MICHIGAN MEDICINE
UNIVERSITY OF MICHIGAN
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Test Update 685

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PDGFB (22q13) Rearrangement by FISH

Order Code: MPDGB

Pro Fee Code: 88377-26

Effective October 2, 2019, MLabs will offer PDGFB (22q13) Rearrangement by FISH (MPDGB).

Test Usage: PDGFB (22q13) encdes the preprotein for platelet-derived growth factor, beta chain. Rearrangements between PDGFB and the gene for collagen type 1, alpha-1 chain preprotein (COL1A1; 17q21) occur in more than 90% of dermatofibrosarcoma protuberans (DFSP) as well as in giant cell fibroblastoma (now considered a juvenile form of DFSP). This rearrangement results in the fusion of a variable number of exons from the 5' end of COL1A1 with all of PDGFB except for exon 1. The COL1A1 promoter drives expression of this fusion transcript whose protein is ultimately proteolytically processed into normal PDGFB ligand. The increased expression of PDGFB results in autocrine activation of its receptor (PDFRB) on surface of neoplastic cells, thereby driving tumorigenesis. The detection of PDGFB rearrangements can be useful in diagnosing DFSP – including uncommon histologic variants – and in distinguishing this sarcoma from other mesenchymal tumors that may be considered in the differential diagnosis such as cellular dermatofibroma. Of note, rare alternative rearrangements in DFSP including those involving PDGFD (11q22) will not be detected with this assay. Preclinical and limited clinical studies have implicated a potential role for targeted therapy with tyrosine kinase inhibitors (e.g. imatinib) capable of blocking activation of PDGFRB for unresectable or metastatic DFSP.

Specimen Requirements: A formalin-fixed, paraffin-embedded tissue block (containing sufficient neoplastic cells) is preferred. Unstained slides (3 slides cut at 4-microns) with associated H&E-stained slide are also acceptable. Decalcified tissue or tissues with other fixatives will be accepted and the assay attempted; however, these specimens may result in failed testing due to degraded nucleic acid. Both blocks and slides should be stored at room temperature.

Methodology: Fluorescence In Situ Hybridization (FISH) Analytic Time: 3-10 Days

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