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# FA Family Newsletter

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

## The Emerging Adult Population with FA: Challenges, Coping and Quality of Life



*Amy Frohnmayer, MA, is a psychosocial researcher at Oregon Health & Science University. She presented the results of her Master's thesis project at the annual FA Family Meeting held in Casco, Maine, in June. Frohnmayer is 24 years old and has Fanconi anemia.*

Amy Frohnmayer presented the results of her 2010 Master's thesis, completed as part of her degree in psychology. She explored the physical, medical, cognitive perceptual and social challenges that adults with Fanconi anemia confront, the coping strategies they use, and how these strategies relate to quality of life.

Frohnmayer interviewed 18 adults with FA and obtained surveys from an additional 96 adults. Both groups ranged in age from

18 to 55. Approximately a third of participants had experienced cancer and half had received a bone marrow transplant. Frequently cited psychological challenges were anticipation of future medical complications, perception of self as fundamentally different from peers, concern about taking responsibility for medical management, and emotions related to the loss of other FA patients.

Survey data indicated that coping strategies used most frequently

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Science News From the Family Meeting

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issue 50

## FA Family Newsletter Hits Milestone Issue

This is the 50<sup>th</sup> issue of the *FA Family Newsletter*. The first newsletter was produced in 1985 in Dave and Lynn Frohnmayer's living room using an early-model computer and a daisy wheel printer. The six-page, black and white newsletter was sent to 30 families.

Today the publication regularly exceeds 20 pages and is sent to about 4,000 families and researchers around the world. Lynn still writes every science article and sends

them to the relevant researcher or clinician to review for accuracy. Dave Frohnmayer; Mary Ellen Eiler, former FARF executive director; and Joyce Owen, PhD, board member emeritus, also review all articles and weigh in with edits.

Back issues of the newsletter serve as helpful references and are available (dating from 1998) on the Fanconi Anemia Research Fund's website, [www.fanconi.org](http://www.fanconi.org). ■

# The Emerging Adult Population with FA: Challenges, Coping and Quality of Life *continued from page 1.*

were acceptance and active coping; those used least often were substance abuse and denial. Active coping, positive reframing and religion (including spirituality) were highly correlated with hope (one of the wellbeing outcome measures). Self-blame, self-distraction and denial were correlated negatively with wellbeing.

Of the 18 patients interviewed, 17 could identify positive aspects of living with FA. Fourteen felt they had more compassion for others, 12 had more self-appreciation, 10 had a deeper appreciation for life and seven felt that dealing with FA gave purpose to their lives. Participants were asked to choose emotion cards that described how this illness had affected their lives. Thirteen chose hope, 12 picked compassion and 12 chose

love. Fewer patients chose negative emotions such as sadness, fear, embarrassment and anger.

Coping mechanisms that helped those interviewed were positive thinking, acceptance and maintaining normalcy, use of external support systems, and actively managing one's medical care. Substance use and passivity were not helpful.

Advice to other FA patients from those interviewed included the following:

- Get involved in your medical care.
- Reach out to the community for support and advice.
- Try to find something positive about having FA.
- Live your life: don't let FA control your life.
- Always be hopeful. ■

## New Directions in FA Research Discussed



Akiko Shimamura, MD, PhD, Fred Hutchinson Cancer Research Center, Children's Hospital, Seattle, discussed research that could be of future benefit to Fanconi anemia patients. Ongoing efforts include the following:

- Scientists are now screening for drugs that might correct the FA pathway in cells. When the FA pathway is working correctly, DNA damage causes a small molecule called ubiquitin to attach to the FANCD2 protein (a process called ubiquitization), enabling DNA damage repair. Researchers can now test for the add-on of the ubiquitin molecule and are screening for compounds that might effectively result in the addition of ubiquitin and therefore correct the FA pathway.
- Scientists can take adult skin cells, introduce four different genes and re-program these cells to become induced pluripotent stem (iPS) cells. IPS cells can replicate themselves and can be cultured to become any tissue in the body. Gene correction combined with iPS cell technology has corrected sickle cell

anemia in mice. Scientists hope that this method could eventually cure the FA deficiency in bone marrow cells.

- Genomics technology allows scientists to sequence the entire genome at a fraction of earlier costs. This methodology could be used to identify quickly and accurately all disease-causing mutations.
- Scientists at the Fred Hutchinson Cancer Research Institute, Seattle, have developed a clinically successful method of increasing the number of blood-forming cells available from a single cord blood unit by expanding the cells in the laboratory. Patients undergoing a cord blood transplant who received these expanded cells recovered their neutrophil counts in half the time compared to those who received cells that had not been expanded. This technology could significantly improve the outcomes for patients undergoing cord blood transplantation, which is particularly important for patients who lack a suitably matched donor. ■

**Scientists are now screening for drugs that might correct the FA pathway in cells.**

## Early Gynecological Care is Essential for Girls with FA



Given the special health risks associated with Fanconi anemia, parents need to introduce their girls with FA to the need for gynecologic care early. The first appointment should be at age 16 or at the onset of menstruation.

Rahel Ghebre, MD,  
University of Minnesota,

stated that establishing trust with a gynecologist is of paramount importance and should be the main purpose of the first visit.

The gynecologist needs to be aware of FA-associated gynecological complications or be willing to learn. Patients should be prepared to discuss potentially sensitive issues such as sexual activity, contraception and risk for sexually-transmitted diseases.

The physical exam should include a careful evaluation of the vulva, vagina and cervix. Annual screening should

include a Pap test and vulvar and vaginal inspection for lesions. Colposcopy is useful to magnify this area, and should be followed by biopsy if suspicious lesions are seen. Lesions of the vulva or vagina should be treated aggressively with surgery, since FA patients may respond poorly to standard radiation and chemotherapy. Other possible therapies include laser surgery of lesions of the vulva or vagina and topical medicine such as Aldara (imiquimod) or 5-fluorouracil for pre-cancerous lesions.

At each annual visit, patients should be tested for sexually-transmitted infections (including gonorrhea and chlamydia) until age 25, a standard of the US Centers for Disease Control and Prevention. Dr. Ghebre recommended use of condoms to prevent sexually-transmitted infections. Contraception should be used when pregnancy is not desired.

The human papillomavirus (HPV) causes cervical cancer and genital warts. It has also been implicated in vulvar, vaginal and anal cancer. FA patients should be vaccinated against HPV as early as age 9. ■

## Early Results Reported from Alternate Donor, Multicenter Transplant Study Using Radiation-Free Approach



Steve Margossian, MD, PhD, Dana-Farber Cancer Institute, reported on a five-center transplant study using alternate donors and busulfan instead of radiation in the conditioning protocol. Participating centers are Memorial Sloan-Kettering Cancer Research Center, Children's Hospital Boston,

Children's Hospital of Wisconsin, Cincinnati Children's Hospital Medical Center and the Fred Hutchinson Cancer Research Center. Fourteen patients have enrolled in this study, ranging in age from 5 to 27, with a median age of 8. One of the concerns of the study was that

removing radiation would lead to poor engraftment, but so far all 14 patients have engrafted. Eleven patients are alive and disease-free at one to 24 months post-transplant. Deaths were attributed to infection, pulmonary hypertension and multi-organ failure.

**One of the concerns of the study was that removing radiation would lead to poor engraftment, but so far all 14 patients have engrafted.**

Dr. Margossian stated that early results in engraftment and survival were encouraging. Longer follow-up is needed to assess the impact of this protocol on chronic graft-versus-host disease and secondary malignancies. ■

## Understanding the Genetics of FA: Carriers, Complementation Groups and Cancer Risk

Certified genetics counselor Heather Zierhut, MS, University of Minnesota Medical Center, gave attendees at Camp Sunshine a primer on the genetics of Fanconi anemia. Approximately one in 181 individuals is a carrier of FA. In the Ashkenazi Jewish population, carrier frequency is one in 90.

To know if relatives of an FA patient are carriers, one must know the FA patient's specific disease mutations. If one mutation has been passed on to a relative, that individual is a carrier of FA. When two carriers of disease mutations in the same gene have children, there is a one in four chance that a child will have FA, a two in four chance that children will be carriers, and a one in four chance that the child will not have FA nor be a carrier.

Fifteen FA genes have been discovered, and patients with defects in the same gene are in the same "complementation group." Most complementation groups are not associated with an elevated risk of cancer in

carriers. However, studies show an elevated risk of cancer in carriers from five different complementation groups.

The *FANCD1/BRCA2* gene mutations confer a 45% risk of breast cancer to carriers by age 70 and up to an 80% risk of breast cancer during one's lifetime. Ovarian cancer is also a major concern for women carriers. Male carriers in this complementation group are at increased risk of breast and prostate cancer. Two complementation groups, *FANCN* and *FANCI*, typically double a carrier's breast cancer risk. One study showed that *FANCC* also doubles the risk of breast cancer for FA carriers. A newly discovered FA gene, *FANCO/RAD51C*, is also associated with ovarian cancer in the general population. The specific gene mutations that a person carries can influence the specific cancer risks. Carriers are encouraged to discuss these risks with a genetic counselor and/or their health care providers. ■

## Dietitian Advises to "Eat the Rainbow!"



Carol Ceresa, Registered Dietitian, Veteran's Administration Medical Center, San Francisco, spoke to Camp Sunshine attendees about the importance of good nutrition in maintaining optimal health and helping to prevent cancer. All three of her sisters had

Fanconi anemia, which inspired her early interest in the protective effects of nutrient-rich foods.

Ceresa highly recommends the book *What Color is your Diet?* by David Heber, MD, PhD, Director of Human Nutrition, UCLA. Choosing fruits and vegetables covering a wide range of bright colors promotes good health. She recommended eating two cups of fruit and two cups of vegetables daily and leaving the skins on whenever possible.

Ceresa believes that certain foods may protect against cancer and cited a recent study by Tong Chen, MD, PhD, Ohio State University, which suggests that freeze-dried strawberries might play a role in preventing esophageal cancer. Other foods thought to have anti-cancer properties include turmeric (which contains the anti-inflammatory curcumin), mushrooms (especially shiitake), walnuts and avocados. Ceresa recommended the book *Healing Spices* by Bharat B. Aggarwal, PhD, University of Texas, M.D. Anderson Cancer Center, which details healing properties of foods and spices.

Butter should be avoided in favor of monounsaturated fats such as olive oil, avocado oil, peanut oil and walnut oil. One should avoid foods rich in fats, such as pizza, cheese, hotdogs, sausage, bacon, ribs and fried food. Avoid foods high in sugar (sodas, fruit drinks, sweet teas, candy, desserts) and choose whole grains such as brown rice over white rice. Eat fish twice a week (especially oil-rich fish such as salmon, sardines and mackerel). And, Ceresa concluded, "don't forget to 'eat the rainbow!'" ■

## Plans Under Way to Study FA Parental Issues Related to PGD

This year marks the 10-year anniversary of the successful use of pre-implantation genetic diagnosis (PGD) with human leukocyte antigens typing for Fanconi anemia. Heather Zierhut, MS, a certified genetics counselor, notes that much has changed since this technology began. She wrote a grant to study the ethical, legal, and psychosocial issues that have surrounded this subject. Through an electronic survey, she and her collaborator will ask all

parents of children with FA, not just those who have undergone PGD, about their decision-making, opinions and experiences with the use of this technology. Researchers will invite FA groups in Canada, the United Kingdom and Australia to participate. The survey will be emailed to FA parents shortly. The study results will be presented at the Brocher Foundation Symposium in Geneva, Switzerland, in November. ■

## Managing Head and Neck Cancer in FA Patients



David Kutler, MD, Weill Cornell Medical College, gave a comprehensive overview of head and neck squamous cell carcinoma (HNSCC) in the FA population. While this cancer is relatively rare in the general population, 21% of FA patients will experience HNSCC by age 40 at a

median age of 31. In the non-FA population, smoking and/or drinking causes HNSCC. FA patients should not drink or smoke and must avoid second-hand smoke.

Sixty-five percent of FA HNSCCs occur in the oral cavity. They appear as ulcers or masses in the mouth that do not heal or go away. The human papillomavirus (HPV) has been implicated in certain HNSCCs in the general population and in 83% of FA tumors, according

to one study. Dr. Kutler strongly recommends that FA patients, male and female, receive the HPV vaccine. This vaccine protects against HPV subtypes 16, 18, 7 and 11; HPV16 causes 90-95% of HPV-related cancers, so this vaccine might protect against many FA malignancies.

Treatment of HNSCCs can cause tremendous morbidity and negatively affect quality of life. Small tumors can be surgically removed; surgical removal of large tumors can be devastating. Doctors sometimes use robotic surgery to remove head and neck cancer, which enables surgical removal through the mouth, thereby causing less morbidity.

Dr. Kutler reported on 12 FA patients who underwent radiation therapy at his center. The average dose of radiation was 5,278 rads. Eight of 12 patients died, four during the course of radiation. Patients experienced high-grade mucositis, inability to swallow, low blood counts, esophageal stenosis and wound breakdown. Radiation therapy should be done only for more advanced tumors and by surgeons experienced in FA. Chemotherapy such as cisplatin should be avoided but Erbitux is a possible option for FA patients.

Early detection is crucial. Head and neck surgeons (otolaryngologists) for adults should conduct surveillance examinations, because pediatric ENTs do not commonly see or diagnosis head and neck cancer.

The youngest FA patient diagnosed with HNSCC was 9. Surveillance should begin around that age and be repeated every six months and more frequently if cancer is diagnosed. ■

### Three take-aways from the presentation:

- Patients should not drink or smoke and must avoid second-hand smoke.
- Dr. Kutler strongly recommends that FA patients, male and female, receive the HPV vaccine.
- Surveillance should begin around the age of 9 and should be repeated every six months and more frequently if cancer is diagnosed.

## Bone Marrow Clonal Abnormalities: Laboratories Agree on Major Findings

Betsy Hirsch, PhD, University of Minnesota Medical School, reviewed findings from three major Fanconi anemia research centers concerning the significance of certain abnormal clones found in the bone marrow of FA patients. These three centers (University of Minnesota, University of Cincinnati and Charité Hospital, Berlin) have reached consensus on the following:

- The 3q gain (a gain of the long arm of chromosome 3) is the most frequent recurring clonal abnormality in FA. This abnormality is rarely seen in pediatric patients who do not have FA.
- The 3q gain is not transient, but persists in the bone marrow.
- Clones with a 3q gain frequently expand over time, often evolve to include monosomy 7, and lead to myelodysplastic syndrome and/or acute myelogenous leukemia (MDS/AML) in the majority of cases. In a German study of 18 FA patients with a 3q gain, 90% evolved to MDS/AML.

- The 3q gain can remain as a sole abnormality for two or more years without evidence of MDS or AML.

Dr. Hirsch provided a lucid explanation of abnormal clones and their significance. A “clone” refers to a group of cells that have the same abnormality. These clones are not present at birth but are acquired over time, and they signify an abnormal process in the bone marrow.

The incidence of abnormal clones increases with age. FA patients over age 20 have a 70% chance of having an abnormal clone compared to 24% of those under age 10.

In individuals with FA, the chromosome abnormalities found in clones are mostly “unbalanced” and result in a gain or loss of a portion of a chromosome.

The 3q gain can be difficult to recognize in standard chromosome studies that do not use techniques such as FISH. Chromosome reports that have the term “add” or “mar” next to a chromosome should be carefully reviewed to see if a hidden piece of 3q is involved. ■

## Fertility and Fertility Preservation in Males and Females with FA

Rahel Ghebre, MD, University of Minnesota, believes that fertility and its preservation are subjects that merit discussion well before patients undergo a medical procedure that usually results in loss of fertility, such as transplant. A survey of cancer survivors showed that many suffered unresolved grief and depression over their infertility and were resentful that fertility preservation options were never discussed. Physicians and family should explore the patient’s interest and potential options in fertility preservation.

Fertility is higher in women with Fanconi anemia than in men. A 1991 study of 110 FA females age 16 or older reported 26 pregnancies between the ages of 18 and 25, with 18 surviving children. A study last year of 300 FA patients transplanted at 15 transplant centers revealed that 10 women had 14 children post-transplant.

Women with FA have fewer eggs, fewer follicles, and experience premature ovarian failure. Hormonal imbalances caused by androgens or chemotherapy reduce fertility due

to loss of ovarian function. Females are most vulnerable to radiation damage of the uterus before puberty.

The most successful method of preserving fertility before transplantation is embryo cryopreservation. One needs a sperm donor, an obstacle for young patients or those without a committed relationship. Survival per thawed embryo ranges from 35% to 90%. For those who do not have a partner, egg cryopreservation can be done in certain centers with expertise. Pregnancy rates from this method are around 20%. Investigational methods include freezing of ovarian tissue.

Fertility preservation in males with FA is less promising. Some FA males have fathered children, but this is rare. The cause of infertility is usually low sperm count, but also includes anatomic problems and primary or secondary hormonal insufficiency.

Options for preserving fertility in males include sperm cryopreservation and hormonal therapies to protect testicular tissue during chemotherapy or radiation therapy. ■

## Psychological/Social Issues Affect Families During Transplantation



Julia Kearney, MD, Memorial Sloan-Kettering Cancer Center (MSKCC), New York, reminded families facing a stem cell transplant (SCT) that the “family is the patient” and to remember that “you are not crazy, what you are going through is crazy!” She gave a comprehensive

presentation on social, educational and psychological considerations before, during and after SCT.

Prior to SCT, Fanconi anemia families and patients often seem more distressed than non-FA families, so MSKCC conducted a study to investigate psychiatric issues in families with FA. Fear of future cancers, congenital malformations, neurologic issues, multiple affected children, effects of androgen use and infertility issues can contribute to psychological issues and increase a family’s stress level.

Goals of a pre-transplant psychiatric consultation are to address current problems, identify medical stressors and

develop a symptom management plan. Families should identify personal and professional sources of support. Children need preparation for what lies ahead. Honest, age-appropriate discussion with parents and helping professionals, books, role-playing medical procedures, and games that help children understand medical issues can be helpful.

At MSKCC, a multidisciplinary psychosocial team helps with issues as they arise throughout the transplant. It’s useful to create a routine by posting goals for each day. Psychiatric symptoms such as depression and anxiety cause suffering and should be treated.

Dr. Kearney identified the challenges that await families post-transplant. These include coping with an extremely complex medication schedule, caregiver burnout, and emotional/behavioral issues. Maintaining contact with peers during transplant (through Skype, for example) can help with re-entry to school and peer contact post-transplant. FA survivors and their families need to understand the importance of adhering to medical advice and follow-up screening, and to avoid behaviors that could risk future health. ■

## Making a Personal Connection to FA Research

Several researchers attended the FA Family Meeting at Camp Sunshine to gather tissue or saliva samples to help further their Fanconi anemia research. Eunike Velleuer, MD, Heinrich Heine University, Children’s Hospital, Düsseldorf, Germany, took oral brush samples from FA patients as a means of detecting early-stage head and neck cancer. Flavia Teles, DDS, MS, DMSc, The Forsyth Institute, Cambridge, took saliva samples for her study on microbial markers of oral cancer; and Melinda Butsch Kovacic, MPH, PhD, Cincinnati Children’s Hospital Medical Center, Cincinnati, collected saliva samples for her work on human papilloma virus (HPV) in people with FA.

In-depth information about families affected by FA was collected by the research team of Blanche Alter,

MD, and Lisa Leathwood, RN, from the National Cancer Institute in Rockville, Md. Dr. Alter and Leathwood took advantage of the high number of new families at Camp this year (14!) to encourage enrollment in the Inherited Bone Marrow Failure Syndromes Cohort Study.

We know from past meeting evaluations that families appreciate the presence of researchers at Camp and clearly understand that their participation in the projects can significantly contribute to the body of knowledge about FA. Likewise, the researchers always express gratitude for the opportunity to meet directly with families and acquire such precious samples. Thanks to both the families and researchers for their efforts to move FA research forward! ■

## DNA Testing for FA Before Pregnancy



Certified genetics counselor Heather Zierhut, MS, gave Camp Sunshine attendees an overview of preimplantation genetic diagnosis (PGD). PGD offers families the option of testing for Fanconi anemia and for human leukocyte antigens status in an early embryo before implantation.

Readers can consult the October 2010 *FA Family Newsletter* for detailed information on the steps involved in this process.

The Reproductive Genetics Institute of Chicago, Illinois, has helped 17 families at risk for FA to try to expand their families. These families have gone through

51 PGD cycles (average is three per family), resulting in the births of six healthy children. The chance that any one PGD attempt will result in the birth of a child is 12%. PGD is expensive, averaging approximately \$20,000 per cycle. Most insurance companies will not cover PGD.

Zierhut discussed the physical and emotional implications of undergoing PGD. Interviews with 14 women who had undergone PGD elicited both positive and negative emotions: women felt in control, empowered and hopeful, but also experienced anxiety while anticipating results, and extreme distress when a cycle failed. It was suggested that couples consider in advance what they will do with fertilized eggs that are not needed when the process ends. Zierhut cautioned that PGD can negatively affect future pregnancies and, on rare occasion, be life-threatening to the mother. ■

## Excellent Transplant Outcomes Continue at Charité Hospital, Berlin



Over more than a decade, Wolfram Ebell, MD, Charité Children's Hospital, Berlin, has transplanted 27 Fanconi anemia patients with protocols using oral busulfan in a cumulative dose of 2 mg/kg instead of radiation in the conditioning regimen. None of these patients

has experienced organ toxicity clearly attributed to the busulfan. Dr. Ebell stated that he uses half of the busulfan dosage currently given in a multi-center, US-based study, which could explain the lack of toxicity.

Alternate donor transplants continue to improve at Charité Children's Hospital (see *FA Family Newsletter* #49 for information on recent protocols and outcomes at this center). All nine patients with alternate donors on the present protocol, called GEFA03, survive, with only one experiencing grade 1 graft-versus-host disease.

One patient on GEFA03 relapsed with his original clonal abnormality. This patient was given a mild dose of fludarabine, Cytosan and ARA-C, and a stem cell boost from his original donor, and is now disease-free.

**At his center, Dr. Ebell finds no survival difference between patients with matched sibling donors and those with alternate donors.**

Dr. Ebell noted that a large number of FA patients, often with androgen support, do not undergo transplant. This group still survives longer than patients who have needed to go to transplant. With improvements in transplant outcomes, this could change. At his center, Dr. Ebell finds no survival difference between patients with matched sibling donors and those with alternate donors. ■



# Minnesota Reports on Transplant Outcomes



The University of Minnesota Amplatz Children's Hospital has transplanted more than 195 Fanconi anemia patients since 1982, using a series of protocols to improve transplant outcomes.

Margaret MacMillan, MD, reported on 40 patients with alternate

donors enrolled in the current trial, which has been open since 2006. This protocol includes 300 rads of total body irradiation, thymic shielding to hasten immune recovery, Cytosan, fludarabine and ATG followed by either T-cell depleted bone marrow or umbilical cord blood. Survival for the entire group is 87%; all 14 patients under the age of 10 survive (median follow-up of 32 months). Dr. MacMillan stated that although the data suggest that children under 10 have a better chance of survival than older patients, age itself is not a criterion for coming to transplant. Transfusion history is also associated with survival.

All 25 patients under the age of 18 with matched sibling donors and a chemotherapy only (no irradiation) protocol are alive and well, with no graft-versus-host disease.

**All 25 patients under the age of 18 with matched sibling donors and a chemotherapy only (no irradiation) protocol are alive and well, with no graft-versus-host disease.**

The University of Minnesota also transplanted eight high-risk patients using a busulfan-based protocol. Seven of these patients had leukemia. Patients experienced high levels of toxicity; four patients survive. This protocol is now open only for patients in the FANCD1/BRCA2 complementation group or for patients who cannot tolerate TBI.

Dr. MacMillan announced plans for a new trial in the near future. This trial will reduce the length of time a patient is on an immunosuppressant post-transplant in an effort to reduce the risk for life-threatening infections. ■



## 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 that are 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 will be 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](mailto:teresa@fanconi.org)

#### Send samples to:

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](mailto:corlessc@ohsu.edu)

## Thriving Through Transplant: Coping Tips\*

- Get as much information as possible. *Knowledge is power.*
- Get professional help. *Psychiatric symptoms cause suffering.*
- Seek multidisciplinary support. *It takes a village...!*
- Don't be afraid to ask your doctor for appropriate medication. *Better living through chemistry.*
- Remember: Everyone is affected. *The family is the patient.*

\*These tips were provided by Julia Kearney, MD, Memorial Sloan-Kettering Cancer Center, New York, at the 2011 FA Family Meeting.

## Family Offers Suggestions to Others Facing Bone Marrow Transplant

Nearly two years ago, Fanconi anemia patient Peter Fiaschetti, now 12, had a bone marrow transplant (BMT). Peter's mother, Mary Ann, shared some tips on preparing for BMT at our recent FA Family Meeting. Here is one of the slides from her presentation:

### EARLY DECISIONS

- Learn as much as possible about Fanconi anemia (FA)
  - Register with FARF and join the e-group
  - FARF: handbooks, newsletters, etc.
  - Camp Sunshine
  - Family online journeys (blogs, CaringBridge, etc.)
  - Regional FA meetings
- Seek treatment at a recognized transplant center (referrals, insurance approval for out of network provider, etc.)
- Move near a recognized transplant center
- Adopt healthy habits
- Raise \$\$\$\$ for FARF to fund a cure
- Appreciate every day—live life



*Peter Fiaschetti (right) and BMT donor, brother Joey, are all smiles with the hospital team on transplant day.*

In preparing for transplant and the possible complications that may arise, Mary Ann reminds families, “Be positive! This is a life-saving experience.” To view Mary Ann’s presentation in its entirety, go to [www.fanconi.org/index.php/family\\_support/annual\\_family\\_meeting](http://www.fanconi.org/index.php/family_support/annual_family_meeting) and click on the “BMT Tips from a Post-Transplant Family” presentation.

For more information about the bone marrow transplant process, consult our *Guidelines for Diagnosis and Management*, available on the Fund’s website. To find out more about what BMT is like for families, register with the Fund and post questions on our secure e-group. ■

# Living with Fanconi Anemia

By Mary Grabher



*Mary (second from the left), attends her first FA Family Meeting, along with (from left to right) her daughter Anne-Marie, her sister Anne and her mother Joan.*

After facing a myriad of “unusual” health problems throughout my life, I found out at the age of 51 that I have Fanconi anemia. My diagnosis came after a bout of anal cancer followed by chemotherapy and radiation. Just a few days into the cancer treatment, my bone marrow crashed with low counts that were off the charts. After a stay in intensive care and in isolation, I stabilized enough to be able to leave the hospital and return home.

Did I ever present a puzzle to my hematologist! He expected that I might have a slight reaction to the chemo and radiation, but he never expected me to bottom out. After months of very low counts and a bone marrow test that proved inconclusive, he searched high and low for a reason, or for a diagnosis, to explain why my body reacted in such a way to the cancer treatment. He eventually diagnosed me with Fanconi anemia.

All the other doctors said that he was “brilliant” for his correct diagnosis of this rare disease. I believe that’s true because no other doctor throughout my entire life ever thought to test me for FA. As a child, I was very sick, with low platelets. I had several bone marrow tests, but all the doctors would say was that the low counts were a mystery. Forty-one years later, the mystery was solved. Most of the illnesses I have had can be traced back to Fanconi anemia: very low birth weight, poor digestive system, no appetite as a child, low blood counts, easy bruising, nose bleeds, squamous cell skin cancer twice, oral cancer twice, early menopause, anal cancer, pre-diabetes (and now diabetes), and elevated liver enzymes.

So now I’ve slowly begun my journey to learn about Fanconi anemia and to put this diagnosis into perspective. I’ve come to the conclusion that life has been very good to me. Despite medical challenges, I’ve looked at each issue one at a time and dealt with what was on my plate at that time. I looked at each surgery as a door that I had to pass through to reach better health on the other side. I’ve lived with the motto that “this too shall pass.” This has helped me face many health issues. I make and keep my doctor appointments, but I live life, too. I’ve always been honest with myself and my doctors, and have made it a point never to be embarrassed to ask my doctor anything.

I have lived a full life with Fanconi anemia. I graduated high school and was a pretty fun teenager. I fell in love and got married. I graduated from college, taught at an inner city high school for 13 years, adopted a beautiful daughter and taught at a local university for 10 years. I did the math once and figured out that through my years of teaching I have influenced more than 5,000 young adults. I have a wonderful support system of family, friends and doctors. My hematologist gently reminds me that I am not FA, rather I am Mary, living with FA. My mother and my sister, Anne, have tirelessly switched hats from caretaker to nurse’s aide, cook and cheerleader for me. I am learning as much about this disease as I can, and I hope to be able to be a part of many research projects. I would love it if something I contribute to research could lead to excellent treatment for those who must make a life with FA.

**My hematologist gently reminds me that I am not FA, rather I am Mary, living with FA.**

For some reason, I survived the chemo and radiation. I think my stubborn nature could be part of it, but perhaps there is something in me, in my DNA, in my Fanconi anemia that could help explain my survival and help other patients. I can only hope. At the end of the day, all we can ever do is hope. ■



Julia Flynn, who has FA, receives a warm welcome to the Family Meeting.

### Campers List One-Word Raves

In the last support session of the 2011 Fanconi Anemia Family Meeting, led by Nancy Cincotta, MSW, MPhil, of Camp Sunshine, participants were asked to say one word to describe their experience at Camp. Following are some responses:

enriched RE-ENGAGED  
 encouraging  
 recharging  
 calm emotional  
 FANTABULOUS  
 priceless Hopeful  
 knowledge ENERGIZING  
 APPRECIATION SUPPORTED  
 SPECTACULAR  
 HOME  
 Extraordinary  
 Bittersweet  
 informative VITAL  
 empowering INSPIRING  
 amazing  
 GIFT  
 grateful  
 Comforted  
 educational  
 THOUGHTFUL  
 UNITY  
 UJEW



### Looking for the Perfect Fanconi Anemia Sundae?

By Stephanie Griggs, FA Parent and First-time FA Family Meeting Participant

Camp Sunshine was....geez, how can I possibly describe Camp Sunshine in words? Camp Sunshine was exactly what our family needed. It was the perfect Fanconi anemia sundae. A scoop of rest and relaxation, a bit of excitement and entertainment, and a whole lot of fun and friends, all covered with ooey gooey gobs of FA information and support, sprinkled with nuttiness and topped off with a cherry of HOPE. ■

## A FANTABULOUS Experience for FA Families



*Fifty-nine families from eight countries enjoy the FA Family Meeting, held at Camp Sunshine in Casco, Maine.*

Fifty-nine families—including 14 first-time families—gathered at Camp Sunshine in Casco, Maine, in June for the Fund's 20<sup>th</sup> annual FA Family Meeting. Once again, Camp Sunshine proved an ideal setting for busy days filled with educational sessions, support groups and fun recreational activities.

Forty-eight children and six adults with Fanconi anemia attended the meeting along with parents, siblings and extended family members, for a total attendance of 227. Attendees traveled from eight countries: Canada, Denmark, Israel, France, Australia, Germany, the United Kingdom and the United States.

Highlights from the educational agenda included updates from several bone marrow transplantation (BMT) centers, sessions on head and neck cancer, fertility preservation, psychosocial issues related to BMT, and a look at exciting FA research currently under way in labs around the world. Amy Frohnmayer, an adult with FA and a long-time Camp Sunshine attendee, shared the results from a study she conducted about adults coping with FA, and two parent panels recounted their personal

journeys related to BMTs and Preimplantation Genetic Diagnosis (PGD). Thanks to Mary Ann and David Fiaschetti, Diane and Mark Pearl, and Lynn and Dave Frohnmayer for participating on the BMT panel, and to Lorne Shelson and Annette Waxberg, Pedro and Marina Ravelo, and Yavin Atzmon and Sharon Harari for sharing their PGD stories.

Camp Sunshine proved an ideal setting for busy days filled with educational sessions, support groups and fun recreational activities.

Scores of caring volunteers, along with dedicated camp staff, made sure that smiles were in abundance, and that attendees were well satisfied with good food and fond memories. ■

# At Last, Success!

By Holly Mirenda



Our family welcomed Georgia Elizabeth Mirenda into the world at 12:01 a.m. on June 26, 2011, after only 16 minutes in the hospital. While her delivery was expeditious, we had been awaiting Gigi's arrival for several years.

We've been blessed with many things in our lives together, including wonderfully happy children, an incredibly supportive and loving family, and exceptional medical care. It's clear now that we were also blessed with patience. We began our *in vitro* fertilization/pre-implantation genetic diagnosis (IVF/PGD) journey in fall 2008 and ultimately completed nine rounds before becoming pregnant in October 2010.

When we first learned about the possibility of pre-selecting a healthy embryo and a human leukocyte antigens (HLA) match through IVF/PGD, we knew *immediately* that this would be our path forward for our family and for our daughter Sofia's health (Sofia has FA). Even with this absolute conviction, however, our patience, reasoning and emotional strength were thoroughly tested: the seemingly endless hours on the road for appointments, two lost early pregnancies, and the many times we arrived at the clinic, hopeful for an embryo transfer—and left brokenhearted.

Having two young kids, Sofia and Teddy, at home while facing this challenge helped to keep us focused, and, well, busy. It also complicated family discussions about health. Looking back, it was Teddy's anxiety about Mommy's health that was perhaps the toughest

emotional part of our process. He was obviously too young to grasp the entire situation, but old enough to notice Mommy's absences, moods and shots. We explained the shots by saying that families sometimes need a little medicine to help make babies. (A funny side story was when Teddy told a friend that babies are made by mommies putting shots in their tummies.) Sofia was young enough not to notice what was happening, except when Mom was gone before dawn on yet another round trip to see the doctors or too tired to read stories at bedtime.

Nothing was easy about the process. Much like the kid's game *Chutes and Ladders*, it seemed that every time we climbed higher, we hit a point where we'd slide down, and sometimes *off* the game board. Deep down, we trusted that someday we would be made whole.

We're now witness to the love and joy that patience and faith has brought to our family. Baby Gigi is not affected by FA, although she is a carrier like we are. She is also an HLA match for Sofia. We collected and stored her cord blood at birth, to be used when Sofia needs a transplant.

Much like the kid's game *Chutes and Ladders*, it seemed that every time we climbed higher, we hit a point where we'd slide down, and sometimes *off* the game board.

Throughout the physical and emotional rollercoaster, we kept faith in Sofia's ability to remain healthy, as well as in our amazing team of nurses, doctors and geneticists. We have to take the opportunity to sing the praises of CNY Fertility and Dr. Mark Hughes at Genesis Genetics. The care, faith and comfort that they extended to us were as important as the science that helped bring us baby Gigi.

Sofia is now 6, beautiful, and has an energy that thrills us every single day. She and Teddy (8) are completely smitten with Gigi. Now that Gigi is starting to smile, we know that the feeling is mutual. ■

# The Sum of All Fears: A Single Mom's Journey Through Fanconi Anemia

By Simone Kieze



I've learned that being a single parent in the Fanconi anemia population is extremely rare. I can't say I was elated to fall into either statistical category, but the moment my son, Mekhi, was diagnosed,

I knew our path on this Earth would be different.

I remember the day my husband left us. Mekhi was a year old. I was overcome with fear that I didn't have what it takes to raise a man alone in today's society. I was at the pinnacle of my accounting career at a large accounting firm with only my family as support. Determined to make it, since failure was not an option, I quickly adopted the attitude "other single moms did this before me; I can get it done!" Then I lost my job. I felt that 2-year-old Mekhi understood everything I was going through. I had to pull it together for him, if no one else.

While unemployed, I decided to continue my studies for the Certified Public Accountant examination. Mekhi stayed perched on my hip as I toiled through accounting problems at night; his job was to keep me awake. And he did such a great job! Eventually, I passed the exam, got a job and qualified for insurance. When I was finally able to take Mekhi for his first check-up in over a year, his platelet count was 25,000. One month later he was diagnosed with Fanconi anemia. That was the straw that broke this camel's back. Losing my husband, my job and getting this diagnosis in the span of one year felt like someone tied my heart to a 10-ton brick and threw it into the ocean as shark bait. I couldn't even begin to process what lessons I could possibly learn from these experiences. It just wasn't fair. There are many things in life I braced myself for as a single mom: talking about sex, love, marriage... but having to tell my child that he has a preleukemic disorder was definitely not on the top of my list. I was devastated. Having no one to help me with that discussion added to the frustration.

Through all that's happened I've learned that there are some things that one should not attempt to go through

alone. The independent spirit I once prided myself on had to be thrown out the window. My thinking had to change; my attitude needed to change; I needed to be a different type of mom to my son to get us both through. The first thing I had to do was admit that I needed help. I sought the best therapist I could find. I later adopted these three basic principles that have helped me immensely:

- Every life has a purpose. Discovering that purpose and going along life's intended path will allow everything to align and will take you to the next level. While being a member of the FA family was something I never thought would happen to me in a million years, I'm elated to be involved.
- Where I am now is exactly where I should be. Nothing happens by chance or coincidence. There are no mistakes on this journey, only opportunities to learn. Every person I've met along this path has been brought into our lives for a reason; every doctor, every nurse and friend has touched our lives in some way and we've learned to appreciate it. Within this past year, we've been able to open the eyes of the Caribbean community to FA and the importance of being a donor. None of this would have happened without Mekhi.
- Love. Love. Love. Teaching Mekhi to love and accept everything around him has become important to me. Just appreciating the simple pleasures of the breeze brushing across our faces, the sound of the ocean and accepting everything nature has to offer us brings a great level of peace to our lives.

**Through all that's happened I've learned that there are some things that one should not attempt to go through alone.**

Life is short. Nothing should be taken for granted and, if our destiny is that I should outlive my son, I want to know that every minute and second of every day spent with him was meaningful. ■

# French Family Support Group Celebrates 20 Years

By Charles Bichet



*The French Family Support Group celebrates 20 years with a trip to Disneyland Paris.*

Under a warm spring sun, Disneyland Paris offered a perfect setting for a two-day celebration of the Association Française de la Maladie de Fanconi’s (AFMF) 20 years in existence.

Thirty Fanconi anemia patients and families came from all over France, Ralf Dietrich and Eunike Velleur came from Germany, and a family even came from the United Kingdom. A few friends and half a dozen volunteers helped us manage the excitement of the kids. All together, about 100 people attended the celebration. We were quite a spirited party, determined to make the happening one of hope, friendship and a celebration of life.

First, we all gathered for a superb buffet lunch on Saturday which gave everyone the necessary energy for

the festivities. We enjoyed meeting old friends again, getting to know new families and hearing news about those who weren’t able to attend. It was fun to blow out the candles on our anniversary cake!

The afternoon was dedicated to the formal support group session, focusing on practical life matters and further FA-related exchanges between the families, including the young FA adults. By early evening, we entered Disneyland Paris theme park and bumped into one another throughout the evening.

On Sunday, our group had breakfast and spent more time enjoying the theme park. It was quite a memorable event for all, especially the children. Thanks to our various sponsors, including Disneyland Paris, as well as to those families who contributed to organizing and financing this wonderful event! ■

## What’s next for AFMF?

The group will continue to provide support for adults with Fanconi anemia and FA family members, develop relationships with other FA groups, and focus on funding FA research in the near future.

## In Loving Memory

*“For some moments in life there are no words.”*

John Hanna..... 5/31/72 – 3/26/11

Justin Barbier..... 4/30/85 – 7/17/11

Edwin Ferreira..... 2/6/84 - 5/18/11

Katrina Aggabao ..... 4/29/05 – 8/30/11



## Kids from FA Families Pitch in to Raise Money To Support Research

Children from families affected by Fanconi anemia are proving that fundraising isn't just for adults. At the FA Family Meeting in June, Kevin McQueen, FA parent and Fanconi Anemia Research Fund board member, and Bev Mayhew, the Fund's Executive Director, handed out awards to "Kids Helping Kids," in recognition of some highly committed junior fundraisers. Here are some highlights of the work kids did to help raise funds for FA research:

- Benjamin Morrison held his second *Chess Tournament* in honor of FA Day.
- Spencer Boggs sold hand cut-outs at school and pitched

- in to help make his family's *Fun Day for FA* a big success.
- Devin Fales participated in *Kicks for FA*, a karate exhibition.
- Austin Jaros-Riley helped his family make their first *Cosmic Bowl-a-thon* an event to remember.
- Matt and Alexandra Pearl spearheaded their second *Kick FA* kickball tournament and, along with Sean McQueen, participated in the *Climb for a Cure* event.
- Peter Fiaschetti held a penny drive while recovering from his bone marrow transplant last year. Please join the Fund in thanking these inspiring fundraisers. ■

## Personal Tragedy Leads to Large Donation



*Melody Ann and Michael Alan Miller, two siblings who passed away decades ago from Fanconi anemia.*

Mrs. Gloria J. Miller of Bethlehem, Pa., who lost two children to Fanconi anemia many years ago, has turned her personal tragedy into hope for many others through a very generous gift of \$250,000 to the Fanconi Anemia Research Fund. Mrs. Miller made this extraordinary contribution to support research as a tribute to her 5-year-old daughter, Melody Ann, whom she lost to FA in 1951, and to her 9-year-old son, Michael Alan, who died of FA in 1962.

According to her son, Jody Miller, "My mother's fondest and deepest desire is that this gift will help

others afflicted with this disease, as well as their families. Hopefully, this contribution will provide monies needed for research, and provide doctors and scientists with support that will allow them to continue to work and to discover a cure for this illness that has taken far too many precious lives already." He noted that at the time of his brother's death in 1962, his family was told by Max Strumia, MD, at Bryn Mawr Hospital in Philadelphia, that there were only 47 living FA patients in the world and that Michael Alan, at age 9, was the oldest survivor.

*"My mother's fondest and deepest desire is that this gift will help others afflicted with this disease, as well as their families. Hopefully, this contribution will provide monies needed for research, and provide doctors and scientists with support that will allow them to continue to work and to discover a cure for this illness that has taken far too many precious lives already."*

"What an amazing, unexpected gift this is," says Lynn Frohnmyer, Fund co-founder and board member, "and how tragic the familiar circumstances leading to this incredible generosity. We are humbled and heartened by Mrs. Miller's spectacular philanthropy." ■

# Second Annual International Fanconi Anemia Day a Big Success



More than 20 families affected by Fanconi anemia helped raise awareness and funds for FA research for *The Second Annual International Fanconi Anemia Day*, observed May 1. Events included a karate exhibition, a “cosmic” bowl-a-thon, a Facebook challenge, an online auction, yard sales, a 5k run/walk, and fundraising letter campaigns. In all, families raised more than \$75,000 for FA research and family support.

*International Fanconi Anemia Day* was the brainchild of the FA community’s own Peg Padden. Padden, an FA

parent, member of the Fund’s board of directors, and FA fundraiser, has some exciting new plans. Peg announced that she will be devoting the next year to raising money for the Fund. We are delighted with her commitment to the cause and grateful for the gift of her time.

Many thanks to these dedicated and imaginative families for all they did to spread the word and raise money for such an important cause. It’s not too early to start thinking about next year! ■

## Family Fundraising Efforts Remain Strong

From Jan. 1, 2011 through Sept. 15, 2011, Fanconi anemia families raised \$1,200,571 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.

### **\$250,000 and up**

Gloria Miller

### **\$150,000 - \$199,999**

Dave and Lynn Frohnmayer

### **\$100,000 - \$149,999**

Kendall & Taylor Atkinson Foundation with the Nash and Atkinson Families

Glen Shearer and Peg Padden

### **\$20,000 - \$49,999**

Kerrie Brannock

Mike and Tracy Brannock

Robert and Barbara Capone

John and Kim Connelly

Ed and Janice Duffy

Fanconi Hope Charitable Trust

Kevin and Lorraine McQueen

Todd and Kristin Levine

### **\$10,000 - \$19,999**

Chris and Susan Collins

Mark De Groot and Hanneke Takkenberg

Carole Felmy

Michael Glas

Kaps for Kendall

Matthew and Evelyn Keyes

Dan and Nikki McCarthy

### **\$5,000 - \$9,999**

Jeffrey and Donna Boggs

Chris and Jennifer Branov

Donald and Danielle Burkin

Roger and Eleanor Herman

Brian Horrigan and Amy Levine

Charles and Katy Hull

Steve and Jennifer Klimkiewicz/Wyatt’s Warriors

Jeremy and Stacey Mefford

Mark and Diane Pearl

Peter and Janice Pless

George and Kathryn Reardon

### **\$1,000 - \$4,999**

Jimmy and Jenny Armentrout

Andrew and Vicki Athens

Mark and Linda Baumiller

Israel and Mary Jo Becerra

Randy and Nancy Bloxom

Richard and Tena Bosen

Patti Carter

David and Kim Chew

Darrel and Kalani DeHaan

Brian and Jennifer Dorman

Lisa Doyle

Ezat and Laila Faizyar

David and Mary Ann Fiaschetti

Ben and Stephanie Griggs

Alan and Rachel Grossman

David and Paula Guidara

Owen Hall and Margaret Kasting

Erik Kjos-Hanssen and Frislid Turid

Christopher and Dana Lamb

Mark and Angela Lamm

Tanner and Jessica Lindsay

Gregory and Lynnette Lowrimore

Deane Marchbein and Stuart Cohen

Tue Marker and Kirstine la Cour

Rasmussen

Orion and Lisa Marx

Sheila Meehan

Jim and Holly Mirenda

Tyler Morrison and Rachel Altmann

Jack and Lisa Nash

Robert and Mary Nori

Ron and Fredi Norris

Joshua and Crystal Pepper

Paul and Rena Rice

Mark Ritchie and Lisa Mingo

Kevin and Katie Rogers/My Best Friend

Rick and Lynn Sablosky

Bob and Andrea Sacks

Bill and Connie Schenone

Bruce and Loreen Timperley

William and Mary Underriner

Mike and Beth Vangel

### **Up to \$999**

Randy and Lauren Armstrong

Ken and Jeanne Atkinson

Gerald Barbier

Julie Barbier

John and Audrey Barrow

Conrad and Joan Bender

James and Tracy Biby

Michael and Diane Bradley

Richard Briga

Lezlie Chesler

Tyler and Teresa Clifton

Brad and Cynthia Curry

Dottie Day

Richard Day

Jeremy and Michelle DellaValle

John Delzell

Wendy Delzell

James and Carol Dillon

Antonino and Marie Di Mercurio

Pat and Mary DiMarino

Lindsay and Sandra Dunn

David and Kelly Dunning

Gene and Lynn Eddy

Ginger Eggers

Sharon Ellis

Billy Jo and Debbie Estep

Kay Eubanks

Curt and Crystal Fales

Edwin Ferreira and Yalitzta Negron

Nancy Finnegan

Scott Finnegan

Doreen Flynn

Brett and Nanette Foster

Liz Funk

Gary and Melody Ganz

Brian and Lisa Gillott

Pat and Maria Gleason

Josh and Maria Godwin

Allen Goldberg and Laurie Strongin

Andrew and Jennifer Gough

John and Raquel Hanna

Jeff Hoffman

Judy Hoffman

Shane and Colleen Irvin

Jeff and Beth Janock

John and Karilyn Kelson

Keith and Krisstina King

Kayla Lackey

Tim and Mary Ann Lana

Eugene and Renee Lemmon

Skip Longstaff and Susan Gannon-Longstaff

Eric and Beth Losekamp

Bill and Jackie Lucarell

Steve and Alison McClay

Kevin and Barbara McKee

Catherine McKeon

Gianna and Lauren Megna

Ian and Tricia Mitchell

Des Murnane and Mai Byrne

Tony and Lina Nahas

Jack and Tammy Neal

Fred and Nancy Nunes

Lorraine O’Connor

Michael and Katharine Ormond

Albina Parente

John and Dianne Ploetz

Michael and Kay Proctor

Lynn and Shirley Quilici

Pedro and Marina Ravelo

Gail Richardson

Leonard and Jan Riley

Richard and Dolores Satterlee

Thomas and Brenda Seiford

Bryan and Karen Siebenthal

Debby Slater

Jeff Slater

Tamara Stephens

LeaAnn Stiller

Greg and Brandi Stuart

Charles and Jennifer Sumrall

Paul and Debra Sundsvold

Mark and Susan Trager

Tom and Kathy Uno

Joe and Wendy Vitirritto

Mike and Wendy Wade

Xiaoqing Wang and Ning Liu

Marc Weiner

Sandy Weiner

Michael and Kim Williams

Norman and Michelle Wilson

Sean and Kristin Young

## Everything Old is New Again

By Jeanne Atkinson



Special friends of ours, Vicky Stone and Christine Flaum, came up with the idea to have a jewelry sale featuring mostly recycled and renewed pieces. They knew that women often tire of wearing the same jewelry or perhaps they no longer wear that cute blue dress with the matching blue earrings. So why not ask people to donate

what they no longer wear, polish the pieces and invite even more people to a jewelry party featuring jewelry designs from today and yesterday at great prices, all to benefit a great cause?

For this event, gently-used jewelry was obtained via e-mail request and networking between friends, coworkers and family. The request included the suggestion that people might have vintage jewelry from a mother or grandmother that they would like to give to a good cause. One woman sent us two boxes of her mother's beloved costume jewelry, saying that she was thrilled to find something meaningful to do with it. We offered a drop-off spot or pick-up service for those providing jewelry.

People were invited to come and enjoy a glass of wine while shopping for great jewelry deals. The list of people came from our past fundraising efforts and from Vicky and Christine's contacts. The sale was held at Christine's home from 3 p.m. to 8 p.m. on a weekday. In the end, around \$3,000 was raised for FA research, and all involved voted it a big success! ■

## Social Networking Pays Off – FA Families Use Online Tools to Raise Funds

Facebook, Twitter, LinkedIn—the list of online social networking tools continues to expand. Families affected by Fanconi anemia are harnessing the power of these tools to increase awareness of FA and to raise funds. Families have participated in Facebook contests and launched online challenges to help spread the word about our worthy cause.

FA parent Mary Nori saw an opportunity to land some much-needed funds for research and submitted the Fanconi Anemia Research Fund's name for the *Chase Community Giving Program's Facebook Contest* in April. This contest sought to identify the top nonprofits in the US based on Facebook user votes. Nori used her online network and the Fund's e-group to spread the word and recruit voters. The Fund placed in the

top 100 vote-getters in the first round, winning \$25,000 from Chase.

Michael and Katharine Ormond, parents of 3-year-old Elias (who has FA), are no strangers to using social media for social good. For the past two years, they've held an online giving challenge in honor of *International Fanconi Anemia Day*. According to the *razoo* donation site, to date the Ormond's challenge has raised more than \$7,500.

The Internet can be an excellent resource for fundraisers. Whether using Facebook to spread the word about a contest or simply emailing contacts about an upcoming event, social networking is an effective way to gain some attention for a good cause. Follow the Fund on Facebook at [www.facebook.com/fanconianemiaresearchfund](http://www.facebook.com/fanconianemiaresearchfund). ■



*Fanconi Anemia Research Fund's Board President, Barry Rubenstein, accepts a \$25,000 check from "Chase" the dog. Thanks to strong social networking efforts from our community, the Fund was a winner in the Chase Community Giving Program's online challenge.*

## Wiggle for Wyatt



*Smiling trophies are awarded to all participants.*

What do you get when you mix a group of preschoolers, a room full of balloons, and some great music? A fun-filled fundraiser! Kim Kenworthy of Schwenksville, Pa., developed this dance party at her son's preschool in honor of Wyatt Klimkiewicz, her young relative with FA. Each participating child received one of the fabulous trophies pictured above. In all, the event raised more than \$4,500. Now that's something to wiggle about!

## Fighting the Fight



*Michael Capone, age 12*

It was a big year for the Capone family. After Michael Capone, age 12, was diagnosed with FA in July 2010, his mother, Barbara, decided to roll up her sleeves and start fundraising. In May, the Capone family called together friends, family and acquaintances to "Fight the Fanconi Anemia Fight along with Michael's Army" in Boothwyn, Pa. The evening featured food, wine, dancing and a silent auction. The Capones raised \$26,732 for the Fund and are already planning a golf tournament for next year. Fighting the fight and walking the walk, Barb Capone is a mother determined to make a difference.

## Knit Away FA



Kaps for Kendall is a nonprofit organization founded in memory of Kendall Atkinson. Kendall had FA and worried about losing her hair during bone marrow transplant. For every \$25 donated to the Fund through the Kaps for Kendall website, [www.kapsforkendall.com](http://www.kapsforkendall.com), the organization sends a handmade hat to a person undergoing medical treatment that results in hair loss. On March 19, Kaps for Kendall held *Knit Away FA*, a first-time event bringing 48 knitters together to knit 467 hats. *Knit Away FA* raised more than \$9,000 for the Fund!

## Leaping for Connor and Dane



*The Mefford Family*

The Mefford family's *International FA Day* turtle race and silent auction event in Carrollton, Ky. was a hit again this year, with community members donating everything needed for the event. Activities for children and delicious food added to the fun. Connor and Dane, who have FA, joined sister Bailey and parents Stacey and Jeremy for a successful and exciting day. The event raised more than \$8,000 for the Fund. Apparently, slow and steady *does* win the race.

## Door to Door Success



Jack Timperley, age 12

Recently, the Fund's staff received a message from 12-year-old Jack Timperley, who has FA, reporting on his fundraising efforts:

Hello there, this is Jack Timperley with FA. On May 1st, I went door to door selling rose bushes for \$30. A lot of people said they didn't want flowers, but gave me \$50 for FARF. By the end of the day I raised over \$1,400. Plus \$333 that I already had, plus \$125 from my mom, plus my grandma collected money, plus we still have to give some.

## A Cosmic Bowl-a-thon



Austin Jaros-Riley, age 15

On Saturday, May 14, Patti Carter invited the St. Peters, Mo., community to "Come Play for FA" at a "cosmic" bowl-a-thon held in honor of her grandson, Austin Jaros-Riley, who has FA. This first-

time event, organized in conjunction with *The Second International FA Day*, drew an enthusiastic crowd and raised more than \$2,100 to help strike out Fanconi anemia.

## How FARF Can Help You Fundraise

More than 80% of the Fanconi Anemia Research Fund's annual budget comes from family fundraisers. The Fund's staff is here to help make your fundraising efforts a success. We can:

- Provide sample fundraising letters and help you edit/design your letter
- Use your photos to personalize your materials
- Use your mailing list to send your letter or invitation from our office
- Provide ideas, information and materials for 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 write their donation checks to the "Fanconi Anemia Research Fund." When we receive a donation, we will generate a letter of thanks from the Fund with a tax receipt, and we'll notify you that a donation has been made in your name.

We appreciate all of your fundraising efforts. You are making a difference. ■

## Fun Day for FA



Members of the Boggs family held their *Fun Day for FA* on May 21 in Coeburn, Va. Activities included a 5K run/walk, face painting, a miniature train and carnival games. This event was held in loving memory of Nicholas

Boggs, and participants received a "Team Nicholas" T-shirt. Nicholas, who had FA, loved having fun and a big crowd of people—which is exactly what this day was all about. Through this event, the Boggs family raised \$1,030 for FA research and family support.

## Fanconi Anemia Research Pioneer Retires



Hans Joenje, PhD, a pioneer in Fanconi anemia research and former member of the Fanconi Anemia Research Fund's Scientific Advisory Board, has retired. Dr. Joenje, the only person ever to receive both the Fund's *Award of Merit* and the *Distinguished Service Award*, made exceptionally

significant contributions to the field of FA research, including participating in the discovery of 12 FA genes, which helped to provide the foundation for current FA science. His research remains integral to the study of FA.

Early in Dr. Joenje's career, he made the groundbreaking discovery that the chromosomal breakage in FA depends on atmospheric oxygen. He has continued to work on FA and the genetic toxicology of oxygen ever since. When Dr. Joenje began studying FA, little was known about the rare disorder and none of the genes responsible for FA had been identified. In 1992, Dr. Joenje was instrumental in

forming a European consortium to clarify the genetic basis of FA. Fifteen FA genes have now been discovered.

In addition to his work in the laboratory, the Fund recognizes Dr. Joenje for his dedicated service to families affected by Fanconi anemia. He has attended the Fund's annual FA Family Meeting to present information to families about the scientific basis of FA and has worked tirelessly with families to help them determine their particular FA gene.

Dr. Joenje spoke in March 2011 at a symposium organized at the Vrije Universiteit in Amsterdam, The Netherlands, to honor him for his work in Fanconi anemia research. Dave Frohnmayer participated by video, and thanked Dr. Joenje on behalf of FARF and FA families worldwide for his innovative work. Although retired and looking forward to spending more time in his garden and with his beloved dachshund, Dr. Joenje has promised to remain actively involved in the FA community.

We thank Dr. Joenje for his tremendous contribution to the field of FA research and wish him well in his active retirement. ■

## Welcome Dr. William to the Fund's Scientific Advisory Board



William N. William, Jr., MD, a medical oncologist from The University of Texas MD Anderson Cancer Center in Houston, accepted an appointment in April to serve a three-year term on the Fanconi Anemia Research Fund's Scientific Advisory Board.

Dr. William is an assistant professor in the Department of Thoracic/Head and Neck Medical Oncology, Division of Cancer Medicine at MD Anderson, a position he has held since 2006. Dr. William came to the US from Brazil for a post-doctoral and then a clinical fellowship. He received his

medical degree in 2001, graduating from the Faculdade de Medicina da Universidade de São Paulo, Brazil. Dr. William has co-authored dozens of articles related to head and neck cancer and has won several prestigious awards, including the American Society of Clinical Oncology Cancer Foundations' Young Investigator Award in 2009.

Dr. William joins a group of distinguished researchers and clinicians on the Scientific Advisory Board. Members of this board (listed on the back cover of this newsletter) guide the Fund's research priorities, advocate on behalf of FA at national and international meetings, and provide guidance and support to the staff and board of the Fund. We welcome Dr. William and are honored to have him join the FARF team! ■

## Researchers and Clinicians to Convene in Barcelona for 23rd Annual Scientific Symposium



The Fanconi Anemia Research Fund's 23<sup>rd</sup> annual Scientific Symposium, to be held this month in Barcelona, promises to be our "most exciting" symposium, according to Grover Bagby, MD, chair of our Scientific Advisory Board. Dr. Bagby's excitement is heightened by the rapidly evolving body of FA science to be shared by the more than 270 researchers and clinicians expected in Spain. Topics of special sessions include gene therapy, induced pluripotent stem cells, the role of the human papillomavirus in squamous cell carcinoma, and novel drug agents. Look for the April 2012 issue of the *FA Family Newsletter* for a full recap of the symposium. ■

## Head and Neck Cancer Experts Will Meet in 2012 To Continue Discussion on FA

Squamous cell carcinoma (SCC), particularly of the head and neck, poses a significant risk to people with Fanconi anemia and remains a top priority for the Fund. The Fund's third meeting devoted to SCC in FA patients was held in April 2010. A follow-up meeting, scheduled for March 2012, is expected to move the discussion

forward, with a focus on prevention, diagnosis and treatment. In addition, several exciting grant applications exploring various aspects of this insidious disease in FA patients are currently under review. Stay tuned for future updates. ■

### Upcoming Events

Contact Pauline Thaler, Project Coordinator at the Fund, at [pauline@fanconi.org](mailto:pauline@fanconi.org) with details of upcoming fundraising events. We will include your information on the Fundraising Events Calendar on our website.

### Help Advance FA Research!

FA researchers are working hard to find effective treatments and a cure for Fanconi anemia, but they can't do it alone. **FA researchers need you.**

Please consider donating research material.

#### For more information, contact:

Teresa Kennedy, Fanconi Anemia Research Fund, Inc.  
Phone: 541-687- 4658 or Email: [teresa@fanconi.org](mailto:teresa@fanconi.org)



*Working together to advance FA research.*

### Visit Our New FA Marketplace

The Fanconi Anemia Research Fund is pleased to announce a new feature on our website—the FA Marketplace. The FA Marketplace is a page featuring items for sale by families affected by FA and a central location where friends, families and donors can purchase items that benefit the Fund. One hundred percent of the net proceeds are donated to the Fund for FA research and family support.

Visit [www.fanconi.org](http://www.fanconi.org), click on the "fundraising" button and shop in the FA Marketplace. Each FA family "shopkeeper" handles his or her own sales.

If you currently sell items and donate the net proceeds to the Fund, and would like to be a part of the FA Marketplace, please email Pauline Thaler, Project Coordinator for the Fund, at [pauline@fanconi.org](mailto:pauline@fanconi.org). ■

## REMINDER

### Use of Logo

A reminder to our families with FA: please use our logo or letterhead only after you have consulted our staff here 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.*

## HOW YOU CAN HELP

**Donations Online:** You can donate via the heart button on the Fund's website ([www.fanconi.org](http://www.fanconi.org)) or through [www.networkforgood.org](http://www.networkforgood.org) or [www.paypal.com](http://www.paypal.com)

**Donations by Phone:** Call us at 541-687-4658 or toll free at 888-FANCONI (888-326-2664) (USA only)

**Donations by Mail:** 1801 Willamette St, Suite 200, Eugene, OR 97401  
*Please go to [www.fanconi.org](http://www.fanconi.org) to learn about other ways to donate.*

1801 Willamette St., Suite 200, Eugene, OR 97401  
phone: 541-687-4658 • 888-FANCONI (888-326-2664) (USA only)  
fax: 541-687-0548 • email: [info@fanconi.org](mailto:info@fanconi.org) • web: [www.fanconi.org](http://www.fanconi.org)

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