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Opinion Article

Molecular Diagnosis of Bone Tumours

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DESCRIPTION

The cornerstone of diagnosing bone tumours is classic histomorphology; however since the development of new molecular tools, this subject has rapidly advanced. The discovery of novel genetic changes in bone malignancies has increased understanding of the genetic basis of these tumours and given molecular pathology a more significant place in clinical treatment. Numerous studies conducted over the past few decades have shown that bone tumours can be roughly divided into those with simple karyotypes and those with complex karyotypes based on molecular alterations. Tumors with particular translocations, somatic gene mutations, or more or less particular amplifications can be sub classified under the first category. In contrast, sarcomas with complicated karyotypes typically don't have any distinctive changes. The detection of recurring genetic changes can be done using a variety of methods, including IHC analysis today. This article, however, concentrates on assays used often in molecular diagnostics. The importance of molecular pathology in routine diagnosis is underlined after discussing and illuminating tumour groups with distinctive genetic abnormalities with more detailed examples.

Bone tumours are a diverse category of benign and malignant neoplasms that pathologists believe are challenging to identify. Clinically significant bone tumors, particularly bone sarcomas, are uncommon, and it can be difficult to distinguish one from another using classic histomorphology because of the overlap in morphology. Additionally, when comparing bone tumours to soft tissue cancers, the use of standard immunohistochemistry analysis for the identification of the line of differentiation is less useful. Fortunately, significant improvements in molecular techniques over the past few decades have allowed for the identification of numerous genetic anomalies in bone cancers. These discoveries have shed light on how bone tumours develop, and as a result, a conceptual framework for molecular sub classification of bone tumours into essentially two groups has been developed. Tumors with translocations and cancers with particular gene mutations and/or amplifications are included in the first group of tumours, which exhibit simple karyotypes. The transcription, signaling, or gene function can all be affected by these particular recurrent gene alterations. Tumors without any distinct changes and complex karyotypes make up the second category. Additionally, combining molecular tests in a diagnostic setting is the consequence of the detection of molecular changes, which has been crucial in understanding the molecular pathways relevant to cancer. Bone tumour diagnostics have been considerably enhanced by the continued advancement of mutation-specific IHC analysis. An overview of the molecular assays now used in the diagnosis of bone cancers is provided in this study. The molecular categorization of bone cancers and altered molecular pathways important to carcinogenesis are then reviewed. Additionally, the clinical use of particular molecular changes pertinent to the differential diagnosis of a number of bone cancers is highlighted.

The need for DNA or RNA from these lesions for molecular research has been one of the major obstacles in the evaluation of genetic abnormalities in bone cancers. The DNA and RNA recovered from formalin-fixed, paraffin-embedded bone cancers frequently deteriorate because decalcification is necessary for an adequate histologic evaluation of bone tissue. Although having access to frozen tumour tissue is ideal, it is not always the case. Strong mineral acids and weaker organic acids, which hydrolyze and fragment nucleotides, respectively, more or less harm DNA and RNA among the acid-based agents. In contrast to lesser acids like formic acids, nitric acid and other powerful mineral acids act quickly, but they severely destroy nucleotides, making molecular testing impossible. The development of molecular techniques has speed up the analysis of the molecular landscape of bone tumors, which has contributed to a better understanding of the mechanisms underlying tumorigenesis and the development of conceptual models for bone tumorigenesis.

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