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
_____ African American
_____ White
_____ Prefer not to indicate
_____ Asian
_____ Alaska Native
_____ Native Hawaiian or Pacific Islander

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

_____

_____

_____

_____

_____

_____

_____

_____

Please return to IFAR

Agata Smogorzewska, MD, PhD
The Rockefeller University
1230 York Avenue Box 182
New York, NY 10065
U.S.A.
(212) 327-7850

Signature of health care provider: _____ Date: _____