Department of Medical and Molecular Genetics
Division of Diagnostic Genomics

Laboratory Test Directory

Chromosome Analysis – Tissue (Skin Biopsy)

CPT Code(s): 88233, 88261

Service Code (IU Health): 53100384, 53100574

Ordering Recommendation: Detection of genetic chromosomal abnormalities in patients with suspected mosaicism and/or when peripheral blood specimen is of poor quality/inconclusive or sampling is not possible. To identify translocations, duplications, deletions, inversions, mosaicism, marker chromosomes, and numerical aberrations.

Synonyms: Skin biopsy, mosaicism, mosaic study, chromosome

Methodology: Tissue culture, microscopic analysis of G-banded chromosomes.

Performed: Monday through Saturday

Reported: 21 days

Specimen Requirements

Patient Preparation: None

Collect: Skin punch/surgery skin specimen aseptically in a sterile, screw-top container filled with sterile transport media (provided upon request).

Specimen Volume: 3-10 mm skin biopsy.

Storage/Transport: Refrigerate in transport media (which can be provided) or sterile screw-top container with sterile media.

Unacceptable Conditions: Frozen or formalin-fixed.

Remarks: If sample is for culturing only for send out testing, a completed requisition form appropriate for the requested testing must accompany the sample.

Stability: Refrigerated: 48 hours; Frozen: Unacceptable; Formalin-fixed: Unacceptable.
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Interpretive Data

**Characteristics:**
Negative: A 46,XX or 46,XY karyotype indicating no apparent chromosomal abnormality is considered negative.
Positive: Identification of any numerical or structural chromosomal abnormality. A report detailing interpretation of results will be provided.

**Limitations:** A normal karyotype, i.e. 46,XX or 46,XY with no apparent chromosome abnormality, does not eliminate the possibility that the birth defect may be caused by submicroscopic cytogenetic lesions, molecular mutations, and/or environmental factors such as exposure to teratogens. Living cells are required for chromosome analysis. As such, sample quality may affect the turnaround time.