MAYO CLINIC LABORATORIES

Test Definition: CHRTI

Chromosome Analysis, Skin Biopsy

Overview

Useful For

Diagnosis of mosaic congenital chromosome abnormalities, including mosaic aneuploidy and mosaic structural abnormalities

Subsequent chromosome analysis when results from peripheral blood are inconclusive

Reflex Tests

Test Id	Reporting Name	Available Separately	Always Performed
_M15A	Metaphases, 1-14	No, (Bill Only)	No
_M19	Metaphases, 15-20	No, (Bill Only)	No
_MG19	Metaphases, >20	No, (Bill Only)	No
_KTG2	Karyotypes, >2	No, (Bill Only)	No
_STAC	Ag-Nor/CBL Stain	No, (Bill Only)	No

Testing Algorithm

This test includes a charge for cell culture of fresh specimens and professional interpretation of results. Analysis charges will be incurred for total work performed, and generally include 2 banded karyograms and the analysis of 20 metaphase cells. If no metaphase cells are available for analysis, no analysis charges will be incurred. If additional analysis work is required, additional charges may be incurred.

Special Instructions

- Informed Consent for Genetic Testing
- Informed Consent for Genetic Testing (Spanish)

Method Name

Cell Culture followed by Chromosome Analysis

NY State Available

Yes

Specimen

Specimen Type

Tissue

Shipping Instructions

Advise Express Mail or equivalent if not on courier service.

Necessary Information

Provide a reason for testing with each specimen. The laboratory will not reject testing if this information is not provided, but appropriate testing and interpretation may be compromised or delayed.

Specimen Required

Specimen Type: Skin biopsy

Container/Tube: Sterile container with sterile RPMI transport media, Ringer's solution, or normal saline-RPMI transport media (T095-Petri dish is not needed for this test).

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

New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:

-Informed Consent for Genetic Testing (T576)

-Informed Consent for Genetic Testing-Spanish (T826)

Specimen Minimum Volume

4 mm punch biopsy

Reject Due To

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

Specimen Stability Information

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

Clinical & Interpretive

Clinical Information

Chromosomal abnormalities cause a wide range of disorders associated with birth defects and congenital diseases. Usually, the abnormalities can be demonstrated in peripheral blood, which is readily available. Chromosome analysis on skin fibroblasts may be indicated when the results from peripheral blood are inconclusive or in clinical circumstances such as suspected cases of chromosome mosaicism, confirmation of new chromosome disorders, or some dermatological disorders.

Subtle structural chromosomal anomalies can occasionally be missed.

Chromosomal mosaicism may be missed due to statistical sampling error (rare).

Reference Values

An interpretative report will be provided.

Interpretation

When interpreting results, the following factors need to be considered:
-Some chromosome abnormalities are balanced (no apparent gain or loss of genetic material) and may not be associated with birth defects. However, balanced abnormalities often cause infertility and, when inherited in an unbalanced fashion, may result in birth defects in the offspring.
-A normal karyotype (46,XX or 46,XY with no apparent chromosome abnormality) does not eliminate the possibility of birth defects such as those caused by submicroscopic cytogenetic abnormalities, molecular mutations, and environmental factors (ie, teratogen exposure).

It is recommended that a qualified professional in Medical Genetics communicate all results to the patient.

Cautions

Interfering factors:
-Transport time should not exceed 2 days.
-Inadequate amount of fluid may not permit adequate analysis.
-Improper packaging may result in broken, leaky, and contaminated specimen during transport.
-Exposure of the specimen to temperature extremes (freezing or > 30 degrees C) may kill cells and interfere with attempts to culture cells.

Clinical Reference

1. McKinlay Gardner RJ, Amor DJ, eds. Gardner and Sutherland's Chromosome Abnormalities and Genetic Counseling, 5th ed. Oxford Monographs on Medical Genetics, 2018
2. Gersen S, Keagle M. The Principles of Clinical Cytogenetics. Springer; 2013
3. Azcona C, Bareille P, Stanhop R. Lesson of the week: Turner's syndrome mosaicisim in patients with a normal blood lymphocyte karyotype. BMJ. 1999;318:856-857
4. Woods CG, Bankier A, Curry J, et al. Asymmetry and skin pigmentary anomalies in chromosome mosaicism. J Med Genet. 1994;31(9):694-701
5. Ribeiro Noce T, de Pina-Neto JM, Happle R. Phylloid pattern of pigmentary disturbance in a case of complex mosaicism. Am J Med Genet. 2001;98(2):145-147

Performance

Method Description

TThe specimen is cut into small pieces and treated with enzymes. The tissue is then placed into tissue culture dishes with Chang and MEM-alpha-medium containing 10% fetal bovine serum and antibiotics to establish a fibroblast culture. The fibroblasts are exposed to ethidium bromide, colcemid, and hypotonic solution and fixed with glacial acetic acid and methanol. Metaphase preparations are routinely stained by G-banding, but other staining methods are frequently employed as needed. At least 20 metaphases are examined. Minimal evidence for the presence of an abnormality is defined as 2 or more metaphases with the same structural abnormality or chromosome gain (trisomy), or 3 or more

metaphases lacking the same chromosome. Five to 10 metaphases are captured using a computer-based imaging system and karyograms are prepared from 2 or more representative metaphases.(Arsham MS, Barch MJ, Lawce HJ, eds. The AGT Cytogenetics Laboratory Manual. 4th ed. John Wiley and Sons; 2017)

PDF Report
No

Day(s) Performed
Monday through Friday

Report Available
23 to 24 days

Specimen Retention Time
4 weeks

Performing Laboratory Location
Mayo Clinic Laboratories - Rochester Main Campus

Fees & 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 account representative. 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. It has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information
88233, 88291- Tissue culture for skin/biopsy, Interpretation and report
88262 w/modifier 52-Chromosome analysis less than 15 cells(if appropriate)
88262-Chromosome analysis with 15 to 120 cells (if appropriate)
88262, 88285-Chromosome analysis with greater than 20 cells (if appropriate)
88280-Chromosome analysis, greater than 2 karyotypes (if appropriate)
88283-Additional specialized banding technique (if appropriate)

LOINC® Information

Test ID	Test Order Name	Order LOINC® Value
CHRTI	Chromosomes, Skin Biopsy	62353-8

Result ID	Test Result Name	Result LOINC® Value
52311	Result Summary	50397-9

52313	Interpretation	69965-2
52312	Result	82939-0
CG768	Reason for Referral	42349-1
52314	Specimen	31208-2
52315	Source	31208-2
52317	Method	85069-3
52316	Banding Method	62359-5
54642	Additional Information	48767-8
52318	Released By	18771-6