

Overview

Useful For

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

Assisting in the diagnosis and classification of malignant neoplasms, including hematolymphoid malignancies

Evaluating the prognosis for patients with malignant tumors

Testing Algorithm

DNA is extracted from the specimen prior to hybridization to the microarray. An unstimulated cell culture will be set up on all specimens with adequate volume and held pending additional testing. If additional testing is requested, such as karyotype analysis or FISH, it will be performed at an additional charge.

Method Name

Chromosomal Microarray (CMA) Using Applied Biosystems (Affymetrix) Cytoscan HD

NY State Available

Yes

Specimen

Specimen Type

Varies

Additional Testing Requirements

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

Necessary Information

1. A reason for referral must be provided for testing to be performed.
2. A pathology report should accompany the specimen. If this information is not available at the time of order, submit as soon as possible for appropriateness of testing and to aid in interpretation of results.

Specimen Required

Submit only 1 of the following specimens:

Specimen Type: Tumor biopsy

Supplies: Hank's Solution (T132)

Container/Tube: Sterile container with sterile Hank's balanced salt solution (T132), Ringer's solution, or normal saline

Specimen Volume: 0.5-3 cm(3) or larger

Specimen Type: Lymph node

Supplies: Hank's Solution (T132)

Container/Tube: Sterile container with sterile Hank's balanced salt solution (T132), Ringer's solution, or normal saline.

Specimen Volume: 1 cm(3)

Specimen Type: Skin biopsy

Supplies: Hank's Solution (T132)

Container/Tube: Sterile container with sterile Hank's balanced salt solution (T132), Ringer's solution, or normal saline.

Specimen Volume: 4-mm diameter

Collection Instructions:

1. Wash biopsy site with an antiseptic soap.
2. Thoroughly rinse area with sterile water.
3. Do not use alcohol or iodine preparations.
4. A local anesthetic may be used.
5. Biopsy specimens are best taken by punch biopsy to include full thickness of dermis.

Forms

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

Specimen Minimum Volume

Tumor Biopsy: 3 cm(3)

Lymph Node: 1 cm(3)

Skin Biopsy: 4 mm diameter

Reject Due To

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

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Varies	Ambient (preferred)		
	Refrigerated		

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 greater than 1.9 million copy number probes and approximately 750,000 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 30 to 60 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.

Cautions

This test is not approved by the US Food and Drug Administration 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 CytoScan HD platform in a study of 58 specimens from a variety of tumors including liver, breast, kidney, lung, and brain. Results were correlated to previous conventional karyotype and FISH analysis, when available.

Clinical Reference

1. Cooley L, Lebo M, Li M, et al: American College of Medical Genetics and Genomics technical standards and guidelines: microarray analysis for chromosome abnormalities in neoplastic disorders. *Genet Med* 2013;15:484-494
2. Ciriello G, Miller ML, Aksoy BA, et al: Emerging landscape of oncogenic signatures across human cancers. *Nat Genet* 2013 Sep 26;45(10):1127-1133

Performance

Method Description

DNA 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

Any remaining specimen is discarded at the time results are reported.

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

Test ID	Test Order Name	Order LOINC Value
CMAT	Chromosomal Microarray, Tumor	In Process

Result ID	Test Result Name	Result LOINC Value
54729	Result	62356-1
54730	Nomenclature	62356-1
54728	Result Summary	50397-9
54731	Interpretation	69965-2
CG905	Reason for Referral	42349-1
54743	Specimen	31208-2
54732	Source	31208-2
54733	Method	49549-9
53424	Additional Information	48767-8
54734	Released By	18771-6