

# Fanconi Anemia Treatment and Management Resource Guide

***Editors' Note and Disclaimer: The resources provided in this guide are not meant to be inclusive. The reader should not consider the resources listed here to be endorsements by the Fanconi Anemia Research Fund, Inc.***

## **Apollo Specialty Hospital**

320 "Padma Complex"

Anna Salai

Chennai 600 035

India

refrain@vsnl.net

### ◆ **Dr. Sheila Mohan, MBBS, DCH, PhD**

*Phone: 91-44-24912943 Mobile: 91-92-82114661*

### ◆ **Dr. Revathi Raj, MBBS, DCH, MRCP (Path)**

*Phone: 91-44-24331741, Ext 2509*

## **Services Offered**

- ❖ Complete evaluation, treatment and follow-up of new and previously diagnosed FA patients;
- ❖ Provide information and assistance for physicians in the care of their FA patients;
- ❖ Cancer screening and specialist consultations;
- ❖ Diagnostic testing (MMC test) and screenings of siblings;
- ❖ Complementation grouping (in process of development);
- ❖ Genetic counseling;
- ❖ Stem cell transplantation;
- ❖ Maintenance of national database: Registry for Fanconi Anemia in India (REFRAIN).

## **Charité, Campus Virchow Klinikum, University Medicine, Berlin**

Children's Hospital

Augustenburger Platz 1

13353 Berlin

Germany

### ◆ **Wolfram H. Ebell, MD**

*wolfram.ebell@charite.de*

*Phone: 49-30-450-566-014 Fax: 49-30-450-566-919*

## **Services Offered**

- ❖ Pediatric bone marrow transplant service;
- ❖ Stem cell laboratory;
- ❖ Stem cell bank;
- ❖ Cytogenetic laboratory;
- ❖ German Fanconi anemia registry.

## **Children's Hospital Medical Center, The Fanconi Anemia Comprehensive Care Center**

3333 Burnet Avenue  
Cincinnati, OH 45229

- ◆ **Richard Harris, MD**, Professor of Pediatrics, Division of Hematology/Oncology, Blood and Marrow Transplant Program, Director of Bone Marrow Failure Clinic  
*richard.harris@cchmc.org*  
Phone: (513) 636-8610 Fax: (513) 636-7951
- ◆ **Frank O. Smith, MD**, Marjory J. Johnson Endowed Chair and Professor of Pediatrics, Director, Division of Hematology/Oncology  
*frank.smith@cchmc.org*  
Phone: (513) 636-8811 Fax: (513) 636-2034
- ◆ **David Williams, MD**, Beatrice C. Lampkin Chair and Professor of Pediatrics, Director, Division of Experimental Hematology, Associate Chair for Translational Research  
*david.williams@cchmc.org*  
Phone: (513) 636-0364 Fax: (513) 636-3768
- ◆ **Robin Mueller, RN**, Nurse Coordinator, Fanconi Anemia Comprehensive Care Center  
*robin.mueller@cchmc.org*  
Phone: (513) 636-3218 Fax: (513) 636-7951
- ◆ **Parinda Mehta, MD**, Assistant Professor of Pediatrics, Division of Hematology/Oncology, Blood and Marrow Transplant Program  
*parinda.mehtar@cchmc.org*  
Phone: (513) 636-5917 Fax: (513) 636-3549
- ◆ **Stella Davies, MB BS, PhD, MRCP**, Jacob G. Schmidlapp Endowed Chair and Professor of Pediatrics, Director, Blood and Marrow Transplant Program, Division of Hematology/Oncology  
*stella.davies@cchmc.org*  
Phone: (513) 636-1371 Fax: (513) 636-3549

### **Services Offered**

- ❖ Evaluation of new and previously diagnosed patients;
- ❖ Personalized treatment regimens by specialists in bone marrow transplantation, cardiology; endocrinology, genetics, hematology, nephrology, urology, orthopedics, hand surgery, social work and family support network;
- ❖ Gene therapy, pharmacological and other clinical trials (such as with cytokines, novel androgens, and etanercept);
- ❖ Fanconi anemia repository for samples and data;
- ❖ Expertise in matched sibling donor and unrelated donor transplantation for Fanconi anemia;
- ❖ Fanconi anemia complementation analysis;
- ❖ Stem cell collection, storage, and reinfusion;
- ❖ Collaborator, International Fanconi Anemia Registry (**Arleen Auerbach, PhD**).

### **Cincinnati Hand Surgery Specialists**

10700 Montgomery Rd., Suite 150  
Cincinnati, OH 45242

- ◆ **Thomas R. Kiefhaber, MD**  
*tkiefhaber@handsurg.com*  
Phone: (513) 961-4263

## ***Cincinnati Hand Surgery Specialists (cont'd)***

### **Services Offered**

- ❖ Hand Surgery Consultant to the Cincinnati Children's Hospital, Fanconi Anemia Comprehensive Care Center;
- ❖ Reconstructive surgery for children with congenital differences of the hand.

## **Constantiaberg Medi-Clinic**

Department of Haematology and Bone Marrow Transplant Unit Incorporating the Searll Research Laboratory for Cellular and Molecular Biology

PO Box 294

Plumstead, Cape Town

South Africa, 7801

### ◆ **Professor Peter Jacobs, MD, PhD**

*haematol@icon.co.za*

*Phone: 27-21-799-2569 Fax: 27-21-761-4278*

### ◆ **Sister Lucille Wood**, Hematology Coordinator and Head Nurse

### **Services Offered**

- ❖ Adult and pediatric bone marrow and peripheral blood hematopoietic stem cell program;
- ❖ Special interest in new conditioning regimens for patients with FA;
- ❖ Participation in gene therapy project with Professor **Christopher Walsh, MD**, Mount Sinai School of Medicine, New York;
- ❖ Association with **Grover Bagby, MD**, Oregon Health & Science University, in transcriptome database;
- ❖ Basic science and translational stem cell research in the Searll Research Laboratory for Cellular and Molecular Biology.

## **Dana-Farber Cancer Institute, Comprehensive Fanconi Anemia Center**

44 Binney Street, Boston, MA 02115

### **Children's Hospital Boston, Hematology Clinic**

300 Longwood Avenue, Boston, MA 02115

### ◆ **Alan D'Andrea, MD**, Professor of Pediatrics, Director, The Comprehensive Center for Fanconi Anemia

### ◆ **Akiko Shimamura, MD, PhD**, Director, Bone Marrow Failure Clinic, Children's Hospital, Boston

*akiko.shimamura@childrens.harvard.edu*

*Phone: (617) 919-2508 Fax: (617) 730-0934*

### ◆ **Lisa A. Moreau, MS, CLSpCG**, Supervisor of Clinical Laboratory

*Phone: (617) 632-6302*

### **Services Offered**

- ❖ Clinical consultation for the diagnosis, management and treatment of FA through the Bone Marrow Failure Clinic, Division of Hematology, Children's Hospital;
- ❖ Coordination of multidisciplinary medical care;
- ❖ Diagnosis of new FA patients through Clinical Laboratory Improvement Act (CLIA) certified diepoxybutane (DEB) test;

***Dana-Farber Cancer Institute, Comprehensive Fanconi Anemia Center and Children's Hospital Boston, Hematology Clinic (cont'd)***

**Research Studies**

- ❖ FA subtyping, gene mutational screening, carrier detection, cell repository (blood, bone marrow, skin and tumor samples);
- ❖ FA therapeutics: hematopoietic stem cell transplant, gene therapy;
- ❖ Molecular mechanisms underlying FA.

**Duke University Medical Center**

Box 3350 DUMC  
2400 Pratt Street, Suite 1400  
Durham, NC 27705

- ◆ **Joanne Kurtzberg, MD**, Professor of Pediatrics and Pathology, Director, Pediatric Blood and Marrow Transplant Program, Director, Carolinas Cord Blood Bank at Duke  
*kurtz001@mc.duke.edu*  
*Phone: (919) 668-1119 Fax: (919) 668-1183*

**Services Offered**

- ❖ Pediatric bone marrow transplant program including a 16-bed inpatient unit, dedicated outpatient clinic and day hospital;
- ❖ Matched sibling, haploidentical, and unrelated stem cell transplants;
- ❖ Comprehensive evaluation and education from initial contact with the program;
- ❖ Family support throughout all phases of care;
- ❖ Umbilical cord blood bank and stem cell laboratory.

**Fred Hutchinson Cancer Research Center**

1100 Fairview Ave N, D1-100  
Seattle, WA 98109

- ◆ **Hans-Peter Kiem, MD**, Associate Member, Fred Hutchinson Cancer Research Center, Investigator, Markey Molecular Medicine Center, Adjunct Associate Professor of Pathology, Associate Professor of Medicine, University of Washington School of Medicine  
*hkiem@fhcrc.org*  
*Phone: (206) 667-4425 Fax: (206) 667-6124*

**Services Offered**

- ❖ Nonmyeloablative stem cell transplantation with related and unrelated donors;
- ❖ Collaboration with Hospital de Clinicas, University of Parana, Brazil (**Ricardo Pasquini, MD, PhD**).

**The Genesis Genetics Institute, Genomic Technology Center**

5555 Conner Ave  
Suite A-2064  
Detroit, MI 48213  
[www.genesisgenetics.org](http://www.genesisgenetics.org)

## ***The Genesis Genetics Institute, Genomic Technology Center (cont'd)***

- ◆ **Mark Hughes, MD, PhD**  
*pgd@genesisgenetics.org*  
*Phone: (313) 544-4006 Fax: (313) 544-4006*
- ◆ **Shannon Wiltse, MS, CGC, Certified Genetic Counselor**  
*counselor@genesisgenetics.org*  
*Phone: (313) 544-4006 Fax: (313) 544-4006*

### **Services Offered**

- ❖ Pre-pregnancy/pre-implantation diagnosis of FANC A, C, G. Helping a couple begin a pregnancy knowing their baby will not have Fanconi anemia;
- ❖ Extensive experience in Fanconi mutation testing with HLA identity matching for families desiring a healthy baby and cord blood stem cells that are a perfect match to their Fanconi-affected child needing stem cell transplantation;
- ❖ Personalized treatment regimens coordinated with the family's hematologist, geneticist, obstetrician, and (if necessary) transplantation doctors. The FA family does not need to travel anywhere for these PGD services. Our clinic collaborates with the leading molecular biologists in Fanconi research, with over 90 *in vitro* fertilization programs, and with internationally respected Fanconi transplantation experts.

## **Hackensack University Medical Center**

30 Prospect Avenue  
Hackensack, NJ 07601

- ◆ **Alfred P. Gillio, MD**  
*agillio@humed.com*  
*Phone: (201) 996-5600 Fax: (201) 996-0521*
- ◆ **Barbara Adler-Brecher, MS**  
*badler@humed.com*

### **Services Offered**

- ❖ Diagnosis and genetic counseling;
- ❖ Comprehensive medical treatment including gynecology and ENT specialists;
- ❖ Matched sibling and unrelated donor transplants.

## **Hadassah Hospital**

Ein Kerem  
Jerusalem 91120 Israel

- ◆ **Shimon Slavin, MD, Department of Bone Marrow Transplantation and Cancer Immunotherapy**  
*slavin@tapuz.co.il*  
*Phone: 972-2-677-7270 Fax: 972-2-642-2731 or 972-2-643-6151*  
*After hours, home: 972-2-643-0279*
- ◆ **Menachem Bitan, MD, Department of Bone Marrow Transplantation and Cancer Immunotherapy**  
*mbitan@hadassah.org.il*  
*Phone: 972-2-677-7270 Fax: 972-2-642-2731 Mobile: 972-2-642-2731*

## ***Hadassah Hospital (cont'd)***

### **Services Offered**

- ❖ Stem cell transplantation using a reduced intensity, non-myeloablative or myeloablative preparatory regimen;
- ❖ Transplantation of every patient in need, including multiply transfused, pre-leukemic, and fully leukemic patients, those with fully matched sibling, matched unrelated, partially or haploidentically mismatched family member donors utilizing stem cells derived from bone marrow, peripheral-blood or cord-blood. Several immunotherapy protocols are available for patients with resistant hematologic malignancy and metastatic solid tumors.

## **Heinrich Heine University Medical Center**

Children's Hospital

Moorenstrasse 5

40225 Düsseldorf, Germany

- ◆ **Helmut Hanenberg, MD**, Department of Pediatric Hematology, Oncology and Clinical Immunology, Department of Pediatrics, Wells Center for Pediatric Research, Indiana University School of Medicine, Indianapolis, IN  
*Hanenberg@uni-duesseldorf.de*  
*Phone: 49-211-811-61037-7680 Fax: 49-211-811-6436*
- ◆ **Eunike Velleuer**, Department of Pediatric Hematology, Oncology and Clinical Immunology  
*Velleuer@uni-duesseldorf.de*
- ◆ **Arndt Borkhardt, MD**, Director, Department of Pediatric Hematology, Oncology and Clinical Immunology  
*lesch@med.uni-duesseldorf.de*  
*Phone: 49-211-811-7680 Fax: 49-211-811-6206*

### **Services Offered**

- ❖ Primary diagnosis, blood work, transfusions and bone marrow harvests of pediatric patients with hematological, malignant or immunological disorders (also for the National Marrow Donor Program - NMDP);
- ❖ Autologous and allogenic stem cell transplantation;
- ❖ FA multidisciplinary out- and in-patient care;
- ❖ FA complementation group analysis on peripheral blood T-cells, bone marrow cells, fibroblasts and B-cell lines for groups A, B, C, D1, D2, E, F, G, L, J, M, N;
- ❖ Close collaboration with **Detlev Schindler**, University of Würzburg, on FA-related clinical and scientific issues;
- ❖ Cooperation with **Ruud Brakenhoff**, University of Amsterdam, on "Progression of FA-SCC and Development of a Non-Invasive Screening Method for Precursor Lesions";
- ❖ Preclinical testing of alternative treatment and gene therapy approaches in animal models in cooperation with **Wade Clapp**, **Laura Haneline**, **Karen Pollok** and **Kenneth Cornetta** at the Indiana University School of Medicine, Indianapolis, IN;
- ❖ Close cooperation with the two German FA family support groups Deutsche FA-Hilfe e.V. (Ralf Dietrich) and Aktionskreis Fanconi Anemia e.V.

### **Hôpital Saint-Louis**

Bone Marrow Transplant Unit  
1 avenue Claude Vellefaux  
75010 Paris  
France

- ◆ **Eliane Gluckman, MD**, Head Professor  
*eliane.gluckman@sls.ahp.fr*  
Phone, direct: 33-142-499-644  
Secretary: 33-142-499-639 Fax: 33-142-499-634
- ◆ **Jean Soulier, MD, PhD**, Associate Professor, Genome Rearrangements and Cancer Group, Hematology Laboratory  
*jean.soulier@sls.aphp.fr*  
Phone: 33-142-499-891 Fax: 33-153-722-217
- ◆ **Attending Physicians:** Pr. Gerard Socie, Dr. Agnes Devergie, Dr. Helene Esperou, Dr. Christelle Ferry, Dr. Thierry Leblanc, Dr. Vanderson Rocha

#### **Services Offered**

- ❖ Bone marrow, peripheral stem cell, and cord blood transplants;
- ❖ FA diagnosis and characterization: breakage tests, cell cycle, FANCD2 analysis, mosaicism on skin biopsy, retroviral group complementation, bone marrow karyotype (**Dr. Jean Soulier**);
- ❖ Cell therapy laboratory (**Dr. Jerome Larghero**);
- ❖ Gene therapy laboratory (**Pr. Yves Beuzard, Pr. Philippe Leboulch**);
- ❖ Head and neck cancer evaluation and treatment (**Dr. Pascal**).

### **Hospital de Clinicas, University of Parana**

Rua General Carneiro, 181 – 15º andar  
80001-970-Curitiba-PR  
Brazil

- ◆ **Ricardo Pasquini, MD, PhD**  
*pasquini@hc.ufpr.br*  
Phone: 55-41-3360-1007 Fax: 55-41-3360-1015

#### **Services Offered**

- ❖ A comprehensive clinic for FA patients;
- ❖ Out-patient unit for diagnostic procedures and treatment;
- ❖ Cytogenetics laboratory performing DEB chromosomal breakage testing and conventional cytogenetics of bone marrow;
- ❖ Transplant unit performing related and unrelated bone marrow and umbilical cord transplants.

### **Hospital Ste-Justine**

3175 Cote-Ste-Catherine  
Montréal, Québec H3T 1C5  
Canada

- ◆ **Josette Champagne, MD**, Bone Marrow Failure Program  
*champaj@medclin.umontreal.ca*

### ***Hospital Ste-Justine (cont'd)***

- ◆ **Martin Champagne, MD, FRCPC**, Bone Marrow Transplant Program  
*martin.a.champagne@umontreal.ca*  
Phone: (514) 345-4830 (appointments), (514) 345-4969 (clinical research) Fax: (514) 345-4884

#### **Services Offered**

- ❖ Diagnostic testing;
- ❖ Related and unrelated marrow and cord blood transplantation;
- ❖ Designated (sibling) cord blood storage when required;
- ❖ Diagnostic and therapeutic services restricted to Canadian citizens.

### **Memorial Sloan-Kettering Cancer Center**

1275 York Avenue  
New York, NY 10021

- ◆ **Farid Boulad, MD**, Pediatric Hematology and Bone Marrow Transplantation  
*bouladf@mskcc.org*  
Phone: (212) 639-6684 Fax: (212) 717-3447
- ◆ **Bhuvanesh Singh, MD, PhD**, Head and Neck Cancer  
*singhb@mskcc.org*  
Phone: (212) 639-2024
- ◆ **Mercedes Castiel, MD**, Gynecology  
*castielm@mskcc.org*  
Phone: (212) 639-5060

#### **Services Offered**

- ❖ Comprehensive care and long-term follow-up of new and previously diagnosed patients, including: hematopoietic stem cell transplantation with related, unrelated, matched or mismatched donors, head and neck cancer evaluation and treatment, gynecological cancer evaluation and treatment, and endocrinology clinic;
- ❖ Direct collaboration with **Arleen Auerbach, PhD**, The Rockefeller University, for diagnosis, mutation screening, complementation group analysis, and somatic mosaicism studies;
- ❖ Cell biology research on hematopoietic stem cells and progenitor populations in neoplastic and genetic diseases.

### **Mount Sinai School of Medicine**

Novel Therapeutics and Stem Cell Transplantation Program  
Division of Hematology-Oncology  
Annenberg 24-42, Box 1079  
One Gustave L. Levy Place  
New York, NY 10029

- ◆ **Christopher Walsh, MD**, Associate Professor of Medicine  
*christopher.e.walsh@mssm.edu*  
Phone: (212) 241-3554
- ◆ **Luis Isola, MD**, Director, Stem Cell Transplantation Program
- ◆ **Anne Hurllet, MD**, Associate Professor of Pediatrics

## ***Mount Sinai School of Medicine (cont'd)***

### **Services Offered**

- ❖ Adult and pediatric FA patients can be seen by specialists in adult medicine and pediatrics;
- ❖ Complete hematologic evaluation/treatment and cancer surveillance;
- ❖ Recommendations for stem cell transplantation (related and unrelated donor) at Mount Sinai School of Medicine and elsewhere;
- ❖ Future clinical trials for FA-associated myelodysplastic syndrome;
- ❖ Collaboration with **Arleen Auerbach, PhD**, The Rockefeller University, on genetic analysis;
- ❖ Complementation screening.

### **National Cancer Institute**

Clinical Genetics Branch, Division of Cancer Epidemiology and Genetics, National Institutes of Health  
6120 Executive Blvd., Executive Plaza South, Room 7020  
Rockville, MD 20892-7231  
[www.marrowsfailure.cancer.gov](http://www.marrowsfailure.cancer.gov)

#### ◆ **Blanche P. Alter, MD, MPH**

[alterb@mail.nih.gov](mailto:alterb@mail.nih.gov)

Phone: (301)594-7642 Fax: (301) 496-1854

### **Services Offered**

- ❖ Comprehensive clinical evaluation by all subspecialties;
- ❖ All clinical laboratory tests;
- ❖ Cancer screening, age and sex appropriate, such as head, neck and esophagus, and gynecological;
- ❖ Bone marrow aspirate, biopsy, and cytogenetics;
- ❖ Chromosome breakage studies in a CLIA-certified clinical laboratory;
- ❖ Genetic counseling and mutation analysis in a CLIA-certified clinical laboratory;
- ❖ Comprehensive evaluation of all immediate family members (with the exception of bone marrow studies);
- ❖ Epidemiologic study of cancer in patients and in family members.

### **National Heart, Lung, and Blood Institute**

*Building 10, CRC 3E-5140*

*10 Center Drive, MSC 1202*

*Bethesda MD 20892-1202*

*Phone: (301) 402-0764*

#### ◆ **Neal Young, MD**, Chief, Hematology Branch

#### ◆ **Richard Childs, MD**, Hematology Branch

### **Services Offered**

- ❖ Second opinion regarding diagnosis and management of constitutional bone marrow failure states.

## **Oregon Health & Science University, Doernbecher Children's Hospital**

3181 S.W. Sam Jackson Park Road  
Portland, OR 97239

- ◆ **Peter Kurre, MD**, Assistant Professor, Division of Pediatric Hematology/Oncology  
*kurrepe@ohsu.edu*  
Phone: (503) 494-1543 Fax: (503) 494-0714

### **Services Offered**

- ❖ Diagnostic evaluation and genetic counseling for FA and other marrow failure syndromes;
- ❖ Comprehensive medical treatment;
- ❖ Stem cell transplantation;
- ❖ Research studies investigating molecular diagnostics, cytogenetics, complementation group subtyping, leukemogenesis and genetics of Fanconi anemia.

## **The Reproductive Genetics Institute**

2825 N Halsted St.  
Chicago, IL 60657  
*www.reproductivegenetics.com*  
*rgi@flash.net*  
Phone: (773) 472-4900 Fax: (773) 871-5221

- ◆ **Allen Horwitz, MD, PhD**
- ◆ **Renee Genovese, MS, CGC**
- ◆ **Dana Pauling, MS, CGC**
- ◆ **Christina Lavin, MS, CGC**

### **Services Offered**

- ❖ Pre-implantation genetic diagnosis for known Fanconi mutations with or without HLA matching for sibling bone marrow donation (from cord blood).

## **The Rockefeller University**

Laboratory of Human Genetics and Hematology (Box 77)  
1230 York Avenue  
New York, NY 10021

- ◆ **Arleen D. Auerbach, PhD**  
*auerbac@mail.rockefeller.edu*  
Phone: (212) 327-7533, or 8862 Fax: (212) 327-8262  
<http://www.rockefeller.edu/labheads/auerbach/auerbach.html>

### **Services Offered**

#### ***Clinical Genetics Services***

- ❖ Postnatal diagnosis of Fanconi anemia, (FA) including reporting of somatic mosaicism;
- ❖ Prenatal diagnosis of Fanconi anemia in first trimester through chorionic villus sampling, (CVS), and second trimester through amniocentesis, or percutaneous umbilical blood sampling;

### ***The Rockefeller University (cont'd)***

- ❖ FA complementation group analysis using retroviral vectors (currently for groups FA-A, B, C, E, F, G, J);
- ❖ Mutation screening for FA families once the complementation group is known;
- ❖ Carrier detection for FA-C mutations in any individual of Ashkenazi Jewish (AJ) origin, or non-Jewish partner of a known carrier of the AJ mutation in FA-C.

### ***Genetic Services on Research Protocols***

- ❖ International Fanconi Anemia Registry;
- ❖ Incidence of cancer in FA carriers;
- ❖ DNA instability and genetic alterations in FA: A model for carcinogenesis;
- ❖ PGD/IVF: a collaboration with **Dr. Mark Hughes** (provide genetic information to enable the development of an assay for PGD);
- ❖ Collaboration with the Fanconi Anemia Gene Therapy Group (**Dr. David Williams**, et al) to provide genetic information for patients eligible for future gene therapy trials.

### **St. Jude Children's Research Hospital**

332 N. Lauderdale St.  
Memphis, TN 38104

- ◆ **Ulrike Reiss, MD**  
*ulrike.reiss@stjude.org*  
Phone: (901) 495-4753 Fax: (901) 495-2952
- ◆ **Martha Rieman, FNP**, Bone Marrow Failure Nurse Practitioner  
*martha.rieman@stjude.org*  
Phone: (901) 495-2173
- ◆ **Melody Cunningham, MD**  
*melody.cunningham@stjude.org*

### **Services Offered**

- ❖ Bone Marrow Failure Clinic;
- ❖ Comprehensive care, including subspecialties;
- ❖ Participation in FA research in conjunction with other centers.

### **Schneider Children's Hospital, Albert Einstein School of Medicine**

Bone Marrow Failure Program  
Division of Hematology/Oncology and Stem Cell Transplantation  
269-01 76<sup>th</sup> Ave., Room 225  
New Hyde Park, New York 11040

- ◆ **Adrianna Vlachos, MD**, Assistant Professor of Pediatrics, Head, Bone Marrow Failure Program and Associate Head, Stem Cell Transplant Program  
*avlachos@lij.edu*  
Phone: 718-470-3611
- ◆ **Johnson M. Liu, MD**, Les Nelkin Professor of Pediatrics, Head, Section of Experimental Hematology and Les Nelkin Memorial Pediatric Oncology Research Laboratory

### ***Schneider Children's Hospital, Albert Einstein School of Medicine (cont'd)***

- ◆ **Jeffrey M. Lipton, MD, PhD**, Professor of Pediatrics, Chief, Division of Hematology/Oncology and Stem Cell Transplantation

#### **Services Offered**

- ❖ Bone Marrow Failure Clinic offering comprehensive evaluation of adult and pediatric FA patients by an experienced team of multidisciplinary specialists directed by medical and pediatric hematologists;
- ❖ Hematopoietic Stem Cell Transplant Program offering expertise in transplantation in bone marrow failure syndromes;
- ❖ Experimental Hematology Program studying molecular mechanisms of hematopoietic failure and oncogenesis in FA and other bone marrow failure syndromes.

### **Shriners Hospital for Children**

3551 North Broad St.  
Philadelphia, PA 19140

- ◆ **Scott Kozin, MD**  
*skozin@shrinenet.org*  
*Phone: (215) 430-4034*
- ◆ **Linda Filipone, Nurse Coordinator**  
*lfilipone@shrinenet.org*  
*Phone: (215) 430-4074*

#### **Services Offered**

- ❖ Evaluation and treatment of FA children with upper limb and other orthopedic problems;
- ❖ Surgical expertise in hand and upper limb differences;
- ❖ Occupational therapy evaluation and treatment, including splints, adaptive equipment, and other needs.

### **Tokai University**

Department of Cell Transplantation and Regenerative Medicine  
Bohseidai, Isheara  
Kanagawa, 259-1193 Japan

- ◆ **Hiromasa Yabe, MD**, Stem Cell Transplantation  
*yabeh@is.icc.u-tokai.ac.jp*  
*Phone: 81-463-93-1121 Fax: 81-463-91-6235*
- ◆ **Miharu Yabe, MD**, Stem Cell Transplantation  
*miharu@is.icc.u-tokai.ac.jp*  
*Phone: 81-463-93-1121 Fax: 81-463-91-6235*
- ◆ **Shunichi Kato, MD**, Stem Cell Transplantation  
*skato@is.icc.u-tokai.ac.jp*  
*Phone: 81-463-93-1121 Fax: 81-463-91-6235*

#### **Services Offered**

- ❖ Evaluation of new and previously diagnosed patients;

### ***Tokai University (cont'd)***

- ❖ Expertise in pediatric and adult stem cell transplantation from matched sibling donor or alternative donor;
- ❖ Special interest in new conditioning regimens for patients with FA to improve long-term quality of life;
- ❖ Umbilical cord blood bank and cell processing center.

### **Universitat Autònoma de Barcelona**

Department of Genetics and Microbiology  
08193 Bellaterra (Cerdanyola del Valles)  
Barcelona, Spain

*Spanish Fanconi Anemia Network: [www.redfanconi.net](http://www.redfanconi.net)*

#### ◆ **Jordi Surrallés, PhD**

*[jordi.surralles@uab.es](mailto:jordi.surralles@uab.es)*

*Phone: 34-935-811-830, or 34-935-812-597 Fax: -34-935-812-387*

### **Services Offered**

- ❖ DEB chromosomal breakage testing;
- ❖ FA subtyping by retrovirus mediated gene transfer, in collaboration with **Dr. J.A. Bueren's** laboratory at CIEMAT, Madrid;
- ❖ Genetic counseling;
- ❖ Maintenance of a database integrating genetic and clinical data;
- ❖ Maintenance of a biobank of Spanish FA patients with cryopreserved transformed cell lines and DNA;
- ❖ An FA registry with approximately 170 (Spanish) families.

*Services for genetic analysis are offered free of charge to **Spanish patients** through the Spanish Fanconi Anemia Network, which is funded by the Spanish Ministry of Health.*

### **University of Minnesota**

MMC 366, University of Minnesota  
420 Delaware Street, S.E.  
Minneapolis, MN 55455

#### ◆ **John E. Wagner, MD, Fanconi Anemia Comprehensive Care Program Leader, Director, Division of Hematology/Oncology and Blood and Marrow Transplantation, CCRF/Hageboeck Family Chair in Pediatric Oncology & Clinical Director, Stem Cell Institute)**

*[wagne002@umn.edu](mailto:wagne002@umn.edu)*

*Phone: (612) 626-2961 Fax: (612) 626-4074*

#### ◆ **Margaret L. MacMillan, MD, Fanconi Anemia Comprehensive Care Program Leader, Assistant Professor, Department of Pediatrics, Division of Blood and Marrow Transplantation**

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## ***University of Minnesota (cont'd)***

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- ◆ **Heather Zierhut, MS**, Genetic Counselor  
*hzierhul@fairview.org*  
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### **Services Offered**

- ❖ Fanconi Anemia Comprehensive Care Program with specialists in bone marrow transplantation, hematology, cancer (particularly head and neck), infectious disease, neurology, nephrology, care partners (volunteer family support), social work, genetics, voice reconstruction (to treat androgen side effects), orthopedic surgery, endocrinology (diabetes, short stature), gastroenterology, and radiation therapy;
- ❖ Consultation with new and previously diagnosed Fanconi anemia patients;
- ❖ Expertise in unrelated marrow and umbilical cord transplantation and matched sibling donor transplantation for Fanconi anemia with a 16-bed inpatient area and outpatient bone marrow transplantation clinic;
- ❖ Genetic counseling and coordination of complementation group analysis, mutation screening, somatic mosaicism studies, and other genetic testing options including prenatal genetic testing and preimplantation genetic diagnosis (PGD);
- ❖ Umbilical cord blood bank and stem cell laboratory;
- ❖ Ongoing research in stem cell biology, genetic correction of stem cells, mechanisms of bone marrow failure, and novel treatments with collaborators around the country.

## **University of Pavia**

Pediatric Haematology and Oncology Unit, Fondazione IRCCS Policlinico San Matteo, Pavia Italy  
Piazzale Golgi, 2  
27100, Pavia, Italy

- ◆ **Prof. Franco Locatelli, MD**  
*f.locatelli@smatteo.pv.it*  
Phone: 39-0382-502607 Fax: 39-0382-501251

### **Services Offered**

- ❖ Hematopoietic Stem Cell Transplantation from HLA-identical or disparate family donors and from unrelated volunteers;
- ❖ Stem cell laboratory;
- ❖ Cell therapy laboratory;

## ***University of Pavia (cont'd)***

- ❖ Stem cell bank;
- ❖ Cytogenetic laboratory;
- ❖ Molecular diagnosis of Fanconi anemia.

## **Vrije Universiteit Medical Center Amsterdam**

Department of Clinical Genetics

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## **Services Offered**

- ❖ Diagnosis by chromosomal breakage analysis, including mosaicism;
- ❖ Molecular diagnosis by mutation screening in all known FA genes;
- ❖ Prenatal diagnosis;
- ❖ Preimplantation genetic screening (in preparation);
- ❖ Genetic counseling.

## **Research**

- ❖ Identification of the genes responsible for FA and “overlapping” conditions;
- ❖ Understanding the FA pathway and how a defect in the pathway causes FA specific clinical symptoms;
- ❖ Discovery of the FA pathway defect’s role on sporadic cancer.

## **Collaboration in The Netherlands**

- ❖ **Dr. C.M. Zwaan, MD, PhD** : Clinical Research Coordinator, Erasmus Medical Center, Rotterdam
- ❖ **Dr. M. Bierings, MD, PhD**: Stem Cell Transplantation, Utrecht University Medical Center
- ❖ **Dr. E.T. Korthoff, MD, PhD**: Stem Cell Transplantation, Leiden University Medical Center