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Perspective

Surgical Pathology of Rare Bone Tumors in Adults

Jeanett Jose*

Departments of Surgical Pathology, University of Milan, Milan, Italy

DESCRIPTION

Rare bone tumors in adults constitute a diverse group of neoplasms that arise from osseous or cartilaginous tissue, exhibiting variable clinical behavior, histopathology, and prognosis. These tumors differ significantly from the more common primary bone tumors, such as osteosarcoma, chondrosarcoma, and giant cell tumor, and their rarity often leads to diagnostic difficulties and management challenges. Accurate pathological characterization is essential for determining prognosis, guiding surgical treatment, and identifying cases that may benefit from adjuvant therapy or targeted interventions. Despite advances in imaging and molecular diagnostics, surgical pathology remains the cornerstone for definitive diagnosis, providing critical information regarding tumor type, grade, margins, and potential aggressiveness.

Clinically, rare bone tumors in adults may present with nonspecific symptoms, including localized pain, swelling, pathological fracture, or reduced mobility, depending on the tumor location and size. These symptoms often overlap with those of more common bone conditions such as metastatic lesions, osteomyelitis, or degenerative disorders, which may delay recognition and biopsy. The anatomical distribution is variable, with long bones, pelvis, vertebrae, and craniofacial skeleton being frequent sites. In some cases, tumors are discovered incidentally during imaging performed for unrelated reasons. Laboratory investigations are often nonspecific, though markers such as alkaline phosphatase may be elevated in osteogenic lesions, and inflammatory markers may be mildly increased in aggressive or necrotic tumors.

Radiological evaluation is an essential step in the preoperative assessment and surgical planning of rare bone tumors. Plain radiographs provide initial information about tumor location, size, matrix mineralization, cortical involvement, and periosteal reaction. Computed tomography and magnetic resonance imaging offer detailed evaluation of intraosseous extent, soft tissue involvement, neurovascular proximity, and marrow infiltration. Certain imaging characteristics can suggest tumor lineage; for instance, a mineralized matrix with chondroid features may indicate a cartilaginous tumor, while lytic, expansile lesions with trabecular disruption suggest osteogenic origin. However, imaging features alone are insufficient for definitive diagnosis, as rare tumors often mimic more common primary bone neoplasms or metastatic lesions, necessitating histopathological confirmation.

Surgical pathology provides the definitive diagnosis through examination of tissue obtained by biopsy or resection. Rare bone tumors in adults exhibit considerable histological diversity. Examples include adamantinoma, a low-grade malignant tumor often arising in the tibia characterized by epithelial nests within a fibrous stroma; chordoma, arising from notochordal remnants with physaliphorous cells and myxoid matrix; clear cell chondrosarcoma, displaying large clear cells within a cartilaginous background; epithelioid sarcoma involving bone secondarily with distinctive nodular architecture and central necrosis; and undifferentiated pleomorphic sarcoma presenting with sheets of atypical spindle cells and variable osteoid or chondroid matrix. In many cases, tumors exhibit mixed or overlapping features, which may complicate classification and necessitate adjunctive studies.

Immunohistochemistry plays a vital role in differentiating rare bone tumors and confirming lineage. Immunohistochemistry is particularly valuable in small biopsies or poorly differentiated tumors where morphology alone may be insufficient for definitive classification.

The biological behavior of rare bone tumors varies widely. Some tumors, such as classical adamantinoma, are slow-growing and locally aggressive but rarely metastasize, whereas chordomas demonstrate indolent growth yet have a high risk of local recurrence and distant metastasis, particularly to lungs or lymph nodes. Clear cell chondrosarcoma may recur locally if excision is incomplete, and undifferentiated pleomorphic sarcoma is highly aggressive, with early metastasis and poor prognosis. Identification of histopathological features associated with aggressiveness, including mitotic activity, necrosis, vascular invasion, and cellular atypia, is critical for surgical planning and risk stratification.

Surgical management is the mainstay of treatment for rare bone tumors in adults. Wide local excision or en bloc resection with

Correspondence to: Jeanett Jose, Departments of Surgical Pathology, University of Milan, Milan, Italy, E-mail: jeanett.jose@i-med.ac.it

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clear margins is preferred, as incomplete resection is associated with high recurrence rates. Limb-sparing procedures are favored when feasible, but amputation may be required in cases involving neurovascular compromise or extensive soft tissue invasion. In tumors with a high risk of recurrence or metastasis, adjuvant radiotherapy or systemic therapy may be considered, although evidence is limited due to the rarity of these tumors. Long-term follow-up with imaging is essential, given the potential for late recurrences or metastasis, and surveillance strategies are tailored according to tumor type, grade, and surgical outcome.

Advances in imaging, immunohistochemistry, and molecular diagnostics have improved the ability to characterize rare bone tumors, yet surgical pathology remains indispensable for definitive diagnosis. Recognition of distinctive morphological patterns, careful evaluation of cellular features, and judicious use of immunohistochemical and molecular markers allow accurate classification and prognostication. Comprehensive assessment of these tumors enhances understanding of their biological behavior, informs surgical planning, and supports individualized patient management, ultimately improving outcomes for adults affected by these uncommon but clinically significant neoplasms.

CONCLUSION

Rare bone tumors in adults represent a heterogeneous group of neoplasms with diverse clinical, radiological, and histopathological characteristics. Surgical pathology provides definitive diagnosis, guiding treatment and prognosis through careful evaluation of morphology, immunohistochemical profile, and, when indicated, molecular features. Accurate characterization of these tumors is essential due to their variable biological behavior, potential for recurrence, and implications for surgical management. Multidisciplinary collaboration, integration of multiple diagnostic modalities, and ongoing research into molecular mechanisms continue to enhance understanding, diagnosis, and management of these rare but clinically significant tumors.