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Commentary

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The clinical impact of molecular techniques on diagnostic pathology of soft tissue and bone tumours

Lulian Spataru
Assistant professor, University of Medicine and Pharmacy "Carol Davila" Bucharest, Romania

ABSTRACT
Molecular techniques have become quite crucial for diagnostic histopathology. Their application in sarcomas, both in bone as well as in soft tissue, have been quite successful. Being sarcomas quite rare and diagnostically challenging, the use of ancillary molecular diagnostic tools is quite useful.

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Introduction

The integration of molecular techniques has been having a substantial impact on the pathology practice. In particular the most relevant changes caused by such approach are: better disease definition and therefore more accurate diagnostics, identification of molecular predictive and prognostic markers, unraveling novel molecular targets for more specific therapeutic approach and eventual drug development.

Molecular tools for soft tissue and bone tumors diagnostics

Primary soft tissue and bone tumors are a heterogeneous group of tumours including benign and malignant lesions. They have a mesenchymal origin, presumably from mesenchymal stem cells. First of all, as a group, they are rare (sarcomas, the malignant mesenchymal tumours, are less than 2% of all cancers) but over 100 subtypes are described. Furthermore morphologic criteria used to recognize malignancy in other fields are not always applicable.

In this setting the use of ancillary molecular techniques is diagnostically beneficial. The successful integration of these techniques relies also on the fact that many of these lesions are known to harbour specific point mutation, gene amplification and specific translocations.

For the time being we will not address every single mesenchymal lesion, its differential diagnosis and the role molecular approach may play in this. We will address paradigmatic cases in which the application of molecular techniques is widely used, crucial for the diagnosis and ultimately for the treatment.

In particular low grade fibromyxoid sarcoma may often be misdiagnosed as neurofibroma, perineurioma, cellular myxoma, nodular fasciitis and desmoid-type fibromatosis.

Recently, data coming from expression profile of low grade fibromyxoid sarcomas allowed to identify a novel marker for its differential diagnosis.

Regarding bone tumours, low grade osteosarcomas either parosteal osteosarcomas as well as central low grade osteosarcomas may closely simulate benign lesions such as desmoid-type fibromatosis and fibrous dysplasia, respectively. Both parosteal osteosarcoma and central low grade osteosarcoma are characterized by amplification of MDM2 gene which may be either assessed by FISH or, since such amplification results in an overexpression of the transcript and ultimately of the relative protein, by immunohistochemistry for MDM2.

Therefore larger studies are needed to confirm/reject an eventual role for GNAS1 mutation screening as an ancillary diagnostic tool for this differential diagnosis. This reminds us to use great caution in interpreting the results of every molecular test. More importantly integration of the acquired results with the clinico-pathological features is mandatory.

It is worthy to mention the same activating mutation in GNAS1 is found also in 50–60% of intramuscular myxoma, also in the cellular variant. Since this mutation is not occurring low grade myxo fibrosarcoma, mutation screening may be used for differential diagnosis.

Secondly, it has to be realized that molecular pathology is characterized by a relatively high methodological complexity. As a consequence it is necessary that inter-laboratory quality assessment

procedures are implemented.

Contact: Lulian S LulianSpataru@yahoo.com Assistant professor, University of Medicine and Pharmacy “Carol Davila” Bucharest, Romania