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)**

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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 *(please circle all that apply):*

Neck Mouth Pharynx Esophagus Skin

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.

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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

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Please return to IFAR

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Signature of health care provider: ____ Date: ___________________________