

Test Update 809

Posted Date 06/22/2022 Effective Date 06/22/2022 Test Name <u>USP6 (17p13) Rearrangement by FISH</u> Update Type <u>New Tests</u> CPT Code 88377-TC, 88377-26

NEW TEST

USP6 (17p13) Rearrangement by FISH

Order Code: MUSP6

Fee Code: NA119

CPT Code: 88377-TC, 88377-26

The MLabs Molecular Diagnostics Laboratory will begin offering USP6 (17p13) Rearrangement by FISH effective June 22, 2022. The *USP6* gene (also known as *TRE17*), localized to chromosome 17p13, encodes a protein involved in deubiquitination and regulation of vesicle-mediated transport. Rearrangements of *USP6* result in promoter swapping, driving increased *USP6* expression, altered Wnt signaling, and imbalance of osteoclastic to osteoblastic activity. *USP6* rearrangements have been described in approximately 59-69% of primary aneurysmal bone cysts (most commonly involving *CDH11*) and 86%-92% of nodular fasciitis (particularly rearrangements involving *MYH9*). So-called secondary aneurysmal bone cysts associated with giant cell tumor, chondroblastoma, osteoblastoma and fibrous dysplasia do not display these rearrangements. *USP6* rearrangements have also been described in tumors diagnosed as so-called cellular fibroma of tendon sheath as well as myositis ossificans. Detection of *USP6* rearrangements may be useful in distinguishing nodular fasciitis and primary aneurysmal bone cyst from histological mimics.

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

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