromosome 22

Chromosome 22 is one of the 23 pairs of chromosomes in human cells. Humans normally have two copies of Chromosome 22 in each cell. Chromosome 22 is the second smallest human chromosome (chromosome 21 being smaller), spanning about 49 million DNA base pairs and representing between 1.5 and 2% of the total DNA in cells.

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Deletion in 22q and mutations in the neurofibromatosis type 2 (NF2) gene are frequent in sporadic meningiomas. The tumor suppressor protein merlin is encoded by NF2, and mutations may promote tumor development. NF2 status is increasingly important in meningioma diagnosis and Tollefsen et al. questioned whether merlin immunohistochemistry could be used as an accessible and affordable surrogate marker for prediction of NF2 mutations. Previous studies on merlin immunoreactivity have reported diverging results. They aimed to describe the immunohistochemical expression of merlin in a large series of meningiomas and relate these findings to clinicopathological features and NF2 status. Standardized immunohistochemistry was conducted on 172 meningiomas using three different merlin antibodies directed toward the N-terminal, C-terminal and phospho-merlin (ser 518). Twenty of the included cases had known NF2 status. All tumor specimens were immunoreactive for the three merlin antibodies. The immunoreactivity of phosphorylated merlin was higher in meningothelial tumors. There were no other significant associations between merlin immunoreactivity and NF2 status, WHO grade, tumor subtype, tumor location or gender. These results indicate that merlin immunoreactivity does not seem to be predictive of NF2 mutation, as merlin was abundantly expressed by all included tumors and independently of NF2 status [1].

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Neurofibromatosis type 2 (NF2), a multiple neoplasia syndrome, is a manifestation of an impaired expression of the merlin protein [2] [3] exerting inhibitory effects on cell proliferation signals due to abnormalities of the NF2 gene located on chromosome 22.

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Biallelic inactivation of the NF2 gene has been established as the principal underlying genetic event in patients with sporadic and syndrome-associated vestibular schwannoma (VS). Two independent teams contemporaneously identified the NF2 gene located on chromosome 22 at 22q12.2 in 1993, which codes for the tumor suppressor protein Merlin, also called schwannomin

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In 1999, researchers working on the Human Genome Project announced they had determined the sequence of base pairs that make up this chromosome. Chromosome 22 was the first human chromosome to be fully sequenced.

Identifying genes on each chromosome is an active area of genetic research, because researchers use different approaches to predict the number of genes on each chromosome, the estimated number of genes varies. Chromosome 22 contains about 693 genes.

Chromosome 22 was originally identified as the smallest chromosome. After extensive research,

however, researchers concluded that chromosome 21 was smaller. The numbering of these chromosomes wasn't rearranged because of chromosome 21 being known by that designation as the chromosome that can lead to Down syndrome.

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Loss of chromosome 22 and gain of 1q are the most frequent genomic aberrations in ependymomas, indicating that genes mapping to these regions are critical in their pathogenesis. Using real-time quantitative PCR, we measured relative copy numbers of 10 genes mapping to 22q12.3-q13.33 and 10 genes at 1q21-32 in a series of 47 pediatric intracranial ependymomas. Loss of one or more of the genes on 22 was detected in 81% of cases, with RAC2 and C22ORF2 at 22q12-q13.1 being deleted most frequently in 38% and 32% of ependymoma samples, respectively. Combined analysis of quantitative-PCR with methylation-specific PCR and bisulphite sequencing revealed a high rate (>60% ependymoma) of transcriptional inactivation of C22ORF2, indicating its potential importance in the development of pediatric ependymomas. Increase of relative copy numbers of at least one gene on 1q were detected in 61% of cases, with TPR at 1q25 displaying relative copy number gains in 38% of cases. Patient age was identified as a significant adverse prognostic factor, as a significantly shorter overall survival time (P = 0.0056) was observed in patients <2 years of age compared with patients who were >2 years of age. Loss of RAC2 at 22q13 or amplification of TPR at 1q25 was significantly associated with shorter overall survival in these younger patients (P = 0.0492 and P = < 0.0001, respectively). This study identifies candidate target genes within 1q and 22q that are potentially important in the pathogenesis of intracranial pediatric ependymomas [4].

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2)
Rouleau GA, Merel P, Lutchman M, et al. Alteration in a new gene encoding a putative membrane-organizing protein causes neurofibromatosis type 2. Nature 1993;363:515–21.
3)
Trofatter JA, MacCollin MM, Rutter JL, et al. A novel moesin-, ezrin-, radixin-like gene is a candidate for the neurofibromatosis 2 tumor suppressor. Cell 1993;72:791–800.
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Karakoula K, Suarez-Merino B, Ward S, Phipps KP, Harkness W, Hayward R, Thompson D, Jacques TS, Harding B, Beck J, Thomas DG, Warr TJ. Real-time quantitative PCR analysis of pediatric ependymomas identifies novel candidate genes including TPR at 1q25 and CHIBBY at 22q12-q13. Genes Chromosomes Cancer. 2008 Nov;47(11):1005-22. doi: 10.1002/gcc.20607. PubMed PMID: 18663750.

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