



INTERNATIONAL FANCONI ANEMIA REGISTRY (IFAR)

(This information is confidential and for research purposes only)

1. a. Today's Date: _____ b. Person completing this form: _____

2. Patient Information

a. Name _____

b. Address _____
 Street City State Zip

c. Home Telephone _____ d. Mobile Telephone _____

e. Email _____ f. Gender _____

g. Date of birth _____ h. Place of birth _____

i. If deceased, date of death _____ j. Cause of death: _____

k. Autopsy performed? Y/N

l. Race:
(Indicate all that apply) _____ American Indian _____ Asian
 _____ African American _____ Alaska Native
 _____ White _____ Native Hawaiian or Pacific Islander
 _____ Prefer not to indicate

m. Ethnicity: Hispanic or Latino? Y/N

n. Alternate contact: _____ o. Relationship to patient: _____
 (Preferable to list someone not living with the patient)

p. Address: _____

q. Telephone/email address: _____

3. Referring Physician Information

a. Referring Physician _____

b. Institution _____

c. Department: _____ d. Specialty: _____

e. Telephone _____ f. Fax _____ g. Email _____

f. Is the patient followed by any other physician(s): Y/N If yes, please state:



Name	Specialty	Hospital	Phone Number
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4. **Diagnosis**

- a. Has the patient been diagnosed with Fanconi anemia? Y/N
- b. Method of diagnosis (**Please attach report**): _____ DEB/MMC test _____ Molecular testing
- c. Date of diagnostic test: _____ d. Location (lab): _____ e. Age at diagnosis _____
- f. Is patient thought to be mosaic? Y/N

5. **Ascertainment** (please circle indication for patient to come to medical attention):

Hematologic abnormalities	Malformations	Family history
Leukemia or other cancer	Prenatal findings	Other _____

6. **Testing**

- a. Has patient had chromosome breakage studies? Yes No Pending Unknown
- b. Has patient had molecular testing for FA? Yes No Pending Unknown
- c. Has patient had complementation testing? Yes No Pending Unknown
- d. Has patient had bone marrow cytogenetic studies? Yes No Pending Unknown
- e. Has patient had other genetic testing? Yes No Pending Unknown
- f. If yes to any of the above, please give date, laboratory and result (**please enclose copy of report**)

7. **Cell lines/Publications**

- a. Have cultured fibroblast strain(s) been established from the patient? Y/N
- b. If yes, please give laboratory, and cell strain designation _____
- c. Have cultured lymphoblast strain(s) been established from the patient? Y/N
- d. If yes, please give laboratory, and cell strain designation _____
- e. Has the patient been reported in the literature? Y/N
- f. If yes, please give reference or enclose reprint _____

8. **Birth History:**

- a. Full term / / Premature / / Gestational age (in weeks) _____
- b. Complications during pregnancy _____

c. Type of delivery: Vaginal/Cesarean section Planned/Emergency



Reason for C-Section: _____

d. Measurements at birth: weight _____ (kg) (%ile _____) length _____ (m) (%ile _____)
head circumference _____ (cm) (%ile _____)

e. APGAR score(s) _____ (1 min) _____ (5 min)

f. Concerns at birth? Y/N *If yes, please circle below all that apply:*

Congenital anomalies (see #10a) IUGR/SGA Respiratory distress

Jaundice Hypotonia Meconium staining

Other: _____

9. **Growth and Development:**

a. Age (months) when: Walked _____ Talked _____

b. Typical developmental "milestones"? Yes / No, delayed /

If delayed, please comment _____

c. Typical onset of puberty and secondary sexual development?

Yes / No / if no, please comment _____ Not applicable /

Has menstruation started? Yes / Age _____ No / Not applicable /

d. Current weight _____ (kg) (%ile _____) height _____ (m) (%ile _____)

Head circumference _____ (cm) (%ile _____) Date of measurements _____

10. **Summary of Medical History** (description of treatment, date, & indicate unilateral/bilateral):

a. Abnormalities noted at birth or in childhood (if abnormality is not congenital please indicate age of onset):

1. Cardiac _____

2. CNS/Neurological (ex/structural abnormalities, learning disabilities, mental health issues etc) _____

3. Ears/Hearing _____

4. Endocrine (ex/abnormal hormone levels, etc) _____

5. Eyes/Vision (including microphthalmia) _____

6. Gastrointestinal (ex/duadonal atresia, malrotation, etc) _____

7. Genital _____

8. Growth (ex/ growth retardation, failure to thrive, microcephaly) _____

9. Kidney and urinary tract _____

10. Reproductive/Gynecological _____

11. Respiratory _____

12. Skeletal: Thumb and radius _____

Skeletal: Other _____

13. Skin (ex/birthmarks, moles, café-au-lait spots) _____



14. Other: _____

b. Has patient ever been hospitalized: Y/N Total # of hospitalizations: _____
Date admitted Date discharged Hospital Reason for hospitalization

1. _____
2. _____

c. Has patient ever had surgery: Y/N If yes, please complete the following:
Date surgery Hospital Reason for surgery Total # of surgeries: _____

1. _____
2. _____

d. Other chronic conditions? Y/N If yes, please provide details:

e. Allergies? Y/N If yes, please provide details:

f. Frequent infections? Y/N If yes, please provide details:

g. Hematologic manifestations? Y/N
If yes, please list patient's most recent blood counts from (date): _____

WBC: _____ ANC: _____ ALC: _____ HGB: _____ MCV: _____ Retic: _____ Plts: _____

Date of onset of hematologic manifestations _____ Age _____

Did the patient have any antecedent illness or medication (e.g. pneumonia, epistaxis, etc.) Y/N

If yes, please describe: _____

Treatment administered for hematologic manifestations? Y/N, If yes, complete the following:

Has the patient had any transfusions? Y/N

Total # of RBC Transfusions: _____ Total # of Platelet transfusions: _____

Date of transfusion: _____ Platelet or RBC Number of units: _____

Date of transfusion: _____ Platelet or RBC Number of units: _____

Date of transfusion: _____ Platelet or RBC Number of units: _____

Date of transfusion: _____ Platelet or RBC Number of units: _____

Androgen therapy administered? Y/N Date started: _____ Date ended: _____



Type of androgen: _____ Dose: _____

Steroid therapy administered? Y/N Date started: _____ Date ended: _____

Bone marrow transplant recipient? Y/N Date of BMT: _____

Location: _____ Type of donation: _____ BM/PSC/cord blood

Is donor a relative of the patient? Y/N If Y, relationship: _____

BMT Prep: Chemo used? Y/N Agent: _____ Dose: _____

Radiation used? Y/N Dose: _____

Immunosuppressive agent: _____ Dose: _____

h. Has patient been diagnosed with cancer? Y/N Date of diagnosis: _____ Age: _____

Cancer type: (please circle all that apply)

- Liver Lung Kidney Prostate Anal Neck
- Mouth Pharynx Esophagus Skin Breast Cervix
- Vulva Ovary Colon Blood Medulloblastoma
- Neuroblastoma Retinoblastoma Other: _____

Was cancer treatment administered? Y/N

Did patient have surgery? Y/N Date: _____ Institution: _____

Did patient receive chemo? Y/N Date: _____ Institution: _____

Agent: _____ Dose: _____ Frequency: _____

Did patient have radiation? Y/N Date: _____ Institution: _____

Radiation dose: _____ Frequency: _____

i. Vaccines: Have any age recommended vaccines been withheld from the patient? Y/N

If yes, which? _____

Vaccines received in addition to routine age-recommended vaccinations? Y/N

If yes: Name of vaccine: _____ Age received: _____

HPV vaccine received? Y/N If yes, age received: _____

j. Exposures:

Alcohol consumption? Y/N # of glasses/pints/cups/day _____ #days/week? _____

Does patient smoke tobacco? Y/N If Y, approx. # cigarettes/day _____

If Y, for how long? _____

Is sunscreen routinely used? Y/N

c. Is patient involved in other research studies? Y/N

Location of other research study: _____ PI: _____

12. **Family History:** (If pedigree is available, please enclose a copy and indicate family history of birth defects, short stature, anemia, leukemia, cancer, diabetes)

- a. Is patient adopted? _____ Y/N
- b. Patient's biological mother known? Y/N
- Mother's name _____ Date of birth _____
- Medical History _____
- Total # of pregnancies: _____ # of miscarriages: _____ # of terminations: _____

Maternal ancestry? _____

Maternal Ashkenazi Jewish ancestry? _____ Y/N

- c. Patient's biological father known? _____ Y/N
- Father's name _____ Date of birth _____
- Medical history _____
- Paternal ancestry? _____
- Paternal Ashkenazi Jewish ancestry? _____ Y/N

- d. Parental consanguinity (are parents of patient related)? _____ Y/N/Unknown
- If yes, please specify relationship _____

- e. Siblings: #full sibs with FA: _____ #full sibs without FA: _____ #half sibs without FA: _____
- List below, in order of pregnancy, all full and half siblings of patient. Please include deceased siblings, stillbirths and abortuses. For additional information, use space provided on next page.

Name	Gender	DOB	Full/Half	Has FA?	Medical history
1. _____					
2. _____					
3. _____					

- f. Children: # of biological children: _____ # of non-biological children: _____
- | Name | Gender | DOB | Biological? | Medical history |
|----------|--------|-----|-------------|-----------------|
| 1. _____ | | | | |
| 2. _____ | | | | |



3. _____

g. Known family history of FA? Y/N
If yes, who: _____

h. Have HLA studies been done in this family? Y/N

13. **Additional Information:**

- Other family history or any other information you think may be helpful. (Please include relatives with malformation, anemia, leukemia, or cancer)
- Other diagnostic, testing, or management information

Please return to IFAR
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Signature of health care provider: _____

Date: _____