



FAMILY Newsletter

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No Longer an Orphan

By Sharon Schuman

My subject was born an orphan in 1927. With no known relatives and no connections to a wider community, what followed were 60 years of abandonment. In 1985, one courageous family decided that this isolation had gone on long enough, and that connections to relatives, if there were any, should be found. This family reached out for help from those who were particularly good at understanding this orphan. Investigators chipped away at the mystery and began to uncover clues. Over a 30-year period, bits and pieces of information emerged about a chain of relatives who at first glance seemed distant, then turned out to be close kin. What was once considered an orphan now has an extensive family and has become a celebrity, recognized worldwide. This is the story that I want to tell you.

Fanconi Anemia

Some of you may have already figured out that the orphan I am talking about is not really a person, but a disease—Fanconi anemia—

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TEN OF THE BEST THINGS ABOUT FA FAMILY CAMP

Every summer, families affected by Fanconi anemia come together for five days at Camp Sunshine in Casco, Maine. The Family Meeting (or Family Camp) is a special event that allows families to hear from expert FA researchers and physicians, to attend support groups for help in coping with the disease, to voluntarily participate in FA research projects, and to connect with other families affected by FA. Meanwhile, Camp Sunshine provides a fun-filled program for children with FA and their siblings.

We asked FA families to share with us some of their favorite moments from the 2018 FA Family Meeting. As memories were shared, we noticed some common themes that families seemed to appreciate most about camp. Here are ten of the best things about FA family camp:

Finding your people

"One of my favorite moments was swaying side to side with our arms around each other singing the Camp Sunshine theme song. It was very special to know that at that moment,

everybody had each other's backs no matter what. We understood each other more in a glance than some people do in a lifetime. It was a peaceful easy feeling!"

Reuniting with friends and Family

"My favorite moment is always the big reunion between our son Dylan and his closest friend, Eli."

Giving everyone in the family a place to belong

"One of the best things about camp is knowing my 13-year-old, who does not have FA, actually wants to be there."

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No Longer an Orphan

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the rare genetic disorder that took the lives of all three daughters of Lynn and Dave Frohnmayer. Fanconi anemia was considered an “orphan” disease because it was so rare and so little was known about its causes, connections to other diseases, or any effective treatment. When Kirsten, Katie, and Amy Frohnmayer were diagnosed in the 1980s, doctors could only offer palliative care. Some people might have reacted to this shock with paralysis and passivity, but the Frohnmayers responded with energy, determination, leadership and sheer grit to this unwelcome challenge. In the beginning they hoped scientists might uncover “the gene” that causes FA, then do something about it.

In 2004, not one, but eight genes had been discovered, one of which was *BRCA2*, a breast cancer gene. That one discovery linked FA to breast cancer, a disease that in 2012 affected 1.7 million women worldwide. Since then, four additional breast cancer genes have also been found to be Fanconi anemia genes. Now we know that *BRCA2* is only one of the genes in the Fanconi anemia pathway, all of which, when they function correctly, are essential to the DNA repair process that keeps us alive.

For all of us, the healthy metabolism of our cells requires an elaborate

network of interconnected pathways made up of chemical reactions that involve proteins, enzymes, and the synthesis or breakdown of amino acids. When cells fail, it is because something has gone wrong in the DNA replication and repair processes that are governed by these pathways and are essential to life. In people with Fanconi anemia, one or more of these 22 genes has a mutation that makes it defective. Thus, people with FA have a tougher time than the rest of us repairing DNA damage, whether it is caused internally by normal processes of metabolism, or externally, by environmental factors like radiation therapy, chemotherapy, alcohol, tobacco, or the sun. It is important to know that none of us can avoid all DNA damage. No matter how much sun block we wear, no matter how many glasses of wine we don’t sip or cigarettes we don’t smoke, no matter how many carcinogens we avoid in our air, water, soil, and in the food we eat, our own bodies create DNA damage every single day, just through the normal process of staying alive.

What this means is that from the moment of conception, when cells are dividing like crazy and life is bursting forth, people with FA begin to fall behind in DNA repair. The

damage accumulates at such a rapid rate that FA babies are sometimes born with missing thumbs, bones, or organs. Though some people with FA show almost no outward signs of the condition, their bodies age prematurely from all this DNA damage. As children they often fall behind on growth charts. As their blood cells struggle to keep up with demand, many kids with FA develop bone marrow failure or they get leukemia. If a way could be found to restore in people with FA the DNA repair processes that operate in healthy people like you and me, Fanconi anemia would become a manageable condition like diabetes or asthma. We would not need to find a cure for FA if the disease itself could be made manageable.

Much of the FA research over the past decades has been devoted to figuring out what it is and to coping after the fact with the physical problems it creates. In small children, surgeons fashion thumbs out of fingers and straighten out bones that are bending. Doctors prescribe growth hormones that help FA kids grow. Still, bone marrow failure remains a huge threat to children with FA. One of the best after-the-fact interventions is a bone marrow transplant that replaces the compromised ability of a person with FA to create healthy blood, with a donor’s healthy bone marrow. Because of improvements in bone marrow transplant protocols between 1990, when 20% of FA patients survived the process, and now, when over 90% survive, most children with FA who need

“We would not need to find a cure for FA if the disease itself could be made manageable.”

a transplant now live into adulthood. This is a remarkable accomplishment. Would that it were a cure!

But for people with FA, the DNA repair problem is not just in the bone marrow, or in the blood. It is in every single cell of the body. A bone marrow transplant can fix the blood, but the rest of the DNA damage grinds on, and the result is all too often cancer. Now that more children with FA are surviving into adulthood, what we are seeing is an epidemic of head and neck cancers in FA adults who have had successful bone marrow transplants. These cancers are particularly awful, because they are painful, disfiguring, and usually lethal. Individuals with FA get head and neck cancer at roughly 500 times the rate in the general population. By the time these cancers are detected, it is often too late. So far, the best treatments for these head and neck cancers involve detecting them at the earliest possible stage, so that they can be surgically removed before they spread. BUT, even if month-by-month and year-by-year diligent patients and doctors catch hundreds of these cancers, the DNA damage continues to pile up, and new cancers appear.

Thank You, Fanconi Anemia!

I realize that this all sounds pretty grim, but there are a number of reasons we should all be grateful to Fanconi anemia and to the research it has inspired. Discovering the link to breast cancer was just the beginning. Now that we know that Fanconi anemia is not just a blood disorder, but rather a DNA repair disorder, and that the 22 Fanconi anemia genes are essential to the DNA repair process in healthy people, these genes turn out to be

the very genes that malfunction in various forms of cancer, which is ALSO a DNA repair disorder. Uncovering the secrets of FA's genes and pathways is a step toward uncovering the secrets of all cancers. Right now if you travel anywhere in the world to a scientific meeting that studies blood, radiation, gene therapy, immunotherapy, or various specific cancers, you will hear papers about Fanconi anemia. In this sense, FA has become a celebrity disease, known to researchers all over the world for the insights it can give them about how cancer destroys lives. What was once an orphan disease is closely connected to a very large family of cancers.

On a therapeutic level, FA research has also found a larger family that includes many people without Fanconi anemia. This is how it works. Because FA patients are extremely sensitive to DNA damage, the toxic regimens of radiation and chemotherapy that are routinely used to treat cancer in the general population can be lethal for people with FA. As researchers develop less toxic therapies to treat FA patients, these same drugs can be used in non-FA patients, avoiding levels of toxicity that otherwise kill the very sick or elderly.

Furthermore, because the accumulation of DNA damage in people with FA leads to premature aging, research into the nature of this process has given scientists insights into aging itself, which turns out to be a gradual loss of our ability to repair the damage to our DNA that accumulates day by day over many years. If we could slow that process, what would that mean for the human life span—not just of people with FA, but for us all?

The most cutting-edge FA research today is devoted to trying to discover ways not just to correct bone marrow failure, remove head and neck cancer, or repair birth defects—all after-the-fact interventions—but to slow down the DNA damage that is accumulating and to restore to good health the DNA repair process that could prevent these problems from arising in the first place.

What can we hope for?

Since we can never hope to prevent all DNA damage (which is part of our natural metabolic processes), we are hoping for a breakthrough that will allow us to enable people with FA to cope with that damage as effectively as you and I do. **That would constitute a cure for Fanconi anemia!**

Fanconi anemia was once an orphan disease that no one knew anything about, except that it was rare, it affected the blood and it was fatal. Thanks to 30 years of research tirelessly promoted by the Fanconi Anemia Research Fund, Fanconi anemia is no longer an orphan disease. The DNA repair problem at its center turns out to be the problem that characterizes all cancers. Thus FA is deeply embedded in a huge family

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Sharon Schuman visits dear friend Amy Winn in the hospital in 2016.

Ten of the Best Things about FA Family Camp

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She makes friendships and memories that carry her the whole year through. It makes my heart smile. Most days are spent dealing and worrying about our daughter with FA, so we love knowing that our children without FA are happy at camp. That is the best feeling in the world!"

Feeling it all in support groups

"The support groups are our favorite part of Camp. It's such a special time, when we can share the best and worst moments of living with FA, with the only other people in the world who really get it."

Hearing from the best FA experts in the world

"We're always grateful for the updates from FA researchers and doctors. The panel on bone marrow transplant is so informative, and we love learning about trials and therapies on the horizon. It's great to see how our fundraising efforts are literally paying off."

Kicking back and having fun

"We love hanging out and swimming at the beach area as a family on Tuesday afternoon (the free day)!"

Releasing balloons and remembering

"The emotional power of the balloon release caught me unawares and certainly made me pause to reflect on what more I could do to help reduce the number of balloons in the future. A very poignant moment."

Leaning on your community

"Wanting to capture some memories before we left camp, we asked a couple of Bella's friends to get in a picture with her. What started out as a group of 3 quickly grew to 4, then 5, then more, all gathered around Bella. As I looked at the picture later, I was reminded what an extraordinary tribe of support Bella has from the FA community. Friends both with and without FA surround her with love and support—physically when they are at camp, but always in spirit throughout her FA journey."

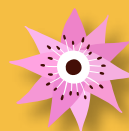
Bonding between FA parents

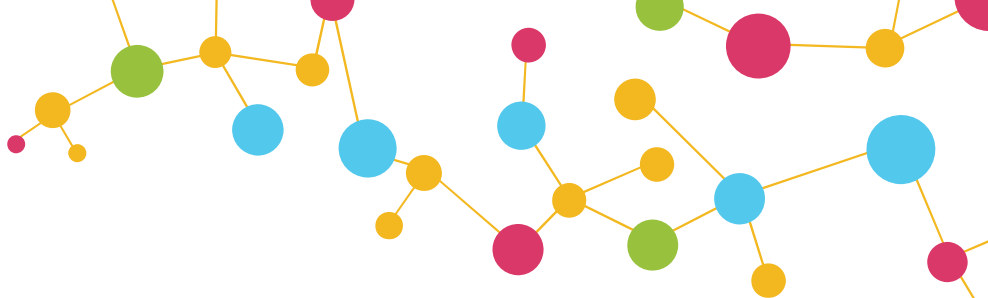
"I love the bonding between FA moms, working out the stress of having a kid with FA."

Welcoming the joy

"Our favorite moments are watching the joy on our son Eli's face. Every day he makes a new friend. Those memories will forever stay with our family."

Please join us at 2019's Family Meeting. Look out for an invitation in early 2019, or contact marie@fanconi.org to learn more. ■





Global Fanconi Anemia Gene Therapy Program Expanded

Gene therapy trials recently conducted at Fred Hutchinson Cancer Center (Seattle, Washington) and at CIEMAT/Hospital del Niño Jesús (Madrid, Spain) were developed to investigate whether gene therapy could potentially prevent bone marrow failure and thus the need for a bone marrow transplant. The Seattle trial had three patients; the Madrid trial had six. All patients had Fanconi anemia complementation group A (FANCA) mutations.

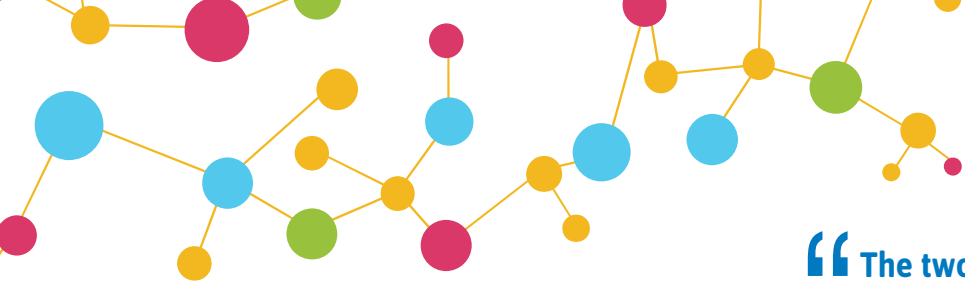
The two independent trials used different protocols to remove, purify, and treat the stem cells (see the diagram on the next page), which allowed the research teams at Madrid and Seattle to test key study protocol variables that would determine the best patient outcomes.

The Madrid study enrolled younger patients (age 3-6) while the Seattle study enrolled patients ranging in age from 5 to 22 years. Additional differences between the studies included the amount of harvested stem cells (the Madrid team isolated higher levels) and stem cell purification methods. Recent results from each trial show that the safety profile of gene therapy is favorable and that the observed side effects in each study were not considered serious and were attributed to the procedure to remove the stem cells.

Evidence of long-term engraftment of the FANCA-corrected hematopoietic stem cells was observed in the Madrid study. The corrected FANCA gene in blood and bone marrow cells was observed in four of four patients at follow up (up to two years post infusion of the corrected cells). The blood counts in these four patients were also stable, which was an improvement from the low platelet and neutrophil levels typically observed during the months/years prior to gene therapy. In contrast, patients enrolled in the Seattle trial did not exhibit long-term engraftment of cells corrected for the FANCA gene (tested at 1.5 years post infusion of the gene-corrected cells), although the patients did demonstrate stable blood counts. The difference in engraftment results from the two trials could be attributed to several factors, including the age of study participants, and differences in the cell isolation, purification and transduction protocols between the institutions.

In the Madrid study, the number of gene-corrected blood and bone marrow cells increased in the patients over time, and these cells became progressively resistant to DNA-damaging agents. For example, two of the four patients on the Madrid study had increased resistance to DEB-induced





chromosomal fragility tests at two years post corrected cell infusion. The level of resistance to the DNA damaging agent was similar to FA mosaic patients who exhibit spontaneous correction of genes. Spontaneous correction is the correction of a faulty gene that happens naturally without the use of gene therapy or gene editing. Results from the two most recently treated patients at Madrid are expected later in 2018 and in 2019.

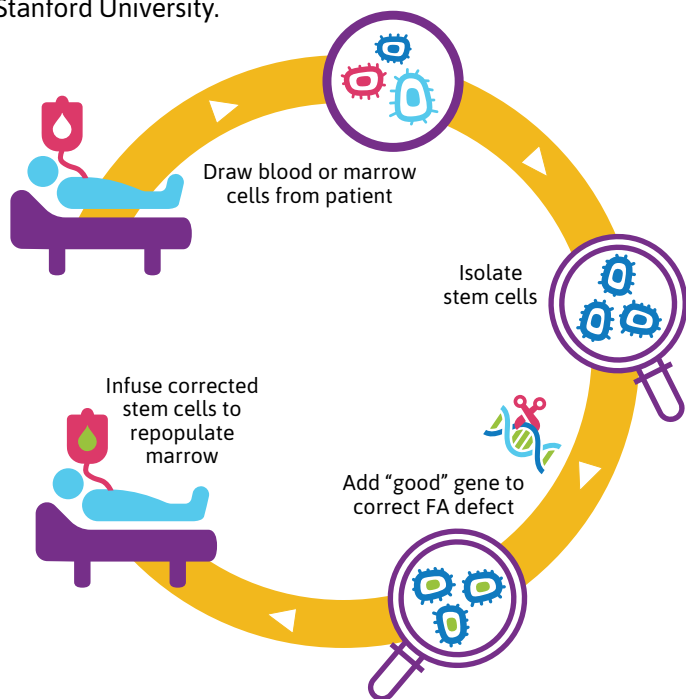
The two trials demonstrate that gene therapy has a high safety profile, and the trial in Madrid specifically showed long-term engraftment of corrected cells. These results are promising, but more research is needed in order to understand how best to administer gene therapy so that all patients have long-term engraftment of gene-corrected cells with minimal side effects.

In order to address this, the global Fanconi anemia gene therapy program will be expanded. There will be new trials available to people with FA in the US and Europe in 2018 and 2019. In Europe, a new study, known as FANCOLEN-2, will be available at CIEMAT/Hospital del Niño Jesús, and then at University College London/Great Ormond Street Hospital in England. In the US, a similar study will become available at Stanford University.

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The new global FA gene therapy trials will seek to enroll patients ages 1 through 12 with FANCA mutations who have not developed severe bone marrow failure. The trials will utilize the lentiviral vector developed by the Madrid research team and will incorporate key learnings from the recent gene therapy studies conducted in Madrid and Seattle. The therapy will be administered without conditioning chemotherapy, and will require a (relatively) short hospital stay to collect stem cells over an approximate 6-day period. This will be followed by a single intravenous administration of gene-corrected hematopoietic cells over the course of one hour or less. Patients treated to-date have been able to leave the hospital within 1-2 days after receiving the therapy. Following treatment, study visits will be scheduled over the course of three years.

Both the US and European programs are sponsored by Rocket Pharma (New York, USA). Financial assistance for travel, housing accommodations, and other support during both the initial treatment and subsequent follow-up visits will be provided to families. ■



If you or your family member may be interested in enrolling in the gene therapy trials and would like to learn more, please contact:

Dr. Juan Bueren: juan.bueren@ciemat.es

Dr. Julian Sevilla: julian.sevilla@salud.madrid.org

Dr. Agnieszka Czechowicz: aneeshka@stanford.edu

Dr. Gayatri Rao: gr@rocketpharma.com

Pilot Study of Metformin Opens at Boston Children's Hospital

For over a decade, leading Fanconi anemia investigators including Dr. Markus Grompe, Dr. Alan D'Andrea, and Dr. Grover Bagby have investigated various drugs that might improve blood production and prevent cancer in FA. In an FA mouse model, Dr. Grompe discovered that, of the six compounds he tested, the **ONLY** drug to both improve blood counts and delay onset of solid tumors was metformin, an aldehyde scavenger.

In an FA cell line, metformin reduced the levels of spontaneous radials and chromosome breakage. Studies of the non-FA population suggest that metformin may have anti-cancer properties for a wide range of malignancies including breast cancer, lung cancer, colorectal cancer and liver cancer.

“ Dr. Grompe discovered that, of the six compounds he tested, the **ONLY drug to both improve blood counts and delay onset of solid tumors was metformin, an aldehyde scavenger. ”**

Metformin was approved by the FDA in 1994, and is the most widely used medication for diabetes in the world. It has a long track record of safety, and has been extensively studied for gestational diabetes, obesity, insulin resistance, and cancer.

Researchers at Boston Children's Hospital have opened a clinical trial of metformin, planning to enroll 22-24 patients at Boston Children's Hospital.

Enrollment criteria include ages 6-35 and a low blood count in at least one of the following: hemoglobin, platelets or neutrophils. Exclusion criteria include bone marrow transplantation (or plans to go to transplant in the near future), pregnancy, use of a medication to improve blood production (an androgen or growth factor), low blood sugar or type 1 diabetes, leukemia or myelodysplastic syndrome, or solid tumor undergoing treatment.

Patients on this study will travel to Boston twice: at the beginning of this six-month trial and at the end of the study period. Reasonable travel expenses will be reimbursed. The study will measure improvement in blood counts and marrow cellularity, and will monitor for safety and tolerability. This clinical trial also includes additional

biologic studies that will assess the potential anti-cancer effects of metformin by studying changes in chromosomal breakage and buccal swab micronuclei and advance our understanding of how metformin might improve the health of patients with FA. For additional information about this trial, contact Dr. Elissa Furutani: by email elissa.furutani@childrens.harvard.edu or by phone at 617-632-1978.

Metformin Safety

Metformin is an oral medication with a long track record of safety.

There have been no cases of lactic acidosis in almost 70,000 patients who were taking metformin in over 300 clinical trials. Reported patients in clinical practice who developed lactic acidosis while taking metformin had other reasons for developing this complication, such as diabetes, heart failure, kidney failure, and/or severe infection. In fact, rates of lactic acidosis in diabetes patients were similar regardless of whether individuals were taking metformin or other diabetes medications.

Even though studies have demonstrated that metformin does not cause lactic acidosis when prescribed appropriately, this trial has set strict eligibility criteria in consultation with an endocrinologist experienced in the care of Fanconi anemia to make sure that the participants of the trial have adequate organ function prior to starting metformin treatment. ■



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

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

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

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

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, or email NCI.IBMFS@westat.com

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: Ashley E Kuniholm | 617-355-6513 | ashley.kuniholm@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. ■



Joel Walker Inspires Scientific Meeting Series to Advance FA Research

Joel and Joanne Walker were talented students, good at sports, sociable and happy. Some of their happiest memories as children were spent visiting family and friends in England and Australia. As seemingly happy, healthy children, no one would have guessed that they both had Fanconi anemia (FA). At age 33, Joanne was diagnosed with FA a few months after she developed esophageal cancer. Following this diagnosis, she immediately began chemotherapy and radiation – standard treatment protocols for cancer but very dangerous to those with FA. These treatments caused an immediate deterioration of her

bone marrow function and blood levels. After many tests to find the cause of this deterioration, she was finally diagnosed with FA in March 2013. Joel volunteered to be tested as a sibling donor match in case a bone marrow transplant became a possibility. It was then that the Walker family learned that Joel also had FA. Joanne's blood counts never recovered sufficiently from the chemotherapy. She passed away on June 24, 2013 at the age of 34.

Watching Joanne throughout her journey, Joel knew what he could potentially face and was closely monitored for bone marrow deterioration and cancers. It was devastating when Joel was diagnosed

with cancer of the hypo-pharynx in August 2015. In November of that year, a radical laryngectomy was performed to remove his voice box. Joel then spoke and breathed through a hole created in his throat, and for a while, it appeared the cancer had gone. However, it returned a few months later, along with esophageal and mouth cancers. He was deemed inoperable shortly thereafter, so he started on a variety of immunotherapy treatments. Joel continued to deteriorate as the tumors grew, but insisted on trying newer experimental treatments and did not want to take hospice care. Joel passed away on November 1, 2016 at age 33.

Research into treatment for FA was a high priority for Joel, along with the hope that he could help others, so he left a large part of his estate to the Fanconi Anemia Research Fund (FARF). The Walker family worked with FARF to set up the **Joel Walker Scientific Meeting Series**, which supports focused scientific meetings on a variety of topics, beginning with head and neck cancer prevention and treatment in FA.

Head & Neck Squamous Cell Carcinoma (HNSCC): Developing Guidelines to Maximize Therapeutic Outcomes

The first Joel Walker Scientific Series Meeting took place on April 9, 2018 at the University of Pittsburgh. The meeting brought together the clinical and scientific expertise of physicians and researchers who treat people with FA or have extensive research programs focused on the disease. The goals of this meeting were to begin developing standard of



Joel Walker Scientific Series Meeting

care guidelines and future research objectives to maximize therapeutic outcomes for treating head and neck cancer in people with FA.

What do we know about head and neck cancer in people with Fanconi anemia?

- People with FA are at an increased risk to develop cancer (500-700 times higher than in the general population)
- Reasons for increased risk are not well understood
- Treatment options are limited

People with FA diagnosed with HNSCC have limited options for treatment because of low tolerance to adverse effects from highly toxic radiation and chemotherapy. This often leaves surgery as the only viable treatment option, which is highly problematic because FA patients are typically diagnosed at an advanced stage with highly aggressive disease. The surgical removal of tumors is currently the best treatment option, but this is usually not curative for FA patients who often acquire secondary tumors or have aggressive tumors that are difficult to remove.

Aside from surgical intervention, the current treatment guidelines for FA HNSCC include preventative measures

such as (1) abstaining from carcinogens including alcohol and tobacco, (2) maintaining oral hygiene, and (3) participating in extensive oral cancer screening measures.

Researchers and clinicians are working with FARF to develop a roadmap for clinical care for FA patients diagnosed with HNSCC. Basic and clinical research is needed to develop an understanding of the molecular mechanisms driving FA HNSCC and tolerance to targeted therapies so that FA-specific treatment plans can be established.

So, what do we do?

- 1) Improve strategies for early detection by expanding current screening protocols and developing a brush-biopsy kit to be used by patients, families, and medical professionals.
- 2) Develop a biorepository so that researchers have access to tissue for study and patients have the possibility for personalized care.
- 3) Research the potential of immunotherapy for FA patients, beginning by characterizing their immune systems and moving into trials.
- 4) Develop more clinical trials specifically for people with FA to truly understand potential and

efficacy of therapies.

- 5) Assess if treatment options in non-FA patients could be applicable for FA patients.
- 6) Establish FA cancer centers of excellence so that people with FA have access to expert teams of physicians and scientists. In addition, a virtual FA tumor board or advisory panel of experts could be enlisted to manage FA patients with HNSCC who would be unable to attend the centers of excellence.

What needs to happen next

The challenges of preventing, detecting, and treating HNSCC in FA patients are many. The Joel Walker meeting identified clear areas that need to be addressed from both the research and clinical treatment perspectives moving forward. FARF is poised to lead these efforts and we will focus immediate efforts on (1) developing the tissue biobank and (2) establishing a personalized cancer prevention/detection brush biopsy kit for all FA families and (3) developing chemoprevention clinical trials using compounds already approved by the FDA for use in other cancers. ■



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To learn more about the specifics discussed at the meeting and for a more detailed plan of action, visit www.fanconi.org/news.



Navigating Financial Resources

By Rachael Alaniz, social worker and FA parent

The Fanconi anemia diagnosis affects every aspect of life, including healthcare, family life, school, social life, and how you make financial decisions. What are the choices when trying to make sure healthcare is covered? For individuals with FA and their families in the United States, here are some options to consider:

01

Private Insurance

Private health insurance offered through an employer is extremely valuable to families of children with special medical needs. It's important that you understand what your health plan covers as well as any out-of-pocket expenses for which your family will be responsible.

If you already qualify for health insurance coverage under the terms of an employer-sponsored plan, you can apply for income-based Medicaid and receive monthly premium support. Families who are over-income for income-based Medicaid may still be able to access Medicaid through a waiver. Families may also be able to supplement their private health insurance plan through Title V funding or with Supplemental Security Income (SSI) payments (see below).

This means it could be possible for a family of a child with Fanconi anemia to have private insurance as their primary insurance and Medicaid through a waiver or the Tax Equity and Fiscal Responsibility Act (TEFRA) as secondary (essentially picking up any copays or co-insurance for the child). In addition, Title V could provide tertiary insurance, picking up any expenses related to a qualifying diagnosis not

covered by the primary or secondary insurance while also receiving SSI payments.

02

Medicaid

For low-income families, it is important to apply for income-based Medicaid if the child is not covered by a private plan. Medicaid is funded by the state and federal governments and provides necessary medical care for low-income individuals. Federal policy requires that each state have a Medicaid program, but since programs are administered at the state level, program names and income qualifying categories will vary from state-to-state. To find out about Medicaid programs in your state, visit www.Medicaid.gov.

When a child with a special medical need does not qualify for Medicaid based on his/her family's income, it can still be accessed in two ways: through a Medicaid waiver or through the deeming process (see the next page).

03

Social Security

The Social Security Administration (SSA) administers two different programs; Social Security Disability Income (SSDI), and Supplemental Security Income (SSI). SSDI is provided to adults who qualify as disabled and

have worked and paid into the social security system long enough to qualify for the program. SSI is offered to low-income people with disabilities who either have not accumulated enough work credits over their lifetime to access SSDI or who have never worked. SSI benefits can be paid to people at any age, whereas SSDI payments are only paid out to adults.

For children under the age of 18, the SSA provides SSI payments to children in families with limited resources who also meet the agency's definition of disability. Families must first qualify financially for benefits through SSA before the child's disability will be considered.

For those age 18 and older, only the income and resources of the person who is disabled counts. That means that even if a child didn't qualify for SSI as a child with a disability due to the family's income, an adult child may qualify. A child can apply for SSI up to 90 days before his/her 18th birthday. If a minor child is already receiving SSI benefits, the child will be reevaluated for SSI within a year of his/her 18th birthday.

The definition of "disability" for an adult is different than that of a child. For a child to be considered disabled, s/he must present with significant functional limitations whereas, for an adult, the focus is on the ability to work. This means, if a child did not meet the agency's definition of "disabled" under the age of 18, s/he may meet the criteria as an adult. To be eligible for SSI as an adult, the individual's disability must make them unable to work at "a substantial level", which at the current time is to make \$1010 a month or more. In addition, the disability must be expected to last at

least a year or to result in death.

Adults receiving SSI benefits are also able to work or attend school. SSI recipients can continue to work and receive benefits until the time their pay and other income exceeds the income limits for SSI. The SSA also offers work incentives to adults living with disabilities who wish to work.

Since Medicaid comes along with SSI, if a person exceeds the SSI income limits, Medicaid will be stopped. Even if your SSI payments stop, your Medicaid can stay in place if you are eligible under a waiver plan. Therefore, it is important to consider applying for a Medicaid waiver even if you are already receiving Medicaid through SSI as this provides more stability in healthcare financing.

To apply for SSI, call 1-800-772-1213 to make an in-person appointment at your local SSA office.

04

Title V Programs

Each state receives a federal block grant to administer a Title V program, often called “Children Special Healthcare Services” or “Children Special Healthcare Needs”. The program works a lot like Medicaid but has more lenient financial criteria for qualifying. The associated diagnoses related to Fanconi anemia are considered qualifying diagnoses (hematological disorders, hearing loss, visual impairment, seizure disorders, etc.). This insurance is offered in addition to Medicaid and private insurance, so it should pick up expenses related to the qualifying diagnosis that might get denied by Medicaid and/or private insurance. Children can be covered until their 22nd birthdays.

05

Institutional Deeming

Under the Tax Equity and Fiscal Responsibility Act (TEFRA) children receiving care in an institution may qualify for programs they were previously unable to access due to being over-income. Institutional deeming is a valuable and underutilized healthcare financing option for families who don't qualify for government funded financial assistance or insurance programs based on income, or who might be on a waiting list for a Medicaid waiver. Medicaid and SSI can offer support to children who have been hospitalized for an extended period of time, usually after 30 days. Through a process called “institutional deeming” these programs can become available to all children who qualify based on medical condition and length of stay in hospital. To learn more, including if yours is one of the 18 states that offer this service, visit www.fanconi.org and look under “Patients and Families”.

06

Medicaid Waivers

Every state has a Medicaid Waiver program, known as Katie Beckett waivers, which allow a child with a disability to qualify for Medicaid regardless of that family's income. In order for a child to be eligible, the child must need the level of care provided in a hospital or a nursing facility or intermediate care facility. While each state must provide waiver services for children meeting the levels of care set forth by the Katie Beckett law, states also have a right to define hospital and intermediate levels of care using their own standards. Therefore, it is important to reach out to an expert in

your state to learn which waivers might be available to your child. To locate Medicaid Waiver programs in your state, visit: www.kidswaivers.org

07

Other Funding Options

Even with all the aforementioned programs, families still sometimes find themselves in need of financial assistance as a result of their child's special medical needs. Community Foundations, Townships Trustees, and Civic Organizations all have a record of helping families in need. On the national level, United Healthcare Children's Foundation is a nonprofit that provides grants to cover out-of-pocket medical expenses for children under the age of 17 that are either not completely covered or not covered at all by a family's private health insurance. This program is offered to families who meet their generous financial criteria and are not currently receiving any government funded healthcare coverage. Additional information and an online application can be found at www.uhccf.org.

08

Important to Remember

Programs and systems change frequently. It is important that you talk to someone whose sole job it is to remain well-versed in all systems and how they intersect in order to ensure your family is accessing all available programs. Hospital financial advocates are an excellent resource. The Federal Department of Health's Health Services and Resources Administration also funds a Health Information Center in each state to help families

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Optimizing Success in School While Living with a Chronic Disease

The complexities and challenges of a chronic illness like Fanconi anemia (FA) tend to focus the attention of parents on the physical effects the disease has on their child. Monitoring of blood counts, bone marrow biopsies, initiating a stem cell donor search, and deciding on a transplant center are only a few of the many physical needs requiring attention. Of great importance that may take a back seat are the educational needs and accommodations of those living with FA. At the 2018 Family Meeting, Valerie Theile, MEd, School Liaison Specialist at Cincinnati Children's Hospital, highlighted some of the challenges faced by children and adolescents with FA and interventions available to help them succeed in school.

Implications

The educational implications of a chronic disease are seen in three areas: 1) physical/medical; 2) social/emotional; and 3) cognitive. Potential or actual challenges in each area must be identified and met with appropriate interventions. Communication with teachers and all those involved in a child's school life is of highest priority for maximizing educational, social and behavioral potential. Making staff

aware of the precise needs of the child, is crucial.

Considering the child's own perceived needs is equally important. While parents and caregivers tend to focus on the medical/physical needs, children and adolescents focus on the social and emotional aspects of their school experience. The physical demands of a chronic disease can easily result in isolation, which leads to a negative affect on social skills and possible behavioral problems. Keeping kids connected to peers and teachers can help minimize negative effects of isolation. This is a challenge when a child is limited to home instruction. Find an extensive list of creative ways to keep children and peers connected during absences at www.fanconi.org under "Patients and Families".

Educational Plans

Public schools

Once the needs of the child are identified, three educational plans are available to accommodate public school students: 1) Individual Education Plan (IEP); 2) 504 Plan; 3) Individual Health Plan (IHP). Each plan addresses different needs. Plans can be used together, but this is not common, as all accommodations

found in a 504 plan can be included in an IEP, and the IEP can provide services not available in a 504 plan. A description of each plan, as well as a comparison of an IEP and a 504 Plan can be found on FARF's website.

Non-public schools

Education plans for private school students are different, in that the school has varying degrees of responsibility in meeting the needs of the student. For instance, home instruction/tutoring for the private school student might come at a cost, whereas the same accommodation for a public-school student is free.

In addition to public and private, other school systems exist (charter, online, homeschooling), and programs are available depending on the system. Information on student rights, school responsibility and resources can be found online by state. Additional help is available through physicians and clinical institutions. Most large medical institutions have education/school intervention programs, such as the one at Cincinnati Children's.

Other Interventions

Early identification and intervention of the needs of the child, adolescent, or even young adult student, combined with ongoing, effective communication between all parties involved, gives the most promise for success in school. Theile recommends starting the education plan as early as possible, even as early as preschool, so that the transition to elementary school is smoother, with needs identified and accommodations provided. In addition to the accommodations and services provided by the different education

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Scientist Spotlight

Name: Agata Smogorzewska, PhD

Institution: The Rockefeller University, New York, USA

Area of expertise: DNA repair

My work:

My lab works on diverse aspects of Fanconi anemia (FA) and repair of DNA. A lot of our work starts with samples from FA patients. We identify new genes and new mutations that may explain how different patients present and progress; for example, why they get bone marrow failure early or late in life, and why they develop cancer. We strive to understand in great detail how the DNA repair processes work in FA and non-FA cells. A strong component of our work is to study the tumor samples from people with FA to understand how the cancers develop, so we can help identify them early and find better treatments.

What motivates me to work on FA:

I became interested in Fanconi anemia due to the critical function of FA proteins in DNA repair. Quite accidentally, I discovered a Fanconi anemia gene (FANCI) when I was characterizing unknown proteins participating in DNA repair in Steve Elledge's laboratory at Harvard. My clinical training exposed me to Fanconi anemia but going to my first FARF Symposium really opened my eyes to all of the unanswered questions in the field.

Meeting families affected by FA was also remarkably motivating for me to continue to be in this scientific field. I was thrilled to be offered an independent position at The Rockefeller University which was the home of the International Fanconi Anemia Registry started by Dr. Arleen Auerbach in 1982. The registry provided me with cell lines from patients who did not know their mutations which allowed us to identify new FA genes and study how they work in DNA repair. Now, we have multiple ongoing studies that use the samples in the registry.

I believe that it is our duty to use what was gathered over the last 36 years to understand the disease better and provide some answers to the families who contributed their cells and clinical information. Our most recent work on cancers is motivated by meeting people with FA and FA families affected by cancer. It feels very personal to know the people behind the samples in the registry. There is no single day that I don't think about Amy, Chris and others with FA who have lost their battle with cancer.

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

In one of the great art institutions in New York: the Metropolitan opera, the NY Philharmonic, the Carnegie Hall, the Metropolitan museum or the Frick Collection. I also like just walking in New York and getting energized by the city.

Anything else you want FA families to know?

The FA families are my heroes. Your resilience always amazes me and motivates me in daily life. You are also such great advocates for research on FA. Your support of our research through participating in the registry and through supporting FARF, which provides us with funds to perform studies with cancer samples, is essential and deeply appreciated. ■



The Smogorzewska lab

The Sibling Journey

By Nancy Cincotta, LCSW, MSW, MPhil
Psychosocial Director, Camp Sunshine at Sebago Lake



Sibling relationships can be the longest, strongest and at times the most complex relationships in a family. During the formative years, siblings share life's journey and all it brings. What is it like to be the sibling of someone with Fanconi anemia? How is life as a sibling influenced by FA?

Being a sibling of a child with FA is a lifelong role. What is understood, and how siblings cope will vary. Siblings' knowledge about FA is determined by their stage of development, the family's explanations of FA, the context of their sibling's current FA treatment, educational materials available to them, communication with professionals, contact with other families, attendance at FA family meetings, information found on the internet, things which are overheard, etc.

Common themes voiced in FA sibling groups

- Worries about siblings with FA and parents
- Pride about their sibling's endurance and capability
- Confusion, concern, sadness about FA and its future course
- Understanding the genetics of FA
- Resentment of the attention that sibling gets
- Guilt about feeling resentment
- Guilt that they do not have FA
- Feeling left out, isolated
- Assuming additional household and caretaker responsibilities

As siblings talk in groups, you hear their emotional responses: "I have too much anxiety to be so far away." There

is a focus on the present: "Right now she's healthy and I think it's better to live in the now," and a subtle alluding to the future cancer risk, "We all wear sunscreen to encourage her to wear sunscreen."

When you listen to siblings along developmental lines you begin to see a continuum of responses. At first, "I thought it (FA) was a passing thing, that it was good she had this and not cancer. I didn't realize this was something that would last her whole life." Then, "It gets scarier as you get older. When you're young, parents seem so calm and they have it together. When you're older, you see through that. You see more how they really feel..." and finally, "You know more about the outcomes and you can think about it in a different way."

Bone marrow transplantation

When a child has a bone marrow transplant, the fabric of daily life is changed, forcing a degree of instability (which may also provoke independence and growth). There are other losses

that may seem invisible: a parent no longer walking a child to school, reading a bedtime story or helping with homework.

As an FA sibling, you are a potential donor for transplant. There are personal, physical, and emotional responses to donation. And there can be disappointment if you are not a match: "Wasn't a match. It's weird to know that I'm nothing, and I want to help, but I can't." And then there is how the transplant looks through the sibling's eyes: "It was weird looking at my brother; he looked totally different than the last time I saw him. He had no hair." And then there are positive memories: "The only thing I remember was going into my sister's room and pressing the button to start the transplant..."

Siblings of all ages need

- to be understood, and to be understanding
- to feel as important and cared about as their siblings with FA
- to feel that things are fair (not necessarily equal)
- to be part of the FA family dialogue
- to have a version of "family time"

Communication

I have heard siblings tell me that parents should ask them how they are doing; they do not want to burden them, but they feel that it is their parent's job to check in on them. Not talking about issues between parents and children takes two parties; children acclimate to what they are exposed to:

“Afford siblings a voice and hear what they have to say.”

"I saw my mom crying about it and I thought asking her questions might make it worse."

"You need to watch for the right time to ask questions and not just ask it as soon as it pops into your head. It's a waiting game."

Sibling groups, locally and at the FA family meetings, are helpful experiences, as they provide support and minimize sibling feelings of isolation. Sibling comments from a recent support group included: "It's easier to talk to people in a similar situation as you." "It makes you feel not so crazy when things get messy."

Developmental and emotional considerations

"It's kind of a waiting game [for sibling to get a bone marrow transplant] ... an uncertainty. It was an added layer of 'what if' to my life when I left for college."

Siblings should be educated about FA in an ongoing way, having explanations grow as they grow. Children of all ages should be afforded the opportunity to talk and allow time for dialogue. Address their medical questions. Prepare them for what is happening; things left to the imagination can be



Support group for siblings at the 2018 Family Meeting.

more frightening than reality. Help them anticipate changes in their sibling's medical status and explain new issues as they arise. Appreciate the challenges of leaving for college, of pursuing their own goals.

Issues such as carrier status become very important to siblings on their life journey, and they benefit from ongoing support and dialogue about this issue.

Family-focused activities and activities with siblings

Families can benefit from focusing on activities and experiences that promote growth and encourage resilience. Develop family rituals in the context of FA and current treatment regimens, i.e., transfusions, pill-taking, procedures. Create family memories: memorable moments which encourage family

cohesion that have nothing to do with FA.

Set aside special time alone with siblings, regardless of how limited the time (grocery shopping, getting ice cream, playing a game). Let them know how much you care about them. Support and reinforce siblings' importance in the family.

In one group when asked for one word to describe being a sibling of someone with FA, the responses were: hardship, hope, hurricane, patience, unusual, inspiration, strength, challenging, stressful, faith, control, growth, appreciation, family, and different.

Coping as a sibling is a marathon, and the terrain changes over the course of time. Afford siblings a voice and hear what they have to say. ■



Get Ready!

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FA DAY | MAY 2019



Marcos, Silvana, and Marina

Let's Meet at the Porch?

By Marcos Teixeira

Our only child, Marina, is a lovely, sweet girl who loves going to the beach and riding bicycles (especially at her grandparents' farm). She is 11 years old and we are so thankful she is doing well.

Just after her Fanconi anemia diagnosis in early 2015, we were informed that she did not have a matched donor for a bone marrow transplant. We rushed to ask our relatives and friends to register as donors. However, the local government agency responsible for the registry informed us that they could not register at that time. We were so baffled. This is because in Brazil, where we live, the registry is state-owned and to reduce the costs associated with HLA testing, the government stipulated an annual limit, or quota, of donors that could be registered. So, beyond dealing with all of the difficulties associated with the FA diagnosis, we also had to face this kind of frustrating bureaucracy.

We knew we had to act, so we got in touch with a lawyer and filed a lawsuit. We've received a favorable decision in our state, but unfortunately it is not applicable to the whole country, meaning that quotas for donors are still in place in most parts of Brazil. However, we continue on, knowing that the Brazilian registry needs many more donors in order to increase its genetic diversity.

Even with our efforts, we still haven't found a match for Marina, so we considered enrolling in a gene therapy clinical trial. We got in touch with the teams in Seattle and Madrid, and the doctors and specialists involved gave us their full

attention and were extremely kind. However, we learned that the chances for best results would be higher when a large number of stem cells could be collected. In Marina's case, this number was barely high enough to meet the threshold of the eligibility criteria. Therefore, we do not know whether she would benefit much from the gene therapy clinical trial.

Given this information, we will rely on Danazol (when needed) and will take a close look at the Eltrombopag, metformin and quercetin clinical trials. We also decided to register in the International Fanconi Anemia Registry (IFAR) and to enroll in the study on cancer risks (www.marrowsfailure.cancer.gov) presented by Dr. Blanche Alter and Dr. Neelam Giri at Camp Sunshine. We encourage everybody to take part in both the registry and the study—let's all be brave enough to fill out their long questionnaires!

We learned about all of these possibilities thanks to FARF. When I was in Seattle, I even had the opportunity to visit the Robison family, who welcomed me so warmly! Earlier this year we went to Camp Sunshine for the very first time. What an amazing and invaluable experience! We really felt at home, as part of a very united group. We want to give special thanks to our mentor family—the Ritchie-Mingo family—who were so great and helpful. We also thank all the doctors, investigators, volunteers and the invaluable staff of FARF and Camp Sunshine.

In Brazil, we went to a pretty large FAMily meeting last year. We clearly understood that a lot needs to be done for FA patients here. For instance, low-income FAMilies depend on lawsuits to obtain vital medication for transplant. It was also necessary to file a request at the Ministry of Health showing that FA meets the criteria to be considered a rare disease (this recognition, finally obtained in May 2018, was crucial). We try to help with these issues, but the most important is yet to be accomplished: a truly functional FA association in Brazil.

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Camp Sunshine porch gathering

Ask the Adults: Advice for the Younger FA Generation



One of the best tools to help cope with the Fanconi anemia diagnosis is talking to and leaning on others who also live with FA. The private FA Family Support Group on Facebook is a great place to ask questions and get feedback from other parents or adults with FA (learn about joining by emailing info@fanconi.org).

Thank you, Jack Timplerley, for organizing this advice column.

Several questions are directed at the growing adult population, so one adult with FA took on the task of sharing questions from parents and teens with the other adults. Here are some of their responses.

Our son is significantly smaller than his same-age peers. We know that this will become more of an issue for him socially as he gets older. Any advice?

The more you accept yourself, the more others will accept you. We smaller folk stand out, but maybe that's a good thing! Help him see all the benefits there are to his situation. Other kids may not get it, but they don't have to. Use standing out as an advantage! Get involved in activities, live your own life, and you'll make true friends along the way!

Where do smaller FA adult guys shop for clothes? Particularly pants with very small waists?

I've found H&M to work for me when it comes to pants. Other than that, I wear track/wind pants all the time.

How do we go about telling our child with FA about his condition? As far as we're concerned, we have a perfectly normal, wonderful child who just happens to require a fair amount of medical care.

You need to do what you think is best for your family dynamic. No one knows your child better than you. You'll do a great job either way.

Are there others like me (with FA) who are all grown up? What do they do for work?

I'm not just grown up now, I'm also close to being 33 years post bone marrow transplant. I have had some issues along the way, but I have a great life. I have lived out a lot of my dreams and now I have settled down with my wife and my dog. I would never pretend it's easy to live with FA. But instead of letting it be a hindrance in life, you can use it as fuel to getting things done and enjoy life much more. Right now I work full-time at a nursery. Also, I'm trying to build a

small business with a friend of mine where we arrange small eSport/game tournaments and leagues both online and in different locations in Copenhagen. But I have also worked with many other things, from customer service and sales jobs, to being a shop manager and working for years as a game journalist.

When you start dating (or really any new relationship, when is an appropriate time to discuss "I have a rare and life threatening genetic disease"? How do you work that into a conversation?

I didn't tell her about it all at once. It was definitely gradual. When she offered me drinks in the beginning I would say no because I just don't like alcohol. But eventually I sat her down and explained to her why alcohol is dangerous for me and that's why I don't drink. I didn't do this when she first offered me a drink because that's really catching her off guard. I waited until we were having a deeper conversation. If you think your partner is ready, I would take him/her to Camp Sunshine or the Adult Meeting. If the time is not right to go together, share with him/her what the meeting is about and why it is important to you.

As a child transitions into young adulthood, how much of your medical care do you want to handle completely independently? Would you prefer that a parent continues medical management and handles the appointment making, follow ups, and in depth understanding of FA so you can focus on school, relationships and beginning a career?

I would prefer to handle my own medical life. My parents and doctors have all helped me understand the science of FA and the potential implications in the future. I am a very independent person so I pride myself with my ability to take care of myself and run my own life. It's all about finding that balance. My parents are always there when I need them though.

Do you have a question you'd like to ask the adult FA community? Send it in by emailing info@fanconi.org. ■

Caregiver Position Description

by Allison Breining, caregiver, mother to Maya, wife to Sean (adult living with Fanconi anemia)



POSITION SUMMARY:

The Caregiver cares for the patient's medical, physical, emotional, spiritual, mental, social-emotional, financial, organizational, and dietary needs.

REPORTING TO: This position reports to the patient, medical staff, the patient's mother, other family members, friends, acquaintances, and the Lord.

PREFERRED DEGREES/

CERTIFICATIONS include, but are not limited to:

- Licensed Social Worker
- Psychiatrist
- Psychologist
- Chaplain
- Healing Touch Practitioner
- Essential Oil Consultant
- Geneticist
- Urologist
- Oncologist
- Dermatologist
- Hematologist
- Orthopedist
- Ear/Nose/Throat Specialist
- Cardiologist
- Radiologist
- Pulmonologist
- Anesthesiologist
- Ophthalmologist
- Pharmacist
- Nurse
- EMT
- Surgeon
- Dietician
- Physical Therapist
- Personal Trainer
- Wound Care Specialist
- Insurance Specialist
- Life Coach
- Researcher
- Professional Organizer
- Fundraiser
- Zen Master
- Public Relations Guru

**Please note: the ideal candidate for this position would have not one, but all of the above certifications.*

GENERAL QUALIFICATIONS:

- Strong capacity to be flexible and adaptable to varied and constantly changing circumstances, paired with a conscientious commitment to keep the patient alive.
- Exceptional interpersonal skills for effectively communicating and problem-solving with medical staff, family and friends, insurance collectors, nosy neighbors, and well-meaning helpers.
- Extraordinary capacity to hold emotions in check throughout difficult procedures, hospital stays, and awkward interactions.
- Proven ability to successfully manage all tasks with little to no preparation, training, sleep, or nourishment.
- Technologically literate with strong computer skills, including comfortable use of email, WebMD, MyChart, multiple bill pay sites, blogs, social media platforms, etc.

SCHEDULE: This is a 24 hour/day, 7 days/week position.

HOLDING CONCURRENT POSITIONS:

It is expected that those who hold this position will concurrently hold other positions, many of which may be full time, such as Parent and/or Breadwinner.

VACATION:

This position allows for two types of vacations:

1. *You may go on vacation with the patient.* In this case you will continue to perform all of the duties listed, including additional duties related to the trip, which include, but are not limited to: renting an electronic scooter, standing in line for a disability pass, hunting down a wheelchair, sharing notes from doctors when questioned by security about liquid formula in a carry-on, carrying and distributing hand sanitizer at all times, making real-time decisions regarding when it would be in the patient's best interest to cut the vacation short.
2. *You may go on vacation without the patient.* In this case you must set up back up care for the entire time you are gone. To do so, find someone with the above-listed qualifications, with whom the patient is comfortable, who is available and willing to perform these tasks at this level of compensation. *Please note: while gone you must be reachable by phone at all times so that you can be notified when the requisite disaster occurs.*

COMPENSATION: There is no monetary compensation for this position. You will, however, receive:

- the joy and satisfaction of knowing you are helping your loved one
- an honorary medical degree (that no one acknowledges)
- pitying looks and awkward questions from co-workers and acquaintances
- endless cups of free coffee, Lipton tea, and stale graham crackers

ADDITIONAL INFORMATION:

This position is on an ongoing contract, renewed automatically each year, until the time of the patient's death. At that point, please see position description for Widow/er.

Serious illnesses do not discriminate on the basis of race, color, creed, religion, national origin, sex, marital status, status with regard to public assistance, familial status, disability, sexual orientation, or age.

ESSENTIAL DUTIES:

1. **Be or become an expert on the patient's illness.** This includes, but is not limited to: listening to, taking notes on, and comprehending all information given during appointments with specialists, asking insightful follow-up questions, and doing independent research on the topic, including weeding out what is an ad, a scam, inaccurate, not applicable, or outdated.
2. **Stay up to date on breaking new research on the illness and/or any other illness that may, in some way, relate to this illness.**
3. **Graciously accept and assess the validity of:** stories (i.e., anything that begins with, "I once knew someone..."), links to buy probiotics in bulk, jugs of water purified to have the perfect pH level, leaves from the Holy Land, names of better doctors, documentaries on Veganism, articles on the benefits of walking barefoot in the grass, TED talks on the power of positivity, names of acupuncturists, marijuana in various forms, treadmills, yoga practices, newspaper clippings of someone else who was once sick and got better, to name a few. With each submission, go through the following steps:
 - weigh its validity vs. the treatment plans of the specialists the patient is currently seeing
 - wonder if the doctors have narrow-mindedly gotten it all wrong and have brainwashed you and the patient
 - consider dumping the doctors and "going East"
 - give up sugar for an hour to help clear your mind and your toxins as you think it over
 - decide to stick with the specialists
 - eat a sundae from Culver's
4. **Assist patient with daily needs.** This may include, but is not limited to: bathing, dressing, feeding, wound changes, laundry, shopping, cooking, cleaning, and dispensing medications.
5. **Attend doctor's appointments and medical procedures:**
 - Get the patient to the correct doctor at the correct time with the correct paperwork and correct amount of food, drink, or medications in or out of the patient's system.
 - Bring/provide snacks, beverages, etc. for the duration of the appointment, both for the patient and yourself. Keep in mind the patient's current dietary restrictions and preferences and that most appointments will last twice as long as expected.
 - Bring any items the patient may need before, during, or after the procedure, i.e., earbuds, phone chargers, medications, insurance cards, etc.
 - Watch every member of the medical team to check for accuracy, precision, and sterile practices.
 - Speak up when necessary, take notes, and ask questions.
 - Fill in details that patient may have left out.
 - Record phone numbers for after-hours emergencies. *Please note: there is no need to write down phone numbers for daytime emergencies, as these never occur.*
 - Set up next appointments.
6. **Assess unexpected symptoms, such as fevers, rashes, pain, confusion, etc., to determine what steps need to be taken, i.e. ambulance, ER visit, call to clinic, etc.**
7. **Create and maintain impeccable records of appointments, insurance, disability paperwork, contact information for medical team, medication inserts listing potential side effects, receipts potentially usable for tax purposes, medical research, etc.**
8. **Order, organize, dispense, and fight with insurance companies over medical supplies and prescriptions.** This task occurs at least monthly.
9. **Build strong knowledge of and relationships with the health insurance field.** You will have daily contact with billing companies, medical supply companies, bill collectors, etc. It is important to research each piece of mail received, as most of them are faulty, but not obviously so. In this role you may decide, after doing a thorough cost-benefit analysis, that it is easier to pay the bill, however faulty, than to continue to fight the system.
10. **Know when and how often to ask for help with meals, groceries, play dates for children in the home, etc.** Ask for help only so often so as not to burn out friends and family. Ask carefully, sounding desperate enough to not be able to fetch your own groceries, but not so desperate that if you answer the door with a shirt that is clean the gift giver will be confused as to why they are there. Make requests specific so that you don't end up with 14 chicken casseroles or play dates for your child with strangers. Keep in mind that many of these tasks will come with interaction with well-meaning humans who often don't know what to say. Before asking, weigh the benefit of the help against the time spent with humans.
11. **Update family and friends on latest information, providing them with enough information so they don't worry, but not so many details that they have something to worry about.**
12. **Actively follow social media pages related to the illness.** This will help maintain a constant level of panic and despair as you watch others with the same diagnosis struggle in ways you didn't realize were possible.
13. **Fundraise for a cure.** Organize bake sales, send holiday letters, and create inspirational videos to raise money, not for your own struggling family, but so that researchers can work to find a cure for this disease. It may seem as if you already have a full plate, but you need to do this if you want your patient to survive.
14. **Join the hospital advisory board and attend monthly meetings.** This is an important part of this role as it gives you practice driving to and from the hospital as well as a monthly opportunity to air the one or two grievances you may have encountered in your time as a caregiver.



Read more of Allison's work about being a caregiver, mom, wife, coach, and writer at www.thenegativespace.life.

FAMILY FUNDRAISING EFFORTS

From January to August 2018, FA families raised an impressive \$741,285 for the Fanconi Anemia Research Fund! 182 families raised funds, with 85 raising at least \$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.

\$140,000 +

Lynn Frohnmayer

\$98,000 +

Kevin and Lorraine McQueen

\$68,000 +

Orion and Lisa Marx

\$20,000 – \$31,000

Mark Frohnmayer
Steve and Jennifer

Klimkiewicz
Tim and Mary Ann Lana
Todd and Kristin Levine
Gerard and Cynthia
Vandermeys
Alex Winn

\$15,000 – \$19,999

Matthew and Evelyn Keyes

\$10,000 – \$14,999

Mauro and Kerrie Cazzari
Chuck and Katy Hull
Peg Padden
Peter and Janice Pless
Neil and Emily Robison

\$5,000 – \$9,999

Jimmy and Jenny Armentrout
David and Sarah Borden
James and Crystal Eubank
Andre Hessels and Rutger Boerema
Ian and Tricia Mitchell
Tyler Morrison and Rachel Altmann
Leah Petsanas
Bill and Connie Schenone
Nigel and Ann Walker

\$1,000 – \$4,999

Brian and Carly Adel

Michael and Jennifer

Aggabao
Assila Al-Marshoudi
Mark and Linda Baumiller
Israel and Mary Jo Becerra
Adam and Marissa Becker
Jeff and Donna Boggs
Rick and Tena Boson
Chris and Jennifer Branov
Sean and Allison Breininger
Ryan and Becky Brinkmann
David and Kim Chew
Jerry and Natalie Christensen
Andrew Coons and Valeen Gonzales
Marie Di Mercurio
Justin and Britteny Ferrin
David and Mary Ann Fiaschetti
Allen Goldberg
Andrew and Jennifer Gough
Gary and Heidi Grassi
Madeline and Patrick Gregg
Ben and Stephanie Griggs
Alan and Rachel Grossman
Owen Hall and Margaret Kasting
John and Martina Hartmann
Brian Horrigan and Amy Levine
Robert and Anna Langtry
Dan and Nikki McCarthy
Jack and Lisa Nash
The Pearl Family
George and Kathy Reardon
Eliot and Parker Rushing
Bob and Andrea Sacks
Erik and Lori Salo
Richard and Dolores Satterlee
Bryan and Karen Siebenthal
Ana Alejandra Tabar Concha
and Elvin Estevez Lopez
Bill and Mary Underriner
Joseph and Natalie Vitrano

Up to \$999

Peter and Donna Abramov
Marzban and Daisy Ardeshir
Juanita and Ron Arroyo
Andrew and Vicki Athens
Jeanne Atkinson
Charles Balow and Xandra Towndrow
Conrad and Joan Bender
John and Francene Berglund
Tracy Biby
Katie Bowe
Donny and Danielle Burkin
The Family of Chris Byrd
Hugo Canalli
John and Launa Clanton
Yvette Coats
John and Kim Connelly
Jeremy and Michelle DellaValle
James and Carol Dillon
Sharon Ellis
Chloe Eminger
Daryn and Carol Franzen
Liz Funk
Laurie Gerhardt
Stan Gilbert
Jeff and Beth Janock
Nancy Jansen
Ben and Mary-Beth Johnson
Randy Jones
Stan and Michelle Kalembe
John and Karilyn Kelson
Tanner and Jessica Lindsay
Keith and Jessica Loo
Col. Gregory and Lt. Col. Lynnette Lowrimore
Bill and Jackie Lucarell
Kory and Julie MacMurray
Paul and Leighsa Makowicz
Deane Marchbein and Stuart Cohen
Aaron and Nicki Marsters
Catherine McKeon

Danny and Angie McMahon
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Alaina Mercer
Kim Mootassem
Tony and Lina Nahas
David and Catherine Neumann
Robert and Mary Nori
Ron and Fredi Norris
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David and Stacy Ownby
Jeff and Arianne Pederson
Michael and Joanna Peros
Ashleigh Pinion
Mike and Kay Proctor
Lynn and Shirley Quilici
Pedro and Marina Ravelo
Mark Ritchie and Lisa Mingo
Kevin and Katie Rogers
Les and Nancy Ross
Rick and Lynn Sablosky
Jennifer and Brian Sadlowe
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Colleen Scholl
Lorne Shelson and Annette Waxberg
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Greg and Brandi Stuart
Charles and Jennifer Sumrall
Sharon Swanson
Ken and Jan Sysak
Mark and Susan Trager
Tom and Kathy Uno
Mike and Beth Vangel
Janet Voorheijen
Marc Weiner
Sandy Weiner
The Family of Clay Williams
Werner and Laetitia Wolfswinkel
Chad and Dawn Wood
Jason and Joan Woodle
Kyle and Madison Wright

A note to our fundraisers: we greatly appreciate your efforts to raise money for FARE, 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. ■



By Ashley Chorneyko

In October of 2015, my husband Colin and I gained a new FAmily. It wasn't the FAmily we were looking for, or ever would have imagined we would be a part of, but this FAmily was inevitable when our son Vonn was diagnosed with Fanconi anemia. When we joined the Fanconi anemia support group, it was apparent that we would really need to pull up our socks and figure out a way to help raise funds in order to further FA research and therapies, and to eventually find a cure for this terrifying disease. Since FA is so rare and most research money comes from the hard work of FA families and their individual fundraising efforts, we wanted to pay it forward and ensure that future FAmilies were given more hope at diagnosis than we were.

Bake sales, steak nights and marathons all seemed like great fundraising ideas, but something inside of me was telling me to dream bigger. I imagined an adult's evening out: a gala, where friends and family could dress up, have a great time, and feel good about helping raise desperately needed funds. In January of 2016 the journey began with a group of 13 hard working women, who formed the FAv Gala Committee. Together we hosted our first annual FAv Gala, seven months after Vonn's diagnosis.

It was remarkable! We were overwhelmed with the love and support shown to our family. The outpouring of love given to one small boy in our small Regina, Saskatchewan community was unimaginable! Throughout the journey we found another new family – our FAv gala supporters. These are people in our community whom we will never take for granted, people who felt compelled to help and continue to

cheer on Vonn and donate to the gala every year. The first year, our gala raised over \$120,000 CAD, then in year two, we raised \$135,000, and this past year \$129,000 was raised for Fanconi anemia research!

It's amazing how generous people are, especially for young children. People are genuinely good and care, but if you don't ask... you don't get!

Putting on this gala has given us a pretty good template to hold a similar fundraiser:

1. Rally a group of the amazing people around you, who are probably more than willing to help your family's cause.
2. Write a compelling letter from the heart, explaining the importance and desperation of raising funds.
3. Find a great venue with wonderful food.
4. Create a pamphlet with sponsorship levels and ticket prices.
5. Create a social media page, where caring people can follow.
6. Brainstorm a thorough mailing list.
7. Choose a theme for your event and try to make it as fun as possible.
8. Collect as many silent auction items as possible. We usually have over 200 items for our 500 guests to bid on. The guests love the shopping experience and can feel good about their purchases.
9. Create a genuine video about Fanconi anemia for the night of the gala and surprise the guests with a special appearance by your FA fighter.
10. Always say thank you, thank you, thank you!

Vonn is one of the most empathetic people around and continues to feel thankful that he is surrounded by people who are constantly rooting for him and concerned about how he is doing. Through it all Vonn remains optimistic and has an unwavering attitude about living life to the fullest, playing his hardest and always making new friends along the way. He has said things like, "we need to help other kids like me with sick blood." Heartbreaking, but inspiring. Along with raising funds, Colin and I are proud to be teaching our son to rise above, do good and feel great about making a difference! ■

FUNDRAISING SCRAPBOOK



More Birthdays for Bella

On April 28th, in honor of FA Day and Bella's 14th birthday, Bella Griggs and her friend Aspen hosted a community garage sale at Bella's home in Castle Rock, Colorado. They exceeded their goal of \$1,400! Thank you, Bella!



Eli's Battle Team

Led by Eli's mom Sarah Borden, Eli's Battle Team raised an incredible \$5,799 as part of the FA Day campaign. Sarah's colleagues all made donations to vote on the executives from their company who would receive a pie in the face from Eli. Thank you, eFaucets and Borden family!



Ryan's Hope – Our Littlest Love

The Adel family kicked off their very first FA fundraiser as part of the #ThisIsHowIFA campaign for FA Day this past May. Ryan's Hope raised over \$2,500, showing that you don't have to be an experienced fundraiser to make a difference. Check out Ryan's Hope on Facebook or at <http://fundraise.fanconi.org/RyanAdel>.



Mitchell Family Fundraisers

The Mitchell family has been busy this year! In addition to a bake sale and walk hosted by Emily's school in May, they were also the top team for the Lana Family's 5K for FA, bringing in over \$5,000. Earlier this year, Tricia Mitchell held a Facebook birthday fundraiser, and in September hosted a Casino Night in New York City. Thank you for all of your efforts, Mitchell Family.



Olivia's Fight to Cure FA

The Eubank family exceeded their goal of raising \$6,000 through their Facebook fundraiser in honor of Olivia Eubank and FA Day. They didn't stop there, however—in October the Eubanks held their first Vineyard Party fundraiser. Thank you!



5K for FA

This year, the Lana family raised over \$28,000 (even more than last year!) through their annual 5K for FA, which was held on May 5th in Hilton, New York. Coming up on May 4, 2019 is their 5th annual event. Visit www.5kforfa.com to join in the fun.



Team BrAvery

How many of you like a challenge? Team BrAvery is always looking for its next extreme challenge. On May 19 – 20, Orion Marx and his father-in-law Charlie Scott ran an impressive 100 miles in the Florida Keys 100 Ultra-Marathon, raising over \$68,000. Way to go, Team BrAvery!



Your Rope Team

Bill McCorey, FARF board member and long-time friend of the McQueen family, climbed Mt. Cotopaxi in Ecuador this past June to raise funds for FA research. The Rope Team faced extreme weather conditions, but never lost sight of the people they were climbing for: FA fighters. Not only did they scale the mountain, they raised more than \$22,000! Incredible!

Dear Families, we need YOU this holiday season

Dear Families,

Hi, my name is McKenna. I joined the Fanconi Anemia Research Fund in April as Philanthropy Director. I have spent my first several months getting to know our community through Camp Sunshine, the Meeting for Adults with FA & Scientific Symposium, and family fundraisers.

I still have a lot to learn about our organization, but this much I know to be true: this community DEEPLY cares. Patients, families, friends, donors, board members—we all have vested interests in fulfilling FARF's mission and have come together to accomplish incredible things.

Whether you are new or seasoned in your journey with FARF, our cumulative impact is heightened when we work together towards a focused goal. This season, our focus is holiday appeals. Did you know that more than 2/3 of FARF's annual budget is generated in November, December, and January? **Last year, in response to family holiday letters, 2,090 gifts were made to the Fanconi Anemia Research Fund.** Each gift, of every size, has a huge impact in what we accomplish as an organization.

70% of FARF's income is generated by holiday gifts

\$60 is the median gift size in 2018 so far

16 FA families did holiday appeals in 2017. How many will join this year?

Here's how you can get involved:

How-To Guide: Family Fundraising Made Easy!

1. Visit <http://fundraise.fanconi.org/2018holidays> and click "become a fundraiser". This will create a custom fundraising page for you to share in your community.
2. Add your photo and a message you want to share about your family. We've already added the impact of fundraising dollars for your donors to see.
3. Share your page in the community via email, Facebook, and other social media outlets.

If you have any questions or ideas about your holiday fundraising efforts, please reach out to us at: 541-687-4658 or info@fanconi.org. We are here to help you every step of the way, from building your fundraising page, to customizing your message, and sharing your holiday appeal in the community.

We simply couldn't do what we do without the power of family fundraising. From all of us at FARF, thank you for your effort, time, and support as we come together and fundraise for this worthy cause.

Happy holidays!

McKenna Knapp

Philanthropy Director





Team FARF in the Oregon countryside this fall

By Mark Quinlan, Executive Director

As I reflect on 2018, it is easy to remember all of the positives: two FA clinical trials opened, another successful annual family meeting at Camp Sunshine, our Meeting for Adults with FA and the 30th Scientific Symposium in Newport Beach, CA. We also filled key positions in our research and fundraising departments. Along with these accomplishments, we funded new research and have made strides to ensure our status as a premiere rare disease support organization. It has been an honor to work with such a committed team, board of directors, scientific advisory board, and of course, our amazing FA families.

Every day I am reminded why we need to remain acutely focused on our mission. This year, some of the first people I met after taking this position received a cancer diagnosis. I knew this would happen when I joined FARF, but I could not have prepared for how it affects me each time one of our young adults receives this devastating diagnosis.

There are many research projects in progress throughout the year, all of them of great importance, and yet I never leave a meeting, presentation, conversation, or read a publication where the issue of cancer in FA does not come up. We all know that individuals with FA are at increased risk for developing acute myeloid leukemia (AML) and solid tumors which include head and neck squamous cell carcinoma (HNSCC) and anogenital squamous cell carcinoma. While the reasons for increased risk are not yet well understood, that does not deter us from aggressively addressing this issue.

In 2018, FA-related cancers were a main focus of our research priorities. FARF provided funding to two clinical trials, one

focused on metformin, an FDA-approved drug that has shown anti-cancer effects in the general population, and quercetin, a naturally occurring compound in food with potential anti-cancer properties. In April, we convened experts in our first ever Joel Walker Scientific Meeting Series to discuss HNSCC. In September we dedicated an entire day at our Scientific Symposium to cancer in FA. These meetings are important because they bring physicians and researchers together to figure out the best directions for advancing treatments.

Moving forward in 2019, a few priorities stand out: we will explore efforts to expand early detection approaches, improve tissue collection protocols to make sure this valuable resource is available for research, explore how immunotherapies may help fight FA cancers, and promote collaboration between FA scientists and clinicians to accelerate the potential therapies available to FA patients.

The challenges of preventing, detecting, and treating cancer in FA patients are real and they are daunting. FARF accepts these challenges and is driven to address the problems and identify solutions. We will work every day to improve and expand the lives of those living with FA, but we can't do it alone. We need your help to raise the funds required to address these challenges. You can get involved by making a holiday appeal to share with your community. Go to <http://fundraise.fanconi.org/2018holidays> or call our office at 541.687.4658 to get started. Thank you for your participation.

Sincerely,

Mark Quinlan

FARF GRANTS AWARDED FEBRUARY – OCTOBER, 2018

From February 2018 to October 2018, the Fanconi Anemia Research Fund awarded **\$1,078,668** to the following projects:

Investigators: Peter Glazer, MD, PhD; Gary Kupfer, MD
Yale University

Title: Use of triplex-forming PNAs as a strategy for correction of the FA phenotype (continuation)

Amount Funded: \$125,000

Investigators: Eunike Velleuer, MD; Ralf Dietrich
German Fanconi Anemia Support Group and Research Fund

Title: Reducing the Burden of Squamous Cell Carcinoma in Fanconi anemia

Amount Funded: \$182,081

Investigator: Agata Smogorzewska, MD, PhD
The Rockefeller University, New York City, New York

Title: Identification of Novel Therapeutic Targets against Fanconi Anemia-associated Squamous Cell Carcinoma

Amount Funded: \$120,000

Investigator: Raymond Monnat Jr., MD
University of Washington, Seattle, Washington

Title: The Fanconi Anemia Cancer Translational Resource

Amount Funded: \$174,987

Investigators: Kevin G. Haworth, PhD and Hans-Peter Kiem, MD
Fred Hutchinson Cancer Research Center, Seattle, Washington

Title: Direct in vivo gene correction of hematopoietic stem cell populations in Fanconi anemia

Amount Funded: \$175,000

Investigator: Agnieszka Czechowicz, MD, PhD
Stanford University, Stanford, California

Title: Development of a safe, completely non-genotoxic anti-Kit antibody-based conditioning regimen for hematopoietic stem cell transplantation in Fanconi Anemia

Amount Funded: \$180,000

Investigator: Jacob E. Corn, PhD
University of California Berkeley

Title: Defining tractable approaches for gene editing of Fanconi Anemia hematopoietic stem cells (continuation)

Amount Funded: \$121,600

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 **229 research grants**, two clinical trials, and one service grant to **152 investigators** at **74 institutions** worldwide. The total amount of research dollars awarded is over **\$23 million!**

No Longer an Orphan

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of conditions that touches us all. Not only that: the cutting-edge discoveries about DNA repair, bone marrow transplant protocols, immunotherapy, gene therapy, and aging, give Fanconi anemia a family so large that it embraces us all. It's still a fatal disease, but the urgency we feel to find a cure is an urgency not felt in isolation, as it was for the Frohnmayers in 1983. It is an urgency that lies at the heart of our shared mortality.

Sharon Schuman, PhD, is Secretary of the Board of Directors of the Fanconi Anemia Research Fund. She was Amy Frohnmayer's violin teacher from age 9 to 15. Sharon holds a benefit concert for FARF every year which has raised more than \$330,000 over 25 years. This presentation was first delivered to the Round Table town-gown organization in Eugene, Oregon, in February, 2018. See full article at www.fanconi.org ■

Optimizing Success in School

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plans, other support is available. Many resources exist on the local and state level. Two such resources provided by Theile are www.parentcenterhub.com and www.understood.org. Theile also provided excellent resources for school re-entry and accommodation recommendations for chronic illnesses (www.fanconi.org). With a comprehensive and well-coordinated plan, schools, parents, healthcare providers and even the community can help provide a supportive educational environment for those with FA to succeed in school and optimize their academic, behavioral and psychosocial development. ■

Let's Meet at the Porch?

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Apart from that, we have no words to express our deep gratitude to Dr. Carmem Bonfim, Dr. Lisandro Ribeiro and the great team that specializes in FA in Brazil. They get truly involved in absolutely everything, from lawsuits to obtaining medicine, to organizing FA meetings and collecting donations. Our FA association must reduce this burden affecting doctors and specialists, despite our challenges. We still have to carry on our full-time jobs and deal with all of the treatments and emotional distress related to FA. For the vast majority of our Families, it would be very difficult to get involved, and those willing to participate usually live thousands of kilometers away from each other.

Sometimes, we just do not know what to do regarding treatments and the emerging association. Maybe the most important aspect is to avoid getting discouraged and to do our best. That's one more reason why FA family meetings are so tremendously important. Against gloom or weariness, our gatherings at the porch while at Camp were a nice deep breath of fresh air. Thank you to all of you! ■

Navigating Financial Resources

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navigate these services. To locate your state's Families-to-Families Health Information Center, visit www.familyvoices.org and click on your state on the homepage.

**This is a shortened version of this article. To access the full text, please visit www.fanconi.org and look under "Patients and Families". ■*

In Loving Memory

Josh Baumiller
7.5.82 – 4.22.18

Robert Clayton "Clay"
Laverne Williams
9.3.93 – 4.26.18

Ashley Kay Crow
8.30.87 – 5.4.18

Anna Maria Dahm
7.1.90 – 6.18.18

Nicola Wooding
12.11.90 – 6.30.18

Elizabeth Walker
8.29.75 – 7.14.18

Adeline Marsters
9.2.10 – 8.29.18

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!

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HAVE YOU BEEN TO THE NEW FANCONI.ORG?

We're excited to share our brand new website, featuring a fresh design, easier and better options to give, a news blog, directories of funded research and supported researchers, and more. Check it out today by visiting www.fanconi.org. ■



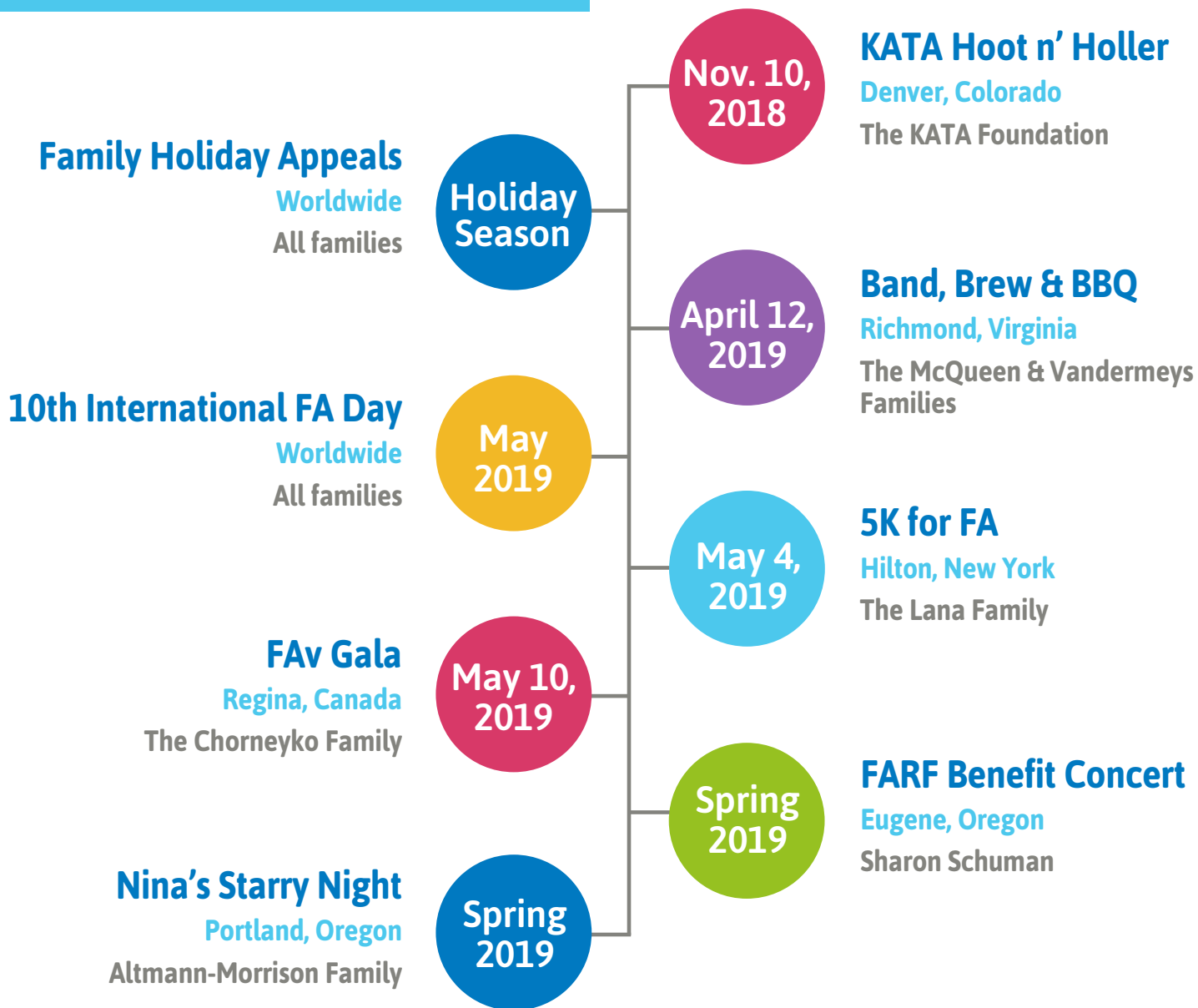
FAMILY DIRECTORY NOW AVAILABLE ONLINE

The new www.fanconi.org features an online family directory in which individuals with FA and families can add and edit their own entries. The online directory allows people in it to see all other individuals/families who have signed up. People can update their info in real time and quickly look online for another family's info when needed. We will still have a print version available. However, the online version will be more up to date since people can update their entries whenever there are changes. Everyone will need to create an entry, even if the paper form has previously been filled out. Signing up only takes a few minutes. Go to www.fanconi.org to add your entry! ■



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



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.

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Donations Online:

Donate via the Fund's website (www.fanconi.org) or through
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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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