



Fanconi Anemia
RESEARCH FUND, INC.

International Fanconi Anemia
Treatment and Testing
Resource Guide

2015

OUR MISSION

To find effective treatments and a cure for Fanconi anemia and to provide education and support services to affected families worldwide.



1801 Willamette Street, Suite 200
Eugene, OR 97401
1-541-687-4658

Toll Free: 1-888-FANCONI (inside the U.S. only)

Fax: 1-541-687-0548

www.fanconi.org

info@fanconi.org

www.facebook.com/fanconianemiaresearchfund

<https://twitter.com/FAresearchfund>

Staff

Laura Hays, PhD, Executive Director

laura@fanconi.org

Teresa Kennedy, MA, Director of Family Support Services

teresa@fanconi.org

Janae Peverill, Executive and Family Support Assistant

janae@fanconi.org

Cynthia Freeman, Special Projects Coordinator

cynthia@fanconi.org

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U.S. Fanconi Anemia Treatment Resource Guide

Editors' note and disclaimer: The resource list provided in this guide is not comprehensive. Inclusion on the list does not imply endorsement by the Fanconi Anemia Research Fund, Inc. Families are encouraged to ask clinicians about their experience treating patients with Fanconi anemia.

BONE MARROW FAILURE

Children's Hospital Central California

Department of Pediatric Hematology SC-62
9300 Valley Children's Place
Madera, CA 93636 -8762
Telephone: 559-353-5460 Fax: 559-353-5095
Email: vbalasa@childrenscentralcal.org

- **Laura Andrews, RN, Transplant Coordinator**
- **Vinod Balasa, MD, Director of Hematology/Oncology**
- **Kelly Folmer, RN, CPNP**
- **Terea Giannetta, DNP, CPNP, Nurse Practitioner**
- **Catherine Hanson, LCSW**
- **Nancy G. Hatcher, MSW, Clinical Social Worker**
- **Mary Beth Jones, Child Life Coordinator**
- **William Koole, Pharm.D**
- **Shari Langer, PhD, Clinical Psychologist**
- **J. Daniel Ozeran, MD, PhD**
- **Latha Rao, MD**
- **Faisal Razzaqi, MD**
- **Ruth Rolen-Martinez, RN, BSN, Clinical Coordinator**
- **Wendy Tcheng, MD**
- **Ruetima Titaphiwatanakun, MD**
- **Pamela Young, RD**

Services Offered:

Children's Hospital Central California provides diagnostic, treatment and referral services to children ages birth to 21 years in California's Central Valley, at the only pediatric hospital between Los Angeles and San Francisco and the largest children's hospital between San Diego and San Francisco. Patients are seen and treated from initial diagnosis to and beyond bone marrow transplant for which patients are referred to nationally recognized transplant centers. Patients receive specialty care within the Fanconi anemia treatment program as well as from Genetics, Orthopaedics, Cardiology, Behavioral Health Sciences and other specialty services. Patients with Fanconi anemia are provided supportive, educational, and consultive services to further enhance care.

Children's Hospital Los Angeles, Keck School of Medicine, University of Southern California

4650 Sunset Boulevard, MS #62
Los Angeles, CA 90027

- **Neena Kapoor, MD, Division of Research Immunology & Bone Marrow Transplant**
Telephone: 323-361-2546 Fax: 323-361-8068
Email: nkapoor@chla.usc.edu

Children's Hospital of Wisconsin

MACC Fund Research Center, Suite 3018
8701 Watertown Plank Road
Milwaukee, WI 53226

Monica Thakar, MD

Telephone: 414-955-4170 Fax: 414-955-6543
Email: 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.

Note: FANCD2 immunoblot=FANCD2 western=FANCD2 ubiquitination.

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Cincinnati Children's Hospital Medical Center Fanconi Anemia Comprehensive Care Center

MLC 11013
3333 Burnet Avenue
Cincinnati, OH 45229
www.cincinnatichildrens.org

- **Erica L. Goodridge, RN, BSN, CPN**, Care Manager, Fanconi Anemia Comprehensive Care Center
Telephone: 513-636-3218 Fax: 513-636-7951
Email: erica.goodridge@cchmc.org
- **Stella Davies, MBBS, PhD, MRCP**, Director, Bone Marrow Transplantation and Immune Deficiency Jacob G. Schmidlapp Endowed Chair
Professor, UC Department of Pediatrics
Telephone: 513-636-2469 Fax: 513-636-3549
Email: stella.davies@cchmc.org
- **Richard Harris, MD**, Professor of Clinical Pediatrics University of Cincinnati College of Medicine Division of Blood and Marrow Transplantation and Immune Deficiency
Cancer and Blood Diseases Institute
Telephone: 513-803-9063 Fax: 513-636-7951
Email: richard.harris@cchmc.org
- **Lesley Breech, MD**, Division Director, Pediatric and Adolescent Gynecology
Associate Professor, UC Department of Surgery University of Cincinnati Department of Obstetrics and Gynecology
Telephone: 513-636-2960 Fax: 513-636-8844
Email: lesley.breech@cchmc.org
- **Roger Cornwall, MD**, Associate Professor of Pediatrics, Division of Orthopaedics (Hand Surgery)
Telephone: 513-803-HAND Fax: 513-636-3928
Email: roger.cornwall@cchmc.org
- **Jens Goebel, MD**, Professor, Nephrology
Telephone: 513-636-0993
Email: jens.goebel@cchmc.org
- **Sara C. Knapke, MD**, Clinical Manager and Certified Genetic Counselor
Telephone: 513-803-0211
Email: sara.knapke@cchmc.org
- **Samuel Kocoshis, MD**, Professor
Telephone: 513-636-4415
Email: samuel.kocoshis@cchmc.org
- **Sarah Lawson, MD**, Assistant Professor, Endocrine
Telephone: 513-736-4465
Email: sarah.lawson@cchmc.org
- **Nancy Doan Leslie, MD**, Director, Biochemical Genetics Laboratory
Director, ABMG-Accredited Clinical Training Program
Professor, UC Department of Pediatrics
Telephone: 513-803-2438 Fax: 513-636-7297
Email: nancy.leslie@cchmc.org
- **Charles T. Mehlman, MD**, Director, Musculoskeletal Outcomes Research
Co-Director, Brachial Plexus Center
Professor, Division of Pediatric Surgery
Telephone: 513-636-4785 Fax: 513-803-3928
Email: charles.mehlman@cchmc.org
- **Parinda Mehta, MD**, Associate Professor, UC Department of Pediatrics
Telephone: 513-636-4913 Fax: 513-803-1969
Email: parinda.mehta@cchmc.org
- **Eugene Minevich, MD, FACS, FAAP**, Director, Stone Program
Professor, UC Department of Pediatrics
Telephone: 513-636-4975
Email: kasiani.myers@cchmc.org
- **Kasiani Myers, MD**, Assistant Professor, U.S. Department of Pediatrics
Telephone: 513-636-7287
Email: kasiani.myers@cchmc.org
- **Susan R. Rose, MD, MEd**, Professor, UC Department of Pediatrics, Endocrine
Telephone: 513-636-4744
Email: susan.rose@cchmc.org
- **Michael Rutter, MD**, Pediatric Otolaryngologist and Director of Clinical Research
Professor, UC Department of Pediatrics Otolaryngology - Head and Neck Surgery
Telephone: 513-636-4881 Fax: 513-636-8133
Email: mike.rutter@cchmc.org
- **Thomas Ryan, MD**, Assistant Professor, Cardiology
Telephone: 513-636-9282
Email: thomas.ryan@cchmc.org
- **Mary Sutton, MD**, Assistant Professor, UC Department of Pediatrics
Telephone: 513-636-4222

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- **Robert E. Wood, MD, PhD**, Director, Pulmonary Bronchoscopy Department
Professor, UC Department of Pediatrics
Telephone: 513-636-2776 Fax: 513-636-3845
Email: rewood@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;
- Pharmacological and other clinical trials (e.g., cytokines and flavonoids);
- Fanconi anemia repository for samples and data;
- Expertise in matched sibling donor and unrelated donor transplantation for Fanconi anemia;
- Fanconi anemia complementation and mutation analysis;
- Stem cell collection, storage, and reinfusion;
- Collaboration with, International Fanconi Anemia Registry (**Arleen Auerbach, PhD**).

City Of Hope

1500 E. Duarte Road
Duarte, CA 91010
Telephone: 626-301-8442

- **Joseph Rosenthal, MD**, Chair, Department of Pediatrics, Pediatric Hematology/Hematopoietic Cell Transplantation
Email: jrosenthal@coh.org
- **Anna Pawlowska, MD**, Director, Pediatric Hematology/Hematopoietic Cell Transplantation
Email: apawlowska@coh.org
- **Nicole Karras, MD**, Staff Physician, Pediatric Hematology/Hematopoietic Cell Transplantation
Email: nkarras@coh.org
- **Saro Armenian, MD, MPH**, Associate Professor, Pediatric Hematology/Oncology and Population Sciences, Division of Outcomes Research
Email: sarmenian@coh.org
- **Julie Wolfson, MD**, Staff Physician, Pediatrics; Assistant Professor, Population Sciences & Pediatrics
Email: jwolfson@coh.org
- **Debbie Hitt, RN, BSN, CPHON**, Nurse-Coordinator, Pediatric Hematology/Hematopoietic Cell Transplantation
Email: jwolfson@coh.org

- **Lisa Gutierrez, RN, MSN, CPNP, CPHON**, Nurse Practitioner, Pediatrics
Email: lisagutierrez@coh.org

- **Nadia Ewing, MD**, Director, Pediatric Hematology
Email: newing@coh.org

- **Lisa Pullens, RN, BSN**, Nurse Coordinator, Pediatric Hematology
Email: lpullens@coh.org

- **Joyce Collins, PhD**, Director, Cytogenetics
Email: jcollins@coh.org

- **Christina Cabanillas, LCSW**, Social Worker, Pediatric Hematology/Hematopoietic Cell Transplantation
Email: ccabanillas@coh.org

- **Renee Ortiz, LCSW**, Social Worker, Pediatric Hematology/Hematopoietic Cell Transplantation
Email: reortiz@coh.org

- **April Sorrell, MD**, Pediatric Hematology/Hematopoietic Cell Transplantation and Pediatric Clinical Cancer Genetics

- **Smita Bhatia, MD**, Pediatrics, Hematology/Hematopoietic Cell Transplantation and Director, Population Sciences

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;
- Hematopoietic stem cell transplantation from siblings, matched unrelated donors and umbilical cord blood units;
- Long-term, Fanconi anemia-focused, post-transplant follow-up.

Cohen Children's Medical Center of New York, Bone Marrow Failure Program

Division of Hematology/Oncology and Stem Cell Transplantation
269-01 76th Avenue, Rm. 255
New Hyde Park, NY 11040

- **Adrianna Vlachos, MD**, Assistant Professor of Pediatrics
Head, Bone Marrow Failure Program and Associate Head, Stem Cell Transplant Program
Telephone: 718-470-3611 Fax: 718-343-2961
Email: avlachos@lij.edu

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- **Johnson M. Liu, MD**, Les Nelkin Professor of Pediatrics
Head, Section of Experimental Hematology and Les
Nelkin Memorial Pediatric Oncology Research
Laboratory
Telephone: 718-470-7174
Email: jliu3@nshs.edu
- **Jeffrey M. Lipton, MD, PhD**, Professor of Pediatrics
Chief, Division of Hematology/Oncology and Stem Cell
Transplantation
Telephone: 718-470-3460
Email: jlipton@lij.edu
- **Nancy Barbach, LCSW**, Social Worker, Hematology
Oncology and Stem Cell Transplantation
Telephone: 718-470-3286
Email: nbarbach@nshs.edu

Services Offered:

- Bone Marrow Failure Clinic offering comprehensive evaluation of adult and pediatric FA patients by an experienced team of multi-disciplinary 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.

Fanconi Anemia and Bone Marrow Failure Multi-disciplinary Clinic at the Dana- Farber/Boston Children's Cancer and Blood Disorders Center

Dana-Farber Cancer Institute

450 Brookline Avenue
Boston, MA 02215

- **Eva C. Guinan, MD**, Associate Professor of Radiation
Oncology
Director, Translational Research BWH/DFCI Department
of Radiation Oncology
Telephone: 617-632-4932 Fax: 617-632-3770
Email: Eva_Guinan@dfci.harvard.edu
- **Lisa L. Brennan, BSN, RN**, Research Nurse
Email: Lisa_brennan@dfci.harvard.edu

Services Offered:

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

Boston Children's Hospital

300 Longwood Avenue
Boston, MA 02115

<http://www.childrenshospital.org/fa-bmf>

- **David A. Williams, MD**
Telephone: 617-919-2697
Email: dawilliams@childrens.harvard.edu
- **Colin Sieff, MB BCH**
Telephone: 617-355-2367
Email: colin.sieff@childrens.harvard.edu
- **Mary Cogan, CPNP, RN, CPHON**
Telephone: 617-355-8246 Fax: 617-730-0641
Email: mary.cogan@childrens.harvard.edu

Dana-Farber Cancer Institute

450 Brookline Avenue
Boston, MA 02215

<http://research.dfci.harvard.edu/dandrealab>

- **Alan D'Andrea, MD**
Telephone: 617-632-2080
Email: alan_dandrea@dfci.harvard.edu
- **Lisa A. Moreau, MS, CLSpCG**
Telephone: 617-632-6302
Email: 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; Mary Cogan, CPNP, RN, CPHON**
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: **Susanna Locke, LCSW**
Urology: **Hiep (Bob) Nguyen, MD**

Services Offered:

- Our Nurse Practitioner arranges a comprehensive

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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.
- Facilitation of DNA mutation identification via CAP/CLIA-certified DNA sequencing in the Laboratory Medicine Department at Boston Children's Hospital coordinated with Dr. Bai-Lin Wu and Ann Reinhard. Current available testing for *FANCA*, *FANCC*, and *FANCG* are listed at www.genetests.org. Genetic Counselor: Ann E. Reinhard, MS, CGC, phone: 617-355-4907, ann.reinhard@childrens.harvard.edu.
- 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.

Fred Hutchinson Cancer Research Center

1100 Fairview Ave N
Seattle, WA 98101
Telephone: 206-667-5000
www.fhcr.org

Seattle Children's Hospital

4800 Sand Point Way NE
Seattle, WA 98105
www.seattlechildrens.org

- **Akiko Shimamura, MD, PhD**, Director, Comprehensive Marrow Failure/MDS Program, Pediatric Hematology-Oncology, Seattle Children's Hospital
Member, Fred Hutchinson Cancer Research Center
Professor of Pediatrics, University of Washington School of Medicine
Telephone: 206-987-2106 Fax: 206-987-3946
Email: ashimamu@fredhutch.org

- **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
Telephone: 206-667-4425 Fax: 206-667-6124
Email: hkiem@fredhutch.org
- **K. Scott Baker, MD, MS**, Member and Director, Pediatric Blood and Marrow Transplantation and Survivorship Programs, Fred Hutchinson Cancer Research Center and Seattle Children's Hospital
Professor of Pediatrics, University of Washington School of Medicine
Telephone: 206-987-2106 Fax: 206-667-5899
Email: ksbaker@fredhutch.org
- **Ann Woolfrey, MD**, Director, Unrelated Donor Program
Member, Fred Hutchinson Cancer Research Center, Pediatric Blood and Marrow Transplant Program
Professor of Pediatrics, University of Washington School of Medicine
Telephone: 206-667-4453 Fax: 206-667-4310
Email: awoolfre@fredhutch.org
- **Jennifer Adair, PhD**, Clinical Research Associate
Fred Hutchinson Cancer Research Center
Telephone: 206-667-7110 Fax: 206-667-6124
Email: jadair@fredhutch.org
- **Pam Becker, MD, PhD**, Associate Professor of Medicine, University of Washington School of Medicine, Department of Hematology
Telephone: 206-288-2294 Fax: 206-543-3560

Services Offered:

- Comprehensive Fanconi Anemia Program including evaluation and management of new and previously diagnosed pediatric and adult patients with FA;
- Personalized treatment regimens by specialists in hematology, bone marrow transplantation, cardiology, endocrinology, genetics, gastroenterology, nephrology, urology, orthopedics, and 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;
- Collaboration with Hospital de Clinicas, University of Parana, Brazil (Carmem Bonfim, MD and Ricardo Pasquini, MD, PhD).

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Hackensack University Medical Center **Tomorrow's Children Institute**

30 Prospect Avenue
Hackensack, NJ 07601
www.HackensackUMC.org

- **Alfred P. Gillio, MD**, Director, Blood and Marrow Transplantation Program
Joseph M. Sanzari Children's Hospital
Institute of Pediatric Cancer & Blood Disorders at
Hackensack University Medical Center
Telephone: 551-996-5600 Fax: 551-996-0521
Email: agillio@hackensackumc.org
- **Barbara Adler-Brecher, MS**
Email: badler-brecher@hackensackumc.org

Services Offered:

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

International Fanconi Anemia Registry (IFAR) **The Rockefeller University**

Part of New York Consortium for Inborn Errors of Hematopoiesis and Immunity (see also, Memorial Sloan Kettering Cancer Center, New York Presbyterian Hospital, and Hospital for Special Surgery)

Laboratory of Genome Maintenance
1230 York Avenue, Box 182
New York, NY 10065
<http://lab.rockefeller.edu/smogorzewska/ifar/>
Facebook: *Fanconi Anemia Registry* or *Agata Smogorzewska*

- **Agata Smogorzewska, MD, PhD**, Principal Investigator
Telephone: 212-327-7850, or 8862 Fax: 212-327-8262
Email: asmogorzewska@rockefeller.edu
<http://lab.rockefeller.edu/smogorzewska>
- **Arleen D. Auerbach, PhD**, Co-Investigator
Telephone: 212-327-7533, or 8862 Fax: 212-327-8262
Email: auerbac@rockefeller.edu
- **Jennifer Kennedy, MS, CGC**, Genetic Counselor/Study Coordinator
Telephone: 212-327-8612, or 8862 Fax: 212-327-8262
Email: jkennedy@rockefeller.edu

Genetic Services on Research Protocol:

- International Fanconi Anemia Registry (IFAR);

- Mutation screening of probands (gene specific sequencing, next generation sequencing, whole exome sequencing, SNP analysis, and/or CHG) for genes under investigation by the laboratory;
- Genomic analysis of tumor tissue from enrolled participants: surgical samples needed;
- Derivation of lymphoblastoid cell line for complementation testing; maintenance of Fanconi anemia cell and DNA repositories;
- Genetic counseling for individuals involved in the IFAR, and their family members.

Testing Services on Research Protocol:

- Mutation analysis (research)
- *FANCD2* western blot (research)

Co-Principal Investigators for the IFAR:

- **Farid Boulad, MD**, Memorial Sloan-Kettering Cancer Center, New York, NY
Telephone: 212-639-6684 Fax: 212-717-3555
Email: bouladf@mskcc.org
- **Stella Davies, MBBS, PhD, MRCP**, Cincinnati Children's Hospital Medical Center
Cincinnati, OH
Telephone: 513-636-1371 Fax: 513-636-3459
Email: stella.davies@cchmc.org
- **Margaret L. MacMillan, MD**, University of Minnesota, Minneapolis, MN
Telephone: 612-626-2778 Fax: 612-626-2815
Email: macmi002@umn.edu
- **David I. Kutler, MD**, Head and Neck Surgeon, Weill Cornell Medical Center/New York Presbyterian Hospital, New York, NY
Telephone: 646-962-4323, 646-962-0100
Email: dik2002@med.cornell.edu

Memorial Sloan-Kettering Cancer Center

Part of New York Consortium for Inborn Errors of Hematopoiesis and Immunity (see also, Rockefeller University, New York Presbyterian Hospital, and Hospital for Special Surgery)

1275 York Avenue
New York, NY 10065
Email: www.mskcc.org

- **Farid Boulad, MD**, Medical Director, Pediatric Day Hospital
Attending Physician
Bone Marrow Transplant Service
Department of Pediatrics

BONE MARROW FAILURE

Telephone: 212-639-6684 Fax: 212-717-3555

Email: bouladf@mskcc.org

- **Bhuvanesh Singh, MD, PhD**, Head and Neck Cancer
Telephone: 212-639-2024
Email: singhb@mskcc.org
- **Julia A. Kearney, MD**, Psychiatry and Pediatrics
Telephone: 646-888-0028
Email: Kearneyj@mskcc.org
- **Jennifer Kennedy, MS, CGC**, Genetic Counselor/Study Coordinator
Telephone: 646-888-4102
Email: Kennedyj@mskcc.org
- **Mercedes Castiel, MD**, Gynecology
Telephone: 646-962-2110
Email: mec9109@med.cornell.edu

Services Offered:

- Comprehensive care and long-term follow-up of new and previously diagnosed patients, including:
- Overall medical and hematological care;
 - Hematopoietic stem cell transplantation with related, unrelated, matched or mismatched donors;
 - Psychological evaluation and treatment;
 - Head and neck cancer evaluation and treatment;
 - Gynecological cancer evaluation and treatment;
 - Endocrinology clinic;
 - Direct collaboration with Dr. Agata Smogorzewska, The Rockefeller University, for mutation screening, complementation group analysis, somatic mosaicism studies, and participation in the International Fanconi Anemia Registry (IFAR);
 - Direct collaboration with Dr. Aaron Daluiski at the Hospital for Special Surgery, Pediatric Hand Registry (IFAR);
 - 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
Telephone: 212-241-3443
Email: christopher-e.walsh@msnyuhealth.org

- **Luis Isola, MD**, Director, Stem Cell Transplantation Program
- **Anne Hurlet-Jensen, 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 Clinical Genetics Branch, National Institutes of Health

9609 Medical Center Drive, Room 6E452, MSC 9772
Rockville, MD 20850

www.marrowsfailure.cancer.gov

- **Lisa Leathwood, RN, BSN**, Research Nurse and Study Coordinator
Telephone: 301-212-5250 and 800-518-8474
Fax: 301-212-5281
Email: lisleathwood@westat.com
- **Blanche P. Alter, MD, MPH, FAAP**
Telephone: 240-276-7239 Fax: 240-276-7836
Email: alterb@mail.nih.gov
- **Neelam Giri, MD, FAAP**
Telephone: 240-276-7256 Fax: 240-276-7836
Email: girin@mail.nih.gov

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.

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Gynecology Consultation Service at the NIH Clinical Center; Eunice Kennedy Shriver National Institute of Child Health and Human Development

10 Center Dr. Suite 1-3140
Bethesda, MD 20892

- **Pamela Stratton, MD**, Chief, Gynecology Consult Service
Telephone: 301-496-9079 Fax: 301-480-6703
Email: strattop@mail.nih.gov

Services Offered:

- Gynecology consulting service is only available to FA patients/family members enrolled in Blanche Alter's protocol.

Head and Neck Surgery Branch at the NIH

Building 10, Room 4-2732
10 Center Drive
Bethesda, MD 20892

- **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
Email: vanwaesc@nidcd.nih.gov

Dermatology Consultation Service at the NIH Clinical Center; Center for Cancer Research

Building 10, Room 12N238
Bethesda, MD 30892-1908

- **Edward Cowen, MD**, Head, Dermatology Consultation Service
Telephone: 301-496-4299 Fax: 301-402-2943
Email: cowene@mail.nih.gov

Services Offered:

- Dermatology consulting service is only available to FA patients/family members enrolled in Blanche Alter's protocol.

Oregon Health & Science University

Doernbecher Children's Hospital

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

- **Peter Kurre, MD**, Professor of Pediatrics
Telephone: 503-494-1543 Fax: 503-494-0714
Email: kurrepe@ohsu.edu

- **Evan Shereck, MD**, Associate Professor of Pediatrics
Telephone: 503-494-0829 Fax: 503-494-0714
Email: shereck@ohsu.edu

Services Offered:

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

Stanford School of Medicine

The Bass Center for Childhood Cancer and Blood Diseases

725 Welch Road
Palo Alto, CA 94304
www.stanfordchildrens.org

Pediatric Hematology/Oncology Academic Office

1000 Welch Road, Suite 300
Palo Alto, CA 94304

Hematology/Oncology

Telephone: 650-723-5535

- **Bertil Glader, MD, PhD**, Professor, Pediatrics and Pathology
Email: bglader@stanford.edu
- **Michael Jeng, MD**, Associate Professor, Pediatrics
Email: mjeng@stanford.edu
- **Anu Narla, MD**, Assistant Professor, Pediatrics
Email: anunarla@stanford.edu
- **Judith Lea, RN, MSN, PNP**, Hematology Nurse Practitioner
Email: jlea@stanfordchildrens.org
- **Kirsten Mouradian, RN, MSN, FNP**, Hematology Nurse Practitioner
Email: kmouradian@stanfordchildrens.org

Stem Cell Transplant

Telephone: 650-725-9250

- **Kenneth Weinberg, MD**, Professor, Pediatrics
Email: kw1@stanford.edu
- **Rajni Agarwal, MD**, Assistant Professor, Pediatrics
Email: rajnia@stanford.edu

BONE MARROW FAILURE

- **Sandhya Kharbanda, MD**, Assistant Professor, Pediatrics
Email: ksandhya@stanford.edu
- **Karen Kristovich, RN, MSN, PNP**, Stem Cell Transplant Nurse Practitioner
Email: kkristovich@stanfordchildrens.org
- **Elizabeth Schnorr, RN, PNP**, Stem Cell Transplant Coordinator
Email: eschnorr@stanfordchildrens.org
- **Donna Reimer, RN**, Stem Cell Transplant Coordinator
Email: dreimer@stanfordchildrens.org
- **Gayle Humphrey, RN**, Stem Cell Transplant Coordinator
Email: ghumphrey@stanfordchildrens.org
- **Margaret L. MacMillan, MD**, Fanconi Anemia Comprehensive Care Program Leader
Associate Professor, Department of Pediatrics, Division of Blood and Marrow Transplantation
Telephone: 612-626-2778 Fax: 612-626-2815
Email: macmi002@umn.edu
- **Jakub Tolar, MD, PhD**, Director, Stem Cell/Gene Therapies
Albert and Eva Corniea Chair
Associate Professor, Department of Pediatrics, Division of Blood and Marrow Transplantation
Telephone: 612-625-2912 Fax: 612-624-3913
Email: tolar003@umn.edu
- **Anna Petryk, MD**, Pediatric Endocrinology
Telephone: 612-624-5409 Fax: 612-626-5262
Email: petry005@umn.edu
- **Sarah Jane Schwarzenberg, MD**, Pediatric Gastroenterology
Telephone: 612-624-1133 Fax: 612-626-0639
Email: schwa005@umn.edu
- **Frank Ondrey, MD, PhD, FACS**, Otolaryngology
Telephone: 612-624-5900
Email: ondre002@umn.edu
- **Rahel G. Ghebre, MD**, Gynecologic Oncology
Telephone: 612-626-2613 Fax: 612-626-0665
Email: ghebr004@umn.edu
- **Ann E. Van Heest, MD**, Dept. of Orthopaedic Surgery
Telephone: 612-273-8086 Fax: 612-273-7959
Email: Vanhe003@umn.edu
- **Jo-Anne H. Young, MD**, Infectious Disease
Telephone: 612-624-9996 Fax: 612-625-4410
Email: Vanbu004@umn.edu
- **Pat Fidler, RN**, Nurse Coordinator
Telephone: 612-273-8480 Fax: 612-273-2919
Email: pfidler1@fairview.org
- **Rebecca Tryon, MS, CGC**, Genetic Counseling
Telephone: 612-624-1510 Fax: 612-624-6645
Email: rtryon1@fairview.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.

Texas Children's Hospital

1102 Bates, FC1200
Houston, TX 77030

- **Alison Bertuch, MD, PhD**
Telephone: 832-824-4579
Email: abertuch@txch.org

University of Minnesota

Fanconi Anemia Comprehensive Care Program

University of Minnesota Medical Center and Masonic Children's Hospital

MMC 366
420 Delaware Street, S.E.
Minneapolis, MN 55455
Telephone: 612-273-2800
<http://bmt.umn.edu/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
Telephone: 612-626-2961 Fax: 612-626-4074
Email: wagne002@umn.edu

Journey Clinic

Center for Pediatric Blood and Marrow Transplantation
East Building, Ninth Floor
2450 Riverside Avenue
Minneapolis, MN 55454
<http://bmt.umn.edu/>
Telephone: 612-273-2800 Fax: 612-273-2919

BONE MARROW FAILURE

Services Offered:

- The University of Minnesota Fanconi Anemia Comprehensive Care Program (FACCP) utilizes a multi-disciplinary 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 in the new Children's Hospital 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.

University of Texas Southwestern Medical Center **Pediatric Bone Marrow Failure Program** **Children's Health Dallas**

Margaret Allen Gill Center for Cancer and Blood Disorders
1935 Medical District Drive
Dallas, TX 75235
Telephone: 214-456-2382 or 214-456-6102
Email: BoneMarrowFailure@UTSouthwestern.edu

- **Zora R. Rogers, MD**, Professor of Pediatrics, Hematology-Oncology, Clinical Director, Hematology and Bone Marrow Failure Program
Email: zora.rogers@utsouthwestern.edu
- **Tiffany Simms-Waldrup, MD**, Assistant Professor of Pediatrics, Hematology-Oncology
Head, Bone Marrow Failure Stem Cell Transplant Program
Email: tiffany.simms-waldrup@utsouthwestern.edu

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, dental/oral-maxillo-facial surgery, nephrology, hand and orthopedic surgery interested in the unique needs of persons with Fanconi anemia;
- Psychology, social work, physical and occupational therapy services.

BONE MARROW DONOR REGISTRIES

Delete Blood Cancer DKMS

100 Broadway, 6th Floor
New York, NY 10005
www.dkmsamericas.org
Telephone: 1-866-340-3567
Email: info@dkmsamericas.org

Delete Blood Cancer 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.

Delete Blood Cancer DKMS:

- is the world's largest bone marrow donor center, with more than 3 million registered donors;
- can register new bone marrow donors anywhere in the U.S;
- 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;
- offers doctors immediate search of our database;
- does not require new donors to pay the \$65 registration fee.

National Marrow Donor Program® / Be The Match®

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

Be The Match helps patients with leukemia, lymphoma and other diseases who need a marrow or umbilical cord blood transplant. People can join the Be The Match Registry® - the world's largest listing of potential marrow donors and donated cord blood units - contribute financially, and volunteer. Patients and their families can also turn to Be The Match for support and resources before, during, and after transplant. Be The Match is operated by the National Marrow Donor Program® (NMDP), a nonprofit organization that matches patients with donors, educates health care professionals, and conducts research so more lives can be saved. Learn more at BeTheMatch.org/patient.

Services Offered:

Be The Match has a team dedicated to supporting patients, caregivers and families before, during, and after transplant. We offer confidential one-on-one support, financial guidance, and free educational resources. Our goal is to get you what you need, when you need it. We offer support in more than 100 languages, including Spanish bilingual staff and translated materials.

CORD BLOOD REGISTRIES

Cord Blood Registry®

Designated Treatment Program (DTP)
Cord Blood Registry
1200 Bayhill Drive, 3rd Floor
San Bruno, CA 94066
www.cordblood.com
Telephone: 1-888-932-6568

Cord Blood Registry® (CBR) is the world's largest and most experienced family cord blood and tissue stem cell bank.

Families wishing to store their newborn's umbilical cord blood stem cells for a family member diagnosed with a life-threatening disease treatable by donor stem cells (including Fanconi anemia) are encouraged to apply for CBR's Designated Treatment Program. If eligible, CBR will provide the collection kit and process and store the newborn's cord blood and cord tissue sample for five years without any cost to the family. The family member must be a first-degree blood relative (full-sibling or parent) to the newborn and have an oncologist/hematologist or other treating physician agree that the stem cells can be used for treatment.

DERMATOLOGY

National Cancer Institute

Clinical Genetics Branch, Division of Cancer Epidemiology and Genetics Clinical Genetics Branch, National Institutes of Health

9609 Medical Center Drive, Room 6E452, MSC 9772
Rockville, MD 20850

www.marrowsfailure.cancer.gov

Dermatology Consultation Service at the NIH Clinical Center; Center for Cancer Research

Building 10, Room 12N238
Bethesda, MD 30892-1908

- **Edward Cowen, MD**, Head, Dermatology Consultation Service
Telephone: 301-496-4299 Fax: 301-402-2943
Email: cowene@mail.nih.gov

Services Offered:

- Dermatology consulting service is only available to FA patients/family members enrolled in Blanche Alter's protocol.

Brigham and Women's Hospital

Brigham Dermatology Associates
221 Longwood Avenue
Boston, MA 02115

- **Jennifer Lin, MD**,
Telephone: 617-732-4918 Fax: 617-582-6060
Email: jylin@partners.org

ENDOCRINOLOGY

Cincinnati Children's Hospital Medical Center **Fanconi Anemia Comprehensive Care Center**

3333 Burnet Avenue
Cincinnati, OH 45229

- **Susan R. Rose, MD, MEd**, Professor of Pediatrics, Pediatric Endocrinology
Telephone: 513-636-4744
Email: susan.rose@cchmc.org

University of Minnesota **Fanconi Anemia Comprehensive Care Program** **University of Minnesota Medical Center and** **Masonic Children's Hospital**

East Bldg Rm MB671
2450 Riverside Avenue
Minneapolis, MN 55454

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

GASTROENTEROLOGY

Cincinnati Children's Hospital Medical Center Fanconi Anemia Comprehensive Care Center

3333 Burnet Avenue
Cincinnati, OH 45229

- **Samuel Kocoshis, MD**, Professor
Telephone: 513-636-4415
Email: samuel.kocoshis@cchmc.org

University of Minnesota Fanconi Anemia Comprehensive Care Program University of Minnesota Medical Center and Masonic Children's Hospital

East Building, 6th Floor
2450 Riverside Avenue
Minneapolis, MN 55454

- **Sarah Jane Schwarzenberg, MD**, Director, Pediatric Gastroenterology, Hepatology and Nutrition
Telephone: 612-624-1133 Fax: 612-626-0639
Email: schwa005@umn.edu

GENETIC COUNSELING

The Rockefeller University

Part of New York Consortium for Inborn Errors of Hematopoiesis and Immunity (see also, Memorial Sloan Kettering Cancer Center, New York Presbyterian Hospital, and Hospital for Special Surgery)

1230 York Avenue Box 182

New York, NY 10065

Fax: 212-327-8262

Facebook: *Fanconi Anemia Registry or Agata Smogorzewska* (PI of IFAR)

- **Jennifer Kennedy, MS, CGC**, Certified Genetic Counselor*
Telephone: 212-327-8612
Email: jkennedy@rockefeller.edu

*Genetic counseling provided for IFAR participants and their family members. Please see page 6 for more information about IFAR.

Journey Clinic

Center for Pediatric Blood and Marrow Transplantation

East Building, Ninth Floor

2450 Riverside Avenue

Minneapolis, MN 55454

Website: <http://bmt.umn.edu/world-class-bmt-program/fanconi-anemia.php>

Telephone: 612-273-2800 Fax: 612-273-2919

- **Rebecca Tryon, MS, CGC**
Telephone: 612-624-1510 Fax: 612-624-6645
Email: rtryon1@fairview.org

GYNECOLOGY

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
Email: strattp@mail.nih.gov

University of Minnesota

Fanconi Anemia Comprehensive Care Program University of Minnesota Medical Center, Masonic Children's Hospital, Fairview

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

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

The New York Presbyterian Hospital Weill Cornell Medical Associates

Part of New York Consortium for Inborn Errors of Hematopoiesis and Immunity (see also, Memorial Sloan Kettering Cancer Center, The Rockefeller University, and Hospital for Special Surgery)
2315 Broadway
New York, NY 10024
Telephone: 214-456-2382 or 214-456-6102

- **Mercedes Castiel, MD**, Gynecology
Telephone: 646-962-2110
Email: mec9109@med.cornell.edu

Services Offered:

Gynecological cancer evaluation and treatment.

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
Email: 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 Fanconi Anemia Comprehensive Care Center

3333 Burnet Avenue
Cincinnati, OH 45229

- **Roger Cornwall, MD**, Associate Professor of Pediatrics, Division of Orthopaedics (Hand Surgery)
Telephone: 513-803-HAND Fax: 513-636-3928
Email: roger.cornwall@cchmc.org

HAND SURGEONS, cont'd

Hospital for Special Surgery

Part of New York Consortium for Inborn Errors of Hematopoiesis and Immunity (see also, Memorial Sloan Kettering Cancer Center, The Rockefeller University, and The New York Presbyterian Hospital)

535 E 70th Street, 5th Floor
New York, NY 10065

- **Aaron Daluiski, MD**, Orthopedics – Hand and Upper Extremity Surgery, Lerner Children's Pavilion
Telephone: 212-606-1284

University of Virginia Health System

- **A. Bobby Chhabra, MD**, Professor and Chair
Department of Orthopaedic Surgery
Lillian T. Pratt Distinguished Professor of Orthopaedic Surgery
Professor of Plastic and Reconstructive Surgery
Telephone: 434-243-0218 Fax: 434-243-0290
Appointments: 434-982-4263
Email: AC2H@hscmail.mcc.virginia.edu
Websites: <http://uvaortho.com/chhabra>
<http://orthodoc.aaos.org/bobbychhabra/>

Services Offered:

- Surgery for congenital hand and upper extremity differences.

Shriners Hospital for Children

3551 North Broad Street
Philadelphia, PA 19140

- **Scott Kozin, MD**, Chief of Staff, Shriners Hospital for Children Philadelphia, Professor, Orthopedic Surgery, Temple University
Telephone: (215) 430-4034
Email: skozin@shrinenet.org
- **Linda Filipone**, Nurse Coordinator
Telephone: (215) 430-4074
Email: lfilipone@shrinenet.org
- **Maureen Fulcher**, Nurse Assistant
Telephone: (215) 430-4204
Email: mfulcher@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.

OPHTHALMOLOGY

Boston Foundation for Sight

464 Hillside Avenue, Suite 205
Needham, MA 02494
www.bostonsight.org

- **Gillian Fontana**, New Patient Affairs Coordinator
Telephone: 781-726-7501 Fax: 781-726-7310
Email: GFontana@bostonsight.org

Boston Foundation for Sight is a non-profit organization with a mission to improve the quality of life for patients and their families by transforming the understanding, treatment, and care of complex corneal disease.

Services Offered:

BostonSight® PROSE treatment is a process by which visual function is restored to a patient with complex corneal disease, enabling them to reclaim their life. During PROSE treatment, doctors customize prosthetic devices to replace or support impaired ocular surface functions, improving vision and reducing pain and light sensitivity.

OTOLARYNGOLOGY (Ear, Nose, and Throat) and HEAD & NECK CANCER

Cincinnati Children's Hospital Medical Center Fanconi Anemia Comprehensive Care Center

3333 Burnet Avenue
Cincinnati, OH 45229

- **Michael Rutter, MD**, Professor of Pediatrics, Division of Otolaryngology
Telephone: 513-636-4356 Fax: 513-636-8133
Email: mike.rutter@cchmc.org

Cornell University, Weill Medical College

Department of Otolaryngology-Head and Neck Surgery
1305 York Avenue, 5th Floor
New York, NY 10021

- **David Kutler, MD**, Associate Professor
Telephone: 646-962-4323
Email: dik2002@med.cornell.edu

Clinical Interests: Head and neck cancer in Fanconi anemia patients, tongue cancer, disorder of nasal sinus, skull base surgery, pharynx, thyroid cancer, benign tumor of oral cavity, thyroid surgery, head and neck cancer surgery, and salivary disorder.

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
Email: 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 Avenue
New York, NY 10021

- **Bhuvanesh Singh, MD, PhD**, Head and Neck Cancer
Telephone: 212-639-2024
Email: 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**, Senior Staff Clinician, Head and Neck Surgery Branch, National Institute on Deafness and Other Communication Disorders
Professor, Department of Otolaryngology, Head and Neck Surgery, Georgetown University Hospital
Telephone: 301-496-5369 Fax: 301-402-0409
Email: 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
Email: vanwaesc@nidcd.nih.gov

Clinical Interests: Diagnosis and investigational treatment of head and neck mucosal lesions and cancers.

University of California at San Francisco

CTSI, Box 0558
550 16th Street, 6th Floor
San Francisco, CA 94158-2549

- **Jennifer Rubin Grandis, MD, FACS**
Telephone: 415-514-8084
Email: Jennifer.Grandis@ucsf.edu

Specialty: Otolaryngology

Sub Specialty: Head and neck surgery

University of Chicago

5841 S. Maryland Avenue, MC6092
Chicago, IL 60634

- **Everett Vokes, MD**, John E. Ulmann Professor, Chairman, Department of Medicine, Physician-in-Chief, University of Chicago Medicine and Biological Sciences
Telephone: 773-702-9306 Fax: 773-702-4427
Email: evokes@medicine.bsd.uchicago.edu

Clinical Interests: Hematology, oncology, head and neck cancer, and lung cancer.

OTOLARYNGOLOGY (Ear, Nose, and Throat) and HEAD & NECK CANCER

- **Tanguy Seiwert, MD**, Assistant Professor of Medicine
Telephone: 773-702-2452 Fax: 773-702-3002

Clinical Interests: Head and neck cancer and lung cancer.

University of Cincinnati, College of Medicine

Department of Otolaryngology
P.O. Box 670528
Cincinnati, OH 45267

- **Keith A. Casper, MD**, Assistant Professor
Telephone: 513-475-8400 Fax: 513-558-4477
Email: 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

Masonic Children's Hospital

MMC 396, University of Minnesota
420 Delaware Street, SE
Minneapolis, MN 55455

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

Clinical Interests: Head and neck cancer, precancerous mouth and throat lesions, comprehensive general otolaryngology, and facial trauma.

University of Texas Health Science Center at Houston

Department of Otorhinolaryngology - Head & Neck Surgery
6400 Fannin, Suite 2700
Houston, TX 77030

- **Ron J. Karni, MD**, Assistant Professor of Otolaryngology, Assistant Professor of Medical Oncology
Telephone: 713-486-5000 and 713-500-5410
Email: ron.j.karni@uth.tmc.edu

Clinical Interest: Head and neck cancer, robotic, and minimally-invasive surgery of the head and neck.

Services Offered: Management of head and neck cancer including surgery, chemotherapy, and radiation oncology.

University of Texas MD Anderson Cancer Center

1515 Holcombe Boulevard, Unit 1445
Houston, TX 77030

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

Clinical Interests: Head and neck cancer, salivary carcinoma, and thyroid cancer, human papillomavirus, and sarcomas of the head and neck.

Services Offered: Multi-disciplinary evaluation and treatment of head and neck tumors.

PRE-IMPLANTATION GENETIC DIAGNOSIS

Genesis Genetics

705 S. Main Street
Plymouth, MI 48170
www.genesisgenetics.org

- **John Kitchen, PhD**
Telephone: 313-579-9650 Fax: 313-544-4006
- **Elizabeth Cameron, MS, CGC**, Certified Genetic Counselor
Telephone: 313-579-9650 Fax: 313-544-4006
Email: counselor@genesisgenetics.org
- **Jeannie Klavanian, MS**, Genetic Counselor
Telephone: 313-579-9650 Fax: 313-544-4006

Services Offered:

- Pre-pregnancy/pre-implantation diagnosis for Fanconi anemia (including types A, C, D2, F, G, I, and J, among others). Helping a couple begin a pregnancy knowing their baby will not have Fanconi anemia.
- Extensive experience in Fanconi anemia 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 anemia-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 for these PGD services. Our clinic collaborates with the leading molecular biologists in Fanconi anemia research, with over 230 in vitro fertilization programs, and with internationally respected Fanconi anemia transplantation experts.

Reproductive Genetic Innovation

2910 MacArthur Boulevard
Northbrook, IL 60062
Telephone: 847-400-1515
www.rgipgd.com
Email: info@rgipgd.com

- **Stephanie Peters, MGC, CGC**
- **Divya Shah, MS, LCGC**
- **Agnes Machaj, MS**

Services Offered:

- Pre-implantation genetic diagnosis for any known Fanconi anemia mutations with or without HLA matching for sibling bone marrow donation;
- Pioneers in PGD; first laboratory in the world to successfully perform PGD for FA and HLA matching. Highly accurate state-of-the-art testing methods;
- Able to test for chromosomal abnormalities in addition to FA and HLA on a single cell from an embryo;
- Free consultations with genetic counselors (in-person or via telephone);
- Travel to our facility is not necessary as we collaborate with hundreds of in vitro fertilization clinics all over the world.

RADIATION ONCOLOGY

University of Pittsburgh Cancer Institute

UPMC Cancer Pavilion, POB 2
5150 Centre Avenue
Pittsburgh, PA 15232

- **Joel Greenberger, MD**
Telephone: 412-647-3602
Email: greenbergerjs@upmc.edu

Clinical Interests:

- Radiation sensitivity in Fanconi anemia

Services Offered:

- Consultation (telephone, email, in person) with families and physicians regarding the use of radiation in a person with Fanconi anemia.
- Treat patients diagnosed with cancer by utilizing various forms of radiation therapy.

U.S. Fanconi Anemia Testing Resources

U.S. Fanconi Anemia Testing Resources

Institution	Test
<p>Cincinnati Children’s Hospital Medical Center Fanconi Anemia Comprehensive Care Center Bone Marrow Failure Clinic Cincinnati, OH</p> <p>Erica L. Goodridge, RN, BSN, CPN, Care Manager, Fanconi Anemia Comprehensive Care Center 513-636-3218 Toll-free: 1-800-344-2462, ext. 3218 <i>erica.goodridge@cchmc.org</i></p>	<p>DEB/MMC diagnostic testing using blood or bone marrow samples.</p> <p>Complementation analysis for <i>A, B, C, E, F, G, L, I</i> (<i>I</i> analysis via skin testing only) and <i>FANCD2</i> (via western blot). Testing done with blood, skin biopsy or amniocytes. Note: This testing must be requested by clinicians or researchers.</p> <p>Molecular testing to find specific mutations for patients with <i>FANCA, FANCC,</i> or <i>FANCG</i>.</p> <p>Prenatal diagnostic molecular testing for <i>FANCA, FANCC,</i> or <i>FANCG</i>.</p> <p>Carrier detection testing for patients with a family history of FA, from complementation groups <i>FANCA, FANCC,</i> or <i>FANCG</i>.</p>
<p>Dana-Farber Cancer Institute Boston, MA</p> <p>Alan D’Andrea, MD 617-632-2080 <i>alan_dandrea@dfci.harvard.edu</i></p> <p>Lisa Moreau, MS 617-632-6302 <i>lisa_moreau@dfci.harvard.edu</i></p>	<p>DEB/MMC diagnostic testing using blood, bone marrow, or skin fibroblasts.</p> <p><i>FANCD2</i> western blot (Research).</p> <p>FA carrier testing.</p>
<p>Indiana University School of Medicine Wells Center, Riley Hospital for Children Indianapolis, IN</p> <p>Helmut Hanenberg, MD 317-278-9290 or 274-2980 Fax 317-274-2852 <i>hhanenbe@iupui.edu</i></p>	<p>Complementation Group (Research): Use of retroviruses for <i>A, B, C, DI, D2, E, F, G, I, J, L, M, N, O, P, Q</i> in primary T-cells, bone marrow cells, or fibroblast cells or in lymphoblast cell lines (LCL) (Research).</p> <p><i>FANCD2</i> western blotting (research).</p>
<p>Indiana University School of Medicine Cytogenetic Lab, Dept. of Medical & Molecular Genetics Indianapolis, IN</p> <p>Gail H. Vance, MD 317-274-2243/317-278-0172 Fax 317-278-1616 <i>ghvance@iupui.edu</i></p>	<p>Diagnostic testing: Cytogenetic breakage studies with DEB on peripheral blood and fibroblast specimens.</p> <p>Mutation testing for <i>FANCA, FANCC,</i> and <i>FANCG</i>. Collaborative mutation testing and research with Gail Vance, MD, and Shaochun Bai, PhD, Medical & Molecular Genetics, Indianapolis, IN.</p>

Institution	Test
<p>Oregon Health & Science University Portland, OR</p> <p>Susan Olson, PhD, FACMG (cytogenetics) 503-494-5964 <i>olonsu@ohsu.edu</i></p> <p>C. Sue Richards, PhD, FACMG (molecular) 503-494-5400 <i>richarsu@ohsu.edu</i></p>	<p>DEB/MMC diagnostic testing - blood and skin fibroblasts.</p> <p>Next-Generation sequencing comprehensive Fanconi panel is now available for all <i>FANC</i> genes. Exon-centric microarray is also available for detection of deletions and duplications in all <i>FANC</i> genes.</p> <p>Complementation and full gene mutation analysis by Sanger sequencing for <i>A, C, G, E, F</i> as well as targeted sequencing of any exon is available. Deletion/duplication analysis for <i>FANCA</i> is also available by MLPA. Prenatal testing available for known mutations. Custom sequencing for known mutations in Fanconi genes other than <i>A, C, G, E, F</i> is also available. Please see GeneTests for further information.</p> <p>FA carrier testing.</p>
<p>Prevention Genetics 3700 Downwind Drive Marshfield, WI 54449</p> <p>Michael Chicka, PhD 715-387-0484 <i>michael.chicka@preventiongenetics.com</i></p>	<p>FA gene sequencing and deletion/duplication analysis for molecular diagnosis. Genes analyzed (at press): <i>FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCI/BRIP1, FANCL, FANCM, FANCN/PALB2, FANCO/RAD51C, FANCP/SLX4, FANCO/ERCC4</i></p> <p>Tests offered include:</p> <ul style="list-style-type: none"> • Next-Generation and Sanger Sequencing FA Panels for identifying the particular FA gene and variant(s) of interest; • Sanger sequencing of individual FA genes when gene of interest is known; • Sanger confirmation of all pathogenic variants and variants of unknown significance found by Next-Generation Sequencing; • Detection of large deletions/duplications using custom designed, high-density, gene-centric array CGH technologies; • Targeted variant analysis for familial variants and for confirming research findings; • Prenatal testing.
<p>Quest Diagnostics Chantilly, VA</p> <p>Dr. Steven Schonberg 800-336-3718 x7285</p> <p>Quest Diagnostics San Juan Capistrano, CA</p> <p>Joy Redman, MS, MBA, CGC 800-642-4657, ext. 4279 <i>Joy.B.Redman@questdiagnostics.com</i></p>	<p>DEB diagnostic testing performed on blood, bone marrow, and skin fibroblasts (in addition to prenatal testing): the doctor needs to specify DEB test, otherwise it is possible that a DNA carrier test for <i>FANCC</i> may be done.</p> <p>Mutation analysis for <i>FANCC</i> (IVS4+4A>T and 322delG).</p> <p>Prenatal diagnosis.</p>

U.S. Fanconi Anemia Testing Resources

Institution	Test
<p>University of Minnesota Physicians Outreach Laboratories Minneapolis MN</p> <p>Bharat Thyagarajan, MD, PhD, Director Matt Bower, MS, CGC, Contact 612-624-8948 <i>mbower1@fairview.org</i></p>	<p>Next Generation sequencing. These methods will detect point mutations and small indel mutations (<18bp).</p> <p>Copy number analysis for large deletions/duplications is under development and will be available in 2015.</p> <p>Targeted mutation analysis available in families with known mutations. Lab requires a positive control to develop primers.</p> <p>Complementation and analyze genes: <i>FANCA, FANCB, FANCC, FANCD1, FANCD2, FANCE, FANCF, FANCG, FANCI, FANJ (BRIP1), FANCL, FANCM, FANCN (PALB2), FANCO(RAD51C), FANCP(SLX4), FANQ (ERCC4)</i>.</p>

Please note:

- The laboratories listed are CLIA (Clinical Laboratories Improvement Amendments)-certified.
- Estimated costs for diagnostic tests range from approximately \$1,000 - \$1,200. Check with your insurance company for coverage. The average turnaround time for testing results is about 10 working days. For more information, please contact the labs directly.
- Additional testing information for Fanconi anemia is available at: <http://geneclinics.org>.
 - Select “Gene Reviews”.
 - Select “Search by Disease”.
 - Enter “Fanconi anemia” into the text box.
- “Research” indicates that the results will be used for research purposes only and may not be shared with referring physician/family.

International Fanconi Anemia Treatment Resources

Editors' note and disclaimer: The resource list provided in this guide is not comprehensive. Inclusion on the list does not imply endorsement by the Fanconi Anemia Research Fund, Inc. Families are encouraged to ask clinicians about their experience treating patients with Fanconi anemia.

AUSTRALIA

The Children's Hospital at Westmead

Sydney, NSW 2145, Australia

www.chw.edu.au/prof/services/fanconi_anaemia

- **Dr. Julie Curtin, MD, PhD**, Head Haematology Department
Telephone: 612-9845-0000 Fax 612-9845-3332
Email: julie.curtin@health.nsw.gov.au
- **Professor Peter Shaw**, Head BMT Service
Telephone: 612-9845-0000 Fax: 612-9845-2171
Email: peter.shaw@health.nsw.gov.au
- **Ida O'Brien**, Clinical Nurse Consultant BMT Follow Up
Telephone: 612-9845-0000 Fax: 612-9845-2171
Email: Ida.obien@health.nsw.gov.au
- **A/Prof. Meredith Wilson**, Clinical Geneticist
Telephone: 612-9845-3273 Fax: 612-9845-3204
Email: meredithwilson@health.nsw.gov.au
- **A/Prof. David Little**, Director Orthopaedic Research
Telephone: 612-9845-3352 Fax: 612-9845-3180
Email: david.little@health.nsw.gov.au
- **Professor Chris Cowell**, Head Endocrinology Department
Telephone: 612-9845-0000 Fax: 612-9845-3170
Email: chris.cowell@health.nsw.gov.au
- **Dr. Carsten E. Palme, FRACS**, Surgeon, Otolaryngology Head & Neck Surgery
Telephone: 612-9845-6025 Fax: 612-9893-7440
Email: Carsten_Palme@specialistservices.com.au
- **Mark Schifter**, Oral Medicine and Oral Pathology
Telephone: 612-9845-8716 Fax: 612-9845-5740
Email: mark.schifter@swahs.health.nsw.gov.au

Services Offered:

This 350-bed tertiary care paediatric institution provides comprehensive care for the majority of children in NSW with all diagnoses. This includes a comprehensive team of medical professionals working in disease-specific multi-

disciplinary teams to manage complex disorders with multi-system manifestations, such as Fanconi anaemia. We offer comprehensive management of new and previously diagnosed FA patients, including:

- Diagnostic testing and screening of siblings;
- Genetic counseling; coordination of gene testing via overseas facilities to identify causative mutations in affected individuals; discussion of family planning options for future pregnancies, including prenatal diagnosis or pre-implantation genetic diagnosis for families with identified mutations;
- Orthopaedic management, including specialist hand, upper limb and congenital hand clinic;
- Haematological management;
- Blood and marrow transplantation: comprehensive program of related, unrelated and haploidentical marrow, PBSC and cord blood transplant;
- Co-ordinated long-term follow-up and ongoing surveillance, with particular reference to BMT follow-up, oral, head and neck cancer screening and endocrine follow-up (Chris Cowell);
- Translational research on non-radiotherapy-based BMT conditioning including a well-established drug pharmacokinetic laboratory for real time tailoring of drug doses in individual patients.

Princess Margaret Hospital for Children

Roberts Road Subiaco

Perth, Western Australia 6008, Australia

- **Catherine Cole, MBBS, FRACP, FRCPA**, Clinical Director of Bone Marrow Transplantation Program
- **Tina Carter, FRACP, PhD**, Bone Marrow Transplant Physician
Telephone: 08-9340-8222
Email: Tina.Carter@health.wa.gov.au
- **Shanti Ramachandran, MBBS, FRACP, MPaeds**, Paediatric Oncologist-Clinical Haematologist
Telephone: 08-9340-8222
Email: Shanti.Ramachandran@health.wa.gov.au

Sydney Children's Hospital

Blood & Marrow Transplant Program
Kids Cancer Centre
Sydney Children's Hospital
High St, Randwick
Sydney, NSW, 2031, Australia

- **A/Prof Tracey O'Brien**, Director, Blood and Marrow Transplant Program, Deputy Director and Head of Clinical Service, Kids Cancer Centre
Telephone: 02-93821733 Fax: 02-93821789
Email: t.obrien@unsw.edu.au
- **Dr. Susan Russell**, Head Haematology, Kids Cancer Centre
Telephone: 02-93821733 Fax: 02-93821789
Email: Susan.Russell@sesiahs.health.nsw.gov.au
- **A/Prof. Richard Cohn**, Head, Oncology, Kids Cancer Centre
Telephone: 02-93821731 Fax: 02-93821789
Email: Richard.Cohn@sesiahs.health.nsw.gov.au
- **Anne Senner**, BMT Coordinator, Blood and Marrow Transplant Program, Kids Cancer Centre
Telephone: 02-93821733 Fax: 02-93821789
Email: Anne.Senner@sesiahs.health.nsw.gov.au

Lucy Maurice, BMT Clinical Nurse Consultant, Blood and Marrow Transplant Program, Centre for Children's Cancer and Blood Disorders
Telephone: 02-93821655
Email: Lucy.Maurice@sesiahs.health.nsw.gov.au

Services Offered:

Sydney Children's Hospital is a large tertiary paediatric hospital with comprehensive multi-disciplinary services. We offer evaluation of new and previously diagnosed patients with specialists in haematology, bone marrow transplantation, genetics, endocrinology, nephrology, surgery and adolescent medicine. We offer on-site DEB testing, facilitation of DNA mutation testing, family genetic screening and referral for pre-implantation genetic diagnosis. In addition, comprehensive social work, psychology, family support networks are available. The transplant program is one of the oldest and largest national programs with over 900 transplants performed since establishment of the in 1975. The centre is a national leader in unrelated marrow and cord transplants with particular interest in transplant for non-malignant diseases. The program offers a well established multi-disciplinary comprehensive pre- and post-transplant service for patients with FA. The centre director has specialized knowledge in FA having trained in a leading U.S. FA transplant centre, and the centre collaborates with

the International Fanconi Anemia Registry (Dr. Arleen Auerbach).

AUSTRIA

St. Anna Children's Hospital

Department of Hematology and Oncology
Kinderspitalgasse 6
Vienna A - 1090, Austria

- **Prof. Wolfgang Holter**, Medical Director
Email: aerztliche.direktion@stanna.at

Services Offered:

- Stem cell transplants for FA patients.

BRAZIL

Hospital de Clinicas, University of Parana

Serviço de Transplante de Medula Óssea - 15º andar
Rua General Carneiro, 181 - 15º andar
80060-900-Curitiba-PR, Brazil

- **Carmem Bonfim, MD**, Director of Pediatric Stem Cell Transplant Program, Stem Cell Transplant in Genetic Diseases, Coordinator, Fanconi Anemia Outpatient Clinic
Telephone: 55-41-3360-1000 Fax 55-41-3360-1015
Email: carmembonfim@gmail.com
- **Ricardo Pasquini, MD, PhD**, Senior Professor
Federal University of Parana
Telephone: 55-41-3360-1000 Fax: 55-41-3360-1015
Email: pasquini@hc.ufpr.br

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;
- Haploidentical transplants;
- Mutation analysis (*FANCA*, *C* and *G*) and *FANCD2* by western blot.

CANADA

Hospital for Sick Children

University of Toronto

555 University Avenue
Toronto, Ontario, Canada M5G 1X8

- **Yigal Dror, MD, FRCP(C)**, Professor of Paediatrics; Director, Marrow Failure and Myelodysplasia Program; Head, Hematology Section; Division of Hematology/Oncology; Scientist, Program in Genetics and Genome Biology, Research Institute
Telephone: 416-813-5668 Fax: 416-813-5327
Email: yigal.dror@sickkids.ca
- **Adam Gassas, MBChB, MSc, MRCP, DCH, MRCPCH**, Associate Professor of Pediatrics; Section of Blood & Marrow Transplant, Division of Hematology/Oncology
Telephone: 416-813-8227 Fax: 416-813-5327
Email: adam.gassas@sickkids.ca

Princess Margaret Hospital

Ontario Cancer Institute
University of Toronto
610 University Avenue
Toronto, Ontario, M5G 2M9, Canada

- **Jeffrey H. Lipton, FRCPC, MD, PhD**, Leukemia and Allogeneic Stem Cell Transplant Programs, PMH Professor of Medicine, University of Toronto
Telephone: 416-946-2266 Fax: 416-946-6585
Email: Jeff.Lipton@uhn.on.ca

University of Manitoba

ON2021a-675 McDermot Avenue
Winnipeg, Manitoba, Canada R3E 0V9

- **John J. Doyle, MD, FRCPC, FAAP**, Associate Professor, Dept. of Pediatrics and Child Health, University of Manitoba; Head, Pediatric Hematology/Oncology/BMT
Telephone: 204-787-4135 Fax: 204-786-0195
Email: john.doyle@cancercare.mb.ca

CHINA

Tianjin First Central Hospital

No. 24, Fukang Road, Nankai
District, Tianjin, China

Mingfeng Zhao, MD, PhD

Capital Institute of Pediatrics

No. 2 Yabao Road
Chaoyang District
100020 Beijing, China

Xiaodong Shi, MD, PhD

Telephone: 13911601076

COLOMBIA

Clínica Las Americas

Diagonal 75 B #2 A80 Ofic. 520
Medellin
Antioquia, Colombia

- **Dr. Amado Karduss**, Adult Hematology/Stem Cell Transplantation
Telephone: 57 4 3409314
Email: amaka@une.net.co
- **Dr. Yadira Coll**, Pediatric Hematology
Telephone: 57 4 3409314
Email: yacoll@une.net.co

Services Offered:

- Evaluation;
- Medical treatment;
- FA stem cell transplantation.

Dr. Adriana Linares

Hospital de La Misericordia
Av Caracas 1-67
Pediatric Hematology/Oncology Unit
Telephone: 571-381-1970 extension 1309
Email: alinaresb@gmail.com

- **Universidad Nacional de Colombia**
Departamento de Pediatría
Unidad de Oncohematologie PEDIATRICA
Av Caracas 1-67 4 piso
Bogotá, Colombia

Services Offered:

- Pediatric hematology oncology, clinical evaluation, medical treatment, and therapy.

Centro Médico Imbanaco de Cali

Bone Marrow Transplant Unit
Carrera 38 A No. 5A 100
Cali, Valle, Colombia

- **Oscar W. Ramirez, MD, MPhil**
Telephone: 572-682-1000 or 572-518-6000
Email: *oscar.ramirez@imbanaco.com.co* or *oramirez1167@gmail.com*

Services Offered:

- Pediatric hematology oncology;
- Hematopathology;
- Clinical evaluation;
- Medical therapy;
- Stem cell transplantation related and unrelated.

CZECH REPUBLIC**University Hospital Motol**

Department of Paediatric Haematology and Oncology V
Uvalu, 84
150 06 Prague, Czech Republic

- **Prof. Petr Sedlacek**
Telephone: 420 2 2443 6552 Fax: 420 2 2443 6519
Email: *petr.sedlacek@lfmotol.cuni.cz*

Services Offered:

- Stem cell transplants for FA patients.

DENMARK**University Hospital of Aarhus at Skejby**

Brendstrupgaardsvej 100
DK-8200 Aarhus N, Denmark

- **Niels Clausen, MD**, Department of Pediatrics
Telephone: 45-7845-1673
Email: *niecla@rm.dk*

Services Offered:

- Diagnosis, treatment and follow-up for children with Fanconi anemia.

FRANCE**Hôpital Robert Debré**

48, boulevard Sérurier
75935 Paris cedex 19, France

- **Thierry Leblanc, MD**, Department of Pediatric Hematology
Secretary: 33-1-7128-2535 Fax: 33-1-4003-4740
Email: *thierry.leblanc@rdb.aphp.fr*
- **Jean-Hugues Dalle, MD, PhD**, Department of Pediatric Hematology
Telephone direct: 33-1-4003-1241
Secretary: 33-1-4003-5388 Fax: 33-1-4003-4740
Email: *jean-hugues.dalle@rdb.aphp.fr*

Services Offered (primarily for children):

- Bone marrow, peripheral stem cell and cord blood transplants;
- FA diagnosis: breakage tests, cell cycle, FANCD2 analysis, mosaicism on skin biopsy, retroviral group complementation, bone marrow karyotype (Dr. Jean Soulier);
- Gene sequencing on grouped patients, in collaboration with Institut Curie (Pr. Dominique Stoppa-Lyonnet);
- Head and neck cancer evaluation and treatment (Dr. Francis Pascal and Dr. Arnaud Rigolet);
- Cell therapy laboratory (Dr. Jerome Larghero);
- Gene therapy laboratory (Dr. Philippe Leboulch).

Hôpital Saint-Louis

1, Avenue Claude Vellefaux
75010 Paris, France

- **Gérard Socié, MD, PhD**, Department of Hematology Transplantation
Telephone direct: 33-1-4249-9824
Secretary: 33-1-4249-9639 Fax: 33-1-4249-9636
Email: *gerard.socie@sls.aphp.fr*
- **Jean Soulier, MD, PhD**, Genome Rearrangements and Cancer Group, Hematology Laboratory
Telephone: 33-1-4249-9891 Fax: 33-1-4249-4027
Email: *jean.soulier@sls.aphp.fr*
- **Arnaud Rigolet, MD**, Department of Oral and Maxillofacial Surgery
Telephone: 33-1-42-49-41-92 Fax: 33-1-1-49-49-91-92
Email: *arnaud.rigolet@sls.aphp.fr*

Services Offered (primarily for teens and adults):

- Bone marrow, peripheral stem cell and cord blood transplants;
- FA diagnosis: breakage tests, cell cycle, FANCD2

- analysis, mosaicism on skin biopsy, retroviral group complementation, bone marrow karyotype (Dr. Jean Soulier);
- Gene sequencing on grouped patients, in collaboration with Institut Curie (Pr. Dominique Stoppa-Lyonnet);
 - Head and neck cancer evaluation and treatment (Dr. Francis Pascal and Dr. Arnaud Rigolet);
 - Cell therapy laboratory (Dr. Jerome Larghero);
 - Gene therapy laboratory (Dr. Philippe Leboulch).

GERMANY

Charité, Campus Virchow Klinikum University Medicine, Berlin

Children's Hospital
Augustenburger Platz 1
13353, Berlin, Germany

- **Jörn-Sven Köhl, MD**
Telephone: 49-30-450 566432
Fax: 049-030-450-566919
Email: jskuehl@charite.de

Services Offered:

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

Hannover Medical School

Department of Pediatric Hematology and Oncology
Carl-Neuberg-Str. 1
30625 Hannover, Germany

- Christian Peter Kratz, MD, Director**
Telephone: 49 (0)511 532 6711 Fax: 49 (0)511 532 9120
Email: kratz.christian@mh-hannover.de

Services Offered:

- Natural history study for patients with Fanconi anemia;
- Guidance in clinical decision making.

Heinrich Heine University Medical Center

Children's Hospital
Moorenstrasse 5
40225 Düsseldorf, Germany

- **Eunike Velleuer, MD**, Department of Pediatric Hematology, Oncology, and Clinical Immunology
Telephone: 49-211-811-7687 Fax: 49-211-811-7634
Email: velleuer@uni-duesseldorf.de

- **Arndt Borkhardt, MD**, Director, Department of Pediatric Hematology, Oncology, and Clinical Immunology
Telephone: 49-211-811-7680 Fax: 49-211-811-6206
Email: lesch@med.uni-duesseldorf.de

Services Offered:

- Clinical care of FA patients;
- Cooperation with Ralf Dietrich, German FA family support group (Deutsche FA-Hilfe e.V.) on "Reducing the burden of squamous cell carcinoma in Fanconi anemia;"
- Cooperation with Birgit Henrich, University of Duesseldorf, Microbiology; Microbiom of the oral cavity in FA;
- Cooperation with Ruud Brakenhoff, University of Amsterdam, on "Progression of FA-SCC and Development of a Non-Invasive Screening Method for Precursor Lesions;"
- Cooperation with Prof. Stefan Biesterfeld, University of Duesseldorf, Cytopathology; non-invasive testing of oral lesions (Cytology, DNA-Cytometry);
- Cooperation with Prof. Torsten Remmerbach, University of Leipzig, development of alternative non-invasive methods for the detection of oral cancers.

University of Tuebingen

Children's University Hospital
Department of Hematology/Oncology and General Pediatrics
Hoppe-Seyler-Strasse 1
72076 Tuebingen, Germany

- **Prof. Rupert Handgretinger, MD**, Chairman
Telephone : 49-7071-298-4744 Fax: 49-7071-294713
Email: Rupert.Handgretinger@med.uni-tuebingen.de

Services Offered:

- Evaluation of new patients;
- Hematopoietic stem cell transplantation from HLA-matched siblings, matched unrelated donors and haploidentical family donors;
- Co-transplantation of bone marrow with mesenchymal stem cells to enhance engraftment and to prevent graft-versus-host-disease (GvHD);
- Extracorporeal photopheresis in small children for the prevention and treatment of GvHD;
- Adoptive transfer of virus-specific T-cells to prevent viral infections post-transplant;
- Long-term follow-up studies (growth, endocrinology, quality of life).

INDIA

Apollo Specialty Hospital

320 Padma Complex
Anna Salai
Chennai 600 035, India

- **Sheila Mohan, MBBS, DCH, PhD**
Telephone: 91-44-2491-2943
Mobile: 91-92-8211-4661
- **Revathi Raj, MBBS, DCH, MRCP (Path)**
Telephone: 00- 91-44-33151294

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 ;
- Genetic counseling;
- Stem cell transplantation;
- Maintenance of national database: Registry for Fanconi Anemia in India (REFAIN).

National Institute of Immunohaematology (ICMR)

13th floor, New Multistoried Building
K.E.M. Hospital Campus, Parel
Mumbai 400012, India

- **Dr. V. Babu Rao**
Telephone: 91-22-24138518 or 24138519
Fax: 91-22-24138521
Email: vbaburao@hotmail.com

Services Offered:

- Chromosomal breakage study using MMC and DEB test;
- Cytogenetic study from bone marrow and FISH analysis;
- FANCD2 western blot;
- Molecular testing of complementation groups *FANCA*, *FANCC*, *FANCG*;
- Prenatal diagnosis of FA after mutation detection in proband.

IRAN

Tehran University of Medical Sciences

Shariati Hospital
Hematology-Oncology and SCT Research Center
Kargar Avenue
14114, Tehran, Iran
<http://horecst.tums.ac.ir>

- **Ardeshir Ghavamzadeh, MD**, Professor of Medicine
Adult Hematologist Oncologist
President of Hematology-Oncology and SCT Research Center
Telephone: 98-21-84902635 Fax: 98-21-88004140
Email: ghavamza@sina.tums.ac.ir
- **Amir Ali Hamidieh, MD**, Associate Professor of Medicine
Pediatric Hematologist Oncologist
Director of Fanconi Anemia Transplant Program
Telephone: 98-21-84902645 Fax: 98-21-88004140
Email: aahamidieh@sina.tums.ac.ir

Services Offered:

- Pediatric and adult FA patients can be seen by specialists in pediatric and adult medicine;
- Pediatrics stem cell transplantation, 11-bed ward;
- Adults stem cell transplantation, 25-bed ward;
- Stem cell laboratory;
- Cord blood bank;
- Cytogenetic laboratory;
- Complete evaluation, treatment and follow-up of new and previously diagnosed FA patients;
- Diagnostic testing (DEB and MMC test) and screenings of siblings;
- Genetic counseling;
- Matching sibling, unrelated, haploidentical and cord blood stem cell transplants;
- HLA registry (Iranian Stem Cell Donor Program, ISCDP).

ISRAEL

International Center for Cell Therapy & Cancer Immunotherapy (CTCI)

Weizman Center, Top Ichilov
14 Weizman Street
Tel Aviv 64239, Israel

- **Shimon Slavin, MD**, Professor of Medicine, Scientific & Medical Director of CTCI
Telephone: 972-77-777-9255 Fax: 972-77-777-9247
Email: slavin@CTCIcenter.com

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. Stem cell transplantation can also be considered from half-matched (haploidentically mismatched) family member while avoiding the risk of graft-versus-host disease.

Schneider Children's Medical Center of Israel

14 Kaplan Street
PO Box 559
Petach Tikvah 49202
Israel

- **Hannah Tamary, MD**, Director of Pediatric Hematology, Professor of Pediatrics
Telephone: 972-3-9253669 Fax: 972-3-9253042
Email: htamary@post.tau.ac.il
- **Joanne Yakobovich, MD**, Senior Pediatric Hematologist
- **Jerry Stein, MD**, Director of Pediatric Bone Marrow Transplantation

Services Offered:

- Mutation detection for all Fanconi anemia genes, genetic counseling, and prenatal diagnosis;
- Comprehensive clinical care.

ITALY

G. Gaslini Children's Institute

Largo G.Gaslini 5
16147 Genova, Italy

- **Carlo Dufour, MD**, Head, Hematology Unit
Telephone: 39-010-5636-694 Fax: 39-010-5636-714
Email: carlodufour@ospedale-gaslini.ge.it
- **Johanna Svahn, MD**, Hematology Unit
Telephone: 39-010-5636-694 Fax: 39-010-5636-714
Email: johanna.svahn@ospedale-gaslini.ge.it
- **Enrico Cappelli, PhD**, Hematology Laboratory
Telephone: 39-010-5636-693 Fax: 39-010-5636-714
Email: enricocappelli@ospedale-gaslini.ge.it

Services Offered:

- Primary diagnosis with DEB test, cell cycle analysis and MMC sensitivity, FANCD2 Ubiquitination (WB) on peripheral blood cells and fibroblasts;
- FA complementation group analysis by retroviral vectors on peripheral blood T-cells, bone marrow cells, fibroblasts and B-cell lines for groups A, C, G, D2;

- Collaboration with Dr. Helmut Hannenberg, Riley Hospital for Children, Indianapolis and Dr. Juan Bueren, CIEMAT, Madrid, Spain for remaining FA complementation group analyses;
- Collaboration with Prof. Juan Carlos Belmonte Ispizua, University LaJolla, San Diego, CA for IPS;
- Collaboration with Dr. Anna Savoia, IRCSS Burlo Garofolo, University of Trieste, Italy, for molecular diagnosis of FA;
- Collaboration with Dr Paolo Degan, Mutagenesis Lab, Scientific Tumour Institute, Genova, Italy, for redox activity;
- Haematopoiesis laboratory (cell cultures, cytokine hypersensitivity tests);
- Cytogenetic laboratory;
- Collaboration with European Centres of the EBMT (European Society for Blood and Marrow Transplantation) for data collection and retrospective studies;
- Close scientific collaboration with Dr. Grover Bagby (OHSU, Portland, OR, USA) on pathogenetic mechanism of marrow failure in FA;
- 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 (Dr. Edoardo Lanino, HSC Unit, G. Gaslini Children's Hospital), head and neck cancer evaluation, endocrinology, orthopedic and nephrology clinic, psychological support;
- Ante-natal diagnosis available for patients in whom mutations are known;
- Experimental protocol for treatment of FA patients with bone marrow failure and no HSC donor available, using Etanercept (anti TNF-alpha); Eudract Code (Italian Ministry of Health Registration Code) 2005-003096-20. This study has now been published.

Second University of Naples

Clinica Pediatrica 1, Department of Paediatrics
Via De Grecchio 4
80138 Napoli, Italy

- **Bruno Nobili, MD**
Telephone and Fax: 39-0815-665432
Email: bruno.nobili@unina2.it
- **Silverio Perrotta, MD**
Telephone and Fax: 39-0815-665421
Email: silverio.perrotta@unina2.it
- **Sofia Maria Rosaria Matarese, MD**
Telephone and Fax: 39-0815-665444
Email: SofiaMariaRosaria.MATARESE@unina2.it

- **Francesca Rossi, MD**
Telephone and Fax: 39-0815-665342
Email: francesca.rossi@unina2.it

Services Offered:

- Complete clinical evaluation, treatment and follow-up of new and previously diagnosed FA patients;
- Diagnostic testing (chromosomal breakage with Mitomycin C, DEB test, cell cycle analysis, karyotype, bone marrow examination, cytogenetic analysis, FANCD2 ubiquitination (WB) on peripheral blood cells and fibroblasts);
- Transfusion of irradiated blood and platelet products if needed;
- Information, education and assistance to families of patients with FA and referring physicians;
- Coordination of consultations with other subspecialties for management of FA-related disorders.

University of Pavia

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

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

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.

JAPAN

Tokai University

Department of Cell Transplantation and Regenerative
Medicine
Shimokasuya 143
Isheara, Kanagawa, 259-1193, Japan

- **Hiromasa Yabe, MD, Stem Cell Transplantation**
Telephone: 81-463-93-1121 Fax: 81-463-95-9567
Email: yabeh@is.icc.u-tokai.ac.jp

- **Miharu Yabe, MD, Stem Cell Transplantation**
Telephone: 81-463-93-1121 Fax: 81-463-95-9567
Email: miharu@is.icc.u-tokai.ac.jp

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.
- Direct collaboration with Prof. Minoru Takata, the Kyoto University, for mutation analysis.

KENYA

Kenvatta National Hospital

Haematology & Blood Transfusion Unit
Department of Human Pathology
University of Nairobi, School of Medicine
Nairobi, Kenya

- **Dr. Grace W. Kitonyi MBChB, FRCPath**
Haematologist/Haematopathologist
Telephone: 254 20 2721815
Mobile phone: 254722385336
Email: gwkitonyi@yahoo.com
Cc: gkitonyi@uonbi.ac.ke
- **Dr. Jessie N. Githanga MBChB, MMED Path**
Haematologist/Haematopathologist
Telephone: 254 20 2721815
Mobile phone: 254721245721
Email: jgithanga3@gmail.com
- **Dr. J. A. Rajab, MBChB, MMED, Path, MPH**
Haematopathologist/Public Health Specialist
Telephone: 254 20 2721815
Mobile phone: 254722707421
Email: amjamilla@yahoo.com

Services Offered:

- Diagnosis and management of all patients with FA including counseling of families;
- Coordination of consultations with other sub-specialities for management of FA related disorders.

LEBANON

St. George Hospital University Medical Center

Dept of Pediatrics and Pediatric Hematology/Oncology
Office 121
Ashrafieh, Beirut, Lebanon

- **Dr. Roula Farah, MD, FAAP**
Associate Professor of Pediatrics and Pediatric Hematology/Oncology
President, CHANCE Association
Cell phone: 961-3-681-309
Email: roula.fs@dm.net.lb

Services Offered:

- Complete clinical evaluation, treatment and follow-up of new and previously diagnosed FA patients;
- Prenatal diagnosis;
- Diagnostic testing (chromosomal breakage with mitomycin C, karyotype, bone marrow examination, etc.);
- Genetic testing for *FANCA* gene mutations;
- Transfusion of irradiated blood and platelet products if needed;
- Support with financial coverage for diagnostic studies if patient cannot afford it;
- Information, education and assistance to families of patients with FA and referring physicians;
- Genetic counseling;
- Maintenance of the National Fanconi Anemia Registry.

MEXICO

Centro Medico Nacional “La Raza”

Instituto Mexicano Del Seguro Social
Division de Trasplantes
Seris y Zaachila
Col. La Raza, 02990
Mexico DF, Mexico

- **Dr. Inès Montero Ponce**
- **Dr. Ana Lilia Hernández Moreno**

Prof. Dr. Clara Gorodezky

Head of Department of Immunology & Immunogenetics, InDRE
Secretary of Health
President of the Board of Fundacion Comparte Vida, A.C.
Francisco P. Miranda #77 - Laboratory Building, 2nd Floor,
Mexico DF, 01480, Mexico
Phone: +5255 6383 0041 & +5255 5062 1600 ext. 59314 & 59397

Email: clarag@unam.mx or
cgorodea@fundacioncompartevida.org.mx
www.fundacioncompartevida.org.mx

Services Offered:

- HLA Lab, ASHI accredited laboratory;
- Access to DONORMO (Mexican Unrelated Bone Marrow Donors Registry) as well as the BACECU (Mexican Unrelated Cord Blood Bank);
- HLA typing and all related studies for solid organ transplantation;
- Search DONORMO, BACECU, and BMDW, etc. and perform all the workup;
- Referrals to physicians for Fanconi anemia patients;
- We work with all the hospitals in Mexico and with many transplant centers abroad;
- We belong to BMDW and are a Coop Registry of NMDP.

Instituto Nacional de Pediatría

Instituto de Investigaciones Biomédicas
UNAM
Insurgentes Sur 3700-C
Col. Insurgentes Cuicuilco
4530 Mexico City, DJ, Mexico

Sara Frias, PhD

Telephone: 52-55-1084-5533
Email: sarafrias@biomedicas.unam.mx

Marisol González, MD

Telephone: 55-320-51010
Email: mary_solg@hotmail.com

Services Offered:

- Clinical and laboratory diagnosis of Fanconi anemia;
- Hematology in bone marrow and blood;
- Analysis of spontaneous chromosomal aberrations and chromosomal breakage test induced with DEB and MMC in lymphocytes, fibroblasts and amniocytes;
- Analysis of G2/M cell cycle halt by flow cytometry in lymphocytes and fibroblasts.

Unidad de Citogenética

Servicio de Hemato-Oncología Pediátrica, Unidad de Citogenética, Nuevo Hospital Civil
Dr. Juan I Menchaca
Laboratorio de Citogenética Genotoxicidad y Biomonitorio
CUCS, Universidad de Guadalajara

Juan I. Menchaca, MD

Nuevo Hospital Civic
División de Pediatría,
Servicio de Hematología Pediátrica
Unidad de Citogenética,
Salvador de Quevedo y Aubieta #750, CP-44340

Guadalajara, Jalisco, México
Telephone: 52-33-3883440 ext. 52866

Enrique Corona Rivera, MD

Universidad de Guadalajara
Centro Universitario de Ciencias de la Salud
Instituto de Genética Humana
Laboratorio de Citogenética genotoxicidad y Biomonitorio
Edificio P 2do Nivel
Sierra Mojara #950, S.L., CP-44340
Guadalajara, Jalisco, México
Telephone: 52-33-10585200 ext. 33647

Lucina Bobadilla-Morales, MD, PhD

Main contact: Diagnosis, research, and patient management
Email: *lucinabo@cucs.udg.mx*

Alfredo Corona-Rivera, PhD

Research and diagnostic
Email: *alcoronar@gmail.com*

Fernando Sánchez-Zubieta, MD

Pediatric Hemato-oncology services
Email: *dfernados59@hotmail.com*

Services Offered:

- Clinical and laboratory diagnosis of Fanconi anemia;
- Hematology in bone marrow and blood;
- Analysis of spontaneous chromosomal aberrations and chromosomal breakage test induced with MMC in lymphocytes;
- MLPA molecular biology studies in Fanconi anemia patient samples;
- Genetic counseling for patients, parents, and relatives;
- Diagnostic evaluation for Fanconi anemia and other bone marrow failure syndromes;
- Consultation and education for new and previously diagnosed patients and their families;
- Comprehensive clinical management of pediatric Fanconi anemia patients;
- Hematopoietic stem cell progenitors transplantation;
- Performance of HLA- A*, B*, C*, DQ*, DR*, high resolution typing;
- Cytogenetics, FISH, molecular biology, cryopreservation and flow cytometry facilities to alternative studies.

NETHERLANDS

Leiden University Medical Center

Department of Pediatrics/BMT unit
PO Box 9600
Leiden, 2300 RC, Netherlands

- **Arjan C Lankester, MD, PhD**, Program Director Stem Cell Transplantation, Department of Pediatrics/BMT unit
Telephone: 31-71-5264131 Fax: 31-71-5248198
Email: *A.Lankester@lumc.nl*

UMC Utrecht

Kinderarts - Hematoloog
Wilhelmina Kinderziekenhuis
Postbus 85090
3508 AB Utrecht
Huispostnr KC 03.063.0, Netherlands

- **Marc Bierings, MD, PhD**, Represents the Dutch Fanconi Anemia Guideline on behalf of the Dutch Childhood Oncology Group (DCOG), and Chairs the Guideline Committee for FA in Netherlands.
Telephone: 31 (0)88 7554344 Fax: 31 (0)88 7555350
Email: *m.bierings@umcutrecht.nl*

VU University Medical Center Amsterdam

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

Diagnosis:

Telephone: 31-20-444-8346 Fax: 31-20-444-8293

- **Hans Gille, PhD**
Email: *jgp.gille@vumc.nl*
- **Resie Vervenne, PhD**
Email: *r.vervenne@vumc.nl*
- **Pino Poddighe, PhD**
Contact for chromosomal breakage testing.
Email: *p.poddighe@vumc.nl*

Research:

Telephone: 31-20-444-8270 Fax: 31-20-444-8285

- **Josephine C. Dorsman, PhD**
Email: *jc.dorsman@vumc.nl*

Services Offered:

Diagnosis:

- Diagnosis by chromosomal breakage analysis, including mosaicism;
- Molecular diagnosis by mutation screening of all known FA genes;
- Prenatal DNA testing (if mutations are known).

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 FA pathway defects and their role in sporadic cancer.

Collaboration in Netherlands:

- **M. Bierings, MD, PhD**, Stem Cell Transplantation, Clinical Coordinator for FA families in The Netherlands, Utrecht University Medical Center
- **E.T. Korthoff, MD, PhD**, Stem Cell Transplantation, Leiden University Medical Center
- **R.H. Brakenhoff, PhD**, Dept. Otolaryngology/Head-Neck Surgery, VU University Medical Center Amsterdam.

NEW ZEALAND

Christchurch Hospital

Children's Haematology Oncology Centre

Private Bag 4710
Christchurch 8140, New Zealand

- **Dr. Siobhan Cross**, Paediatric Haematologist
Telephone: 64 3 364 0640, ext. 87500
Email: Siobhan.Cross@cdhb.govt.nz

Services Offered:

- FA diagnosis (samples are sent to Australia), follow up/surveillance for FA patients under age 18.

Starship Children's Hospital

Grafton
Auckland 1023, New Zealand

- **Dr. Nyree Cole**
Telephone: 64 9 307 4919
Email: Nyreec@adhb.govt.nz

REPUBLIC OF KOREA

Seoul St. Mary's Hospital

The Catholic University of Korea

Blood & Marrow Transplantation Center
#222 Banpo-daero, Seocho-gu
Seoul, 137-701, Republic of Korea
Telephone: 82-10-9407-2220 Fax: 82-2-537-4544

- **Nack-Gyun Chung, MD**, Professor, Department of Pediatrics, Hematology/Oncology/BMT
Telephone: 82-2-2258-6188 Fax: 82-2-537-4544
Email: cngped@catholic.ac.kr

- **Myungshin Kim, MD, PhD**, Professor, Department of Laboratory Medicine
Telephone: 82-2-2258-1645 Fax: 82-2-2258-1719
Email: microkim@catholic.ac.kr

Services Offered:

- Evaluation of new and previously diagnosed patients;
- Breakage test, sequence analysis;
- Diagnosis and management of all patients with FA including counseling of families;
- Expertise in stem cell transplantation from matched sibling donor or alternative donor.

SAUDI ARABIA

King Faisal Specialist Hospital & Research Center

Riyadh, Saudi Arabia

- **M. Ayas, MD**, Head, Section of Pediatric Stem Cell Transplantation Department of Pediatric Hematology Oncology
Telephone: 966-1-442-3934 Fax: 966-1-442-4662
Email: mouhab@kfshrc.edu.sa

Services Offered:

- Diagnosis: cytogenetics testing including DEB chromosomal breakage testing and conventional cytogenetics of bone marrow and peripheral blood;
- Comprehensive care for FA patients, including blood and platelet transfusion;
- Allogeneic stem cell transplant: matched related bone marrow and matched/partially matched unrelated cord blood transplants;
- Haploidentical stem cell transplantation for FA patients with no matched related donors.

SINGAPORE

National University Hospital, Singapore

Department of Paediatrics
1E Kent Ridge Road
NUHS Tower Block Level 12
Singapore 119228
Republic of Singapore
<http://www.nuh.com.sg/ucmi/>

- **Poh-Lin Tan, MBBS, MMED** (Paediatrics)
Email: poh_lin_tan@nuhs.edu.sg

International Patient Liaison Centre:

Telephone (24hr Helpline): (65) 67792777

Email: iplc@nhg.com.sg

Services Offered:

- FACT-accredited paediatric blood and marrow transplant;
- Paediatric sub-specialties: adolescent medicine, allergy, cardiology, developmental paediatrics, endocrinology, gastroenterology, genetics, haematology, infectious diseases, intensive care, nephrology, neurology, oncology, otolaryngology, psychiatry, pulmonology, rheumatology, surgery.

SOUTH AFRICA

Chris-Hani Baragwanath Hospital

Division of Haematology and Oncology

Johannesburg, South Africa

- **Dr. Linda Wainwright**

Clinic Telephone: 0800333555

Office Telephone: 0119339594

Cell: 0828219985

Email: rosalind.wainwright@wits.ac.za

Johannesburg Hospital

Division of Haematology and Oncology

Dept. of Pediatrics

Private Bag X39

Johannesburg 2000, South Africa

- **Prof. Janet Poole**

Telephone: 27(0)114883294/0 Fax: 27(0)114884152

Cell: 27(0)824695762

Email: Janet.Poole@wits.ac.za

NHLS, Division of Human Genetics

University of the Witwatersrand

PO Box 1038

Johannesburg 2000, South Africa

- **Dr. Amanda Krause**

Telephone: 27-11-489-9219/9224

Mobile: 27-82-809-5797

Fax: 27-11-489-9226

Email: amanda.krause@nhls.ac.za

Services Offered (Chris-Hani Baragwanath, Johannesburg Hospital, and NHLS):

- Genetic counseling service for families with FA;
- Diagnostic, carrier, preclinical and prenatal testing.

Unitas Hospital

PO Box 15225

Lyttleton, 0140, Pretoria

Gauteng, South Africa

- **Dr. Charmaine Jacobs, MD**, Pediatric

Oncology/Haematology Unit

Telephone: 27-12-6778076

Mobile Phone: 27724195346 Fax: 27-12-6647848

Email: cjaconko@mweb.co.za

Services Offered:

- Diagnosis and management of FA patients and their families of all age groups;
- Coaching and counseling families towards a healthy and optimum lifestyle with FA;
- Coordination of consultations with other sub-specialties for management of FA-related disorders;
- Motivational speaking presentation on living a life with Fanconi with meaning and purpose;
- Workshops on: Lifestyle management for Fanconi families - empowering the patients to live their best life.

University of the Free State

PO Box 339(G69)

Bloemfontein 9300, South Africa

- **Prof. D.K. Stones**, Paediatric Oncology and Haematology, Department of Paediatrics and Child Health

Telephone: 27-51-4052820

Mobile: 083-444-7233

Fax: 27-51-4443230

Email: StonesDK@UFS.ac.za

Services Offered (Universitas Hospital, Bloemfontein and NHLS):

- Genetic counseling service for families with FA;
- Diagnostic, carrier, preclinical and prenatal testing in conjunction with genetics department and NHLS.

SPAIN

Hospital General Universitario Gregorio Marañón

Pediatric Hematology

C/Doctor Esquerdo, 46

28007 Madrid, Spain

- **Dr. Cristina Belendez**

Telephone: 91-529-00-37

Email: cristina.belendez@salud.madrid.org

Hospital Universitario de la Princesa

C/Doctor Esquerdo, 45
28028 Madrid, Spain

- **Dr. Ángela Figuera**
Telephone: 91-520-24-31
Email: afiguera.hlpr@salud.madrid.org

Hospital Vall d'Hebron

Pediatric Oncology
P. de la Vall d'Hebron 119-129
Barcelona 08035, Spain

- **Dr. Cristina Díaz de Heredia**
Telephone: 93-488-30-93

University Hospital Niño Jesús

Avenida Menéndez Pelayo, 65
28009 Madrid, Spain

- **Julian Sevilla, MD**, Department of Hematology and Oncology
Telephone: 91-503-5938 Fax: 91-503-5960
Email: jsevilla.hnjs@salud.madrid.org
- **Dr. Marta Prudencio**, Department of Hematology and Oncology
Telephone: 91-503-5938 Fax: 91-503-5960

CIEMAT/CIBERER

Division of Hematopoietic Innovative Therapies (HIT)
CIEMAT/ CIBER of Rare Diseases (CIBERER)
IIS Jiménez Díaz Foundation
Madrid, Spain
www.ciemat.es

Spanish Fanconi Anemia Network: www.redfanconi.org

- **Juan Bueren, PhD**, Head of Division of Hematopoietic Innovative Therapies (HIT)/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Díaz Foundation
Telephone: 34-91-346-6518
Email: juan.bueren@ciemat.es
- **José A. Casado**, Senior Researcher of Division of Hematopoietic Innovative Therapies (HIT)/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Díaz Foundation
Telephone: 34-91-496-2525
Email: jose.casado@ciemat.es
- **Paula Río**, Researcher of Division of Hematopoietic Innovative Therapies (HIT)/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Díaz Foundation
Telephone: 34-91-346-0890
Email: paula.rio@ciemat.es

- **Susana Navarro**, Senior Researcher/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Díaz Foundation
Telephone: 34-91-346-0891
Email: s.navarro@ciemat.es
- **M^a Luz Lozano**, Technical Assistance/CIBER of Rare Diseases (CIBERER)/IIS Jiménez Díaz Foundation
Telephone: 34-91-346-6255
Email: luz.lozano@ciemat.es
- **Aurora de la Cal**, Secretary, Spanish FA Research Network
Telephone: 34-91-496-2518
Email: aurora.delacal@ciemat.es

Services Offered:

- Coordination of the FA Spanish registry;
- Coordination of the FA gene therapy clinical trial;
- FA subtyping by retrovirus mediated gene transfer;
- Analysis of the hematopoietic reserve in bone marrow samples from FA.

Universitat Autònoma de Barcelona

Chromosome Fragility Service (UAB-CFS)
Department of Genetics and Microbiology
08193 Bellaterra (Cerdanyola del Valles)
Barcelona, Spain

<http://safcro.uab.es/>

Spanish Fanconi Anemia Network: www.redfanconi.org

- **Jordi Surrallés, PhD**, Professor of Genetics (UAB-CFS Scientific Director)
Telephone: 34-935-811-830 Fax: 34-935-812-387
Email: jordi.surralles@uab.es
- **Maria Roser Pujol, PhD**, Cytogeneticist (UAB-CFS Technical Director). Accredited by Spanish Society of Human Genetics
Fax: 34-935-812-387
Email: MariaRoser.Pujol@uab.cat

Services Offered:

- DEB chromosomal breakage testing in blood and skin fibroblasts;
- Diagnosis of mosaicism;
- MMC-induced G2/M-cell phase arrest assay;
- FANCD2 monoubiquitination and foci formation assays;
- Spanish contact for prenatal diagnosis;
- Genetic counseling;
- Maintenance of a biobank of Spanish FA patients with cryopreserved transformed cell lines and DNA.

Services for genetic analysis are offered free of charge to Spanish FA patients through the Spanish Fanconi Anemia Network, which is funded by the Spanish Ministry of Health. Chromosome fragility testing in blood lymphocytes or skin

fibroblast is free of charge to Spanish patients when the results are positive. The cost of the DEB test in blood lymphocytes to non-Spanish patients or to Spanish patients with a negative result is 290 euros. The cost of the DEB test in fibroblasts to non Spanish patients and to Spanish patients with a negative result is 450 euros.

TURKEY

Hacettepe University

Inherited Bone Marrow Failure Center
Ankara, Turkey 06100

- **Sule Unal, MD**, Director, Associate Professor
Telephone: +90 312 305 1170
Email: suleunal@hacettepe.edu.tr

Services Offered:

- NGS screening is available for genetic diagnosis of FA.
- FA patients are followed by experienced physicians in their fields. Each patient is seen by the same hematologist, ENT, O&G, plastic surgeon on visits. Both adult and pediatric patients are followed and the facility is approved by the Turkish Ministry of Health.
- Hematopoietic stem cell transplantation unit has good results with FA patients.

Istanbul University

Bone Marrow Bank & Department of Medical Biology
Istanbul Medical Faculty
Çapa 34390 Istanbul, Turkey

- **Nevin Yalman, MD**, President, Fanconi Anemia Association, Professor, Pediatric Hematologist
Telephone: 9-212-414-20-00, x 31208
Fax: 9-212-635-11-68
Email: nevinyalman@gmail.com

UNITED KINGDOM

Birmingham Children's Hospital

Steelhouse Lane
Birmingham, West Midlands B4 6NH, United Kingdom

- **Phil Darbyshire, MD**, Consultant Pediatric Hematologist
Telephone: 44-121-333-9999
Email: Phil.Darbyshire@bch.nhs.uk

Bone Marrow Transplant & Late Effects Clinic

Department of Haematology
Royal Hallamshire Hospital
Glossop Road
Sheffield S10 2JF, United Kingdom

- **Professor John Snowden, BSc (Hons), MBChB, MD, FRCP, FRCPath**
Consultant Haematologist
Telephone: 0114-271-3357 Fax: 0114-271-3997
Email: John.Snowden@sth.nhs.uk
- **Dr Diana Greenfield, BSc (Hons), RN, PhD**
Consultant Nurse
Telephone: 0114-226-5592 Fax: c/o 0114-271-3754
Email: Diana.Greenfield@sth.nhs.uk

Services Offered:

- The joint Bone Marrow Transplant and Late Effects Clinic provides adult Fanconi anaemia patients access to a multi-disciplinary team providing initial general assessment, confirmation of diagnosis and subsequent coordination of screening and support. The multi-disciplinary clinic provides access to other specialists at our hospital including: endocrinology, respiratory, gynaecology, urology, gastroenterology, maxillo-facial, radiology, genetics, nutrition, psychiatry, psychology and lifestyle advice. There is close transitional working with the paediatric services run by Prof. Ajay Vora at Sheffield Children's Hospital NHS Foundation Trust.
- Patients are assessed and reviewed in Sheffield with communication to their referring clinician. Nurse-led follow-up and support is recommended on an annual basis for all those patients who are medically stable.
- Where appropriate, haematopoietic stem cell transplantation can be arranged in Sheffield. Both the Sheffield Teaching Hospitals NHS Foundation Trust and Sheffield Childrens' Hospital are JACIE-accredited centres.

Bristol Royal Hospital for Children

Oncology Day Beds
Level 6
Upper Maudlin Street
Bristol, BS2 8BJ, United Kingdom

- **Colin Steward, MA, BM, BCh, PhD, FRCPCH, FRCP**, Department of Paediatric Haematology and Bone Marrow Transplantation
Telephone: 44-117-3428044 Fax: 44-117-3420170
Email: colin.steward@uhbristol.nhs.uk

Services Offered:

- Comprehensive diagnostic and routine haematological care in specialized clinic;

- Coordination of complementation group analysis, mutation analysis and somatic mosaicism studies;
- Expertise in androgen and transplantation therapy;
- Specialists in genetics, haematology, bone marrow transplantation (from sibling, matched and mismatched unrelated, haploidentical and cord donors), endocrinology, gastroenterology and radiotherapy;
- 16-bed inpatient area and outpatient bone marrow transplantation clinic.

St. Mary's Hospital

Praed Street, Paddington
London W2 1NY, United Kingdom

- **Josu de la Fuente, PhD FRCP MRCPath FRCPa**
Consultant Paediatric Hematologist
Telephone: 44-20-788-66164
Email: *Josu.delaFuente@imperial.nhs.uk*

Yorkshire Paediatric Haematology Unit

Leeds General Infirmary
Fanconi Anaemia – Multi-disciplinary Clinic
Great George Street
Leeds LS1 3EX, United Kingdom

- **Professor Sally E. Kinsey, MD**, Paediatric Haematologist

Telephone: 0113-392-8191 Fax: 0113-392-8488
Email: *sally.kinsey@nhs.net*

- **Ben Sykes**, Nurse Co-ordinator
Telephone: 0113-392-7379 Fax: 0113-392-6629
Email: *benjamin.sykes@nhs.net*

Services Offered:

The Fanconi Anaemia multi-disciplinary clinic provides initial general assessment of cases and confirmation of diagnosis. The multi-disciplinary clinic provides direct access to various specialists.

Patients are assessed and reviewed in Leeds with communication to their referring clinician. Follow-up in the multi-disciplinary clinic is recommended on a yearly basis. Geographically local individuals can have routine follow-up (including blood counts, biochemistry, ultrasound examinations, bone marrow review etc) in Leeds. Individuals living more distant can have care coordinated via their referring clinician.

Where appropriate, work-up for stem cell transplantation can be arranged in Leeds.

International Fanconi Anemia Testing Resources

International Fanconi Anemia Testing Resources

Country	Institution	Test
Australia	<p>Children's Hospital at Westmead Sydney, Australia</p> <p>Zhan he Wu, MD, PhD, FFSc (RCPA) 0061-2-98453236 <i>zhan.wu@health.nsw.gov.au</i></p>	<p>DEB/MMC diagnostic testing for patients and family <i>(Accredited by the National Association of Testing Authorities, Australia).</i></p> <p>Next generation sequencing on trial basis.</p>
Canada	<p>Hospital for Sick Children Toronto, Canada</p> <p>Yigal Dror, MD (416) 813-5630 <i>yigal.dror@sickkids.ca</i></p>	<p>DEB/MMC diagnostic testing (<i>Licensed under the Ontario Ministry of Health and Long Term Care</i>).</p> <p>Canadian Inherited Marrow Failure Registry (Enrollment of patients on this research study includes data collection and sample repository).</p>
Germany	<p>Heinrich Heine University Medical Center ENT (HNO) Lab Düsseldorf, Germany</p> <p>Helmut Hanenberg, MD 49-211-811-0832 or 0830 Fax: 0839 <i>Hanenberg@uni-duesseldorf.de</i></p>	<p>Complementation Group (Research).</p> <p>Use of retroviruses for <i>A, B, C, D2, E, F, G, I, J, L, M, N, O, P</i> in primary T-cells, bone marrow or fibroblast cells or in lymphoblast cell lines (LCL).</p> <p>FANCD2 western blot.</p> <p>Coordinator of the Fanconi Anemia Registry (FAR01) in Germany, Austria, and Switzerland, together with Christian Kratz, MD, Hannover Medical School, Germany.</p>
	<p>University of Würzburg Würzburg, Germany</p> <p>Detlev Schindler, MD, PhD +49-931-888 ext. 4088, 4098, or 4075 <i>schindler@biozentrum.uni-wuerzburg.de</i></p>	<p>MMC Diagnostic Testing (<i>Certified under Governmental and Societies</i>).</p> <p>FANCD2 western blot.</p> <p>Prenatal Diagnosis.</p> <p>Mutation Analysis (<i>Certified under Governmental and Societies</i>).</p> <p>Complementation Group.</p> <p>Carrier testing.</p>

Country	Institution	Test
Netherlands	<p>VU University Medical Center Amsterdam, Netherlands</p> <p>Hans Gille, PhD 31-20-444-8346 <i>jip.gille@vumc.nl</i></p>	<p>Mutation analysis <i>FANCA</i> (<i>Clinically Certified</i>).</p> <p>Mutation analysis <i>FANCC</i>, <i>-E</i>, <i>-F</i>, and <i>-G</i> (<i>Clinically Certified</i>).</p> <p>Mutation analysis of other <i>FANC</i> genes (<i>Clinically Certified</i>).</p> <p>Prenatal DNA diagnosis (<i>Clinically Certified</i>).</p>
Republic of Serbia	<p>Laboratory for Medical Genetics Mother and Child Health Care Institute of Serbia St. Radoja Dakica 6-8 11070 Belgrade, Republic of Serbia</p> <p>Marina Djuriscic, PhD 381-11-3108273 <i>genetika@imd.org.rs</i></p>	<p>DEB diagnostic testing performed in patients with symptoms of FA and on request of the physician/hematologist. Test siblings of DEB-positive patients, but only if requested.</p>
Spain	<p>Hematopoiesis and Gene Therapy Division, CIEMAT/CIBER of Rare Diseases (CIBERER) Madrid, Spain <i>www.ciemat.es</i></p> <p>Spanish Fanconi Anemia Network: <i>www.redfanconi.org</i></p> <p>Juan Bueren, PhD, Head of Hematopoiesis and Gene Therapy 34-91-346-6518 <i>juan.bueren@ciemat.es</i></p> <p>José A. Casado, Senior Researcher 34-91-496-2525 <i>jose.casado@ciemat.es</i></p> <p>Aurora de la Cal, Secretary of the Spanish FA Research Network 34-91-496-2518 <i>aurora.delacal@ciemat.es</i></p>	<p>Coordination of the Spanish FA Registry.</p> <p>FA subtyping by retroviral complementation (<i>A, C, G, E, F</i>) in T-cells (Research).</p> <p>Studies of G2/M cell cycle arrest in fibroblasts in FA patients with mosaicism in peripheral blood and subtyping by reversion of G2/M arrest with retroviral vectors (<i>A, C, G</i>) (Research).</p>

Country	Institution	Test
Spain	<p>Universitat Autònoma de Barcelona/CIBER of Rare Diseases (CIBERER) Barcelona, Spain Spanish Fanconi Anemia Network: www.redfanconi.org http://safcro.uab.cat Jordi Surrallés, PhD 34-935-811-830 jordi.surralles@uab.cat Maria Roser Pujol, PhD Cytogeneticist (UAB-CFS Technical Director) MariaRoser.Pujol@uab.cat</p>	<p>Coordination of the Spanish FA Biorepository (biobank of DNA and cell lines from FA patients).</p> <p>DEB diagnostic testing, diagnosis of mosaicism (certified by Societies).</p> <p>Studies of G2/M cell cycle arrest in fibroblasts, FANCD2 ubiquitination and foci formation, western blot of FA proteins. (Research)</p> <p>Mutation analysis (Research) for Spanish gypsy population.</p> <p>Carrier testing is available for family members when mutations are known in the affected case (sibling, etc.) Genetic characterization (subtyping and mutation analysis) of FA patients is done in the lab for Spanish families under the financial umbrellas of research grants. Service is not free of charge to non-Spanish patients.</p>

Please note:

- The laboratories listed are CLIA (Clinical Laboratories Improvement Amendments)-certified.
- Estimated costs for diagnostic tests range from approximately \$1,000 - \$1,200. Check with your insurance company for coverage. The average turnaround time for testing results is about 10 working days. For more information, please contact the labs directly.
- Additional testing information for Fanconi anemia is available at: <http://geneclinics.org>
Select "Gene Reviews"
Select "Search by Disease"
Enter "Fanconi anemia" into the text box
- "Research" indicates that results may only be used for research purposes, meaning results may not be shared with referring physician/family.

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