Nervous system: Medulloblastoma

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Clinics and pathology

Disease

Medulloblastomas are malignant invasive embryonal tumours of the cerebellum with a tendency to metastasize in the central nervous system (CNS). This tumor is more frequently found in children.

Epidemiology

It represents 10 at 20 % of brain tumours and 30 % of tumours localized in posterior fossa; annual incidence is 0,5 per 100000 children; peak of occurrence at 7 years.

Pathology

Belongs to the primitive neurectodermal tumours (PNET): highly malignant embryonal tumours of the CNS with predominant neuronal differentiation. Several variants medulloblastoma are recognized in the OMS classification:

Classic medulloblastoma composed of densely jacked round-cells with round to oval hyperchromatic nuclei.

Desmoplastic medulloblastoma represents a variant with abundant reticulin and collagen.

Large cell medulloblastoma is a rare variant composed of cells with large round nuclei.

Histological features of a typical medulloblastoma: Homer-Wright rosettes - Anne Marie Capodano.
Immuno histochemistry: Classic medulloblastoma is strongly immuno-reactive for Vimentin. Some tumours are immunoreactive for NSE, Synaptophysine and GSAP.

Treatment

The treatment associates total surgical resection and radiotherapy or, according to the age, chemotherapy.

Prognosis

Survival without recurrence is 50 at 70 %; depends on the quality of surgical resection and on the presence of metastases at the time of diagnosis.

Cytogenetics

The most common specific abnormality in medulloblastomas, which is present in approximately 50 % of cases, is isochromosome 17q [i(17q)]. The breakpoint is in the proximal portion of p-arm at 17p11.2, so that the resultant structure is dicentric. In a few cases, partial or complete loss of 17p occurs through interstitial deletion, unbalanced translocation or monosomy 17. Chromosome 1 is also involved in medulloblastomas. The most frequent abnormalities are unbalanced translocations, deletions and duplications. Rearrangements of chromosome 1 often result in trisomy 1q without loss of the p-arm. Others less common chromosomal changes are: deletions of 6q, 9q, 10q, 11q, 11p and 16q, monosomy 22 and in rare cases double minutes.

Cytogenetics Molecular

Isochromosome 17q has been observed in interphase nuclei using fluorescence in situ hybridization. This technique is used in particular when only a few metaphases are obtained or when only normal diploid cells are obtained in culture.

Genes involved and proteins

Note

Studies on loss of heterozygosiy (LOH) have confirmed loss of portions of 17p in 30-45 % of cases. Some studies showed a correlation between LOH for 17p and a poor response to therapy and shortened survival. Mutations of p53 gene located on 17p13 have been found in only 5-10 % of these tumors. Expression of PAX5 and PAX6 mRNA was shown in 70 % of medulloblastomas. The precise mechanism by which these genes are involved remains unknown. Inactivation of PTCH tumor suppressor gene occurs in a subset of medulloblastomas.

References


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