

**Beth Israel Deaconess Medical Center**  
**Cytogenetics Laboratory For Fanconi Anemia Testing**  
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**FANCONI ANEMIA - CHROMOSOME BREAKAGE TEST FOR FIBROBLAST CULTURE  
SUBMISSION INSTRUCTIONS**

A negative MMC/DEB-induced chromosomal breakage test for Fanconi anemia performed on peripheral blood lymphocytes does not necessarily rule out the diagnosis of Fanconi Anemia. The patient may have developed somatic mosaicism. To rule out mosaicism, a skin fibroblast culture can be used to perform the chromosomal breakage test.

**This instruction is for locations outside the BILH network.**

**Note:** *We do not accept the fibroblast culture without first performing the test on a blood sample that has been previously processed at BIDMC or DFC Fanconi lab.*

1. Notify the laboratory by phone (# 617-667-3403) before shipping.
2. Sample is processed twice a week on Tuesday and Friday afternoon. We request that the fibroblast culture to be shipped from **Monday through Wednesday** for next day delivery via FedEx priority overnight.
3. Complete the patient, referring physician and hospital/practice billing information as indicated on the requisition form.
4. The charge for Chromosome Breakage Test will be client billed directly to the referring physician or hospital.
5. Two **confluent** fibroblast cultures (**T25 flasks**) are required to send for the chromosome breakage test.
6. Fill each flask completely with media, cap tightly with a non-filter cap and sealed with ParaFilm.
7. Label each flask with the patient's full name, date of birth and/or social security number, and date of the culture set up.
8. A completed requisition must be included with the sample. The requisition form can be requested by phone or downloaded from the web site:
9. Check with FedEx safety requirements in shipping biological materials. Carefully pack the flasks in an insulated Styrofoam container to maintain constant temperature and to protect the culture against any damage during transit.
10. Samples should be sent at **ROOM TEMPERATURE** via FedEx Priority overnight to the mailing address below:

**Mailing address:**

**Attn:**

**Cytogenetics Lab – Fanconi Anemia**  
**Beth Israel Deaconess Medical Center**  
**Lab Control, Finard Rm305**  
**330 Brookline Ave. Boston MA 02215**

**Note:** *For locations within the BILH network, please refer to the EPIC procedure catalog for submission details.*