Place Patient Label Here

(Place additional labels on the back of requisition)

Chromosome Breakage Requisition

Cincinnati Children's
Cytogenetics, Molecular Genetics & TTDS Laboratories
3333 Burnet Ave. NRB 1013. Cincinnati, OH 45229-3039
For test inquiries or courier service please call:
Ph: 513-636-4474 or FAX: 513-636-4373
www.cincinnatichildrens.org/cytogenetics

□ Patient Presented for Lab Draw □ Specimen Only

Patient/Physician Information

Specimen Collection Date:	Specimen Collection Time:	Patient Name (last, First, Middle Initial)	Sex	Date of Birth:
/ /			M F	/ /

Hospital MR #	Referring Institution	Referring Physician	Physician Fax #

Call Abnormal Results To:	Phone:	Authorized Signature (**REQUIRED**):

Email Address for interim assay status reports

Billing Information (Institutional Billing Preferred- call 1-866-450-4198 for insurance/self-pay inquiries)

Institution Name	Phone:	Email Address:

Fanconi Anemia Test(s) Requested

Samples must be sent overnight to arrive Monday - Thursday for Priority Overnight delivery.
Please call 513-636-4474 if you have questions about which test(s) to order or acceptable specimen types.

Check box(es) to order testing. Tests can be run □ sequentially, □ concurrently, or □ individually.
*Default is to run tests sequentially Chromosome Breakage -> Molecular Sequencing. DNA extraction charges may apply.

Chromosome Breakage

Specimen Type for Chromosome Breakage:

- □ 5-10 mLs peripheral blood in NaHep
- □ 5-10 mLs bone marrow in NaHep
- □ Skin biopsy (Send in media or saline-formalin fixed tissue is **not** accepted)
- □ Cultured fibroblast (2) T25 flasks

❑ Chromosome Breakage

Molecular Sequencing

Specimen Type for Molecular Sequencing:

- □ 3-5 mLs peripheral blood in EDTA

- ❑ Fanconi Anemia 22 gene panel
- ❑ *FANCA* full gene sequencing
- ❑ *FANCC* full gene sequencing
- ❑ *FANCC* c.456+4A>T (IVS4+4 A>T) common Ashkenazi mutation
- ❑ *FANCG* full gene sequencing
- ❑ Family study
  Default is FA panel

Fanconi Anemia Complementation testing is available for research/investigational purposes only. Call 513-636-5998 for details.

Bloom Syndrome Test Requested

□ Sister Chromatid Exchange (SCE) analysis (3-5 mLs blood in NaHep)

□ Chromosome Breakage Disorders Sequencing Panel *(ATM, BLM, LIG4, NBN, NHEJ1)* (At least 5mLs blood in EDTA)

Indications for Testing (Indication or ICD-9 Codes REQUIRED for processing)

PHYSICAL FINDINGS:

- □ Hypopigmentation/ hyperpigmentation
- □ Short stature
- □ Dysmorphic features
- □ Limb malformation
- □ Eye anomaly
- □ Erythematous "butterfly" lesion on face

HEMATOLOGIC/ ONCOLOGIC FINDINGS:

- □ Pancytopenia
- □ Aplastic anemia
- □ Myelodysplastic syndrome (MDS)
- □ Acute myelogenous leukemia (AML)
- □ Immune deficiency
- □ Other cancer diagnosis _____

FAMILY HISTORY:

- □ Ashkenazi Jewish descent
- □ Family history of Fanconi anemia

✦ Has another family member had genetic testing? __Y __N
Name: _____
Relationship/Proband: _____
Mutation identified: _____

OTHER:

□ _____
_____
_____

Specimen handling after completion of testing - please check all that apply (N/A for internal requests)

□ After the tests above are completed, part of this sample may be left over. Normally these leftover samples would be thrown away. Instead, we would like to store them so that they can be used for research. We will not keep information like the patients name, birth date, medical record number or other information that could be used to link the sample back to the patient once it has been stored for research purposes. Please check here to indicate (1) that you have discussed this request with your patient and (2) that it is acceptable for us to keep these leftover samples.

Medical Necessity Regulations - At the government's request, the Genetics Laboratory would like to remind all physicians that when ordering tests that will be paid under federal health care programs, including Medicare and Medicaid, that these programs will pay only for those tests the relevant program deems to be (1) included as covered services (2)

Procedures for FA specimen collection and shipping to CCHMC

Samples must be sent overnight to arrive Monday through Thursday.

Please notify the following of Intent to Ship for reflex or complementation testing only:

TTDSL Lab: 513-636-5998 or 513-803-1115

ttdsl@cchmc.org

***Complementation testing done in a tiered approach:**

1. **Cell line derivation (Fresh sample may be needed)**
2. **Complementation for groups *A, C, G***
3. **Complementation for groups *E, F, L (B). FANCB* will only be tested in males. *FANCI* will only be tested in fibroblast/skin biopsy samples.**

For complementation testing, indicate the type of sample submitted:

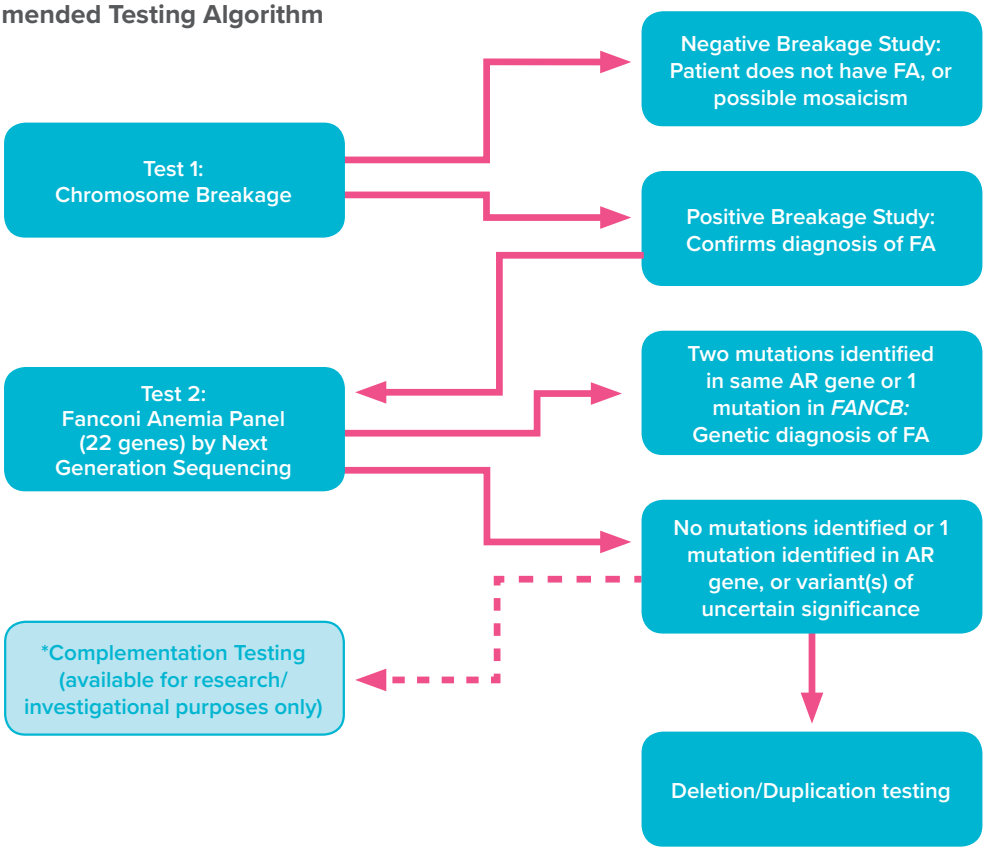
A. LCL immortalized cell line: Submit cryopreserved per standard procedure and transport on dry ice. LCL cells can also be shipped in growth media in a 15 ml tube at ambient temperature in an insulated container. Provide 20 ml extra growth media when shipped ambient.

B. Fibroblast primary cell line: Submit cryopreserved per standard procedure and transport on dry ice. Fibroblast cultures can also be shipped in a T25 flask filled with growth media at ambient temperature in an insulated container. Provide 20 ml extra growth media when shipped ambient.

C. 3 mL whole blood for EBV Transformation: Anticoagulant: ACD-B – collect in a yellow stopper Vacutainer tube. Submit ambient in an insulated container.

D. Skin biopsy for establishing a fibroblast cell line: *Cleaned biopsy, 3-4 mm (shaved of hair follicles) in transport medium comprised of Base medium (RPMI,DMEM) buffered with Sodium Bicarbonate + 0.04 mg/mL Gentamycin.* Submit ambient in an insulated container.

Recommended Testing Algorithm



This is the suggested testing algorithm. Please note that any test can be requested in any order.

*Contact 513-636-5998 for details regarding complementation testing on a research/investigational basis.