

PATHOLOGIC DIAGNOSIS: BCOR ASSOCIATED SARCOMA

DISCUSSION:

BCOR associated sarcoma (*BCOR::CCNB3* rearranged sarcoma) is a recently defined genetic entity among undifferentiated round cell sarcomas, which are characterised by *BCOR* and *CCNB3* fusion¹. It was previously classified as the Ewing's-like sarcoma and treated similarly². This tumor commonly affects bone and soft tissue of children and young males with a mean age of 15 years and account for 4% to 13% of undifferentiated small round cell sarcomas without a *EWSR1* rearrangement³. A subset of these sarcomas present in the soft tissues of the trunk and extremities^{6,13}.

The fusion of *BCOR* and *CCNB3* protein as recently discovered by Pierron et al, is oncogenic and drives proliferation in this sarcoma⁴. Radiologically, these tumors present as permeative lytic or sclerotic lesions associated with cortical thickening in the metadiaphyseal region of long bones⁷.

Microscopically, the tumor shows predominantly proliferating atypical spindle and/or small round cells with a variety of morphologic features such as small whorls, myxoid stroma, a hemangiopericytomatous appearance, and/or hyalinized collagen resembling a solitary fibrous tumor, and angiomatous or slit-like spaces containing extravasated erythrocytes^{1,10,13}. Close morphological differentials in this age group and location include Ewing's sarcoma, *CIC::DUX4* rearranged sarcoma, small cell osteosarcoma, synovial sarcoma, rhabdomyosarcoma, sclerosing epithelioid fibrosarcoma, malignant peripheral nerve sheath tumor, and neuroblastoma¹². An appropriate immunohistochemical panel for diagnosis includes CD99, *BCOR*, SATB2, TLE1, CyclinD1, EMA WT1, NKX2.2, DESMIN, WT1, CD45, ALK, CK, Synaptophysin, SALL4, S100, MYOD1, ERG, CD117. Diffuse nuclear *CCNB3* expression is highly specific and sensitive for the diagnosis of *BCOR::CCNB3* rearranged sarcoma⁹. *BCOR* and cyclin D1 are highly sensitive markers and show diffuse nuclear staining in all *BCOR::CCNB3* sarcomas. Cytoplasmic and focal membranous CD99 expression is seen, in contrast to the diffuse strong and membranous CD99 in Ewing's sarcoma. SATB2 and TLE1 co-expression is frequently seen⁸. For confirmation RT-PCR assay or FISH that in conjunction with *CCNB3* immunohistochemistry can be useful in diagnosing these tumors and differentiating the from *CIC-DUX4* rearranged sarcomas which are clinically more aggressive than *BCOR-CCNB3* rearranged sarcomas.^{5,7,11}