Overview

Useful For
Genomic characterization of tumor for copy number imbalances and loss of heterozygosity

Assisting in the diagnosis and classification of malignant neoplasms

Evaluating the prognosis for patients with malignant tumors

Testing Algorithm
This test does not include a pathology consult. If a pathology consultation is requested, PATHC / Pathology Consultation should be ordered and the appropriate FISH test will be ordered and performed at an additional charge.

Hematoxylin and eosin stain review of the paraffin-embedded sample is performed to identify the area of invasive tumor prior to DNA extraction and microarray analysis. If additional FISH testing is requested, it will be performed at an additional charge.

If a fresh specimen or specimen in fixative is submitted, ANPAT / Anatomic Pathology Consultation, Wet Tissue will be added by the laboratory, at an additional charge, to facilitate the performance of this test. See Aggressive B-cell Lymphoma Diagnostic Algorithm in Special Instructions.

Special Instructions
- Cytogenetic Analysis of Glioma
- Aggressive B-cell Lymphoma Diagnostic Algorithm

Method Name
Chromosomal Microarray (CMA) Using Applied Biosystems (Affymetrix) Oncoscan

NY State Available
Yes

Specimen

Specimen Type
Varies

Additional Testing Requirements
If a fresh specimen or specimen in fixative is submitted, ANPAT / Anatomic Pathology Consultation, Wet Tissue will be added by the laboratory, at an additional charge, to facilitate the performance of this test.

Necessary Information
A reason for referral and pathology report are required in order for testing to be performed. Send information with specimen. Acceptable pathology reports include working drafts, preliminary pathology or surgical pathology reports.

Specimen Required
Submit only 1 of the following specimens:
Specimen Type: Tissue

Container/Tube: Formalin-fixed, paraffin-embedded tumor tissue block

Specimen Type: Slides

Specimen Volume: 10 Consecutive, unstained, 5-micron-thick sections placed on positively charged slides and 1 hematoxylin and eosin-stained slide

Forms

If not ordering electronically, complete, print, and send an Oncology Test Request (T729) with the specimen.

Specimen Minimum Volume

See Specimen Required

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Stability Information

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<th>Specimen Type</th>
<th>Temperature</th>
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Clinical and Interpretive

Clinical Information

The importance of identifying chromosome abnormalities in malignant neoplasms is well established, and often provides important diagnostic, prognostic, and therapeutic information critical to proper patient management. Although many chromosomal abnormalities are large enough to be detected with conventional chromosome analysis, many others are below its limits of resolution, and conventional chromosome analysis does not detect copy-neutral loss of heterozygosity.

Chromosomal microarray (CMA) improves the diagnostic yield to identify genetic changes that are not detected by conventional chromosome analysis or FISH studies. CMA utilizes copy number probes and single nucleotide polymorphism probes to detect copy number changes and regions of copy-neutral loss of heterozygosity.

CMA analysis is appropriate to identify gain or loss of chromosome material throughout the genome at a resolution of 50 to 100 kilobases. CMA can:

- Define the size, precise breakpoints, and gene content of copy number changes to demonstrate the complexity of abnormalities
- Characterize unidentified chromosome material, marker chromosomes, and DNA amplification detected by conventional chromosome and FISH studies
- Determine if apparently balanced chromosome rearrangements identified by conventional chromosome studies
have cryptic imbalances

-Assess regions of copy-neutral loss of heterozygosity, which is common in neoplasia and often masks homozygous mutations involving tumor suppressor genes

The limit of detection is dependent on size of the abnormality, type of abnormality (deletion or duplication) and DNA quality. When a deletion or duplication exceeds the reporting limits, mosaicism can confidently be detected as low as 25% and may be lower if the abnormality is large and DNA quality is good.

Reference Values
An interpretive report will be provided.

Interpretation
The interpretive report describes copy number changes and any loss of heterozygosity that may be associated with the neoplastic process. Abnormal clones with subclonal cytogenetic evolution will be discussed if identified.

The continual discovery of novel copy number variation and published clinical reports means that the interpretation of any given copy number change may evolve with increased scientific understanding.

Although the presence of a clonal abnormality usually indicates a neoplasia, in some situations it may reflect a benign or constitutional genetic change. If a genetic change is identified that is likely constitutional and clearly pathogenic (eg, XYY), follow-up with a medical genetics consultation may be suggested.

The absence of an abnormal clone may be the result of specimen collection from a site that is not involved in the neoplasm, or may indicate that the disorder is caused by a point mutation that is not detectable by chromosomal microarray (CMA).

CMA, FISH, and conventional cytogenetics are to some extent complementary methods. In some instances, additional FISH or conventional cytogenetic studies will be recommended to clarify interpretive uncertainties.

See Cytogenetic Analysis of Glioma in Special Instructions for common questions and answers.

Cautions
This test is not approved by the FDA and it is best used as an adjunct to existing clinical and pathologic information.

This test does not detect balanced chromosome rearrangements such as reciprocal translocations, inversions, or balanced insertions.

This test does not detect point mutations, small deletions or insertions below the resolution of the assay, or other types of mutations such as epigenetic changes.

This test may not detect mosaic abnormalities in a minor proportion of cells, as such it is not recommended for minimal residual disease monitoring or for specimens with tumor proportions less than approximately 20% of sample.

The results of this test may reveal incidental findings unrelated to the original reason for referral.

Supportive Data
The chromosomal microarray was validated on the Affymetrix OncoScan platform in a study of 50 specimens from a variety of tumors including glioma, breast, and melanoma. Results were correlated with the pathology report, FISH, or other results.

Clinical Reference


**Performance**

**Method Description**

The selection of tissue and the identification of invasive tumor on the hematoxylin and eosin (H and E)-stained slide are performed by a pathologist. Using the H and E slide as a reference, the target areas are marked on the unstained slide, the DNA is extracted from the tumor is labeled and hybridized to the microarray. Following hybridization, the microarray is scanned and the intensity of signals is measured and compared to a reference data set. These data are used to determine copy number changes and regions with loss of heterozygosity. Chromosomal microarray data alone does not provide information about the structural nature of an imbalance. Thus, it may be of benefit to utilize FISH or additional techniques to further characterize a patient sample.(Unpublished Mayo method)

**PDF Report**

No

**Day(s) and Time(s) Test Performed**

Specimens are processed Monday through Sunday.

Results reported Monday through Friday, 8 a.m.-5 p.m.

**Analytic Time**

10 days

**Maximum Laboratory Time**

21 days

**Specimen Retention Time**

Slides and H and E used for analysis are retained by the lab indefinitely. Client provided paraffin blocks and extra unstained slides (if provided) will be returned after testing is complete.

**Performing Laboratory Location**

Rochester

**Fees and Codes**

**Fees**

- Authorized users can sign in to Test Prices for detailed fee information.
- Clients without access to Test Prices can contact Customer Service 24 hours a day, seven days a week.
- Prospective clients should contact their Regional Manager. For assistance, contact Customer Service.

**Test Classification**

This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with
CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

**CPT Code Information**
81277

**LOINC® Information**

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