Chromosome Analysis – Blood Breakage Study

CPT Code(s): 88230, 88248, 88249

Service Code (IU Health): 53100293, 53100491, 53100525

Ordering Recommendation: Investigation of possible chromosomal breakage in patients exposed to various agents known to cause chromosome damage.

Synonyms: DEB, Breakage syndrome, Fanconi anemia

Methodology: Tissue culture, breakage analysis.

Performed: Monday through Saturday

Reported: 7-10 days

Specimen Requirements

Patient Preparation: None

Collect: Whole blood, Green (Sodium Heparin).

Specimen Volume: 10-15 mL.

Storage/Transport: Room temperature. Do not freeze or expose to extreme temperatures.


Remarks: Patients with low white blood cell counts may have a greater chance of testing failure.

Stability: Ambient: 48 hours; Refrigerated: 48 hours; Frozen: Unacceptable.

Interpretive Data
Characteristics: Patients with Fanconi anemia will have breakage values from 1.06-23.9 mean breaks/cell in the 0.1 μg/mL DEB culture. Normal control individuals should have from 0.00 to 0.10 breaks/cell in the 0.1 μg/mL DEB culture. Chromosomal breakage in unstressed cultures from both Fanconi positive individuals and normal control individuals, may not differ significantly.

Inheritance: Autosomal recessive and X-linked recessive

Cause: Fanconi anemia is a genomic instability disorder caused by mutations in genes regulating replication dependent removal of interstrand DNA crosslinks. Sixteen FA or FA-like genes have been identified - FANCA, FANCB, FANCC, FANCD1 (BRCA2), FANCD2, FANCE, FANCF, FANCG, FANCI, FANCJ, FANCL, FANCM, FANCN, FANCP, RAD51C, and XPF. All of these genes except FANCB are autosomal, FANCB is on the X chromosome.

Incidence: 1 in 131,000 births in the U.S

Clinical sensitivity: Induction of chromosome breaks by DEB is considered diagnostic in FA testing.

Limitations: This does not eliminate the possibility of low frequency mosaicism or small structural abnormalities. Living cells are required for chromosome analysis. As such, sample quality can affect the turnaround time. 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.

References: