Solid Tumour Section

Short Communication

Bone: Chordoma

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Abstract

Review on Chordoma, with data on clinics, and the genes involved.

Keywords
Chordoma; Bone tumor; Notochord; Spinal; CDKN2A; CDKN2B; T (T Brachyury Transcription Factor)

Identity
Chordoma is a malignant tumour derived from remnants of the fetal notochord; it occurs along the spinal axis, predominantly in the sphenoccipital (35%), vertebral (15%) and sacroccygeal (50%) regions

Phylum
Bones:Notochordal tumors:Chordoma

Clinics and pathology

Etiology
Although most chordomas are sporadics, five families with chordoma occurrence have been reported, two of them displaying an autosomal dominant transmission with incomplete penetrance (MIM. *215400).

Epidemiology
Chordomas accounts for 1-4% of all primary bone tumours; the sacroccygeal lesions are more common in the fifth decade of life, whereas the sphenoccipital tumours occur predominantly in children.

Clinics
Chordoma is a slowly-growing tumour, characterized by local destruction of bone and rarely distant metastatic spread.

The differential diagnosis includes renal tumours, chondrosarcomas and myxopapillary ependymoma.

Pathology
Microscopically, it resembles normal fetal notochord in its different stages of development; it is composed of extremely large cells (known as physaliferous) and other small tumour cells; areas of cartilage and bone may be present.

Dedifferentiated chordoma is a biphasic tumor, with features of a chordoma NOS and an abrupt transition to a high grade undifferentiated spindle cell tumor. Chordomas express keratins and brachyury is a highly specific marker for chordoma and helps to distinguish chordoma from chondrosarcoma and clear cell renal cell carcinoma. Brachyury as well as keratins are not expressed in a dedifferentiated component.

Cytogenetics
Bone: Chordoma

**Cytogenetics Morphological**

The most common cytogenetic abnormality in chordoma is monosomy of chromosome 1 and gain of chromosome 7.

**Genes involved and proteins**

Note

Approximately 70% of chordomas harbour either homozygous or heterozygous loss of CDKN2A and CDKN2B. Additional copy-number gain of brachyury (gene: T (T Brachyury Transcription Factor)) on chromosome locus 7q33 are commonly found in chordomas. In the coding region of brachyury somatic mutations have been found as well. Hotspot mutations in the tyrosine kinase receptor gene EGFR and in KRAS, NRAS, HRAS, BRAF are also described in chordomas. Furthermore, deletions and point mutations in SMARCB1 gene can be found.

**References**


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