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.

**INSTRUCTIONS:**

- **We do not accept a fibroblast culture without first performing the test on a blood sample that has been processed by DFCI.**
- Send two growing confluent fibroblast cultures (T25 flasks). (Do not send the punch biopsy)
- Fill the flasks completely with media, cap tightly with a non-filter cap and seal with parafilm.
- Label each flask with the patient’s full name, and one other patient identifier, i.e. hospital record number, birth date, etc.
- Carefully pack the flasks to protect against breakage. Place in a cushioned, insulated box (i.e. so it does not get damaged in shipping). A styrofoam box works well to keep the temperature constant.
- Ship the cells FED EX priority overnight to the address below.
  - Shipments should be sent on a **Monday through Wednesday** for next day delivery.
  - Please notify the laboratory at 617.632.6302 before sending
- A completed requisition should be included with the sample. A requisition form may be downloaded from our web site: [http://research.dfci.harvard.edu/dandrealab/kinesis/template.php?page=fanconi](http://research.dfci.harvard.edu/dandrealab/kinesis/template.php?page=fanconi).
- Samples should be sent at **ROOM TEMPERATURE (not on ice)** Fed Ex Priority overnight to:
  - The Comprehensive Center for Fanconi Anemia
  - Attn: Lisa A. Moreau
  - Dana Farber Cancer Institute
  - HIM 208
  - 77 Avenue Louis Pasteur
  - Boston, MA 02115
- **Contact information:**
  - Lisa A. Moreau
  - Dana Farber Cancer Institute
  - HIM 244
  - 450 Brookline Avenue
  - Boston, MA 02215
  - Phone: 617.632.6302
  - Fax: 617.632.5757
  - Lisa_Moreau@dfci.harvard.edu

[https://www.dandrealab.org/comprehensive-center-for-fanconi-anemia.html](https://www.dandrealab.org/comprehensive-center-for-fanconi-anemia.html)