



FAMILY Newsletter

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issue
65

30 years

FARF celebrates 30 years advancing research and supporting families

A note from Mark Quinlan, Executive Director



The Story of FARF: Lynn Frohnmayer Speaks at 30th Symposium

In 1983, Oregon parents Lynn and David Frohnmayer learned their two daughters, Kirsten and Katie, had Fanconi anemia (FA). They would later find out that their third daughter, Amy, also had the disease. At that time, the Frohnmayers were told that patients with FA rarely live to adulthood and that there is no cure for this illness.

In 1989, the Frohnmayers founded the Fanconi Anemia Research Fund (FARF). Thirty years later, all three Frohnmayer daughters have died from complications of FA. At the 30th Scientific Symposium in late 2018, Lynn Frohnmayer addressed the

continued on page 2

Thirty years ago, David and Lynn Frohnmayer invited a group of Fanconi anemia researchers to a meeting in Portland, Ore. The 13 researchers that attended who meeting, along with the Frohnmayers, developed the guiding principles that have driven the work we do here at FARF. These principles laid a foundation based on collaboration that continues to define the research we fund, the partnerships we form, and the way we engage with one another as a community. Throughout this newsletter, you will read about how our community works together, the impact of 30 years of research and family support, and statistics that measure our effectiveness in advancing our mission.

One of the most profound statistics is that, currently, we have more adults with FA registered at FARF than children. Why is this important? Because until recently, this had never occurred. This new reality indicates that the research

continued on page 2

“ We are in this together, and there is power in numbers. ”

FARF celebrates 30 years of advancing research and supporting families

continued from page 1

FARF has supported has increased the quantity of life for individuals with FA overall. This disease is no longer just a childhood disease, and because of this shift, we must address the needs of our growing adult population with FA – especially cancer – while continuing to support efforts that improve bone marrow transplant outcomes.

This past February, our Board of Directors and Scientific Advisory Board

met in Scottsdale, Ariz. for two days. This annual planning meeting gives the FARF staff the opportunity to inform and educate our leadership on the initiatives we have planned, and in turn, scientists and board members have the opportunity to advise us on the best path forward toward meeting our mission. I'm excited to share with you a few of the projects that we discussed and plan to carry out in 2019.

We have been partnering with the National Organization of Rare Diseases (NORD) to develop a clinical registry.

The rollout of this registry is underway and will be fully operational later this year. It will help us track the natural history of disease in people with FA and provide valuable information to scientists and clinicians as they work to advance treatments.

FARF has made several connections at Stanford University and is helping to support and develop a variety of FA projects there. These projects include research on reducing toxicity associated with bone marrow transplants, as well as a gene therapy

The Story of FARF: Lynn Frohnmayer Speaks at 30th Symposium

continued from page 1

audience, speaking about her family's story and the progress made in three decades of FA research. Her whole speech is available as a video at www.fanconi.org. What follows are highlights from her address.

The Beginning

In the early 1980s, things were going extremely well in our family. We had wonderful children who were smart, kind, and seemingly healthy. I remember going to the beach in Oregon, looking at my lovely family and thinking: 'I have got to be about the luckiest person I know.' Alas, that happy picture was not to last.

Over the course of the 1980s, we were to learn that of our five children,

all three of our daughters had Fanconi anemia. We were told it was a fatal disease, that kids usually died in the first two decades of life, and very few ever lived to adulthood. Therapies then were practically nonexistent. I just do not have the words to tell you the extent of the anguish we felt.

One of the harsh realities of life is that we're not always in control of the things that matter to us the very most of all. We all do the best we can with what we have been given, and sometimes we're fortunate to see that progress, and even great good, can come from our own misfortune.

Gene Discovery

David and I founded the FA Research Fund in 1989. Our first priority was to discover *the* FA gene. We now know that mutations in any one of 23 different genes cause FA. Five

of our 23 genes are breast cancer susceptibility genes (including *BRCA1* and *BRCA2*), discoveries that put our rare genetic disease in the mainstream of cancer research. All our FA genes function in the *FA/BRCA* pathway, and their main job is to protect and repair the genome. But the proteins encoded by these genes have functions independent of DNA repair. Some are involved in controlling oxidative stress, some in protecting against endogenous aldehydes and inflammation, and others in enhancing stem cell survival.

Bone Marrow Transplant

We've come very far in improving bone marrow transplant outcomes. In 1986, our daughter Katie needed a transplant, but she did not have a matched sibling donor and no center would perform the transplant. When daughter Kirsten developed leukemia



clinical trial. Stanford is emerging as a major FA research center, and we are excited to be part of this effort.

In March, we partnered with the Aplastic Anemia and MDS International Foundation to advocate in Washington D.C. for more federal funding to support bone marrow failure research. While these discussions are ongoing, we hope to double the \$3 million appropriation for this year.

In April we held our second Joel Walker Scientific Meeting on Head and Neck Cancer. This year our focus is to

continue developing strategies for early detection, prevention, and treatment while establishing the framework for a virtual tumor board for individuals with FA diagnosed with cancer.

Last fall we received 52 applications for the David B. Frohnmayer Research Grant Awards, and are currently reviewing 15 finalists. In June we will determine which projects will receive funding.

These are just a few projects that illustrate our efforts, but rest assured we have many other endeavors

underway to support research and families, like publishing revised Clinical Care Guidelines and producing two new family support guides about living with FA (one for families and one for adults with FA). We are in this together, and there is power in numbers. Thank you all for your contributions, and I look forward to continuing this effort with each one of you. Go, Fight, Cure!

—Mark Quinlan ■

in 1995, centers were willing to transplant her but gave her only a 20% chance of survival.

In 1999, Dr. John Wagner did a trial to see if one drug, fludarabine, could make a difference. Practically overnight, success rates went from 20% to 60%! Today in the United States at our three major FA transplant centers, success rates for unrelated donor transplants are approximately 90%. As a result, we are seeing a huge improvement in survival.

Cancer

I wish we could declare victory and go home, but because of increased longevity, the next shoe to drop is cancer. Back when we founded FARF in 1989, one FA researcher estimated that 2% of the FA population would get cancer. Our 2014 Clinical Care Guidelines stated that from birth to age 40, about 14% of FA

patients would develop oral squamous cell carcinoma. But when you consider only the *adult* FA population, the numbers become alarming. A German study of 142 individuals from ages 18 to 45 showed that 35% had developed cancer, with the likelihood increasing steadily with age.

What is FARF doing to get ahead of this huge challenge? We now have two full-time scientists on our staff working on this problem. We're committed to holding small cancer workshops annually. We're developing an adult cancer registry. We're giving grants to researchers who are working hard to identify ways to prevent and treat FA cancers.

A Request to Researchers and Doctors

I suspect that every single FA family member in this room has an

overwhelming sense of urgency. Families depend on us to find answers, to identify drugs and better approaches to prevent and treat malignancies.

I make one request of all you scientists and treating physicians: that you share our families' sense of urgency. It's not enough to identify a hopeful compound or to write an article and get it published in a scientific journal. Those efforts won't help patients until we put promising drugs into clinical trials. Not everything we do will be successful, but we must take reasonable chances and move very aggressively to tackle this problem. Our young adults desperately want and deserve a chance at a full lifespan, and depend on all of us to make that hope a reality. ■

FARF Celebrates 30 Years Advancing Research at Scientific Symposium



When FARF created guiding principles back in 1989, one was that each year, we would bring together researchers working on Fanconi anemia (FA) and physicians treating people with FA. They would spend three days sharing updates and concerns, brainstorming ideas, and creating collaborations. In September 2018, the 30th Scientific Symposium took place in Newport Beach, Calif.

In addition to the three days of talks by prominent and upcoming FA researchers and clinicians, the 30th Symposium featured new and exciting sessions and grant opportunities, inspiring talks by adults with FA, and the celebration of 30 years of progress. Nearly 400 researchers, clinicians, individuals with FA, family members, staff, and volunteers attended the conference. Here are some of the highlights.



FARF Tank

The first ever FARF Tank took place to allow up and coming researchers an opportunity to pitch their innovative ideas to the scientific community and win a \$10,000 grant. Ten contestants had only five minutes each to describe their ideas to nearly 400 audience members and our panel of judges. At the end, the audience voted for the project they thought most deserved the \$10,000 grant. The judges also chose a winner.

People's Choice Winner: Allison Bartlett for "Serotonin - A Novel Target in FA."

Judges' Choice Winner: Lianne Vriend for "Addition of mild hyperthermia to improve treatment of FA/BRCA pathway deficient head and neck cancer in Fanconi anemia patients."



Unforgettable opening session

In a thoughtful interview, Dr. Stella Davies (Cincinnati Children's Hospital, FARF Board of Directors) asked Mary-Beth Johnson and Sean Breininger about their lives with Fanconi anemia: how they manage stress around healthcare decisions, how they balance FA in their everyday lives, and their hopes and fears about the future. Mary-Beth told the audience about taking ownership over her health and her journey to growing her family. Sean's wife, Allison, also spoke about what it means to be a caregiver to someone living with a chronic illness, and the impact FA has on their family. Visit www.fanconi.org to watch videos from the opening session.





Record number of poster presentations

Sixty researchers presented their projects as posters. Posters are displayed throughout the conference and presenters are given the chance to share their research during two poster receptions. During the banquet, awards were given to Jordi Minguillón for best translational abstract, Sonia Ruiz-Torres for best basic science abstract, and Gilda Garza-Mayan for best clinical abstract (all pictured in center above).



International Fanconi Anemia Summit

This year, more than 40 researchers, clinicians, FA family members, and representatives from FA organizations around the world got together to build a more intentional global FA network. Participants split into three groups to address organizational and fundraising capacity, family services, and access to doctors and medicine. Together, the groups determined needs, priorities, and action steps in each of these areas. The second summit will take place at the 2019 Symposium in Chicago to continue progress in each area.



Kevin McQueen accepts Distinguished Service Award

Kevin is the father of an adult son with Fanconi anemia. He has been involved with FARF since 2000, as a board member, board president, fundraiser, and advocate. Together with his wife and close friends, Kevin has raised more than \$1 million for FA research and family services. At the Symposium banquet, current board president Mark Pearl presented Kevin with the distinguished service award. Thank you to Kevin for the years of incredible service.

Lynn Frohnmayer, FARF co-founder, addresses the scientific community

When Lynn and David Frohnmayer first heard of Fanconi anemia after their daughter was diagnosed in 1983, they were told that children with FA rarely survive to adulthood and that there was no cure. They quickly reached out to any researcher who had ever worked on Fanconi anemia. Six years later, in 1989, they formed the Fanconi Anemia Research Fund so that three institutions could use funds raised by the Frohnmayers for FA research.

Thirty years later, Lynn spoke to the FA scientific community about the tremendous progress made over the last three decades, and the formidable hurdles still to overcome (see pages 2-3). ■

Key Scientific Takeaways from the Scientific Symposium

Cancer in FA:

- Early detection using brush biopsy can detect the development of cancer. Detection and prevention measures are not enough to combat the prevalence of HNSCC tumors, even in the general population. Treatments also need to be developed.
- Currently, surgery is the best option for treating solid tumors in FA patients, but radiation therapy can be used in some cases. Certain people are more sensitive to radiation, so this kind of therapy has to be evaluated on an individual basis. When an FA patient does have a tumor, the earlier the surgery is performed, the better the outcome.
- There is a higher risk of cancer in FA patients who have had graft-versus-host disease (GvHD).
- Next generation sequencing will be necessary to identify genomic signatures (changes in genes) of FA patients and will enable us to define which drugs can be used

for precision medicine. There are currently several potential FA cancer drugs that are in pre-clinical testing.

- Preclinical models of FA squamous cell carcinoma will help identify pathways and mechanisms that can be targeted clinically.
- Heterozygote carriers of most mutated FA genes do not have an increased risk of cancer.
- Researchers are looking at changes in chromosomes in tumors. Changes in chromosomes can lead to having genes in the wrong place, which can increase the chance of getting cancer.

New Approach to Stem Cell Transplantation

- FA individuals undergoing bone marrow transplant are typically given radiation and/or chemotherapy to eliminate the host bone marrow, in order to make space

for healthy donor marrow. These conditioning agents damage DNA, and are believed to increase the later risk of cancer.

- A new method, using antibodies to eliminate the patient's marrow and defective stem cells, could be much safer, and is now under investigation at Stanford University. If preliminary studies validate the use of this method, Stanford plans to initiate a clinical trial in the near future.

Improving Hematopoiesis

- The FA pathway is necessary for DNA repair. People with FA do not have a functional FA pathway, because a crucial gene in this pathway has mutations that prevent it from functioning normally.
- Researchers are investigating whether blocking or activating non-FA proteins could reduce some of the detrimental effects of FA.
- Researchers are conducting preclinical tests of targeted inhibitors to determine if they can improve stem cell growth.

Gene Therapy

- Gene therapists in Spain have treated eight FA patients in the FA-A complementation group over the past three years. Four individuals are now post-therapy long enough to be evaluated, and all four show a progressive increase in the percentage of corrected cells, both in the peripheral blood and in the bone marrow. Patients have experienced no severe adverse effects.
- Because of the encouraging results from the Spanish gene therapy trial, Rocket Pharmaceuticals, Inc. is now launching new clinical trials in both Spain and at Stanford University. Early progenitor cells from the

patient will be transduced and infused immediately after collection with no prior cryopreservation, in an effort to obtain the largest number of cells possible. Stanford has recently transplanted two FA patients using this new methodology. It is too early to know the success of this approach.

Gene Discovery

- Scientists from the University of Wurtzburg, Germany, the National Institute on Aging and the University of Miami announced the discovery of the 23rd FA gene, *FANCY*. To date, only one individual has been placed in this complementation group.

Gene Therapy by Gene Editing

- CRISPR-Cas9 is a unique technology that enables researchers to edit parts of the genome by removing, adding or altering sections of the DNA sequence. An enzyme called Cas9 acts as a pair of "molecular scissors" that can cut the two strands of DNA at a specific location, so that bits of DNA can be added or removed. A piece of RNA called guide RNA makes sure that the DNA is cut in just the right place. Scientists are very hopeful that this system of gene editing can be used to correct genetic diseases.
- Preliminary data shows that this precise method of gene editing depends on a functional FA pathway for success. The doubled stranded cut in DNA needs to be repaired, and FA genes are crucial in achieving this repair. Our FA scientists are now exploring possible methods to compensate for this problem.



Could anticancer drugs work in FA?

Presented by Jordi Surrallés and Jordi Minguillón

Photo: Jordi Surrallés (bottom left) and Jordi Minguillón (bottom second from left) with other Spanish researchers at the Scientific Symposium

People with Fanconi anemia (FA) have a 500- to 700-fold increased risk of developing head and neck squamous cell carcinoma (HNSCC) tumors when compared to the general population. Conventional treatments available for non-FA-HNSCC such as radiation therapy and chemotherapy are highly toxic to people with FA, making alternative therapeutic options desperately needed.

The research

In a recent FARF-funded study, Drs. Jordi Surrallés and Jordi Minguillón of the Hospital de Sant Pau and Universitat Autònoma de Barcelona performed a screen of 3,800 drugs (including 1,200 FDA approved drugs) on HNSCC cell lines derived from patients with FA. The goal of the study was to identify drugs that could kill tumor cells without harming normal cells or causing DNA damage.

At the 30th Scientific Symposium, the Barcelona team showed promising results on two potential drugs: Gefitinib (Iressa) and Afatinib (Gilotrif), which block activity of the epidermal growth factor receptor (EGFR). EGFR is a cell-surface receptor protein that, when active, leads to increased cell growth, survival of tumor cells, and metastasis (when cells grow beyond the primary tumor). EGFR is highly expressed in head and neck tumors in the general population and is active in FA patient-derived tumor cell lines. EGFR has long been a cancer-specific target for drug therapies, and Gefitinib and Afatinib currently have FDA-approval for treating a specific kind of lung cancer (non-small cell) in the general population. The efficacy of these drugs has never been tested before in the context of FA cancers, however.

Drs. Surrallés and Minguillón's study is the first to show that Gefitinib and Afatinib inhibited FA HNSCC cell line growth without damaging DNA. When mice with FA HNSCC tumor cells were treated with these drugs, the research team observed that the drugs inhibited tumor growth efficiently. In addition, no adverse hematologic effects were seen in a mouse model of Fanconi anemia, suggesting that these drugs could be safe in patients.

Why is this significant?

The findings from this study are exciting because targeted therapies for HNSCC that do not induce DNA damage are desperately needed for FA patients. Dr. Surrallés and Dr. Minguillón have since obtained Orphan Drug Designation (ODD) for both Gefitinib and Afatinib to treat HNSCC in Fanconi anemia by the European Medicines Agency (EMA). These are early but encouraging steps to reach the goal to develop and initiate a clinical trial to treat HNSCC in FA patients. FARF has recently funded the Spanish team to develop this project into a clinical trial. ■

“ The findings from this study are exciting because targeted therapies for HNSCC that do not induce DNA damage are desperately needed for FA patients. ”

How might we apply cancer-fighting strategies from the general population to the FA population?

Presented by Carter Van Waes and Ruud Brakenhoff



The head and neck cancer panel at the Scientific Symposium. From left to right: Drs. John Wagner, Ruud Brakenhoff, Joel Greenberger, and Carter Van Waes.

To fast-track therapies for people with Fanconi anemia (FA), it's often best to start by examining progress that's already been made in the general population. Large-scale studies such as The Cancer Genome Atlas (TCGA) have made it possible to detect abnormal chromosomes and mutations that lead to cancer in people without FA. Dr. Carter Van Waes, the clinical director and chief of head and neck surgery at the National Institute on Deafness and Other Communication Disorders in Bethesda, Md., gave a keynote address at the 30th Scientific Symposium on the promise of using genomic profiling studies such as the TCGA to identify therapeutic targets for Fanconi anemia squamous cell carcinoma (SCC).

According to Dr. Van Waes, data from the TCGA study of 1,400 squamous cancers, including lung, head and neck, esophageal, cervical, and bladder cancer, can provide clues for FA researchers as to which abnormalities in FA tumor cells can be exploited for treatment purposes. He discussed TCGA results that showed that all SCC tumor cells – despite their site of origin – share common characteristics, exhibit DNA mutations, and have changes in the number of chromosomes that can lead to cancer.

SCCs analyzed in the study showed changes in the 3q and 5p chromosomes. A gain of the 3q chromosome is also used to test for the emergence of myelodysplastic syndrome (MDS) and acute myeloid leukemia (AML) in FA patients. The TCGA study showed that tumors express genes that play a role in growth, survival, inflammation, and the DNA damage response—processes all essential for tumor growth and survival. Interestingly, some SCC tumors with human papillomavirus show inactivation of the *FANCF* gene by providing another link between FA and the development of non-FA SCC. This finding could make them more sensitive to certain therapies where DNA damage helps kill cancer cells.

What does this mean in FA?

FARF-funded researchers and others are currently studying whether the characteristics observed in SCC cells in the TCGA study also occur in FA squamous cell carcinoma. If similarities exist, the goal would be to test targeted therapies that work in the general population on FA patients with cancer.

One such researcher, Dr. Ruud Brakenhoff of the Cancer Center of Amsterdam, also shared his work focused on noninvasive diagnosis and targeted treatment of oral precancers. Detecting precancer before it becomes

continued on page 10

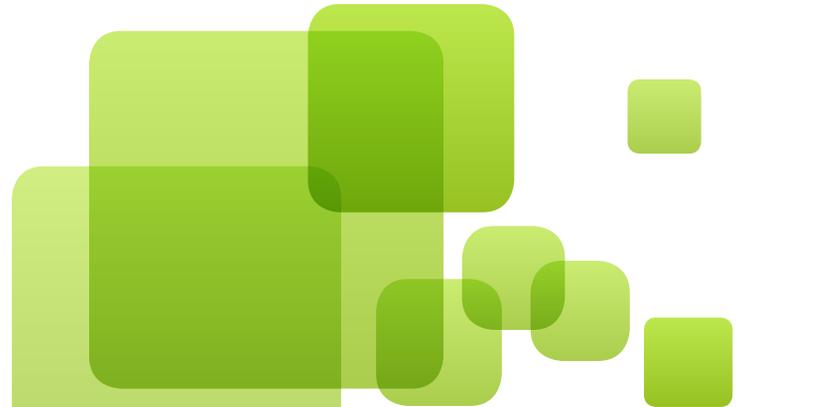
How might we apply cancer-fighting strategies from the general population to the FA population?

continued from page 9

cancerous gives us the opportunity to stop it before it requires aggressive treatments that could be detrimental for patients with FA. One goal of Dr. Brakenhoff's study was to determine whether noninvasive screening tests, such as the brush biopsy of the oral cavity, can be used to identify precancerous changes that can then be targeted with therapy.

In his study, results showed that brush biopsies are a feasible way to analyze precancerous cells and compare to tissue biopsies. Dr. Brakenhoff's study typically showed that FA tumors have many DNA mutations, making them similar to the SCC tumors in the general population analyzed in the TCGA trial discussed by Dr. Van Waes.

Dr. Brakenhoff showed that certain proteins in FA and non-FA HNSCC cells, like serine/threonine-protein kinases (PLK1) and Wee1-like kinase (WEE1), are potential drug targets, that when blocked, would specifically kill cancer cells. Dr. Brakenhoff's group is now investigating the potential toxicity of drugs targeting these proteins in FA patients. Once the safety profile is determined, the intent is to develop clinical trials in people with FA. This therapeutic method would : 1) screen for precancers, 2) treat precancerous lesions once they're detected, and 3) monitor for progression to cancer. ■



Aldehyde inhibitor may help protect stem cells

Presented by Jennifer Tsai

What are aldehydes and what do they do?

Research has shown that small molecules called aldehydes cause DNA damage and increase the potential for bone marrow failure.

Aldehydes are formed within our bodies (endogenous) and from sources outside our bodies (exogenous). Exogenous aldehydes are found in sources such as automobile and cooking fumes and cigarette smoke, and dietary sources such as alcoholic beverages. Aldehydes are metabolized (or detoxified) in the body by a group of enzymes called aldehyde dehydrogenases (ALDH2).

In the context of FA?

Studies have shown that aldehydes (which cause DNA damage) have accelerated progression to bone marrow failure and aplastic anemia. In animal studies, mice that do not express ALDH2 and *FANCD2* have spontaneous leukemia and bone marrow failure.

Dr. Jennifer Tsai and Dr. Kenneth Weinberg from Stanford conducted

a recent study funded by FARF to investigate whether a small molecule activator of ALDH2 could protect against bone marrow failure. The goal of the study was to develop pre-clinical data and tools that will be needed to support a clinical trial for ALDH2 activators in people with FA.

Dr. Tsai presented results from their study at the Symposium and demonstrated that Alda-1, a small molecule ALDH2 activator, protected and expanded the number of hematopoietic stem cells (HSC) in mice and decreased hematopoietic stem cell (HSC) death. The drug was safely tolerated, non-toxic, and actually expanded stem cells regardless of the presence of mutations in ALDH2. A three-week treatment of mice with Alda-1 improved stem cell function that persisted for six weeks post treatment, indicating the potential for longer-term protective effects. Results from this study will be used to develop the feasibility and efficacy of this treatment strategy for a clinical trial in people with Fanconi anemia. ■



Inhibiting Transforming Growth Factor beta (TGF- β) may help combat bone marrow failure

Presented by Alfredo Rodriguez

Treatment options for bone marrow failure that do **not** rely on transplantation are urgently needed for individuals with FA. Dr. Alan D'Andrea and his lab at the Dana Farber Cancer Institute have been working to uncover ways to prevent bone marrow failure and recently identified the role of the Transforming Growth Factor beta (TGF- β) pathway in the process. Experiments in his lab have shown that FA mice exhibit a hyperactive TGF- β pathway and that FA patient cells have increased expression of proteins in this pathway.

At the 30th annual Scientific Symposium, a postdoctoral fellow in Dr. D'Andrea's lab, Dr. Alfredo Rodríguez, reported on exciting advancements on the TGF- β story. His work, funded by FARF, focused on using preclinical experiments to identify whether clinically relevant drugs that inhibit the TGF- β pathway can potentially be used to inhibit bone marrow failure in FA patients. Dr. Rodríguez discussed promising results that showed that a TGF- β pathway inhibitor known as AVID200 rescued stem cell defects in FA patient bone marrow samples. Additional preclinical testing showed

that AVID200 increased the number of bone marrow cells and decreased the cellular defects that lead to bone marrow failure in FA mouse models.

AVID200 was very effective in cells from patients who showed progression to Myelodysplastic Syndrome (MDS). This is important because it shows we could potentially use AVID200 or other TGF- β pathway inhibitors to prevent MDS. This work suggests that the use of TGF- β pathway inhibitors may provide a therapeutic approach to inhibit bone marrow failure in FA patients, and particularly for those who may be at increased risk for developing MDS. Dr. D'Andrea's team is currently working on developing these therapies for clinical use in FA patients. ■

“ This work suggests that the use of TGF- β pathway inhibitors may provide a therapeutic approach to inhibit bone marrow failure in FA patients, and particularly for those who may be at increased risk for developing MDS. ”

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.

Tumor Testing Available to Help Identify Therapy Options

If you or your loved one develops a tumor and would like guidance about potential personalized therapies, please contact FARF team members Dr. Sudhir Borgonha or Dr. Isis Sroka to help you navigate options and next steps. FARF has a relationship with the Knight Cancer Institute at Oregon Health & Science University to make sequencing available to Fanconi anemia patients who develop a malignancy. The Knight Diagnostic Laboratories (KDL) specialize in molecular diagnostic testing that may lead to targeted drug therapy options for patients based on the identification of DNA mutations in cancer samples.

Dr. Sudhir Borgonha, FARF Translational Science Director | 541.687.4658 | sudhir@fanconi.org

Dr. Isis Sroka, FARF Director of Scientific Operations | 541.687.4658 | isis@fanconi.org

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

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 | currently recruiting participants

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: Evette Barranta | 301-827-4421 | barrantae@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
Sara Loveless | 513-803-7656 | Sara.Loveless@cchmc.org

A Clinical Trial to Evaluate the Safety of RP-L102 in Pediatric Subjects With Fanconi Anemia Subtype A

Stanford University and Lucille Packard Children's Hospital, Stanford, CA | currently recruiting participants

The objective of this study is to assess the therapeutic safety and preliminary efficacy of a hematopoietic cell-based gene therapy consisting of autologous CD34+ enriched cells transduced with a lentiviral vector carrying the FANCA gene in subjects with Fanconi anemia subtype A (FA-A). Blood stem cells collected from an FA-A patient are genetically modified to introduce an intact copy of the FANCA gene using a virus that has been changed in the laboratory so that it cannot grow or spread to cause an infection. The genetically modified cells are then returned back into the patient. <https://clinicaltrials.gov/ct2/show/NCT03814408>

Contact: Sandeep Soni, MD | 650-725-9250 | ssoni1@stanford.edu
Agnieszka Czechowicz, MD, PhD | 650-497-2218 | aneeshka@stanford.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

Natural History of FANCD1/BRCA2

National Cancer Institute (NCI), Bethesda, MD

We previously determined that published cases with two mutated FANCD1/BRCA2 genes appeared to have a very high risk of cancer before age 6. We are now aware of individuals with these mutations who are much older and have not had cancer. In order to determine the natural history of patients with FA associated with mutations in FANCD1/BRCA2, we have created a subgroup within the National Cancer Institute study of Cancer in Inherited Bone Marrow Failure Syndromes (above). <http://www.marrowsfailure.cancer.gov/>

Contact: IBMFS Study Team | 1-800-518-8474 | NCI.IBMFS@westat.com

Pilot Study of Metformin for Patients with Fanconi Anemia

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

Preclinical studies from OHSU suggest 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: Ashley E Kuniholm | 617-355-6513 | ashley.kuniholm@childrens.harvard.edu
Jacob Cotton | 617-919-4227 | jacob.cotton@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. ■



Angela Bedoya Receives Amy Winn and Christopher T. Byrd Award for Adults with Fanconi Anemia

FARF Executive Director, Mark Quinlan, presents Angela Bedoya the Amy Winn and Christopher T. Byrd Award for Adults with Fanconi Anemia

At the 2018 Meeting for Adults with FA, the Amy Winn and Christopher T. Byrd Award for Adults with Fanconi Anemia was presented to Angela Bedoya. Angela is a young researcher who developed an interest in science during her bone marrow transplant as a teenager. After completing her degree in biomedical engineering, Angela began a PhD program in microbiology, immunology, and cancer biology at the University of Minnesota. She hopes to be part of the transformative medical research in Minnesota and to provide hope for patients and families. You can watch her acceptance speech at www.fanconi.org.

The Fanconi Anemia Research Fund honors the inspirational lives of two beloved former board members with the **Amy (Frohnmayr) 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. ■

“ She hopes to be part of the transformative medical research in Minnesota and to provide hope for patients and families. ”



ONE MONTH. ONE CAUSE.

How Will You Make an Impact?

FA families started International Fanconi Anemia Day 10 years ago to raise funds for research and family support services. We know that longer and better lives for people with FA are possible when we all get involved.

The FA Day slogan is #ThisIsHowIFA. Inspired by Matt Pearl, an adult with FA, #ThisIsHowIFA reflects the many people it takes to make a difference and find a cure. Matt says "FA means Find Answers. FA means Fight Always." People with FA, family members, scientists, doctors, volunteers, friends, and staff members are all part of the FA community. Whether you're Finding Answers or Fighting Always, or both, show us how you FA!

That's right, it starts with you. Whether it's \$50 or \$5,000, the money you raise or give goes to fund our incredible FA researchers, bring our FA families together, and keep our organization running.

We celebrate FA Day all throughout May.

Visit <http://fundraise.fanconi.org/faday2019> to learn more.

Go, Fight, Cure!



Are you with us?

Together with dedicated community members like you, we research and supported affected families worldwide for 30

1980s

THE ACCIDENTAL ORGANIZATION

In 1989, three institutions approach the Frohnmayers to raise money for their investigations. Lynn and Dave again write to their friends and ask them to make checks out to "Fanconi Anemia Research Fund". However, the institutions can't cash them as written! The Frohnmayers quickly decide to form a 501(c)(3) charitable corporation, name it the Fanconi Anemia Research Fund (FARF), form a board of directors, and establish articles of incorporation. They cash the checks, send each of the researchers \$30,000, and the FA Research Fund is launched!

A DEVASTATING DIAGNOSIS

Oregon parents Lynn and David Frohnmayer learn their two daughters, Kirsten and Katie, have Fanconi anemia (FA), a rare genetic disorder that leads to bone marrow failure, leukemia, and cancer. They would later find out that their third daughter, Amy, also has the disease. The Frohnmayers learn that patients with FA rarely live to adulthood and that there is no cure for this illness. They also discover that the disease is scarcely known in the medical community.



The Frohnmayer family in 1985. From left to right, David, Lynn, Mark, Jonathan (baby), Kirsten, Katie.



The Frohnmayer children in 1987. From left to right, Amy, Kirsten, Jonathan, Mark, Katie.

THE FIRST SCIENTIFIC SYMPOSIUM

The Frohnmayers invite scientists who have published about FA or are recommended by those scientists to a scientific meeting. Thirteen researchers are asked to "bring their brains" to Portland, Ore., where, together with the Frohnmayers and FARF board, they draft what would become FARF's guiding principles. These same principles continue to define the Fund's work today: encourage scientific collaboration and a multidisciplinary approach, hold annual scientific symposia, identify and support the best science possible, and fund researchers who would go on to leverage FARF's start-up grants into much larger grants from their institutions or government entities. The group endorses a five-year gene identification project with the goal of identifying one or more genes responsible for this disorder.



Dr. Buchwald accepts a discovery award from Dave Frohnmayer.

1990s

THE FIRST FA GENE IS DISCOVERED

One of the first efforts of FARF is to identify the FA gene(s). Gene discovery enables scientists to study and begin to understand the function of the protein(s) encoded by a FA gene(s), which might suggest how best to treat the disorder. In 1992, Manuel Buchwald and his team at the Hospital for Sick Children in Toronto, Canada, discover the first FA gene, *FANCC*. This is a major breakthrough for FA science and paves the way for many more research discoveries in the decades to follow.

1990S: MOVING FORWARD

FARF surges forward in the 90s, funding new research grants each year and expanding family support services. Annual science and family meetings result in the growth of the FA community, consisting of researchers, clinicians, people with FA and their families, volunteers and staff. Educational resources and publications like the patient handbook and family & scientific newsletters keep the community informed and engaged.



Dave Frohnmayer and Ralf Dietrich with Hans Joenje (right) after he accepts the Award of Merit at the Family Meeting in 1996.



Grover Bagby addresses the audience at the 10th Annual FARF Symposium in Denver in 1998.

GENE DISCOVERY

GENE DISCOVERY
FANCA

FANCG

1996

1997

1998

1999

THE FIRST FA FAMILY MEETING

As a scientific community begins to take shape, the Frohnmayers also focus on family engagement as a central strategy. Although they were at first skeptical that families would be interested or able to gather together, in 1991 more than 100 FA parents and children attend the very first "FA Family Symposium" in Washington, D.C. This gathering marks the beginning of what would become a cornerstone of the FA community: an annual meeting of individuals with FA, their families, and physicians and researchers, with the purpose of educating and supporting families, forming connections and improving and extending the lives of those affected by FA.

EDUCATIONAL SUPPORT FOR FAMILIES & PHYSICIANS

With the help of the emerging FA community, Lynn and Dave Frohnmayer publish *Fanconi Anemia: A Handbook for Families and Their Physicians*, the first edition of what would later become the *Fanconi Anemia Guidelines for Diagnosis and Management*.



Right: Dave Frohnmayer presents John Wagner an award at the 14th Scientific Symposium in Philadelphia in 2002.



John Wagner, MD, University of Minnesota, Minneapolis, receives the first Fanconi Award for Outstanding Achievement in Research from Dave Frohnmayer and Lynn Frohnmayer at the 14th Scientific Symposium in Philadelphia in 2002. The award honors Dr. Wagner's research on the genetic basis of FA and his leadership in the development of the first FA patient handbook, *Fanconi Anemia: A Handbook for Families and Their Physicians*. Dr. Wagner's research has led to the identification of the first FA gene, *FANCA*, and the discovery of the first FA patient with a *FANCA* mutation.

Above: headlines from the Fall 1999 issue of the FA Family Newsletter.

1981 1982 1983 1984 1985 1986 1987 1988 1989 1990 1991 1992 1993 1994 1995 1996 1997 1998 1999 2000

CONNECTING WITH OTHER FA FAMILIES

Feeling anxious, frightened and lonely, the Frohnmayers follow the advice of a researcher at The Rockefeller University and reach out to other FA families to create a support group. Seventeen families respond with interest. Lynn and Dave begin writing the FA Family Newsletter to share information they glean from scientists with other families.

EARLY OUTREACH TO EXPERTS

A scientist asks the Frohnmayers to raise \$50,000 for research. They promptly write to family, friends, and acquaintances to solicit donations to The Rockefeller University. The unofficial research fund is born.

have funded Fanconi anemia years...and counting. Thank you.



FANCONI ANEMIA RESEARCH FUND
Advancing Research. Supporting Families.

2000s

2000s: GROWTH CONTINUES



Children and teens at the Family Meeting in 2000.

Throughout the 2000s, FARF continues to extend its outreach, funding several research projects each year, welcoming more scientists and families to regular meetings, and publishing clinical care guidelines, newsletters and family directories. During this decade, FARF sponsors seven regional family meetings in addition to the annual FA Family Meeting at Camp Sunshine. In addition to the annual Scientific Symposium, FARF holds nine focused workshops on topics such as bone marrow transplantation, small molecules as possible therapeutics, and squamous cell carcinoma.

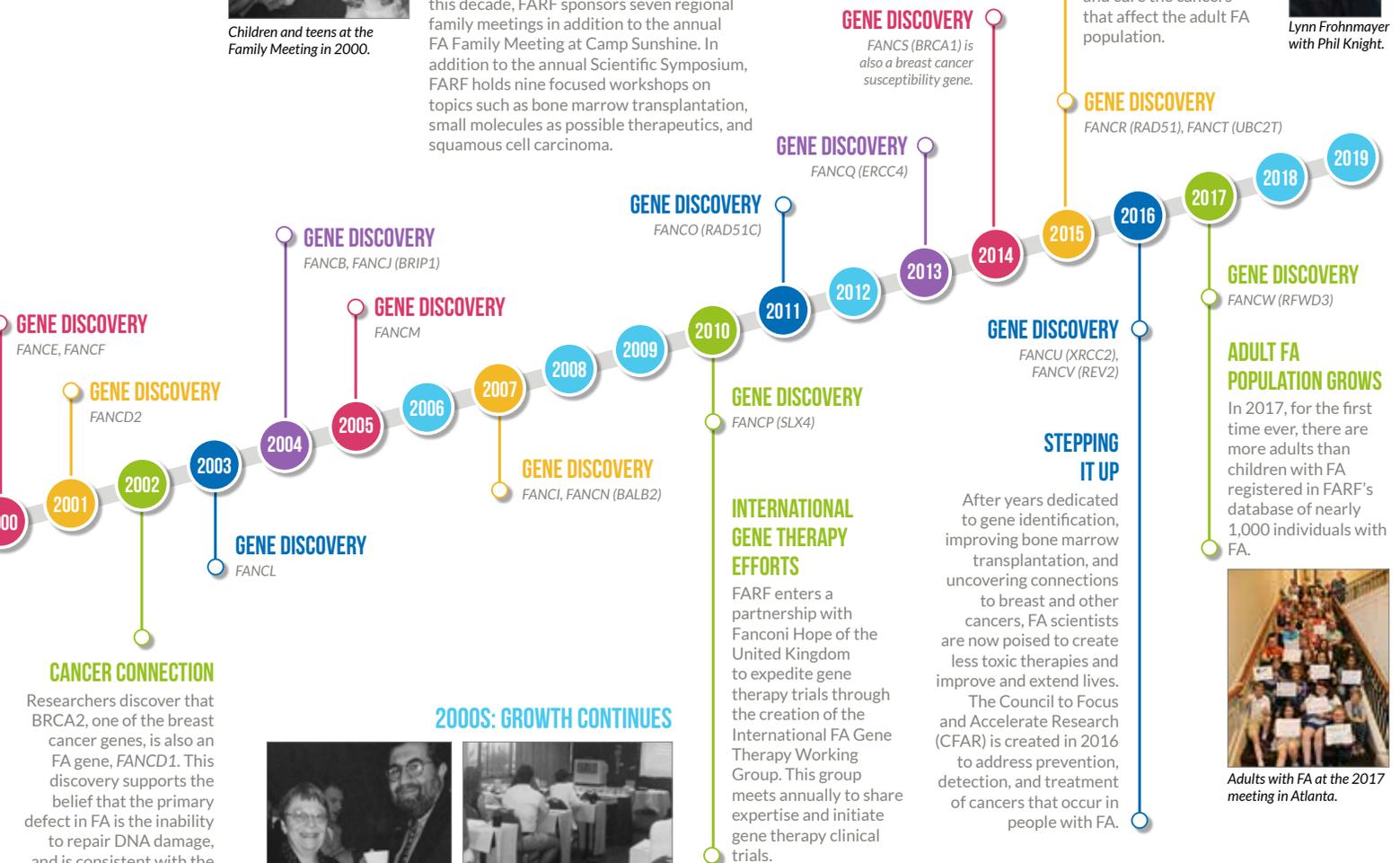
2010s

A RECORD-BREAKING YEAR

Longtime FARF supporters Phil and Penny Knight pledge \$10 million to advance research over the next 10 years. Their incredible gift focuses on bold initiatives to prevent and cure the cancers that affect the adult FA population.



Lynn Frohnmayer with Phil Knight.



CANCER CONNECTION

Researchers discover that BRCA2, one of the breast cancer genes, is also an FA gene, FANCD1. This discovery supports the belief that the primary defect in FA is the inability to repair DNA damage, and is consistent with the finding that those with FA are at far greater risk for cancer. This cancer connection puts FA on the radar of oncologists around the world.

2000s: GROWTH CONTINUES



Arlen Auerbach and Farid Boulad at the 14th Symposium in 2002 in Philadelphia.



Blanche Alter presents at the first Meeting for Adults with FA in 2003 in Virginia. Seven adults with FA and one teen with FA are in attendance.



FA families and community members at the 2010 Family Meeting.



INTERNATIONAL GENE THERAPY EFFORTS

FARF enters a partnership with Fanconi Hope of the United Kingdom to expedite gene therapy trials through the creation of the International FA Gene Therapy Working Group. This group meets annually to share expertise and initiate gene therapy clinical trials.



The FA Gene Therapy Working Group in Madrid in 2016.

STEPPING IT UP

After years dedicated to gene identification, improving bone marrow transplantation, and uncovering connections to breast and other cancers, FA scientists are now poised to create less toxic therapies and improve and extend lives.

The Council to Focus and Accelerate Research (CFAR) is created in 2016 to address prevention, detection, and treatment of cancers that occur in people with FA.

ADULT FA POPULATION GROWS

In 2017, for the first time ever, there are more adults than children with FA registered in FARF's database of nearly 1,000 individuals with FA.



Adults with FA at the 2017 meeting in Atlanta.

LOOKING FORWARD

FARF continues to advance FA research and provide support to families. Gene therapy trials are now open on two continents and two new clinical trials were approved in early 2018, one of metformin (Dana Farber Cancer Research Center) and the other of quercetin (Cincinnati Children's Hospital). The incredible progress achieved over these years can be attributed to the hard work, dedication, and commitment of our FA families, researchers, clinicians, donors, staff and volunteers.



Adults with FA at the 2018 Family Meeting.



Eli and Lucy at the Family Meeting in 2018.



The Karras Lab at MD Anderson Cancer Center.

Top ten reasons to attend the Meeting for Adults with FA

1



"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 in the moment while worrying about possible illness in the future."

"It's an incredible experience to feel like we all belong and to be able to share our stories with other patients, doctors and researchers. It's that undeniable sense of belonging to a family that just 'gets it.'"



2

3



"My reason for attending these meetings is simple: the pure joy that comes from seeing all these wonderful people come together, united, as a community, by the single thing we share, rather than separated by any differences. We can all put aside our personal hardships and struggles to enjoy the pleasure of a friend's company. I am always glad to meet everyone there, as they inspire me to involve myself in these battles we wage individually and, at the same time, as one."



"I was a little overwhelmed in the support groups initially. However, the more that I interacted with other adults with FA, the more I realized that I needed to be here and that I have support from others who know what I feel and am going through."

"I don't have any FA specialists where I live, so attending this meeting, hearing from the experts, and getting their advice is invaluable to me. I take what I learn back to my own doctors, and I feel empowered to direct my medical care."



"Having the Adult Meeting combined with the Scientific Symposium is such a great idea. Even though the Symposium sessions can be hard to understand, it is inspiring to see so many scientists working on this disease, and hearing the things they're working on gives us hope for our future."

"We sat with researchers from Spain at dinner one night, and their passion for our little disease gave me so much hope! It made me a little bolder in my dreaming of a long life. It made me SO thankful that they have persisted, sacrificed, and pushed through failure, to bring us closer to long-term solutions."



8



Meet in real life.

"I'm thankful that we have such a supportive community, spread all across the world. It's so exciting to come together and meet people in person, whom you've encouraged or supported online, or who have encouraged and supported you. For me, the meeting is about unity. I love hearing about and acknowledging other patients' unique experiences and realizing we're all individuals and resilient, while having so many of the same troubles and fears. We are both united and unique."

"I will forever be indebted to the adults with FA. For creating a community of generosity and hope. For sharing the pit of feelings. And for finding the resilience to climb out of the pit, with arms reaching down to pull others up, and sit around a pool late into the night sharing life and laughs."



Those nights by the pool.

9

10



Have fun!

"My personal favorite thing about the FAdult meeting is hearing other people's stories and being there to support them and receive support from individuals like me. I enjoy the late nights, the long conversations, the self-planned outings and the small groups that just seem to form. I think they are lot of fun, and look forward to going back year after year."

FANCONI ANEMIA RESEARCH FUND

2019 MEETING FOR ADULTS WITH FANCONI ANEMIA

RENAISSANCE DOWNTOWN CHICAGO 09/18 - 9/21

Scientific Advisory Board



Blanche P. Alter, MD, MPH
National Cancer Institute
Senior Clinician



Amy DeZern, MD, MHS
Johns Hopkins Hospital
Assistant Professor of Oncology &
Medicine, Dept. of Oncology;
Attending Physician



Jennifer Grandis, MD
University of California, San Francisco
Professor in the Department
of Otolaryngology – Head and
Neck Surgery



Melissa Haendel, PhD
Oregon State University
Director of Translational Data Science,
Linus Pauling Institute



Premal Patel, MD, PhD
Lyell Immunopharma
Scientific Advisory Board Chair;
Chief Medical Officer



Kathryn Pennington, MD
University of Washington
Faculty Member in
Gynecologic Oncology



Jeffrey Siegel, MD
Gilead Sciences
Executive Director, Translational
Medicine, Inflammation Clinical
Research



Agata Smogorzewska, MD, PhD
The Rockefeller University
Associate Professor



John Wagner, MD
University of Minnesota
Professor, Dept. of Pediatrics;
Pediatric Hematologist Oncologist

Welcome new members to both FARF boards and thank you, departing members



We would like to extend our gratitude to **Peter Pless, MD**, and **Kevin McQueen, MBA** for their many years of service on the FARF board of directors. Peter Pless spent six years on the board, offering his medical expertise and serving on two grant review committees and FARF's budget and investment committee. Throughout Kevin's 15 years of service, he lent his expertise to a number of finance and fundraising committees in addition to leading the organization as board president from 2016 until 2018. We are a stronger organization thanks to the contributions of Kevin and Peter and look forward to their continued involvement in FARF projects.



Peter Pless, MD



Kevin McQueen, MBA

As we thank departing members, we also welcome two new additions to the board of directors, **John Connelly** and **Lisa Mingo, CPA**.

John and his wife, Kim, are the proud parents of six children: Evan, Claire, Rachael, Leah, Sarah, and Becca.

John is the Owner and President of D2 Ingredients LP, a manufacturer and distributor of food ingredient technology. Since his oldest child Evan was diagnosed in 2002 at six weeks old, John and Kim have dedicated themselves to helping advance research and working for a cure by hosting numerous fundraisers over the years. In addition to serving on FARF's board, John is very active in the community of Green Bay, Wis. serving on the school board and church finance council.



John Connolly

Lisa joined the "Family" in 2007, when her 5-month-old son, Dylan, was diagnosed with FA. A world traveler, Lisa has a love of meeting new people and experiencing other cultures. She has lived and worked in four continents and currently resides in Vancouver, Canada with her New Zealand-born husband, Mark, and their two boys, Connor and Dylan.



Lisa Mingo, CPA

As a Canadian CPA (Chartered Professional Accountant) and CPHR (Chartered Professional of Human Resources), Lisa has experience working across a variety of industries in the Finance, HR, IT and Project Management spaces. Presently, she owns and operates a small architectural practice with her husband. Lisa is looking forward to her continued involvement in advancing FA research as a board member and fundraiser. She and her family have held community-based fundraisers in Vancouver since 2011. Of particular interest to her is expanding FARF's connection to the international community.

2019 marks a year of renewed energy and motivation to FARF's Scientific Advisory Board (SAB). Four new members joined the SAB in addition to the five new members that joined in 2017. This group of scientific, clinical, and industry experts serves to guide FARF's scientific priorities and offer counsel to the board of directors and staff. We would like to welcome new members **Jennifer Grandis, MD** (University

of California, San Francisco, head and neck cancer), **Melissa Haendel, PhD** (Oregon State University, translational data science), **Kathryn Pennington, MD** (University of Washington, gynecologic oncology), and **Jeffrey Siegel, MD** (Gilead Sciences, translational medicine). They join existing members **Blanche P. Alter, MD, MPH** (National Cancer Institute, epidemiology), **Amy DeZern, MD, MHS** (Johns Hopkins, oncology), **Agata Smogorzewska, MD, PhD**, The Rockefeller University (DNA repair), and **John Wagner, MD** (University of Minnesota, pediatric hematology-oncology).

We would also like to welcome **Premal Patel, MD, PhD** as the new chair of the SAB. Dr. Patel is chief medical officer at Lyell Immunopharma in California and plays an integral role in determining FARF's scientific direction. He steps up as chair following the retirement of **Dr. Ray Monnat** after 20 years of service on the SAB.

Dr. Monnat joined the SAB in 2001 and served as chair from 2015 – 2019. We are tremendously grateful to him for his years of steadfast service and strong leadership as FARF took major steps forward.

“ Working with FARF over the last 30 years has been deeply engaging and rewarding, and I plan to continue with this through the end of my

career. As an SAB member I've tried to guide and grow FA science where I could, to bring useful new knowledge to the many remarkable and courageous patients and families I've been privileged to meet through FARF. There's not been a day these past 20 years without my thinking of how better to deliver on the trust they've placed in me – we can't have answers too soon. ” —Ray Monnat ■



Ray Monnat, MD

FAMILY FUNDRAISING EFFORTS

In 2018, FA families stepped up to fundraise. More than 240 families raised funds, with 135 raising over \$500! Altogether, families raised \$2.91 million for the Fanconi Anemia Research Fund! 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.

\$1,454,000

Lynn Frohnmayer

\$285,000

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

\$89,000+

Kevin and Lorraine McQueen

\$68,000 - \$71,000

Orion and Lisa Marx
Gerard and Cynthia
Vandermeys

\$20,000 – \$31,000

Mark Frohnmayer
Stephen and Jennifer
Klimkiewicz
Timothy and Mary Ann Lana
Todd and Kristin Levine
Alex Winn

\$15,000 – \$19,999

Mauro and Kerrie Cazzari
Mark De Groot and Hanneke
Takkenberg
Matthew and Evelyn Keyes
Peggy Padden
Rose and David Pennell

\$10,000 – \$14,999

Herminia Carvalheira
David and Mary Ann
Fiaschetti
Charles and Kathleen Hull
The Levine-Horrigan Family

Ian and Tricia Mitchell
Tyler Morrison and Rachel
Altmann

Peter and Janice Pless
Neil and Emily Robison
Nigel and Ann Walker

\$5,000 – \$9,999

Brian and Carly Adel
James and Jennifer
Armentrout
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David and Sarah Borden
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Joseph and Nancy Chou
Sultana Graham and Brian
Anderson
Gary and Heidi Grassi
Alan and Rachel Grossman
Andre Hessels and Rutger
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John and Karilyn Kelson
Susan and Skip Longstaff
Daniel and Angie McMahon
The Pearl Family
Leah Petsanas
Andrea and Robert Sacks
William and Connie Schenone

\$3,000 – \$4,999

Michael and Jennifer
Aggabao
Chris and Jennifer Branov
James and Crystal Eubank
Kevin Gatzlaff and Rachael
Alaniz
Daniel and Nicole McCarthy
Adam and Olivia Mindle

Lisa and Jack Nash
Bryan and Karen Siebenthal
Colleen Satterlee

\$1,000 – \$2,999

Assila Al-Marshoudi
Amanda Barber
Joshua Baumiller
Tracy Biby
Israel and Mary Jo Becerra
Adam and Marissa Becker
Jeffrey and Donna Boggs
Richard and Tena Boson
Sean and Allison Breininger
David and Kim Chew
Jerry and Natalie Christensen
John and Kim Connelly
Andrew Coons and Valeen
Gonzales
Donna DellaRatta
Egil Dennerline and Nanna
Storm
Marie Di Mercurio
Justin and Britteny Ferrin
Daryn and Carol Franzen
Allen Goldberg
Andrew and Jennifer Gough
Michael Greenberg
Madeline and Patrick Gregg
Benjamin and Stephanie
Griggs
Gary Haftek
Owen Hall and Margaret
Kasting
John and Martina Hartmann
Jeff and Beth Janock
Erik Kjos-Hanssen and Turid
Frislid
Robert and Anna Langtry

Keith and Jessica Loo
Col. Gregory and Lt. Col.
Lynette Lowrimore
Alaina Mercer
Mark Ritchie and Lisa Mingo
Ronald and Fredi Norris
Fernando Nunes
Nancy Nunes
Ortiz/Jackson Family
George and Kathryn Reardon
Paul and Rena Rice
Craig and Alisha Rushing
Richard and Marilyn Sablosky
Emily Salo and Kenn
Lonnquist
Richard and Dolores Satterlee
Ron and Alice Schaefer
Colleen School
Janice and Kenneth Sysak
Ana Alejandra Tabar Concha
and Elvin Estevez Lopez
William and Mary Underriner
Joseph and Natalie Vitrano
Michael and Kimberly
Williams
Werner and Laetitia
Wolfswinkel
Jessica and Jonathan Young

Up to \$999

Peter and Donna Abramov
Alvaro Acosta
Lea Ann and Jeff Stiller
Marzban and Daisy Ardeshir
Ronald and Junita Arroyo
Andrew Athens and Dr. Vicki
Anton-Athens
Charles Balow and Xandra
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John and Audrey Barrow
Conrad and Joan Bender
Jasmine Bennetsen
John and Francene Berglund
Giovan Domenico Bertolucci
and Federica Bonati
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Katie Bowe
Lisa Buglehall
Donald and Danielle Burkin
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Tony and Monica Cabral
Hugo Canalli

Robert and Barbara Capone
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Mary Eilleen Cleary and
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Marchbein
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Stitely-Lamm
Eddie and Maly Lee
Mayra Lemus
Larry and Gayle Licari

Tanner and Jessica Lindsay
Michael London
William and Jacquelyn
Lucarell
Kory and Julie MacMurray
Leighsa and Paul Makowicz
Aaron and Nicki Marsters
Peggy McDaniel
Kevin and Barbara McKee
Catherine McKeon
Jeremy and Stacey Mefford
P. Michael and Kay Proctor
Carmine Mignone and Albina
Parente
James and Holly Mirenda
Georg and Sabine Mohr
Manuel and Heather Molina
Sidney and Betsy Moore
Kim Mootassem
Griff and Cecelia Morgan
John and Betty Mozisek
Tony and Lina Nahas
Louis and Virginia Napoles
Jack and Tammy Neal
Philip Nelson and Candy
Lindsey
David and Catherine
Neumann
Alice Nicholson
Robert and Mary Nori
Lorraine O'Connor
Joonseok Oh and Sukyung
Jung
David and Stacy Ownby
Seth Parelman
Arianna Pederson and Robert
Bright
Michael and Joanna Peros
Tim and Ashleigh Pinion
Ashley Power
Lynn Quilici
Mario and Yolanda Ramirez
Aaron and Jean Randolph
Pedro and Marina Ravelo
Kelsey Robinson
Paul and Catherine Rodwell
Kevin and Katherine Rogers
Daniel and Bonnie Rosen
Les and Nancy Ross
Stanley and Lisa Routh
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Devon and Rob Tes
James and Kerri Tilson
Bruce and Loreen Timperley
Mark and Susan Trager
Thomas and Cathy Uno
Michael and Beth Vangel
Janet Voorheijen

Marc Weiner
Williams (Clay) Family
David and Erica Williams
Chad and Dawn Wood
Jason and Joan Woodle
Sharonda Woods
Kyle and Madison Wright
Wesley and Susan Wycoff
Sean and Kristin Young
Thomas and Marjorie
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. ■



Holiday Letters

Best Holiday Campaign to Date!

This past holiday season, a record 23 FA families wrote appeal letters to their families and friends to raise money for FARF! Some made online giving pages through FARF's website, some sent letters, some sent family holiday photo cards, and some did a combination of those choices. Thank you to the following families: Sacks, Robison, Janock, Altmann-Morrison, Di Mercurio, Borden, Sysak, Fiaschetti, Horrigan, Hessels, Pearl, Frohnmayer, Franzen, Brannock-Cazzari, Nori, Barber, Ferrin, Walker, Starner, McQueen, Loo, Lana, and Adel!
Thank you!



FUNDRAISING SCRAPBOOK



Mackenzie's Run for Emma Jane

When Mackenzie heard about family friend Emma Jane's Fanconi anemia diagnosis, she wanted to do something to help. On March 9, Mackenzie ran a 5K to raise funds in Emma's honor. Her campaign gained traction and after the race, she sent Emma's family a check to FARF for \$2,300! Mackenzie is proof that anyone, child or adult, can make a difference to research. Way to go, Mackenzie!



Emma's Hope

When one-year-old Emma Jane Barber was diagnosed with Fanconi anemia in late 2018, her parents immediately got in touch with FARF. They wanted to be proactive and involved in Emma's journey and understood that research is vital. Emma's family made a holiday appeal page on FARF's website to raise funds. Since they received Emma's diagnosis, their friends and family have rallied around them to raise nearly \$2,000. Diagnosis can be terrifying, but often families find fundraising to be a source of empowerment and advocacy. Thank you, Barber Family!



Everyday Heroes, Fighting FA

Since 2015, the Graham-Anderson and Robison families in the Seattle area have put on the annual FARF 5K Run to raise funds, bring their communities together, and celebrate their sons. Blake, Isaac and Avery all have FA and all love superheroes. Last summer, both Blake and Avery went through bone marrow transplants. Their incredible families and community came together once again to rally around them, as they juggled transplant and organizing a fundraiser. Over the last four years, the FARF 5K Superhero Run has raised over \$77,000. Thank you, Graham-Anderson and Robison Families!



Planting for a Cure

Four-year old Teddy loves trucks, gardening, painting, and playing at the playground. Last year Teddy helped plant some flowers at his home. Since then, he has developed a green thumb and a lot of pride in caring for his flowers. This spring, Teddy's mom organized a fundraiser to bring beauty to people's gardens. They sold flower and plant seeds, spreading Teddy's joy of gardening and raising \$650 for research. Thank you, Teddy!



Pell Bridge Run

Team FARF does it again! The Fiaschetti Family brought a team together to run in the 5th annual Pell Bridge Run in Newport, RI last fall. David, an FA parent, joined together with FA researcher Dr. Niall Howlett as well as Dr. Howlett's wife and daughters to participate in the run. By the end of the race, the team raised \$8,030. Thank you Team FARF! Interested in joining the team in 2019? The 6th Pell Bridge Run will take place Sunday, October 20.

#GIVING
TUESDAY™

Giving Tuesday

Thank you to all 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 \$20,000 for FARF! That's \$8,000 more than you raised in 2017 – way to go!

You've decided you want to make an impact by raising funds for FARF. Thank you! Now the fun part:

WHAT KIND OF FUNDRAISER SHOULD I HAVE?



EVERY YEAR, FA FAMILIES

STEP UP TO RAISE FUNDS. THANK YOU!

Got your idea? Need help deciding? Get in touch: info@fanconi.org | 541.687.4658

2018 HEALTHLINE AND NORD STRONGER SCHOLARSHIP RECIPIENT

Congratulations to **Matthew Pearl**, 2017 recipient of the Winn/Byrd award, for winning the 2018 Healthline and NORD Stronger Scholarship. This award is given by the National Organization of Rare Diseases (NORD) to those who've demonstrated dedication to the advancement against a rare or chronic disease either through research, patient advocacy, raising awareness, or community building. FARE is grateful for the advocacy and fundraising work Matt has done in the Fanconi anemia community worldwide. **Keep it up, Matt!** ■



DONATE WHILE YOU SHOP ON AMAZON

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



In Loving Memory

Manuel Molina
8.6.04 – 11.2.18

Lelania (Church) Innes
4.13.73 – 12.20.18

Gidion Reuben du Toit
12.27.08 – 11.27.18

Joel Gimenez
6.4.84 – 2.3.19

Deborah Baffour
3.3.13 – 12.3.18

Zephan Cannon
3.5.87 – 2.9.19

Raymond Nelson
4.6.15 – 12.9.18

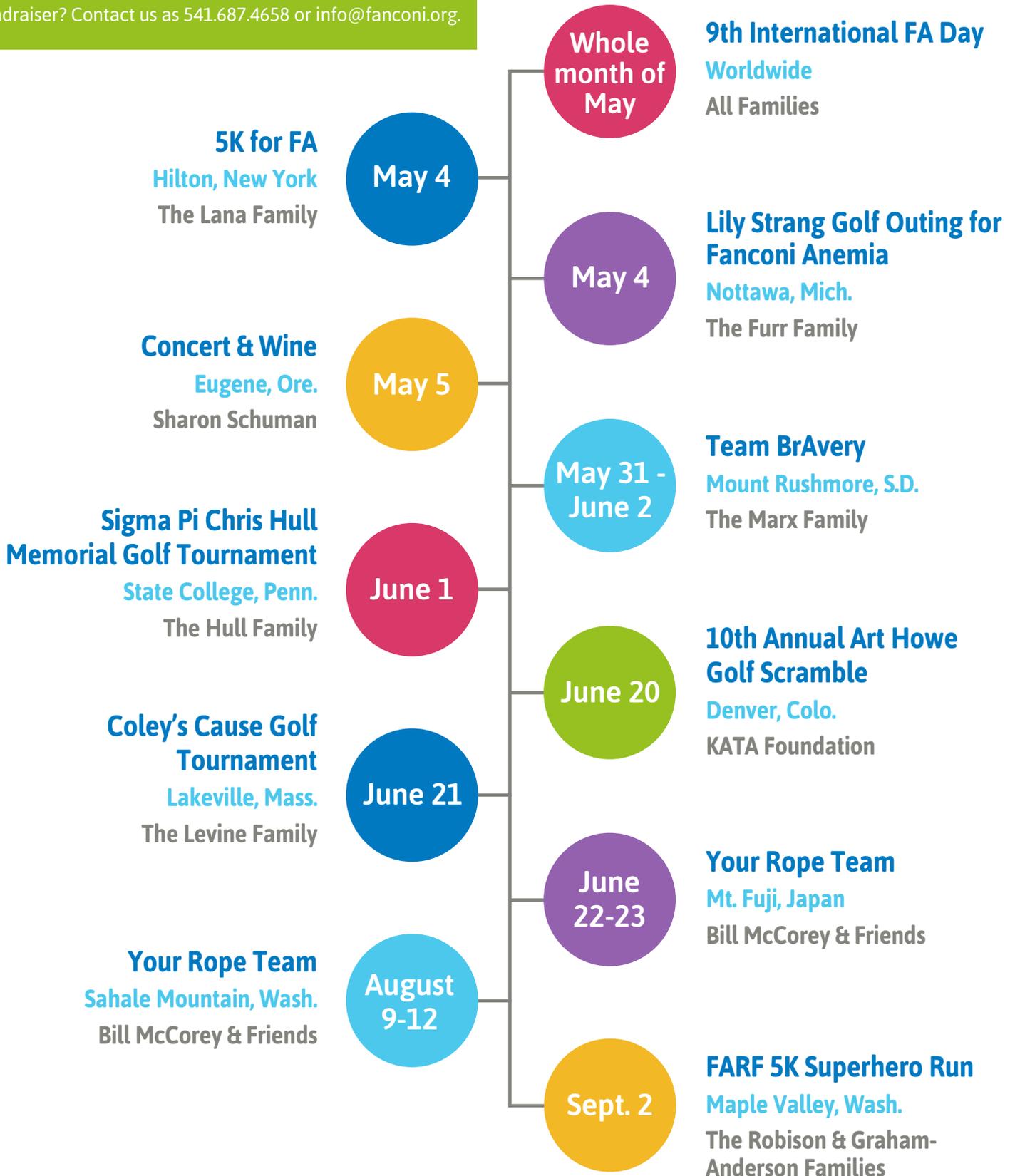
Lerica Vercellotti
8.7.75 – 2.15.19

Colleen Satterlee
10.29.78 – 12.12.18

Anne-Marie Malmgren
1.5.71 – 3.17.19

The Fanconi anemia community spans the entire globe, with events in several different locations. The Fund encourages everyone to participate in FA fundraisers. 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



Sept. 7

Old Quincy Barn Dinner

Denver, Colo.

KATA Foundation

October 20

Pell Bridge Run

Newport, R.I.

The Fiaschetti Family & Friends

Nov. 9

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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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.

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