Rockefeller University Institutional Review Board
IRB NUMBER: AAU-0112
IRB APPROVAL DATE: 02/23/2016
IRB EXPIRATION DATE: 02/03/2017

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

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

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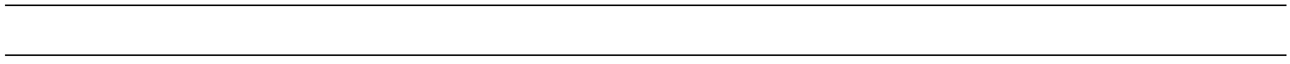

Please return to IFAR

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Signature of health care provider: ____ Date: __________________________