Fanconi Anemia Registry 01
FAR01 Initial Registration

Please ensure that the consent form is signed prior to transmitting this form!

Patient initials (first name / last name) |__|__|

Quarter and year of birth (q/yyyy) |__| / |__|__|__|__|

Sex: ☐ male ☐ female

Date signed inform consent |__|__||__|__||__|__| (dd/mm/yy)

Date initial registration |__|__||__|__||__|__| (dd/mm/yy)

Diagnosis

Reasons for FA testing

- ☐ physical abnormalities
- ☐ bone marrow failure/aplastic anemia
- ☐ myelodysplastic syndrome/acute leukemia
- ☐ solid tumor
- ☐ excessive toxicity to chemo or radiotherapy
- ☐ familial testing
- ☐ other, specify I___________________________________I

Date of FA diagnosis |__|__||__|__||__|__| (dd/mm/yy) **Please submit original report**

History of FA Patient

Pregnancy complications ☐ no ☐ yes, specify I_________________________________I

Complications at birth ☐ no ☐ yes, specify I_________________________________I

Gestational age at birth (weeks) I_____I

Birth weight (kg) I__________I

Birth height (cm) I__________I

Birth head circumference (cm) I__________I

Apgar scores 1 min I__________I 5 mins I__________I 10 mins I__________I

History of endocrine abnormalities

Thyroid hormone abnormalities ☐ no ☐ yes, specify I________________I

Growth hormone abnormalities ☐ no ☐ yes, specify I________________I

Cortisol abnormalities ☐ no ☐ yes, specify I________________I

Glucose/insulin abnormalities ☐ no ☐ yes, specify I________________

Dyslipidemia ☐ no ☐ yes, specify I________________I

Overweight/obesity ☐ no ☐ yes, specify I________________I

History of malignancy ☐ no ☐ yes, ***for details please use form Malignancy history***

For female patients

(if applicable) age at puberty (years) I_______I
(if applicable) age of menses (years) I_______I
(if applicable) age at menopause (years) I_______I
(if applicable) regular menstrual cycles ☐ no ☐ yes
Gravida (pregnancies) I_______I
Para (viable/nonviable births) I_______I
Abortus (miscarriages/abortions) I_______I

For male patients

(if applicable) age at puberty (years) I_______I

History of

gastrointestinal dysplasia ☐ no ☐ yes, specify I_______________________________I

esophageal atresia ☐ no ☐ yes, specify I_______________________________I

duodenal atresia/stenosis ☐ no ☐ yes, specify I_______________________________I

annular pancreas ☐ no ☐ yes, specify I_______________________________I

intestinal atresia ☐ no ☐ yes, specify I_______________________________I
dysplasia of other small or large intestine

☐ no ☐ yes, specify I_______________________________I

rectal atresia, anal atresia ☐ no ☐ yes, specify I_______________________________I

esophagotracheal fistula ☐ no ☐ yes, specify I_______________________________I

Surgeries ☐ no ☐ yes, specify I____I

Date of procedure /__/__//__/__//__/__/
Institution I____I
Complications related to surgery I___________________________I
specify I____I
Date of procedure /__/__//__/__//__/__/
Institution I____I
Complications related to surgery I___________________________I
specify I____I
Date of procedure /__/__//__/__//__/__/
Institution I____I
Complications related to surgery I___________________________I
specify I____I
Date of procedure /__/__//__/__//__/__/
Institution I____I
Complications related to surgery I___________________________I
specify I____I
Date of procedure /__/__//__/__//__/__/
Institution I____I
Complications related to surgery I___________________________I

Previous transfusions (initial) ☐ no ☐ yes

Red blood cells ☐ no ☐ yes Number ☐ <5 ☐ 5-10 ☐ 11-20 ☐ >20

Platelets ☐ no ☐ yes Number ☐ <5 ☐ 5-10 ☐ 11-20 ☐ >20

***A*ndrogens** ☐ no ☐ yes, please use form **Androgen Therapie** for details

Family History
(biological parents and sibling)

Family History

Father, ethnic group I___________________________I

Mother, ethnic group I___________________________I

Parents are consanguineous ☐ no ☐ yes

Previous or current cancer Father ☐ no ☐ yes, specify I___________________________I

Previous or current cancer Mother ☐ no ☐ yes, specify I___________________________I

Previous or current cancer Sibling ☐ no ☐ yes, specify I___________________________I

If cancer please submit report after obtaining approval

Vaccination history

Vaccination for HPV ☐ no ☐ yes, how often |_____|

☐ Gardasil® ☐ Cervarix®

Immunizations ☐ no ☐ yes, please specify

☐ DTPa ☐ Hib ☐ IPV ☐ HepA ☐ HepB

☐ MMR ☐ PCV ☐ MCV ☐ VZV ☐ FSME

☐ Rota

DTPa=Diphterie/Tetanus/Pertussis, Hib=Haemophilus, IPV=polio, MMR=Masern/Mumps/Röteln, PCV=Pneumokokken, MCV=Meningokokken, VZV=Varizellen, FSME=FrühSommerMeningoEnzephalitis

Physical examination at initial registration

Date of examination |__|__||__|__||__|__| (dd/mm/yy)

Current Body weight (kg) I__________I

Current height (cm) I__________I

Current head circumference (cm) I__________I

Abnormalities of the head ☐ None

☐ Head dysplasia ☐ Microcephaly ☐ Macrocephaly

☐ Cleft lip or palate ☐ Microretrognathia

☐ Macroglossia ☐ High palate ☐ Flat nose

☐ Low set ears ☐ Low hairline (hair dysplasia)

Other, specify I_____________________________I

Abnormalities of the eyes ☐ None

☐ Eye dysplasia ☐ Hypertelorism ☐ Epicanthus (eyelid dysplasia)

☐ Ptosis ☐ Strabismus ☐ Blue sclerae

☐ Congenital cataract ☐ Microphthalmia

Other, specify I________________________________I

Abnormalities of the ears ☐ None

☐ Outer ear dysplasia (left) ☐ Outer ear dysplasia (right)

☐ Middle ear dysplasia (left) ☐ Middle ear dysplasia (right)

☐ Interior ear dysplasia (left) ☐ Interior ear dysplasia (right)

Abnormalities of the neck ☐ None

☐ Neck dyplasia ☐ Short neck ☐ Torticollis ☐ Pterygium colli

Other, specify I________________________________I

Abnormalities of the skin and nails ☐ None

Skin dysplasia ☐ no ☐ yes, location, number and size ______________________________________I

Café-au-lait spots ☐ no ☐ yes, location, number and size ______________________________________I

Hyperpigmentation ☐ no ☐ yes, location, number and size ______________________________________I

Hypopigmentation ☐ no ☐ yes, location, number and size ______________________________________I

☐ Nail dystrophia ☐ Simian crease

Other, specify I________________________________I

Abnormalities of the chest and lungs ☐ None

☐ Mammary dysplasia

Other, specify I_______________________________I

Abnormalities of the urogenital system ☐ None

☐ Hypogonadism ☐ Microgenitosomia

Other, specify I_______________________________I

Abnormalities of the extremities ☐ None

☐ Extremities (incl hand/feet) dysplasia ☐ Triphalangeal thumb

☐ Polydactylia ☐ Cleft hand ☐ Hypoplasia thumb

☐ Thenar hypoplasia ☐ Radial aplasia/hypoplasia ☐ Toes/foot dysplasia

Other, specify I_______________________________I

Abnormalities of the skeletal system ☐ None

☐ Skeletal dysplasia ☐ Scoliosis ☐ Hypoplasia/aplasia of radius

☐ Scapula abduction ☐ Hip dysplasia

Other, specify I_______________________________I

Clincal features and physical examination at initial registration

If abnormal, please submit report

Abdomial Ultrasound ☐ not done ☐ done

☐ normal ☐ abnormal Date |__|__||__|__||__|__|

(splenic dysplasia, asplenia, accessory spleen, splenic atrophy, renal agenesis, dystopic kidney, dysplastic kidney, horseshoe kidney,

ECHO ☐ not done ☐ done

☐ normal ☐ abnormal Date |__|__||__|__||__|__|

(cardiovascular dysplasia, VSD, ASD, tetralogy of Fallot, stenosis/insufficiency of the mitral valve, pulmonary V. stenosis/insufficiency, aortic isthmus stenosis, pulmonary hypertension)

EKG ☐ not done ☐ done

☐ normal ☐ abnormal Date |__|__||__|__||__|__|

(cardiovascular dysplasia, VSD, ASD, tetralogy of Fallot, stenosis/insufficiency of the mitral valve, pulmonary V. stenosis/insufficiency, aortic isthmus stenosis, pulmonary hypertension)

Eye examination ☐ not done ☐ done

☐ normal ☐ abnormal Date |__|__||__|__||__|__|

(congenital cataract, glaucoma, visual acuity)

Hearing test, ENT ☐ not done ☐ done

☐ normal ☐ abnormal Date |__|__||__|__||__|__|

Gynecology exam. ☐ not done ☐ done

☐ normal ☐ abnormal Date |__|__||__|__||__|__|

Head MRI ☐ not done ☐ done

☐ normal ☐ abnormal Date |__|__||__|__||__|__|

X-rays, left hand ☐ not done ☐ done

☐ normal ☐ abnormal Date |__|__||__|__||__|__|

(Bone Age)

Other X-rays ☐ not done ☐ done, specify I______________________________

☐ normal ☐ abnormal Date |__|__||__|__||__|__|

Laboratory data

Complete blood counts

At diagnosis Date |__|__||__|__||__|__|

WBC ________ unit _______ MCV ________ unit _______

ANC ________ unit _______ Hb ________ unit _______

Platelets ________ unit _______ Reti ________ unit _______

Most recent Date |__|__||__|__||__|__|

WBC ________ unit _______ MCV ________ unit _______

ANC ________ unit _______ Hb ________ unit _______

Platelets ________ unit _______ Reti ________ unit _______

Hb-Electrophoresis prior to transfusion ☐ not done ☐ done

HbF I______I (%) Date |__|__||__|__||__|__|

Immunological testing

Leukocyte subsets ☐ no ☐ yes, Date |__|__||__|__||__|__|

Monocytes (CD14+) I______I (%) I________I (absolut/µl)

Granulocytes (CD15+ or CD66b+) I______I (%) I________I (absolut/µl)

T-cells (CD3+ CD56-) I______I (%) I________I (absolut/µl)

NK-cells (CD56+ CD3-) I______I (%) I________I (absolut/µl)

B-cells (CD19+ or CD20+) I______I (%) I________I (absolut/µl)

T-helper cells (CD3+ CD4+) I______I (%) I________I (absolut/µl)

T-suppressor cells (CD3+ CD8+) I______I (%) I________I (absolut/µl)

Activated T-cells (CD3+ HLADR+) I______I (%) I________I (absolut/µl)

Activated T-cells (CD3+ CD25R+) I______I (%) I________I (absolut/µl)

Specify results ____

Endocrine testing (result prior to initiation of therapy i.e. free T4/TSH level prior to initiation of thyroid hormone supplement or most recent results if no supplements)

Thyroid system

Free T4 ________ unit _______ Date |__|__||__|__||__|__|

TSH ________ unit _______ Date |__|__||__|__||__|__|

Oral glucose tolerance test ☐ not done ☐ done

☐ normal ☐ abnormal Date |__|__||__|__||__|__|

25OH vitamin D ________ unit _______

HbA1c ________ unit _______

Growth rate/hormone ☐ not done ☐ normal ☐ abnormal Date |__|__||__|__||__|__|

IGF-1 ________ unit _______ Date |__|__||__|__||__|__|

IGFBP-3 ________ unit _______ Date |__|__||__|__||__|__|

FSH ________ unit _______ Date |__|__||__|__||__|__|

LH ________ unit _______ Date |__|__||__|__||__|__|

Estradiol ________ unit _______ Date |__|__||__|__||__|__|

Testosterone ________ unit _______ Date |__|__||__|__||__|__|

Renal and hepatic function, if completed

AST (SGOT)	_______ unit ______	Date |__|__||__|__||__|__|
ALT (SGPT)	_______ unit ______	Date |__|__||__|__||__|__|
Total bilirubin	_______ unit ______	Date |__|__||__|__||__|__|
AP	_______ unit ______	Date |__|__||__|__||__|__|
Albumin	_______ unit ______	Date |__|__||__|__||__|__|
BUN	_______ unit ______	Date |__|__||__|__||__|__|
Creatinine	_______ unit ______	Date |__|__||__|__||__|__|

Fasting lipid profile ☐ not done ☐ done

HDL	_______ unit ______	Date |__|__||__|__||__|__|
LDL	_______ unit ______	Date |__|__||__|__||__|__|
TG	_______ unit ______	Date |__|__||__|__||__|__|

Humoral immunity ☐ no ☐ yes Date |__|__||__|__||__|__|

IgM	_______ unit ______	Date |__|__||__|__||__|__|
IgG	_______ unit ______	Date |__|__||__|__||__|__|
IgA	_______ unit ______	Date |__|__||__|__||__|__|
IgE	_______ unit ______	Date |__|__||__|__||__|__|

Virus specific serology Date |__|__||__|__||__|__|

		pos	neg	unknown			pos	neg	unknown
CMV	IgG	☐	☐	☐	Herpes Simplex	IgG	☐	☐	☐
	IgM	☐	☐	☐	HHV 6	IgG	☐	☐	☐
EBV	IgG	☐	☐	☐		IgM	☐	☐	☐
	IgM	☐	☐	☐	Parvovirus B 19	IgG	☐	☐	☐
EBV-VCA	IgG	☐	☐	☐		IgM	☐	☐	☐
	IgM	☐	☐	☐		PCR	☐	☐	☐
EBV-EA	IgG	☐	☐	☐	HCV	IgG	☐	☐	☐
EBV-EBNA	IgG	☐	☐	☐		IgM	☐	☐	☐
Anti-HBs		☐	☐	☐	HBsAg		☐	☐	☐
Anti-HBc		☐	☐	☐	HIV		☐	☐	☐

Urine analysis ☐ normal ☐ abnormal Date |__|__||__|__||__|__|

If abnormal: ☐ glucosuria ☐ hematuria ☐ proteinuria

☐ other, specify ____

Further comments: ____

Referring physician name and institution |____|

Address |____|

Telephone and fax |____|

email |____|

Date I__I__II__I__II__I__I Signature ___________________________________