

DDIT3 (CHOP) (12q13) Gene Rearrangement by FISH

Indications for Ordering

Use in conjunction with histologic and clinical information for the diagnosis of round cell/myxoid liposarcoma

Test Description

- Fluorescence in situ hybridization (FISH) analysis on formalin-fixed, paraffin-embedded tissue
- Break-apart DNA probes flank the 12q13 locus of the *DDIT3* gene
- 100 cells evaluated from regions of tumor identified on histopathologic review of a matching hematoxylin and eosin stained section

Tests to Consider

Primary test

[DDIT3 \(CHOP\) \(12q13\) Gene Rearrangement by FISH 2007223](#)

Related test

[MDM2 Gene Amplification by FISH 2003016](#)

- Aids in the differential diagnosis between well-differentiated liposarcoma and benign lipoma
 - Individuals diagnosed with or suspected of having well-differentiated liposarcoma based on tissue morphology

Disease Overview

Incidence

Liposarcomas account for 10-16% of all soft tissue sarcomas

- Myxoid and round cell liposarcomas account for 50% of all liposarcomas

Diagnostic/prognostic issues

- Myxoid and round cell liposarcomas may be pathologically confused with a variety of neoplasms, including
 - Myxoid malignant fibrous histiocytoma
 - Myxoma
 - Myxoid chondrosarcoma
- Round cell differential also includes synovial and rhabdomyosarcomas
- Differentiation of these tumors from lipoblastoma in children is imperative because there is little malignant potential in lipoblastomas
- Differentiation of these tumors from possible other neoplasms is important prognostically and therapeutically

Genetics

Gene – *DDIT3*

Variants

DDIT3 gene can fuse with *FUS* (16p11) or *EWS* (22q12) to form a complex translocation

- Not found in lipoblastoma

Test Interpretation

Results

- Positive – *DDIT3* rearrangement detected in $\geq 25\%$ of nuclei
 - Round cell/myxoid liposarcoma likely
- Negative – *DDIT3* rearrangement not detected
 - Does not exclude diagnosis of round cell/myxoid liposarcoma

Limitations

- Results may be compromised if the recommended fixation procedures have not been followed
- Cannot be used to assess dedifferentiation of liposarcomas