FA Courier issue 20

a publication of the Fanconi Anemia Research Fund, Inc.

This is a summary of studies, clinical trials, and current requests for materials for Fanconi Anemia Research Fund (FARF). Visit the Publications section of our website at www.fanconi.org to find complete information about each request or clinical trial, including eligibility and protocol descriptions. Contact us about scholarships for travel assistance to clinical trials. If you do not have online access, please call us at 1-888-FANCONI for assistance.

NDRI

FA researchers are working hard to find effective treatments and a cure for Fanconi anemia, but they can't do it alone. FA researchers need you. Researchers need samples to study, such as tumor samples and biopsied tissue.

Please consider donating research material. All it takes is a phone call to FARF and completion of paperwork for the National Disease Research Interchange (NDRI).

For more information, contact:
Laura Hays, PhD
Executive Director
Fanconi Anemia Research Fund
Telephone: 541-687-4658 or 1-888-FANCONI (888-326-2664)
Email: laura@fanconi.org

Testing Services for FA Patients: Molecular Testing

The Knight Diagnostic Laboratories at Oregon Health & Science University in Portland, Ore., have made available molecular tumor tissue tests that are designed to identify potential treatment targets in cancer, and to predict the likelihood of benefit for patients treated with the latest therapeutics. If your insurance company will not pay for the service, or if you have co-insurance or a deductible that needs to be met, FARF will cover the out-of-pocket expenses.

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Laura Hays
Executive Director
Fanconi Anemia Research Fund
Telephone: 541-687-4658 or 1-888-FANCONI (888-326-2664)
Email: laura@fanconi.org

Send tumor tissue samples to:
Knight Diagnostic Laboratories
2525 SW. 3rd Avenue, Suite 350
Portland, OR 97201
Phone: 855-KDL-1LAB (535-1522)
Fax: 855-KDL-1FAX (535-1329)
Email: KDLClientServices@ohsu.edu

Clinical Trials:

Clinical Trial: Quercetin (dietary supplement)

Quercetin in Children with Fanconi Anemia: A Pilot Study

This is a pilot study aiming to assess feasibility, toxicity, and pharmacokinetics of oral quercetin therapy in patients with FA.

Principal Investigator: Parinda A. Mehta, MD, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio. Telephone: 513-636-5917 (toll-free in the U.S. at 800-344-2462, ext. 65917)
Alternative Contact: Stephanie Edwards, BSN
Telephone: 513-636-9292; Email: stephanieL.edwards@cchmc.org

Needed: Patients with Fanconi anemia complementation group A without current indication of MDS

Gene Transfer for Patients with Fanconi Anemia Complementation Group A (FANCA)

This is a Phase I study designed to develop gene transfer as a treatment for Fanconi anemia complementation group A patients. The objective of this study is to transfer a functional copy of the Fanconi anemia gene for complementation group A by lentiviral vector into bone marrow stem cells from Fanconi anemia patients and then re-infuse these cells with the ultimate goal of improving patient blood counts. The study is currently open for adults and children age 4 or older.

Principal Investigator: Pamela S. Becker, MD, PhD, University of Washington, Seattle, Wash.
Contact: Jennifer E. Adair, PhD, Clinical Research Associate, Fred Hutchinson Cancer Research Center, Seattle, Wash.
Telephone: 206-667-7110; Email: jadair@fhcrc.org

Clinical Trials continued

Clinical Trials: Hematopoietic Stem Cell Transplants

Hematopoietic Stem Cell Transplants

Multiple centers perform hematopoeietic stem cell transplants and are listed at www.clinicaltrials.gov and described in our International Fanconi Anemia Treatment and Testing Resource Guide available at www.fanconi.org. Please refer to FARF's *Fanconi Anemia Guidelines for Diagnosis and Management* for recommentations when selecting a transplant center.

To request a FARF publication, contact:
Cynthia Freeman
Special Projects Coordinator
Fanconi Anemia Research Fund
Email: info@fanconi.org
Telephone: 541-687-4658 or 1-888-FANCONI (888-326-2664)

Needed: Patients with advanced cancers with Fanconi Anemia mutations

Phase II Study of the PARP Inhibitor BMN 673 (talazoparib tosylate) in Advanced Cancer Patients with DNA Mutations in FA Genes

This protocol is a signal seeking study of BMN673, a potent oral PARP inhibitor, in patients with multiple genetic aberrations including Fanconi Anemia mutations. In addition to the required genetic mutations for enrollment onto the study, patients should be older than 18 years old, with adequate organ function, and measurable and biopsiable tumor lesions; Patients also have no history of hepatitis B, hepatitis C or HIV, or a history of stroke or heart attack. Also patients should not have unstable angina, brain metastasis or active infections.

Principal Investigator: Sarina A. Piha-Paul, MD
University of Texas, MD Anderson Cancer Center, Department of Investigational Cancer Therapeutics
Contact: Rosa Mostorino
Telephone: 713-563-1230; Email: rmmostor@mdanderson.org

Samples Requested

Needed: Saliva samples from FA patients with newly diagnosed oral cancer, given after the biopsy and before medical treatment begins

Salivary Biomarkers in Oral Cancer

Researchers have identified biomarkers in saliva that detect oral cancer. They hypothesize that, in the future, these oral cancer salivary biomarkers may be used to help detect early cases of oral cancer in FA patients. Dr. Wong is collecting saliva samples from FA patients with newly diagnosed oral cancer—after the biopsy and before medical treatment begins.

Principal Investigator: David Wong, DMD, DMSc, UCLA School of Dentistry, Los Angeles, Calif.
Contact: Laura Hays, PhD, Executive Director, Fanconi Anemia Research Fund
Telephone: 541-687-4658 (toll-free in the U.S. at 888-326-2664); Email: laura@fanconi.org

Needed: Lesion sample; blood sample; oral rinse with saline

Molecular Surveillance for Squamous Cell Carcinoma of the Upper Aerodigestive Tract

Development of a screening test for cancer of the mouth and throat (head and neck cancer): cancers of the mouth and throat may go undetected in their earliest and most treatable stages. A new test that analyzes the DNA from cells released when an individual gargles is being developed at Johns Hopkins.

Principle Investigator: Wayne Koch, MD, Johns Hopkins, Baltimore, Md.; Telephone: 410-955-4906
Alternate Contacts:
Jessica Bondy, BS, Study Coordinator
Telephone: 410-614-9231; Email: jbondy1@jhmi.edu
Zubair Khan, MD, MPH, Assistant Professor
Telephone: 410-955-3157; Email: zkhan@jhmi.edu

Needed: Blood sample; bone marrow

Laboratory Studies of Gene Transfer for Fanconi Anemia

In order to optimize the delivery of a normal FANCA or FANCC gene to abnormal cells and to test its ability to correct the Fanconi anemia defect in the laboratory, researchers need bone marrow and/or blood samples from patients with FANCA or FANCC.

Principal Investigator: Pamela S. Becker, MD, PhD, University of Washington, Seattle, Wash.
Telephone: 206-616-1589; Email: pbecker@u.washington.edu

Needed: Consent to be in the study; completion of questionnaire; blood sample; sometimes skin sample; medical records

The International Fanconi Anemia Registry (IFAR)

The IFAR is a longitudinal study of patients with FA, which investigates the genetics of FA, the natural history of FA, and factors predisposing to bone marrow failure and cancers. We are also identifying genetic changes that occur in cancers of FA patients, which may be helpful in developing future treatments.

Principal Investigator: Agata Smogorzewska, MD, PhD, The Rockefeller University, New York, N.Y.
Telephone: 212-327-7850; Email: asmogorzewska@rockefeller.edu

Needed: Completion of questionnaires, saliva, and blood from all participating family members, and bone marrow, skin biopsies, and tumor tissue from patients with FA

Etiologic Investigation of Cancer Susceptibility in Inherited Bone Marrow Failure Syndromes

This project seeks to identify all study participants with FA, since some patients are at high risk of developing bone marrow failure, leukemia, and solid tumors prior to the appearance of these adverse events, in order to determine the risks and improve outcomes. Family members are included because of the potential for malignant complications in carriers of mutated FA genes. We will classify patients according to their mutated genes, and determine new genes by next generation sequencing; gene-outcome associations will be evaluated, leading to better understanding of complications and management. All participants are welcome, including those without or with bone marrow failure, those who have had a stem cell transplant, and those who have had cancer.

Principal Investigator: Blanche Alter, MD, MPH
Clinical Genetics Branch, National Cancer Institute, National Institutes of Health, Bethesda, Md.
Contact: Lisa Leathwood, Research Nurse
Telephone: 800-518-8474 or 301-212-5268; Email: NCI.IBMFS@westat.com
Website: www.marrowfailure.cancer.gov

For FA Researchers

FA Antibody Project and Cell Repository

FARF has sponsored the development of high-titer, affinity-purified rabbit polyclonal antisera against the Fanconi anemia complementation group proteins in order to facilitate research on Fanconi anemia.

Affinity-purified antisera are currently available against the following FANC proteins: FANCA, B, C, D1(BRCA2), D2, E, F, G(XRCC9), I, J(BRIP1/BACH1), M, N(PALB2), O(RAD51C), P(SLX4) and the deubiquitinating enzyme USP1. In addition to affinity-purified antisera, unpurified sera are available for many FANC proteins for investigators interested in trying alternative purification approaches.

The FA Cell Repository has immortalized human FA fibroblast cell lines for complementation groups *FANCA*, *FANCC*, *FANCD2* and *FANCG*, as well as immortalized mouse embryonic fibroblasts (MEFs) from knock out mice of the following genotypes: *Fanca*, *Fancc*, *Fancd2*, *Fanca/c*, *Aldh2* and *Fancd2/Aldh2*.

Investigators requesting antisera or cell lines must complete a request via the website http://www.ohsu.edu/research/fanconi-anemia/index.cfm and be willing to meet FARF's use agreement. We depend critically on user feedback on antisera, so will require this as well from all end-users.

General inquiries contact: Ray Monnat, MD
University of Washington, Seattle, Wash.
Telephone: 206-616-7392; Email: monnat@u.washington.edu
Antibody and cell line distribution contact: Leslie Wakefield
Oregon Health & Science University, Portland, Ore.
Telephone: 503-494-6889; Email: wakefiel@ohsu.edu

Availability of Head and Neck Cancer Cell Lines for FA Research

The FA researchers listed below may make FA SCC cell lines available upon request to qualified colleagues. Please note that material transfer standards, policies, and costs may differ between laboratories.

Cell line type	Cell line #	Contact
SCC (FA-A)	OHSU974	Grover Bagby, MD (grover@ohsu.edu); Kim-Hien Dao (daok@ohsu.edu)
SCC (FA-C)	VU-SCC-1131	Hans Joenje (h.joenje@bumc.nl); Josephine Dorsman (jc.dorsman@vumc.nl)
SCC (FA-A)	VU-SCC-1365	Hans Joenje (h.joenje@bumc.nl); Josephine Dorsman (jc.dorsman@vumc.nl)
SCC (FA-L)	VU-SCC-1604	Hans Joenje (h.joenje@bumc.nl); Josephine Dorsman (jc.dorsman@vumc.nl)

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issue 20