



FAMILY Newsletter

Spring 2018

A semi-annual publication of the Fanconi Anemia Research Fund

issue
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Calling all Families & Friends of the Fund: FA Day is May 1 and we need you!

By Peg Padden, FA Parent

It's not too late to get involved! FA Day is a day (ok, month) where FAMILIES all over the world do whatever they can, to raise whatever they can, for research of this cruel disease, better treatments, and someday what we all dream of... a cure! How will it happen? Only one way: research. Without a doubt, we all have hope because of research. It has brought much progress these last 30 years and we all know we need it to bring more. Unfortunately, it takes money. A lot. And since we're an orphan disease, we get little federal funds. Almost all the money for research

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Two Clinical Trials Funded

Pilot Study of Metformin to Treat Fanconi Anemia

While allogeneic stem cell transplantation can cure the hematologic complications of FA, transplant is associated with potential short-term and long-term risks, and has been associated with an increased risk of solid tumors. Safe and effective oral therapies to treat or prevent marrow failure and cancer are urgently needed. With this goal, collaborative laboratory studies by some of the leading FA investigators, including Dr. Markus Grompe, Dr. Alan D'Andrea, Dr. Grover Bagby, and others, have been ongoing for over a decade. These lab studies have identified a few candidate compounds and molecular pathways that protect against DNA damage in FA models and improve hematopoiesis. The most promising of the compounds studied to date is metformin.

Metformin (N,N-dimethylbiguanide) has been shown to be a safe oral medication widely used for many decades to treat high blood sugar. Recently, there has been renewed interest in metformin given its anti-oxidant properties, aldehyde scavenging capability, and potential cancer-protective effects. A decreased incidence of breast cancer, lung cancer, colorectal cancer, and hepatocellular carcinoma has been observed in patients treated with metformin. There are ongoing trials testing metformin both for cancer prevention and cancer treatment in the general population. Promisingly, preclinical FA studies have demonstrated that metformin protects against DNA damage, and improves blood counts while also delaying tumor formation in mice with Fanconi anemia.

Based on these data, researchers at Boston Children's Hospital have developed a pilot study of metformin for patients with FA.



Akiko Shimamura



Elissa Furutani

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Two Clinical Trials Funded

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This study is now open. Eligibility criteria include age 6-35 years and at least one of the following cytopenias: hemoglobin <10g/dL, platelets <100k/uL, ANC<1000/uL. The study will assess improvement in blood counts after six months of treatment. This pilot trial also incorporates biological studies investigating the effect of metformin on DNA damage and oral cancer risk. The results of this study will inform potential future trials investigating the long-term use of metformin for cancer prevention or marrow failure prevention, as well as combination therapies with androgens or other agents to treat marrow failure, or as adjunct therapy in combination with transplant to reduce cancer risk.

Quercetin Chemoprevention for Squamous Cell Carcinoma in Patients with FA

Excessive toxicity from chemotherapy and radiation makes treatment for squamous cell carcinoma (SCC) in FA quite challenging and leads to negative outcomes in most patients. There is clearly a need for a new approach for prevention and/or treatment that has fewer and less severe side effects. A previous study by Cincinnati Children's Hospital Medical Center showed that the naturally occurring antioxidant quercetin is safe and well tolerated in pre-transplant patients with FA. Quercetin is a plant polyphenol with multiple pharmacological properties including anti-cancer, anti-inflammatory and anti-oxidant effects in preclinical models. Additionally, there was evidence of decreased DNA damage in oral mucosa brushings from patients with FA (pre-transplant) after treatment with quercetin for one month.

Based on these promising data, this new study will examine whether quercetin treatment in post-transplant patients may prevent or delay the development of SCC. Participants in the study will take quercetin for at least six months, with the possibility to take it for up to two years. It is hoped that quercetin treatment will result in decreased oxidative stress and ongoing DNA damage of the mucosa, leading to the elimination or delay of the development of squamous cell carcinoma of the oral mucosa. ■



Parinda Mehta

THE AMY WINN AND CHRISTOPHER T. BYRD
**AWARD FOR ADULTS
WITH FANCONI ANEMIA**
NOW ACCEPTING APPLICATIONS | DEADLINE 7.13.18

The Fanconi Anemia Research Fund honors the inspirational lives of two beloved former board members with the Amy (Frohmayer) Winn and Christopher T. Byrd Award for Adults with Fanconi Anemia (FA). Individuals with FA aged 17 or older are welcome to apply.

Chris and Amy set high goals, devoted their time and energy to making a positive difference, and

lived their lives enthusiastically in spite of the challenge of FA. This \$5,000 award is given annually to someone who, like Chris and Amy, is striving to make a difference and has set high goals for him/herself. Does this sound like you? Would this award help you reach your goals? For more information, including how to apply, visit www.fanconi.org.

Andrew Deans Wins David B. Frohnmayer Award for Early Investigators

An excerpt from Dr. Deans' acceptance speech at the 2017 Scientific Symposium

When I first joined Steve West's lab in London as a postdoc in 2006, I set about learning everything I could about FA. It wasn't until I attended my first FARF meeting in 2008 that I discovered that I had become a part of an amazing community. This meeting and the researchers, clinicians, and families that I met here ultimately inspired me to continue research into FA when I moved back to Australia to start my own lab. Steve suggested I attempt to purify and characterize the FA core complex as part of my new lab's mission. I thank FARF so much for the seed funding that they provided for this project, which then formed the basis for a larger grant from the Australian equivalent of the NIH. My team now consists of 13 people who all work on Fanconi anemia. We mostly study the mechanistic basis of FA – but we're now starting the process of translating our basic discoveries into therapies, with several projects on gene correction, drug screening and diagnostic tests that can all be traced back to ideas that have come from the FARF meetings and some supported through FARF funding.



Lynn Frohnmayer, FARF co-founder, congratulates Dr. Deans

“...be proud that almost every advance in understanding and treatment of FA in the last two decades can be traced back to the influence of FARF.”

I cherish this recognition but point out that my work and the very success of my team is a product of the success of this organization. To all the young scientists and clinicians in the audience – welcome to this amazing group. These few days provide an opportunity to meet lifelong colleagues, test your own crazy ideas on people who know more than you and make connections that will accelerate your research. To those who've been here from the beginning, thank you for sharing your stories, ideas and reagents so freely and congratulations on getting it so right. To families, we're working for you – I'm sorry research and discovery take so long, but be proud that almost every advance in understanding and treatment of FA in the last two decades can be traced back to the influence of FARF. ■

Calling all Families & Friends of the Fund: FA Day is May 1 and we need you! *continued from page 1*

comes from us: FAMILIES who want it with every fiber in our bodies. Amazing and wonderful Lynn and Dave Frohnmayer raised the VAST majority of money all these years.

Now it is up to us.

Please think of what part you can play in this. Whether you're able to put on a fundraiser or simply ask your friends and family to make a donation on behalf of your family, it all counts and all adds up to fund research. We're a small group, but a determined and mighty one. Together we will find a cure. I firmly believe that. If I didn't, I wouldn't be doing this.

FARF has put together a FANTASTIC, easy to use online platform to set up a fundraising page or donate. Check it out: <https://fundraise.fanconi.org/FADay2018>. ■

DONATE WHILE YOU SHOP ON AMAZON

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FARF Executive Director Mark Quinlan and board member Kevin McQueen present Stella Davies with the Distinguished Service Award



Orion and Avery Marx challenge the audience to "do something epic"

29th Scientific Symposium Brings the FA Community Together

The 29th Scientific Symposium took place in Atlanta, Ga. last September, narrowly missing Hurricane Irma's path up the southeast coast. Thankfully, more than 200 researchers and clinicians were able to make it to Georgia to participate

in this crucial meeting. Attendees presented their latest findings on a number of topics, including the FA pathway (p. 16), preclinical models, immunotherapy (p. 11), advances in transplantation, cancer in FA (p. 8-12), genetics and gene editing, and rare disease & drug development.

A favorite part of the Symposium is always the banquet dinner Friday night.

This is an opportunity to acknowledge researchers for their outstanding work and for participants to hear from FA families. This year, Dr. Stella Davies from Cincinnati Children's Hospital Medical Center received the award for distinguished service for her role in pioneering advances in transplantation and improving survival rates.

Dr. Andrew Deans of St. Vincent's

Institute in Australia accepted the David B. Frohnmayer Early Investigator Award with an inspiring speech that had the audience on its feet in applause (see an excerpt from Dr. Deans' speech on p. 3).

Attendees heard from the Kalemba family about the FA diagnosis and journey of now 15-year-old Andie. While FA is something no one wants, they cited their connection with the FA community as one of the best aspects of their journey and their lives. Orion Marx and his 14-year-old daughter Avery, who has FA, then challenged the audience to 'do something epic' to improve the lives of those with FA. For the Marx family, this involved creating Team BrAvery, a group that takes on intense physical challenges – like biking across seven states in one day –



200+

Attendees



42

Oral talks



64

Poster presentations



17

Countries represented



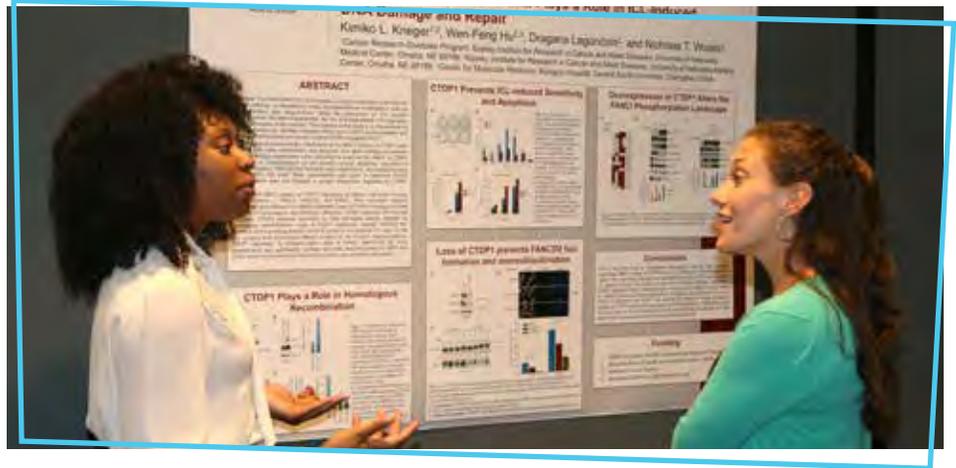
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Family members

to raise awareness for FA and funds for FARF. Matthew Pearl, the winner of the first Amy Winn and Christopher T. Byrd Award for Adults with Fanconi Anemia, shared his FA life lessons and explained to the audience how FA is really a call to action that stands for “Find Answers” and “Fight Always”.

What really made this year’s meeting stand out was the presence and participation of several adults with FA and other FA family members throughout the entire meeting, not just the banquet. The Meeting for Adults with FA was held at the same hotel over the same weekend, allowing these two groups to share time and experiences with one another. This valuable overlap is characteristic of why the FA community is so strong: when people with FA, family members, researchers, doctors, staff and volunteers come together, all voices are heard and each group learns from the others. This is how we move forward, together.

To highlight this significant convergence, for the first time this past year, the opening session of the Symposium was not the scientific FA 101, but rather a panel discussion



Poster sessions inspire discussion and collaboration

bringing FA clinicians and family members together to share their experiences. The panel, appropriately titled “Living with FA: Natural History of Disease & Clinical Perspectives,” set the tone of the entire conference by handing the mic over to FA parents and an adult with FA, who encouraged and reminded researchers and clinicians in the room of how vital their work really is. For a full recap of this special session, see the next page.

This year we will hold our 30th Symposium! It will take place September 27-30 in Newport Beach, Calif. Registration opens May 1 at www.fanconi.org. We hope to see you there! ■

Congrats, 2017 Poster Award Winners

Best basic science:

Kah Suan Lim

Dana-Farber Cancer Institute

Best clinical:

Adam Nelson and

Sonya Ruiz-Torres

Cincinnati Children’s Hospital

Medical Center

Best translational:

Alessio Ligabue

University of Washington



Matthew Pearl shares his commitment to the FA community



Michelle, Stan and Andie Kalemba tell about their journey with FA

Living with FA: Natural History of Disease & Clinical Perspectives



The “Living with FA” panelists (left to right): Stella Davies, Jason Brannock, Dede Harris, Lisa Mingo, Akiko Shimamura, Farid Boulad

Attendees of this year’s Scientific Symposium would be forgiven for thinking they had stumbled onto the set of a talk show, with three comfortable chairs replacing the usual speaker’s lectern at the opening session. Over the course of the next hour and a half, participants gained new understanding of Fanconi anemia across the lifespan, with perspectives of clinicians, FA parents and an adult with FA. In this unique session, clinicians paired up with those affected by FA to share information about the disease during early childhood (0-10 years), late childhood/young adulthood (11-20 years), and the adult years.

Early Childhood

FA parent Lisa Mingo spoke about the challenging process of diagnosis and her family’s decision to switch from a “why us?” attitude to “why not us?” They became proactive in learning about FA, in connecting with other families, and with fundraising. Lisa Mingo and her husband Mark Ritchie have two children, Connor and Dylan. Dylan



was identified as having multiple physical anomalies even before he was born. He was diagnosed with FA at five months old. Now 10 years old, Dylan’s health has been relatively good so far. He has checkups four times a year. Lisa said that while their family might seem “normal” from the outside, she thinks about FA every day. They do try to maintain some normalcy—Dylan stays active, eats well, and goes to school. Lisa and Mark worry about making the wrong decisions and they worry about running out of time. “We’re pre-transplant, pre-cancer. We need all the collective brain power in this room,” she told scientists.

Dr. Akiko Shimamura described the early years with FA from a clinical point of view. The physical manifestations of FA are usually what lead to its eventual diagnosis. Forty percent of those with FA have skin pigmentations, 40% have decreased growth, over a third have issues with upper limbs, 20% have problems with the kidneys and urinary tract, 20% have skeletal issues, 20% have eye issues and 10% have issues with the gonads. By age 40, 90% are in bone marrow failure. Low neutrophils can lead to frequent infections, sending those with a fever straight to the ER. Low hemoglobin can lead to fatigue, headaches, and dizziness. Low platelets can lead to bleeding in the nose, gums, and GI tract, and can even result in a stroke. Bone marrow failure is treated with androgens and/or with stem cell transplant. Other potential major hematological issues are myelodysplastic syndrome (MDS) and acute myeloid leukemia (AML). AML in FA is difficult to cure—survival is better if a transplant is done prior to leukemia. Unfortunately it is hard to predict who will develop leukemia. The *Guidelines for Diagnosis and Management*, available at www.fanconi.org, recommends complete blood counts 3-4 times per year, and bone marrow biopsies once a year.

Late Childhood/Early Adulthood

Dede Harris’ son Trevor wasn’t diagnosed until he was eight years old. Up until that point, Dede felt like she was collecting pieces of a puzzle in an exhausting search to try to understand his medical situation. While the diagnosis was a

shock, it also put those pieces together. However, the relief of having answers didn't last long, as Trevor was already in bone marrow failure when he was diagnosed. A year later he received a stem cell transplant. Like Lisa, Dede spoke to the fear that surrounds making medical decisions. "There are so many decisions to be made [as a parent of] any child, but these decisions are harder and 'more real.'" For example, they started out thinking they would go through transplant with their local doctors, but that team had only done five FA transplants. With FARF's help Dede learned more about other centers, and after meeting Dr. Stella Davies, she realized that by transferring their care to Cincinnati they would have the support and care of a team who really knew FA.

Dr. Davies described the process of a stem cell transplant, from the first few days of chemotherapy (and sometimes radiation) to remove the patient's bone marrow, to the infusion of new stem cells, the wait during the first few weeks for the new cells to start to grow, and within a year, completely new bone marrow. Dede said that watching Trevor go through chemo and cope with mucositis was incredibly hard, but that the return home was a hardship as well. Being in isolation for a year was very difficult for an active nine-year-old boy! Going through transplant impacts the entire family. Trevor and his sibling went to counseling prior to transplant to have someone else to talk to, and to learn tools for relaxation, like breathing and guided imagery. Now that Trevor is in middle school, FA impacts his life in different ways. Like most kids his age, fitting in with his peers is important, so he doesn't talk about FA as much as he used to. On the other hand, his experiences have given him a level of confidence that sets him apart—in a good way!

Adulthood

Finally, Dr. Farid Boulad spoke about FA in adulthood, while Jason Brannock shared his own experiences as an adult living with FA. The past 20-30 years have seen great advances in the treatment of bone marrow failure, MDS and AML. Dr. Boulad anticipates that the next 20-30 years will bring



similar improvements in the treatment of solid tumors. He hopes that the FA community will continue to focus on medical issues, while creating programs for FA adolescents and young adults with FA that include strong psychosocial

support. Dr. Boulad pointed out that doctors see what they think are the issues, but that these may be—and often are—different than what is pressing for the patient. Doctors are trained to attend to the physical aspects of the disease—the blood, the limbs, the endocrine system, the tumors. While the patient's life is influenced by these factors, there are many others that require attention.

“ Dr. Boulad hopes that the FA community will continue to focus on medical issues, while creating programs for FA adolescents and young adults with FA that include strong psychosocial support. ”



Jason brought that message home as he spoke about living with FA. He was diagnosed at five years old and received a matched sibling transplant at age eight. Now he is a fifth-year student at North Carolina State University, studying biochemistry. He spoke about some of the challenges he faces. Short and young-looking, he is often overlooked in the workplace. He counters this by working extra hard to prove himself to his boss and his peers—always saying “yes,” helping his peers when he can, and generally building a reputation for being reliable. “We all have struggles. I try not to let mine be any bigger than anyone else’s.” Outside the workplace, FA impacts his social life as well. Knowing how dangerous alcohol is for those with FA, Jason chooses not to drink, though he feels both peer pressure and internal pressure to do so, to have the connection he feels others have when they drink together. When Dr. Boulad asked Jason what he would tell someone just diagnosed with FA, Jason advised: “Live in the moment. And plan like you’re going to live a long time.” When asked about his fears, there was not a dry eye in the audience when he replied that one was infertility, and the other—more than the pain of cancer or even dying—was leaving a family he started behind. ■

Epidemiology of Cancer in FA



What is epidemiology?

Epidemiology is the study of how disease is distributed in populations. Epidemiologists use data obtained from case reports (retrospective, in the medical literature) or from cohorts (prospective, following groups of patients). Analyses include simple descriptions, crude rates (numbers of events/numbers of participants), cumulative incidences, or competing risks of adverse events.

When was the FA-cancer connection first made?

Cancer cases in patients with Fanconi anemia (FA) were first reported in the 1950s and 1960s. Cohort reports and literature summaries became available in the 1990s, and several cohort reports were published in the last two decades.

What about today?

Dr. Blanche Alter, MD, MPH, National Cancer Institute, delivered a keynote address at our 2017 Scientific Symposium on the epidemiology of cancer in FA. The talk focused on the methods used in the National Cancer Institute (NCI) FA cohort, reported first in 2010 and updated in 2017. The NCI cohort has enrolled more than 160 patients with FA since 2002. To join the study, see p. 15: "Cancer in Inherited Bone Marrow Failure Syndromes."

What can we glean from the updated study?

- The cumulative incidence of severe aplastic anemia (leading to transplant or death) was more than 70%, solid tumors 20% and leukemia ~5% by age 50 years.
- The relative risk of cancer was more than 20-fold higher than in the general population, and the median age was under 40 years.
- The most frequent type of cancer was head and neck squamous cell carcinoma (HNSCC), followed by gynecological SCC.
- Patients who had undergone a stem cell transplantation (SCT) had a ~3-fold higher risk of cancer, at a younger age (median under age 30).
- The HNSCC tumors in the NCI patients were not associated

with human papilloma virus (HPV).

Dr. Alter speculated about possible causes for cancer in FA:

- genetics, including the type of inherited gene and specific mutations in that gene;
- gene by gene interaction (e.g. FA gene and gene for aldehyde metabolism as an example);
- acquired mutations in FA or other genes;
- the role of components of stem cell transplantation: Graft-versus-Host Disease, inflammation, radiation, intrinsic frailty;
- environment, which includes the following: tobacco, alcohol, reactive oxygen species (a natural byproduct of normal metabolism of oxygen, which can increase dramatically during times of environmental stress, leading to cell damage), aldehydes, immunodeficiency, inflammation, and the microbiome.

Important issues include prevention (no smoking or drinking), routine screening, and extra-sensitive screening if there are persistent symptoms. The most effective current management relies on surgery while tumors are still small and have not spread. An ongoing study (funded by FARF) is focused on whether there is an increased risk of cancer in relatives of patients with FA who themselves have one mutated FA gene (carriers). ■

4/20 - 6/15 Accepting abstract submissions
5/1 Registration open

SEPT 27-30

30th FANCONI ANEMIA RESEARCH FUND

SCIENTIFIC SYMPOSIUM



FA Carriers and Cancer Risk

Researchers and FA families have long wondered if FA carriers are at increased risk for cancer. Some genes, long known to be breast cancer susceptibility genes in the non-FA population, were later found to cause Fanconi anemia if both parents harbored mutations in the same gene. Therefore, FA parents who are carriers of these mutated genes are at increased risk of breast cancer. These genes include *FANCD1/BRCA2*; *FANCS/BRCA1*; *FANCI/BRIP1*; *FANCN/PALB2* and *FANCO/RAD51C*.

Do other FA genes increase the risk of cancer in carriers?

Arleen Auerbach, PhD, The Rockefeller University, and collaborators conducted the largest carrier study to date, to determine if FA genes apart from *FANCD1/BRCA2* and its interacting partners (*FANCI* and *FANCN*) give carriers a higher likelihood of developing cancer. Her study appeared in *Cancer Research*, October 1, 2007. Relatives of FA patients (784 grandparents and 160 other relatives) participated in this study. There was no increase in overall cancer incidence in this population of carriers and noncarriers. However, Auerbach detected a significantly higher rate of breast cancer than expected among carrier grandmothers. Among carriers of the common FA genes (*FANCA*, *FANCC* and *FANCG*) only carriers of *FANCC* mutations were found to be at increased

risk. An analysis of all 154 carrier grandmothers revealed 18 breast cancers, significantly higher than the 10.7 expected. This increase was not observed among the 161 noncarrier grandmothers. There were 6 breast cancers among the 33 grandmothers who were carriers for a *FANCC* mutation, compared with 2.5 expected. When all 47 female carriers of *FANCC* (33 grandmothers and 14 other female relatives) were analyzed, the same trend was apparent. Auerbach found three *FANCC* mutations among the 8 *FANCC* carriers who developed breast cancer: p.L554P (n = 1), IVS4 (n = 5) and c.322delG (n = 2). FA parents of Ashkenazi Jewish ancestry are often carriers of the IVS4 mutation, putting them at increased risk of breast cancer. Auerbach concluded that overall, there is no increased risk for cancer among FA carriers. However, in addition to the risk of breast cancer in carriers of

known breast cancer susceptibility genes, there is some evidence that *FANCC* mutations increase the risk of breast cancer.

A second study, appearing in the December 2007 issue of the *Israel Medical Association Journal*, contradicted part of Auerbach's findings. The aim of this study was to estimate the cancer rate, particularly for breast and colon cancer, among Ashkenazi Jewish *FANCC* and Bloom Syndrome (*BLM*) carriers and their families over three previous generations.

Researchers studied 42 *FANCC* carriers, 28 *BLM* carriers, and 43 controls, all of Ashkenazi Jewish ancestry. Controls were participants in a prenatal genetic screening program who tested negative for *FANCC* and *BLM*. Participants filled out a questionnaire regarding their own and a three-generation family history of cancer. In addition to the carriers, the study included 463 relatives of *FANCC* carriers, 326 relatives of *BLM* carriers, and 503 family members of the control subjects. The researchers found no significantly increased prevalence of malignancies among carriers and three generations of their relatives compared to the controls. ■

Oral Cancer Screening Project



Ralf Dietrich, Bruno Araujo, Carmem Bonfim and Eunike Velleuer after screenings at the Brazilian FA Family Meeting last November

Background

In 2013, FARF first funded the oral cancer screening project proposed by Eunike Velleuer, MD, then at Heinrich Heine University, and Ralf Dietrich, Executive Director, German Fanconi Anemia Support Group. The aim of the project is to screen the oral cavities of individuals with FA in an effort to prevent oral cancers and identify lesions that are at high risk for pre-cancer or cancer. During visits with participants, they discuss the importance of oral health care and take brush samples of oral lesions. Experts in Dusseldorf, Germany, examine these samples for DNA

abnormalities that suggest possible evolution to pre-cancer or cancer. Physicians and family members are informed of the findings and the recommendation for a biopsy of the suspicious lesion(s) if appropriate.

In 2017, Velleuer and Dietrich examined the oral cavities of 289 FA participants. Most of these individuals (120) live in the US, but 101 live in Brazil and 68 in other countries. They saw individuals at family or adult meetings, but also visited FA individuals in their own homes. Since 2006, this team has examined the oral cavities of 915 adults and children with FA!

Findings from 2017

Of the 289 FA individuals examined, 35% of participants (n=102) had a total of 231 visible oral lesions; 65% (n=187) had no lesions. Transplanted patients had more lesions than those who had not undergone transplant, but most of these lesions did not raise a concern of pre-cancer or cancer. Of the 231 lesions, 165 were negative (not worrisome); 45 were equivocal with close follow-up recommended, and 21 were of sufficient concern to warrant a biopsy.

Of all 289 individuals examined, only 14 had one or more lesions with “worrisome” findings, which meant that the sample was highly suspicious for cancer or there were early signs of evolution to cancer. Nine of the 14 individuals had been transplanted. They ranged in age from 11 to 38. Five individuals had not been transplanted, and their age range was 16 to 35. In all 14 cases, the recommendation from experts in Dusseldorf was a biopsy of the suspicious lesion(s). ■

Update (August 2018): Following our 2018 family meeting at Camp Sunshine, Ralf Dietrich provided data on 524 FA individuals examined by the German team over a three-year period of time. Of this number, 171 had never been transplanted, whereas 353 had undergone transplant. Transplanted patients had far more visible lesions than those never transplanted, but most of these were not “worrisome.” Of the individuals never transplanted, 7 or 4.1% had worrisome lesions, compared to the 25 individuals or 7.1% with worrisome lesions who had been transplanted. These percentages were different, but given the small sample size, they were not statistically significant.

Advances in Immunotherapies to Treat Cancer



Robert Ferris



Premal Patel

In the last decade we have witnessed huge improvements in the effectiveness of immunotherapies in treating and even curing different cancers. Immunotherapy uses the body's immune system to fight cancer cells. Cancer cells do not die normally, but find ways to survive: they mutate, and they use special signals to hide from the immune system. Immunotherapy drugs are designed to alert the immune system to these cancer cells so it can locate and destroy them.

Using Checkpoint Inhibitors for Head and Neck Cancer

Dr. Robert Ferris, Director of the University of Pittsburgh's UPMC Hillman Cancer Center, gave a keynote address at our 2017 Scientific Symposium on the promise, challenges, and current status of using immunotherapies to treat head and neck squamous cell carcinoma (HNSCC). One category of immunotherapy is called "checkpoint inhibitors." These inhibitors disrupt signals that allow cancer cells to hide from the immune system. By blocking those signals, the immune system can attack the cancer cells. PD-1 and CTLA-4 are protein receptors that function as immune checkpoints. Drugs that block PD-1 and CTLA-4 receptors have been especially effective in freeing the immune system to kill cancer cells.

The FDA approved a PD-1 inhibitor, pembrolizumab, in August 2016 for patients with recurrent or metastatic

HNSCC that had progressed in spite of standard therapy. In a HNSCC clinical trial of pembrolizumab, the response rate was about 20%; some of the responses were long-lasting. Using two different drugs that affect the immune response in slightly different ways could be especially promising. Trials using inhibitors to both PD-1 and CTLA-4 are currently underway.

Dr. Ferris discussed a recent trial using the PD-1 inhibitor nivolumab in combination with liriumab, which activates anti-tumor "natural killer" cells. This trial had a response rate of 24% in patients with HNSCC. Of 29 patients in the study, three had a complete response and four had a partial response. Two of the four partial responses were near complete responses, with an 80% or more reduction in tumor size. This combination of liriumab with nivolumab was most encouraging, because it produced deep and durable responses in some patients.

Checkpoint Inhibitors Improve Outcomes in a Wide Range of Tumor Types

At our 2017 Scientific Symposium, Dr. Premal Patel, Senior Vice President at Juno Therapeutics, described the sea change that has occurred over the past decade in the use of immune checkpoint inhibitors in treating different cancers. A combination therapy of two different checkpoint inhibitors (nivolumab, a PD-1 inhibitor and ipilimumab, which blocks CTLA-4) has revolutionized the treatment of metastatic melanoma, with combination therapy achieving a cure rate of approximately 50%. Another study of nivolumab in patients with metastatic non-small cell lung cancer improved long-term survival in these patients. Dr. Patel noted that cancers with the highest number of mutations did the best. Another PD-1 checkpoint inhibitor called pembrolizumab has been effective in treating some colon cancers, particularly those with high mutation rates. And a small phase 1 ovarian cancer trial using another combination therapy (PD-1 inhibitor plus PARP inhibitor) shows a 20% response rate.

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Critical Thoughts and Future Directions

The recent success of cancer immunotherapy illustrates the power of the immune system to control cancer, but the therapeutic benefit is limited to a subset of patients. We need to understand why some patients are resistant to immunotherapy.

Dr. Patel emphasized the importance of understanding each tumor and the T-cell response to that particular tumor. T-cell inflamed tumors respond better to immunotherapy than non-T-cell inflamed tumors. Also, tumors that express the protein PD-L1 are more responsive to anti-PD-1

immunotherapy than those that don't express this protein. Scientists are working hard to find ways to overcome these barriers to successful therapy.

Dr. Ferris raised the following questions: Should a PD-1 inhibitor be given during the one-month window between diagnosis and surgery? Should we be sequencing the genes in tumors to determine which genes are mutated, since that information might suggest a targeted therapy? Tumors with the greatest number of mutations respond better to PD-1 inhibitors; can this be exploited to treat FA-associated cancers? Combination therapies are more effective than monotherapies,

but involve more toxicity. Should two drugs be given at the same time, or sequentially?

We do not yet know if immunotherapy using checkpoint inhibitors could be effective in treating the HNSCCs that affect FA patients. Enrollment of FA patients in immunotherapy clinical trials and development of an FA Cancer Registry in an effort to document the effectiveness of various therapies are important steps in an effort to answer this crucial question. FARF is working with leading physician scientists to study the feasibility of immunotherapy for FA-associated cancers. ■

Cancer Cells in FA: Observations from The Rockefeller University



At this time, our understanding of how squamous cell carcinomas in people with Fanconi anemia develop is poor. Success in treatment still relies on early detection and surgical removal of small tumors. In order to learn more about progression from normal cells to cancer cells, the team from The Rockefeller University, led by Dr. Agata Smogorzewska (pictured above), performed whole genome analyses of a small number of tumors and tumor cell lines derived from FA

patients. Although the studies are still preliminary, the team made a number of observations:

1. First, the group did not find any traces of the human papilloma virus (HPV) in the specimens by deep sequencing. Instead, they found that a majority of the tumors had mutations in a major caretaker of the DNA, p53. This finding indicates that p53 mutations, rather than HPV, may play a crucial role in the cause of disease in the samples already examined.

2. The second observation was that besides p53, a number of other mutations were present in the tumors, just like they are present in non-FA patients with squamous cell carcinoma. **This knowledge is important because any new treatments that are being developed for non-FA patients might also be useful for FA patients.**

3. Another observation the group made was that overall, the DNA of FA cancers had high numbers of changes within the genome. Some of these changes were very complex, as if the genome was scrambled while the tumor was developing. Although this finding might help explain why the tumors seem to be aggressive in FA patients, it also warrants testing if immunotherapy – which tends to work on cancers that have many changes in their genome – could be successfully used in FA patients. The group is currently assessing this possibility. ■



Scientist Spotlight

Name: Andrew Deans, PhD

Institution: St. Vincent's Institute, Australia

Area of expertise: protein chemistry and cell biology of Fanconi anemia

My work:

We are working on biochemical reconstitution of the Fanconi anemia DNA repair pathway outside of cells, using only FANC proteins. We have so far been able to use this system to find out the exact biochemical function of several of the genes that cause Fanconi anemia. We can also now see what the FANC proteins look like at the molecular level, using powerful new electron microscopy techniques. Our current major goal is to discover what the critical ubiquitination modification* does to FANCD2 and FANCI. This modification is absent in almost all FA patients, but we still don't know its normal function in protecting cells from DNA damage, and people from bone marrow failure. We also have a program searching for genetic and chemical activators of FANC proteins that might one day be able to restore FA pathway function in some FA individuals.

What motivates me to work on FA:

As a scientist, I was originally motivated to work on FA because it results from failure of a very complex system that is very interesting to understand. But meeting families and hearing their stories of heartache and triumph is now easily

my biggest motivator to succeed in my work. The more I have worked on FA, the more I have realized that the work I do in the lab could have important outcomes for FA patients and families.

When I'm not in the lab, you could find me:

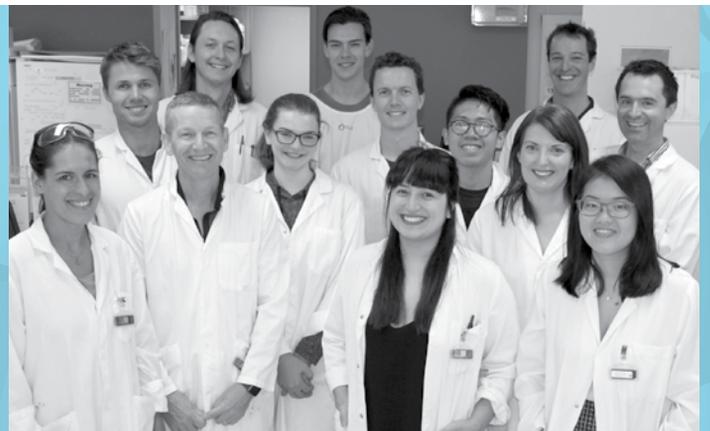
At one of Melbourne's many beautiful beaches or parks with my wife and son.

Just a note:

My research now has 13 scientists working on FA, which would never have been possible without the financial and scientific support of FARF and its network in my early days as a lab head. So, thanks!

* Ubiquitin is a small regulatory protein. It must be added to both the D2 and I proteins, a process called ubiquitination, for them to function normally and repair damage to DNA. If an FA patient is defective in any one of the core complex genes (A, B, C, E, F, etc), the protein products of the genes D2 and I cannot be ubiquitinated, resulting in the DNA repair defect. It is therefore crucial to understand this process. ■

“ Meeting families and hearing their stories of heartache and triumph is now easily my biggest motivator to succeed in my work. ”



The Deans lab

Current **CLINICAL TRIALS** and **RESEARCH OPPORTUNITIES**

New treatments and therapies for people with FA are not possible without research. Listed below are current clinical trials and research opportunities available.

Visit the links listed to learn more about eligibility and protocol descriptions. If you're interested in participating in a clinical trial, scholarships are available from FARF in order to help offset the cost of transportation and housing. Please contact Marie Sweeten, Family Services Director: marie@fanconi.org or 541-687-4658.

National Disease Research Interchange (NDRI)

FA researchers are working hard to find effective treatments and a cure for Fanconi anemia, but they can't do it alone. They need you. Researchers need samples to study, such as tumor samples and biopsied tissue. Please consider donating research material. All it takes is a phone call to FARF and completion of paperwork for the National Disease Research Interchange (NDRI). Contact: Marie Sweeten, FARF | 541-687-4658 | marie@fanconi.org

Tumor Testing Service for FA Patients

The Knight Diagnostic Laboratories at the Oregon Health & Science University specializes in molecular diagnostic testing of tumor tissue that may lead to targeted drug therapy options for patients based on the identification of DNA mutations in cancer samples. The OHSU Knight Cancer Institute, led by Dr. Brian Druker, and the Fanconi Anemia Research Fund have collaborated to make next-generation sequencing available to Fanconi Anemia patients who develop a malignancy. This testing is especially suitable for squamous cell carcinoma of the lung, head and neck, esophagus, and cervix. It may also be used to screen for rare mutations in breast carcinoma and gliomas. The Fanconi Anemia Research Fund will pay for the portion of the test fee not covered by the patient's insurance plan.

www.knightdxlabs.com

Contact: OHSU Knight Diagnostic Laboratories | (855) 535-1522 |

KDLClientServices@ohsu.edu

Marie Sweeten, FARF | 541-687-4658 | marie@fanconi.org

Study of Pembrolizumab (MK-3475) for High Risk Oral Intra-Epithelial Neoplasias

M.D. Anderson Cancer Center, Houston, TX | currently recruiting participants

The goal of this clinical research study is to compare pembrolizumab to standard of care observation (no treatment) in controlling oral pre-malignant lesions. Pembrolizumab is FDA approved and commercially available for the treatment of certain types of melanoma and non-small cell lung cancer. It is currently being used for research purposes in head and neck cancer. FA patients who have not had a transplant and who have a history of oral lesions may be eligible to apply for this trial.

<https://clinicaltrials.gov/ct2/show/NCT02882282>

Contact: Renata Ferrarotto | 713-792-6363 |

CR_Study_Registration@mdanderson.org

Eltrombopag for People with Fanconi Anemia

National Heart, Lung, and Blood Institute (NHLBI), Bethesda, MD | not yet recruiting

Objective: To find out if a new drug, eltrombopag, is effective in people with Fanconi anemia and to know how long the drug needs to be given to improve blood counts.

<https://clinicaltrials.gov/ct2/show/NCT03206086>

Contact: Sophia Grasmeder | 301-827-0367 |

grasmeders@mail.nih.gov

A Study of Prexasertib in Patients with Solid Tumors with Replicative Stress or Homologous Repair Deficiency

Dana-Farber Cancer Institute, Boston MA | currently recruiting participants

This is a research study of a checkpoint kinase 1 (CHK1) inhibitor as a possible treatment for advanced solid tumors that harbor genetic alterations in the homologous repair (HR) pathway or with genetic alterations that indicate replication stress.

<https://clinicaltrials.gov/ct2/show/NCT02873975>

Contact: Geoffrey Shapiro | 617-632-4942 |

Geoffrey_Shapiro@dfci.harvard.edu

Quercetin in Children with Fanconi Anemia; a Pilot Study

Children's Hospital Medical Center, Cincinnati, OH | currently recruiting participants (pre-transplant)

This is a pilot study aiming to assess feasibility, toxicity and pharmacokinetics of oral quercetin (a dietary supplement) therapy in patients with FA and is a first step towards a clinical study of the efficacy of quercetin therapy in delaying progression of bone marrow failure in FA.

<https://clinicaltrials.gov/ct2/show/NCT01720147>

Contact: Stephanie Edwards | 513-636-9292 | stephanieL.edwards@cchmc.org

Gene Therapy for Fanconi anemia

Fred Hutchinson Cancer Research Center, Seattle, WA | currently recruiting participants (FANCA)

This pilot clinical trial will assess the toxicity and efficacy of infusion of gene modified cells for patients with Fanconi anemia (FANCA). Infusion of autologous patient blood stem cells that have been corrected in the laboratory by introduction of the normal gene may improve blood counts in patients with FA.

<https://clinicaltrials.gov/ct2/show/NCT01331018>

Contact: Pamela S. Becker | 206-288-7234 | pbecker@u.washington.edu

Lentiviral-mediated Gene Therapy of Fanconi Anemia Patients Subtype A (FANCOLEN-1)

Hospital Infantil Universitario Niño Jesús, Madrid, Spain & Hospital Vall d'Hebron, Barcelona, Spain | currently recruiting participants

This is an open clinical trial to evaluate the safety and efficacy of a hematopoietic gene therapy procedure with an orphan drug consisting of a lentiviral vector carrying the FANCA gene for patients with Fanconi anemia of subtype A.

<https://clinicaltrials.gov/ct2/show/NCT03157804>

Contact: Julian Sevilla | +34 915035938 | julian.sevilla@salud.madrid.org

Cancer in Inherited Bone Marrow Failure Syndromes

National Cancer Institute (NCI), Bethesda, MD | currently recruiting participants

This is a study to provide information regarding cancer rates and types in inherited bone marrow failure syndromes (IBMFS), including Fanconi anemia. It is a natural history study, with questionnaires, clinical evaluations, clinical and research laboratory tests, review of medical records, and cancer surveillance.

<https://clinicaltrials.gov/ct2/show/NCT00027274>

Contact: Blanche P. Alter | 240-276-7239 | alterb@mail.nih.gov

Pilot Study of Metformin for Patients with Fanconi Anemia

Boston Children's Hospital, Boston, MA | currently recruiting participants

There have been preclinical studies from OHSU suggesting that metformin may improve blood counts in an FA animal model. This clinical trial is being conducted to determine if metformin improves blood counts in people with FA. The study also looks at the effects of metformin on DNA damage and aldehydes. You may be eligible for this study if you have FA and low blood counts, are between the ages of 6-35 years, and have not had a bone marrow transplant. As a participant in this study, you will be provided with metformin for 6 months and your blood counts, other laboratory tests, and clinical symptoms will be monitored while you are in the study. There are 2 required visits to Boston Children's Hospital and compensation for reasonable travel expenses is provided.

<https://clinicaltrials.gov/ct2/show/NCT03398824>

Contact: Dr. Elissa Furutani | 617-632-1978 | Elissa.Furutani@childrens.harvard.edu

Quercetin Chemoprevention for Squamous Cell Carcinoma in Patients with FA

Cincinnati Children's Hospital Medical Center, Cincinnati, OH | currently recruiting participants

In the lab, quercetin, a natural antioxidant, kills tumor cells in FA head and neck squamous cell carcinoma (SCC) cell lines and also prevents development of SCC tumors in non-FA mice. Based on these strong and promising data this study will look at the beneficial effects of oral quercetin treatment for 2 years, in post-transplant patients with FA. It is hoped that treatment with quercetin will result in decreased oxidative stress and ongoing DNA damage of the mucosa, leading to the prevention of, or at least delay the development of squamous cell carcinoma.

<https://clinicaltrials.gov/ct2/show/NCT03476330>

Contact: Stephanie Edwards | 513-636-9292 | StephanieL.Edwards@cchmc.org

TRIALS vs. TREATMENT

It is critical to understand the difference between medical treatments and clinical trials. A medical treatment is a regimen specific to an individual patient and his/her condition, administered by doctors. A trial tests a potential drug, procedure, or medical device in people. Participants in trials play an integral role in determining the safety and efficacy of drugs or procedures. It is important to remember that clinical trials are meant for research, not to administer proven medical care. ■

A Closer Look: the FA Pathway

What is the FA pathway and why does it matter?

The “FA pathway” is a complex, multi-step cascade of genes that protects our cells from chromosomal damage. More than 22 healthy FA genes are required for this pathway to function. When any one of these genes is defective, the pathway fails and the consequence is disease.

At the 2017 Scientific Symposium, 15 presentations were dedicated to the FA pathway (almost a third of all presentations), as well as 19 poster presentations (over a quarter of all posters). This reflects the interest in and importance of understanding the pathway. Every time scientists learn more about the FA pathway, or other pathways that affect FA, we learn more about how the disease operates. This knowledge opens the door for the development of specific, targeted therapies that may one day turn FA into a very manageable condition.

What pathway-related work are scientists carrying out?

Dr. Jung Eun Yeo of South Korea is studying how two different FA genes that were thought to work together to promote DNA interstrand crosslink (ICL) repair actually share some roles and operate separately for others. These experiments used CRISPR/Cas9 gene editing techniques to knock out specific genes whose functions these scientists wanted to examine. They discovered that FANCD2 and FANCI act in concert during DNA ICL repair, but they function separately to accomplish other important tasks. This kind of information shows that FA patients of complementation groups FA-D2 and FA-I may have different responses to current treatments. Understanding better the roles of these and other FA

proteins will be crucial to developing personalized strategies in the treatment of FA.

Sylvie van Twest, working with Dr. Andrew Deans at St. Vincent’s Institute of Medical Research in Australia, developed a biochemical system that can analyze the functional impact of any FA mutation. Their analyses may one day be able to predict the severity of symptoms and, once again, develop more effective personalized treatments.

Dr. Anna Motnenko of the United Kingdom explained how her work in Dr. Martin Cohn’s lab has identified a particular protein, UHRF2, which is a sensor for interstrand crosslink repair. This protein is necessary for recruitment of FANCD2 at sites of DNA damage. Thus it is important for activation of the FA pathway. This study illustrates one of the many links between FA and cancer, in that the protein they have identified is frequently deregulated in various cancers. When FA scientists study it, their discoveries may wind up pushing

the boundaries of cancer research and helping cancer patients worldwide.

In addition to the FA pathway, FA scientists are discovering that other pathways may also repair interstrand crosslinks. Dr. Dan Semlow of Harvard described the discovery of one such pathway, which operates before and instead of the FA pathway, with the FA pathway serving as back up. Dr. Puck Knipscheer of the Netherlands described a possible third pathway. This new information about additional pathways for DNA repair is extremely important and suggests novel approaches for developing new FA therapies.

Therapeutic applications are on the horizon. Dr. Lauren van Wassenhove of Stanford described how certain small molecule activators of an enzyme that detoxifies aldehydes might be able to prevent or delay bone marrow failure, leukemia, and other cancers in people with FA.

The bottom line is that these studies of the FA pathway and other pathways acting in concert hold great promise in the development of personalized treatments that may be able to reverse or prevent the DNA damage that is at the heart of FA. At FARF we are pushing hard to accelerate this research that is happening all over the world, and to promote the development of the therapies suggested by these discoveries. ■

“ The bottom line is that these studies of the FA pathway and other pathways acting in concert hold great promise in the development of personalized treatments that may be able to reverse or prevent the DNA damage that is at the heart of FA. ”



Lucy Grassi

Wading into FA

By Heidi Grassi

“I happy Mommy.”

This is a heartwarming declaration I hear so often from our Lucy. It not only brings a smile to my face but also a tear to my eye as I think about her life ahead with Fanconi anemia (FA). There are many benefits of having learned of Lucy’s diagnosis early—she was just two and a half. The fact that she doesn’t yet have to face the fear and emotional stress of having a life-threatening incurable disease is right there at the top. Right now we get watch Lucy grow, love, embrace life and be blissfully happy. We get to send Lucy to preschool, take her swimming, watch her jump off the furniture—and place her in timeout for doing so. We are enjoying all the things that help make her happy and healthy and normal for the time being. But then

there are the reminders: the quarterly CBCs, the yearly biopsy, the two Pediasure she drinks every day, and the new regimen of quercetin we started in February. These reminders force us to come to terms with the fact that while we may not be treading water, we are wading into her life with FA.

“ The dread at this stage is not for transplant or what might come after, but for the day when Lucy’s ignorance of FA is lost. ”

The dread at this stage is not for transplant or what might come after, but for the day when Lucy’s ignorance of FA is lost, when SHE will be faced with the fear of understanding that she has what is currently an incurable disease that can ravage the body and the spirit. The fear at present is for the

day that we will no longer be able to shoulder that burden of fear for her.

I won’t pretend we are fully prepared for those days but I do feel like we are collecting insights that might help us better predict how she might feel or react and what we can do to make it more bearable. This is in large part thanks to those families and fighters who are at a different or a later stage in their FA journey and have graciously shared their stories. I think we are more aware of what will likely cause Lucy anxiety, like feeling different than other kids at school, or when her CBC counts drop. She might look upon transplant – if that is her fate – with some sense of anticipation, for wanting to feel better and to move past the waiting phase. Perhaps most importantly, I learned that her mental health will need to be as closely monitored as her physical health. The research has suggested and FAmilies have confirmed: children with chronic and incurable illnesses are more vulnerable to depression, and depression is a major factor in suicide. While some may think it should have been obvious to me that this would be a side effect that we might confront, it wasn’t. We have been so focused on the physical concerns that, until others

had shared their experiences, mental health was not even on my radar.

For us, wading into FA does come with a more prolonged period of worry, but it also comes with the benefit of time to learn, to better prepare, and to cherish those “I happy Mommy” declarations all the more. ■



Amelia Hawkshaw

Shock and Relief

By Amelia Hawkshaw

Hi there, I'm Amelia! I live in Sydney, on the east coast of Australia. I'm 25, and I found out I had Fanconi anemia less than two years ago. For many people, learning that you or your child has FA can be a daunting and frightening experience. For me, it was a relief.

It was towards the end of 2016 and I had just been through the toughest year of my life. I grew up not knowing that I had a genetic condition that would affect my future. I lived and laughed, read constantly, learned an instrument, travelled across the world, found my faith, studied and graduated, and had just begun a career in social research. So, when I was diagnosed with bowel cancer in January 2016, it came as a shock. Getting cancer at any age can be a kick in the guts. **At 23, I hadn't yet realized that suffering**

is present in all people's lives, and it would exist in mine.

Two weeks later, I had my first major surgery to resect my bowel and get the tumor out. In April, I would undergo another major surgery with only hours' notice, due to an endoscopic complication. After recovery, we prepared for chemo. And if you know anything about FA, you know what I'm going to say next – chemo didn't exactly agree with me. Two days into my first cycle, I was back in the hospital. My bone marrow and immune system

stopped doing their job. I spent eight weeks in the hospital as my body cycled through several issues, some life-threatening. The doctors were bewildered by what was happening. They had no explanation for my reaction to treatment. I didn't have much capacity for being myself at this time, so I shut myself down and focused on staying alive.

Thankfully, my bone marrow kicked in again and I did get better. I transitioned to a rehab hospital and built up my strength. I began to feel like myself again, and I went home. In November of 2016, my geneticist diagnosed me with Fanconi anemia. After spending 2016 as an anomaly – a patient with an unusual cancer for my age, unexpected symptoms, unexplainable reactions, endless complications, it was comforting to finally have a reason. FA also explained other strange things about me, like how I'm the shortest in a very tall family, my hydrocephalus and endocrine problems, and the café-au-lait spots I am still discovering on my skin daily.

However, the FA diagnosis also came with more confusion. Every patient is unique. And I didn't present like a normal FA patient anyway. There are so many possible manifestations of the condition. We had a lot to learn, and very few specialists in Australia we could contact. On top of dealing

“ I left that meeting with new hope for my future, a clearer idea of how to manage my medical screenings, a lot of new friends, and a tight bond that I feel tugging me across the sea to the next meeting. ”

with the emotional and physical repercussions of cancer, I was learning to live with the constant anxiety and anticipation of hardship that comes with FA. I found just one person with FA living in Sydney (shout out to David!). After the relief I'd felt with a diagnosis, I was suddenly feeling isolated and lost. I was dealing with the unknown once again.

This began to change as I got connected to the Fanconi anemia family. Through FARF and Facebook groups, I gained advice from others dealing with FA, a handbook on the most recent research and treatments, support from fellow patients, and learned of the Adult and Family Meetings. In September 2017, (despite jogging around the hurricane), I attended the Adult Meeting/Scientific Symposium. It was overwhelming and brilliant! I went from knowing one person with FA to knowing over 40! They know exactly what it's like to have a dozen different doctors and some kind of test every month or so. They know what it's like to balance living life in the moment while worrying about possible illness in the future. We spent five days sharing our experiences with FA, giving each other advice about the illness that connected us, talking about

our lives, and having fun. We took advantage of every moment we had hanging out with each other, because next week, we would go back to our own lives. I left that meeting with new hope for my future, a clearer idea of how to manage my medical screenings, a lot of new friends, and a tight bond that I feel tugging me across the sea to the next meeting.

I think back to my family as I was going through the toughest year of my life. I remember that it was family – those who love me most – that got me through. My mum, my dad, my sister. My extended family. My church family. The friends I grew up with. It's family who gets us through. And now my family is bigger.

The FAmily is a strange one to be part of. We are united by common goals – support and a cure. I receive support, and I also support others, because we all go through difficult times at some point. I am often scared for my friends. But I'm also so glad I know them. They inspire me, they make me laugh, they make me want to grab life while I can, because that's what they have learned to do. It turns out there is strength in numbers, even with FA. ■

In Loving Memory

Jessica Paulson Stone
2.18.81 – 12.22.17

Luis Miguel
Morales Torres
12.10.09 – 12.26.17

Imroze Ardeshir
4.15.83 – 12.27.17

Shreyas Vaze
6.12.11 – 1.10.18

Aaron Shelson
5.17.93 – 2.13.18

Registration open
www.fanconi.org

Newport
BEACH
CALIFORNIA

Meeting for Adults with FA
SEPTEMBER 26-29 | Adults with FA 18+ welcome



A group of kids and adults with FA at the Brazilian meeting

New Perspectives at the Brazilian Family Meeting

By Marie Sweeten, Family Services Director

In November, I had the opportunity to travel to Curitiba, Brazil for the 5th Brazilian FA Family Meeting. Matt Pearl and Jason Brannock, two adults with FA who spoke at the Scientific Symposium in Atlanta last September, also traveled to Brazil at the invitation of Dr. Carmem Bonfim to reprise their inspiring talks for the Brazilian families. FARF board president Mark Pearl joined us as well.

While in Curitiba, I got to see first-hand the various resources available for FA families in Brazil. I visited the Hospital de Clinicas da Universidade Federal do Paraná, where nearly all FA patients in Brazil travel to receive care from Dr. Carmem Bonfim and her team. This program is not only a world leader in the treatment of FA, it was also the first place in Latin America to perform bone marrow transplants.

I met with social workers from several nonprofit organizations, including Instituto TMO, which provides support for bone marrow transplantation in Brazil. They

support Dr. Bonfim’s program and invest in clinical research and training of professionals in the field. In addition, they provide aid for patients going through transplant, including those with FA. They run a house called Casa Malice with 22 beds where patients can stay near the hospital before and after transplant, similar to a Ronald McDonald house.

“With 101 individuals with FA in attendance, this meeting was the largest gathering of people with FA anywhere, ever!”

Despite the world class FA program and other resources available to patients, and the fact that there are around 250 known individuals with FA in Brazil, there is currently no formal FA family organization there. However, that may soon change. During our stay in Curitiba, Mark Pearl and I met with two FA parents who are working to establish a Brazilian FA organization. We shared the story of how FARF got started, discussed their needs and challenges, and suggested they start small. FARF will continue to do what we can to support the establishment of the FA organization in Brazil as part of our goal to increase our support of FA families around the world.

With 101 individuals with FA in attendance, this meeting was the largest gathering of people with FA anywhere, ever! It was an amazing opportunity to meet so many Families, hear their stories, and witness all of the wonderful work being done there. ■



Brazilians, Germans & Americans working together to help people with FA

Adults with FA Gather in Atlanta for 7th Meeting

The 7th Meeting for Adults with FA was special in several different ways. Held this past September in Atlanta, Ga., it almost didn't happen at all, as Hurricane Irma moved through the area just days before the meeting was scheduled to begin. Luckily the wind and rain died down and the sun came out just in time. There were 42 adults with FA in attendance, the most ever at a FARF meeting! They came from 23 states and seven countries and ranged in age from 18 to 51. For 18 of them, it was their first time attending an adult meeting. One attendee said "It was a great first time and I loved meeting others like me. The meetings were very insightful."

Another first this year was holding the Meeting for Adults with FA in conjunction with the Scientific Symposium. This allowed for interaction between the researchers and the adults with FA, as well as allowing for the adults to attend Symposium sessions in addition to Adult Meeting sessions. The

“It was amazing to see all the adults and scientists together.”

combination of the two meetings was a big hit with both groups. As one participant said, "It was amazing to see all the adults and scientists together. Very hopeful. Would love it to be together always." Going forward, the

plan will be to hold these two meetings together, which means the Meeting for Adults with FA will now be held annually instead of every 18 months.

Over the course of the meeting, seven speakers delivered presentations on a range of topics. One of the highlights was the presentation entitled "Living our Best Life in the Face of Uncertainty" given by Stella Davies, MBBS, PhD. She inspired the audience with her use of humor combined with practical advice. Her takeaway message for the audience was trifold: (1) free yourself to live your best life—don't waste it—uncertainty is always going to be there; (2) value the people in your life; material stuff doesn't matter; (3) avoid alcohol and cigarettes; take control and participate in clinical trials that will help the FA community.

Another important aspect of this meeting was the chance for attendees to participate in various research opportunities, including oral cancer screenings. Researchers were available to meet one-on-one with adults with FA and address individual concerns and questions. Attendees also had the opportunity to participate in support groups, led by psychosocial director of Camp Sunshine, Nancy Cincotta, MSW, MPhil.

FARF received numerous positive responses from the meeting and



The next Meeting for Adults with FA will be held September 26-29, 2018 in Newport Beach, CA. Registration is now open.

appreciates the participation of so many adults with FA, loved ones, researchers, doctors, and other experts. We are grateful to all who made this meeting so successful! The next Meeting for Adults with FA will be held September 26-29, 2018 in Newport Beach, Calif. Registration is now open. Go to www.fanconi.org or email Marie Sweeten at marie@fanconi.org. We look forward to another great meeting and hope you can join us! ■

FAMILY FUNDRAISING EFFORTS

In 2017, FA families raised an incredible \$3,291,160 for the Fanconi Anemia Research Fund! More than 225 families raised funds, with 117 raising over \$500. Each dollar donated advances 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.

Over
\$3 million
raised!

\$1,558,000 and up

Lynn Frohnmayer

\$587,000 and up

Kendall & Taylor Atkinson
Foundation with the
Nash, Griggs and Atkinson
Families

\$70,000 - \$80,000

John and Kim Connelly
Kevin and Lorraine McQueen
Gerard and Cynthia
Vandermeys

\$35,000 - \$56,000

Peter and Tara Himmelreich
Orion and Lisa Marx
Emily and Neil Robison
Nigel and Ann Walker

\$20,000 - \$31,000

Robert and Barbara Capone
Brannock and Cazzari
Families
Kevin Gatzlaff and Rachael
Alaniz
Steve and Jennifer
Klimkiewicz
Tim and Mary Ann Lana
Todd and Kristin Levine
Tyler Morrison and Rachel
Altmann
Pedro and Marina Ravelo
Peg Padden
Brian and Susan Wiseman

\$15,000 - \$19,999

Adam and Marissa Becker
Mark De Groot and Hanneke
Takkenberg

David and Mary Ann
Fiaschetti
Brian Horrigan and Amy
Levine
Charles and Katy Hull

\$10,000 - \$14,999

Jimmy and Jenny Armentrout
Donald and Danielle Burkin
Herminia Carvalheira
Alan and Rachel Grossman
Andre Hessels and Rutger
Boerema
Ian and Tricia Mitchell
Mark and Diane Pearl
Rick and Lynn Sablosky

\$5,000 - \$9,999

Ryan and Becky Brinkmann
Joseph and Nancy Chou
Susan and Skip Gannon-
Longstaff
John and Martina Hartmann
John and Karilyn Kelson
Daniel and Angie McMahon
Sheila Meehan
Rose and David Pennell
Peter and Janice Pless
Bob and Andrea Sacks
Brian and Jennifer Sadlowe
Bill and Connie Schenone

\$1,000 - \$4,999

Michael and Jennifer
Aggabao
Israel and Mary Jo Becerra
Jeffrey and Donna Boggs
David and Sarah Borden
Richard and Tena Boson

Chris and Jennifer Branov
David and Kim Chew
Colin and Ashley Chorneyko
Ana Concha and Alejandra
Tabar Concha
Donna DellaRatta
Egil Dennerline and Norma
Storm
Antonino and Marie Di
Mercurio
Ed and Janice Duffy
Chloe Eminger
James and Crystal Eubank
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Kasting
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Randy and Lisa Jones
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Sejin Kwon and Jee-Ai Kim
Gregory and Lynnette
Lowrimore
Jim and Holly Mirenda
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Jack and Lisa Nash
Robert and Mary Nori
Ron and Fredi Norris
Fred and Nancy Nunes
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David Ownby
George and Kathryn Reardon
Paul and Rena Rice

Ron and Elesa Schaefer
Matt and Diane Senatore
Bryan and Karen Siebenthal
Jan and Ken Sysak
The Family of Chris Byrd
Joseph and Natalie Vitrano
Anthony and Elisa Walsh
Marc Weiner
Michael and Kim Williams
Robert and Julie Williams
Cecelia Zurhellen

Up to \$999

Peter and Donna Abramov
Al and Janeth Acosta
Juanita and Ron Arroyo
Andrew and Vicki Athens
John and Audrey Barrow
Mark and Linda Baumiller
Jasmine Bennetsen
John and Francene Berglund
Tracy Biby
Randy and Nancy Bloxom
Sean and Allison Breininger
Tony and Monica Cabral
Anita Casani
Jerry and Natalie Christensen
Tom and Mary Eilleen Cleary
Tyler and Teresa Clifton
Daniel and Melinda Coleman
Chris and Heidi Collings
Andrew Coons and Valeen
Gonzales
Bradley Curry and Lea Ann
Stiller
Brian and Margaret Curtis
Michael and Kim Curvey
Darrel and Kalani DeHaan
Jeremy and Michelle
DellaValle
Wendy Delzell
Scottie and Jessica Dill
James and Carol Dillon
Delbert and Linda Dotson
Alex Eddy
Sharon Ellis
Billy Jo and Debbie Estep
Curt and Crystal Fales
Nancy and Scott Finnegan

Holiday Letters

This past holiday season, 17 FA families wrote appeal letters to their families and friends to raise money for FARF. Together, 16 raised more than \$55,000! Thank you to the following families: Griggs, Robison, Di Mercurio, Lana, Altmann-Morrison, Janock, Levine-Horrigan, Vandermeys, Sacks, Franzen, Pearl, Brannock-Cazzari, Boson, Ravelo, and Walker. Of course, we must acknowledge the phenomenal effort by Lynn Frohmayer, who raised more than \$500,000 for FA research! Thank you!

Liz Funk
 Mitch and Erin Furr
 Emmanuel and Dana Gallegos
 Manuel Gaytan
 Brian and Lisa Gillott
 Maria and Josh Godwin
 Hilary Gosztonyi
 Johnathan and Dionne Granados
 Michael Greenberg
 Madeline and Patrick Gregg
 Ben and Stephanie Griggs
 David Guidara
 Mitchell and Tirzah Haik
 Eric and Elisabeth Haroldsen
 Bob and Victoria Hathcock
 Patti and Mike Hilbert
 Tammy and Ben Hilton
 Jeff Hoffman
 Frances Hutchins-Huff
 Lester and Nancy Jansen
 Stan and Michelle Kalemba
 Lila Keleher
 Dan Klug and Elizabeth Bertrandt-Klug
 Kayla Lackey
 Christopher and Dana Lamb
 Martin Lamo
 Ernie and Nancy Landwehr
 Eddie and Maly Lee
 Hynchul Lee and Young Ran Choi
 Eugene and Renee Lemmon

Peg LeRoux
 Tanner and Jessica Lindsay
 Joey Linsenmann
 Michael and Regina London
 Dennis and Sharon Lower
 Bill and Jackie Lucarell
 Deane Marchbein and Stuart Cohen
 Aaron and Nicki Marsters
 Leandre Marx
 Dan and Nikki McCarthy
 Kevin and Barbara McKee
 Catherine McKeon
 Gianna and Lauren Megna
 Naomi Miller
 Adam and Olivia Mindle
 George and Sabine Mohr
 Manuel and Heather Molina
 Sydney and Betsy Moore
 Griff and Cecilia Morgan
 Kenny and Lisa Myhan
 Louis and Virginia Napoles
 Jack and Tammy Neal
 David A. Neumann
 Alice Nicholson
 Jeff and Arianne Pederson
 Michael and Joanna Peros
 Derek and Ginger Persson
 Ashleigh Pinion
 John and Dianne Ploetz
 Michael and Kay Proctor
 Lynn and Shirley Quilici
 Aaron and Jean Randolph

Ben and Shirley Ricker
 Mark Ritchie and Lisa Mingo
 Kevin and Katie Rogers
 Dan and Bonnie Rosen
 Les and Nancy Ross
 Craig and Alisha Rushing
 Maureen Russo
 Erik and Lori Salo
 Mike Sanders
 Richard and Dolores Satterlee
 Ruthie and Sharon Saunders
 Severt and Beatrice Score
 Thomas and Brenda Seiford
 Sharon Saunders
 Jack and Debbie Siegel
 Alain Silverston
 Su Kyoung Sim
 Mokrane Simoussi
 Jim Siniawski
 Kevin and Joanne Smith
 Jessica Speelman
 Adam and Jennifer Stewart
 Scott and Nina Stopen
 Charles and Jennifer Sumrall
 Paul and Debra Sundsvold
 Bruce and Loreen Timperley

Mark and Susan Trager
 Charles and Doris Trotta
 Tom and Kathy Uno
 Mike and Beth Vangel
 Louis and Theresa Viola
 Joe and Wendy Viritto
 Brenda Witherspoon
 Victor Wizman
 Kyle and Madison Wright
 Wesley and Sue Wycoff
 Jian Yang and Jing Nie
 Sean and Kristin Young
 Tom and Marge Zaborney

A note to our fundraisers: we greatly appreciate your efforts to raise money for FARF, and we want to recognize you all accordingly and with 100% accuracy. If we have inadvertently made an error, please let us know by emailing info@fanconi.org. Thank you. ■



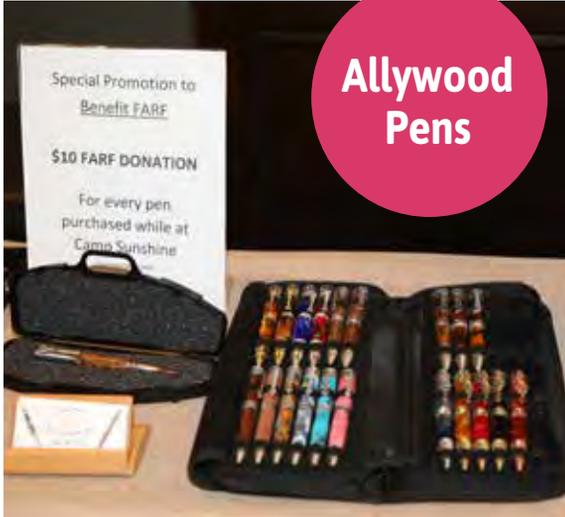
Thank you to all 100+ donors who made this past Giving Tuesday a huge success! Giving Tuesday is a global giving movement built by individuals, families, organizations and communities all around the world. We have two days for deals—Black Friday and Cyber Monday. On Giving Tuesday, we have a day for giving back. This past year, you all helped raise more than \$12,000 for FARF! Look for next year's campaign right around Thanksgiving 2018!

FUNDRAISING SCRAPBOOK

Last August, Sonja Horne knew she wanted to do something special and important for her close friend, Sarah, whose 3-year-old son had recently been diagnosed with FA. A runner and race veteran, she knew she could honor Eli and support his family by raising money for FA research & family services. Sonja created a fundraising page, started a blog, and spread the word. On November 12, she ran the Surfer's Point Marathon in Ventura, Calif. By the end of the race, she'd raised more than \$1,600 for FARF. Thank you, Sonja!



Racing for Time



Allywood Pens

Alex Pearl made her first wood project at age six, started woodshop in 7th grade and continued classes throughout high school. As someone with FA, she was born without a functioning thumb and limited hand muscles and has endured multiple hand surgeries. Now 22 years old, her love of woodworking has blossomed into a business: Allywood Pens. Alex creates beautiful wooden and acrylic ink pens. In her own words, "each pen is made with determination for life, love of the art and hope for more creations in the future." Alex donates a portion of sales from each pen to FARF and recently gave \$1,000. Thank you, Alex! Learn more about the pens at Allywood's website: www.facebook.com/AllywoodPens

Big thanks to St. George's Youth Group of Piscataway, NJ, for their incredible fundraiser in November to benefit FARF. Taverna Night brought nearly 400 people together to raise \$10,000 for FA research & family support! The event was held in honor of 7-year-old FA fighter Thanasis. He was joined by another FA fighter, Shannon, sister of NFL player Scott Simonson, who was Shannon's bone marrow donor. Thanks to all who made this event happen, especially to the organizer, Sophie!



Taverna Night

The KATA Foundation held its annual 'boot stompin' good time' fundraiser, Hoot n' Holler, this past November in Denver, Colo. Between ticket sales, donations, the cow plop, silent and live auctions, wall of wine, and bingo, an astounding \$230,000 was raised for FA research! FARF extends mega thanks to the KATA Foundation and all of the supporters of the Hoot n' Holler for this incredible contribution. 2018's Hoot will be Nov. 10. Visit www.katafoundation.org to learn more.



Hoot n' Holler



KATA 26.2: Running for Their Lives

Last summer, Allison Adams and her life-long friend Emily Krall knew they wanted to raise money to support those with FA and to honor the memory of Allison's dad, Ken Atkinson, brother, Taylor and sister, Kendall. They decided to take on the New York City Marathon on November 5th and made a goal to raise \$1,000 for each of the 26 miles of the marathon. While Allison was training in Colorado, Emily trained in South Carolina, and throughout these months, they shared stories of FA fighters each day. By the time they crossed the finish line in New York, they had exceeded their goal and raised more than \$27,000! Well done, Allison & Emily! Thank you!

In March 2014, Jonathan G. Wiseman lost his battle to FA at age 31. His family writes, "Jon was someone who always had a way of connecting and influencing the lives of others. His strength, courage and bravery always persevered over the hand life dealt him." The JGW Golf Open is an annual golf outing to bring family, friends and the community together to help raise money for FA and cancer research, and to honor Jon's life and legacy. Last year's tournament took place October 2 in Newbury, Mass. The Wiseman family raised \$25,000 for FARF and dedicated the donation to the Walter Lab at Harvard Medical School, where the team uses frog eggs in their FA research. It seemed especially fitting to support this lab, since Jon absolutely loved frogs! Thank you for your support, Wiseman family! Learn about the event here: www.jgwgolfopen.com.



JGW Golf Tournament



Jennifer and Jared Aggabao

Transforming Grief into a Legacy of Love

I am a mother on earth and a mother in heaven. What do you do when your heart is split in two? My kids, Katrina and Jared, are a dynamic duo. Both extraordinary and equally precious, both were also born with Fanconi anemia (FA).

Katrina had challenges from the day she was born. In 2008, at three and a half years old, she developed leukemia. After enduring two years of chemotherapy and its gruesome side effects, she went into remission, only to relapse a few months later. Katrina had a bone marrow transplant in April 2011. She successfully engrafted and her marrow no longer showed signs of leukemia. However, the transplant was just too rough on her already fragile body. On August 30, 2011, at only six-years-old, Katrina earned her angel wings.

I miss my Katrina so much. She always had a positive attitude, was always content. She never felt sorry for herself. What other kids could do, she would show that she could do, too.

She was a brave, vibrant, confident, and loving little girl who was so appreciative of the simplest gifts. A joyful spirit, beautiful smile, and deep faith became her trademark. Katrina was an inspiration for her strength, perseverance, and ability to overcome. Thinking beyond herself during her most challenging moments, Katrina, with a relentless spirit, looked at us and gave us a thumbs-up. That was her last gesture to us, the last moment we saw her awake.

Jared is now nearly 10 years old and is closely watched by FA specialists. He routinely endures bone marrow biopsies, kidney ultrasounds, and brain MRIs. Recently, a cystic structure was found in his brain. At a glance, you wouldn't know he is fighting a life-threatening condition because Jared is always bursting with joy, showing us that each day, even the most challenging one, is a blessing. Jared enjoys dancing, swimming, and riding his skateboard and hoverboard. At

church, he is well-known by everyone as the "mighty lizard hunter." He continually surprises us. Recently, he learned how to ride a bike all by himself in minutes. There is no limit to what he can do because of his drive and "I can do this and never give up" attitude. Although his condition is stable at this time, I know we are racing against the clock.

It is always hard to raise money for a disease like FA that affects so few people directly. It's been a small, private fight, shared with other afflicted families—and with our friends. However, in the last few years, scientists have discovered that FA genes play a key role in the repair of DNA and genomic stability. New treatments are being developed, and breakthroughs are on the horizon, but defeating this illness will require the help of many generous souls.

Every day I wake up, I grieve the loss of my sweet Katrina and the uncertainty of what the future holds for Jared. Someone once said, "grief is really just love with no place to go." I agree but I also know that I can transform it into a legacy. On March 18, in honor of Katrina and Jared, I ran the Los Angeles Marathon to benefit FARF. I have run a few marathons before, and not because I have the love for running. I run because I am a mother on earth and a mother in heaven. As my heart is split in two, I run to honor Katrina's memory and I run to raise funds, in hopes that I am contributing to an eventual cure for Jared. I run for a cause. I run to transform my grief into a legacy of love... ■

The Aggabao family has held several events, raising more than \$24,000 since registering with FARF!

FARF THANKS TWO LONG-TIME BOARD MEMBERS AND WELCOMES TWO NEWCOMERS

FARF would like to extend our sincere gratitude to Drs. David Fiaschetti and Richard Gelinas for their years of dedicated service on the Board of Directors.

David and Rich spent six years as board members, both bringing scientific expertise to the larger Board. We are grateful for their continued participation and guidance on the proposal review committee to help determine the best grants to push FA research forward. Rich will also continue his instrumental role on the Scientific Advisory Board. Thank you, Rich and David!

We are very pleased to welcome friend and supporter Aileen Carlos and long-time FARF fundraiser and FA parent Orion Marx to the Board of Directors.

Aileen is a lawyer based in Portland, Ore. She was introduced to Fanconi anemia while attending law school with Jon Frohnmayer and taking classes from Dave Frohnmayer. She has worked in the legal nonprofit and education sector, and continues to support nonprofits with her legal and dispute resolution experience. Aileen grew up in Pittsburgh, Penn., and continues to be an avid Pittsburgh sports fan despite her distance. She and her partner recently bought their first home, with a fenced in yard that their dog adores. Aileen enjoys hiking and photography, and volunteers for several nonprofits in her free time.

A longtime resident of Sarasota County, Fla., Orion volunteers with numerous local foundations and organizations. Following years of experience in the economic sector, Orion founded Atlas Financial, a firm which helps clients with financial, estate and business owner planning. He and his wife, Lisa, have two daughters, Avery and Violet, and two rescue dogs. An avid sports fan, Orion trains for and participates in various multi-sport competitions, including running, biking, and kayaking. In 2010, he turned these sports challenges into a way to raise funds for Fanconi anemia research. "Team BrAvery" is a fundraising team named for his daughter, Avery. The team takes on extreme challenges such as running two marathons in one weekend, biking across seven states in a day, and pushing a 3,100 pound truck around a two-mile track. ■



PRIVATE FACEBOOK GROUP FOR TEENS WITH FA NOW OPEN

Calling all teens with FA: there's a new, private Facebook group just for you. Teens with FA ages 13-19 are welcome to join. The group is moderated by a few adults with FA. No parents or other adults may join. To join, visit: <https://www.facebook.com/groups/189012454987665/> or contact Marie Sweeten: marie@fanconi.org. ■



Do you want a simple way to increase giving to the Fanconi Anemia Research Fund? Welcome to the PayPal Giving Fund.

EBay sellers are encouraged to give a percentage of their proceeds to a nonprofit certified by PayPal Giving Fund each time they list an item for sale. EBay members can also choose to make an online gift with PayPal. The Donate Now tab lets anyone with a PayPal account donate. For more information, see www.paypalgivingfund.org.

FARF GRANTS AWARDED FEBRUARY – OCTOBER



From October 2017 to February 2018, the Fanconi Anemia Research Fund awarded **\$887,448 to two clinical trials and one research project:**

Investigator:

Parinda Mehta, MD
Cincinnati Children's Hospital Medical Center

Title:

Quercetin Chemoprevention for Squamous Cell Carcinoma in Patients with Fanconi Anemia

Funding Amount: \$75,000

Investigator:

Jacob E. Corn, PhD
University of California Berkeley

Title:

Defining Tractable Approaches for Gene Editing of Fanconi Anemia Hematopoietic Stem Cells

Funding Amount: \$125,600

Investigators:

Akiko Shimamura, MD, PhD & Elissa Furutani, MD
Dana-Farber/Boston Children's Cancer and Blood Disorders Center

Title:

Pilot Study of Metformin for Patients with Fanconi Anemia

Funding Amount: \$686,848

FARF is committed to supporting research to further our mission of finding new treatments and a cure for Fanconi anemia. Over our 29-year history, we have funded 225 research grants, two clinical trials, and one service grant to 149 investigators at 74 institutions worldwide. The total amount of research dollars awarded is over \$20 million!

UPDATE FROM HEADQUARTERS

By Mark Quinlan

Dear Families,

This month marks one year since I joined the Fanconi anemia community as Executive Director of FARF. Together, this community has accomplished so much; I wanted to give you all an update about what we've been up to at FARF headquarters. Over the past six months, we have paid particular attention to how we should position the organization to make significant strides in achieving our mission. This process has been deliberate, thoughtful, and exciting. We have focused the effort on looking strategically at the four pillars of our organization: governance, fundraising, family services, and research.

Under the guidance of the FARF Board of Directors, we as a staff have been working to strengthen the organizational infrastructure to ensure that we have the tools necessary to be agile and efficient in an ever-changing nonprofit environment. We have to make sure we are responsive to the needs of our stakeholders and can communicate efficiently with the FA community. To this end, a couple of our significant achievements over the last six months include implementing a new donor tracking system and the development of our brand new website.

We have professionalized our fundraising efforts. In late 2017, we were awarded a grant to assist in funding the organization's first Philanthropy Director, a position filled by McKenna Knapp. McKenna will bring expertise to our fundraising efforts by focusing on the development of individual donors, strengthening our family fundraisers, and developing a sustained giving program. This new role will allow us to expand systems to maintain and increase our donor base, which, in turn, will lead to increased funding for research and family support services.



The team with visitors John Connelly and Sharon Schuman on Rare Disease Day, Feb. 28th

Two things are apparent in our family services pillar: the number of adults who have FA is increasing, and more and more individuals with FA who live outside the United States are looking to FARF as a resource. In 2018 we will look at ways to better serve these populations. These efforts include developing approaches to better identify the needs of adults and collaborate with international FA support groups to make sure all individuals with FA know where to turn to receive support.

And finally, our research pillar. We started 2018 by providing funding to two clinical trials (read about them on the cover). Our Scientific Advisory Board gained five new members; this revitalized group consists of leaders in their fields who share their expertise and provide vital scientific guidance. We've taken steps to strengthen our scientific and research staffing and are pleased to welcome Isis Sroka, PhD, as our Director of Scientific Operations. We are also exploring the development of a clinical registry. We had our first meeting in the Joel Walker Scientific Meeting Series earlier this month. This important meeting brought experts together to discuss therapeutic approaches for head and neck cancer.

In my first year with FARF, I have learned, I have made new friends, and, I have felt the sadness of losing someone in the Family. Through all these experiences, I have been energized to make sure we move this organization forward with urgency, expertise, and compassion. I am excited for 2018; I hope you are, too. ■

STAFF

STAFF

Mark Quinlan, Executive Director
 Abigail Havens, Development Coordinator
 Sandra Hilliard, Finance Coordinator
 McKenna Knapp, Philanthropy Director
 Suzanne Planck, Family Services Coordinator
 Isis Sroka, PhD, Director of Scientific Operations
 Marie Sweeten, Family Services Director
 Helen Taggart, Research and Operations Coordinator
 Sherri Van Ravenhorst, Communications Director

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Fundraising events are a great way to get involved and make a difference in the FA community. And, they're fun! Check this list to see upcoming fundraisers near you! Visit FARF's website to see more events and follow links to find out more information. Do you know of an upcoming fundraiser? Contact us as 541.687.4658 or info@fanconi.org.

UPCOMING FUNDRAISERS

9th International FA Day
Worldwide
All families

Nina's Starry Night
Portland, Ore.
The Altmann-Morrison Family

50 for Liam
Durango, Colo.
The Towndrow/Balow Family

Your Rope Team
Peru
Bill McCCorey & Friends

May
2018

May 12,
2018

May 26,
2018

June 22-
30, 2018

April 29,
2018

May 5,
2018

May 25,
2018

June 2,
2018

Nov. 10,
2018

Eugene Half Marathon
Eugene, Ore.
Sharon Schuman

5K for FA
Hilton, New York
The Lana Family

Team BrAvery Keys 100
Key West, Fla.
The Marx Family

**Sigma Pi Chris Hull
Memorial Golf
Tournament**
State College, Penn.
The Hull Family

Hoot n' Holler
Denver, Colo.
KATA Foundation

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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Eugene, Oregon 97401

RETURN SERVICE REQUESTED

Our mission is to find effective treatments and a cure for Fanconi anemia and to provide education and support services to affected families worldwide.

HOW YOU CAN HELP

Donations Online:

Donate via 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

Donations of appreciated stock:

Please contact our office at 541-687-4658 or email info@fanconi.org.

fax: 541-687-0548 • info@fanconi.org • www.fanconi.org