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FA Family Newsletter

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Phil and Penny Knight Pledge Support for Fanconi Anemia Research as Part of \$20 Million Fundraising Campaign



Lynn Frohnmayer and Phil Knight

Longtime supporters of the Fanconi Anemia Research Fund, Phil and Penny Knight have pledged an incredible gift of \$10 million over the next ten years to support the David B. Frohnmayer Scientific Research Fund, a dedicated

fund under the umbrella of FARE. This Fund was established by the FARE Board of Directors to honor co-founder David Frohnmayer, who passed away last March. The Frohnmayer dedicated fund is intended to accelerate, not replace, FARE's ongoing efforts to raise money each year to support scientific research grants and symposia, and to provide education and support services to families devastated by this disease.

The Knight pledge is part of FARE's new \$20 million campaign, which is committed to speeding up the pace of clinical trials, drug testing, gene therapies and more effective treatments for children and adults with Fanconi anemia. A major focus of these game-changing initiatives will be preventing

and curing the cancers that are now the primary cause of death in adults with FA. The support by the Knights is vital to helping achieve fundraising goals and carry out the Fund's mission. Their pledge serves to inspire and challenge the FA community to push forward its fundraising efforts, which have made possible more than 25 years of FA research and family support.

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27th Annual Scientific Symposium Celebrates History and Marks New Traditions

The 27th Annual Fanconi Anemia Research Fund Scientific Symposium was a meeting of many firsts. It was the first symposium held in Canada (Toronto), the first meeting co-sponsored by another Fanconi anemia organization (Fanconi Canada), the first conference that involved the participation of numerous FA individuals and family members, and the first year without the impactful presence of Fund co-founder David Frohnmayer,

who passed away in early 2015. While David's absence was noticeably felt, his legacy of mentorship and investigation carried on, as the symposium inspired engaging conversation and animated debates, and embodied an overall spirit of active learning.

More than 220 researchers, doctors, FA families, Fund board members and staff from over 15 countries attended

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Connections and collaborations made at symposium

27th Annual Scientific Symposium

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last September's conference. Forty-five oral presentations covered a range of subjects, including FA Genetics, Immunity and Inflammation, Squamous Cell Carcinoma, FA Protein and Function, Experimental Hematology, Replication: Initiation, Forks and RNA, Recombination Pathways, Protein Structure and Function, Hematology, Repair Pathways, and Potential New Roles for FA Proteins. Seventy-five abstracts were presented during the poster sessions and three keynote speakers drew in the audience with talks on inflammation and DNA damage (Glen Barber, PhD, University of Miami School of Medicine), drug discovery and development for novel targets (Patrick Gray, PhD, Omeros Corp), and CAR-T cells (Michael Jensen, MD, University of Washington School of Medicine, Seattle Children's Research Institute/BTCCCR. See pg. 4 in this newsletter).

As in past symposia, the event provided an opportunity for scientists, researchers, doctors and other Fanconi anemia experts to share their insights and to form new connections and collaborations. This interaction was fostered through a mentorship lunch which matched seven Fanconi anemia experts with groups of 10 early investigators and students, allowing for intimate discussion as well as teaching and learning moments.

The involvement of FA individuals and family members made this symposium special. Not only did they form a top-notch team of volunteers that helped Fund staff run the event smoothly, they made connections with researchers and doctors for whom Fanconi anemia may have seemed to be a faceless disease. The interaction between the families and the researchers impressed upon scientists the importance of their research, according to many present.

The influence of FA families was particularly poignant during the annual dinner banquet. FA parent and now Fund Board President Kevin McQueen, MBA, served as master of ceremonies, a role he fulfilled expertly and naturally. Heidi and Chris Collings, parents of 12-year-old Theo, who has FA, shared some of their family's journey, captivating the audience with emotion. Matt Pearl, a young adult with FA, inspired everyone with his powerful talk about living with FA and how it is a source of strength rather than weakness. Lorne Shelson, president of Fanconi Canada and an FA parent, drew upon Toronto's role in the history of Fanconi anemia research, acknowledging the contributions of a team led by Manuel Buchwald, PhD, OC, a pioneer in FA research, who was in the audience that evening.

History was both recognized and made during the banquet. Following a very moving tribute to David Frohnmayer, his wife and Fund co-founder Lynn Frohnmayer presented the inaugural David B. Frohnmayer Early Investigator Award to Agata Smogorzewska, MD, PhD, of The Rockefeller University. To honor David's commitment to mentorship and to young investigators, the award is meant to acknowledge an early investigator who has significantly impacted FA science and contributed to the FA community at large. Finally, the evening concluded with a video entitled *Faces of FA*, a compilation of all the faces of the disease, in particular highlighting the children and adults who rely on and benefit from FA research.

To all those who made this event such a success, especially the Fund's dedicated co-sponsor Fanconi Canada, *thank you*. The 28th Annual FA Scientific Symposium is September 15-18 in Bellevue, Wash. For more details, including information on registration and abstract submission, please visit www.fanconi.org.



Canadian and American FA families inspired researchers. Pictured here from left to right: Charlotte Heath, Tim Lana, Karen Bentley, Chris Bentley, Mary Ann Lana, Mary Hopper



FA families at a poster session. From left to right: Lorraine McQueen, Mary Ann Fiaschetti, Peter Fiaschetti



Fund co-founder Lynn Frohnmayer, MSW, presents the David B. Frohnmayer Early Investigator Award to Agata Smogorzewska, MD, PhD of The Rockefeller University

Phil and Penny Knight Pledge Support for Fanconi Anemia Research *continued from page 1*

“The urgency of this campaign to intensify scientific research is a direct consequence of FARE’s success. Fanconi anemia, once thought to be primarily a fatal bone marrow disorder of childhood, has now been unmasked as a DNA repair disorder that causes cancer in young adults,” said Grover C. Bagby, Jr., M.D., Chair Emeritus, OHSU Knight Cancer Institute, FARE Scientific Advisory Board member. “Thanks in part to research supported by FARE, 80-94% of children with FA now survive bone marrow transplants. As adults, though, they are at extraordinarily high risk for developing cancer, at an average age of 33. Since they can’t tolerate chemotherapy, treatments that are effective for others are out of the question for them,” Bagby said.

As grim as this prognosis sounds, it comes in the midst of an explosion of FA research — especially research that leads

to treatment. After 25 years devoted to identifying 19 FA genes, improving bone marrow transplants, and uncovering connections to breast and other cancers, FA scientists are now poised to create less toxic therapies that radically extend lives. These advances will help not only adults with FA, but millions of others with cancer.

Lynn Frohnmayer, Fund co-founder, elaborated on the implications of FA research: “Cancer is the largest cause of death in the FA young adult population. It is my profound hope that this Fund in David’s honor will identify new, effective therapies to prevent and treat these cancers, and give young people with FA the chance to experience a full adulthood.”

“Phil and Penny Knight’s generosity is a fitting tribute to David,” she continued. “More than financial, it is a gift of hope and compassion. It honors David’s boundless optimism and reinforces his firm conviction that dedicated people, working together, can change the world.”

New FA Gene Discovery: UBE2T



Speakers: Asuka Hira, MD, Radiation Biology Center, Kyoto University (sixth from right); Kimberly Rickman, MD-PhD student (fifth from left) The Rockefeller University; Helmut Hanenberg, MD, Heinrich Heine University (second from right) with others involved in the UBE2T discovery.

Three presenters at this year’s Scientific Symposium reported on the finding of a new FA gene, *UBE2T*, also called *FANCT*. With this discovery there are now 19 known FA or FA-like genes, in which mutations lead to clinical symptoms of FA.

Dr. Asuka Hira noted that the distribution of FA genes in Japan is quite different than in the rest of the world, with no reported *FANCC*, a higher percentage of *FANCA*, *FANCG* and potentially more FA genes not yet identified. In the process of trying to classify patients without defects in known FA genes, Dr. Hira discovered two individuals with mutations in the *UBE2T/FANCT* gene. Both individuals showed the typical clinical symptoms of FA. The function of the *UBE2T* gene early in the FA pathway was already known, however no FA patients with mutations had been previously identified.

In a North-American FA patient of Italian descent, Ms. Kimberly Rickman and Dr. Helmut Hanenberg independently reported mutations in *UBE2T/FANCT* by two different approaches. Importantly, this patient had inherited two unique mutations that cannot be detected by standard sequencing strategies used in clinical settings. In addition, this patient developed a reversion in the hematopoietic system in his first year of life and therefore had normal blood counts ever since. Dr. Hanenberg also reported that mutations in *UBE2T* are not associated with an increased breast/ovarian cancer risk.

Now that the *UBE2T* gene has been identified as an FA gene, other cases not yet assigned to a known FA complementation group could be tested for this gene.

A Promising Immunotherapy for a Specific Type of Acute Lymphoblastic Leukemia



Michael Jensen, MD, University of Washington School of Medicine, Seattle Children's Research Institute/BTCCCR

Excitement is rapidly growing for a new way to treat cancer: harnessing a patient's own immune system to fight the disease. Early results from small trials have produced remarkable, sometimes long-lasting results, suggesting that fine-tuning different methods of immunotherapy may have applicability to kill cancers that defy conventional therapies.

Immunotherapy improves the body's capacity to detect and kill cancer cells. One new method, called Adoptive Cell Transfer (ACT), involves a series of steps:

1. T-cells are collected from the patient.
2. These T-cells are genetically engineered in the laboratory to produce chimeric antigen receptors (CARs) on their surface. CARs are proteins that allow the T-cells to recognize a specific protein (antigen) on targeted tumor cells.
3. These CAR-T cells are multiplied many times over in the laboratory.

4. The expanded CAR-T cells are infused into the patient. Once in the patient, they continue to multiply rapidly.
5. CAR-T cells recognize and kill the cancer cells that have the targeted antigen on their cell surface.

CAR-T immunotherapy has been used with remarkable success in small clinical trials involving patients with acute lymphoblastic leukemia (ALL), characterized by a specific antigen (CD19) on the surface of the cancer cells. Patients undergoing this therapy had relapsed in spite of standard therapies including transplantation. In one trial of 33 ALL patients, 30 patients (91%) went into remission, and many patients experienced long-term survival. Side effects included neurotoxicity in 20% of patients, due to the release of cytokines when the T-cells encountered their targets. Corticosteroids were used to manage this side effect.

At the present time, this type of immunotherapy is not being used for solid tumors, as there is no equivalent of the CD19 antigen on the surface of these tumors. Additional research will determine if modifications could make this a viable therapy for head and neck cancers.

Some FA patients in the BRCA2 complementation group have developed ALL, and relapse rates following transplantation are high. This type of immunotherapy might provide a therapeutic option for these patients.

Diminished Immune Function in FA over Time



Melinda Butsch Kovacic, MPH, PhD, Cincinnati Children's Hospital Medical Center

"A person is, among all else, a material thing, easily torn and not easily mended" (Ian McEwan). It seems that a person with FA has a particularly vulnerable immune system.

Over the past five years, Dr. Melinda Butsch Kovacic and her team at Cincinnati Children's Hospital Medical Center have collected blood and health histories from over 29 cancer-free individuals with FA ages 2 to 47 years, who have not yet had a bone marrow transplant. The goals of the study are:

- to determine if the numbers and functions of a broad spectrum of immune cells change over time; and
- to assess disease and lifestyle factors that influence the timing and characteristics of the immune defects, in relation to increasing age and stage of bone marrow failure.

The team has already discovered that both children and adults with FA have significant abnormalities in some of their immune cells compared to age-matched healthy individuals. As the longitudinal study continues over time, they hope that the information learned will eventually have predictive and therapeutic value in individuals with FA.

A Promising New Immunotherapy for Head and Neck Cancer



William William, MD, University of Texas MD Anderson Cancer Center

Dr. William William summarized the outcome of this trial at the 2015 FA Scientific Symposium.

In this study, 132 heavily pretreated head and neck

An exciting study of a new immunotherapy drug gives hope for patients suffering from recurrent or metastatic head and neck cancer.

These cancers are currently considered incurable.

Results of a clinical trial of pembrolizumab were presented at the 2015 meeting of the American Society for Clinical Oncology (ASCO).

Dr. William William

cancer patients received infusions of pembrolizumab every three weeks. Nearly 25% of these patients were considered “responders” to this drug, and 57% experienced some level of tumor shrinkage, far greater than expected. Responses were seen in both HPV positive and HPV negative tumors, and efficacy lasted more than 30 weeks in some patients.

Pembrolizumab was well tolerated. The most common side effects were fatigue, rash and itching of the skin.

Could this therapy be effective in treating the head and neck cancers that affect individuals with FA?

Dr. William speculated that FA tumors may be highly mutagenic, a condition that correlates with a higher efficacy of this type of immunotherapy. Unknowns include the competency of an FA patient’s immune system, and how well this special group of patients might tolerate this therapy. These issues need to be explored aggressively, given the few good treatment options for FA individuals with aggressive cancer.

Not the Spitting Images of Their Siblings!



Flavia Teles, DDS, MS, DMSc, University of North Carolina at Chapel Hill

susceptibility to oral cancer. The team examined the collected saliva for small proteins called cytokines. Cytokines are released by immune and non-immune cells and act as “messengers” called intercellular mediators. They have multiple functions,

If you have been to Camp Sunshine in the last few years, you might have been puzzled by the odd sight of FA families spitting into tiny containers. Dr. Flavia Teles and her team have been collecting saliva from FA individuals and their non-FA siblings to examine whether there is a difference in the mouths of FA individuals that could account for their higher

ranging from regulating immunity and the formation of blood cells (hematopoiesis) to controlling tissue growth, migration, development and differentiation. In addition, they have also been linked to the development of oral cancer, inflammation and tissue dysregulation.

In her study of 24 pairs of siblings and 86 cytokines, Dr. Teles found differences in some of these cytokines in the saliva of FA individuals versus their non-FA siblings. The results suggest there is a link between inflammation and the increased risk for oral squamous cell carcinoma observed in FA. The cytokines GRO and MCP-1 seem to be particularly involved. Even though Dr. Teles found this association, further cell and animal studies are needed to determine if the cytokines really do contribute to oral cancer development. This information could ultimately lead to strategies for earlier detection, prevention or treatment for oral squamous cell carcinoma specific to FA.

Until that time, very good oral care is recommended.

Hallo Ciao
이 안녕하세요
Hola Hello Bonjour
Guten tag

The Fanconi anemia community extends far beyond the English-speaking world, but information in other languages is scarce and can be difficult to find. The Fanconi Anemia Research Fund seeks to expand access to information about FA by making it available in other languages. If you speak another language and are willing to translate newsletter articles, please email sherri@fanconi.org. We are especially seeking those who can translate from English into Spanish, French, Italian, German, Dutch, Portuguese and Korean.

Metformin



Qingshuo Zhang, PhD, Oregon Health & Science University

Dr. Qingshuo Zhang reported on a low-cost FDA-approved drug called metformin, which shows promise in its ability to both improve peripheral blood counts and postpone the formation of solid tumors in an FA mouse model.

Dr. Zhang used a *Fancd2* mouse model to show that mice treated with dietary metformin had a mild, but significant increase in tumor-free survival time compared to those consuming a placebo. Furthermore, after six months of metformin treatment, *Fancd2* mice showed significantly higher platelet counts and hemoglobin levels than those that were given a placebo.

Additionally, metformin suppressed the formation of radials and chromosome breaks in cultured FA cells from FA patients.

This work shows that metformin has potential as a treatment option for cancer prevention and bone marrow failure in FA. Pending funding approval, the next step will be clinical trials with human FA patients.

Update on the International Fanconi Anemia Gene Therapy Working Group

By Jakub Tolar, MD, PhD, University of Minnesota Medical School

“I grew up with a real sense that an individual can make a difference, and that, if you can, you should.” The legacy of David Frohnmayer’s commitment to make a difference resonates throughout the Fanconi Anemia Research Fund’s efforts, from the beginning – when he and his wife, Lynn, started a Fanconi Anemia Family Support Group – to now, when FA is one of the best-researched rare diseases. He led with personal and intellectual charisma, and with a leadership style that was always focused on sharing information and working together for the best outcome. His influence can be seen in how the International Fanconi Anemia Gene Therapy Working Group was built, bringing together an integrated team of teams, where the culture of connectivity is deliberately working to unleash the originality and innovation of others. His inspiration is behind the idea that we will have the same platform for gene therapy trials around the world, so that we can combine knowledge, learn more quickly, and make gene therapy for FA the next standard of care.

The International FA Gene Therapy Working Group focuses on increasing the quality of life and survival, decreasing the incidence of head and neck cancer, treating bone marrow failure, and preventing leukemia for these patients. The Working Group has united both clinical and basic science teams. The three teams on the forefront of clinical trials of gene therapy in FA are:

- Fred Hutchinson Cancer Research Center in Seattle, led by Jennifer Adair and Hans-Peter Kiem, who have already initiated treatments with FANCA-modified autologous hematopoietic cells.
- Centro de Investigaciones Energéticas, Medio Ambientales y Tecnológicas (CIEMAT) in Madrid, led by Juan Bueren,

Susana Navarro, and Paula Rio. They have initiated two trials, one in mobilization and collection of hematopoietic stem cells (called FANCOSTEM), and, as in Seattle, a trial of transducing FANCA hematopoietic stem cells with a FANCA lentiviral vector (called FANCOLEN). Their preliminary data suggest that there is a strong proliferation advantage of gene-corrected FANCA cells after xenotransplantation in mice.

- Institute of Child Health, at University College London, where the trial led by Claire Booth, Adrian Thrasher, and Bobby Gaspar has already identified several patients they plan to treat this year.

On the pre-clinical front, Els Verhoeven in Nice and Lyon, France, has developed new viral envelopes that enable transduction of hematopoietic stem cells without cytokine stimulation. Preliminary data show that these envelopes allow correction of alkylator sensitivity in bone marrow cells from *FANCA* patients. Mark Osborn, in Minneapolis, has shown that gene editing of FA cells is possible and can be made very efficient by adapting the design of the gene-editing nuclease. Manfred Schmidt in Heidelberg has been the group’s guardian of off-target effects, and Tim Osborn in Boston has laid the groundwork for expanding gene therapy approaches into non-hematopoietic tissues, namely in the oral cavity.

Dave once said that when your child is stricken with a fatal disease, you can either turn inside or turn outside. We are fortunate that he turned outside and led by example, so that we can turn all the available science—evidence-based, data driven, adaptive, and international—into focused strategies for safe, assertive and efficient gene therapy of FA.

The High Risk of Cancer of the Anogenital Tract in Women with FA



Krupa Patel, The Rockefeller University & Weill Cornell Medical College

Young women with Fanconi anemia are at extraordinarily high risk of developing anogenital squamous cell carcinoma (SCC), especially of the vulva, and these cancers are often fatal. Krupa Patel described the treatment and outcomes of nine FA female patients in the International Fanconi Anemia Registry diagnosed with

anogenital SCC. Patel believes that these cancers are under-reported.

Median age at cancer diagnosis was 29, with a range of 23 to 45 years. At the time of diagnosis, patients' primary complaints included genital warts, vulvar lesions and constipation. Three patients had a history of genital warts and three had used tobacco. The tumor site was vulva in five patients, the vulva and vagina in one patient, the rectum and vagina in two patients and the cervix and vagina in one patient.

Seven of the nine patients underwent surgical removal of the cancer, which was generally well-tolerated. However,

the cancer recurred in four patients at a median time interval of 16 months post-surgery. All three patients who underwent radiation therapy developed significant treatment complications such as low blood counts, severe infections and second-degree burns. Seven of the nine patients died with a median survival after diagnosis of 21 months.

How can these cancers be prevented?

Gynecological cancer screening is imperative. According to our 2014 Guidelines, females with FA should have visual examinations of the external genitalia beginning at age 13. Sexually active FA women at any age, and all females with FA by age 18, should undergo annual comprehensive exams, including Pap smears and HPV testing. A colposcopy and biopsy should be done if any precancerous or cancerous lesions are identified. Even if a Pap smear and HPV testing both come back negative, the screening should still be repeated at least every year. Anyone previously diagnosed with dysplasia should undergo comprehensive gynecologic exams including Pap smears every 4 to 6 months. Obtain the HPV vaccine, abstain from smoking and undergo regular gynecologic screening!

SAVE ^{the} DATE

28th Annual SEPT 15-18, 2016
SCIENTIFIC SYMPOSIUM

Hyatt Regency | Bellevue, Wash.

www.fanconi.org

Meeting for Adults with Fanconi Anemia Inspires Hope and Action



“I can’t thank you enough for the weekend. Truly, for the first time in a long time, my heart is full of hope!”—Adult with Fanconi anemia

Ninety-five individuals from 13 states and 10 countries gathered in Orlando, Fla. for the sixth Meeting for Adults with Fanconi Anemia. From February 26-29, nearly 40 adults with FA came together with loved ones, doctors, researchers, and other experts to connect with each other, attend educational talks, and participate in research opportunities. For some adults, this meeting presented the first chance they had to meet others their age living with FA. The bonding over shared experiences was tremendously impactful, and not just for those with FA: “As a mother, I was brought to tears by the happy, welcoming friendliness of the FA family.”

Over the course of the weekend, speakers delivered both informational and inspirational presentations on a range of topics. Richard Gelinas, PhD, Institute for Systems Biology,

presented about the progress in FA research and potential future therapies. Farid Boulad, MD, Memorial Sloan-Kettering Cancer Center, delivered concrete information about bone marrow transplantation for patients with FA. Another highlight of the weekend was the presentation on healthy living for adults with FA by Margaret MacMillan, MD, University of Minnesota. Support groups took place throughout the weekend, led by psychosocial director of Camp Sunshine, Nancy Cincotta, MSW, MPhil. Fanconi Anemia Research Fund board members Lynn Frohnmayer, MSW, and Sharon Schuman, PhD, gave a motivating talk about the importance of fundraising, leaving attendees inspired to take action: “There was great audience participation and creative new ideas.”

Another exciting aspect of this meeting was the chance for attendees to participate in various research opportunities. Researchers were available all weekend to meet one-on-one with adults with FA. One attendee commented on the benefit of this time, stating “it’s great because it helps [to have] different FA experts be willing to teach what they have learned, and more opportunities to have further care during our journey.” Research opportunities will also be available to FA families this year at Camp Sunshine, June 24-29.

The Fund is overjoyed by the numerous positive responses from the meeting and appreciates the participation of so many adults with FA, loved ones, researchers, doctors, and other experts. A sincere thanks to all who made this weekend wonderfully successful! The next meeting for adults with FA will be in the fall of 2017 (exact dates and location to be announced soon).

Mission: Telling Our Stories



My name is Jasmine Bennetsen. I am an adult with Fanconi anemia. I’ve been fighting it since I was diagnosed 11 years ago. I’ve never attended Camp Sunshine or the adult meeting until now. Being able to meet other patients and the amazing people who support them was one of the most incredible experiences of my life. While I was attending this meeting, we dedicated time to discussing fundraising and how important it was for what the Fund does. It was very inspiring and it made me think “what could I do to help?” That’s when it hit me. Although there are a few videos online and a Facebook page about Fanconi anemia, I feel that we can do so much more to create a global awareness of FA. My idea is to use social media as more of a tool to help create awareness, because without more people standing behind us, we

can only go so far, so fast. That’s why I have made it my mission to travel and meet more patients and their loved ones and have them share with me their stories, their struggles, and their hopes. If anyone would like to participate, please reach out to me by email: Stellic@yahoo.com with a subject titled FA-Video. I invite everyone to take part in this because there is no way I can do it alone.

Together we can make a difference, together we can create a movement, and shed light on this invisible war we are fighting.

Bone Marrow Biopsies and Popsicles

By Wesley Young



I'm Wesley Young. I'm 12 years old and I have Fanconi anemia. Although having this life-threatening disease can be tedious sometimes, there is one thing about it that I absolutely adore. This thing is bone marrow biopsies. If you don't believe me, I can give you a list of reasons why I think they're like Christmas. First off, when I go to sleep, it feels really good because to get there I have to wake up really early. Secondly, when I was younger and went to the hospital, I got a small toy or something from a treasure chest. Next, when I do wake up I get a popsicle, which normally feels good, especially with my sore back. Then, they put me in a wheel chair and roll me to my mom's car. Lastly, when I get home, I get to sit in bed all day eating snacks and watching TV. My mom tells me I'm easy to please, but who doesn't love popsicles and movies?



Your Article Could Be Here!

The FA Family Newsletter would love to have more young people contribute stories. Do you have something to share about your experience with Fanconi anemia? We want to hear from you! You could submit an article, a drawing or painting, a poem, or even a joke. Email it to sherri@fanconi.org or send to our office and you could be in the next issue of the newsletter!

FARF's Annual
Family Meeting
June 24-29, 2016
Camp Sunshine
Casco, Maine

Contact: Marie Sweeten, Family Services Director • 541-687-4658 • marie@fanconi.org

www.fanconi.org
www.campsunshine.org

Our Journey to Diagnosis

By Angela Keaton



On August 18, 2015, I found out that my beautiful daughter, Stella, seven years old, has Fanconi anemia. From the minute I heard those words, my world stopped spinning. My outlook on everything changed. It's very hard to explain. I feel as if I'm walking around in an empty shell. I think that stems from being so helpless against this disease. It's so hard for me knowing there's nothing I can do to fix this for my baby. I'm learning to take things day by day, but I'm still having a very hard time.

I am very thankful for the people who are a part of the Fanconi Anemia Research Fund. This group has helped me so much. I honestly don't know where I'd be without their help and understanding.

Stella has had more than her share of medical appointments, even before we found out about FA.

Now, looking back, almost everything she went through can be attributed to FA.

When Stella was three, she burned her thumb on her sister's flat iron. It just so happened it was the thumb that was a little smaller than the other. No one ever noticed because it was such a small difference. When her burn healed, the doctors noticed she couldn't bend her thumb. They did surgery, thinking her ligament was stuck from the way the burn healed. Turns out, the surgery was all for nothing! Not only did it not help, it made the thumb worse. Now she has discomfort sometimes. We now know this was due to FA.

Stella also had speech therapy from the ages of three to six,

when we found out she had conductive hearing loss. The ENT assumed it was from fluid buildup, so we did the surgery to place in tubes. Again, this was all for nothing. After a CT scan, the doctors realized her hearing loss was due to her bone structure...due to FA.

When she was four, she went through a phase where almost every type of material itched her, so I took her to a dermatologist. During the exam, he noticed the café au lait spots and referred us to genetics (we never did figure out the cause of her itching). The first test they ran was for Neurofibromatosis. When the nurse called and told me it came back negative, I jumped for joy. She then said the doctor wanted to run more tests, but at this point they couldn't tell me which tests. I refused because I felt they were just doing unnecessary tests, and Stella had already been through so much. I wasn't going to allow more poking and prodding without good reason.

This past year Stella wasn't gaining any weight, so I began to worry. I took her to a nutritionist, who reviewed Stella's diet and said I was feeding her well. She also told me that Stella should be gaining some type of weight regardless of what she ate. Once again, they referred me to genetics. This time genetics ran a test for Russell-Silver syndrome. When I researched that, FA popped up and I was terrified that was her diagnosis. Everything fit. Her thumb, conductive hearing loss, café au lait birthmarks, speech problems, and her being so small.

My worst fear was then realized. The doctor said the test for RSS was negative and they were referring me to hematology/oncology because he was afraid all signs were pointing toward Fanconi anemia. Since then, we have learned that Stella will need a bone marrow transplant in the next six months to a year.

It's scary how fast life can change. Living with the diagnosis of FA is definitely a challenge for Stella, myself, and my other children. There are so many little things that went unnoticed before and were taken for granted. Now I try to take more time to notice the little things, appreciate what blessings we have and cherish every moment with my beautiful children.

Stella is only seven but she is very mature for her age. The other day she said, "Mom, I didn't know having Fanconi anemia was going to be this hard, but I see now that I can do it!" She is the toughest little girl I know! With her courage and her strength we will get through this together.

The Sibling's Struggle

By Brian Anderson



“Dad, will I die if I eat peanuts?”

That was Eli, my 9-year-old son. I explained to him the reality of his allergy, just like any other parent of an allergy-laden child would. It was a shock to hear these words from him, yet easily calmed with some discussion. He felt better and I felt great. In my opinion, a peanut allergy is a good problem to have. We can control the danger and, more importantly, the reaction if something should happen. Eventually it will become Eli's issue to manage. And if we succeed in his rearing, he will go on to succeed himself.

However, he does have another burden to bear. One that is not normal. A burden he has yet to fully realize. I remember that age. Life was small. The world was small. I worried about very little. I can only assume Eli feels somewhat similar. Somewhat. You see, Elias is special because he has two brothers with a disease. Due to its extreme rarity, the disease is shrouded in misunderstanding. Even after four years of nonstop study and involvement, when someone asks me to explain it, I stumble.

If you ask Eli about his brothers, he will tell you their blood does not work. He knows as much as he wants to know about it. If he asks, we tell him. He knows his blood can help only one of his brothers. (Eli is a perfect bone marrow match for Isaac). When the time comes to give his blood to his brother, Eli knows we will give him an undisclosed sum of cash for doing so to offset his fear of needles. He knows that his peanut allergy can transfer to his brother through his bone marrow. Fascinating, right? But these are just things he has to wonder about. Possibilities cannot define his reality. Children live in moments much better than we do. So, while I do everything I can to not fall apart at the thought of the future, my son

likely worries more about normal childhood stuff. I take solace in that. He is a strong young human who cares deeply for his brothers. They carry a different bond. One I will never understand.

I work hard and care for my family. I strive to be a better husband, father, and human. But I do worry. I worry about possibilities. As time passes, possibilities become likelihoods. I really do not want to know how it feels when the likelihoods become realities. Unfortunately, I fear someday I will. Worse yet, so will Eli.

We learned recently that Isaac's blood counts are failing rapidly. A transplant feels not so elusive anymore and in beginning to prepare for the procedure, my wife and I asked Eli if he had any questions. This conversation was the hardest yet. The fear rested on his face when we spoke about the time

If you ask Eli about his brothers, he will tell you their blood does not work. He knows as much as he wants to know about it. If he asks, we tell him.

frame and our absence from home. He expressed some jealousy and seemed really bothered that Isaac could lose his hair. I continue to wonder how he can handle all this. If my wife Sultana and I cannot make sense out of this, how can we expect a nine year old to do so? Short answer: we can't. I envy his resilience. We can all agree that every sibling in this unique situation is owed the world for their very special place in it.



facebook

Find us on Facebook at

www.facebook.com/fanconianemiaresearchfund

For Michael



Michael Kwon (1991 - 2015)

By Sarah Baker (aunt)

Michael,
The day you were born,
the Sun and the Moon must have
been jealous.
For you had the biggest heart in the
universe,
but a body that didn't know how to
make blood.

Michael,
I still remember the first time I met
you.
You were not even a year old,
But you were as fast as a lightning
bolt,
Zipping around in a walker with a
huge, drooly grin on your face.

Michael,
When your family came to live
just down the street from us in
Greensboro, NC,
So your brother David could get treated
at the Duke Medical Center,
You were our weekend son, to give your
Mom a short reprieve.
You learned to sing lullabies, play video
games, and do the crab dance that
made us laugh so much.
But most of all, you were a loving 4-year
old boy.

Michael,
We lost your older brother, David,
when he was two weeks past his 8th
birthday,
But you fought on through your bone
marrow transplant at age 15.
The grueling battle against your own
body never dimmed your smile,
Nor your love for life and for those who
loved you.

Michael,
How precious are those moments I
shared with you.
A summer visit in Kentucky where you
played in the creek in the back yard,
A sunset we shared on the front porch of
our home in Maryland,
My 60th birthday party in Seoul where
you had to make an impromptu
speech,

And those Thanksgiving dinners in
Virginia.

Michael,
Defying everyone's expectation, you
grew into a young man.
You'll never know how I loved our time
together in Florida,
You never complained that your spring
break was spent with old aunt and
uncle.
Your touching birthday card for Roger's
80th birthday that put his children to
shame.

You were so happy when you brought
your girlfriend along for a few June
days.

Michael,
Wherever you are now, you must be as
free and happy as you were born to be.
Your failing body could not contain your
soaring soul,
We who are left behind promise you
this,
You will light our hearts bright whenever
we are sad,
For you loved life, and we loved you.

Your forever loving Aunt,
Sarah

In Loving Memory

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

Michael Kwon..... 12/19/91 – 10/13/2015

Anthony Rojas.....12/29/07 – 12/3/15

Vishav Gaurav10/13/93 – 11/10/15

Lucas Krieg.....1/29/96 – 12/14/15

My FA Story

By Yan



I'm 34 going on 35 and I am so glad to have found the Fanconi Anemia Research Fund. I met a lot of people who have shared similar experiences and are going through the same harrowing diagnosis and situations that Fanconi anemia puts us through. It is great to have support for something so few know about.

I was 32 when I was diagnosed with uterine cancer, specifically Uterine Papillary Serous Carcinoma (the rare and aggressive type), stage three. It was devastating, as my husband and I had been trying for a child for many years. After surgery, during which they removed the uterus, ovaries and other areas (including lymph nodes), I stayed in the hospital for another 21 days because the wound got infected. This was in part because my white blood cells were low. At the same time, I was told I had to wear compression stockings for the rest of my life and had to deal with the prospect of lymphedema. I thought I was at the lowest point in my life then and thought about different ways to escape reality.

After my wound healed, the next step was chemotherapy. I went for two sessions and crashed big time. My blood counts were so wiped out that I had to be admitted for another 42 days before I recovered enough to out of the hospital. I was on total parenteral nutrition, had six blood transfusions, almost five weeks of granulocyte-colony stimulating factor (GCSF) injections daily, suffered almost all side effects of treatment you can imagine, and tolerated the thought of impending doom and death during that period.

My aunt (who was going through lung cancer at the same time) consulted her doctor to understand what could have caused such a massive drop in my cell counts while hers had always been excellent even through her rounds of chemotherapy. The oncologist suggested genetic testing for Fanconi anemia. Back at the hospital, I mentioned this to my team of doctors but was brushed off. They mentioned that FA was so rare that I need not bother to test for it at all, and

that all my symptoms were chemotherapy induced, yet they remained perplexed as to why my counts dipped so low and took so long to recover.

Just before I was well enough to be discharged, my oncologist mentioned that as soon as my counts came back up to normal, they would continue with the same chemotherapy regime minus carboplatin. I was so afraid that the same episode would repeat itself, so I decided to change to another hospital. When I got discharged, I consulted with my aunt's oncologist and he sent my blood samples to be tested for FA. The results were inconclusive.

At a different hospital, my blood counts were monitored monthly and I had pet scans every six months. It took three years for my blood counts to get back within normal range. I had a relapse last September and had a single lymph node removed. I was lucky it was contained. However, the doctors were eager to start on chemotherapy again. I asked the new doctor about testing for FA and yet again, was told it was very rare and that she had only come across one patient with FA. I wanted to be sure I would not suffer the same distress as before, so I got a second and then third opinion and decided to repeat the genetic testing for Lynch Syndrome and FA. Thanks to my geneticist at yet another hospital, my diagnosis of FA was confirmed.

I've had good days and I've had bad days, but for the sake of sanity and all the people who love me, I have decided and make it a point every single day to be grateful, to be happy, and to still be me.

I've since had another relapse and the cancer has spread. And yet, I'm in a good place because I am surrounded by the best doctors and care, my loving family and husband, love from my brothers and sisters in Christ and support from my FAmily.

I resolve to live and enjoy life to the fullest despite my circumstances. I believe we all have what it takes to conquer all odds and make life worth living. Cancer and FA have definitely changed my life. But for better or for worse, we all have to make a conscious effort in deciding which route we take in life. I've had good days and I've had bad days, but for the sake of sanity and all the people who love me, I have decided and make it a point every single day to be grateful, to be happy, and to still be me.

How FA Inspired Lifestyle Changes and a Fundraising Artist

By Carol Franzen



Abbey was diagnosed with Fanconi anemia one day after her sixth birthday. My husband, Daryn, and I had never heard of FA. Besides Abbey's short stature, she looks like a healthy eight-year-old child. No one could know that her platelets are dangerously low or that being tired is an everyday norm for her.

Fanconi anemia is such a devastating diagnosis because it affects the entire body. With this in mind, our family changed many of our everyday habits. Abbey is now on an all-natural and high antioxidant diet, as antioxidants have repairing properties. We avoid artificial colors and flavors, preservatives, fried foods, boxed foods, and processed meats. We limit gluten and refined sugar, using honey and maple syrup in most recipes that call for refined sugar. We avoid ingredients like carrageenan, soy lecithin, partially hydrogenated oils, high fructose corn syrup, and disodium EDTA, and many more. We do not buy anything without first checking the ingredients. We avoid skin products that contain sodium laurel sulfate and parabens. These ingredients are commonly found in soaps, shampoos, and toothpaste. Because there are limited products that do not have these ingredients, or they are too expensive, we make our own

versions. We make our own toothpaste, laundry soap, and hand soap. We can make the hand soap antibacterial by adding a few drops of tea tree oil. We use Shea Moisture shampoo (adding 2Tbs of baking soda and drops of lemon) and Shea Moisture bar soaps. We use coconut oil for a moisturizer and use homemade deodorant by a local merchant. We are trying to limit Abbey's exposure to things that could cause her body harm over time.

Outside of her strict diet, routine blood tests and hospital visits, we try to live as normally as possible. Abbey takes ballet twice a week. She loves it and has a recital coming up in May. Abbey has a younger brother named Drew. They are about 17 months apart and have a very close relationship. In an effort to make the hospital visits less disruptive to normal routine, and in the best interest of Abbey's care, our family decided to move four and half hours closer to Cincinnati Children's Hospital.

Last year, Abbey and I came up with the idea of selling her artwork to raise funds for research. Abbey loves to paint. Recently, she was the featured artist at our local art gallery. She painted 34 pieces of artwork and sold all but three paintings! Abbey was very proud of her accomplishment. She raised a total of twelve hundred dollars for the Fanconi Anemia Research Fund (FARF) and Abbey's Army Medical Fund, which was formed in Altamont, the small community we moved from in Illinois. Abbey's Army now fights from Illinois and Ohio.

We will be traveling back to Altamont for International Fanconi Anemia Awareness Day. Abbey's Army will be a vendor at a local winery Saturday,

April 30 and Sunday, May 1. Abbey is currently working hard on her artwork. Again, proceeds from the sale of her paintings will be split between the Fanconi Anemia Research Fund (FARF) and Abbey's Army Medical Fund. Wish her luck!

Learn more about Abbey's Army and see some of Abbey's artwork at www.abbeyarmy.org!

Franzen Family Recipes

Toothpaste

Yields about one tube

2 Tbs. Baking Soda
3 Tbs. Coconut Oil
15-20 Drops Peppermint Oil
5 Drops Liquid Stevia

Admittedly, the toothpaste does not taste the best. It took a few weeks for the entire family to get used to this version. I put it in a small container and use a miniature spoon to put it on the toothbrush.

Laundry Soap

Yields about sixty loads using 4-8 teaspoons.

2 Cups Borax
2 Cups Washing Soda
1 1/3 Dr. Bronner's Castille Bar Soap

Freeze bar soap, as this makes it easier to grate. Grate as fine as possible. I use the small medicine cups that come with the children's Tylenol. I normally use 1 1/2 to 2 scoops.

Foaming Hand Soap

1/5 Cup Dr. Bronner's Liquid Castille Soap (We've used Peppermint, Orange and Almond)
4/5 Cup Distilled Water
5 Drops Glycerin (optional)
5 Drops Tea Tree Oil (optional for Antibacterial)

Cute foaming hand soap dispensers are available from Marshall's for \$6.99.

Four Artists, One Cause

By Crystal Pepper



When our son, Carter, was diagnosed with Fanconi anemia in 2007, my husband Josh and I were determined to do everything we could to raise money for the Fanconi Anemia Research Fund. Our efforts were placed on hold shortly thereafter, when Carter's counts began failing and transplant was evident. Carter underwent a bone marrow transplant in 2010 and for the next few years, all of our time and effort were spent getting him "well".

Overall, Carter has been doing relatively well for the past little while now, so again we began racking our brains trying to find a way to make a difference. Josh is a singer/songwriter who has been writing and performing music for the majority of his life. A good friend of his, a member of a national touring band based out of Raleigh, N.C. contacted him about Paper Clouds Apparel Company and their founder Robert Thornton, whom he met at a show in Phoenix, Ariz.

Paper Clouds was formed to showcase the creative minds and artistic abilities of those with special needs while raising funds to provide financial support for special needs schools and organizations. Ecstatic, Josh and I awaited a call from them. When Josh originally spoke with him, like most anyone that is not directly affected, Robert was completely unaware

of Fanconi anemia. Together we came up with a plan and the fundraising efforts began.

Josh and I sent a general request to everyone via the Fanconi anemia family Facebook page and requested drawings from FAmilies, with an emphasis on getting a few that were music related. Once the drawings were compiled, we sent them to Paper Clouds where a panel of judges chose the four designs that they felt best showcased the talent and spirit of FA and would make great designs for t-shirts, hats and bags.

For two weeks in late January and early February these designs were displayed at www.papercloudsapparel.com and promoted through all major social networking sites. We were humbled at the amount of support from FAmilies, friends and neighbors worldwide.

In the end, the campaign raised \$2,078 for the Fanconi Anemia Research Fund and spread awareness across the web to many people who may have never even heard of this dreaded disease.

Throughout the campaign, Josh and I worked closely with Paper Clouds founder, Robert Thornton, and even managed to meet up at the hometown show of Raleigh, N.C. band American Aquarium on February 5, a fitting end to a fundraising effort that was brought together in part by a general love for music and helping people.

Robert has already suggested doing an annual fundraiser for FARF through Paper Clouds and we are looking forward to keeping this relationship going for years to come.

Crystal Pepper, mother to Carter, FA-C (6 years post MUD transplant)



Artists: Matt Morrow (Uncle to Cale Ferrin-FA) (moon music), Ben Bland (brother to Nate-FA) (shark), Mary Jo Fisher Becerra (mom to Israel-FA) (flower), Carter Pepper (FA) (headphones)

Michael's Army Fights for a Cure

By Valerie Monnier-Capone



My name is Valerie Monnier-Capone and my husband is Tony Capone. Our story began in July of 2010 when my husband's nephew Michael Capone was diagnosed with Fanconi anemia at the age of 12. I don't need to tell you how devastated our entire family was, but Michael's parents, Robert and Barbara Capone, didn't waste any time helping to find a cure. They held their first fundraiser within six months.

In 2013, Michael's mother confided in me that fundraising proved to be too much for them to continue, as the disease is very demanding of their time and energy, so I made a promise to Barbara that I would take over the fundraising for them. I'm happy to say that we have had two very successful fundraising events.

Tony and I are blessed to live in the amazing community of Sawgrass in Rehoboth Beach, Delaware. This past year I

I encourage all families to try some type of fundraising. We as healthy adults owe it to all of these young, innocent children affected by FA to do what we can to raise funds so we can find a cure.

formed a host committee with eight of my friends to help with the event. This was so helpful, as I was able to pull from their different talents and resources in our town. This year we designed a Michael's Army logo, we formed a Facebook page (Michael's Army fights Fanconi Anemia) where we posted all of the event information and listed all the businesses that gave donations for silent and live auction items. This page would also link you to a secure Qgiv.com website where people could either purchase tickets to the event or make a donation that would be credited towards our event. This was a tremendous

help because the qgive.com website worked directly with FARF and that was one less thing for me to keep track of. We also asked for sponsors whom we then recognized both online and at the event. **We have been truly blessed by the generosity of I & I Sling. This past year they were our top donor, giving \$11,000, and to date, have donated \$50,000 to our cause.** Also, I can't say enough about my friends on the host committee. It is incredibly hard and time consuming to go door-to-door asking for item donations. Luckily for us, we live in a wonderful town. The business owners were so generous to donate a plethora of items and services.

As for the event itself, it was called "Party Puerto Vallarta Style". We held it at our community club house. In 2013 I was lucky to have been introduced to a generous woman, Gretchen Hanson, who owns a local restaurant (Hobo's) in our town. She graciously prepared an amazing buffet of island style food, basically at her own cost. We have a spectacular Italian restaurant in town (Touch of Italy) and owner Bob Ciprietti donated mouthwatering homemade cookies. We also reached out to a local favorite "Tune Your Palate" with Mr. Paul Cullen (former bassist for Bad Company). He was gracious enough to play acoustic music for us. We found a retired auctioneer, Mr. Frank Craghen, who kindly donated his time as well. We ended up with over 100 guests, all family, friends and neighbors. The evening was filled with laughter and friendly bidding. It's heartwarming to see how everyone was happy to open their wallets for such a good cause. **In the end, we raised over \$45,000!**

I encourage all families to try some type of fundraising. We as healthy adults owe it to all of these young, innocent children affected by FA to do what we can to raise funds so we can find a cure. The FARF team will be a tremendous help to get you started. I had absolutely no idea how or where to begin. I started by asking a couple friends to help. As we talked with people and asked questions, we were surprised with how many people wanted to help. I think you'll find that fundraising is happening all around you and once you start asking, there will be people in your area that want to help as well.

Valerie Monnier-Capone, Chairman, Michael's Army

Host Committee: Christine Cox, Jo Ann Gibbons, Margret Feola, Mary Poulis, Karen Gustafson, Lori Stonton-Dinger, Wendy Maclay and Sheree Davis.

Lucky Duck Foundation Supports Fanconi Anemia Research and Education



The Fanconi Anemia Research Fund would like to extend our most sincere thanks to the Lucky Duck Foundation and its founders, Pat and Stephanie Kilkenny, for their tremendous continued support. Their very successful Swing & Soirée event last September in San Diego, Calif. made the Fund the extremely lucky recipient of more than \$300,000! Fund co-founder Lynn Frohnmayer attended the event and accepted the donation on behalf of the organization (pictured right). During the evening, the Kilkennys recognized the remarkable contributions of Fund co-founder David Frohnmayer, as well as Mark Sciarretta, a long-time supporter of the Fund, both of whom passed away in 2015

Established in 2005, Lucky Duck strives to raise funds and awareness for charitable causes. Since then, the Foundation has donated more than \$1,000,000 to Fanconi anemia research and education. In the past year, Pat and Stephanie Kilkenny reinforced not only their commitment to the Fund, but also to cancer research in general, in part to honor Dave Frohnmayer.

To read more about the Lucky Duck Foundation and its support of the Fund, see the FA Family Newsletter issue #55, pg. 17.



Lynn Frohnmayer, Stephanie and Pat Kilkenny

7th Annual International Fanconi Anemia Day



FA Day, which is really FA month, is a fundraising and awareness effort initiated and carried out by FA families. In 2015, 27 families from 14 states and five countries raised an impressive total of \$150,000! The effort, time, and care that families dedicated to FA Day were the reasons it was so successful. It's this same motivation and spirit that will continue to propel FA research forward and increase family support. This year, let's reach even higher and aim to raise \$200,000. The FARF team is ready to assist, including making

flyers, mailing letters, sending banners and brochures, promoting the event, and sending tax-receipt letters.

Once you have decided on a fundraising idea, visit www.crowdrise.com and create a page with just a few clicks. Choose FARF as the recipient charity, and donations will come directly to the Fund. To find out more about FA Day fundraising, visit www.crowdrise.com/2016FADay, email info@fanconi.org, or call the office at 541-687-4658.

Here are some of the Fund's favorite FUNdraising ideas:

Events

- Mystery dinner theater
- Dodge ball tourney
- Baking/cooking competition
- Bowling competition
- Talent show or Dance for a cure
- BBQ or block party
- Casino night

Sales & auctions

- Bake sale
- Raffles
- Totes/tees
- Basket auction
- Dinner & dessert auction
- Yard sale

Creative asks & pranks

- Envelopes (at church or any group gathering)
- Gameshow
- The un-fundraiser
- Scavenger hunt
- Xbox challenge
- Flamingo flocking



The Fanconi Island Red Hots

Fanconi Canada's Red Hot Team

Fanconi Canada was an official charity of the Scotiabank Toronto Waterfront Marathon/Half Marathon/5K held October 16, 2015. The event attracts over 26,000 participants from 50 countries and shuts down most of downtown Toronto. Fanconi Canada fielded a team of two dozen people, called the Fanconi Island Red Hots, including members of the Lauzier, Collings, Lana, Bentley and Shelson/Waxberg families. Together, they raised over \$20,000 for FA research! This year's run will take place October 16. Contact Fanconi Canada to get involved! www.fanconicanada.org



The FARM team at the Citizens Bank Pell Bridge Run

Run Brings Together Families and FA Researchers

Thousands of Rhode Islanders got together for the Citizens Bank Pell Bridge Run on October 25, 2015 in Newport, R.I. In the previous year's race, the Fiaschetti Family was the only FA family to participate. In 2015, thanks to the FARM Facebook page, another Rhode Island Family, the Emingers, connected with the Fiaschettis to join the cause. Team FARM continued to grow, as three FA researchers joined in from the lab of Niall G. Howlett, PhD, of the University of Rhode Island. Not only did the run give two Families the chance to connect, it gave FA researchers the opportunity to meet those their research may one day benefit. In total, Team FARM raised \$12,220 for FA research and family support! This year's race is on October 23. Registration opens May 1: www.pellbridgerun.com



The Nashville Community High School Hornets

Bump, Set, Score!

The Nashville Community High School Volleyball team in Nashville, Ill. gives back to the community every year by supporting a cause during one home game. This past October, they sponsored a "Volley for a Cure" game to benefit the Fanconi Anemia Research Fund. FARM was chosen by the senior team members to show support for their fellow sophomore team member, Sydney Brinkmann, and her brother, Zach Brinkmann, who have Fanconi anemia.

The team organized a special Fanconi anemia t-shirt sale that profited \$800 before the game even began! They also held a bake sale and basket raffle, as well as selling FA awareness bracelets. All together, they raised \$1,500! Becky Brinkmann, mom to Sydney and Zach, said the family was "very humbled and appreciative of the team's support." FARM shares their appreciation! Go, hornettes!



Having fun at Austin's House of Mouse fundraiser

Mice Race to Raise Funds

Ready, set, go! The second annual Mouse Race Fundraiser gathered around 100 participants in St. Charles, Mo. last fall for an exciting evening of mouse races, roulette games, raffles, a dice wheel, lottery board, and silent auction. Mike Hilbert, Patti Carter Hilbert, and Marti Pearl organized the event in memory of the Hilbert's grandson, Austin, who passed away in 2013. Mike and Patti's mouse, named Angel, in memory of Austin, even won the championship race! Austin's House of Mouse raised more than \$7,300 for FA research and family support. Thank you, Hilbert family and all those who put on such a fun event to support the efforts of the Fund!

FA Family Holiday Letter Campaign

Last fall, the Fanconi Anemia Research Fund called upon FAmilies to participate in a holiday appeal letter campaign. Thirteen FAmilies enthusiastically responded, together raising an impressive \$50,000 for FA research and family support! We would like to acknowledge and thank the following FAmilies for participating: Altmann-Morrison, Brannock-Cazzari, Di Mercurio, Fiaschetti, Franzen, Horrigan-Levine, Janock, Nori, Nunes, Pearl, Sacks, and Sysak-Petersen. The holidays are a busy time for everyone; we appreciate the time and effort that went into writing heartfelt letters to families and friends.

We would also like to thank co-founder Lynn Frohnmayer for her participation in the holiday appeal letter campaign, and of course, all those who donated to honor Dave Frohnmayer's legacy, raising an amazing \$320,000 for the Fund.

We will begin the 2016 holiday letter campaign the first week in November and look forward to many participants! Want to get a head start? Call the FARF office at 541-687-4658.



FA families at the first annual FARF Run

1st Annual FARF Run a Success

On a crisp autumn day in late September 2015, two FA families held the first annual FARF Run 5k in Renton, Wash. The dual effort of the Robison and Graham-Anderson families proved very successful, as more than 150 runners came out for the race. In addition, more than 25 FA family members took part in the event, coming from all over the Pacific Northwest and one all the way from New York! By the end of the day, the FARF Run brought a community together to support three young boys and raised around \$8,000 for the Fanconi Anemia Research Fund. Co-organizer Emily Robison was touched by the turnout: "We had so much support from the community, family, friends and FAmily! We hope to keep putting on this event for many years to come." Check out the FARF Run's Facebook page for information on the 2016 race: www.facebook.com/FARFrun

Family Fundraising Efforts

In 2015, FA families raised an incredible \$3,137,815 for the Fanconi Anemia Research Fund! More than 220 families raised funds, with 130 raising over \$500! More than 87 cents of each dollar donated went directly to research and family support, making a difference for all those affected by FA and their families. Sincere thanks to every family and individual who worked so hard to raise funds in honor or in memory of loved ones.

\$1,990,000 and up

Dave, Lynn, and Amy Frohnmayer

\$100,000 – \$215,000

Fanconi Canada
Kendall & Taylor Atkinson Foundation
with the Nash and Atkinson Families

\$46,000 – \$84,000

Robert and Barbara Capone
John and Kim Connelly
Steve and Jennifer Klimkiewicz
Kevin and Lorraine McQueen
Glen Shearer and Peg Padden

\$20,000 – \$37,000

Matthew and Evelyn Keyes
Tim and Mary Ann Lana
Todd and Kristin Levine
Kevin, Katie, and Chloe Rogers
Gerard and Cynthia Vandermeys

\$15,000 – \$19,999

Rachael Alaniz and Kevin Gatzlaff
Kerrie Cazzari
Mark De Groot and Hanneke
Takkenberg
John and Martina Hartmann
Brian Horrigan, Amy Levine, and Delia
Levine-Horrigan

\$10,000 – \$14,999

Jimmy and Jenny Armentrout
Owen and Cindy Bagaason
Herminia Carvalheira and Claudia
Fernandes
Justin and Britteny Ferrin
David and Mary Ann Fiaschetti
Andre Hessels and Rutger Boerema
Kaps for Kendall
Orion and Lisa Marx
Bob and Andrea Sacks

\$5,000 – \$9,999

Chris and Jennifer Branov
Joseph and Nancy Chou
Chris and Susan Collins
Susan and Skip Gannon-Longstaff
Alan and Rachel Grossman
David Guidara and Ceresa Family
Patti and Mike Hilbert
Charles and Katy Hull
Andries and Helga Kruger
Tyler Morrison and Rachel Altmann
Mark and Diane Pearl
Peter and Janice Pless
William and Mary Underriner
Nigel and Ann Walker

\$1,000 – \$4,999

Michael and Jennifer Aggabao
Brian Anderson and Sultana Graham-
Anderson

Jeanne and Ken Atkinson
Israel and Mary Jo Becerra
Biby Family
Randy and Nancy Bloxom
Jeffrey and Donna Boggs
Sean and Allison Breininger
Ryan and Becky Brinkmann
David and Kim Chew
Tom and Mary Eileen Cleary
Stuart Cohen and Deane Marchbein
Daniel and Melinda Coleman
Ana Concha and Alejandra Tabar
Brian and Margaret Curtis
Darel and Kalani DeHaan
Donna DellaRatta
Antonino and Marie Di Mercurio
Chloe Eminger
Ezat and Laila Faizyar
Daryn and Carol Franzen
Andrew and Jennifer Gough
Michael Greenberg
Gary Haftek
Owen Hall and Margaret Kasting
Lisa and John Hayden and Chad
Ramser
Jeff and Beth Janock
John and Karilyn Kelson
Jee-Ai Kim and Sejin Kwon
Daniel Kold
Christopher and Dana Lamb
Mark and Angela Lamm
Gregory and Lynnette Lowrimore
Dan and Nikki McCarthy
Sheila Meehan
Adam and Olivia Mindle
Tony and Lina Nahas
Jack and Lisa Nash
Alice Nicholson
Robert and Mary Nori
Ron and Fredi Norris
Fred and Nancy Nunes
Susan Ortiz
Michael and Joanna Peros
John and Dianne Ploetz
Joel and Jennifer Ramirez
Pedro and Marina Ravelo
Mark Ritchie and Lisa Mingo
Emily and Neil Robison
Les and Nancy Ross
Ron and Alice Schaefer
Bryan and Karen Siebenthal
Jan and Ken Sysak
Esther Thompson
Mike and Beth Vangel
Louis and Theresa Viola
Robert and Julie Williams

Up to \$999

Peter and Donna Abramov
Al and Janeth Acosta
Leighsa Anderson
Juanita and Ron Arroyo

Cherie Bank
Julie and Gerald Barbier
John, Audrey, and Kelsey Barrow
Mark, Linda, and Josh Baumiller
Adam and Marissa Becker
Conrad and Joan Bender
Zachary Blecher, Darryl Blecher and
Diana Fitch
Richard and Tena Bosen
Dale and Chris Bossy
Michael and Diane Bradley
Donald and Danielle Burkin
Colin and Ashley Chorneyko
Bradley and Cynthia Curry
Family of Paige D'Angelo
Richard Day
Charles Deeks
Jeremy and Michelle DellaValle
Wendy Delzell
Scottie and Jessica Dill
DiSandro Family
David Doctor
Brian and Jennifer Dorman
Jonathan and Sharon Drew
Ed and Janice Duffy
David and Kelly Dunnock
Gene and Lynn Eddy
Mir Saleem and Umber Ellahi
Sharon Ellis
Billy Jo and Debbie Estep
William and Terry Estes
Curt and Crystal Fales
Nancy and Scott Finnegan
Israel and Rivka Friedlander
Fabio and Sune Frontani
Liz Funk
Gary and Melody Ganz
Mitzi Gerber
Brian and Lisa Gillott
Anthony Glavac and Sabrina Bowmer
Pat and Maria Gleason
Jessica Grady
Doreen Gummoe
Abdul Hameed
Frank and Kelly Hamilton
Bob and Victoria Hathcock
Greg and Diane Hayes
Jeff Hoffman
Bonnie Hutchins
Shane and Colleen Irvin
Lester and Nancy Jansen
Randy and Lisa Jones
Lila Keleher
Kristine la Cour Rasmussen and Tue
Marker
Kayla Lackey
Martin Lamo
Eddie and Maly Lee
Eugene and Renee Lemmon
Peg LeRoux
William and Amy Lewis
Robert and Darla Lindenmayer

Tanner and Jessica Lindsay
Eric and Beth Losekamp
Donnie and Jerri Lott
Bill and Jackie Lucarell
Kevin and Barbara McKee
Catherine McKeon
Gianna and Lauren Megna
Jim and Holly Miranda
Ian and Tricia Mitchell
George and Sabine Mohr
Louis and Virginia Napoles
Jack and Tammy Neal
Jing Nie and Jian Yang
Michael and Katherine O'Halloran
Michael and Katharine Ormond
Joshua and Crystal Pepper
Lynn and Shirley Quilici
Mario and Yolanda Ramirez
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Brenda Witherspoon
Sean and Kristin Young
Cecelia Zurhellen

Meet the Team



Pamela Norr, Executive Director

Pam's calling to make a difference has shaped her professional path and motivated her to be an agent of positive change. As leader of the Fund, Pam is committed to doing whatever it takes to support

the team and the mission of the organization. Born and raised in southern California, Pam first came to Oregon as an undergraduate at the University of Oregon, and eventually moved her family there. With more than 25 years in the health and nonprofit sectors, Pam is driven by her desire to "leave the world a better place than I found it. I really enjoy being busy and productive and seeing a change. When we've served a Family well, when the team is happy, when we make a difference together, that is a good day's work." A book lover and avocado connoisseur, Pam enjoys discovering Oregon's wonders with her two kids, husband, and adorable westie, Wallee.

Which three words do you aspire to embody? Kindness, caring, selflessness



Marie Sweeten, Family Services Director

A lifelong Oregonian, Marie loves exploring nature with either her running shoes or her camera. She has always been motivated to help others, which led her to pursue a degree in Human Development

& Family Sciences from Oregon State University. With over 13 years of experience in social services, Marie brings resourcefulness and compassion to the team. At the Fund, she dedicates her time to helping Families navigate the Fanconi anemia world in any way she can: "It's a great feeling to be someone's hero for a moment." She strives to make a difference in people's lives and also has a huge heart for animals. In her free time, she enjoys running, yoga, going to the beach, and spending time with friends, family, and her cat, Chloe.

What are you known for around the office? Finding answers and resources. Also, finding the chocolate.



Brad Preston, Scientific Director

Brad is passionate about cancer research and scientific discovery. He studied biology and music at Michigan State University before earning a PhD in Oncology at the University of Wisconsin Mc Ardle

Laboratory for Cancer Research. After postdoctoral training in Seattle, he joined the faculty in human genetics at the University of Utah and then returned to the northwest as Professor of Pathology at the University of Washington and Scientific Director at Amgen. Brad is excited to join the Fund and help advance its important mission. As Scientific Director, he works to focus and accelerate FA research into the clinic. He is inspired by his colleagues at the Fund and the many outstanding scientists around the world working toward a cure. In addition to science, Brad enjoys time with his wife Diana and their two sons and pursuing music, cycling, sailing, time with friends, deductive reasoning and a good book.

Who or what inspires you? Creativity, drive, and encouragement.



Suzanne Planck, Family Services Coordinator

Suzanne is a native Californian who moved to Oregon in search of a greener, more peaceful place to raise her family. She earned her Bachelor of Science in Nursing and a Bachelor's in Nutrition,

then spent 10 years working as a nurse before she joined the nonprofit world. "I believe in working to improve the lives of others and to make a true difference. I'm inspired by true, active compassion; this compassion asks us to get uncomfortable and inconvenienced. I admire those who are willing to put the needs of others before their own desires and comforts." Suzanne is driven by this compassion, working with Marie to support Families by fostering connections, providing educational materials, finding resources, and planning meaningful meetings. A self-professed coffee addict, Suzanne loves spending time with her family and friends, serving the community, and being outdoors. Her perfect Sunday involves taking a nap after a big brunch at church and anticipating her kids coming home for dinner.

What are you known for around the office? Baking goodies! Feeding people – it's one way I show love.



Janae Peverill, Office Manager

Five years ago, Janae packed up her family and belongings and left sunny Florida for the Pacific Northwest, where she and her other half pursued their degrees. Janae earned hers in Business Administration and has exercised

her aptitude for management and human resources at the Fund. Simply stated, Janae keeps the office running. She supports the Fund's overall organizational objectives, oversees the research grant program, as well as the Fund's administrative services. Janae is motivated by organizations and people that serve the community. "What excites me most about my work is the connections I've made with the Families and the team, the abundant opportunities to learn and grow, and the focus on our mission." When she's not working or studying, Janae likes to spend her time hiking, cooking and shopping with her daughter, and trying to keep her plants alive.

Which three words do you aspire to embody? Impactful, effective, passionate



Sherri Van Ravenhorst, Communications and Conference Coordinator

Sherri first fell in love with Oregon when she attended the University of Oregon in pursuit of a Bachelor's in International Relations and Spanish. After graduation, her

wanderlust led her to go explore. She returned to Oregon after spending five years abroad, where she studied foreign languages and earned her Master's degree in Communications. She is responsible for all of the Fund's communications, publications, and conferences. "What I love about working at the Fund is that I'm able to contribute to the common good and be proud of my work. I'm excited by the growth of the organization and honored to be a part of expanding its outreach and impact." A wannabee foodie, Sherri is enthusiastic about trying new recipes and sharing meals and great conversation with family and friends. She spends weekends working on overambitious do-it-yourself projects, reading, or binge-watching a series.

What are three things you have no patience for? Intolerant people, radio commercials, the font Comic Sans.



Helen Taggart, Office Support Assistant

Born and raised in Oregon, Helen comes from a large and lively Lebanese family. Inspired by working with children, Helen pursued a degree in Family and Human Services at the University

of Oregon. Her drive to work for a cause she cares about led her to the Fund. "I'm excited about the work and the environment. The team is incredibly talented and passionate. There are so many people who play a role within the Fund; I'm honored to contribute to the larger picture." Helen brings her positive energy and can-do attitude to the team and makes sure the daily operations of the Fund run smoothly. She's an expert organizer and natural people-person who is goal driven. In her own words: "I whistle, I'm whacky, I get things done." In her free time, you might find Helen trekking up a hillside, running with her husband, or curled up with a book or Netflix.

What and who inspires you? My husband, Tyler, for his kind heart and determination to always work hard. He also always make me laugh!



Abigail Havens, Donor Administrative Assistant

Though originally from Oregon, Abigail traveled all over the country after completing her Bachelor's degree in Interdisciplinary Studies. She even lived in South Korea, where she taught English as a

second language. This sparked an interest in linguistics, leading her to get a master's degree in the subject from the University of Oregon. Abigail manages the donor database and the issuing of tax receipts. "Our donors make the mission of the Fund possible. We're so grateful for their contributions, which really do make a difference. I'm happy to play a role in the work the Fund accomplishes." Around the office, Abi is known for her impeccable sense of style and quiet, biting wit. She's also an avid reader and salsa dancing aficionado.

What are three things you have no patience for? Circular logic, pettiness, and hard-to-peel oranges.

Your FA Research Dollars at Work

From October 2015 to April 2016, the Fanconi Anemia Research Fund awarded \$1,641,008 in research grants to the following projects:

Investigator: Rui Yu, PhD, University of North Carolina, Chapel Hill, NC

Title: *Identifying the nature of the endogenous aldehydes-induced DNA damage that Fanconi anemia DNA repair pathways counteract*

Amount: \$160,000

Investigators: William Fleming, MD, PhD; Markus Grompe, MD, Oregon Health & Science University, Portland, Ore.

Title: *A Porcine Model of Fanconi Anemia*

Amount: \$392,690

Investigator: Anna Kajaste-Rudnitski, PhD, San Raffaele Telethon Institute for Gene Therapy (TIGET), Milan, Italy

Title: *Investigating the impact of lentiviral transduction on Fanconi anemia hematopoietic stem cells for improved gene therapy*

Amount: \$175,000

Investigator: Susan Lindquist, PhD, Whitehead Institute for Biomedical Research,

Title: *How environment shapes the consequences of mutation within the Fanconi anemia pathway*

Amount: \$160,000

Changes to the FARF Board of Directors

The Fanconi Anemia Research Fund Board of Directors held elections at the 2016 Annual Planning Meeting in Portland, Ore. this past January. Newly elected officer positions include: Kevin McQueen, MBA, President; Mark Pearl, MBA, Vice President; and Annette Waxberg, MBA, Treasurer. Sharon Schuman, PhD, will continue as secretary. Lynn Frohnmayer, MSW, now serves as board advisor. Barry Rubenstein, JD, stepped down as board president, a role he capably filled since 2008. He has been on the board since 1998 and remains a member.

Investigator: Jordi Surrallés, PhD, University Autonomá of Barcelona, Barcelona, Spain

Title: *Drug screening and repurposing in Fanconi anemia therapeutics (acronym: REPAIR-FANC)*

Amount: \$160,000

Investigator: Susa Wells, PhD, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio

Title: *Targeting lipid metabolism in FA for the prevention and treatment of squamous cell carcinoma*

Amount: \$250,000

Investigators: Eunike Velleuer, MD, Heinrich-Heine University, Düsseldorf, Germany; Ralph Dietrich, German Fanconi Anemia Family Support Group and Research Fund, Unna, Germany

Title: *Reducing the burden of squamous cell carcinoma in Fanconi anemia*

Amount: \$183,318

The Fund is committed to supporting research to further our mission of finding new treatments and a cure for Fanconi anemia. Over its 27-year-history, 212 research grants and two service grants have been funded to 114 investigators worldwide. The total amount of research dollars awarded is over \$18 million!

Online Fundraising Tools Available

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



Donate While You Shop on Amazon

AmazonSmile donates 0.5% of the purchase price of eligible products to selected charities. Visit smile.amazon.com, select the Fanconi Anemia Research Fund as your charity, and start shopping!

RETURN SERVICE REQUESTED

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

Use of Logo

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

Editors' Note and Disclaimer

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

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Suzanne Planck, Family Services Coordinator
Bradley Preston, PhD, Scientific Director
Marie Sweeten, Family Services Director
Helen Taggart, Office Support Assistant
Sherri Van Ravenhorst, MA, Communications and Conference Coordinator

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HOW YOU CAN HELP

Donations Online: Donate via the heart button on the Fund's website (www.fanconi.org) or through www.networkforgood.org or www.paypal.com

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

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

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