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FA Family Newsletter

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issue 52

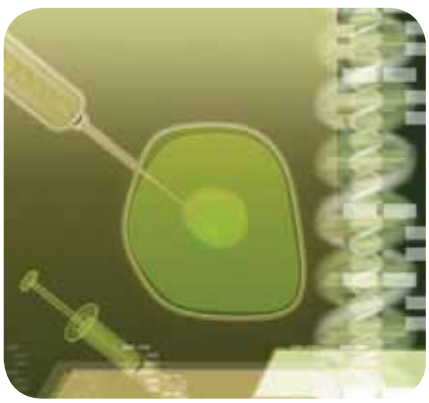
Science News from the Family Meeting

Gene Therapy: Premises and Promises

At the August Fanconi Anemia Family Meeting at Camp Sunshine, Jakub Tolar, MD, PhD, University of Minnesota, Minneapolis, predicted that there will be tangible results in gene therapy for FA patients in 2013.

Dr. Tolar stated that the goal of gene therapy for FA patients is to treat bone marrow failure, prevent leukemia, reduce endocrine problems, lower the risk of cancer and improve quality of life. While a matched sibling donor transplant cures the bone marrow for approximately 90% of FA patients, it can also cause graft-versus-host disease, immune complications, physical injury from chemotherapy and a host of "friendly fire" issues that harm the patient.

For more than a decade, gene therapy has been successful in treating severe combined immunodeficiency disease (SCID). Since the first SCID trial, methods for inserting normal genes into cells have become much safer. FA patients will benefit from that work. But FA poses unique challenges. The stem cells of FA patients are extremely fragile, and the steps necessary to



accomplish gene therapy in SCID patients can cause FA cells to die. We need a method to achieve gene therapy without losing stem cells.

There are different approaches to gene therapy. Scientists can insert a virus carrying an entire normal gene into a cell. This process requires

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INSIDE

Fund Co-founders Recognized for Outstanding Service

David and Lynn Frohnmayer, co-founders of the Fanconi Anemia Research Fund, have been selected by the American Society of Hematology (ASH) as the recipients of the 2012 ASH Outstanding Service Award. The prestigious award is presented by ASH to recognize consummate leadership on issues of importance to hematology research and practice.

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Gene Therapy: Premises and Promises

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penetrating two cellular membranes, incorporating the gene into a chromosome and protecting the virus containing the corrected gene. Since our immune system tries to eliminate viruses, this last complication must be overcome.

A second method involves gene editing. Rather than inserting an entire gene, this method seeks to correct just the "typo" or specific patient mutation. The "mistake" must be cut out and replaced with a corrected DNA sequence. This technology will be a reality in three to five years.

Two years ago, gene therapists from around the world joined forces to create the International FA Gene Therapy Working Group, an effort sponsored jointly by the Fanconi Anemia Research Fund and Fanconi Hope Charitable Trust in the U.K. Scientists now meet annually to collaborate on issues such as choice of vector, mobilization of stem cells and use of conditioning prior to infusing corrected cells. Sharing data and working towards consensus are goals of this group. Their third meeting took place in France in October. ■

Fund Co-founders Recognized for Outstanding Service

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The award announcement honors the Frohnmayers' vision in founding the Fund and applauds their advocacy work "which has resulted in significant improvements in the Fanconi anemia clinical care environment, advancements in information for families and physicians, and increased public awareness of and support for research initiatives that will improve treatment options and lead to a cure for FA."

In nominating the Frohnmayers for the award, Grover Bagby, MD, wrote, "As chair of the Fund's Scientific Advisory Board for more than 20 years, I remain inspired by David and Lynn's dogged pursuit of a better understanding of Fanconi anemia and humbled by their profound commitment to improving the lives of others."

David and Lynn will travel to Atlanta in early December to receive the award at ASH's 54th Annual Meeting, attended by more than 20,000 scientists and physicians. ■

Expert Answers Diverse Questions About FA

Jakub Tolar, MD, PhD, University of Minnesota, Minneapolis, engaged in a lively discussion with Fanconi anemia families, answering questions on diverse issues affecting individuals with FA. Here's a sampling of the questions and answers:

Question: What blood counts are safe for airline travel?
Answer: Platelets of 20,000 or greater should be fine.

Question: Are there natural products without side effects that would be helpful for individuals with FA?
Answer: Every month we learn about yet another compound that might be both safe and therapeutic for FA patients. The important issues are: How do we test this compound, and can we rank-order different compounds to see which is most beneficial? A drug might be helpful in mice but not in humans. We need to test different compounds systematically in a laboratory setting or conduct quick, focused clinical trials to determine which drugs might be most helpful. This is a crucial challenge that should now be a priority for all of us.

Question: Who is eligible for the gene therapy trial at the Fred Hutchinson Cancer Research Center in Seattle?
Answer: Patients must be age 18 or older and in the FA-A complementation group. They cannot have leukemia or a cytogenetic abnormality in the bone marrow and must be free of active infection. This trial has been built to be safe and should not cause organ damage. Patients will need to have a sufficient number of stem cells for this trial to have a chance of success.

Question: Can gene therapy ever be used to treat the oral mucosa of FA patients?
Answer: This is theoretically possible, but we are not there yet.

Question: If you could choose between a stem cell transplant and gene therapy, what would you choose?
Answer: I would go with gene therapy. There will always be toxicities associated with a transplant. We may need to suppress a patient's bone marrow before doing gene therapy, but the chemotherapy dosage will be much lower than with a stem cell transplant. ■

Head and Neck Cancer: Prevention, Screening and Treatment

Dr. Bhuvanesh Singh, Memorial Sloan-Kettering Cancer Center, New York, discussed the causes and evolution of head and neck squamous cell carcinoma (HNSCC) in the general population, and compared these factors to what occurs in Fanconi anemia patients. In the non-FA population, these cancers usually take decades to develop and are caused by the following:

1. Environmental factors: Tobacco and alcohol damage DNA and cause cancer.
2. Inherited susceptibility: We inherit genes that activate carcinogens, fail to eliminate carcinogens from our bodies or do not efficiently repair damage caused by tobacco, alcohol and other cancer-causing agents.
3. Role of the human papillomavirus (HPV): Due to an increase in oral sexual behavior, there is a recent epidemic in cancers of the oropharynx caused by HPV. In New York City, more than 75% of young adults report oral sexual contact. If exposed to HPV, 95% of these young adults will clear the virus and less than 1% will get cancer.
4. HPV is now the most common cause of oropharyngeal cancer in the general population.
5. Patients in the general population with HNSCC caused by HPV experience a high cure rate. Persons whose cancers are related to tobacco and alcohol do poorly, possibly due to the increased severity of genetic abnormalities in the cancers caused by these carcinogens.

Head and neck cancers affecting FA patients show major differences from those that affect the general population:

1. FA patient cancers evolve faster, in years rather than decades.
2. HNSCCs appear early in the lives of FA patients (in their teens, 20s and 30s).
3. The number of genetic changes in the tumors is higher than in non-FA patients.
4. FA cancers are more aggressive and are often at an advanced stage when detected.
5. FA patients do not easily tolerate the main therapies used to treat HNSCC, such as radiation and chemotherapy.

Dr. Singh emphasized the huge importance of early detection in FA HNSCCs, noting "a cure requires early detection." Surgery is the preferred treatment. In select cases, new tools such as robotic surgery can facilitate the removal of cancerous tissue and speed recovery. Patients must undergo very close surveillance following tumor removal.

Cancer patients should seek treatment at a major medical center with a multidisciplinary treatment team. The FA patient has just one opportunity after cancer detection to maximize the chances for the best outcome. In addition to surgery, epithelial growth factor antagonists such as Erbitux might be effective against these cancers. Treatment decisions should be made in consultation with experienced physicians. ■

FA patients should follow strict guidelines in an effort to prevent these cancers:

1. Abstain from tobacco! Smoking is highly addictive: Once one starts to smoke, it's exceedingly difficult to stop. There are more than 4,000 different chemicals in tobacco and at least 60 of these cause cancer.
2. Marijuana is just as harmful as tobacco.
3. Parents should never smoke, especially around their children. Secondhand smoke exposure can also cause cancer.
4. Abstain from alcohol. While one alcoholic drink a month is unlikely to cause cancer, abstention is safest. Beer, wine and hard liquor all have the same negative effects.
5. Aggressive monitoring and routine screening is mandatory. Individuals with FA should examine their own mouths carefully once a month (not daily).
6. A specialist in head and neck cancer should carefully examine patients' mouths and oropharynx twice a year.

Transplant Overview from Cincinnati; Progress Report on Multi-Center Study Eliminating Radiation

Transplant Overview:

Stella Davies, MBBS, PhD, MRCP, Cincinnati Children's Hospital Medical Center, gave a very helpful overview of the steps involved in a stem cell transplant.

Transplant timing varies considerably from patient to patient, and some individuals will never need a transplant. Severe infections prior to transplant and some specific gene mutations help predict when bone marrow might fail. But even siblings with the same genetic mutations can experience bone marrow failure at different ages.

Cincinnati Children's Hospital Medical Center has done 35 matched sibling donor transplants, and 88% of these patients survive. The present protocol eliminates radiation in matched sibling donor transplants. Nine patients have been transplanted without using radiation, and all nine survive. Survival time in this cohort ranges from eight years to two months.

Eighty-five percent of patients who lack a sibling match can find a suitable alternative donor. The National Marrow Donor Program has 9 million donors registered, and 19 million donors are available worldwide. Five hundred thousand cord bloods are frozen in banks throughout the world.

Multi-Center Study:

Four transplant centers (Memorial Sloan-Kettering Cancer Center, New York; Children's Hospital of Wisconsin; Cincinnati Children's Hospital Medical Center; and Boston Children's Hospital) are participating in a study to determine if busulfan can safely replace radiation in unrelated donor transplants, thereby decreasing the risk of later malignancies. This trial has been open for four years, but accrual of patients was slow until two and a half years ago, when Cincinnati admitted patients to this trial. To date, 28 patients have enrolled in this trial and 24 survive. Survival time ranges from four years to two months. Three patients died of infection and one patient died of regimen-related toxicity.

> Dr. Davies believes that radiation is no longer needed in unrelated donor transplants.

After the first 25 transplants, researchers decided to administer a lower dose of busulfan to the next cohort of 10 patients. Three have been transplanted to date on this protocol. If this reduced dosage is successful, the next 10 patients will receive an even lower dose of busulfan. Dr. Davies believes that radiation is no longer needed in unrelated donor transplants. ■

New Trial Using an Antioxidant Opens at Cincinnati Children's Hospital Medical Center

Stella Davies, MBBS, PhD, MRCP, transplanter and researcher at Cincinnati Children's Hospital Medical Center, reported that her center has opened a new trial to determine whether a particular flavonoid can preserve stem cells in individuals with Fanconi anemia. Flavonoids are naturally occurring compounds abundant in fruits and vegetables that have strong antioxidant properties. In mice bred to have FA, the flavonoid under investigation cures pre-diabetes, does not cause leukemia and improves bone marrow function.

Dr. Davies noted that individuals with FA produce high levels of a protein called tumor necrosis factor alpha (TNFa), and that TNFa kills stem cells by making reactive oxygen species (ROS). Dr. Davies hypothesizes that removing ROS by ingesting this flavonoid might prevent bone marrow failure in FA.

The first three patients in this trial must be age 12 or older. For further information about this study, contact Parinda Mehta, MD, Cincinnati Children's Hospital Medical Center. Email: parinda.mehta@cchmc.org; telephone: 513-636-5917.

Related and Unrelated Stem Cell Transplant for FA: Current Results and Future Directions

By John E. Wagner, MD
University of Minnesota

Since 1976, the University of Minnesota has performed more than 200 Fanconi anemia patient transplants. This center has conducted a series of informative clinical trials that have resulted in markedly improved survival rates compared to those expected in the mid-1990s.

For the entire group of FA patients with a human leukocyte antigen (HLA)-matched unrelated marrow donor or partially matched umbilical cord blood donor, treated on six different protocols since 1990, the probability of survival was 61%. Median follow-up was nine years. Mortality was higher in patients older than 10 years or who received any transfusions or developed infection prior to transplant. Mortality has been lowest in patients treated on the current regimen of cyclophosphamide, fludarabine, low-dose total body irradiation (300 rads) and thymic shielding. Survival was 92% at five years post-transplant for patients treated on this regimen who were age 10 or younger, and who had not had transfusions or prior invasive infections.

Similarly, FA patients with an HLA-matched sibling donor have experienced marked improvements in survival over time. Today, the probability of survival is 96% for patients treated with fludarabine, low-dose cyclophosphamide and no irradiation.

The next changes in treatment are geared to reducing the side effects of transplant. For example, mycophenolate mofetil (MMF) has replaced prednisone to reduce the risk of steroid-related complications, such as avascular necrosis of the joint bones, high blood sugar levels and the need for insulin. In addition, treatments are tailored for the patient. For example, patients with mutations in *BRCA2* require higher dose chemotherapy.

The next changes in treatment are geared to reducing the side effects of transplant.

The next significant changes will be replacement of cyclosporine A with rapamycin, which has fewer side effects, and the introduction of regulatory T-cells, which control the immune system and prevent graft-versus-host disease. The hope is that use of regulatory T-cells will eliminate or reduce the need for any immune suppression after transplant.

While longer follow-up is needed, the cancer risk in the Minnesota group thus far has remained low. Only 2% of patients have developed cancer, with a follow-up time of up to 15 years. ■

Sources Offer Information About Alternative Therapies

For those interested in researching the efficacy of herbs, nutraceuticals and other alternative therapies, Memorial Sloan-Kettering Cancer Center and the National Institutes of Health's National Center for Complementary and Alternative Medicine are two respected sources. Below are related links:

- **Memorial Sloan-Kettering Cancer Center:** http://www.mskcc.org/cancer-care/integrative-medicine/about-herbs-botanicals-other-products

  FREE iTunes app for Memorial Sloan-Kettering's site: https://itunes.apple.com/us/app/about-herbs/id554267162?mt=8
- **National Center for Complementary and Alternative Medicine**: http://nccam.nih.gov/

The Fanconi Anemia Research Fund publishes these resources as information only. Always check with your physician(s) before using any supplements. And bear in mind that "natural" doesn't always mean "safe."

Physician Discusses GI and Feeding Issues Related to FA

Sarah Jane Schwarzenberg, MD, University of Minnesota, Minneapolis, spoke on gastrointestinal (GI) and feeding issues in Fanconi anemia, noting that 22% of individuals with FA are underweight, 27% of people with FA are overweight or obese and 5% have congenital GI abnormalities.

Dr. Schwarzenberg recommended a daily 5-2-1-0 plan as a good idea for everyone; five or more servings of fruits and vegetables; fewer than two hours of screen time; one hour of moderate exercise; and zero sweet drinks. If a patient is vitamin-deficient, supplementation may reduce risk. Dr. Schwarzenberg noted, however, that vitamins, minerals, flavonoids and antioxidants are drugs that in some cases may lead to unintended results. An appetite stimulant or supplemental feeding may be considered for underweight individuals after a medical evaluation for causes of poor intake.

GI problems in FA include nausea, abdominal pain, diarrhea, liver adenomas from androgen use and complications from stem cell transplant. Gastroesophageal reflux, which is more common with age, can cause heartburn, burping, vomiting and belly button pain. Small bowel overgrowth can be exacerbated by medications and antibiotics, and causes excessive gas and abdominal pain.

Evaluation of GI symptoms should include a good history and physical exam followed by specific diagnostic testing. A GI evaluation should be performed prior to stem cell transplant so that any issues may be addressed.

Dr. Schwarzenberg stressed seeking medical attention for any of the following symptoms: involuntary weight loss, decrease in past growth rate, GI blood loss, significant vomiting, chronic severe diarrhea, unexplained fever, persistent abdominal pain or family history of inflammatory bowel disease. ■

FA Often Involves Endocrine Issues

The endocrine system involves several glands, each of which secretes different types of hormones directly into the bloodstream to regulate the body. As such, a problem in one part of the body affects other parts of the body. Most children and adults with Fanconi anemia have an endocrine issue related to FA itself or to treatment. Susan Rose, MD, Cincinnati Children's Hospital Medical Center, shared her data on endocrine issues from 120 people with FA. Common endocrine problems in FA include weight issues, impaired insulin release, hypothyroidism, and early or late puberty. Dr. Rose reported that growth hormone deficiency is rare and bone mineral density is normal in FA regardless of whether the patient has had a bone marrow transplant.

Dr. Rose stressed the importance of an annual endocrine evaluation for people with FA. Low weight may be addressed with diet and supplements. Glucose or insulin treatment can adjust impairments and help with weight problems. Hypothyroidism may be treated with hormone therapy, and puberty issues can be addressed with estrogen, progesterone or testosterone supplementation. ■

Physician Discusses Options for Treating Arm and Hand Anomalies

Scott Kozin, MD, Shriners Hospital for Children, Philadelphia, discussed options for treating physical anomalies in Fanconi anemia. Up to 80% of FA physical anomalies are skeletal; of those, 70% are in the upper extremities, primarily the radius and thumb.

Radial deficiencies can affect nerves, tendons and arteries as well as the bone itself. Treatment goals include maximizing forearm length and alignment, improving function and appearance, and stablilizing the wrist. Splinting to stretch the arm is best started early and helps increase mobility. Dr. Kozin reported that he no longer routinely advises centralization (surgery to straighten forearms) because, although it may improve appearance, it has not been shown to increase function. Certain children remain candidates for centralization. He also does not lengthen bones in small children, preferring to wait until the adolescent child requests the procedure.

Dr. Kozin considers treatment of thumb anomalies critical since the thumb provides 50% of hand function. Treatment includes reconstruction or pollicization (creating a thumb from an existing finger, typically the index). In FA children the index finger is usually mobile and robust. Hence, when pollicized into the thumb position it works well in pinching small items and grasping large objects. There is little therapy needed after pollicization, and very little risk of nerve damage or later arthritis. Dr. Kozin pointed out that tasks become increasingly difficult as children grow older and limitations may then become more apparent. Accommodations, such as using a larger pen, can help in certain tasks. Additional therapy or surgery may also be necessary to maximize function. ■

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Testing Service for FA Patients

Testing for Potentially Beneficial Cancer Therapy

The Knight Diagnostic Laboratories at Oregon Health & Science University have recently made available new molecular tumor tissue tests designed to identify potential treatment targets in cancer and to predict the likelihood of benefit for patients treated with the latest therapeutics.

This new testing is available at NO CHARGE to FA patients.

For more information, contact:
Teresa Kennedy, Director of Family Support Services
Fanconi Anemia Research Fund, Inc.
Phone: 541-687-4658 or
1-888-FANCONI (888-326-2664)
Email: teresa@fanconi.org

Or contact:
Christopher Corless, MD, PhD, Medical Director
OHSU Dept. of Pathology (mailcode L113)
3181 SW Sam Jackson Park Road
Portland, OR 97239
Phone: 503-494-6834
Email: corlessc@ohsu.edu

Girls and Women with FA Face Gynecological and Reproductive Issues

Fanconi anemia poses unique gynecological and reproductive issues for girls and young women, according to Jill Huppert, MD, MPH, Cincinnati Children's Hospital Medical Center. These include potential effects on puberty and fertility, and a significantly increased predisposition to gynecological cancers.

The most common effects of FA related to puberty are delayed menstrual periods and delayed onset of puberty, which may be influenced by low weight, chronic illness or androgen/thyroid treatment. Heavy periods may result from low platelets; some patients could benefit from use of oral contraceptives.*

Evidence indicates that women with FA are at greater risk for early menopause and decreased fertility. Dr. Huppert recommends that women interested in starting a family should not delay childbearing. Young women facing a bone marrow transplant (BMT) may consider consulting a reproductive endocrinologist before beginning treatment. One recent study found a higher incidence of pregnancy than expected among women with FA post-BMT: of 101 women over age 16 and post-transplant, 10 women experienced 14 pregnancies.

Women with FA have a significantly greater risk of developing gynecological malignancies, including cervical, vulvar and anal cancers. The human papillomavirus (HPV), especially HPV subtypes 16 and 18, has been implicated in almost all cervical cancers and roughly half of vulvar cancers in the general population. HPV 16 and 18 may also be linked to malignancies of the oral cavity. Dr. Huppert recommends that young women with FA receive the HPV vaccine that protects primarily against these strains beginning at age 11 and practice safe sexual behavior. This includes abstinence, safe sexual practices (limiting the number of sexual partners and using condoms) and regular screenings. Pap smears should begin within one year after first sexual intercourse, or at age 18. Annual gynecological exams after age 18 should include pap smears. Vulvoscopy/colposcopy is indicated following an abnormal pap or visible lesions. Women with FA should also begin mammography screening for breast cancer at age 25.

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Expressing sexuality is a normal part of development. Dr. Huppert recommends safe, open conversation about sexual issues among adolescents and their parents and physicians. ■

**If there are concerns about regularity of periods, Dr. Huppert recommends using a calendar system to log periods. She also recommends the following free app for people with app-friendly technology (smart phones, etc.): period tracker.*

New Study Underway to Detect Oral Cancer in FA

If you or someone in your family is diagnosed with oral cancer, please consider participating in a new research study funded by FARF to determine if saliva can be an early detection tool for oral cancer. Contact Teresa Kennedy as soon as possible after diagnosis and before treatment at teresa@fanconi.org or 888-FANCONI. Teresa will coordinate your participation with David Wong, DMD, DMSc, the study's principal investigator. For more information, visit Research Highlights on our website.

PGD Offers FA and HLA Testing Before Pregnancy

Preimplantation genetic diagnosis (PGD) offers families at risk for a genetic disease like Fanconi anemia the option of embryonic testing for FA and human leukocyte antigen (HLA) matching before pregnancy. Andria Besser, MS, CGC, Reproductive Genetics Institute (RGI), Chicago, discussed the process to interested parents at the Family Meeting.

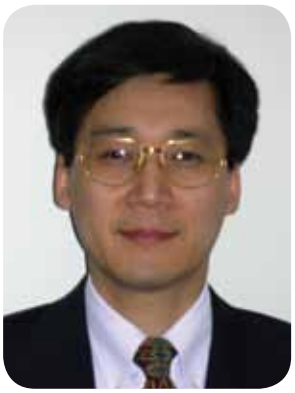
PGD uses *in vitro* fertilization (IVF) in which the woman's ovaries are stimulated to produce multiple eggs. These eggs are removed and fertilized by the man's sperm in the laboratory. The resulting embryos are tested for FA mutations, and HLA if requested. Healthy embryos can be transferred into the woman's uterus with the hope of achieving pregnancy. FA and HLA testing is 95-98% accurate; IVF success rates vary, depending on the woman's age, hormone levels and other factors.

PGD testing has been an option for FA and HLA for more than 12 years. RGI has assisted more than 20 FA families through more than 60 PGD cycles. Eleven babies have been born after PGD for FA, all FA-free. Of the families who chose to do HLA matching, seven babies have been born who are both free of FA and an HLA match to an affected sibling.

> PGD is an option to reduce dramatically the risk of having an affected child and to identify an HLA-matched embryo.

PGD is expensive as well as physically and emotionally difficult, and the IVF process can be associated with low pregnancy rates. However, PGD is an option to reduce dramatically the risk of having an affected child and to identify an HLA-matched embryo as a potential donor for a sibling in need of transplant. ■

Ear and Hearing Problems in FA Patients



H. Jeffrey Kim, MD, National Institute on Deafness and Other Communication Disorders, National Institutes of Health (NIH), Bethesda, Md., discussed ear and hearing problems in individuals with Fanconi anemia. These issues affect approximately 11% of FA patients and may lead to early diagnosis of FA. A comprehensive ear, nose and throat evaluation including an audiogram is recommended for all FA patients; regular follow-ups are necessary if problems are present.

Dr. Kim reported that of 31 FA patients seen at the NIH, 33% of the 62 ears showed some degree of hearing loss, most commonly mild to moderate conductive hearing loss. Ear drum or middle ear abnormalities—small ear drum, abnormal presence of bony plates on ear drum, and malformed middle ear bones—were seen in 56% of the ears.

Hearing loss can be managed with hearing aids or other devices. Behind-the-ear hearing aids are preferred for young children since they are easiest to use and allow for ear growth. An FM auditory trainer is strongly recommended for school settings; the teacher wears a microphone which transmits to the student's receiver. The implantable bone-anchored hearing aid (BAHA) and the SoundBite system are options for some patients with hearing loss or even absent ear canal. Surgery can correct middle ear bones and widen the ear canal. Surgically creating an ear drum or canal is challenging, but can be successful with an experienced surgeon. ■

> These issues affect 11% of FA patients and may lead to early diagnosis of FA.

How to Advocate for Students with FA

Renee Ortiz, MSW, City of Hope, Duarte, Calif., provided parents at the Family Meeting with tips on advocating for their child at school. The family of a student with a chronic or life-threatening condition may have concerns or questions about how the student can best benefit from the school experience. School personnel may have concerns as well. Ortiz noted that since each situation is unique, no one plan fits all. She stressed that communication and patience are vital in all cases. She advised parents to utilize available school services such as individual education plans, school psychologists or special education coordinators. A shortened school day, one set of books at school and another at home to prevent carrying a heavy load, and flexibility with physical education requirements are among accommodations that might be considered. If the student is absent due to a hospital stay, it's helpful to have a liaison between the hospital and school. Attending school helps a child feel "normal" and productive. Communication and advocacy help address any challenges toward a rewarding education. ■

> No one plan fits all, but communication and patience are vital in all cases.

Parent-to-Parent: Making the Transition from Pediatric to Adult Care

Parents Amy Levine, Brian Horrigan, and Beth and Mike Vangel shared tips from their experience in transitioning from pediatric to adult care with their children with Fanconi anemia. They advised preparing teens by being frank about the hows and whys of managing their health care, modeling good working relationships with doctors and training teens to advocate for themselves. The parents warned that although such efforts may be met with resistance, it's important to maintain a goal of open communication.

After patients reach the age of 18, doctors cannot release medical information to parents without patient consent. At age 26, a child is no longer covered by parents' health insurance, which can be a big issue. Transition from pediatric to adult care can be gradual and often depends on the doctor. Keeping track of appointments can be overwhelming for young adults. Parental encouragement is important. Let the young adult know that managing care for such a complex disease "is hard because it *is* hard."

> Let the young adult know that managing care for such a complex disease "is hard because it *is* hard."

The parents' own transitions varied from feeling impatient for their child to become independent to struggling to let go. The process was seen as striking a fine balance—treating the child as an adult, but always being there when needed. ■

Save the Date!

25th Annual Fanconi Anemia Research Fund Scientific Symposium
October 24-27, 2013 • Houston, Texas • www.fanconi.org

Researchers Gather to Investigate FA-related Cancers

At the request of the Fanconi Anemia Research Fund, more than 70 researchers and clinicians specializing in squamous cell carcinoma (SCC) gathered in Chicago last March to discuss screening, prevention and treatment strategies in Fanconi anemia as well as the role of the human papillomavirus.

Continuing a format initiated at the April 2010 SCC meeting, participants focused on one of the four areas noted above and presented numerous recommendations to move research and clinical standards forward:

- Establish and maintain a biospecimen resource specifically for Fanconi anemia specimens such as tumor tissue, saliva and blood
- Conduct a longitudinal molecular characterization study of SCC progression in adults with FA
- Conduct a study to determine adequate margins in SCC surgery for the FA patient
- Conduct an analysis of targeted molecular therapies for the treatment of SCC in individuals with FA
- Develop a photo repository of head and neck cancers to distribute to clinicians and families, and to facilitate early detection and treatment of SCC
- Assemble a board of SCC clinical advisors to draft recommendations to share with both patients and treating physicians
- Produce a video for self-detection of suspicious mouth and tongue lesions
- Ensure that dental hygiene and healthy lifestyle choices are emphasized in materials produced by the Fund and by speakers at every patient/family meeting

In addition to progress on several of these recommendations, the FARF Board has recently approved several SCC-related grants. Please refer to page 23 for a list of projects funded as of the publication date, and watch for updates on Facebook and on the family e-group. We continue to devote significant resources toward a better understanding of how to screen, prevent and treat these devastating cancers in people with FA. ■

Researchers Recognized for Discovery of 15th FA Gene

The work of several laboratories in the United States and Europe led to identification of the 15th Fanconi anemia gene, *SLX4*, or *FANCP*. The following researchers were recognized with a Discovery Award at the Fund's Scientific Symposium held in October 2011 in Barcelona, Spain:

- Chantal Stoepker, MSc; Martin A. Rooimans; Jurgen Steltenpool, PhD; Anneke B. Oostra; Aggie W. M. Nieuwint; Hans Joenje, PhD; Johan P. de Winter, PhD; Vrije Universiteit, Amsterdam
- Karolina Hain; John Rouse, PhD; University of Dundee, UK
- Beatrice Schuster; Katharina Eirich; Detlev Schindler, MD, PhD; University of Wuerzburg, Germany
- Yvonne Hilhorst-Hofstee, MD; Elisabeth T. Korthof, MD; Leiden University Medical Center, Leiden, Netherlands
- Nicolaas G.J. Jaspers, Erasmus Medical Center, Rotterdam, Netherlands
- Thomas Bettecken, MD, Max-Planck-Institut für Psychiatrie, Munich, Germany
- Yonghwan Kim, PhD; Francis P. Lach; Rohini Desetty, PhD; Agata Smogorzewska, MD, PhD; Arleen D. Auerbach, PhD; The Rockefeller University, New York
- Helmut Hanenberg, MD, Indiana University School of Medicine, Indianapolis; Heinrich Heine University, Duesseldorf, Germany ■

Since 1989, the Fanconi Anemia Research Fund has awarded 181 research grants to 95 researchers at 53 institutions totaling $15.1 million. For details, go to www.fanconi.org.

FA Family Meeting Draws Record Number of Attendees

Fifty-seven families from seven countries gathered at Camp Sunshine in Casco, Maine, in August for the Fanconi Anemia Research Fund's 21st annual FA Family Meeting. Despite a few heavy downpours, Camp Sunshine lived up to its name by the measure of smiles, laughs and hugs over the five days.

Forty-eight children and 10 adults with FA, ranging in age from 10 months to 52 years, attended the meeting along with parents, siblings and extended family members.

Written evaluations conveyed enthusiasm about the scientific and medical presentations at Camp Sunshine. Participants learned about progress in gene therapy, had the opportunity to ask questions of an FA expert, and heard updates from two bone marrow transplantation (BMT) centers, plus the multi-center BMT trial. Other presentations focused on post-BMT follow-up, preimplantation genetic diagnosis, hearing, hand, gastroenterology and endocrine issues. Teens and young adults with FA and their parents were encouraged to attend sessions on prevention and treatment of head and neck cancer, and young FA women attended a presentation on gynecology. One speaker addressed ways to help navigate the school experience. As always this was a comprehensive and highly educational meeting!

Brian Horrigan, Amy Levine, and Beth and Mike Vangel shared thoughts and tips about transitioning children from pediatric to adult care. Bev Mayhew gave an overview on Fund expenditures and fundraising, and Lynn Frohnmayer described current FARF-funded research projects. Kevin McQueen enthusiastically led the group in a light-hearted game designed to emphasize the importance and relative ease of fundraising.

A record number of attendees kept the volunteers and camp staff busy, but by all accounts they handled the workload with grace and humor. Campers created fond memories with new and old friends and left with the promise of future connections. ■

In Loving Memory

"For some moments in life there are no words."

Kenneth Evans ..... *8/18/65 - 2/28/12*
Amy Wang ..... *1/21/08 - 6/5/12*
Alyah Haynes ..... *12/8/03 - 6/20/12*
James Hayes ..... *6/14/04 - 7/25/12*
Luella Dueck ..... *9/2/61 - 7/30/12*
Rebecca Tsimmerman Pritchett ..... *8/12/82 - 8/28/12*
Natalie Curry ..... *1/11/85 - 8/30/12*

FA Family Meeting Supports a Journey

Daniel, Mindy and Isaac Coleman

By Mindy Coleman

Camp Sunshine. A fitting name for the hope and sense of community we would feel upon leaving, and also appropriate for our own little ball of sunshine. For us as a family of three, the Fanconi Anemia Family Meeting at Camp Sunshine could not have come at a more perfect time.

Our son, Isaac, was born Oct. 3, 2011. He was diagnosed with FA three weeks later. We were grateful for such a quick diagnosis and heartbroken at the same time. As a young couple (known as "the babies with the baby" at camp), we found ourselves thrust into not just being new parents, but advocates, experts and caregivers, too.

Isaac was born with multiple anomalies: absent radii and thumbs, hydrocephalus, imperforate anus, fused kidneys, severe hearing loss and others. We quickly delved into the world of doctors and therapies and learned as much as we possibly could about FA, as we tried to help our family and friends understand, too. Fast forward 10 months and four surgeries, and we were at Camp Sunshine.

For us, the Family Meeting was about people. Seeing other children throughout the week, we found ourselves thinking about Isaac and what he will be like as he grows older—and even if he will grow to be as old as some of the kids we met. We could see him in a boy playing piano at the talent show or in another little one running past us towards the lake. Our hearts came near to exploding, quietly, with love for each person we met. This year was about soaking in bits of other peoples' journeys. All different. Some looked similar to ours, but they were all unique.

We experienced many firsts on our trip: first flight with a baby, first time in Maine, first time decapitating and eating a lobster, first time singing karaoke for either of us (if you can believe it after my husband Daniel's stellar performance of "Stayin' Alive"), the first time we handed our son off to complete strangers—multiple times, knowing he would be okay—and the first time we felt completely understood. Until last week, I don't think we even realized what a wonderful feeling it was to be around people who were intimately familiar with the way Isaac and our family now functioned. There was something glorious about not having to explain ourselves and our son to the people around us. Everyone just knew. We would never wish our circumstances on others, but there is truly such comfort and encouragement found in knowing that others have felt exactly what you have and have made it through.

There was something glorious about not having to explain ourselves and our son to the people around us. Everyone just knew.

For us, the journey has just begun. We don't know what the future will look like, but we know that Isaac's life, just like ours, has purpose. And we have our loving God and now our wonderful new FA family to keep us strong. We hope the future will include many, many trips to Camp Sunshine for the FA Family Meeting to learn, to encourage, to be encouraged and to reconnect with our FAmily! ■

Find us on Facebook at www.facebook.com/fanconianemiaresearchfund

How to Talk with Children about Fanconi Anemia

By Nancy F. Cincotta, MSW, MPhil

Psychosocial Director, Camp Sunshine

At different stages in the course of having a child diagnosed with Fanconi anemia the question arises, "How do I talk to my child about FA?" How you talk to your child parallels your own coping and your family's communication style. The conversation also will vary depending on the age of the child and the treatment ahead.

Here are some suggestions for discussing FA with your child:

Give it a name: Fanconi anemia. Give it a description. Encourage kids to ask questions, ask them questions and minimize guessing.

Fanconi anemia is…

- *Something that is in the genes in your body.*
- *Something that affects your blood, so we have to check to see how your counts are doing. Counts are… bone marrow is…*

Some people with FA…

- *Have hands that look different, because some children with FA have thumbs that develop differently.*
- *May be shorter than their classmates because of FA.*

Discussions appropriate to age, knowledge base and situation provide the best potential for understanding and effective coping. The description and dialogue about FA must be updated regularly as a child's cognitive and emotional abilities grow. Very little is static in a child with FA. Changes in medical status, a child's comprehension of FA, and advances in treatment approaches and research can make managing communication seem overwhelming. Still, change can serve as the groundwork for an ongoing dialogue, addressing what is imminent as the priority.

Honest discussion and information sharing from the beginning establishes trust and open communication. When accidental learning becomes a pattern of communication, it can complicate the trust children experience within their family. Family members may not have chosen this illness journey, but they can make choices about how to travel on it.

Young children who have noticeable differences will need simple explanations to understand why they are different. When they reach school age, they'll learn that other children don't have the same medical experiences and will require explanations of the differences.

A child can feel many things, including loneliness, isolation, depression and confusion about FA. Acknowledge that FA is confusing, but that you're there as a resource to help sort it all out. It's okay for children to know that adults may not always know the answers, and also to know that adults will try to find the answers.

Parents often worry more about the future than young children do. Burdening children with adult worries is fraught with unnecessary complications. It's important to understand what's on children's minds, what they want or need to know, what they're capable of understanding, and how they process information. Some children need to be prepared far in advance of a procedure, whereas others do better receiving information in a more limited time frame. Learn what is best for your child.

Keep the dialogue ongoing and hopeful. Kids will come to you with questions first, and will continue to come if you're responsive. Children will often ask questions that reflect their own tolerance for information. Siblings, as well as children with FA, need information shared with them in an age-appropriate way.

As children get older, some of the discussions about FA get easier and others more complicated. At certain ages children and teens may not want parents to discuss their situation with others, wanting to "own" the information about their current issues. It becomes important for children, now emerging adults, to be in the ongoing dialogue about the sharing of information. They still have the right to some privacy about their medical information and how and where it's discussed.

Kids talking to other kids with FA allows for a type of "kid wisdom," a camaraderie that is unequalled and an understanding that mitigates a sense of isolation. Knowing that they're not the only one in the world with FA can go a long way towards maintaining the mental health of children, teens and adults with FA. ■

My Life as an Adult with FA: Never a Dull Moment

By Elizabeth Walker

My name is Elizabeth Walker. I am 37, married, live in Scotland, and I have Fanconi anemia. My childhood was normal until I was admitted to hospital at age 9 and misdiagnosed. At 16, I was told there was nothing wrong with me. I met my husband when I was 17. We later bought our first home together and were married in 1996.

In March 1997, I was given a blood test when I had an infection that wouldn't go away. The next day I was told that I had FA and that my blood counts were very low. I was quickly given a bone marrow aspiration which showed FA, myelodysplastic syndrome and the start of leukemia. The diagnosis was a shock to us. My first question was if I could still have children.

I needed an urgent bone marrow transplant (BMT). My siblings were tested, and my oldest brother, who was in New Zealand, was a match. I was in hospital for a month getting transfusions and was told I didn't have much time left. I was admitted for transplant in August 1997. My brother flew home to donate his marrow, and stayed until it was over. I got out in mid-September, very good timing for a BMT patient.

I was then diagnosed with chronic graft-versus-host disease (GvHD) which can be a major problem. The first year I was in and out of hospital with various serious infections. In 2001, doctors tried photopheresis for my GvHD. In 2005, the GvHD got worse and we tried several treatments including cyclosporine. By 2010, I had kidney failure due to years of cyclosporine and my body couldn't cope with the GvHD anymore. My doctors decided to try rituximab, an experimental therapy for GvHD. After a month my blood results improved. I was doing well until I developed cytomegalovirus. The virus attacked my eye, causing a detached retina and requiring two surgeries.

In 2011, I was diagnosed with dysplasia, or precancerous changes, of the mouth. I had two surgeries to remove part of my tongue, and am also treated for a patch of dysplasia on my face. I get renal anemia due to failing kidneys and will eventually need dialysis. I have osteoporosis and a slipped disc in my lower back which can be very painful. I'm still immune-suppressed and catch bugs easily.

So what's life like as an adult with FA? My husband Graham says it's never a dull moment. I try to take care of myself as best I can, but I also want and need some kind of normal life. I have good days and bad days. I try to stay positive, but living with a terminal illness isn't easy.

> FA doesn't stop me. I'm stubborn and push myself for normality.

FA doesn't stop me. I'm stubborn and push myself for normality. My husband and I are like any other normal married couple; we just have to make some compromises. We don't plan too far ahead. When we have plans, I rest up so I'm able to enjoy my special occasions out. We don't have kids (after BMT I wasn't able to), and I can't adopt here due to having FA. My husband says we have each other and that's fine by him; he says I still make him laugh every day. The only physical sign I have of FA is my height, I'm only 4'10". When things are going well, you wouldn't know I have anything wrong with me.

I'm scared of the future, but I can't control what's going to happen. I just enjoy life in the now and do what I can. I'm glad that I can help the parents of FA children, and I've become close friends with some FA adults. We help each other through the bad times. And I am so thankful to have a wonderful husband. The fight continues... ■

Who's Behind the Blue Mask?

Chloe with brother Liam

By Chloe Rogers, Age 11

Have you ever seen someone with a blue mask or no hair? Have you ever wondered why? I was like that; I wore a blue mask and now have short hair (I did lose all my hair at one point). I am like this because I have Fanconi anemia, a blood disorder that lowers my ability to make blood cells and fight infections.

I wore a blue mask not because I have Fanconi anemia but because I had a bone marrow transplant. A donor gave bone marrow to the hospital by minor surgery, then the bone marrow was put in my body through an IV. My body allowed the new marrow to take control, because I had chemo medicine and radiation to kill off my own blood cells. Because of my own cells dying and my donor cells just coming in, I was vulnerable to infections. So I wore a mask and took a lot of medicine so I didn't get sick. My mask was a little annoying and could get in the way.

I had my bone marrow transplant in a Minnesota hospital about a year ago, and I had to live in isolation for seven weeks. After a while, I started getting my strength back and started taking short walks. I am getting stronger every day. I go to school four days a week, and I no longer have to wear the mask—hooray! If I could, I would go to school every day so I wouldn't miss out on all the fun and work. If things go well, in two years I should be just like every other kid.

I do have fun. I still have play dates. I just have to be careful. So now when you see someone with short hair or a blue mask, you don't have to wonder why. Instead, just say "Hi!" ■

Editor's Note: This article appeared in the June 7, 2012, issue of *Newsday*.

Congratulations!

Egil (FA) and Nanna, whose daughter, Flora, was born Aug. 1.*

**Last names withheld on request.*

Katie Criss (FA) and Jonathan Jensen's son, Bentley, arrived on May 16.

Lisa Doyle Kinsella (FA) and Terry Kinsella were married June 22.

Nicole Fauver (FA) and Rondo Fraley married on Aug. 20.

Belinda Bruster-Angel (FA) and Shan Angel's daughter, Natalie, was born March 28. Natalie joins siblings Cobin, 10, and Emily, 5

Mom Creates 'Michael's Army' to Raise Funds for FARF

Barbara, Michael and Robert Capone

By Barbara Capone

The day was July 23, 2010—the day that forever changed my life and the very being I once was. That was the day I found out that my son Michael had Fanconi anemia. Even though I also have two daughters, Alisha and Samantha, my life seemed to come to an end. It took me several months to let everything sink in and learn as much as I could about Michael's FA before I could even think about facing the world. I admit that even though I initially resisted medication, I had no choice but to turn to a psychiatrist and medication.

Once I was able to pull myself together, I jumped into action. I was not going to let this disease take my son! I had a much greater understanding of how desperately funds were needed because FA was considered an orphan disease and as such received little to no federal assistance. I decided to stop at nothing until we find a cure. Despite having no fundraising experience, I was able to draw on my accounting background and started "Michael's Army" to raise funds to find a cure.

My first fundraiser was a beef and beer event, which was a big success and raised more than $40,000. I reached out to family, friends, co-workers and local businesses for donations. I mailed letters, made phone calls, used Facebook and even walked into local businesses. I approached anyone who would listen. Much to my amazement, many people not only donated merchandise, but also made financial contributions. We had an abundance of items and sold tickets for silent, live and tombola auctions. We had silicone bracelets made with "Michael's Army: We are FAmily" embossed on them. Every person who attended received one as a reminder of the cause.

With that success under my belt, I began thinking of ways to raise funds and awareness the following year. After much thought, I decided to hold Michael's Army Golf Tournament for FA. Despite having little knowledge of golf, I relied on the expertise of friends and family again. In addition to the efforts that worked last year, I contacted airlines, cruise ships, hotels… you name it and they were contacted. Much to my surprise, I received two airline tickets from American Airlines, a weekend stay at the Hyatt Regency in Jacksonville, Fla., event tickets and specialty baskets. We also received many other donations including of hotel stays, wine tastings, beer kegs for the golf course, snacks and water bottles. The event was a huge success and raised more than $30,000.

We are already planning next year's event, "Be the Match." We want to find more donors for people who need transplants and don't have a match. We were shocked to learn that a lot of people in the donor pool back out when they are called upon for varying reasons. We will also hold events to raise funds for FARF; we just haven't established the venues yet.

I couldn't move forward in my life if it weren't for the support of my family and friends. They keep me going when I'm down and remind me to never give up! Additionally, I have learned how much the generosity of others means to me and it has given me a much better perspective. I realized I couldn't do this alone. The bottom line is that every little bit of money raised brings us closer to finding a cure for families afflicted with FA. ■

Sisters Alisha and Samantha support the cause.

Girl Scouts Make a Difference for FA

Haley and Amanda with Sean McQueen

By Lorraine McQueen

For those of you who thought the Girl Scouts just sold cookies, I'd like to tell you about two very special girls in Troop 791 who do much more and truly (as stated in the Girl Scout motto) "make the world a better place."

Eleven-year-olds Amanda Pendelton and Haley Barefoot from Midlothian, Va., recently initiated, organized and executed a bike-a-thon to raise money for the Fanconi Anemia Research Fund. After learning about FA from their parents, who attended our Band, Brew and BBQ for FA event, the girls decided they wanted to help in the search for a cure.

They first went door to door in their neighborhood soliciting pledges and donations. The pair then hopped on their bikes and rode 8.5 miles through a local park. Their efforts raised $500 for the Fund! We recently had the pleasure of meeting Amanda and Haley and were able to thank them for their hard work.

> ...two very special girls in Troop 791 who truly (as stated in the Girl Scout motto) "make the world a better place."

These girls truly exemplify the spirit of the Girl Scout law since they have been "considerate and caring, courageous and strong." Girls, the FA community would like you to know that we truly appreciate your good turn for FA. Keep up the good work! By the way, we still love your cookies! ■

Planned Gifts Another Option for Giving to the Fanconi Anemia Research Fund

Gifts to charitable organizations like the Fanconi Anemia Research Fund can be made either during one's lifetime or at the time of death through a will or trust. All charitable gifts, whenever they are made, can provide advantages to the donor that may include tax savings as well as the personal satisfaction of contributing to a worthy cause.

There are a number of ways to make a charitable gift during one's lifetime, for example:

- Transfers of cash or appreciated securities or real property
- Transfers from an individual retirement account (subject to statutory limitations)
- Use of a charitable lead or remainder trust
- Charitable gift annuity

Examples of charitable gifts made after death include:

- Direct cash bequests or a bequest of a specific asset to a charity
- Naming a charity as the designated beneficiary of an IRA or other retirement plan
- Creation of a charitable remainder or charitable lead trust for the benefit of family members and the charity
- Charitable gift annuity
- A percentage of the entire estate

Regardless of the type of planned gift, it is essential that you involve a tax advisor and/or estate planning attorney to ensure accuracy and adherence to related state and federal law. Go to www.fanconi.org for more information. ■

Family Fundraising Efforts January Through September 2012

From Jan. 1, 2012 through Sept. 30, 2012, Fanconi anemia families raised $770,571 for the Fanconi Anemia Research Fund. Thank you for your fundraising efforts so far this year. Remember that the Fund's staff is available to assist you with your holiday fundraising.

$170,000 and up
Kendall & Taylor Atkinson Foundation with the Nash and Atkinson families

$100,000 - $119,999
Dave, Lynn and Amy Frohnmayer
Peg Padden
Glen Shearer

$50,000 - $79,999
Kevin and Lorraine McQueen
Chris and Susan Collins

$30,000 - $39,999
Robert and Barbara Capone
Steve and Jennifer Klimkiewicz/Wyatt's Warriors

$10,000 - $19,999
Kerrie Brannock
Mike and Tracey Brannock
Kaps for Kendall

$5,000 - $9,999
Patti Carter
Mary Eilleen Cleary
Brian Horrigan and Amy Levine
Charles and Katy Hull
Jack and Lisa Nash
Mark and Diane Pearl
Mark Ritchie and Lisa Mingo

$1,000 - $4,999
Peter and Donna Abramov
John and Audrey Barrow
Mark and Linda Baumiller
Israel and Mary Jo Becerra
Darryl Blecher and Diana Fitch
Randy and Nancy Bloxom
Jeffrey and Donna Boggs
Richard and Tena Boson
Chris and Jennifer Branov
Donald and Danielle Burkin
David and Kim Chew
Natalie Curry memorial gifts
Darrel and Kalani DeHaan
Brian and Jennifer Dorman
David and Kelly Dunnock
David and Mary Ann Fiaschetti
Ben and Stephanie Griggs
Alan and Rachel Grossman
Owen Hall and Margaret Kasting
Bob and Victoria Hathcock
Jeff and Beth Janock
Mark and Angela Lamm
Tim and Mary Ann Lana
Gregory and Lynnette Lowrimore
Tue Marker and Kirstine la Cour Rasmussen
Dan and Nikki McCarthy
Adam and Olivia Mindle
Ian and Tricia Mitchell
Tyler Morrison and Rachel Altmann
Ron and Fredi Norris
Fred and Nancy Nunes
Michael and Katharine Ormond
Peter and Janice Pless
Kevin and Katie Rogers
Stanley and Lisa Routh
Bob and Andrea Sacks
Bryan and Karen Siebenthal
Adam and Jennifer Stewart
William and Mary Underriner
Sean and Kristin Young

Up to $999
Michael and Jennifer Aggabao
Leighsa Anderson
Jimmy and Jenny Armentrout
Ken and Jeanne Atkinson
Yavin Atzmon and Sharon Harari
Jason and Robin Baldwin
Cherie Bank
Julie Barbier
Gerald Barbier
Tyren and Kelly Bennett
John and Francene Berglund
John and Elaine Beyer
James and Tracy Biby
Ryan and Becky Brinkmann
Merry Cable
Daniel and Melinda Coleman
John and Kim Connelly
Bill and Pat Danks
Tony and Phyllis Dellapenta
Wendy Delzell
James and Carol Dillon
Pat and Mary DiMarino
Antonino and Marie DiMercurio
Ed and Janice Duffy
Gene and Lynn Eddy
Ginger Eggers
Sharon Ellis
Billy Jo and Debbie Estep
James and Crystal Eubank
Curt and Crystal Fales
Doreen Flynn
Israel and Rivka Friedlander
Gary and Melody Ganz
Andrew and Jennifer Gough
David Guidara
Racquel Hanna-Purser
Marzban Hathiram and Dastur Mahazareen
Peter and Tara Himmelreich
Jeff Hoffman
Judy Hoffman
Mary Hopper
Christopher and Dana Lamb
Ernie and Nancy Landwehr
Eugene and Renee Lemmon
Todd and Kristin Levine
Larry and Gayle Licari
Bill and Jackie Lucarell
Roberta and Glenn Magill
Deane Marchbein and Stuart Cohen
Gil and Peggy McDaniel
Catherine McKeon
Paul and Brooke Miles
Jim and Holly Mirenda
Sydney and Betsy Moore
Tony and Lina Nahas
Louis and Virginia Napoles
Jack and Tammy Neal
Bob and Alice Nicholson
Robert and Mary Nori
Michael and Katherine O'Halloran
Joshua and Crystal Pepper
Derek and Ginger Persson
John and Dianne Ploetz
Michael and Kay Proctor
Lynn and Shirley Quilici
Pedro and Marina Ravelo
Paul and Rena Rice
Gail Richardson
Leonard and Jan Riley
Paul and Catherine Rodwell
Dov Rom
Les and Nancy Ross
Rick and Lynn Sablosky
Richard and Dolores Satterlee
Bill and Connie Schenone
Thomas and Brenda Seiford
Lillian Sherman
Ariel Sitzerman and Gabriela Bernal
Dan and Ashley Spradling
Alfons and Karin Staab
Greg and Brandi Stuart
Charles and Jennifer Sumrall
Steve and Melissa Turner
Bob and Stacy Van Singel
Mike and Beth Vangel
Joe and Wendy Vitiritto
Xiaoqing Wang and Ning Liu
Marc Weiner
Sandy Weiner
Michael and Kim Williams
Brenda Witherspoon
Victor Wizman

For the fundraising events calendar and helpful fundraising materials and tools, visit www.fanconi.org

New Fundraising Tools Are Now Available

Qgiv and Hobnob are new online fundraising tools available through the Fanconi Anemia Research Fund. Through Qgiv, we can accept online donations directly on our website. Hobnob offers people a customizable fundraising page for events, enabling online registrations and donations in advance and at the event. Please contact FARF for details on how Qgiv and Hobnob can enhance your fundraising! ■

Zack Blecher

UnFAzed: A Benefit Concert for Fanconi Anemia Research

On March 31, nine Pittsburgh bands turned out and tuned up to show their support for local musician Zack Blecher, 26, an adult with FA. Zack and his friends organized this musical fundraiser and publicized it on Facebook, raising $1,593 for the Fund.

Climb Every Mountain

Bill McCorey of Winterpark, Fla., doesn't have a child with FA, but his friends Kevin and Lorraine McQueen do (Sean, age 13). For the past five years, this friend in deed has been climbing mountains to raise money for FA research. This journey began when Bill established Your Rope Team and tackled the daunting Mt. Rainier in 2007. The intrepid climber sustained a serious fall during that expedition, but remained motivated to pursue his goals and inspired others to join Your Rope Team. In June, Bill and his sons, Bill III, 29, and Drew, 14, climbed to the 14,179-foot summit of Mt. Shasta. Now that's dedication! To date, Your Rope Team has raised $43,579 for the Fund.

Bill, Bill III and Drew McCorey

Eli Boson

Garage Sale

Spring cleaning took on a whole new meaning in May, when the Boson family of Lewiston, Idaho, decided to gather items to sell and put together a fundraiser for International FA Day. Eli, 3, FA, and sister, Sophia, 10, helped out. The sale netted $865 for the Fund.

Bunco for a Cure

Israel Becerra

When planning her first fundraising event, Mary Jo Becerra of Tulsa, Okla., decided to team up with a friend. Mary Jo's son Israel, 3, has FA and her friend's son has cystic fibrosis. The women put on a Bunco tournament at their church. During the event, the two mothers took to the stage to speak about their sons' illnesses. Mary Jo reported that the public speaking wasn't easy, but it was the highlight of the evening and really helped to raise awareness. Proceeds from the event were split between both worthy causes, generating more than $3,000 for the Fund. This first-time event was such a success that the women plan to do it again.

Play for FA

Wendy and David Delzell

Remember family game nights? Wendy Delzell of Westwood Hills, Kan., whose son David, 13, has FA, took this cozy classic one step further and made it a fundraising event. Admission was $5, with a maximum of $20 charged per family. Attendees played a variety of board and card games and dominoes while enjoying lemonade and cookies. The Fund was the big winner of this fun family event, reaping more than $550.

A Full Heart

Justin Barbier was a young adult with FA who passed away last year at the age of 26. Justin touched many lives and has not been forgotten. His friend April Leonard sold heart-shaped decals printed with Justin's name to raise funds for FA research. April sent this message along with the $550 she raised, "I look toward a brighter future with a cure for Fanconi anemia, and I look forward to the Justins of this world living longer, healthier lives."

Charitable Consignment

Jennifer Branov of Vancouver, British Columbia, had a bright idea while cleaning out her daughter's closet. Jennifer took 6-year-old Lea's outgrown items to a consignment shop, yielding $150. She donated the money to the Fund to help fund a cure for Lea's FA.

Lea Branov

The Economics of Philanthropy

Each year, the fifth grade class at Wickliffe Progressive School, Upper Arlington, Ohio, holds a marketplace as part of their study of economics. Proceeds are donated to a charity of the students' choice. This year, the class selected the Fanconi Anemia Research Fund and made a donation of more than $600. Wickliffe is attended by the siblings of Samantha "Sam" McCarthy who had FA and passed away in 2009.

Board of Directors Comings and Goings

Annette Waxberg

Brian Horrigan

The Fanconi Anemia Research Fund welcomed two new members to its board of directors in mid-2012: Annette Waxberg and Brian Horrigan.

Annette, an FA parent and Treasurer of Fanconi Canada, joined the board for a three-year term on June 1. She and her husband, Lorne Shelson, are the parents of Sarah, 21; Aaron, 19; and Livia, 15. Aaron has FA. The family lives in Toronto, Ontario.

Brian, also an FA parent, began a three-year term on Aug. 1. He is the Exhibits Curator at the Minnesota Historical Society in Minneapolis, Minn. He and his wife, Amy Levine, have two children—26-year-old Colin and 24-year-old Delia. Delia has FA.

Both Annette and Brian bring extensive professional and personal experience to the board. We are delighted to have them as part of the team at FARF.

The board said good-bye and thanks to Peg Padden earlier in the year. Peg served on the board for a number of years and provided sound counsel about fundraising and other key issues. Peg continues her role as a fundraising cheerleader on the e-group and Facebook, encouraging all families to join with her to raise funds for research. Thanks, Peg! ■

FARF Can Help You Fundraise

More than 90% of the Fanconi Anemia Research Fund's annual budget comes from family fundraising. We're here to help make your events a success. We can:

- Provide sample fundraising letters and help you edit your letter
- Use your photos to personalize your letter, event invitation or brochure
- Use your mailing list to send your letter or invitation from our office
- Provide ideas, information and display materials for events
- Provide a PowerPoint or video presentation to use at your events
- List your event on our website
- Send a thank-you letter and tax receipt to your donors

We ask that all fundraising events be covered by liability insurance. Insurance for a one-time event is often available through a family's homeowner's insurance policy as a relatively inexpensive insurance rider. Please contact the Fund if you need assistance obtaining or paying for this required insurance.

Please ask your donors to make checks payable to the Fanconi Anemia Research Fund. When a donation is received, we'll generate a letter of thanks with a tax receipt, and we'll notify you that a donation has been made in your name.

We appreciate all your efforts to raise funds for FA research and family support. You are making a difference! ■

SCC Fact Sheets Available

Regular screenings for oral cancer are critically important for FA patients. The Fund has helpful fact sheets about squamous cell carcinoma to share with your dentist and ear, nose and throat doctor (ENT). **We recommend that you take a fact sheet to every dentist and ENT visit.** Fact sheets—in English, Spanish, Afrikaans, Dutch, French, German, Hebrew and Italian—are available on our website or by calling our office.

Your FA Research Dollars at Work, January through September 2012

The Fund awarded $1,034,507 in research grants to the following projects between January and September 2012:

Investigator: Ruud Brakenhoff, PhD, Free University, Amsterdam, Netherlands
Title: *Targeted Treatment of Oral Cancer and Precancer in FA Patients*
Amount: $324,000

Investigator: Michael Spiotto, MD, PhD, University of Chicago, Chicago
Title: *Non-genotoxic Inhibitors for HPV-induced HNSCC in FA*
Amount: $198,910

Investigator: Dong Zhong, PhD, University of South Dakota, Vermillion, S.D.
Title: *Investigate the Molecular Mechanisms of How FANCJ Regulates Centrosome Cycle*
Amount: $186,545

Investigators: Robert Sclafani, PhD, University of Colorado, Denver, Colo.
Title: *Potential Therapeutic Use of Resveratrol for Head and Neck Carcinogenesis in FA*
Amount: $100,000

Investigators: Parinda Mehta, MD, Cincinnati Children's Hospital Medical Center, and Rachel Katzenellenbogen, MD, Seattle Children's Research Institute/University of Washington
Title: *Serology Biomarkers for Immune Response to HPV Vaccination and Exposure in FA*
Amount: $109,028

Investigators: Joel Greenberger, MD, and Hebist Berhane, University of Pittsburgh, Pittsburgh, Pa.
Title: *GS-Nitroxide (JP4-039)/F15 Liposome Oral Radioprotective Therapy for FA Patients Requiring Chemoradiotherapy for HNSCC*
Amount: $33,045

Investigator: Markus Grompe, MD, Oregon Health & Science University, Portland, Ore.
Title: *Testing Clinically Relevant Compounds in FA*
Amount: $83,817

Last but not least, in early September the Fund agreed to support Eunike Velleuer, MD, Heinrich Heine University, and Ralf Dietrich, Executive Director and Family Support Coordinator, German Fanconi Anemia Support Group, both from Duesseldorf, Germany, in their ongoing efforts to prevent oral cancer in FA patients for a cost of $198,506 for two years. Dr. Velleuer and Dietrich collect brush samples of the mouths of FA patients, provide one-on-one education about the prevention of head and neck cancer, collect and distribute oral cancer tumor samples to research groups around the world, and collaborate with David Wong, DMD, DMSc at UCLA by helping collect saliva samples. While the pair's work is more of a *service* than *research* project, the Fund's board of directors feels strongly that their efforts are critical to preventing oral cancers as well as to our understanding of the disease. ■

Fund Schedules Small Molecules and *Guidelines* Meetings

The Fanconi Anemia Research Fund is sponsoring two important meetings in the next six months. The first meeting, Small Molecules in Fanconi Anemia, will be held in Portland, Ore., in early November. Approximately 35 researchers and clinicians, most actively working on various small molecules, or drug compounds, for FA will gather for a day and a half to discuss the following three topics:

1. What small molecules might modify the FA cellular phenotype *in vitro* or *in vivo*?
2. What insights might guide subsequent work?
3. What is needed to move promising agents to clinical trials?

The second meeting, scheduled for April 2013, is a consensus conference to develop the fourth edition of *Fanconi Anemia: Guidelines for Diagnosis and Management*. Committee work for each of the main chapters began earlier in the fall. We hope to publish the new edition in the first quarter of 2014.

Stay tuned for updates from both meetings in future editions of the FA Family Newsletter as well as on Facebook and the e-group. ■

Fanconi Anemia
RESEARCH FUND, INC.

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RETURN SERVICE REQUESTED

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

Use of Logo

A reminder to our families with FA: Please use our logo or letterhead only after you have consulted staff at the Fanconi Anemia Research Fund and received approval. This step is necessary to be sure our messages are accurate and consistent and it helps avoid legal complications. We are happy to collaborate on fundraisers and mailings.

Editors' Note and Disclaimer

Statements and opinions expressed in this newsletter are those of the authors and not necessarily those of the editors or the Fanconi Anemia Research Fund. Information provided in this newsletter about medications, treatments or products should not be construed as medical instruction or scientific endorsement. *Always consult your physician before taking any action based on this information.*

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Donations Online:	You can donate via the heart button on the Fund's website (www.fanconi.org) or through www.networkforgood.org or www.paypal.com
Donations by Phone:	Call us at 541-687-4658 or toll free at 888-FANCONI (888-326-2664) (USA only)
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