BONE MARROW FAILURE

Children’s Hospital Boston & Dana-Farber Cancer Institute
The Fanconi Anemia and Bone Marrow Failure Multidisciplinary Clinic

Children’s Hospital Boston
300 Longwood Ave.
Boston, MA 02115
http://www.childrenshospital.org/fa-bmf

Dana-Farber Cancer Institute
44 Binney St.
Boston, MA 02115
http://research.dfci.harvard.edu/dandrealab

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  Program Coordinator
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• Lisa A. Moreau, MS, CLSpCG
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  E-mail: lisa_moreau@dfci.harvard.edu

Specialists:
• Adolescent Medicine/Gynecology: Amy DiVasta, MD
• Blood and Marrow (Stem Cell) Transplantation: Leslie Lehmann, MD
• Cardiology: David Harrild, MD, PhD
• Endocrinology: Laurie Cohen, MD
• Gastroenterology: Leonel A. Rodriguez, MD, MS
• Genetics: Jonathan Picker, MD, PhD; Pam Hawley, MS, CGC
• Hand Surgery: Brian Labow, MD
• Hematology: David A. Williams, MD; Colin Sieff, MB BCh; Tara Barlow, PA-C
• Molecular Diagnostics: Alan D’Andrea, MD; Lisa Moreau, MS, CLSpCG; Bai-Lin Wu, PhD, M Med FACMG, Ann E. Reinhard, MS, CGC
• Nephrology: Michael Ferguson, MD
• Otolaryngology: Reza Rahbar, DMD, MD, FACS
• Pulmonary Medicine/Aero-Digestive Disorders: Catherine Sheils, MD
• Social Work: Susan Robinson, LICSW
• Urology: Hiep (Bob) Nguyen, MD

(Continued on the next page)
Services Offered:

♦ Our Program Coordinator arranges a comprehensive evaluation for each new patient, including confirmation of diagnosis, genetic typing, HLA typing, and a multi-system evaluation. The team carefully examines previous diagnostic test results and past medical history to guide the initial evaluation in the multi-disciplinary clinic. This results in a written summary and recommendations for the referring physician and the patient’s family. Ongoing care may continue at Children’s or in partnership with the referring physician. Psychosocial support is available and an important part of our long-term care of families of patients with serious diseases.

♦ A variety of innovative clinical trials are available for enrollment. Two current studies for patients with Fanconi anemia are: 1) the use of danazol, an androgen and 2) a hematopoietic stem cell transplantation trial for those lacking a genotypically identical donor.

♦ Diagnosis of new FA patients through Clinical Laboratory Improvement Act (CLIA) certified diepoxybutane (DEB) test.


♦ Complementation analysis via retrovirus gene transfer.

♦ Fundamental research in DNA repair pathways, induced pluripotent stem cells and gene therapy.

♦ Referral for pre-implantation genetic diagnosis services at two sites in Boston if we find disease causing mutations.

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Children’s Hospital Los Angeles, Keck School of Medicine
University of Southern California
4650 Sunset Boulevard, MS #62
Los Angeles, CA 90027

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BONE MARROW FAILURE, cont’d

Children’s Hospital of Wisconsin
MACC Fund Research Center, Suite 3
8701 Watertown Plank Road
Milwaukie, WI 53226

- Monica Thakar, MD
  Telephone: 414-456-4170    Fax: 414-456-6543
  E-mail: mthakar@mcw.edu

Services Offered:
- Evaluation of new and previously diagnosed patients;
- Personalized treatment regimens and coordination of care by specialists in hematology, bone marrow transplantation, cardiology, endocrinology, genetics, gastroenterology, nephrology, urology, orthopedics, social work;
- Stem cell collection, storage, and infusion;
- Expertise in matched sibling donor, unrelated donor, and haploidentical (alternative donor) transplantation for Fanconi anemia;
- Collaborations with Fred Hutchinson Cancer Research Center and Memorial Sloan-Kettering Cancer Center in alternative donor clinical trials.

Cincinnati Children’s Hospital Medical Center,
The Fanconi Anemia Comprehensive Care Center
3333 Burnet Ave.
Cincinnati, OH 45229
www.cincinnatichildrens.org

- Robin Mueller, RN, Nurse Coordinator, Fanconi Anemia Comprehensive Care Center, Division of Bone Marrow Transplantation and Immune Deficiency, Cancer and Blood Diseases Institute
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  E-mail: robin.mueller@cchmc.org

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- Stella Davies, MB BS, PhD, MRCP, Jacob G. Schmidlapp Endowed Chair and Professor of Pediatrics, University of Cincinnati College of Medicine, Director, Division of Bone Marrow Transplantation and Immune Deficiency, Cancer and Blood Diseases Institute
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  E-mail: stella.davies@cchmc.org

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(Continued on the next page)
Cincinnati Children’s Hospital Medical Center, cont’d

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  E-mail: mike.rutter@cchmc.org

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  E-mail: nada.yazigi@cchmc.org

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**).
City Of Hope
1500 E. Duarte Rd.
Duarte, CA 91010
Telephone: 626-301-8442
E-mail: jrosenthal@coh.org

- Joseph Rosenthal, MD, Director, Pediatric Hematology/Hematopoietic Cell Transplantation
- Debbie Hitt, RN, Nurse-Coordinator, Pediatric Hematology/Hematopoietic Cell Transplantation
- Nadia Ewing, MD, Director, Pediatric Hematology
- Anna Pawlowska, MD, Pediatric Hematology/Hematopoietic Cell Transplantation
- April Sorrell, MD, Pediatric Hematology/Hematopoietic Cell Transplantation and Pediatric Clinical Cancer Genetics
- Julie Wolfson, MD, Pediatric Hematology/Hematopoietic Cell Transplantation
- Joyce Collins, PhD, Cytogenetics
- Smita Bhatia, MD, Pediatrics, Hematology/Hematopoietic Cell Transplantation and Director, Population Sciences
- Christina Cabanillas, LCSW, Social Worker, Pediatric Hematology/Hematopoietic Cell Transplantation

Services Offered:
- Diagnostic testing and screenings of siblings;
- Complete evaluation, treatment and follow-up of new and previously diagnosed FA patients;
- Information and assistance for physicians in the care of their FA patients;
- Cancer screening and specialist consultations;
- Genetic counseling;
- Hematopoietic stem cell transplantation from siblings, matched unrelated donors and umbilical cord blood units;
- Long-term, Fanconi anemia focused, post-transplant follow-up.

Dana-Farber Cancer Institute
44 Binney St.
Boston, MA 02115

- Eva C. Guinan, MD
  Telephone: 617-632-4932     Fax: 617-632-3770
  E-mail: Eva_Guinan@dfci.harvard.edu

Services Offered:
- Comprehensive evaluation;
- Second opinions;
- Pre- and post-transplantation evaluation;
- Follow up care and planning.
Services Offered:

- Evaluation of new and previously diagnosed patients;
- Complementation group analysis (research basis);
- Personalized treatment regimens by specialists in hematology, bone marrow transplantation, cardiology, endocrinology, genetics, gastroenterology, nephrology, urology, orthopedics, social work;
- Gene therapy trial; Fanconi anemia repository for samples and data;
- Stem cell collection, storage, and reinfusion;
- Expertise in matched sibling donor, unrelated donor, and haploidentical (alternative donor) transplantation for Fanconi anemia;
- Collaborator, Hospital de Clínicas, University of Parana, Brazil (Carmem Bonfim, MD and Ricardo Pasquini, MD, PhD).
Hackensack University Medical Center
Tomorrow’s Children Institute
30 Prospect Ave.
Hackensack, NJ 07601
www.tcikids.com

- Alfred P. Gillio, MD, Director Bone Marrow Failure Clinic, Pediatric Hematology/Oncology
  Telephone: 201-996-5600   Fax: 201-996-0521
  E-mail: agillio@humed.com
- Barbara Adler-Brecher, MS
  E-mail: badler@humed.com

Services Offered:
- Diagnosis and genetic counseling;
- Comprehensive medical treatment including Gynecology and ENT specialists;
- Matched sibling and unrelated donor transplants.

Memorial Sloan-Kettering Cancer Center
1275 York Ave.
New York, NY 10021
www.mskcc.org

- Farid Boulad, MD, Medical Director, Pediatric Day Hospital, Pediatric Hematology and Bone Marrow Transplantation
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  E-mail: singhb@mskcc.org
- Mercedes Castiel, MD, Gynecology
  Telephone: 212-639-5060
  E-mail: castielm@mskcc.org
- Julia A. Kearney, MD, Psychiatry and Pediatrics
  Telephone: 646-888-0028
  E-mail: Kearneyj@mskcc.org

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
  Telephone: 212-241-3443
  E-mail: christopher-e.walsh@msnyuhealth.org
- 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.

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.marrowfailure.cancer.gov

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- Pamela Stratton, MD, Chief, Gynecology Consult Service
  Telephone: 301-496-9079  Fax: 301-480-6703
  E-mail: stratton@nih.gov
- Carter Van Waes, MD, PhD, Chief Head and Neck Surgery Branch; Clinical Director, National Institute on Deafness and other Communication Disorders
  Telephone: 301-402-4216  Fax: 301-402-1140
  E-mail: vanwaesc@nidcd.nih.gov

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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.

Oregon Health & Science University, Doernbecher Children’s Hospital
3181 S.W. Sam Jackson Park Road
Portland, OR 97239

• Peter Kurre, MD, Associate Professor, Division of Pediatric Hematology/Oncology
  Telephone: 503-494-1543   Fax: 503-494-0714
  E-mail: kurrepe@ohsu.edu

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.

International Fanconi Anemia Registry (IFAR)
The Rockefeller University
Laboratory of Genome Maintenance
1230 York Ave., Box 182
New York, NY 10065-6399

• Arleen D. Auerbach, PhD
  Telephone: 212-327-7533, or 8862   Fax: 212-327-8262
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  http://www.rockefeller.edu/labheads/smogorzewska/

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International Fanconi Anemia Registry (IFAR), cont’d

Services Offered:

Genetic Services on Research Protocol:

♦ International Fanconi Anemia Registry (IFAR);
♦ Derivation of lymphoblastoid cell line for complementation testing; maintenance of Fanconi anemia cell and DNA repositories;
♦ Mutation screening of probands for FA genes under investigation by the laboratory.

Co-Principal Investigators for the IFAR:

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Schneider Children’s Hospital, Albert Einstein School of Medicine

**Bone Marrow Failure Program**

Division of Hematology/Oncology and Stem Cell Transplantation

269-01 76th 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
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  Telephone: 718-470-3460
  E-mail: jlipton@lij.edu

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Schneider Children’s Hospital, Albert Einstein School of Medicine, cont’d

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.

Stanford School of Medicine, Lucille Packard Children’s Hospital
Hematology Clinic    Pediatric Hematology/Oncology Academic Office
770 Welch Road, Suite 261   1000 Welch Road, Suite 300
Palo Alto, CA 94304    Palo Alto, CA  94304
www.lpch.org

Hematology/Oncology    Telephone: 650-723-5535
• Bertil Glader, MD, PhD, Professor, Pediatrics and Pathology
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Stem Cell Transplant    Telephone: 650-725-9250
• Kenneth Weinberg, MD, Professor, Pediatrics
  E-mail: kw1@stanford.edu
• Rajni Agarwal, MD, Assistant Professor, Pediatrics
  E-Mail: rajnia@stanford.edu
• Sandhya Kharbanda, MD, Assistant Professor of Pediatrics
  E-mail: ksandhya@stanford.edu
• Karen Kristovich, RN, MSN, PNP, Stem Cell Transplant Nurse Practitioner
  E-mail: kkristovich@lpch.org
• Tracey Stoll, RN, Related Donor Stem Cell Transplant Coordinator
  E-mail: tstoll@lpch.org

Services Offered:
♦ Diagnostic evaluation for Fanconi anemia and other bone marrow failure syndromes;
♦ Comprehensive care and long-term follow-up of new and previously diagnosed patients;
♦ Stem cell transplantation with matched related donor, matched unrelated donor, umbilical cord blood, and peripheral blood stem cells.
University of Minnesota, Fanconi Anemia Comprehensive Care Program
University of Minnesota Medical Center and Amplatz Children’s Hospital
MMC 366, University of Minnesota
420 Delaware St., S.E.
Minneapolis, MN 55455
Telephone: 612-273-2800
www.bmt.umn.edu/disease/fanconi-anemia

• **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 Medical Center and Amplatz Children’s Hospital, cont’d

Services Offered:

♦ The University of Minnesota Fanconi Anemia Comprehensive Care Program (FACCP) utilizes a multidisciplinary team approach to ensure that the care we provide is individually tailored to meet the unique needs of each patient and family.

♦ The FACCP is composed of expert health care providers who specialize in blood and marrow transplantation, hematology, cancer (head and neck, gynecological), infectious disease, neurology, nephrology, cardiology, care partners (volunteer family support), social work, genetics, voice reconstruction (to treat androgen side effects), nutrition therapy, orthopedic surgery, endocrinology (diabetes, short stature), dietician, gastroenterology, gynecology and fertility counseling, and radiation therapy;

♦ Consultations with new and previously diagnosed Fanconi anemia patients;

♦ Individualized treatment planning for newly diagnosed children and adults who do not need transplant;

♦ Blood and marrow transplantation: expertise in unrelated marrow and umbilical cord transplantation and matched sibling donor transplantation for Fanconi anemia with a 24-bed pediatric inpatient unit and a 30-bed adult inpatient unit and state of the art 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 pre-implantation genetic diagnosis (PGD);

♦ Family support services including nurse coordination, social work, genetic counseling, care-partners, and child and family life specialists to assist families throughout the diagnostic, transplantation, and post-transplantation processes;

♦ Family umbilical cord blood banking;

♦ Ongoing research in stem cell biology, mosaicism, genetic correction of stem cells, mechanisms of bone marrow failure, and novel treatments with collaborators around the country.
BONE MARROW FAILURE, cont’d

The University of Texas Southwestern Medical Center
Pediatric Bone Marrow Failure Program
Children’s Medical Center Dallas
Center for Cancer and Blood Disorders
1935 Medical District Drive
Dallas, TX 75235

To Contact us:
Telephone: 214-456-2382 or 214-456-6102
Email: BoneMarrowFailure@UTSouthwestern.edu

Nurse Coordinator:
- Deborah J. Boger, MSN, RN, CPNP Bone Marrow Failure Nurse Practitioner
  Telephone: 214-456-6102     Fax: 214-456-8469

Medical Staff:
- Zora R. Rogers, MD, Professor of Pediatrics, Hematology-Oncology
  Clinical Director, Bone Marrow Failure Program
  E-mail: zora.rogers@utsouthwestern.edu
- Andrew Koh, MD, Assistant Professor of Pediatrics, Hematology-Oncology
  Director, Stem Cell Transplant Program

Services Offered:
♦ Consultation and education for new and previously diagnosed patients and their families;
♦ Comprehensive clinical management of pediatric Fanconi anemia patients;
♦ Stem cell transplantation for Fanconi anemia; stem cell collection and reinfusion;
♦ Disease based follow up post transplantation for Fanconi anemia;
♦ Coordinated referral to institutional subspecialists in cardiology, endocrinology, gastroenterology, nephrology, hand and orthopedic surgery interested in the unique needs of persons with Fanconi anemia
♦ Psychology, Social Work, Physical and Occupational Therapy services
**DKMS Americas**

33 East 33rd Street, Suite 501  
New York, NY 10016  
www.dkmsamericas.org  
Telephone: 1-866-340-DKMS (3567)  
E-mail: info@dkmsamericas.org

DKMS is committed to the fight against blood diseases like leukemia and Fanconi anemia, and strives to give every blood cancer patient a second chance at life by recruiting bone marrow donors from every race and ethnicity. We inspire hope by matching our donors with patients from around the world who are in need of a bone marrow transplant.

As the world’s largest bone marrow donor center, DKMS:

♦ Organizes thousands of Bone Marrow Donor Drives across the U.S. to recruit healthy, committed donors; We do not make the testing fee mandatory, but rely on donations to support our life-saving work;

♦ Provides patients with access to more than 2 million registered donors; more than 25% of all transplants are provided by DKMS;

♦ Performs HLA-A*, B*, C*, DR* high-resolution typing on every registered donor, minimizing the search time for finding the best possible donor match for a patient;

♦ Coordinates all transports of bone marrow and blood stem cells from Germany to USA.

**National Marrow Donor Program® Be The Match Registry℠**

3001 Broadway Street NE, Suite 100  
Minneapolis, MN 55413-1753  
www.marrow.org  
Telephone: 1-888-999-6743. Outside of the US: 612-632-3410  
Email: patientinfo@nmdp.org

The National Marrow Donor Program (NMDP) has been helping people who need a potentially life-saving marrow or cord blood transplant for more than 20 years. Be The Match Registry℠ is the new name of the NMDP’s registry

**Services Offered:**

♦ We connect patients, doctors, donors and researchers to the resources they need to help more people live longer and healthier lives;

♦ Assist patients and families in Donor Drives with event planning and coordination, media support, and fundraising;

♦ The NMDP’s Office of Patient Advocacy (OPA) offers resources and services to help patients and their families from diagnosis through survivorship. All OPA materials and services are free and confidential.
Cord Blood Registry®
Designated Transplant Program (DTP)
Cord Blood Registry
1200 Bayhill Drive, 3rd Floor
San Bruno, CA 94066
www.cordblood.com
Telephone: 1-888-Cord-Blood

Services Offered:
♦ Cord Blood Registry® (CBR) is the world’s largest and most experienced family cord blood stem cell bank. As a public service, CBR provides the Designated Transplant Program (DTP) to families wishing to store their newborn’s umbilical cord blood stem cells for a family member diagnosed with a life-threatening disease treatable by stem cell transplantation, including Fanconi anemia. CBR provides the collection kit, processing and storage of the newborn’s umbilical cord blood without any cost to families qualifying for the DTP.
♦ For more information on the DTP, including the qualification criteria and the application process, please contact Cord Blood Registry at 1-888-Cord-Blood or visit www.cordblood.com.

National Marrow Donor Program® (NMDP) Related Donor Cord Blood Program
3001 Broadway St. NE, Suite 100
Minneapolis, MN 55413
www.marrow.org
Telephone: 1-888-999-6743. Outside of the USA: 612-632-3410
Email: patientinfo@nmdp.org

Services Offered:
♦ For families with a first-degree relative diagnosed with Fanconi anemia or with an other medical condition that might benefit from a cord blood transplant in the USA;
♦ No charge to the family when a new baby is born into an eligible family, to have that baby’s umbilical cord blood collected and stored until it can be used for a transplant;
♦ The NMDP’s Office of Patient Advocacy patient services coordinators can answer questions about the Related Donor Cord Blood Program, eligibility guidelines, insurance and how to contact a participating cord blood bank;
♦ Note: the cord blood bank determines final eligibility for this program.
ENDOCRINOLOGY

Cincinnati Children’s Hospital Medical Center,
The Fanconi Anemia Comprehensive Care Center
3333 Burnet Ave.
Cincinnati, OH 45229

• Susan R. Rose, MD, MEd, Professor of Pediatrics, Pediatric Endocrinology
  Telephone: 513-636-4744
  E-mail: susan.rose@cchmc.org
• Meilan Rutter, MD, Associate Professor of Pediatrics, Division of Endocrinology
  Telephone: 513-636-4744
  E-mail: meilan.rutter@cchmc.org

University of Minnesota, Fanconi Anemia Comprehensive Care Program
University of Minnesota Medical Center and Amplatz Children’s Hospital
MMC 366, University of Minnesota
420 Delaware St., S.E.
Minneapolis, MN 55455

• Anna Petryk, MD, Pediatric Endocrinology
  Telephone: 612-624-5409     Fax: 612-626-5262
  E-mail: petry005@umn.edu

GASTROENTEROLOGY

Cincinnati Children’s Hospital Medical Center,
The Fanconi Anemia Comprehensive Care Center
3333 Burnet Ave.
Cincinnati, OH 45229

• Nada Yazigi, MD, Assistant Professor of Pediatrics, Pediatric Gastroenterology
  Telephone: 513-636-4415     Fax: 513-636-7805
  E-mail: nada.yazigi@cchmc.org

University of Minnesota, Fanconi Anemia Comprehensive Care Program,
University of Minnesota Medical Center and Amplatz Children’s Hospital
MMC 366, University of Minnesota
420 Delaware St., S.E.
Minneapolis, MN 55455

• Sarah Jane Schwarzenberg, MD, Director, Pediatric Gastroenterology, Hepatology and Nutrition
  Telephone: 612-624-1133     Fax: 612-626-0639
  E-mail: schwa005@umn.edu
GENETIC COUNSELING

University of Minnesota, Fanconi Anemia Comprehensive Care Program
University of Minnesota Medical Center and Amplatz Children’s Hospital
MMC 366, University of Minnesota
420 Delaware St., S.E.
Minneapolis, MN 55455
Telephone 612-273-2800
www.bmt.umn.edu/disease/fanconi-anemia

• Heather Zierhut, MS, CGC, Certified Genetic Counselor
  Telephone: 612-626-6743    Fax: 612-273-2919
  E-mail: hzierhu1@fairview.org

GYNECOLOGY

Cincinnati Children’s Hospital Medical Center,
The Fanconi Anemia Comprehensive Care Center
3333 Burnet Ave.
Cincinnati, OH 45229

• Jill Huppert, MD, MPH, Pediatric and Adolescent Gynecology
  Telephone: 513-636-7042    Fax: 513-636-8844
  E-mail: jill.huppert@cchmc.org

Eunice Kennedy Shriver National Institute of Child Health and Human Development
Bldg 10, CRC, Room 1-3140
10 Center Dr. MSC 1109
Bethesda, MD 20892-1109

• Pamela Stratton, MD, Chief, Gynecology Consult Service, Program in Reproductive and Adult Endocrinology
  Telephone: 301-496-9079    Fax: 301-480-6703
  E-mail: Stratton@nih.gov
Memorial Sloan-Kettering Cancer Center
1275 York Ave.
New York, NY 10021

- **Mercedes Castiel, MD**, Gynecology
  Telephone: 212-639-5060
  E-mail: castielm@mskcc.org

University of Minnesota, Fanconi Anemia Comprehensive Care Program
University of Minnesota Medical Center, Amplatz Children’s Hospital, Fairview
MMC 366, University of Minnesota
420 Delaware St., S.E.
Minneapolis, MN 55455

- **Rahel G. Ghebre, MD**, Gynecologic Oncology
  Telephone: 612-626-2613     Fax: 612-626-0665
  E-mail: ghebr004@umn.edu

HAND SURGEONS

Ann Van Heest, MD, Professor

- **University of Minnesota, Department of Orthopaedic Surgery**
  2450 Riverside Avenue, Rm R200
  Minneapolis, MN 55454
  Telephone 612-273-8086
  E-mail: vanhe003@umn.edu

- **Gillette Children’s Specialty Healthcare**
  200 University Avenue East
  St. Paul, MN 55101
  Telephone: 651-291-2848

- **Shriners Hospital for Children, Twin Cities Unit**
  2025 East River Parkway
  Minneapolis, MN 55414
  Telephone: 612-596-6100

**Services Offered:**
- Hand and upper extremity surgery in the pediatric population
Cincinnati Children’s Hospital Medical Center,  
The Fanconi Anemia Comprehensive Care Center  
3333 Burnet Ave.  
Cincinnati, OH 45229

- Roger Cornwall, MD, Assistant Professor of Pediatrics, Division of Orthopaedics (Hand Surgery)  
  Telephone: 513-636-7319     Fax: 513-636-3928  
  E-mail: roger.cornwall@cchmc.org

Shriners Hospital for Children  
3551 North Broad St.  
Philadelphia, PA 19140

- Scott Kozin, MD, Professor, Orthopedic Surgery, Temple University, Hand & Upper Extremity Surgeon  
  Telephone: (215) 430-4034  
  E-mail: skozin@shrinenet.org

- Linda Filipone, Nurse Coordinator  
  Telephone: (215) 430-4074  
  E-mail: lfilipone@shrinenet.org

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.

Boston Foundation for Sight  
464 Hillside Ave., Suite 205  
Needham, MA 02494  
www.bostonsight.org

- Brenda Diaz, New Patient Coordinator  
  Telephone: 781-726-7337, ext. 101     Fax: 781-726-7310  
  E-mail: bdiaz@bostonsight.org

Boston Foundation for Sight is a non-profit organization dedicated to helping patients whose eye functions are impaired by damaged/diseased corneas.

Services Offered:  
Custom fitting of the Boston Ocular Surface Prosthesis. This device acts like a corneal liquid bandage, able to provide significant relief to those patients who are unresponsive to low risk treatments for severe dry eyes due to graft versus host disease, etc. This service is offered to all who may benefit, regardless of their ability to pay.
OTOLARYNGOLOGY (Ear, Nose, and Throat) and HEAD AND NECK CANCER

Cincinnati Children’s Hospital Medical Center, The Fanconi Anemia Comprehensive Care Center
3333 Burnet Ave.
Cincinnati, OH 45229

- **Michael Rutter, MD**, Associate Professor of Pediatrics, Division of Otolaryngology
  Telephone: 513-636-4881    Fax: 513-636-8133
  E-mail: mike.rutter@cchmc.org

Johns Hopkins Medical Institutions
601 N. Caroline Street, 6th Floor
Baltimore, MD 21287-0910

- **Joseph Califano, MD**, Professor, Otolaryngology, Head & Neck Surgery & Head & Neck Research Division
  Telephone: 410-955-6420    Fax: 410-955-8510
  E-mail: jcalifa@jhmi.edu

**Clinical Interests:** Tumors of the oral cavity, larynx, pharynx, neck, skull base, salivary glands, thyroid cancer, premalignant conditions of the upper aerodigestive tract, and unknown primary head and neck squamous cell carcinoma, skin cancer and melanoma.

Memorial Sloan-Kettering Cancer Center
1275 York Ave.
New York, NY 10021

- **Bhuvanesh Singh, MD, PhD**, Head and Neck Cancer
  Telephone: 212-639-2024
  E-mail: singhb@mskcc.org

**Clinical Interests:** All head and neck cancers, with a particular focus on cancers of the mouth (tongue, floor of mouth, and buccal mucosa), skull base, and salivary and thyroid glands, as well as melanomas and skin cancers of the head and neck region.
National Institutes of Health
NIDCD/NIH
10 Center Drive, Building 10
Bethesda, MD 20892

- **H. Jeffrey Kim, MD**, Associate Professor, Department of Otolaryngology (Ears, Nose, Throat), Head and Neck Surgery
  Telephone: 301-496-5369
  E-mail: kimj@nidcd.nih.gov

**Clinical Interests:**
- Dizziness, Hereditary Hearing Loss, Balance Disorders, Acoustic Neuroma and skull base surgery, Otosclerosis, Cholesteatoma

- **Carter Van Waes, MD, PhD**, Chief Head and Neck Surgery Branch; Clinical Director, National Institute on Deafness and other Communication Disorders
  Telephone: 301-402-4216    Fax: 301-402-1140
  E-mail: vanwaesc@nidcd.nih.gov

**Clinical Interests:**
- Associate investigator on Dr. Blanche Alter’s protocol, for diagnosis of head and neck mucosal lesions and cancers for participating patients.

University of Chicago Medical Center
5841 S. Maryland Avenue
Chicago, IL 60634

- **Ezra Cohen, MD**, Associate Professor of Medicine
  Telephone: 773-702-4137    Fax: 773-702-2835
  E-mail: ecohen@medicine.bsd.uchicago.edu

**Clinical Interests:**
- Head and Neck Cancer, Lung Cancer, Esophageal Cancer

- **Everett Vokes, MD**, John E. Ultmann Professor of Medicine and Radiation and Cellular Oncology and Chairman, Department of Medicine
  Telephone: 773-702-9306 Fax: 773-702-4427
  E-mail: evokes@medicine.bsd.uchicago.edu

**Clinical Interests:**
- Hematology, Oncology, Head and Neck Cancer, Lung Cancer, Esophageal Cancer
University of Cincinnati, College of Medicine
Department of Otolaryngology
234 Albert Sabin Way
ML 0528
Cincinnati, OH 45267

- Keith A. Casper, MD, Assistant Professor
  Telephone: 513-558-1890   Fax: 513-558-4477
  E-mail: Keith.casper@uc.edu
- Keith M. Wilson, MD
- Lee A. Zimmer, MD

University of Minnesota, Fanconi Anemia Comprehensive Care Program
University of Minnesota Medical Center and Amplatz Children’s Hospital
MMC 396, University of Minnesota
420 Delaware St., SE
Minneapolis, MN 55455

- Frank Ondrey, MD, PhD, FACS, Associate Professor of Otolaryngology, Director, Molecular Oncology Program
  Telephone: 612-624-5900
  E-mail: ondre002@umn.edu

Clinical Interests: Head and Neck Cancer, Precancerous Mouth and Throat Lesions, Comprehensive General Otolaryngology, and Facial Trauma

University of Pittsburgh School of Medicine Eye & Ear Institute
Suite 300
203 Lothrop Street
Pittsburgh, PA 15213

- Jennifer Rubin Grandis, MD, FACS, Professor
  Telephone: 412-647-2100
  E-mail: jgrandis@pitt.edu

Specialty: Otolaryngology
Sub Specialty: Head & Neck Surgery
University of Texas MD Anderson Cancer Center
1515 Holcombe Blvd., Unit 441
Houston, TX 77030

- Erich Madison Sturgis, MD, MPH, FACS, Associate Professor of Surgery, Associate Professor of Epidemiology
  Telephone: 713-792-5432     Fax: 713-794-4662

Clinical Interests: Head & Neck Cancer, Salivary Carcinoma, and Thyroid Cancer, Human Papillomavirus, and Sarcomas of the Head and Neck

Services Offered: Multidisciplinary evaluation and treatment of head and neck tumors.

The Genesis Genetics Institute, Genomic Technology Center
Genomics Center at Samaritan - A1100
5555 Conner
Detroit, MI 48213
www.genesisgenetics.org

- Mark Hughes, MD, PhD
  Telephone: 313-544-4006     Fax: 313-544-4006
  E-mail: pgd@genesisgenetics.org
- Shannon Wiltse, MS, CGC, Certified Genetic Counselor
  Telephone: 313-544-4006     Fax: 313-544-4006
  E-mail: counselor@genesisgenetics.org

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 230 in vitro fertilization programs, and with internationally respected Fanconi transplantation experts.
The Reproductive Genetics Institute
2825 N Halsted St.
Chicago, IL 60657
Telephone: 773-472-4900 Fax: 773-871-5221
www.reproductivegenetics.com
E-mail: rgiworld@gmail.com

- Dana Pauling, MS, CGC
- Christina Lavin, MS, CGC
- Andria Besser, MS
- Anna Cisler, MS

Services Offered:
- Pre-implantation genetic diagnosis for known Fanconi mutations with or without HLA matching for sibling bone marrow donation.
<table>
<thead>
<tr>
<th>Institution</th>
<th>Test</th>
<th>Cost</th>
<th>Covered by Insurance?</th>
<th>Approximate Time for Results</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Cincinnati Children’s Research Foundation, Translational Trials Development Support Laboratory (TTDSL)</strong>&lt;br&gt;Cincinnati, OH&lt;br&gt;• Stella Davies, MD&lt;br&gt;513-636-1371&lt;br&gt;<a href="mailto:stella.davies@cchmc.org">stella.davies@cchmc.org</a></td>
<td>Complementation analysis for 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)&lt;br&gt;CLIA Certified*&lt;br&gt;FANCD2 western blot and monoubiquitination&lt;br&gt;Mutation analysis (in collaboration with The Rockefeller University)</td>
<td>Varies</td>
<td>Varies</td>
<td>Varies, dependent upon gene</td>
</tr>
<tr>
<td>Dana-Farber Cancer Institute&lt;br&gt;Boston, MA&lt;br&gt;• Alan D’Andrea, MD&lt;br&gt;617-632-2080&lt;br&gt;<a href="mailto:alan_dandrea@dfci.harvard.edu">alan_dandrea@dfci.harvard.edu</a>&lt;br&gt;• Lisa Moreau, MS&lt;br&gt;617-632-6302&lt;br&gt;<a href="mailto:lisa_moreau@dfci.harvard.edu">lisa_moreau@dfci.harvard.edu</a></td>
<td>DEB/MMC diagnostic testing&lt;br&gt;CLIA Certified*&lt;br&gt;Complementation group (Research)&lt;br&gt;Mutation analysis (Research)&lt;br&gt;D2 western blot (Research)</td>
<td>Varies (contact lab)</td>
<td>Generally</td>
<td>10 working days</td>
</tr>
<tr>
<td>Oregon Health &amp; Science University&lt;br&gt;Portland, OR&lt;br&gt;• Susan Olson, PhD, FACMG (cytogenics)&lt;br&gt;503-494-5964&lt;br&gt;<a href="mailto:olsonsu@ohsu.edu">olsonsu@ohsu.edu</a>&lt;br&gt;• Markus Grompe, MD (complementation)&lt;br&gt;503-494-6888&lt;br&gt;<a href="mailto:grompe@ohsu.edu">grompe@ohsu.edu</a>&lt;br&gt;• C. Sue Richards, PhD, FACMG (molecular)&lt;br&gt;503-494-5400&lt;br&gt;<a href="mailto:richarsu@ohsu.edu">richarsu@ohsu.edu</a></td>
<td>DEB/MMC diagnostic testing&lt;br&gt;CLIA Certified*&lt;br&gt;Complementation group (Research)&lt;br&gt;Mutation Analysis for A, C, G, E, F (Please see GeneTests for further information at geneclinics.org)&lt;br&gt;CLIA Certified*</td>
<td>Contact lab</td>
<td>Varies</td>
<td>1-2 weeks</td>
</tr>
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</tbody>
</table>

*CLIA Certified* indicates that the test is certified by the Clinical Laboratory Improvement Amendments (CLIA) program, which is required for laboratory testing in the United States.
<table>
<thead>
<tr>
<th>Institution</th>
<th>Test</th>
<th>Cost</th>
<th>Covered by Insurance?</th>
<th>Approximate Time for Results</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Quest Diagnostics</strong></td>
<td>DEB diagnostic testing: the doctor needs to specify DEB test, otherwise it is possible that a DNA carrier test for FANCC may be done. <strong>CLIA Certified</strong></td>
<td>Varies</td>
<td>Generally</td>
<td>10 days-3 weeks</td>
</tr>
<tr>
<td>Chantilly, VA</td>
<td>Prenatal diagnosis <strong>CLIA Certified</strong></td>
<td>Varies</td>
<td>Generally</td>
<td>2-3 weeks</td>
</tr>
<tr>
<td>• Aimee Martin, MGC, CGC</td>
<td></td>
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<td></td>
<td></td>
</tr>
<tr>
<td>800-336-3718, ext. 7148</td>
<td></td>
<td></td>
<td></td>
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</tr>
<tr>
<td><a href="mailto:aimee.d.martin@questdiagnostics.com">aimee.d.martin@questdiagnostics.com</a></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Quest Diagnostics, San Juan Capistrano, CA</strong></td>
<td>Mutation analysis for IVS4=4-&gt; and 322delG <strong>CLIA Certified</strong></td>
<td>$205</td>
<td>Generally</td>
<td>8 days</td>
</tr>
<tr>
<td>• Joy Redman, MS, MBA, CGC</td>
<td></td>
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<tr>
<td>800-642-4657, ext. 4279</td>
<td></td>
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</tr>
<tr>
<td><a href="mailto:Joy.B.Redman@questdiagnostics.com">Joy.B.Redman@questdiagnostics.com</a></td>
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<tr>
<td><strong>The Rockefeller University</strong></td>
<td>Mutation analysis (Research)</td>
<td>No charge</td>
<td>N/A</td>
<td>Varies</td>
</tr>
<tr>
<td>New York, NY</td>
<td></td>
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<tr>
<td>• Arleen Auerbach, PhD</td>
<td></td>
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<tr>
<td>212-327-7533</td>
<td></td>
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<tr>
<td><a href="mailto:auerbac@rockefeller.edu">auerbac@rockefeller.edu</a></td>
<td></td>
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<td></td>
<td></td>
</tr>
<tr>
<td>• Francis Lach, Research Specialist</td>
<td>D2 western blot (Research)</td>
<td>No charge</td>
<td>N/A</td>
<td>Varies</td>
</tr>
<tr>
<td>212-327-8862</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><a href="mailto:lachf@rockefeller.edu">lachf@rockefeller.edu</a></td>
<td></td>
<td></td>
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</tr>
</tbody>
</table>

*CLIA Certified: Clinical Laboratories Improvement Amendments (USA)*

Additional testing information for Fanconi anemia is available at:
http://geneclinics.org
Select “Gene Reviews,” then “Search by Disease”