Oct. 2010

FA Family Newsletter

A semi-annual publication of the Fanconi Anemia Research Fund, Inc.

issue 48

Science and Medical News

The Fund Sponsors Meeting on Squamous Cell Carcinoma

The Fanconi Anemia Research Fund sponsored a meeting in April 2010 on squamous cell carcinoma (SCC) in Fanconi anemia patients, an area of increasing concern. Approximately 35 physicians and researchers, plus members of the Fund's Scientific Advisory Board and Board of Directors, attended the two-day meeting. Philip Rosenberg, PhD, US National Institutes of Health, presented compelling evidence concerning the relatively high incidence of SCC. Ralf Dietrich, the Executive Director of the German Family Support Group; Eunike Velleuer, pediatric oncologist; and Alfred Boecking, MD, Heinrich-Heine University, Düsseldorf, discussed the collection and analysis of oral brush samples of 349 patients. Meeting participants divided into four small groups to work on the following topics:

- Screening for cancer and the importance of prevention
- Carcinogenesis: differences between FA and non-FA patients
- Carcinogenesis: location of FA cancers and the role of HPV
- Clinical Trials: design and agents

Each group generated a draft written report with specific recommendations. Those reports will be circulated among the meeting participants this fall and a decision will be made on the need for, and timing of, a follow-up meeting. One concrete result of the meeting so far: Erich Sturgis, MD, a member of the Fund's Scientific Advisory Board, informed the group that an ongoing clinical trial at MD Anderson Cancer Center for erlotinib (Tarceva) would be opened to FA patients. See related article on page two. ■

John Hanna, an FA patient with SCC, reflects on his lifestyle choices related to head and neck cancer:

" I take great pride, and have a sense of relief, in knowing that I did nothing to speed up this cancer. It makes it easier to live with. I would be so angry at myself if I had done things like smoking and drinking knowing full well that I'd possibly brought this on myself. I'm fortunate that I had excellent role models in my parents. As kids and adults if we see our parents doing these things we will most likely end up doing them as well."

INSIDE

Science & Medical News.... 1-3
Science News from Family Meeting 4-11
Family News..... 12-17
Fundraising..... 18-22
Of Note 23

National Institutes of Health Grant Expands 15-Year Effort Initiated by the Fund to Prevent Cancer

At a Fanconi Anemia Research Fund workshop in 2007, participants agreed to collaborate on an effort to identify and test drugs that could potentially prevent complications associated with Fanconi anemia. Donations to the Fund provided nearly $650,000 to pursue ideas developed at the workshop. Now, a new $10.7 million grant from the National Heart, Lung, and Blood Institute, a branch of the US National Institutes of Health, will expedite research into new and existing drugs and compounds. The research teams are based at Oregon Health & Science University, the University of Oregon and Harvard Medical School. They will screen thousands of drug candidates in mice (OHSU), zebra fish (UO) and human cell lines (Harvard).

A $10.7 million grant from the NIH will expedite research into new and existing drugs and compounds.

Many of the drug candidates to be screened have already been approved by the US Food and Drug Administration (FDA) to treat other medical conditions. This prior FDA approval could speed applications in FA. Another 10,000 molecules randomly modified for various biological reactions also will be screened for therapeutic benefits. ■

THE UNIVERSITY OF TEXAS
MD Anderson Cancer Center

Clinical Trial Availability

Fanconi anemia patients with head and neck cancer who are candidates for surgery may be eligible to participate in a clinical trial at the MD Anderson Cancer Center in Houston, Texas. The trial uses erlotinib (brand name Tarceva), a drug that works by blocking the action of an abnormal protein that otherwise signals cancer cells to multiply. A trial of this drug precedes surgery; after surgery a tumor sample is analyzed for changes in the cancer cells in response to the drug. Contact William N. William, Jr., MD, at wnwillia@mdanderson.org or Raquel Ayuste, RN, at 713-792-6363 for more information. ■

More on SCC...

"For an adult with FA, the probability of an SCC occurring is about 1 or 2 percent per year. Most of these tumors appear to arise from precursor lesions, so although the risk adds up over time, there appear to be promising options in terms of early diagnosis and treatments."

—Philip Rosenberg, PhD.,
National Institutes of Health

Researchers Discover a 14th Gene Linked to FA

FARF-funded researchers in Germany and England, led by Helmut Hanenberg, MD, Chris Mathew, PhD, and Detlev Schindler, MD, have discovered a 14th gene linked to Fanconi anemia, named RAD51C. Research revealed that mutations in this gene may also lead to an increased risk of breast and ovarian cancers. This discovery is the latest in a long line of important findings yielded by 20 years of research supported by the Fanconi Anemia Research Fund. ■

Current FARF-funded Research

Following is a list of FA research projects that are currently receiving (or will receive) support from the Fund. Several new grant applications are under review and may be added to this list for support in the near future.

Farid Boulad, MD
MultiCenter Pilot Trial of HSCT Lacking a Genotype Identical Donor (A collaborative study involving Memorial Sloan-Kettering Cancer Center, Cincinnati Children's Hospital Medical Center, Children's Hospital of Wisconsin and Boston Children's Hospital)

Philip Connell, MD
Restoration of Homologous Recombination in Fanconi Anemia

Neelam Giri, MD
Studies of Immune Function in Patients with Fanconi Anemia

Majlinda Lako, MD
Using iPSC Technology to Understand Early Hematopoietic Development in FA Patients

Susan Mallery, DDS, PhD
Mucoadhesive Patch Delivery of Fenretinide and Berry Anthocyanins for Oral Cancer Chemoprevention

K.J. Patel, MD, PhD
Reconstituting and Dissecting Monoubiquitination in the FA Tumor Suppressor Pathway

John Postlethwait, PhD
Screening for Therapeutics in Fanconi Anemia

Susanne Wells, PhD
HPV Replication and Transformation in FA Squamous Cell Carcinomas

Stephen West, PhD & Andrew Dean, PhD
Coordination of Fanconi Anemia and Bloom Syndrome Complexes by FANCM

Feng-Chun Yang, MD, PhD
Investigating the Role of Microenvironment in the Development of Bone Marrow Failure in Fanconi Anemia

Lynn and Dave Frohnmayer with Andreas Fanconi, MD, son of Guido Fanconi, MD, the physician who first identified the genetic disorder that bears his name. The Frohnmayers, Dr. Fanconi and others in the worldwide Fanconi anemia research community gathered at the University of Würzburg in Germany last May for a scientific meeting on the occasion of the 80th birthday of Traute Schroeder-Kurth, MD. Dr. Schroeder-Kurth was a pioneer in the study of FA, the first to describe the "formal genetics" of FA and to show that FA caused chromosome abnormalities. ■

Top 10 Takeaway Messages from the 2010 Family Meeting at Camp Sunshine

1. Seek information and advocate for yourself or your child.
2. When considering a BMT at a particular center, ask how many FA transplants are performed annually and what the current outcome data are, particularly for unrelated FA BMTs.
3. Inspect the mouth twice a year beginning at age 9 or 10. If any suspicious lesions are present, increase screening to every six weeks and seek a biopsy.
4. Avoid alcohol and tobacco, including secondary smoke and oral products with alcohol.
5. Maintain good oral hygiene. Brush and floss!
6. Consider vaccinating against HPV at age 6 for both boys and girls.
7. Test at a young age for hearing loss.
8. See a gastroenterologist if the following symptoms persist: weight loss, decrease in growth, blood loss, significant vomiting, daily multiple bowel movements, diarrhea, and pain under right ribs or in right lower quadrant.
9. Consider donating tissue samples through NDRI as a way to help move FA research and therapies forward.
10. Stay vigilant! Complications can arise relatively quickly and without warning. ■

Parents and FA Patients Must Be Strong Advocates

A huge gamut of medical issues can accompany the diagnosis of Fanconi anemia. Parents of children with FA and adults with FA must be their own best advocates according to Eva Guinan, MD, Dana-Farber Cancer Institute, Boston, Mass.

Dr. Guinan reviewed a long list of underlying and acquired complications that can confront individuals with FA, including iron overload caused by transfusions; hearing loss related to structural anomalies; endocrine and GI nutrition problems; drug interactions; and living with the threat of cancer, which she likened to "the ever-present light of an oncoming train." Adults with FA generally transition to adult physicians who may know very little about FA. When possible, a multidisciplinary medical center is "the way to go," and a program that integrates local FA experts with adult care providers is a real plus.

Dr. Guinan concluded that parents and patients alike have an enormous responsibility to understand the various ramifications of this disorder, be alert to new studies, educate caregivers and advocate strongly for their own needs. She acknowledged that no one should have to shoulder so much, which is in many ways "not fair." Dealing with this complicated disorder is very "nerve wracking"—but sometimes there is simply no choice. ■

Take home message: Be a very educated advocate and consumer every step of the way.

Minnesota Transplant Outcomes

Over more than a decade, the University of Minnesota transplant program led by John Wagner, MD, has implemented a series of protocols in an effort to improve outcomes. The number of transplanted patients continues to grow while survival remains high.

Dr. Wagner reports that his center has now transplanted 201 Fanconi anemia patients. All 26 pediatric patients with matched sibling donors on the present non-radiation protocol survive, with no GvHD.

Unrelated donor transplants are more challenging, since these patients face greater resistance to engraftment, elevated risk of GvHD and greater delay in immune recovery. To date, 35 patients have been transplanted with thymic shielding and reduced radiation of 300 rads. Survival is 89%.

Minnesota has transplanted seven "high risk" FA patients, which Dr. Wagner defines as patients with kidney failure, infections prior to transplant, leukemia, or age 35 or older. These "high risk" patients were given busulfan instead of radiation. Of these seven patients, four survive.

Quality of life issues post transplant can be complicated by factors such as bone necrosis necessitating hip replacement, infection and the need for insulin therapy. Dr. Wagner reported that only three of 169 Minnesota patients have had cancer post-transplant, occurring at ages 20, 35 and 46. Fifteen years after transplant, cancer risk is 2% at this center.

Future directions include use of mesenchymal stem cells to prevent GvHD and to hasten engraftment. ■

Transplant Outcomes Data from Cincinnati and New York

When faced with the difficult decision to undergo a bone marrow transplant (BMT), the Fund encourages all FA families to contact the various medical centers that perform FA BMTs and inquire about outcomes data. The Fund does not endorse one center or physician over another.

Cincinnati Children's Hospital Medical Center[1]:

Time period Jan. 1, 2006 to Aug. 31, 2009

Type of BMT	Total Patients	Alive 1 Year	Survival Rate
Matched Sibling Donor	4	4	100%
Unmatched cord blood	4	3	75%
Unmatched donor[2]	16	10	63%
TOTAL	24	17	71%

[1]The total number of FA BMTs performed at CCHMC is 123.
[2]Data include unrelated bone marrow and cases with overt leukemia as well as more favorable cases.

Memorial Sloan-Kettering Cancer Center:

Time period Jan. 1, 2006 to Aug. 31, 2009

Type of BMT	Total Patients	Alive 1 Year	Survival Rate
Matched Sibling Donor	1	1	100%
Unmatched cord blood	1	1	100%
Unmatched donor[1]	10	5	50%
TOTAL	12	7	58%

[1]Data include unrelated bone marrow and mismatched related bone marrow transplants. Seven of the 10 patients had MDS or AML; four were older than 18 years of age (including two who were 35); three were very high risk coming into transplant.

Oral Brush Samples Reveal Progression to Cancer of the Oral Cavity

Between 2006 and 2010, Eunike Velleuer, MD, Children's Hospital, Heinrich Heine University Medical Center, Düsseldorf, Germany and Ralf Dietrich, Executive Director, German Family Support Group, collected brush samples from the oral cavities of 365 patients with Fanconi anemia for a study led by Ruud Brakenhoff, PhD, Free University, Amsterdam. Dr. Velleuer presented their findings at our Family Meeting at Camp Sunshine.

Cytological testing of the brush samples, performed by Dr. Alfred Böcking, Düsseldorf, revealed 12 malignant tumors from 10 separate patients. Not one of the tumors had been previously recognized. Two of the 10 patients previously had oral cancer, but were not monitored regularly after tumor removal. Eight patients had never been diagnosed with cancer.

Dr. Velleuer stated that all persons with FA should do four things to help prevent oral cavity cancers:

1. Get information! "If you close your eyes, you can't see the enemy."
2. Undergo regular inspection, including self-inspection and evaluation by an ear, nose and throat (ENT) physician. Document areas of concern and ask your ENT to take pictures of possible lesions for later comparison.
3. Avoid ALL alcohol and tobacco, including secondary smoke. Try to avoid anything that causes irritation to the tissues of the mouth.
4. Maintain good oral hygiene.

Brush sample screening is also useful in identifying precancerous changes that occur over time in the lining of the mouth cavity. Loss of heterozygosity (LOH) means that a patient who has already lost the function of one allele of a gene has now lost the function of the second allele as well. Dr. Brakenhoff and Dr. Velleuer believe that patients who progress to malignancy will first exhibit specific areas of LOH. Screening and testing focus attention on high-risk areas and help identify very early malignancies. ■

Oral Bacteria — a Possible Diagnostic Screen for Oral Cancer

Flavia Teles, DDS, MS, DMSc, The Forsyth Institute, Boston, informed Family Meeting attendees that bacteria are sometimes associated with different cancers. Elevated levels of common oral bacteria have been found, for example, in esophageal cancer lesions. It appears that bacteria may contribute to carcinogenesis by activating carcinogenic chemicals or by causing infection and inflammation.

Studies conducted at Forsyth have shown that the mouth can harbor more than 700 types of bacteria, some of which may be associated with cancer. Forsyth scientists have observed that non-Fanconi anemia patients with oral squamous cell carcinoma (SCC) have elevated salivary levels of specific bacteria. Thus, saliva might be a useful non-invasive tool for the diagnosis of oral cancer.

Dr. Teles plans to characterize the oral bacterial profiles of FA patients, their non-FA siblings and parents, and non-FA oral SCC patients. She will determine the most prominent bacteria in the mouths of individuals with FA and investigate whether these bacteria induce infection, inflammation or activate carcinogenic chemicals. She will also compare the bacterial profiles found in FA patients and in non-FA SCC patients. She hopes this study will lead to an early diagnostic test for patients at greatest risk of developing head and neck cancer.

Dr. Teles obtained saliva samples from 18 individuals with FA and from eight non-FA family members in attendance at Camp Sunshine. ■

Treatable Hearing Loss is Common in FA Patients

Earlier medical articles under-reported the incidence of hearing loss in FA patients, according to H. Jeffrey Kim, MD, National Institute on Deafness and Other Communication Disorders, National Institutes of Health (NIH). The NIH is conducting a multi-disciplinary study of FA patients that shows that this problem is far more prevalent in FA patients than previously believed.

From 2003 to 2010, physicians evaluated ears of 28 patients. All patients received a comprehensive ear exam, audiologic evaluation and an imaging study (computer tomography).

Of the 49 ears studied, 39% had hearing loss. Most were mild (29%), but 10% had more severe hearing loss. The most common problem was conductive hearing loss.

Fifty-five percent of the ears showed middle ear abnormalities. Forty-five percent had bony plates on the eardrum; 30% had grossly small eardrums. These malformations are congenital and probably result from abnormal embryonic development. A long history of chronic ear infections, iron chelating agents and IV antibiotics can also lessen a child's capacity to hear.

Children with mild to moderate conductive hearing loss experience difficulty understanding when there is background noise, have difficulty hearing certain sounds and have trouble with pronunciation. When not corrected, these issues can affect language development and performance at school or work.

For all FA-affected individuals, Dr. Kim highly recommends early detection and intervention for hearing loss. Options ranging from hearing aids and listening devices to ear surgery can greatly improve hearing. ■

A Precious Gift: the Donation of Tissue for Research

Researchers often depend on tissue samples to understand a disease and to devise new therapies specific to that particular disease. The National Disease Research Interchange (NDRI), funded by the National Institutes of Health, collects and preserves human tissues for distribution to researchers. Daniel Remer, Rare Disease Program Manager, NDRI, says that 10 individuals with Fanconi anemia have donated 48 tissue samples to NDRI. Six of those individuals donated tumors. The tumors were divided into 21 samples, and 16 have been shipped to FA researchers. The remaining five are stored in NDRI's biorepository for later distribution.

Individuals can sign up at any time to be a donor, even if the donation comes at a much later date. For patients facing removal of a tumor, the stress of surgery can make it very difficult to remember to donate tissue. Yet this gift can be invaluable in helping to find useful therapies. Inform your physician and family members of your intention and sign up early.

To learn more about NDRI or how to become a tissue donor, visit www.ndriresource.org or contact Daniel Remer at dremer@ndriresource.org. ■

Just Three Steps Required to Donate Tissue Samples to NDRI

There are just three easy steps to donate blood, bone marrow or tumor tissue samples to the National Disease Research Interchange:

Before the biopsy/surgery is scheduled:

1. Contact Tyler Musselman at NDRI: 800-222-6374, ext. 268
2. Contact Teresa Kennedy at the Fund: 800-326-2664
3. Complete NDRI consent and paperwork.

NDRI does the rest by coordinating with your physician, pathologist and FA researchers.

—Thank you!

Human Papillomavirus and FA: Evidence Builds for a Link

Susanne Wells, PhD, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio, discussed the possible link between the human papillomavirus (HPV) and the cancers that affect persons with FA. Dr. Wells noted that approximately one-fifth of all cancers diagnosed each year in the general population are caused by viruses, and HPV is responsible for 500,000 of these.

HPV infection contributes to cancers by improving cancer cell survival and causing malignant cells to grow faster. In the general population, HPV causes almost all cervical cancers and many cancers of the vulva, vagina, anus, penis, and head and neck. Based on laboratory studies, Dr. Wells believes that HPV infection might increase the susceptibility to cancer in persons with FA.

HPV is usually transmitted through sexual contact. However, there is evidence that HPV is found in children. In a US study, HPV was detected in 6% of healthy children. HPV was found in the mouths of 42% of healthy children in a Finnish study and around 52% of prepubertal children in a United Kingdom-based study. Variance in sensitivity of tests explains these very different numbers.

With assistance from FARF, Dr. Wells studied saliva samples from 55 FA patients and 54 normal controls. She hoped to determine if HPV is more common in FA and how HPV might alter cancer risk in FA patients. Her preliminary conclusions are that HPV is present in a greater proportion of FA individuals over controls and that younger patients are affected. Nine of 55 individuals with FA were HPV-positive and four of those were under the age of 12 (two were six years old), compared to two normal controls positive for HPV, ages 13 and 18.

Dr. Wells' ongoing studies focus on the mechanisms by which HPV might alter cancer risk in persons with FA and how HPV infection can be prevented and treated in this at-risk population. ■

Physical and Psychological Complications Post-Transplant

Eva Guinan, MD, Dana-Farber Cancer Institute, reminded attendees of the Family Meeting at Camp Sunshine that FA is forever and "a lot of baggage comes with it," both physical and psychological. A bone marrow transplant won't take care of all the problems associated with FA and can often create new issues. She discussed a range of complications that can be caused by the transplant itself.

Agents used to eliminate a patient's own marrow and immune system, such as radiation and busulfan, can cause regimen-related toxicities. Dr. Guinan observed that busulfan is not uniformly less toxic than radiation. Additional agents that suppress the patient's immune system can cause kidney and blood pressure problems.

One of the biggest concerns post-transplant is graft-versus-host disease, which can be acute (aGvHD), chronic (cGvHD) or both. Transplantation requires the patient to re-constitute an immune system, leading to this serious complication. Donor cells attacking the host cause both forms of GvHD, but cGvHD is more like an autoimmune disease, in which one's immune system attacks various parts of the body. The skin, liver, gut and lungs can all be affected. There are no completely satisfactory therapies for cGvHD. This complication can predispose to malignancy, lead to scarring of the lungs (a chronic cough or poor exercise tolerance can suggest scarring) and can be fatal.

Other post-transplant complications include endocrine disorders (growth impairment, thyroid problems, delayed puberty); bone problems including osteoporosis and avascular necrosis (bone death associated with steroid exposure, caused by an abnormal flow of blood to the bone). Bone pain, not always experienced at the exact location of bone loss, may suggest avascular necrosis.

Managing these sometimes chronic medical demands places an enormous psychological burden on patients and caregivers alike. Given the tendency to emphasize transplantation as a curative therapy for bone marrow disease, one can easily overlook the many complications that can ensue. ■

Diagnosing and Treating Chronic Abdominal Pain

Individuals with FA often suffer from a variety of digestive problems, including poor appetite, failure to thrive and abdominal pain. Sarah Jane Schwarzenberg, MD, University of Minnesota, focused her talk this year on the diagnosis and treatment of chronic abdominal pain.

She defined chronic pain as lasting at least 12 weeks and noted that the pain is not necessarily continuous. She discussed three conditions that can account for chronic abdominal pain:

1. Functional dyspepsia, or pain in the upper part of the abdomen that can't be diagnosed by endoscopy or changes in the stool. This disorder does not respond to medications for reflux (such as Prilosec) and is diagnosed by eliminating other disorders.
2. Irritable bowel syndrome (IBS) is characterized by mucous in the stool, a bloated feeling and distended abdomen. Changes in the frequency or appearance of the stool can suggest this disorder, and producing a stool provides relief.
3. Functional abdominal pain (FAP) is nearly continuous pain in the stomach around the navel. Producing a stool usually does not provide relief.

Patients with chronic abdominal pain need to undergo a thorough physical exam with complete history. Tests for erythrocyte sedimentation rate (ESR), for parasite eggs in the stool, an endoscopy of the stomach and intestine and, in females, ruling out a urinary tract infection are appropriate diagnostic measures. This work-up will eliminate more serious causes of chronic abdominal pain.

Symptoms which cause alarm are loss of weight, decrease in growth, blood loss, significant vomiting (more than once/day), greater than five to six stools a day, diarrhea, and pain under right ribs or in right lower quadrant (which could suggest gall bladder disease or appendicitis). A patient with severe abdominal pain from a family with a history of inflammatory bowel disease warrants immediate medical attention.

There are few good therapies for chronic abdominal pain, and therapies appropriate for the general population may not be advised for individuals with FA. Coated capsules containing peppermint oil may be effective in irritable bowel syndrome. Drugs that prevent the release of acid in the stomach and intestine may be useful. Dr. Schwarzenberg recommends proton pump inhibitors such as Prilosec, but states that H2 antagonists like Pepcid (famotidine) or Zantac (ranitidine) are not appropriate for FA patients, as these drugs may suppress bone marrow. Zophran (ondansatron) is effective in treating nausea.

Chronic abdominal pain curtails a patient's functioning and can be enormously challenging for the entire family. Working with a skilled gastroenterologist is crucial in attempting to identify the cause and appropriate therapy for this multi-faceted disorder. ■

Connect with the Fund Online

The Fanconi Anemia Research Fund now has a re-designed website. The revamped site features a new "Global Partners" section, providing links to informal international partner organizations, an easy-to-complete mailing list form and all the educational materials and support information featured on the Fund's former website.

www.fanconi.org

Please also find the Fund on Facebook! Go to www.facebook.com/fanconianemiaresearchfund for links to the latest scientific articles about FA and stories about families affected by the disease.

Most important, if you're a parent of a child with FA, or an adult with FA and would like to join our online support group, please email teresa@fanconi.org.

Head and Neck Cancer—Prevention, Screening and Treatment

Dr. Bhuvanesh Singh, Memorial Sloan-Kettering Cancer Center, New York, noted that head and neck squamous cell carcinoma (HNSCC) occurs in persons with FA about 500 times more often than in the general population. It appears at a much earlier age and presents unique therapeutic challenges. HNSCC is a progressive disease and often evolves from normal tissue to hyperplasia, dysplasia, carcinoma in situ and finally invasive cancer. Prevention and early detection are imperative, given the difficulty of treating this life-threatening complication.

Prevention Strategies

- Abstain completely from the use of tobacco and extended exposure to second-hand smoke. If parents or friends smoke, don't be in their presence when they do. This is an absolute imperative!
- Abstain from use of alcohol. Dr. Singh states that one drink, once a month, plus a drink on one's birthday or New Year's Eve should be the absolute limit.
- Consider the HPV vaccine at an earlier age than the federal guidelines suggest. Several recent studies find that some FA patients have been found to be HPV-positive as young as age 6 and that the mode of transmission is not necessarily sexual, as is true of the general population. HPV is associated with 26% of the head and neck cancers in the general population and probably plays an important role in FA head and neck cancers as well.

Screening Strategies

- Beginning at age 9 or 10, children with FA should be screened twice a year by a well-trained head and neck physician. If any abnormal areas appear, screening should increase to every six weeks. Experts need to biopsy areas of concern.

Treatment

- If cancer is diagnosed, patients should be treated at a major center with an experienced team of experts. Small centers may have excellent individual physicians but lack a team approach, which FA patients need. Surgery is the mainstay of treatment, because radiation and chemotherapy are especially toxic for FA patients.
- Surveillance following surgery should be at least every three months. The risk of a secondary cancer is 30% in the general population, but over 60% in FA patients.
- Patients diagnosed with HNSSC need a chest x-ray to rule out lung metastasis. Diagnostic chest x-rays and dental x-rays do not cause cancer. ■

facebook

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

Drs. Blanche Alter, Pamela Stratton and Susan Rose also presented at the 2010 Family Meeting. Copies of their presentations on cancer epidemiology (Alter), gynecologic issues (Stratton) and endocrine issues (Rose) are available in the annual Family Meeting section of the Fund's website.

DNA Testing for FA Available Before Pregnancy

By Christina Lavin, MS, CGC, Reproductive Genetics Institute

Preimplantation genetic diagnosis (PGD) offers families at risk for a genetic disease like Fanconi anemia the option of DNA testing for FA and, in many cases, human leukocyte antigen (HLA) testing, prior to pregnancy.

The process involves in vitro fertilization (IVF), during which the female's ovaries are encouraged to produce multiple eggs. These eggs are removed prior to ovulation, fertilized in the laboratory and tested by single cell analysis for the FA mutation(s) and HLA. Only embryos identified as unaffected by FA will be recommended for embryo transfer. Embryo transfer occurs approximately five days following egg retrieval.

PGD testing has been an option for FA for more than 10 years, and Reproductive Genetics Institute (RGI) has helped 17 families at risk for FA try to expand their families. As of May 2010, a total of 51 cycles of PGD

5K/8K Run/Walk! Registration for the meeting is free and travel scholarships will be available. Registration opens in November. Contact Teresa Kennedy at teresa@fanconi.org or 1-888-FANCONI for more information or to register.

FA Family Encourages Fundraising Efforts

By Mike Vangel

On September 6, 2010, we celebrated the eighth anniversary of our daughter Amy's bone marrow transplant. It is hard to believe so much time has gone by. We feel very blessed she has done so well.

Our son, Dennis, has FA, too. He is now in the eighth grade. Because of FA he is growth hormone-deficient, tires easily and his immune system is not as strong as it should be.

He had a bone marrow biopsy procedure in March. Dennis' bone marrow is beginning to fail and his blood counts are getting weaker. Because Amy's bone marrow failed, we know what it's like to go through a bone marrow transplant and worry for him.

He knows, too, what his sister went through when she received a bone marrow transplant and is concerned. He doesn't like to talk about it, but we know it hangs heavily on him.

In December 2009, Amy had pre-cancerous lesions removed from inside her mouth. There were many and they could not remove them all. The surgeon removed what she could. Malignancies are quite common in young adults with FA and there is little that can be done other than close monitoring and surgery to remove the tumors when they develop. It's a scary way for a young woman and a young boy to have to live each day of their lives.

Amy's health remains relatively stable and most of the time she experiences joy in the beauty around her and in the warm friendships she nurtures every day. Her ability to live in the moment and treasure each and every day she is given is amazing and truly inspiring.

Dennis loves school and tries never to miss classes even though he usually doesn't feel that well. He eagerly wakes up every morning to get to school as early as he can. He enjoyed playing in the Middle School Jazz Band. He turns to his guitar for expression and escape from worry.

Like Amy and Dennis, we worry a lot about their health, and the serious health issues experienced by so many young FA adults and children. When Peg Padden came up with the idea of an International Fanconi Anemia Day, we thought we would join in and do a letter-writing campaign to help raise money to fund Fanconi anemia research. With a job change and the slump in the economy, it had been a couple of years since we had last reached out. The hardest part was just committing to do it and then everything else fell into place. The staff at FARF provided us with a list of folks who had donated on our behalf in the past and we added to the list, too. They provided us with some examples of other FA families' fundraising letters which made writing our own family's letter much easier. Although we mailed the letters on our own, they offered to do that for us, too. Four months later, checks are still coming in and more than $11,000 has been raised. We are so grateful.

> *" There are a lot of things in the world we have no control over. Our children's medical issues are uncertain at best. However, it felt good to do something positive to help raise money for research that could benefit them and the rest of the FA community."*

There are a lot of things in the world we have no control over. Our children's medical issues are uncertain at best. However, it felt good to do something positive to help raise money for research that could benefit them and the rest of the FA community. We encourage other families who may not have fundraised in the past couple of years to do so. We are glad that we did and feel strongly you will, too. ■

"Bounce for Fanconi Anemia"

Not many kids would give up the chance to open presents on their sixth birthday, but Shane Ciolfi did just that. He wanted to hold a very special birthday party this year—to raise money for FARF on behalf of his very special cousin and best friend, Wyatt Klimkiewicz, who has FA. Shane held a "Bounce for Fanconi Anemia" party and fundraiser for his sixth birthday party. In the Ciolfi family, great minds think alike, as Shane was following the example set by his brother, Cameron, who held a "Bounce for Fanconi Anemia" birthday party in 2008, in honor of cousin Wyatt and in memory of Trey Dougherty. Together, the Ciolfi brothers have raised more than $10,000 for FA research!

Thank you ...for coming to my 6th Birthday Party and "Bouncing for fanconi anemia" ...and my best friend and cousin wyatt. Together we raised $3,305 for the fanconi anemia research fund!

thank you!! your friend Shane Ciolfi

Climb for a Cure/Your Rope Team

In 2008, the McQueen family joined Bill McCorey and other friends to form Your Rope Team and developed a mountain-climbing fundraising event. In 2009, the Lindsay family joined in. This year, both families participated in Your Rope Team's Climb, soliciting sponsorship/funds when they climbed Old Rag Mountain in Virginia on Aug. 28. This year's event has raised more than $9,600 to date.

Musical Theater Fundraiser

Ready for her close up—Molly Nash, age 16, has performed with PHAMALy (Physically Handicapped Actors and Musical Artist League) for the past five years. The theater troupe is composed of players with a variety of challenges, who teach each other to never let a disability or disease stop them from doing what they love to do the most—perform! This summer, Molly took center stage in an FA fundraiser, as more than 150 supporters came to watch her perform as Chip in "Beauty and the Beast." To date, the fundraiser has generated more than $4,300. Take a bow, Molly!

Kick FA

Thirteen-year-old Matt Pearl, who has FA, wanted to do something to help the cause and his 15-year-old sister, Alex, who has struggled with FA-related health issues this year. Matt put together a kick ball tournament, got the word out on the local news, helped write a fundraising letter, handed out brochures door-to-door, held a raffle, made a presentation about FA at a school assembly and managed to raise more than $20,000 for FA research. Matt's question to potential donors was, "WWYD—What Would You Do?"

Schuman Concert

For the past 11 years, dedicated Fanconi Anemia Research Fund supporter and accomplished violinist Sharon Schuman has organized a classical concert and wine auction fundraiser in Eugene, Ore. This year's concert generated $11,190, bringing Sharon's cumulative fundraising total to more than $213,000 for FA research.

Hull Golf Tournament

Family members, friends and fraternity brothers turned out in force for the 11th Annual Chris Hull Memorial Sigma Pi Open on the Penn State Blue Course, held in honor of Chris Hull, Theta '89, who passed away in 1999 of FA. This event raised $7,900 for FARF this year, reaching a cumulative total of $75,000.

Candy Bar Fundraiser/Wal-Mart Match

Five-year-old Alise Williams has the best seat in the house as she helps mother Debra with a candy bar fundraiser outside their local Wal-Mart. The retailer provided matching funds, bringing the total raised to $1,930.

Coley's Cause

In 2004, 6-year-old Nicole "Coley" Levine passed away due to FA. Every year since then, the Levine family has held Coley's Cause Memorial Golf Tournament fundraiser benefitting FARF in memory of feisty, fun-loving Coley. So far, this event has raised more than $122,000.

Upcoming Fundraising Events Calendar

Check to see what's going on in your area.

Oct. 22, 2010
Play for FA Casino Night and Silent Auction Richmond, Va.
This event will be held at the Renaissance Conference Center in Richmond. For more information, please visit www.playforfa.org or contact Lorraine McQueen at lmcqueen01@verizon.net or 804-247-1459.

Nov. 20, 2010
Hoot n' Holler Denver, Colo.
This event features an auction, dancing, Texas Hold 'Em and more at the Hilton Garden Inn Denver Tech Center. For more information, visit www.katafoundation.org.

Feb. 13, 2011
Seventh Annual Valentine Fanconi Anemia 5K/8K Run/Walk Portland, Ore.
The run will take place in downtown Portland at 8 a.m. For more information, visit http://valentine5k.com.

May 1, 2011
International Fanconi Anemia Day Global!
The 2nd Annual International Fanconi Anemia Day is shaping up to be even more spectacular than the first. Now is the time to start brainstorming and planning events!

For more information about any of these events, or to have your event added to this page, please call us toll free at 1-888-FANCONI or email info@fanconi.org.

Ongoing Events:

Kaps for Kendall
Donate $25 to FARF through Kaps for Kendall and a knitted hat will be donated to patients who have lost their hair from chemo and radiation treatments. If you are a knitter, you can help by supplying a hat. Contact Allison and Whitney Atkinson at www.kapsforkendall.com.

Kendall and Taylor Atkinson Foundation (KATA)
The Kendall and Taylor Atkinson Foundation raises money for Fanconi anemia research. Visit www.katafoundation.org for more information, to donate or to purchase from their online store.

CaddyForACure.com
Caddy For A Cure Inc. raises funds for designated charitable organizations while offering the opportunity to be "inside the ropes" as a caddy for a Tour player at a PGA Tour event. This perfect gift for a golf fanatic offers a one-of-a-kind professional sports fantasy while contributing to genetic disease research. The Fanconi Anemia Research Fund receives 20% of the proceeds from Caddy For A Cure. Contact Russ Holden at www.CaddyForACure.com.

Maddie's Angels Cookbooks
These cookbooks are for sale to support FA research for $15 each. For more info, please contact Nancy and Ernie Landwehr at maddiesmom50@yahoo.com or ernie_landwehr@msn.com.

Sandy Carter Photography
Sandy Carter Photography donates 100% of proceeds sold through her website at www.etsy.com/shop/sandycarterphoto to FA research.

Global Partners Help Support the Fund's Mission

The Fanconi Anemia Research Fund's ability to finance the work of researchers is leveraged by several FA research and support groups outside of the United States:

Fanconi Hope, a United Kingdom-based charitable trust, has made several significant donations to the Fund targeted specifically to support research that emanates from United Kingdom research facilities. Fanconi Hope also hosts scientific meetings, including one this November in London on developing an international FA gene therapy trial.

Steven's Association Moonrise, an organization set up in Belgium in memory of Steven Keyes, a young man who died of FA at 24, similarly looks to the Fund to screen research and identify suitable Europe-based projects. Like Fanconi Hope, Steven's Association Moonrise donates to the Fund with a request to use its donation for a specific project.

Fanconi Canada is very active, raising funds for Canadian-based FA research, and holding scientific and fundraising events throughout the year.

For more information about these organizations, as well as other country-specific organizations that provide support to FA families, go to the Global Partners section of the Fund's website, www.fanconi.org. ■

Family Fundraising Efforts

From, Jan. 1, 2010 through Sept. 30, 2010, Fanconi anemia families raised $996,186 for the Fanconi Anemia Research Fund. The Fund also received $2,493 from the United Way and $5,288 through the Combined Federal Campaign, for a grand total of $1,003,967 for this nine-month period—a very strong showing, especially in this still-challenging economic climate. It's important to note that ninety-two cents of every dollar you raise goes directly toward the Fund's mission of research and family support services. Thank you very much for your efforts so far this year, and remember, the Fund's staff is available to assist you with your holiday fundraising efforts.

$175,000 and up
Dave and Lynn Frohnmayer
Kevin and Katie Rogers/My Best Friend

$50,000 - $99,999
Kendall & Taylor Atkinson Foundation with the Nash and Atkinson Families
Paul and Rena Rice
Glen Shearer and Peg Padden

$20,000 - $49,999
Chris and Susan Collins
Dan and Nikki McCarthy
Mark and Diane Pearl

$10,000 - $19,999
Kerrie Brannock
Mark De Groot and Hanneke Takkenberg
Ed and Janice Duffy
Susan Gannon
Michael Glas and Carol Felmy
Steve and Jennifer Klimkiewicz/One for Wyatt
Kevin and Lorraine McQueen
Pedro and Marina Ravelo
Mike and Beth Vangel

$5,000 - $9,999
John and Kim Connelly
David and Mary Ann Fiaschetti
Alan and Rachel Grossman
Brian Horrigan and Amy Levine
Charles and Katy Hull
Tanner and Jessica Lindsay
Jeremy and Stacey Mefford
Jack and Lisa Nash

$1,000 - $4,999
Mark and Linda Baumiller
James and Tracy Biby
Jeffrey and Donna Boggs
Chris and Jennifer Branov
Donald and Danielle Burkin
David and Kim Chew
Tyler and Teresa Clifton
Curt and Crystal Fales
Justin and Britteny Ferrin
Allen Goldberg and Lori Strongin
David and Paula Guidara
Owen Hall and Margaret Kasting
Jeff and Beth Janock
Erik Kjos-Hanssen and Turid Frislid
Gregory and Lynnette Lowrimore
Bill and Jackie Lucarell
Tue Marker and Kirstine la Cour Rasmussen
Steve and Alison McClay
Sheila Meehan
Jim and Holly Mirenda
Ian and Tricia Mitchell
Tyler Morrison and Rachel Altmann
Robert and Mary Nori
Peter and Janice Pless
Mark Ritchie and Lisa Mingo
Maureen Russo
Rick and Lynn Sablosky
Bob and Andrea Sacks
Bryan and Karen Siebenthal
Charles and Jennifer Sumrall
William and Mary Underriner
Troy and Debra Williams
Michael and Kim Williams
Wesley and Sue Wycoff
Sean and Kristin Young

up to $999
Chris and Ellen Allums
Ken and Jeanne Atkinson
Yavin Atzmon and Sharon Harari
Gerald and Julie Barbier
John and Audrey Barrow
Randy and Nancy Bloxom
Roel and Diane Brand
Lezlie Chesler
Jeanette Clark
Daniel Conde
Richard Day
Tony and Phyllis Dellapenta
Donna DellaRatta
Wendy Delzell
Mike Dennis and Ginger Eggers
Kim and Stephanie Dillow
Pat and Mary DiMarino
Antonino and Marie DiMercurio
Brian and Jennifer Dorman
Delbert and Linda Dotson
Sandra and Lindsay Dunn
David and Kelly Dunnock
Gene and Lynn Eddy
Sharon Ellis
Doreen Flynn
Brett and Nanette Foster
Gary and Melody Ganz
Brian and Lisa Gillott
Andrew and Jennifer Gough
Mitchell and Tirzah Haik
Bob and Victoria Hathcock
Sean and Helen Healey
Roger and Eleanor Herman
Peter and Tara Himmelreich
Jeff Hoffman
Alan Howard-Jones
Bonnie Hutchins
Lester and Nancy Jansen
Randy and Christie Kelley
John and Karilyn Kelson
Keith and Krisstina King
Joseph Konikowski
Kayla Lackey
Ernie and Nancy Landwehr
David and Shawn Leonardson
Peg LeRoux
Todd and Kristin Levine
Anne Llewellyn
Eric and Beth Losekamp
Deane Marchbein and Stuart Cohen
Gil and Peggy McDaniel
Adam and Olivia Mindle
Matt and Andrea Morris
John and Betty Mozisek
Des Murnane and Mai Byrne
Kenny and Lisa Myhan
Tony and Lina Nahas
Jack and Tammy Neal
Bob and Alice Nicholson
Fred and Nancy Nunes
Lorraine O'Connor
Michael and Katharine Ormond
Joshua and Crystal Pepper
Steve Perkins and Karen Magrath
Lori Petersen
John and Dianne Ploetz
Michael and Kay Proctor
Lynn and Shirley Quilici
Gail Richardson
Jan and Leonard Riley
Richard and Dolores Satterlee
Sharon Saunders
Bill and Connie Schenone
Thomas and Brenda Seiford
Matt and Diane Senatore
Lorne Shelson and Annette Waxberg
Jim and Carol Siniawski
Mitzi Speelman
Tamara Stephens
Greg and Brandi Stuart
Mark and Susan Trager
Steve and Melissa Turner
Al and Michelle Valenzuela
Xiaoqing Wang and Ning Liu
Marc Weiner
Norman and Michelle Wilson
Gerald and Elizabeth Wisz

In Loving Memory

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

Matt Hale.....9/30/04 – 5/4/10
Abiye Gambo.....3/25/94 – 7/19/10
Catelyn Duffy.....10/19/87 – 7/25/10
Ashlynn Nicholson.....11/10/03 – 8/7/10
Scott Dorman.....8/16/88 – 8/16/10
Jackson Van Singel.....8/10/05 – 8/25/10

Fanconi Anemia
RESEARCH FUND, INC.

1801 Willamette St, Suite 200
Eugene, Oregon 97401

REMINDER

Use of Logo

A reminder to our families: 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.

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.*

STAFF

Staff

Beverly Mayhew, Executive Director
Teresa Kennedy, Director of Family Support Services
Kristi Keller, Bookkeeper/ Administrative Assistant
Kim Larsen, Grant Writer/Conference Coordinator
Pauline Thaler, Administrative Assistant

Newsletter Editors

Lynn Frohnmayer
Laurie Gerloff
Mary Ellen Eiler
Pauline Thaler

HOW YOU CAN HELP

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

Donations by Mail: 1801 Willamette St, Suite 200, Eugene, OR 97401

Please go to www.fanconi.org to learn about other ways to donate.

1801 Willamette St., Suite 200, Eugene, OR 97401
phone: (541) 687-4658 • (888) FANCONI (USA only)
fax: (541) 687-0548 • e-mail: info@fanconi.org • web: www.fanconi.org

BOARDS

Board of Directors

Barry Rubenstein, JD, President
David Frohnmayer, JD, Vice President
Ruby Brockett, Secretary/Treasurer
Mary Ellen Eiler
Richard Gelinas, PhD
Deane Marchbein, MD
Kevin McQueen
Peg Padden
Mark Pearl
Kevin Rogers
Robert Sacks
Michael Vangel
Peter von Hippel, PhD
Joyce Owen, PhD, Director Emeritus
Lynn Frohnmayer, Advisor to the Board

Scientific Advisory Board

Grover Bagby, Jr., MD, Chair
Joseph Califano, MD
Marc Coltrera, MD
Richard Gelinas, PhD
Eva Guinan, MD
Hans Joenje, PhD
Christopher Mathew, PhD
Stephen Meyn, MD, PhD
Raymond Monnat, Jr., MD
Elaine Ostrander, PhD
Bhuvanesh Singh, MD
Erich Sturgis, MD, MPH
Jakub Tolar, MD, PhD
Neal Young, MD

Layout and Design: www.KerryLutzDesign.com
Please recycle this paper.