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's information*

a. Patient's name_____

b. Address_____

Street City State Zip

c. Home Telephone_____ d. Mobile Telephone_____

e. Email_____ f. Gender_____

g. Date of birth_____ h. Race_____ i. Hispanic or Latino? Y/N

j. Place of birth_____

City State Country

k. If deceased, date of death_____ l. Cause of death: _____

3. *Referring Physician's Information*

a. Referring Physician Name_____

b. Institution_____

c. Department: _____ d. Specialty: _____

e. Telephone_____ f. Fax_____ g. Email_____

4. *Diagnosis*

a. Has the patient been diagnosed with Fanconi anemia? Y N

b. Method of diagnosis: DEB/MMC test Molecular testing ***Please attach report***

c. Date of diagnostic test:_____ d. Location of test:_____ e. Patient's age at diagnosis_____

f. Is patient thought to be mosaic? Y N

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

Hematologic abnormalities Malformations Family history

Leukemia or other cancer Prenatal findings Other_____

6. *Testing*

a. Has the patient had chromosome breakage studies? Yes No Pending Unknown

b. Has the patient had molecular testing for FA? Yes No Pending Unknown

c. Has the patient had complementation testing? Yes No Pending Unknown

d. Has the patient had cytogenetic studies of the bone marrow? Yes No Pending Unknown

e. Has the patient had any other genetic testing? Yes No Pending Unknown

f. If yes to any of the above, please give date, laboratory, and result **(please enclose a copy of the report)**

_____

_____

7. *Cell lines/Publications*

a. Have cultured fibroblast strain(s) been established from the patient? Yes No

b. If yes, please give laboratory, and cell strain designation._____

c. Have cultured lymphoblast strain(s) been established from the patient? Yes No

d. If yes, please give laboratory, and cell strain designation_____

e. Has the patient been reported in the literature? Yes No

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. Were there any concerns at birth: Y/N Please circle all that apply:

Congenital anomalies (see #12) IUGR/SGA Respiratory distress

Jaundice Hypotonia Meconium staining

Other: _____

9. *Growth and development:*

a. Age (in months) when walked_____ talked_____

b. Were developmental "milestones" normal? Yes /__/ or delayed /__/

If delayed, comment_____

_____

c. Was the onset of puberty and secondary sexual development normal?

Yes /__/ No /__/ Not applicable /__/ If no, comment_____

Has menstruation started? Yes /__/ No /__/ Not applicable /__/ If yes, age of onset_____

d. Current weight_____(kg) (%ile_____) height_____(m) (%ile_____)

head circumference_____(cm) (%ile_____) Date of measurements_____

10. *Summary of medical history (please give description, treatment, date, & indicate unilateral or 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 the patient ever been hospitalized: Y/N Total # of hospitalizations: _____

	Date admitted	*Date discharged*	*Hospital*	*Reason for hospitalization*
1.				
2.				

3._____

4._____

c. Has the patient ever had surgery: Y/N If so please complete the following:

Date surgery *Hospital* *Reason for surgery* Total # of surgeries: _____

1._____

2._____

3._____

4._____

d. Has the patient had hematologic manifestations? Yes No

If yes, what were the patient's most recent blood counts?

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

Date of onset of hematologic manifestations_____ Age_____

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

If yes, please describe:_____

e. Has the patient been diagnosed with cancer? Yes No

Date of cancer diagnosis: _____ Age: _____

If yes, site/type of cancer	Neck	Mouth	Pharynx	Esophagus	Skin
(please circle all that apply):	Liver	Lung	Kidney	Prostate	Anal
	Colon	Breast	Cervix	Vulva	Ovary
	Blood	Other: _____			
	Medullobastoma		Neuroblastoma		Retinoblastoma

f. Does the patient have any other chronic conditions: Y/N If yes, please provide details:

_____

_____

g. Does the patient have any allergies? Y/N If yes, please provide details:

_____

_____

h. Does the patient get frequent infections: Y/N If yes, please provide details:

_____

_____

i. If deceased, please provide information and autopsy report if available.

_____

11. *Management/Treatment:*

a. Vaccines:

1. Have any age recommended vaccines been withheld from the patient? Y/N

If yes, which? _____

2. Has the patient had any vaccines in addition to their regular age recommended ones? Y/N

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

3. Has the patient had the HPV vaccine? Y/N If yes, age of vaccination: _____

b. Has the patient had any treatment for the hematologic manifestations? Yes No NA

If yes, please complete the following:

Has the patient had any transfusions? Yes No If yes, please provide the following details:

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:_____

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

Has the patient had androgen therapy? Y/N Date started: _____ Date ended: _____

Type of androgen: _____ Dose: _____

Has patient had steroid therapy Y/N Date started: _____ Date ended: _____

Has patient had a bone marrow transplant? Y/N Date of BMT: _____

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

Was the donor a relative of the patient: Y/N If Y, relationship to proband: _____

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

Radiation used? Y/N Dose: _____

Immunosuppressive agent: _____ Dose: _____

c. Has the patient had any treatment for cancer? Yes No NA

If yes, did the patient have surgery? Y/N Date:_____ Hospital:_____

Did the patient receive chemo? Y/N Date:_____ Hospital:_____

Agent: _____ Dose: _____ Frequency: _____

Did the patient have radiation? Y/N Date:_____ Hospital:_____

Radiation dose: _____Frequency: _____

d. Is the patient followed by any other physician(s): Y/N If yes, please provide their information

_____

Name Specialty Hospital Phone Number

_____

Name Specialty Hospital Phone Number

e. Is the patient involved in any other research studies? Y N

Location of other research study: _____ PI: _____

12. *Family History: If a pedigree is available, please enclose a copy. (Medical history for the family members should include history of birth defects, short stature, anemia, leukemia, cancer, diabetes).*

a. Is the patient adopted? Y N

b. Is the biological mother known? Y N

Mother's name_____ Date of birth_____

Medical History_____

Total # of pregnancies: _____# of miscarriages: _____ # of terminations: _____

What is the mother's ancestry? _____

Does mother have any Ashkenazi Jewish ancestry? Y N

c. Is the biological father known? Y N

Father's name_____ Date of birth_____

Medical history_____

What is the father's ancestry? _____

Does the father have any Ashkenazi Jewish ancestry? Y N

d. Parental consanguinity (are the parents of the patient related)? Y N

If yes, please specify_____

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 the patient. Please include deceased siblings, stillbirths and abortuses. For additional information, use space provided on next page.

Name Gender Date of birth Full/half Has FA? Medical History

1._____

2._____

3._____

f. Children: # of biological children: _____ # of non-biological children: _____

Name Gender Biological? Date of birth Medical History

1._____

2._____

3._____

g. Is there any 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: _____