
Lab Dept: **Anatomic Pathology**

Test Name: **CHROMOSOME, BLOOD, BREAKAGE SYNDROME**

General Information

Lab Order Codes: BREAK

Synonyms: Spontaneous Chromosomal Breakage

CPT Codes: 88230 – tissue culture for non-neoplastic disorders
88248 – Chromosome analysis for breakage syndromes; baseline breakage, score 50-100 cells, count 20 cells, 2 karyotypes (e.g., for ataxia telangiectasia, Fanconi anemia, fragile X).
88291 – Chromosome analysis, interpretation and reporting

Test Includes: Determined by laboratory director based on clinical information.

Logistics

Test Indications: Evaluation of baseline (sometimes referred to as spontaneous) chromosomal breakage may be indicated in patients who:

Have had intentional or accidental exposure to radiation or other clastogen exposure.

Are suspected of having a chromosomal instability disorder such as ataxia telangiectasia or Nijmegen Breakage Syndrome.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: Fairview University of Minnesota Medical Center – Cytogenetics

(UM Test Code: LAB4692/BLBRCG)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: Results within 7-21 days

Special Instructions: Special tube required. [See Container](#), contact the laboratory for appropriate tube.

Specimen

Specimen Type:	Whole blood
Container:	Green top (Sodium Heparin), no gel, tube
Draw Volume:	5 mL (Minimum: 3 mL) blood
Processed Volume:	Same as Draw Volume
Collection:	Routine blood collection
Special Processing:	Lab Staff: Do not centrifuge. Forward specimen to Send Outs. Store and ship at room temperature. Must arrive at reference lab within 24 hours of collection.
Patient Preparation:	None
Sample Rejection:	Improper storage, mislabeled or unlabeled specimen, clotted specimens, incorrect anticoagulant

Interpretive

Reference Range:	An interpretive report will be provided
Critical Values:	N/A
Limitations:	Specimens must be received in the Cytogenetics Laboratory Mon-Fri by 5:30 pm. Weekends and holidays by 4:30 pm. Specimens received after these cutoffs will be processed the following day.
Methodology:	Chromosome breakage analysis; determined by laboratory director based on clinical information.
References:	Fairview University Laboratories April 2023
Updates:	04/20/2023: Updated optimal and minimum specimen volume, CPT codes, reference lab code, and methodology