Chromosome Analysis: Skin/Other Tissue

Test Code: CSKNC
Turnaround time: 10 days - 30 days
CPT Codes: 88233 x1, 88262 x1, 88291 x1

Condition Description

This test will detect abnormalities in chromosome number and large deletions/duplications of chromosome material as well as balanced chromosome rearrangements. It can also detect mosaicism, which is often an indication for skin chromosome analysis.

For most indications for cytogenetic testing (intellectual disability, developmental delays, autism spectrum disorders, multiple congenital anomalies, etc.) a chromosomal microarray has replaced the G-banded karyotype as the first-tier diagnostic test. For more information about the benefits of the microarray, please click here. Additionally, skin samples often do not culture well. The microarray does not use cultured cells whereas standard G-banded analysis requires cultured cells.

Indications

Chromosome analysis performed on a tissue biopsy is warranted when mosaicism is suspected and not found in a peripheral blood sample.

Methodology

Chromosome analysis on non-cancerous solid tissue.

Detection

Chromosome analysis by ISCN and ACMG guidelines; minimum band resolution of 450.

Specimen Requirements

Additional Specimen Collection/Handling Instructions Required for this Test

Tissue fixed in formalin cannot be used.

Type: Tissue Biopsy

Specimen Requirements:

Obtain 1-2 cm piece of skin and place in sterile container with EGL transport media or other sterile culture media.

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample overnight at room temperature for receipt at EGL within 24 hours of collection.

Related Tests

- The EmArray 60K (VA) may detect microdeletions/duplications that are not visible on a peripheral blood chromosome analysis.
- When mosaicism is suspected but not detected on a skin chromosome analysis, a chromosome analysis for mosaicism (MM) in peripheral blood may be warranted.
- If there is a known chromosome abnormality in the family, such as a translocation, a targeted, family member chromosome study (FS) may be indicated.