

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

## FOR THE DIAGNOSIS OF ROUND CELL/MYXOID LIPOSARCOMA

### Test Highlights

- *DDIT3* (*CHOP*) FISH is a sensitive and specific method for the detection of *DDIT3* gene rearrangements, which aid in the diagnosis of round cell/myxoid liposarcoma.

### Disease Overview

- Liposarcoma is one of the most frequent sarcomas in adults, representing 10–16 percent of soft-tissue sarcomas.
- Myxoid and round cell liposarcomas account for 50 percent of all liposarcomas. Myxoid and round cell liposarcomas may be pathologically confused with a variety of neoplasms, including myxoid MFH, myxoma, and myxoid chondrosarcoma.
- Differential diagnosis of round cell liposarcoma also includes synovial sarcoma and rhabdomyosarcoma.

### Epidemiology

Round cell/myxoid liposarcoma is a relatively uncommon neoplasm, with approximately 500 new cases diagnosed each year in the United States.

### Genetics

Round cell/myxoid liposarcoma is strongly associated with a t(12;16) or t(12;22) translocation; both involve the *DDIT3* gene on chromosome 12.

### Pathophysiology

- The exact cause of round cell/myxoid liposarcoma is not known. Most patients with myxoid liposarcoma have an acquired t(12;16) or t(12;22) translocation, both of which involve the *DDIT3* gene on chromosome 12. These translocations can increase the rate of cell division and growth.
- The distinction of round cell/myxoid liposarcoma from other sarcomas is important and provides prognostic and therapeutically relevant information.

### Indications for Ordering

Patients diagnosed with or suspected of having round cell/myxoid liposarcoma based on morphology or immunophenotypic studies.

### Contraindication

This test is not recommended for detection of minimal residual disease.

### Additional Ordering Notes

- The biopsy site and fixative used should be provided.
- The submitted sample should contain sufficient viable tumor.

### Interpretation

The presence of a *DDIT3* gene rearrangement is strongly supportive of a diagnosis of round cell/myxoid liposarcoma.

### Limitations

Tissues fixed in alcohol-based or non-formalin fixatives have not been tested using this method. The *DDIT3* FISH probe will detect rearrangements that involve 12q13 but will not identify the specific translocation partner.

### Methodology

The detection of *DDIT3* gene rearrangements in formalin-fixed, paraffin-embedded tissue uses a commercially available DNA FISH probe.

### References

1. Vysis<sup>®</sup> LSI *CHOP* Dual Color Breakapart Probe. Package insert. 2010. Des Plaines, IL: Abbott Molecular.
2. Antonescu CR. The role of genetic testing in soft tissue sarcoma. *Histopathology* 2006;48:13–21.

## Test Information

0049378

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For specific collection, transport, and testing information, refer to the ARUP website at [www.aruplab.com](http://www.aruplab.com).

For information on test selection, ordering, and interpretation, refer to ARUP Consult® at [www.arupconsult.com](http://www.arupconsult.com).

### AUTHOR

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