

# 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: 91-44-2491-2943 Mobile: 91-92-8211-4661*
- ◆ **Dr. Revathi Raj, MBBS, DCH, MRCP (Path)**  
*Phone: 91-44-2433-1741, 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 Ave.

Cincinnati, OH 45229

- ◆ **Robin Mueller, RN**, Nurse Coordinator, Fanconi Anemia Comprehensive Care Center  
*robin.mueller@cchmc.org*  
*Phone: (513) 636-3218 Fax: (513) 636-7951*
- ◆ **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*
- ◆ **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*
- ◆ **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*

### **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*

#### **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**

Director: The Department of Haematology

◆ **Sister Lucille Wood, BA (Nursing Science) (UNISA), MSc (Medicine)(Haematology)(UCT) RN, Hematology Coordinator**

*haematol@icon.co.za*

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

#### **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/Children's Hospital Cancer Care Program (DFCI/CHCC): The Comprehensive Fanconi Anemia Center**

**Children's Hospital Boston, Hematology/Oncology and Bone Marrow Transplantation**

300 Longwood Ave.

Boston, MA 02115

- ◆ **Colin Sieff, MD**, Director, Marrow Failure Program  
*Phone: (617) 355-7700*
- ◆ **S. Deborah Chirnomas, MD**, Stem Cell Transplant program and Hematology  
*Phone: (617) 355-7700*
- ◆ **David A. Williams, MD**, Chief, Division of Hematology/Oncology, attending physician, Bone Marrow Failure Program  
*Phone: (617) 919-2697*
- ◆ **Ellis Neufeld, MD**, Director, Boston Center for Genetic Blood Diseases, Hematology and Genetics  
*Phone: (617) 355-8246*
- ◆ **Brooke Hoffman, PNP**, Nurse Practitioner  
*Phone: (617) 355-8304*
- ◆ **Cassie Ryan RN**, Hematology program nurse/intake coordinator  
*Phone: (617) 355-8778*

**Services Offered**

Provides comprehensive diagnostic, pre-transplant hematology and transplant services within the Children's Hospital Boston and Dana-Farber Cancer Institute combined Pediatric Hematology/Oncology clinical program.

- ❖ Clinical consultation for the diagnosis, management and treatment of FA through the Bone Marrow Failure Clinic and the Bone Marrow Transplant Program, Division of Hematology/Oncology, Children's Hospital Boston/Dana-Farber Cancer Institute;
- ❖ Coordination of multidisciplinary medical care.

**Dana-Farber Cancer Institute**

44 Binney St.

Boston, MA 02115

- ◆ **Alan D'Andrea, MD**, Chief, Division of Genomic Stability and DNA Repair, The Comprehensive Center for Fanconi Anemia  
*Phone: (617) 632-2112*
- ◆ **Lisa A. Moreau, MS, CLSpCG**, Supervisor of Clinical Laboratory  
*Phone: (617) 632-6302*

**Services Offered**

- ❖ Diagnosis of new FA patients through Clinical Laboratory Improvement Act (CLIA) certified diepoxybutane (DEB) test;
- ❖ Complementation analysis via retrovirus gene transfer.

### **Duke University Medical Center**

Box 3350 DUMC  
2400 Pratt St., 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 which is open 7 days per week;
- ❖ Matched sibling, haploidentical, and unrelated cord blood and adult 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;
- ❖ Expertise in transplantation of infants and young children;
- ❖ Expertise in transplantation of children with inherited metabolic, immunologic, and hematological diseases.

### **Fred Hutchinson Cancer Research Center**

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

- ◆ **Hans-Peter Kiem, MD**, 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, haploidentical and unrelated donors;
- ❖ Collaboration with Hospital de Clinicas, University of Parana, Brazil (**Ricardo Pasquini, MD, PhD**).

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

Genomics Center at Samaritan - A1100

5555 Conner

Detroit, MI 48213

www.genesisgenetics.org

### ◆ **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 FANCA, 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 Ave.

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.

## **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- 6103 or -6568 Fax: 49-211-811-9436*
- ◆ **Eunike Velleuer, MD**, Department of Pediatric Hematology, Oncology and Clinical Immunology  
*velleuer@uni-duesseldorf.de*  
*Phone: 49-211-811-7662 Fax: 49-211-811-7634*
- ◆ **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, I, J, L, 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.aphp.fr*  
Phone, direct: 33-142-499-644  
Secretary: 33-142-499-826 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: 33-142-494-027
- ◆ **Attending Physicians:** Dr. Agnes Devergie; Dr. Thierry Leblanc; Dr. Regis Peffault de la Tour; Dr. Arnaud Rigolet (head and neck cancer); Dr. Marie Robin; Dr. Vanderson Rocha; Pr. Gerard Socie.

### **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**);
- ❖ *FANCA*, *C*, and *G* gene sequencing on grouped patients, in collaboration with Institut Curie (**Pr. Dominique Stoppa-Lyonnet**);
- ❖ 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**

- ❖ 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*
- ◆ **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.

### **International Center for Cell Therapy & Cancer Immunotherapy (ICTC)**

6 Weizman St.  
Tel Aviv 64239  
Israel

- ◆ **Shimon Slavin, MD**, Professor of Medicine, Scientific & Medical Director of ICTC, The Tel Aviv Medical Center  
*slavinMD@gmail.com*  
Phone: 972-3-697-4010 Fax: 972-3-697-4011

### **Services Offered:**

- ❖ Allogeneic stem cell transplantation using well tolerated reduced intensity conditioning towards minimizing the risk of the transplant procedure by using nonmyeloablative stem cell transplantation (NST). Stem cell transplantation can also be considered from half-matched (haploidentically mismatched) family member while avoiding the risk of graft-versus-host disease.

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

1275 York Ave.  
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

## ***Memorial Sloan-Kettering Cancer Center (cont'd)***

### **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 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 Pl.

New York, NY 10029

- ◆ **Christopher Walsh, MD**, Associate Professor of Medicine  
*Christopher-E.Walsh@mountsinai.org*  
*Phone: (212) 241-3554*
- ◆ **Luis Isola, MD**, Director, Stem Cell Transplantation Program
- ◆ **Anne Hurlet, MD**, Associate Professor of Pediatrics

### **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*

- ◆ **Blanche P. Alter, MD, MPH**  
*alterb@mail.nih.gov*  
*Phone: (301)594-7642 Fax: (301) 496-1854*

## ***National Cancer Institute (cont'd)***

### **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 Dr., MSC 1202  
Bethesda MD 20892-1202  
*Phone: (301) 496-5093*

- ◆ **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;
- ❖ Testing for telomerase deficiency.

## **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](http://www.reproductivegenetics.com)

[rgi@flash.net](mailto: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 Ave.

New York, NY 10065

- ◆ Arleen D. Auerbach, PhD  
[auerbac@mail.rockefeller.edu](mailto: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***

- ❖ 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. Mutation screening for families with known mutations in the probands.

##### ***Genetic Services on Research Protocol***

- ❖ International Fanconi Anemia Registry;
- ❖ Confirmation of DEB testing including mosaicism, derivation of lymphoblastoid cell line for complementation testing; maintenance of Fanconi anemia cell and DNA repositories;
- ❖ Mutation screening for particular FA genes under investigation by the laboratory;
- ❖ PGD/IVF: a collaboration with Dr. Mark Hughes (provide genetic information to enable the development of an assay for PGD);
- ❖ Derivation of Human Embryonic Stem Cells from Fanconi anemia embryos (affected embryos diagnosed after PGD/IVF). Cell lines will be available to qualified researchers.

### **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
- ◆ **Russell Ware, MD**  
*russell.ware@stjude.org*
- ◆ **Martha Rieman, FNP**, Bone Marrow Failure Nurse Practitioner  
*martha.rieman@stjude.org*  
Phone: (901) 495-2173

#### **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., Rm. 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 Fax: (718) 343-2961
- ◆ **Johnson M. Liu, MD**, Les Nelkin Professor of Pediatrics, Head, Section of Experimental Hematology and Les Nelkin Memorial Pediatric Oncology Research Laboratory  
*jliu3@nshs.edu*  
Phone: (718) 470-7174
- ◆ **Jeffrey M. Lipton, MD, PhD**, Professor of Pediatrics, Chief, Division of Hematology/Oncology and Stem Cell Transplantation  
*jlipton@lij.edu*  
Phone: (718) 470-3460

#### **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  
Shimokasuya 143  
Isheara, Kanagawa, 259-1193  
Japan

◆ **Hiromasa Yabe, MD, Stem Cell Transplantation**

*yabeh@is.icc.u-tokai.ac.jp*  
*Phone: 81-463-93-1121 Fax: 81-463-95-9567*

◆ **Miharu Yabe, MD, Stem Cell Transplantation**

*miharu@is.icc.u-tokai.ac.jp*  
*Phone: 81-463-93-1121 Fax: 81-463-93-8607*

◆ **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;
- ❖ 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**, Full Professor

*[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;
- ❖ Diagnosis of mosaicism;
- ❖ MMC-induced G2/M-cell phase arrest assay;
- ❖ FANCD2 monoubiquitination assay;
- ❖ Spanish contact for prenatal diagnosis;
- ❖ 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 St., 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)

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

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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  
Piazzale Golgi, 2  
27100, Pavia  
Italy

#### ◆ **Prof. Franco Locatelli, MD**

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

### **VU University Medical Center Amsterdam**

Department of Clinical Genetics  
Van der Boechorststraat 7  
NL-1081 BT Amsterdam  
The Netherlands

#### ◆ **Hans Joenje, PhD**

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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;
- ❖ Genetic counseling.

## *VU University Medical Center Amsterdam (cont'd)*

### **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;
- ❖ **Dr. R.H. Brakenhoff, PhD**, Dept. Otolaryngology/Head-Neck Surgery, VU University Medical Center, Amsterdam.

## Fanconi Anemia Testing Resources

Institution	Test	Cost	Covered by Insurance	Approximate Time for Results	Results go to: Physician/Patient
CIEMAT, Hematopoiesis and Gene Therapy Division, Madrid, Spain <i>Juan Bueren, PhD</i> 34-91-346-6518	Complementation Group <i>(Research)</i>	Free for Spanish Patients	N/A	1 month	Physician
Cincinnati Children's Research Foundation, Translational Trials Development and Support Laboratory (TTDSL), Cincinnati, Ohio <i>Stella Davies, MD (513) 636-1371</i>	Complementation Analysis for <i>A, B, C, E, F, G, J, L (and FANCM analysis under development) in primary T-cells, lymphoblast cell lines (LCL, or fibroblasts) (Available to clinicians and researchers) (CLIA Certified)</i>	Varies	Varies	Varies, dependent upon gene	Physician/Researcher
	FANCD2 western blot and monoubiquitination	Varies	Varies	Varies	Physician/Researcher
	Mutation analysis (in collaboration with The Rockefeller University)	Varies	Varies	Varies	Physician/Researcher
Dana-Farber Cancer Institute, Boston, Massachusetts <i>Alan D'Andrea, MD (617) 632-2112</i> <i>Lisa Moreau (617) 632-6302</i>	DEB/MMC diagnostic testing <i>(CLIA Certified)</i>	Variable (contact lab)	Generally	10 working days	Physician
	Complementation Group <i>(Research)</i>	No charge	N/A	Minimum 3-6 months	Physician
	Mutation Analysis <i>(Research)</i>	No charge	N/A	Minimum 3-6 months	Physician
	D2 Western Blot <i>(Research)</i>	No charge	N/A	1 week	Physician
Heinrich Heine University Medical Center, Düsseldorf, Germany <i>Helmut Hanenberg, MD</i> 49-211-811 ext. 6103 or 6568 fax - 9436	Complementation Group <i>(Research)</i> <i>*Use of retroviruses for A, B, C, D1, D2, E, F, G, I, J, L, M, N in primary T-cells, bone marrow or fibroblasts or in lymphoblast cell lines (LCL)</i> <i>FANCD2 western blotting</i> <i>**Collaborative mutation testing and research with Univ. of Würzburg</i>	No charge	N/A	2-4 weeks	Physician, Patient with signed consent

**CLIA CERTIFICATION:** Clinical Laboratories Improvement Amendments (U.S.)

Institution	Test	Cost	Covered by Insurance	Approximate Time for Results	Results go to: Physician/Patient
Hospital for Sick Children, Toronto, Canada <i>Yigal Dror, MD</i> (416) 813-5630	DEB/MMC diagnostic testing (Licensed under the Ontario Ministry of Health and Long Term Care)	No charge	Generally	2-3 weeks	Physician
Oregon Health & Science University, Portland, OR <i>Susan Olson, PhD,</i> <i>FACMG</i> (diagnostic testing) (503) 494-5964 <i>Markus Grompe,</i> <i>MD,</i> (complementation/ mutation testing) (503) 494-6888 <i>C. Sue Richards,</i> <i>PhD, FACMG</i> (503) 494-5400	DEB/MMC diagnostic testing (CLIA Certified)	Contact lab	Contact lab	1-2 weeks	Physician, Patient
	Complementation Group (Research)	No charge	N/A	Unknown	Physician, patient
	Mutation Analysis (CLIA Certified) FANCA, C, G, E, (Please see GeneTests for further information at <a href="http://geneclinics.org/">http://geneclinics.org/</a> )	Contact lab	Contact Insurance Carrier	3-4 weeks	Physician
Quest Diagnostics Chantilly, VA <i>Andrea Wray</i> (800) 336-3718 ext. 7148	DEB diagnostic testing (CLIA Certified)	Variable	Generally	10 days-3 weeks	Physician
	Prenatal diagnosis (CLIA Certified)	Variable	Generally	2-3 weeks	Physician
Quest Diagnostics San Juan Capistrano, CA <i>Joy Redman</i> (800) 642-4657, ext. 4279	DEB diagnostic testing (CLIA Certified)	\$705.00	Generally	16 days	Physician
	Mutation Analysis: for IVS4=4->T and 322delG (CLIA Certified)	\$205.00	Generally	8 days	Physician
The Rockefeller University New York, NY <i>Arleen Auerbach,</i> <i>PhD</i> (212) 327-7533	Complementation Group (Research)	No charge	N/A	N/A	Physician
	Mutation Analysis for FA Patients (select genes) (Some CLIA Certified, some under Research)	Variable	Variable	Variable	Physician
	Carrier Testing: for known mutations in FANCA/C/F/G in FA relatives, or Ashkenazi Jewish FANCC mutation (CLIA Certified)	\$350.00 (single mutation)	Yes	Yes	Physician
	Screening for common non Ashkenazi Jewish FANCC mutations (CLIA Certified)	\$500.00	Yes	2-3 weeks	Physician

**CLIA CERTIFICATION: Clinical Laboratories Improvement Amendments (U.S.)**

<b>Institution</b>	<b>Test</b>	<b>Cost</b>	<b>Covered by Insurance</b>	<b>Approximate Time for Results</b>	<b>Results go to: Physician/Patient</b>	
<b>Universitat Autònoma de Barcelona, Spain</b> <i>Jordi Surrallés, PhD</i> 34-935-811-830	<b>DEB diagnostic testing (Research), diagnosis of mosaicism</b>	No charge (Spanish patients)	N/A	1 month	Physician	
	<b>Mutation Analysis (Research)</b> For Spanish Gypsy population	No charge (Spanish patients)	N/A	1 week	Physician	
<b>University of Würzburg, Würzburg, Germany</b> <i>Detlev Schindler, MD, PhD</i> 49-931-888-4088-4098 *Collaborative testing and research with University of Düsseldorf	<b>MMC diagnostic testing (Certified under Governmental and Societies)</b>	Varies	No	1-2 weeks	Physician and Patient	
	<b>FANCD2 Immunoblots</b>	\$100.00	Varies	Approx. 2 weeks	Physician and Patient	
	<b>Prenatal Diagnosis</b>	Varies	Varies	Varies	Varies	
	<b>Mutation Analysis (Certified under Governmental and Societies)</b>	\$1000.00-\$2000.00 varies with extent of sequencing	Varies	Varies	Varies dependent upon gene	Physician and Patient
	<b>Complementation Group (Collaborative testing and research with University of Düsseldorf)</b>	Varies	Varies	Varies	Varies	
<b>VU University Medical Center, Amsterdam, The Netherlands,</b> <i>Hans Gille, PhD</i> 31-20-444-8346	<b>Mutation analysis FANCA (Clinically Certified)</b>	€ 650	Generally	3 months	Physician	
	<b>Mutation analysis FANCC, -E, -F and -G (Clinically Certified)</b>	€ 900	Generally	3 months	Physician	
	<b>Mutation analysis of other FANC genes (Clinically Certified)</b>	€ 650 per gene	Generally	Varies	Physician	
	<b>Prenatal DNA diagnosis (Clinically Certified)</b>	€ 650	Generally	1-2 weeks	Physician	

**CLIA CERTIFICATION: Clinical Laboratories Improvement Amendments (U.S.)**

**Additional testing information for Fanconi anemia is available at  
 GENETEST.COM  
 (Search by disease)**