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An overall view of the new genetic developments that changed the classification of tumours of central nervous system

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The last WHO classification of tumours of the nervous system of 2016 stresses the importance of genetic alterations that permitted the inclusion of new diagnostic categories. This is the case of tumours of the diffuse astrocytic type: Identification of mutations in the genes IDH-1 and IDH-2, either by immunohistochemistry or by genetic evaluation of mutations are important tools that have prognostic implications. A new entity has been added to this group with mutation of the H3K27M gene – diffuse midline glioma. The diagnosis of an oligodendroglial tumour needs the presence of co-deletions of the chromosomes 1p and 19q and mutation of the IDH-1 gene. Deletions or translocations resulting in BRAF fusion proteins are pathognomonic of pilocytic astrocytomas; Mutation of the FGFR1 are found in midline pilocytic astrocytomas. Many molecular alterations have been described in the ependymoma group: Cytogenetic aberrations, fusion genes involving the RELA or YAP1 genes are found in supratentorial ependymomas whereas for the spinal cord group present with genome wide polyploidy. These characteristics have prognostic implications. MGMT promoter methylation was described in all choroid plexus papilloma and mutations of the TP53 gene was found in almost all choroid plexus carcinoma and those exhibiting loss of the chromosome 12q are associated to a shorter survival. MYCN gain and overexpression of genes of the WNT signaling pathway were described in central neurocytomas. Medulloblastomas may be genetically or histologically defined with important prognostic implications. Finally, inversion on chromosome 12q13 generates the fusion of the NAB2 et STAT6 genes and induces the nuclear expression of STAT6 necessary for the diagnosis of the solitary fibrous tumour/hemangiopericytoma. Since 2016 a number of new genotype varieties of tumours of the central nervous system have been added.

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