**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
- 50-100 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 3001304**

**Related Tests**
- **MDM2 Gene Amplification by FISH 3001301**
  - 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

- **FUS (16p11) Gene Rearrangement by FISH 3000548**
  - Use in conjunction with histologic evaluation
  - Positive FUS rearrangement may support the diagnosis of low-grade fibromyxoid sarcoma (LGFMS)

**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 ≥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