



FAMILY Newsletter

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issue
61

Welcome, Mark Quinlan

New Executive Director of the FA Research Fund

A Note from the Board President

Dear FAMILIES, scientific community, volunteers, friends and donors,

Please allow me to introduce our new Executive Director, Mark Quinlan. We are excited to bring Mark on board to lead the Fund into the future. Mark comes to FARF with years of nonprofit and Executive Director experience. Most currently, he served as Program Director at the University of Oregon School of Law. This is following six years as the Executive Director for Habitat for Humanity in Bend, Ore., and 11 years as a Program Manager for the High Desert Education Service District in Redmond, Ore. He earned a Bachelor of Science degree in Social and Behavioral Science and a Master in Public Administration. He participated in the Strategic Perspectives of Nonprofit Management executive learning program at Harvard Business School in 2012 and last year, he completed the Foundations of Collaborative Governance program at Portland State University. Most recently, he participated in the University of Oregon's Financial Stewardship Institute course.

Please join me and the rest of the Board of Directors in welcoming Mark to the FA community. We all look forward to his contributions and insights as FARF continues to advance its mission.

Kevin McQueen

Board of Directors, President



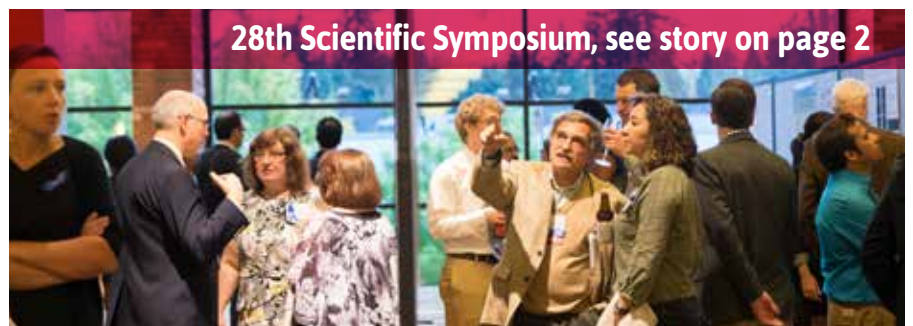
I am thrilled to join this dedicated group as the Fund's Executive Director. I am humbled and honored by the opportunity to carry on the

incredible work that you have all achieved over the last 28 years. What attracted me to the organization was its focused approach to identifying high-quality and meaningful research, and the way FARF puts donor funds to work where they have the greatest impact on families affected by Fanconi anemia. I look forward to building new partnerships, developing innovative programs, and fostering positive relationships within the FA community to advance the Fund's mission.

I am excited about what the future holds for the FA community. Together, we are going to **build** on the incredible successes achieved over the years and **expand** our impact. We are going to **empower** researchers and doctors to continue to make game-changing breakthroughs. We are going to **grow** our support services to families affected by FA worldwide. As a united community, we will work to **achieve** our mission of finding better treatments and a cure for FA.

I look forward to meeting you at Family Camp, the Scientific Symposium, or one of the many great fundraisers for our cause! In the meantime, feel free to reach out to me any time at 541.687.4658 or mark@fanconi.org.

Thanks for your support and for *all* you do,
Mark Quinlan



28th Scientific Symposium, see story on page 2

Attendees discuss research presented during the poster reception



Carmem Bonfim, MD, PhD accepts the award for Distinguished Service



Emily Robison, FA parent, captures the audience with her family's journey with FA



Jack Dempsey performs his song, "He Makes Me Happy", inspired by Cale Ferrin

28th Scientific Symposium Inspires and Motivates

Last September, nearly 200 researchers, doctors, families affected by Fanconi anemia, FARF staff and board members came together for four days of collaboration at the 28th Annual Scientific Symposium in Bellevue, Wash. Attendees were greeted by bright, colorful banners featuring faces of children and adults with Fanconi anemia. This was part of the launch of FARF's brand redesign, including a new logo and color scheme, which helped set a tone of renewed energy and vibrancy at the conference.

Over the course of the weekend, attendees heard 43 presentations on a range of topics, including: Novel Mechanisms & New Approaches, Aldehydes, Genotoxicity & Disease (p. 6), Oral Cancer – Treatment, Risk Factors & Early Detection, The FA Pathway (p. 8), Transplantation & Disease Challenges (p. 12), Preclinical Models & Drug Development, and New Genes & Gene Therapy (p. 9). Four keynote talks engaged the audience and 56 poster presentations gave way to discussion, debate and networking.

This is such an exciting time for FA research because of the influx of young, bright and motivated investigators in the field. Over the past few years, FARF has organized

a mentorship lunch during the Symposium. This is an opportunity for students and young investigators to spend time with experts in various fields of FA research and clinical care. Going forward, FARF plans to cultivate this interaction and provide more opportunities for mentorship to this new population of FA researchers and clinicians.

The 2016 Symposium saw record attendance by FA family members. In total, 23 FA parents and one adult with FA attended the conference, in addition to two young boys with FA. Representatives from international FA family support organizations were also present, including those from Canada, Germany, the Netherlands, and Italy. Emily and Neil Robison, parents of 4-year-old Blake, who has FA, shared their emotional journey at the Symposium banquet dinner, bringing the inspired audience to their feet in applause. FARF Philanthropy Council members and FA parents Lorraine McQueen and Mary Ann Lana spoke about their commitment to raising funds to support FA research and implored researchers and doctors in the audience to make that same commitment. Another highlight of the banquet was a live performance by Hollywood-based musician Jack Dempsey.



190

participants



17

countries represented



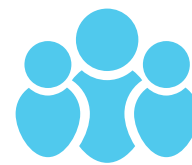
43

oral presentations



56

poster presentations



23

family members



9

staff members



14

board members



Discovery awards are presented to Dominique Bluteau, PhD, Jean Soulier, MD, PhD, and Alan D'Andrea, MD, for their work to identify FANCV



Ray Monnat, PhD, presents the Pioneer Award for Therapeutic Advancement to Grover Bagby, MD



Best poster abstract awards are presented to Wei Tong, PhD (best basic science), Lisandro Ribeiro (best clinical poster), and Agata Smogorzewska, PhD (best translational poster)

He shared the story of how he was overcome with inspiration after meeting 10-year-old Cale Ferrin, who has FA, prompting him to write the song "He Makes Me Happy" and even film a music video with Cale (see pg. 16 for more).

Several researchers and doctors were acknowledged at the banquet for their outstanding contributions in the field. First, those involved in the discovery of the 21st FA gene were acknowledged. The awards for best poster abstracts were presented by FARF Scientific Director Brad Preston, PhD. The second annual David B. Frohnmayer Early Investigator Award was presented to Michael Milsom, PhD, of the Heidelberg Institute for Stem Cell Research and Regenerative Medicine in Germany. Two awards for Distinguished Service were presented to Carmem M. Bonfim, MD, PhD, and Ricardo Pasquini, MD, both from the Federal University of Paraná in Brazil, for their remarkable care of people with FA throughout Brazil. Finally, Grover Bagby, MD, of Oregon Health & Science University and Knight Cancer Institute was presented with the Pioneer Award for Therapeutic

Advancement for his tireless efforts to develop targeted therapies for FA leukemia patients.

Throughout the conference, the energy and excitement around new research and possible new treatments was strongly felt, yet a distinct and heavy feeling of sadness also persisted. The FA community knows too well the weight of tragedy and 2016 was no exception, as Dr. Ken Atkinson (co-founder of the Kendall and Taylor Atkinson Foundation) and Chris Byrd, Esq. (FARF Board of Directors and adult with FA) had recently passed away. Attendees wore blue awareness ribbons in their memory and observed a moment of silence at the banquet dinner. Another FARF board member and adult with FA, Amy (Frohnmayer) Winn, was notably absent, as she was near the end of her battle with leukemia. She died on October 2. As attendees reflected on the absence of these three pillars of the FA community, a deep appreciation of their impact sank in. Inspired, attendees left the conference with a renewed energy to honor Ken, Chris, and Amy's legacies by moving forward in this fight for a cure. ■

29th Annual
Fanconi Anemia
SCIENTIFIC SYMPOSIUM

Grand Hyatt Atlanta, Georgia | www.fanconi.org
September 14-17, 2017

CALL FOR ABSTRACTS
Submit abstracts 4/14 - 6/2

FANCONI ANEMIA RESEARCH FUND
Advancing research, supporting families.



Campaign to Improve Clinical Outcomes for Individuals with FA

The Fanconi Anemia Research Fund has supported several cancer-related projects over its 28 years, but 2016 marked a major expansion of the Fund's commitment to this area of research. Last March, FARF launched an effort dedicated to speeding up the pace of clinical trials, drug testing, gene therapies and more effective treatments for children and adults with Fanconi anemia. A major focus of this initiative is to prevent and cure the cancers that are now a primary cause of death in adults with FA.

The Council to Focus and Accelerate Research (CFAR) was created in March 2016 with the mission to improve clinical outcomes of individuals with FA and to do so as fast as possible. This dedicated panel of scientists and parents first critically assessed FARF's research priorities and then identified scientific projects with the highest promise of improving clinical results for people with FA. Prevention and treatment of cancer and bone marrow failure were identified as the highest priorities. In the last year, the Fund's

CFAR awarded grants to five projects, including the first stage of a clinical trial.

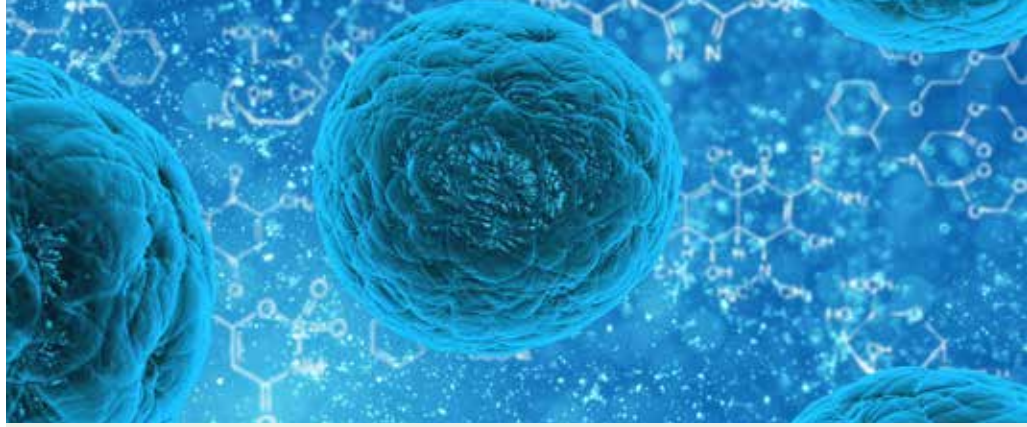
Dr. Eunike Velleuer of Heinrich-Heine University and Ralf Dietrich of the German FA Support Group are evaluating the feasibility of replacing traditional surgical biopsies with non-invasive brush biopsies to diagnose early oral cancer. In 2016, they examined the mouths of 286 people with FA and found four with cancer and three with pre-cancerous lesions. None of these cancers was detected by the patients' treating physicians.

Thus, the diagnosis of cancer would have been delayed, perhaps fatally, if Dr. Velleuer and Dietrich had not conducted their examinations as part of this funded project to reduce the burden of squamous cell carcinoma in Fanconi anemia.

Dr. Wei Tong's laboratory at the University of Pennsylvania studies the role of a protein called "Lnk" in regulating growth factor signaling and hematopoietic stem cell expansion. This newly funded project is based on Dr. Tong's novel finding that Lnk deficiency restores blood cell function to normal levels in FA mutant mice. Her research team will determine if suppressing Lnk can similarly improve blood cell function in FA patients. These studies started in October 2016 and will run for two years.

In addition, FARF is supporting two new, exciting gene therapy projects. These projects use cutting-edge technologies to replace FA genes with normal genes in a process called "gene editing". Dr. Jacob Corn and his team at the University of California-Berkeley recently pioneered a "CRISPR-Cas9" method to repair sickle cell genes; they will now harness this method to repair FA genes. Using another novel approach, Drs. Peter Glazer and Gary Kupfer at Yale University

“ The Council to Focus and Accelerate Research (CFAR) was created in March 2016 with the mission to improve clinical outcomes of individuals with FA and to do so as fast as possible. ”



will use “triplex PNAs” to repair FA genes. The Yale team will also explore the use of nanoparticles to deliver the repaired genes to FA patients by simple injection. Both projects will focus initially on repairing FA genes in the bone marrow. If successful, this will prevent bone marrow failure and leukemia.

Finally, for the first time ever, the Fund is sponsoring the initial stage of a clinical trial to prevent or delay bone marrow failure and leukemia in FA. The Phase I trial will be conducted under the direction of Dr. Akiko Shimamura at the Dana-Farber/Boston Children’s Cancer and Blood Disorders Center. This trial builds on extensive preclinical studies by Dr. Markus Grompe and his group at Oregon Health & Science University (OHSU) showing that metformin (a well-established drug for diabetes) improves hematopoiesis and reduces DNA damage in FA cells and mice. Dr. Grompe’s preclinical studies were supported by grants from FARF, and the administrative aspects of developing the Phase I trial will be paid by a National Institutes of Health (NIH) Fanconi Anemia Program Project Grant awarded to Dr. Grompe and his collaborators at the Dana-Farber Cancer Institute in Boston. Thus, new funding will leverage both FARF and NIH grant dollars. The Phase I trial will explore whether metformin is safe and effective in improving hematopoiesis in patients with Fanconi anemia. This will be the first clinical trial that repurposes an established, safe drug to treat FA. If successful, the impact on FA patients could be very high. ■

Expansion of Cord Blood Stem Cells to Speed Recovery

John E. Wagner, MD, University of Minnesota

Umbilical cord blood (UCB) is a proven source of blood-forming stem cells that has been used in more than 40,000 transplants worldwide. UCB, however, contains only a small number of stem cells. The pace of blood cell and immune recovery is slower than that seen with bone marrow (25 days vs. 17 days in patients with leukemia without Fanconi anemia).

Over the past 20 years, we and others have searched for ways to speed recovery regardless of donor source but especially UCB. Recently, a chemical called StemRegenin-1 (SR-1) has been shown to promote a dramatic (300-500 fold) expansion in the number of blood-forming stem cells. Over the past six years, three clinical trials have been completed at the University of Minnesota in patients with various blood cancers. Compared to identically treated patients transplanted with UCB without expansion, recipients of SR-1 expanded UCB recovered about two weeks faster, with recovery as early as five days in those receiving the highest cell doses. In addition, the donor cells engrafted in all 39 patients (100% as compared to 86% in those transplanted UCB without expansion). To date, one patient with FA was transplanted with SR-1 expanded UCB. After the infusion of 75 million (per kilogram body weight) SR-1 expanded UCB cells, recovery occurred on day 6 with early discharge from the hospital, as observed in prior patients.

As a result of these findings, we will open a study in patients with FA and other bone marrow failure syndromes with the goal of speeding neutrophil and platelet recovery in particular. The next step is to speed up recovery of the immune system by giving the patient thymic progenitors (the parent cells of T cells), grown from the SR-1 expanded UCB graft. ■

What could this mean for people with FA?

Some physicians believe that the long period of immune deficiency that occurs after transplant could be a significant culprit in the risk of cancer after transplant. A future study with thymic progenitor cells to enhance immune recovery is planned for late 2017.

Progress in Combating Aldehydes

K.J. Patel, MD, PhD, MRC Laboratory of Molecular Biology



One of the highlights of the 2016 Symposium was a keynote address by Dr. KJ Patel, a prominent Fanconi anemia researcher based in Cambridge, United Kingdom. He talked about a topic that has important consequences for all those with Fanconi anemia—aldehydes. His lab studies the toxic effects of **aldehydes, which are small organic molecules with reactive atoms that can bind to DNA and block cell replication**. Aldehydes are produced in two ways: by our own bodies, in the course of metabolism (endogenous); or by sources outside our bodies, like alcohol, cigarette smoke, and various

chemicals and foods (exogenous). These reactive aldehydes are a potent source of DNA damage.

Dr. Patel explained that all healthy mammals, including humans, are protected against these genotoxic metabolites through a two-tier system. The first defense is to eliminate aldehydes with oxidizing enzymes also produced by the body. If this Tier 1 defense fails to eliminate an aldehyde (which happens even in the healthiest people), a second tier of protection kicks in, repairing the DNA damage that aldehydes cause. In Fanconi anemia, this second-tier defense also fails.

The activities of aldehydes explain why people with FA accumulate DNA damage, causing developmental abnormalities, bone marrow failure, and an enormous lifetime risk of cancer. The current research at Dr. Patel's lab aims to uncover the origins of endogenous aldehydes, understand how cells remove them, explore the nature of the DNA damage they cause, and study how this damage is repaired. Scientists in Dr. Patel's lab also seek to understand how certain stem cells and organ systems are damaged by these aldehydes, as well as the nature of the consequences to those cells and systems. cont.



Juan Garaycochea



Guillermo Burgos-Barragan

FARF and the Patel Lab

Dr. Patel interrupted his presentation to thank the Fanconi Anemia Research Fund for saving his career on two different occasions when the Fund provided bridge funding for his lab. He specifically thanked Lynn and David Frohnmayer for their dignity, courage and farsightedness. Now Dr. Patel is an eminent scientist, and a new generation of young scientists in his lab, mentored by him and others, are giving presentations about aldehydes at FARF's scientific symposia. At this meeting, Dr. Juan Garaycochea spoke about how "Aldehydes Mutate the Genome of Blood Stem Cells," and Ph. D. student Guillermo Burgos-Barragan spoke about "Vitamin B-9 is a Source of Genotoxic Formaldehyde." Much remains to be done, but these scientists and others are bringing closer the day when people with FA will be able to solve the DNA repair problems at the heart of Fanconi anemia.



From Cloning to Clinic: A Report from the Gene Therapy International Working Group

Fanconi anemia presents unique challenges to the field of hematopoietic stem cell gene therapy. Patients have both reduced quantity and quality of these cells. Because of this unusual level of complexity, combined with the relative rareness of the disease, the Fanconi Anemia Research Fund (FARF) and Fanconi Hope, United Kingdom, established the Fanconi Anemia Gene Therapy Working Group in 2010. Led by Dr. Jakub Tolar (University of Minnesota), the goal of creating this group was to establish a worldwide platform for gene therapy clinical trials in FA.

Translational research has shown that DNA repair to restore cellular fitness in FA can be accomplished by either gene addition of the wild-type (normal FA gene) or by editing the genome. Both methods allow autologous cell therapy. This means that the therapy uses the person's own cells, thus avoiding the severities of pre-transplant conditioning and post-transplant complications.

The most recent meeting of the gene therapy working group, hosted by Juan Bueren in Madrid last October, showcased the tremendous advance from planning and preclinical research to clinical trials, as well as ongoing research investigations. The group explored a number of technical strategies to increase the efficiency of FA gene therapy. They also discussed the potential of gene therapy for head and neck cancers. The work of the gene therapy group is truly an international effort, with experts representing Spain, Italy, France, Netherlands, Germany and the USA.

This year the group will meet in Heidelberg, Germany, and continue collaboration to generate robust, autologous, and disease-free cells for people with FA. ■

“ The work of the gene therapy group is truly an international effort. ”

For example, formaldehyde, the simplest aldehyde, is produced in all of us in our blood from the metabolic process demethylation of RNA. Fortunately, formaldehyde is eliminated by the enzyme ADH5, which we also produce. This enzyme turns formaldehyde into the non-toxic formic acid. Another aldehyde produced by our metabolic processes, acetaldehyde, is removed by ALDH2, a protein that turns the toxic acetaldehyde into the non-toxic acetate.

Dr. Patel's lab studies aldehydes by creating and exploring aldehyde problems in mice. They do this by knocking out ALDH2 and ADH5, the mouse's first line of defense against the toxic effects of formaldehyde (ALDH2) and acetaldehyde (ADH5). The grim results—leukemia, bone marrow failure, liver cancer, and death—confirm the protective functions of ALDH2 and ADH5. However, many questions remain. Are there other sources of DNA repair problems besides formaldehyde and acetaldehyde? What other aldehydes matter? How much can we extrapolate from mouse models to humans? Dr. Patel's lab has identified at least 20 enzymes in the first tier of protection against aldehydes. The most fundamental and urgent question they are now trying to answer is, how can we help people with FA restore to full functionality their second tier defense against the toxic aldehydes that they produce-- their ability to repair damaged DNA? Are there other repair pathways that are protective, beyond the FA pathways? ■



A New Mechanism to Repair Genes

Johannes Walter, PhD, Harvard Medical School



Genes are essential for life. They contain the information needed to create and maintain our bodies: our height, hair color, immune system, the pitch of our voice, the strength of our heart — almost everything.

Because genes are so crucial, they must be protected from the elements that damage our cells everyday: sunlight, oxygen, foods, natural products, germs and more. A healthy Fanconi anemia pathway protects the DNA of our genes by repairing a specific type of damage called “cross-links.” In FA, the pathway is defective and repair is incomplete; cells fail and many genes become permanently damaged (“mutated”).

Dr. Johannes Walter, Harvard Professor of Biological Chemistry and an expert on DNA repair, was honored as a keynote speaker at the 2016 Symposium. He presented a series of molecular studies showing for the first time that cells have a second pathway to repair cross-link damage. The potential impact of this discovery on FA therapy is intriguing.

Lessons from the African Frog

Dr. Walter approaches research from the perspective of a biochemist. That is, he teases out molecular pathways that occur in cells by working with the molecules themselves in a test tube. His approach might surprise the non-scientist. He studies molecules from eggs of the African frog called *Xenopus laevis*. Why frogs?! As it turns out, the FA pathway in *Xenopus* is very similar to that in humans. And it’s much easier to study biochemistry in this model organism; *Xenopus* eggs are relatively large and easy to collect from a simple aquarium tank, yielding substantial amounts of material for biochemical study.

Cross-Links and Repair Pathways Come in Multiple Flavors

Dr. Walter and his research group recognized that not all DNA cross-links are alike. Some are caused by drugs and natural products such as cisplatin and psoralen, while others result from spontaneous decay of our DNA that creates “abasic” cross-links. Dr. Walter and his postdoctoral fellow, Dan Semlow, hypothesized that these different cross-links

with their different chemical structures might be repaired by different pathways. We know the FA pathway is important, but they asked: might there be other pathways?

The answer is a definitive yes! Using clever molecular techniques, Walter’s team found that the repair of psoralen and abasic cross-links occurs even when the FA pathway is inactive. Unlike the FA pathway where cross-links are “unhooked” and repaired by cutting the DNA strands, this new repair mechanism does not cut DNA and uses an enzyme called “NEIL3” to unhook cross-links. Thus, cells have at least two pathways to repair DNA cross-links: the well-known FA pathway and the newly discovered NEIL pathway.

Repair Pathways Have Each Other’s Back

The Walter team further showed that the FA and NEIL pathways can serve as backups for each other. They found that under normal conditions NEIL is the preferred pathway to repair abasic and psoralen cross-links, with the FA pathway playing a lesser role. But when NEIL is inactive, these cross-links are now repaired by the FA pathway. Conversely, when FA is blocked, all repair occurs via NEIL. Therefore, there is considerable flexibility in which pathway is utilized for cross-link repair.

Frogs? Biochemistry? What Does It All Mean?

The discovery of a second pathway that repairs cross-links is big news! This suggests there may be a way to reduce FA symptoms by increasing the activity of the second pathway. The first challenge is relatively easy: show that the NEIL pathway exists in humans. The bigger challenge will be to harness this new pathway to treat FA.

And what about frog biochemistry!?! Of course, we’re not trying to cure frogs. But biology is amazing; lessons learned from the simplest creatures (bacteria, worms, flies, frogs, mice...) provide critical insight into human disease. The frog is particularly good for studying gene repair, and the biochemical details we discover in frogs tell us where to look in humans. These details matter! They lead to cures. ■

Gene Therapy for FA Now a Reality; Gene Correction by Editing Next?



Hans-Peter Kiem



Jen Adair

Gene Therapy

Gene therapy trials for people with defects in the *FANCA* gene are now open in the United States, thanks to Drs. Hans-Peter Kiem and Jennifer Adair (Fred Hutchinson Cancer Research Center, Seattle) and in Spain, thanks to Drs. Juan Bueren and Paula Rio (CIEMAT/CIBERER, Madrid, Spain). Some of the encouraging early results from these trials were shared at the 2016 Scientific Symposium.

Both of these protocols use a sophisticated viral vector to add a normal *FANCA* gene to stem cells derived from the person with FA. These protocols differ in the age of the patients treated with gene therapy (younger in the Spanish trial), and also in how the target blood stem cells are collected and manipulated outside the person, acknowledging that these cells are neither as abundant nor as healthy as they are in people without FA.

Unlike conventional bone marrow transplantation, patients in these trials are not given conditioning treatments before they receive their own gene-corrected blood stem cells. After the procedure, gene-corrected blood cells are initially rare, but with time, corrected cells become more abundant in some patients. In all patients treated, overall blood cell counts are stable after the treatment.

Two people have been treated so far in the US and three have been treated in Spain. The longest follow-up has been more than two years for one of the Seattle patients. The hopeful news is first that this procedure is safe and second, the finding that corrected cells can engraft in some FA recipients. The Spanish trial data suggest that younger patients are more likely to benefit from gene therapy. The Seattle trial data suggest that minimizing manipulation of the stem cells outside of the body improves the efficacy of the gene therapy.



Gene therapy in FA: an international effort. Above: the Madrid team at the 2016 Symposium.

Gene Editing

FA mutations can be corrected by another approach: gene editing. By this method, the exact mutation can be repaired in a living bone marrow cell, rather than replacing the entire gene with a virus. Investigators including teams led by Dr. Francisco Roman-Rodriguez (CIEMAT, Spain) and Dr. Henri van de Vrugt (Vrije Universiteit, Netherlands) showed that defective FA genes in FA cells could be corrected by gene editing. Both labs used an editing system that was derived from bacteria (the CRISPR-Cas system). Using a version of this method, Dr. Mark DeWitt from the lab of Dr. Jacob Corn (UC Berkeley) described how he was able to correct the mutation that leads to sickle cell anemia in human bone marrow cells, return these corrected human cells to a mouse, and over time measure the rise in normal hemoglobin and the decline in sickle globin protein. Remarkably, the replacement of sickle globin by normal globin derived from the edited gene would be curative, if it were achieved in a human. One possible safety concern, off-target changes due to the editing process, are quite rare. Dr. Dewitt is working closely with a team of experts in hematology and bone marrow transplantation to develop a clinical protocol for people with sickle cell disease.

We now know the genes that are the basis of most Fanconi anemia around the world. These hopeful – but very preliminary – advances in gene therapy show the potential for alternatives to conventional bone marrow transplantation from one person to another. Come to the 2017 Symposium in Atlanta to hear the latest results! ■



Mark DeWitt



Francisco Roman-Rodriguez



Promise of Personalized Medicine for Oral Cancer

Eddie Méndez, MD, University of Washington & Fred Hutch Cancer Research Center

The prospect of cancer is a significant challenge facing adults with Fanconi anemia. For reasons not yet understood, the mouth and upper throat are particularly prone to disease. Dr. Eduardo (Eddie) Méndez, a leading clinical investigator at the Fred Hutch Cancer Research Center in Seattle, addressed this important topic in his keynote presentation at the 2016 Symposium. He talked about advances in treating head and neck cancer and his pioneering research to develop precise medicines that are “personalized” for each individual cancer.

Leveraging the Promise of Personalized Medicine

Until recently, cancer treatments were largely based on the location of the cancer (lung, breast, blood, etc.) and what it looks like under the microscope. However, we now know that this approach is overly simplistic.

Cancers that look alike on the outside are often vastly different inside the cancer cells themselves. These differences are shown through modern methods of “genomics” in a process called “genomic profiling” that analyzes all of the genes and gene products in a cancer cell. This involves the collection of millions

(sometimes billions!) of pieces of data from each cancer.

The result is a detailed molecular “signature” of each cancer. Dr. Méndez and his colleagues are examining these signatures in head and neck cancers to find weaknesses (Achilles’ heels) that can be targeted by drugs. Finding the right drug to target the right molecule in each individual cancer is called “precision oncology” and is an example of modern personalized medicine.

Genomic Signatures in Head and Neck Cancer

Dr. Méndez described the results of his “oralchip” study that identified a

131-gene signature in oral cancers that predicts survival. A 13-gene signature subset was subsequently found to be highly predictive of survival in HPV-negative oral cancers. These gene signatures are now being used in the clinic to guide treatment options in head and neck cancers.

Finding the Right Drug

The most frequently mutated gene in head and neck cancers is the *TP53* gene. Normally *TP53* prevents



damaged cells from growing. But when mutated, this “checkpoint” function is lost, and cells grow unchecked to become a cancer. Dr. Méndez and his colleagues hypothesized that mutated *TP53* may be an Achilles’ heel if these cancers rely on other genes to survive. If these other genes can be inhibited by drugs, then *TP53*-mutated cancers would fail to grow.

Their hypothesis was correct! Using high-throughput screens and big-data analyses, the Méndez team discovered several candidate genes to target in head and neck cancers with mutated *TP53*. Most notable was a gene called “*WEE1*”. Similar to *TP53*, *WEE1* also serves a checkpoint function to prevent damaged cells from growing. So when both *TP53* and *WEE1* are mutated or blocked in cancer, the cells die and the cancer dissolves.

Better still, the cancers disappear even faster when treated with a damaging agent in combination with a *WEE1* inhibitor. The results are dramatic. Dr. Méndez showed two cases of inoperable head and neck

cancers that shrunk to pea-sized tumors after treatment with cisplatin and a *WEE1* inhibitor called “AZD1775”. Studies are ongoing to determine how these drugs cooperate to cause cancer cell death and how to maximize the effects of *WEE1* therapy in head and neck cancers.

Precision Oncology in Practice: The Future is Now

Dr. Méndez spent the last few minutes of his presentation describing how his team is personalizing the treatment of patients with head and neck cancer. This is an intense process involving surgeons, hospital staff, research scientists, systems biologists, veterinarians and laboratories full of robots, computers, microscopes and state-of-the-art genomics and drug screening technologies. The procedure involves taking a small piece of a patient’s cancer, determining

its molecular signature, finding its Achilles’ heel, screening for drugs that attack this weakness, and quickly coming back to the patient with the best possible drug for his/her specific cancer.

These are exciting times for precision oncology. Leading cancer centers around the world are developing similar approaches to treat other types of cancer. The recent revolutions in cancer genetics, genomics and translational sciences provide enormous opportunities and hope. Dr. Méndez and his team are a big part of this revolution, leading the way to personalize the treatment of oral cancer and substantially improve the lives of people with this challenging disease. ■

“The recent revolutions in cancer genetics, genomics and translational sciences provide enormous opportunities and hope.”



Contact Marie Sweeten, Family Services Director | 541.687.4658 | marie@fanconi.org

Updates in Stem Cell Transplantation

With improvements in stem cell transplants and other treatments, the number of people with Fanconi anemia surviving well into adulthood worldwide has increased dramatically. Not all patients have equal access to effective treatments, however, and even those who do face a variety of health issues as they age. Because of the increased risk of cancers for those with FA, detecting cancers early and treating advanced cancers have become significant focus points of FA research. At the 2016 Scientific Symposium, Dr. Eva Guinan (Dana-Farber Cancer Institute) chaired a session on transplantation and disease challenges. Clinicians and researchers from around the world provided a number of updates:

- Dr Jordi Surrallés (Autonomous University of Barcelona, Barcelona, Spain) reported on a potential early biomarker for increased cancer risk and poor survival in FA. Blood DNA can be tested for chromosomal mosaic events (CMEs) which happen during embryonic development.

CMEs have recently been associated with aging and increased risk of hematological and solid cancers in the general population. Follow up of these patients 0-15 years after testing showed that hematologic and solid cancers were more common in CME carriers. Dr. Surrallés proposed that CMEs might be a useful biomarker to identify individuals at higher risk of cancer and should be evaluated as a monitoring strategy.

- Dr. Stefan Meyer (University of Manchester, United Kingdom) spoke about kidney abnormalities and dysfunction in 30 FA patients he studied over the last 20 years. Nearly half had kidney abnormalities, and half of those had recurrent urinary tract infections requiring antibiotic treatment and long-term prophylaxis. Kidney dysfunction was less common, but he found that kidney dysfunction increased with age and after stem cell transplant. Dr. Meyer therefore recommends regular monitoring of kidney function in the long-term management of FA.
- Dr. Marc Bierings (Utrecht University Children's Hospital, Netherlands)

reported on an analysis of a large, international cohort of adults with FA transplanted between 1991 and 2014. During this time there have been many changes in transplant protocols, most significantly the introduction of fludarabine (a chemotherapy medication used to treat leukemia). In this analysis, the two factors that significantly improved outcomes were the use of fludarabine and the availability of a matched sibling donor.

- Dr. Miharu Yabe (Tokai University School of Medicine) shared his experience with FA in Japan, where *FANCA* and *FANCG* are most common. Dr. Yabe has observed that *FANCA* patients more often develops myelodysplastic syndrome (MDS) and acute myeloid leukemia (AML) while *FANCG* patients tend to develop bone marrow failure earlier. Dr. Yabe found significant differences in 5-year survival between those with clonal abnormalities (53% survival) compared to those without (82%). Likewise, 5-year survival after transplant was poor (52%) for those with AML.

“ Bone marrow transplantation continues to be a vital subject for the FA community, with increasing success rates wherever the best protocols can be followed. ”



Carmem Bonfim

- Dr. Carmem Bonfim (Federal University of Paraná, Brazil) reported on 122 transplants done with HLA matched related and unrelated donors. The two groups were enrolled on different protocols, with fludarabine used for those with unrelated donors. All transplants were done without radiation. Overall, the matched transplants had an 88% survival rate at five years, and a 97% survival rate for those who had no prior transfusions. However, the limited availability

of immunosuppressive medications may be contributing to higher rates of graft-versus-host disease (GvHD), which reduces survival rates.

Many Brazilian families face the challenge of finding a matched donor. This is common in countries with many ethnic “minorities,” for whom it may be more difficult to find a match in the donor pool. Doctors at Johns Hopkins have developed a (non-FA) transplant protocol for use with a partial HLA match. Dr. Bonfim has modified this protocol for FA patients, with some success. Unfortunately, GvHD and mucositis are significant problems and the lack of immunosuppressive medication again complicates the outcomes. Transplant is often delayed, resulting in the potential for disease progression and reducing the potential for a successful outcome.

Bone marrow transplantation continues to be a vital subject for the FA community, with increasing success rates wherever the best protocols can be followed. Yet these very successes open the door to future challenges for adults with FA, who face enormous risks for cancer. FARF is now devoting significant resources to finding better ways to avoid, detect and treat these cancers. ■

HLA matched donor:

Human leukocyte antigen. A protein found on the surface of cells in the body; this protein helps the body determine what is “self” and what is “foreign”. An HLA-matched donor increases the chances that the patient’s body will accept the transplant as “self.”

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Advice from a Pharmacist

Tips to Ensure You're Not Overpaying For Prescriptions

By Alex Winn, PharmD (atwinn.pharmd@gmail.com)

In 2016, the Food and Drug Administration (FDA) approved 22 new drugs¹, including new treatments for patients with ovarian cancer, bladder cancer, soft tissue sarcoma, and chronic lymphocytic leukemia — as well as two new diagnostic agents for detecting certain forms of cancer. Several of these new drugs are entirely novel drug classes, which include the first treatment for patients with spinal muscular atrophy and the first drug approved to treat Duchenne muscular dystrophy.

While exciting new therapies may bring new hope for a cure, they can also bring high costs and significant insurance hurdles. Fortunately, there are resources available to help you and your family. Here are a few tips to get you started.

Patient Assistance Programs

Many drug manufacturers have patient assistance programs (PAPs) that provide brand-name medication at no cost to qualifying families. Most underinsured and low-income families will qualify. Two nonprofit websites provide help searching for a specific PAP application: www.rxassist.org and www.needymeds.org. Both websites display program eligibility

requirements and regularly update the PAP applications, which are available for download.

Prior Authorizations

Health insurance plans may require your prescriber's signature and the ICD-10 code (diagnosis billing code) for high-cost medications on a form known as prior authorization (PA) before allowing your pharmacy to fill the medication and bill insurance. You can always pay the "cash price", but be aware you may not be able to recover any of that money if the insurance company ultimately denies the claim. If your pharmacy tells you that a PA is required, then the best practice is to call the prescriber's office and let them know that the prescription requires a PA. Usually PAs are faxed or electronically submitted to the office from your pharmacy, and you can ask for confirmation that the office has received yours. It's possible that your PA was placed amid a stack of paperwork in the provider's office, so a gentle reminder that yours is awaiting attention can help speed up the process.

Off Label

Simply put, "off-label" means the prescriber wrote a prescription for an indication (a disease or diagnosis) that hasn't been FDA-approved for that particular medication. For

example, NexAVAR[®] (sorafenib), an antineoplastic tyrosine kinase inhibitor, is FDA-approved for the treatment of hepatocellular cancer, advanced renal cell cancer, and differentiated thyroid cancer, but is considered off-label for angiosarcoma and gastrointestinal stromal tumors. Insurance companies will often consider your request for an off-label use on a case-by-case basis, especially if the provider can provide published evidence that it has worked for the off-label indication from a peer-reviewed journal article or case studies. Ask your physician or the clinic's patient navigator to contact the insurance company and request that they approve off-label coverage.

Clinical Trials

Ask your physician if there are any clinical trials available that could benefit you or your loved one with Fanconi anemia. You can also search www.clinicaltrials.gov for active trials. According to their website, "*ClinicalTrials.gov is a registry and results database of publicly and privately supported clinical studies of human participants conducted around the world.*"²

Expanded Access

Also called "compassionate use," expanded access provides a

pathway for patients to gain access to investigational drugs, biologics and medical devices for serious diseases or conditions. These differ from clinical trials in that they are usually individual applications for single patients who do not qualify for a trial, often due to exclusion criteria, or when there are no trials available. Investigational drugs and devices have not yet been approved by the FDA and they have not been proven to be safe and effective. Therefore, they may be effective in the treatment of a condition, or they may not. It is important to remember that the drug/biologic/medical device may have unexpected serious side effects and that patients need to consider all the possible risks when seeking access to an investigational medical product.³ For more information visit www.fda.gov and navigate Home> News & Events> Public Health Focus> Expanded Access (Compassionate Use).

Editor's notes:

While specific to the healthcare landscape in the United States, these tips may also be relevant in other countries. Check with your pharmacist and insurance provider.

Contact Marie Sweeten, Family Services Director at the Fanconi Anemia Research Fund to learn about current or upcoming FA clinical trials (marie@fanconi.org). ■

- 1 U.S. Food and Drug Administration, Center for Drug Evaluation and Research. 2016 NOVEL DRUGS Summary, December 31, 2016. Retrieved January 19, 2017, from www.fda.gov/downloads/Drugs/DevelopmentApprovalProcess/
- 2 Retrieved January 23, 2017, from <https://clinicaltrials.gov/DrugInnovation/UCM536693.pdf>
- 3 Retrieved January 19, 2017, from www.fda.gov/ForPatients/Other/ExpandedAccess/ucm20041768.htm



By Peg Padden

“Never doubt that a small group of thoughtful, committed citizens can change the world; indeed, it’s the only thing that ever has.” —Margaret Mead

To the FA Community,

There is absolutely no doubt in my mind that all of us share the same goal: to find better treatments and a cure for this devastating disease that continues to impact our lives. As much as I want to lose 10 lbs., get my house organized and learn Italian, those goals mean nothing in comparison to this all-encompassing one.

So, what can I do? What can you do? From the moment my two sons Jake and Spencer were diagnosed in 2003, I wished I had paid attention in biology class, so I could do research. But since I didn’t and I can’t, these past 13 years I have done what I **can** do: raise money for research. Our ticket out of this lies with research. Nothing else.

The 8th Annual International Fanconi Anemia Day is May 1st. This day was started by families eight years ago to raise funds for research. Let’s get as close as we can to our goal! We won’t reach it with just a few of us. Since Fanconi anemia is an orphan disease, it gets very little federal money. If FAmilies don’t raise the money, who will?

Join us this FA Day! 2017’s slogan is “Stick it to FA!” Whether you host an event, write a letter, or simply make a page online to raise funds by sharing your story – every single effort makes an impact. The Fanconi Anemia Research Fund (FARF) is ready to help us with every step. You can start by visiting the official FA Day page (http://fanconi.org/index.php/fundraising/fa_day), emailing info@fanconi.org, or adding your fundraiser to this year’s Crowdrise page: www.crowdrise.com/2017FADay/.

Together we will find a cure. For my son Spencer, for your sons, daughters, grandchildren, and yourselves. I truly believe this. Together. ■



Cale Ferrin

From Diagnosis to Chasing Dreams – It’s All in a Day’s Work for Cale Ferrin

By Britteny Ferrin

Cale was diagnosed with Fanconi anemia (FA) when he was only 10 days old. However, before he was born, we knew something was not right. At 20 weeks, after our first ultrasound, we were given news that our son had hydrocephalus, or water on the brain. The water buildup on his brain was so severe, it was causing extreme pressure. We were told by our doctors that due to the pressure on his brain, the likelihood that our baby would be a functioning child was slim to none. But we knew God had a plan.

Fast forward to today...despite over 100 hospital visits and 27 surgical procedures, Cale is a thriving 10-year-old in the 4th grade. He loves music and gym, but does not like math. He is great at reading and socializing, but worries about getting homework done. Even though his battle with FA is a daily struggle, along with his occupational and physical therapy and other medically-related concerns, he is on an amazing adventure! You see, Cale is also an actor. In 2014, Cale auditioned for talent scouts and was

selected to attend the International Presentation of Performers (iPOP) showcase in Los Angeles. At the showcase, Cale earned high awards in seven categories, and won best child fashion model of the year. He has since signed with a talent agent and manager in the Los Angeles area.

Cale continues to pursue his passion for acting. His recent work includes earning guest star roles on a popular Nickelodeon kid’s show and the top-ranking series on TV Land. He has modeled for a national magazine, appeared in a national Target ad campaign, and has even starred in a music video that highlights his dynamic spirit (and his amazing dance skills). Cale has also had several local acting/modeling jobs. He has worked with a local university for promotional work for an athletic team, and has even co-hosted and emceed live events for major fundraising promotional campaigns. Later this year, Cale is scheduled to begin filming for a movie, in which he will star as one of the main characters.

Cale loves trying new things and meeting wonderful people. Even

though the work of an actor is hard, and not always fun, he still gets wholeheartedly excited when he books an audition. “You have a lot of pressure when auditioning, especially for a big role. You have to go over the lines, over and over and over again, until it is something that you can do ‘off script’. It is all about memorizing and knowing what to say next”, says Cale. Once he has the lines memorized, it is then that he has to add the emotional connection to the character. Cale works closely with a coaching team out of Los Angeles, who help him connect with the characters when practicing for a role. With Cale’s condition, he doesn’t get a lot of chances to audition for your everyday-type of child role. But, when he does fit the part, he continually receives high accolades on his acting skills, his charisma and his glowing personality.

In addition to his other health-related concerns, Cale is also missing bones in his hands and arms. Cale makes annual visits to the University of Minnesota’s Masonic Children’s Hospital for a bone marrow biopsy and follow-up visits with FA specialists. He also has a long list of local specialists he visits with annually. To Cale, he says his condition doesn’t really affect him at all. “I have to go to the doctor a lot, but don’t worry, it’s not a big deal.”

Cale hopes that his story inspires other kids his age to pursue their dreams, no matter how challenging they may be. “You can do anything you dream of, you just need to take the chance!” ■

Check out Cale’s music video with Jack Dempsey, who performed live at FARF’s 2016 Symposium. www.youtube.com/watch?v=Qak520sCfuY



Andie Kalembe and the Valparaiso University softball team

Team IMPACT

By Stan and Michelle Kalembe

We first heard about Team IMPACT from friends at Camp Sunshine, whose daughter had greatly benefited from the experience. Team IMPACT is a nonprofit organization whose mission is to improve the quality of life for children facing life-threatening and chronic illnesses through the power of team (www.goteamIMPACT.org). They do this by matching kids with local college athletic teams. The kids become official team members and participate in practices, games, and bonding activities.

Since no two experiences are ever the same, we really did not know what to expect when we signed up our 13-year-old daughter, Andie. Our friends told us how their daughter had bonded with her team and that she would attend team practices and go to games. They told us how the team members cared for their daughter and stuck up for her when she was having trouble at school. This was the type of relationship we wished for Andie, so we applied at the first chance we could.

Soon after, she was paired up with the Valparaiso University's women's softball team. From the start, the team has always made Andie feel welcome. From treating her to karaoke and dancing at their initial meet and greet, to running drills at practice, they have made her feel like one of their own. We attended as many practices and games as we were able to during the season, which was hard at times due to games scheduled during Andie's school hours. When she did attend a game, it was in full uniform, performing her duties as the team's batgirl. The season did not begin well for the team. There were many tough losses, but Andie was right there in the dugout cheering her team on as enthusiastically as the most diehard fan. To keep their spirits high, they made up chants and they took turns wearing a large orange safety cone on their heads for good luck. With the good luck cone on her head or her rally cap on, the team rallied around Andie for inspiration. The team overcame a hard season and came back to win the Horizon league championship! After each game they would have Andie first in line for handshakes

and she would be in the middle of every huddle. They call Andie their "secret weapon." Her love for softball has never been greater.

The benefits of being part of Team IMPACT reach beyond the softball diamond: these girls have become like big sisters to Andie. Throughout the offseason, they follow each other on social media, they text and share snapchats. During the summer, teammates invite her over to go swimming or invite her out to lunch. When Andie got a part in the school play, the team came in mass to cheer her on. Like good older sisters, they take an interest in her schoolwork and her hobbies, they gossip about boys and celebrities. Some of our favorite times with the team are when they invite Andie out to the university to hang out and cheer on the men's basketball team. GO CRUSADERS!

Besides watching Andie grow and interact with the team, what means the most to us is having the team and their families make our cause their own. Last summer, at an annual softball tournament in Indianapolis that is organized by one of the softball teammate's father, Andie was asked to throw out the first pitch to start the tournament and they donated a portion of the admission fees to Team IMPACT and FARF. This April at the team's home opener and two days before Andie's BMT anniversary, the team will be hosting their first ever bone marrow drive.

We mentioned earlier that no two people's experiences are ever the same. Andie's experience has definitely been unique and rewarding. Signing her up for Team IMPACT has been one of the best decisions we have ever made for her.

Interested in signing your child up for Team IMPACT? Want to volunteer? Learn more at www.goteamIMPACT.org ■





Gallegos family

Diving into an FA Diagnosis: Despair, Isolation, Hope and Gratitude

By Dana Gallegos

As many parents do, my husband and I would frequently find ourselves talking about our children after they went to sleep for the night. Our three children are Esmé, age nine; Lennon, six; and Luka, two and a half. On most occasions, those conversations included our immense gratitude for having healthy children. Most days, our biggest challenges would be to make sure school lunches were prepared, uniforms were washed, and teeth were brushed. Our world was completely knocked off kilter with Lennon's diagnosis of Fanconi anemia. He was diagnosed on June 10, 2016 and he received a bone marrow transplant on November 9, 2016. To say the past eight months have been an emotional whirlwind would be a gross understatement.

Lennon has always been small for his age. He was a tiny baby, weighing just four pounds and five ounces and measuring 16 inches in length at birth. Although he continued to grow more slowly, he met all of his physical and cognitive milestones. It was only after his diagnosis that we began to recognize markers that had never thrown up any serious red flags either for us or for his pediatrician. I first noticed that his right thumb did not look like his left just before he turned

of hypopigmentation on his neck could be additional clues. As he grew older we also noticed bruising, especially on his legs, which gave us slight pause. Of course, it was difficult to determine what amount of bruising was normal for an active young child and what might be a sign of something else.

When his growth really began to slow down and it became apparent that something was not quite right inside his body, I would have never guessed it was due to something genetic, to something I had never heard of. During the period of time when the only symptom was short stature, my personal worst-case scenario was a faulty thyroid, and I remember being so frightened by the possibility of daily growth hormone injections. Once blood work was done with a hematologist and it was discovered that all of his counts were low, my mind immediately went to the possibility of leukemia. I remember thinking that maybe it wouldn't be so bad. It has a beginning, an end, and a clear path of treatment. Plus, we live in Houston, where hundreds of children are successfully treated for leukemia every year at our local, nationally renowned children's hospital.

Lennon had a bone marrow biopsy shortly thereafter and it was confirmed that he was in bone marrow failure. We

also learned that leukemia was not part of the picture for him and we needed to accept a different possibility. Our hematologist taught us the existence of Inherited Bone Marrow Failure Syndromes, all of which

we had never heard of before. Upon doing some research, I gasped at the description of Fanconi anemia and its inclusion

“ This gratitude looks and feels a little different than it did even a year ago and it has been hard-fought; however, it has shaped us, changed us, and forced us to grow in ways that we never knew we needed. ”

a year old. I brought it to his pediatrician's attention, who assured me that it appeared to be normal and functioning. I had no idea that his few café au lait spots and small area

Diving into an FA Diagnosis, cont. on page 20

Growing up with Fanconi Anemia: Results of a Survey of Adults with FA



By Annette Urbanski and Stella M. Davies, MD,
Cincinnati Children's Hospital

Thirty years ago, a minority of children with Fanconi anemia survived transplantation and reached adulthood. Currently, people with FA are experiencing improved life expectancy and there is good reason to believe this improvement will continue. Children surviving to adulthood face a new set of challenges. Aside from the many medical worries, adults with FA face social obstacles such as acceptance from peers and being underestimated by society. There now exists the first large group of people with FA experiencing adulthood, and the struggles they face are virtually unknown to the world and even to the FA community, making their journey that much more valuable to share. We hypothesized that the constant uncertainty of future medical events and recent changes in outcomes might impact life choices for people with FA.

We designed a 20-question electronic survey to define social, educational and economic achievement of adults with FA, and to describe the influence of an FA diagnosis on stress, risk-taking, and personal concerns. The Fanconi Anemia Research Fund identified almost 200 adults with FA and distributed the questionnaire via email. Sixty FA adults with a median age of 29 and with age ranging from 18-64 responded to the survey.

Perspective

Having a serious, sometimes life-threatening illness undeniably impacts a person's view of the future. The results of the survey made it clear that FA was often on the minds of these adults. Forty percent of individuals who completed the questionnaire agreed that there was not a day that went by where they did not think about FA. In contrast, 13% said they rarely think about it. We recognize that results are highly subjective, and may differ by age and seriousness of diagnosis. We also asked how the FA diagnosis had shaped and changed our respondents, and generally, responses were positive. For example, many said FA helped make

them "caring" and "positive [people]." Despite this, 16% of participants said FA turned them into a cynical person. After noting this contrast, we wanted to know what led to these feelings of either happiness or cynicism.

Education

One of our objectives was to understand the influence of FA on choices and success in education. Overall, the group is impressively well-educated, with all but two having at least some college experience. Three were in graduate school, with eight having finished graduate school. When asked to rank a list of four variables, academics averaged second as most valued while growing up. However, it is possible that those who valued education most were more likely to respond to our questionnaire. Nevertheless, the results suggested that FA does not impair individuals cognitively or prevent them from pursuing academics. Moreover, our respondents were very successful in completing undergraduate and higher degrees.

Relationships

Having determined education was important, we chose to explore the value put on romantic relationships. Our survey revealed that romantic relationships were least valued during the teenage years. However, when asked about the current value respondents put on romantic relationships, 57% said they have a prominent role. In fact, 40% of adults who participated in the survey specified they were married, indicating that serious and long-lasting relationships were just as frequent as in the general population.

Friends play a key role in a person's life. We wanted to see if individuals with FA told their friends about having FA; and if they had told their friends, we wondered to what extent friends would comprehend what FA meant to them. The results showed 80% of the adults had told their friends they had FA. Of those who told their friends, only 20% said their friends fully understood. However, 46% indicated that their friends did understand, just not to the same degree as

they wished they had. These results highlight that having understanding and supportive friends is possible, but the majority, despite best efforts, may never fully understand

“Our most consistent finding is that FA did not prevent survey participants from living a happy life, with academic success, and with both love and acceptance.”

what FA means to someone who is diagnosed with it.

Many adults with FA have had a bone marrow transplant. Our data showed 30% of the people surveyed were related to their bone marrow donor. We were curious how it made our participants feel to have a healthy sibling. The reaction to having a healthy sibling was diverse. Many people were grateful that their sibling did not suffer in the same way they did. However, a few responses indicated it was difficult watching their sibling do all the activities they could not do.

Behavior

We also sought to learn if higher medical risks would prevent individuals with FA from living their lives to the

absolute fullest. We found that not to be true. Twenty-nine percent of people indicated that they lived their earlier years recklessly, then shifted to a more cautious lifestyle.

Participants might have been more reckless because many did not believe they would live a long life, as indicated by some answers. Behaviors individuals with FA participated in were skydiving, drugs, excessive drinking, tattoos, traveling, falling in love, and creating a family. The biggest regret reported by adults with FA was always worrying and not taking enough time to focus on relationships.

Conclusions

Our data show that FA has a profound influence on the majority of adults. The impact was reported to be both positive and negative, depending on the person and what period in their life the question was referencing. Our most consistent finding is that FA did not prevent survey participants from living a happy life, with academic success, and with both love and acceptance. The successful lives and achievements of most adults with FA are important to the parents raising children with FA, and for healthcare providers to share with their patients. ■

[Diving into an FA Diagnosis](#) *cont. from page 18*

of “short stature” and “abnormal thumbs” among the list of characteristics. I immediately knew that FA was what we were dealing with even before we received the official diagnosis nearly a month later. I spent the next weeks sick to my stomach, terrified, and reluctantly reading everything I could about this disease and, more immediately, the bone marrow transplant process.

On the day we received Lennon’s official FA diagnosis, I contacted FARF and joined the FAmily Facebook page soon after. It feels impossible to adequately describe what these resources have provided for our family. To have a child with a disease is isolating; to have a child with a rare disease adds an additional layer of isolation. To know and feel supported by others also walking this path helps to lighten our load and expand our perspective of knowing we are in this together.

Once we began to understand more about FA, we quickly made the decision to seek treatment away from

home. Our family temporarily relocated to Minneapolis in October 2016 so that Lennon could be under the care of Dr. John Wagner for his transplant. Although it is not an easy thing to do, we receive constant reminders that this was the right place, right people, and right treatment for Lennon. In addition to being among FA experts and other amazing medical professionals, we have had the joy and privilege of meeting and building relationships with other FAmilies during our time here. Having the ability to talk to others who truly know what you are going through can make the difference between despair and hope.

We are now celebrating Lennon’s 100 days post-transplant and are preparing to return home...and we remain grateful. Immensely grateful. This gratitude looks and feels a little different than it did even a year ago and it has been hard-fought; however, it has shaped us, changed us, and forced us to grow in ways that we never knew we needed. ■

Characterizing Hope

By Nancy Cincotta, LCSW, MSW, MPhil,
Psychosocial Director, Camp Sunshine

How does hope exist, and from where does it come? Where does hope live and with whom does it live? Does it live within an individual's constitution, in connection to others, or within the framework of a group? Is hope born, created, enabled, implied, inferred, intended, deferred? Does hope have its own path? Can hope be a reliable partner?

There is a unique avenue for hope that people impacted by Fanconi anemia, those living with FA and their family members, help engender for each other through sharing personal narratives. At times, it is about being in the same place, acknowledging a struggle, a success, or an aspiration. Just knowing that someone else is facing a similar challenge, in the context of living with such a rare illness, can both "enlighten and lighten" the road ahead. Sometimes it is simply about having one person in the world who truly understands. Perhaps not changing reality, but understanding it, can compel hope.

There are internal components of hope, those which contain the fabric of one's own inner experience (the things you think and feel when you are alone), and the external components of hope (those you share with friends and family, and even medical staff). Combined, these dimensions of hope sustain you. Perhaps throughout life we all reframe hope; we work to understand what we need and what is realistic at a given moment, and then how we need hope to change. We learn when we need to lean on hope to cope.

There is hope for today, to embrace and understand the present moment. There is hope for the future, what you would like to see happen, where you think you might be. There is hope from the past having lived every day, and seeing the strides made in science. There is hope compelled by growth. For instance, the Fanconi Anemia Research Fund's successes are the personification of hope. And then there is hope that grows as a result of challenge, those components

that enable hope in the face of adversity. Hope responds when it is called upon, so we can imagine that it lives in an eternal, accessible place.

As I write this, two parent posts have popped up on Facebook, one from a mother about to be the bone marrow and kidney donor for her child, who says "keep your fingers crossed" and the second from a mother moving towards gene therapy for her son; she says she is "nervous, excited and hopeful." Hope comes in the crossing of fingers, in the asking for support, and in seeing the potential of today's science and tomorrow's.

Hope comes couched in different ways on different days. The sunrise may be today's untold secret, whereas the friendship that allows for understanding may represent hope's enduring nature, and perhaps the life that is lived to its fullest and the quality of a life remembered may be the greatest embodiment of hope.

For decades, when writing about FA, I have had Amy to share my ideas with. I still feel as though I have that, and I still hold those memories close by. For me, they will always engender hope.

“...perhaps the life that is lived to its fullest and the quality of a life remembered may be the greatest embodiment of hope.”

It is with the dreams and admiration within a community of support and with a deep sense of connection that hope grows. To all of those who are enveloped in this world, hope lives here.

Looking forward to connecting,

Please contact me at nancycincotta@gmail.com to share your ideas about FA and how hope enables your journey. ■



a guest room, a warm shower and possibly a meal for the night.

Connections: If you live by our route, we'd love to meet you!

Encouragement: We would certainly appreciate a bit of cheering along the way as we tackle tough hills, obstacles, weather, and exhaustion. You can also follow us on Facebook and Instagram.

Fundraising: Throughout our journey, we will partner with Greater Giving (a crowdfunding website). This will provide FA families, friends, relatives and neighbors the opportunity to participate in the fundraising as well. If you would like to donate, go to: <http://tinyurl.com/C2C4KATA>

Purpose: We have 58 days on our journey. We will dedicate each day of our journey to a special FA fighter. Seeing the faces and reading the stories of the heroes we are riding for will provide us with motivation and purpose. Please share your story (or your child's, friend's, loved one's—living or deceased) by email to Stephanie Griggs: griggsvfive@gmail.com.

We sincerely thank you for supporting our Coast to Coast Bike Ride for Fanconi anemia. Please do not hesitate to contact me if you have any questions or feel you could offer support in any way.

Check out our website: www.KATAfoundation.org and follow our journey on Facebook www.facebook.com/KATArise/. Together, we can continue to fight Fanconi anemia, one mile at a time! ■

Coast to Coast Bike Ride for FA

By Steve Rice, President, KATA Foundation

Over 10 years ago, my good friends Ken and Jeanne Atkinson lost two children to Fanconi anemia.

Despite their tremendous grief, they were inspired to create the Kendall And Taylor Atkinson Foundation (KATA) to raise money to continue the fight against Fanconi anemia and enhance the lives of children. My name is Steve Rice and I am currently President of the KATA Foundation in Denver, Colo. To date, KATA has raised over \$1.8 million to fund Fanconi anemia research! Last April, Ken was senselessly killed outside his own home as he saved the life of a neighbor who was a victim of domestic violence. While the loss of our great friend was devastating, we know he would want us to continue the fight in his honor.

This summer, my close friend Dave Kummer will join me on a Coast to Coast bike ride from Oregon to Maine. We will cover over 4000 miles in 58 days, and travel across 12 states in addition to a few cities in Canada. Our purpose is to connect with FA families across the country, increase awareness

of Fanconi anemia, and raise \$125,000 to fund FA and cancer research. 100% of all money raised for the Coast to Coast ride will go to the Fanconi Anemia Research Fund (FARF).

The first day of our journey will take us from the Pacific shore into Eugene, Ore., home of FARF headquarters. This first leg will signify the strong partnership between KATA and FARF and our combined desire to improve the lives of those affected by this disease.

As we desire to donate every single dollar raised to FARF, it is our hope that costs incurred on the ride—food, lodging, bike repairs, etc.—will be minimized with the assistance of the incredibly resourceful and generous FA community. Please take a moment to see how you are able to assist us during this cross-country adventure.

Lodging, food, etc.: We would greatly appreciate any assistance with places to stay near the ride route (see below),





Hoot N Holler Celebrates 10 Years Raising Funds for Fanconi Anemia Research

By Stephanie Griggs

On a chilly Saturday evening in November, hundreds of people from across the country gathered at The Wildlife Experience in Parker, Colo. They were greeted by dozens of smiling faces looking down from banners hanging from the rafters. They are the faces of heroes and warriors - the faces of people with Fanconi anemia. "I'm why you're here" reads the banner of Taylor, just below the picture of a young man with a gentle smile. Taylor passed away in 2006 at the age of 18. He was the only son of Ken and Jeanne Atkinson, founders of the Kendall and Taylor Atkinson Foundation (KATA). His sister, Kendall, passed away two years before him. She has a banner, too.

Ten years ago, with immense support from family, friends, and their community, Ken and Jeanne launched the Hoot N Holler, a fundraising event like no other. Another Fanconi family in the Denver area, the Nash Family, jumped right in to help. Touted as a "Boot Stompin' Good Time," guests are encouraged to wear jeans, boots, and western attire as they generously open their hearts and wallets to support

KATA's purpose of "Fighting Fanconi, Giving Hope, and Enhancing Lives."

What draws people to this event year after year is not only the amazing food and experience, but also the knowledge that they are truly making a difference. Since its first year, the number of cowboys and cowgirls in attendance has grown to nearly 500! Guests are treated to delicious local cuisine and entertainment, which included everything from Fun Play Poker tables to fantastic dueling pianos. To remind guests of why they're in attendance, table tents feature the picture and story of children and adults with FA and those smiling faces on banners watch over guests throughout the evening. Additionally, a guest speaker discusses the impact this fundraiser has made on the lives of those affected by FA. Towards the end of the evening, Fanconi warrior Molly Nash often steps up on stage to thank guests and inevitably receives a standing ovation.

In addition to an entry fee for each guest, attendees are given ample opportunities to donate in unique ways, including a silent auction and a live auction featuring high value items and

trips, the Boots 'n Booze and Wall of Wine, a game of chance opportunity for guests to win top shelf liquor or high value bottles of wine, and Ponies and Stud Ponies, which is a way for guests to win a variety of gift cards. The evening also includes a special appeal to close out the event.

The 2016 Hoot N Holler was remarkable for many reasons. Not only was it the 10th annual event, but it was also an event to honor and "Tip Your Hat" to Ken Atkinson. Ken was senselessly killed last April. He died in the same manner as he lived, as a hero. The live auction included a very special item this year: Ken's bicycle. Ken's bike was unremarkable save for its owner—it was well loved and had been ridden many miles. Attendees saw the true value in Ken's bike and in the ultimate "Tip Your Hat to Ken," the live auction bidding for Ken's bike soared to an incredible \$10,000! A video tribute to Ken was the highlight of the evening and encouraged those who knew him to continue the fight Ken began many years ago. There wasn't a dry eye in the house.

With the assistance of dozens of volunteers, including family, friends, and other lives touched by the Atkinson and Nash families, and the generous donations from community members, businesses, and attendees, the 2016 Hoot was an incredible success. Hundreds of hours of preparation from dozens of volunteers go in to the preparation for this event, and it certainly paid off. In less than six hours, the 2016 Hoot N Holler raised over \$280,000 for Fanconi anemia research! ■

FAMILY FUNDRAISING EFFORTS

In 2016, FA families raised an incredible \$3,001,529 for the Fanconi Anemia Research Fund! More than 220 families raised funds, with 115 raising over \$500. Each dollar donated advances research and family support, making a difference for all those affected by FA and their families. Sincere thanks to every family and individual who worked so hard to raise funds in honor or in memory of loved ones.

Over
\$3 million
raised!

\$1,720,000 and up

Lynn Frohnmayer and Amy (Frohnmayer) Winn

\$240,000 and up

Kendall & Taylor Atkinson
Foundation with the Nash and Atkinson Families

\$100,000 – \$125,000

Nigel and Ann Walker
Peg Padden and Glen Shearer

\$45,000 – \$99,000

Steve and Jennifer Klimkiewicz
Kevin and Lorraine McQueen
Gerard and Cynthia Vandermeys

\$20,000 – \$30,000

Hermira Carvalheira
Kerrie Cazzari
Mark De Groot and Hanneke Takkenberg
Alan and Rachel Grossman
Tim and Mary Ann Lana
Todd and Kristin Levine

\$15,000 – \$19,999

Rachel Altmann and Tyler Morrison
Brian Horrigan and Amy Levine
Charles and Katy Hull
Ian and Tricia Mitchell

\$10,000 – \$14,999

Michael and Jennifer Aggabao
Adam and Marissa Becker
Chris and Susan Collins
David and Mary Ann Fiaschetti
Ronald and Vicki Gibson
John and Martina Hartmann
Rose and David Pennell
Emily and Neil Robison

\$5,000 – \$9,999

Brian Anderson and Sultana Graham-Anderson
Jimmy and Jenny Armentrout
Ryan and Becky Brinkmann
Cristie Chesler and Mark Bettilyon
Andrew Coons and Valeen Gonzales
Antonino and Marie Di Mercurio
Chloe Eminger and Family
Justin and Britteny Ferrin
Susan and Skip Gannon-Longstaff
Andrew and Jennifer Gough
Jeff Hoffman
John and Karilyn Kelson
Deane Marchbein and Cohen Stuart
Sheila Meehan
Mark and Diane Pearl
Peter and Janice Pless
Bob and Andrea Sacks

\$1,000 – \$4,999

Juanita and Ron Arroyo
Israel and Mary Jo Becerra
Randy and Nancy Bloxom
Jeffrey and Donna Boggs
Richard and Tena Bosen
Chris and Jennifer Branov
Chris Byrd
Anita Casani
David and Kim Chew
Colin and Ashley Chorneyko
Ana Concha
John and Kim Connelly
Brian and Margaret Curtis
Charles Deeks
Darrel and Kalani DeHaan
Donna DellaRatta
Egil Dennerline and Nanna Storm
Scottie and Jessica Dill
Daryn and Carol Franzen
Kevin Gatzlaff and Rachael Alaniz
Laurie Gerhardt
Owen Hall and Margaret Kasting
André Hessels and Rutger Boerema
Jeff and Beth Janock
Stan and Michelle Kalemba
Erik Kjos-Hanssen and Turid Frislid
Kwon Sejin and Jee-Ai Kim
Mark and Angela Lamm
Col. Gregory and Lt. Col. Lynnette Lowrimore
Tue Marker
Gianna and Lauren Megna
Ron and Fredi Norris
Fred and Nancy Nunes
David Ownby
Joshua and Crystal Pepper
Michael and Joanna Peros
George and Kathryn Reardon
Mark Ritchie and Lisa Mingo
Les and Nancy Ross
Rick and Lynn Sablosky

Ron and Elesa Schaefer
Bill and Connie Schenone
Bryan and Karen Siebenthal
Jan and Ken Sysak
The Family of Kelly Muschlitz-Taylor
William and Mary Underriner
Mike and Beth Vangel
Sean and Kristin Young

Up to \$999

Peter and Donna Abramov
Al and Janeth Acosta
Dorian and Kelly Adams
Cherie Bank and Michael Greenberg
John and Audrey Barrow
Jasmine Bennetsen
John and Francene Berglund
Tracy Biby and Family
Michael and Diane Bradley
Sean and Allison Breininger
Donald and Danielle Burkin
Robert and Barbara Capone
Jaclyn Catlett
Michael Christian
Tom and Mary Eilleen Cleary
Tyler and Teresa Clifton
Daniel and Melinda Coleman
Bradley Curry and Lea Ann Stiller
Wendy Delzell
Rob and Dawn Desmond
James and Carol Dillon
Pat and Mary DiMarino
Brian and Jennifer Dorman
Alex Eddy
Sharon Ellis
Billy Jo and Debbie Estep
Ezat and Laila Faizyar
Curt and Crystal Fales
Nancy and Scott Finnegan
Kim and Kevin Frock
Liz Funk
Mitch and Erin Furr
Emmanuel and Dana Gallegos
Gary and Melody Ganz
Mitzi Gerber
Brian and Lisa Gillott
Jessica Grady
Heidi and Gary Grassi
Ben and Stephanie Griggs
Abdul Hameed
Eric and Elisabeth Haroldsen
Bob and Victoria Hathcock
Greg and Diane Hayes
Patti and Mike Hilbert



The Griggs, Nash, Pearl and McQueen families at Hoot N Holler

Sam Hoffman
 The Family of Francesca Hutchins-
 Hull
 Shane and Colleen Irvin
 Steve and Cheryl Janowiak
 Lester and Nancy Jansen
 Lila Keleher
 Dan Klug and Elizabeth Bertrand-
 Klug
 Daniel Kold
 Christopher and Dana Lamb
 Martin Lamo
 Eddie and Maly Lee
 Eugene and Renee Lemmon
 Rene LeRoux
 Robert and Darla Lindenmayer
 Tanner and Jessica Lindsay
 Bill and Jackie Lucarell
 Leighsa Makowicz
 Dan and Nikki McCarthy
 Kevin and Barbara McKee
 Catherine McKeon
 Jeremy and Stacey Mefford
 Adam and Olivia Mindle
 Jim and Holly Miranda
 George and Sabine Mohr
 Sydney and Betsy Moore
 Griff and Cecilia Morgan
 Ashleigh and Tim Pinion
 Kenny and Lisa Myhan
 Tony and Lina Nahas
 Louis and Virginia Napoles
 Jack and Lisa Nash
 Jack and Tammy Neal
 Philip Nelson and Candy Lindsey
 David A. Neumann
 Robert and Mary Nori
 Lorraine O'Connor
 Michael and Katharine Ormond
 Albina Parente and Carmen
 Mignone
 Anne Park
 Stacia Pattison
 Derek and Ginger Persson
 John and Dianne Ploetz
 Lynn and Shirley Quilici
 Joel and Jennifer Ramirez
 Pedro and Marina Ravelo
 Leonard and Jan Riley
 Dov Rom
 Stanley and Lisa Routh
 Brian and Jennifer Sadlowe
 Richard and Dolores Satterlee
 Sharon Saunders
 William Schaecher
 Beatrice Score

Matt and Diane Senatore
 Lorne Shelson and Annette
 Waxberg
 Jack and Debbie Siegel
 Carol Siniawski
 Kevin and Joanne Smith
 Karen Steingarten
 Adam and Jennifer Stewart
 Charles and Jennifer Sumrall
 Gail and Steven Swanson
 Mary Tanner
 Bruce and Loreen Timperley
 Mark and Susan Trager
 Charles and Doris Trotta
 Steve and Melissa Turner
 Tom and Kathy Uno
 Louis and Theresa Viola
 Joe and Wendy Vitoritto
 Joseph and Natalie Vitrano
 Anthony and Elisa Walsh
 Marc Weiner
 Michael and Kim Williams
 Jeff and Nicole Winchester
 Wesley and Sue Wycoff
 Jian Yang and Jing Nie
 Tom and Marge Zaborney
 Cecelia Zurhellen

In Loving Memory

Alex Krinitzky4.15.02 – 5.29.16
 Joel Walker.....4.18.83 – 11.1.16
 Andrew Lake.....8.1.89 – 11.14.16
 Katrina Joy Jenkinson3.29.79 – 12.18.16
 Mirabel Klug.....1.19.14 – 3.14.17

SEVENTEEN FA FAMILIES RAISE RECORD FUNDS WITH HOLIDAY LETTERS

Last fall, The Fund asked FA families to participate in the annual holiday appeal for support. Seventeen families responded and cumulatively raised more funds than any previous year! We know the holidays can be busy and stressful, so we want to give extra thanks to the following families for their support and participation in 2016:

Altmann-Morrison	Griggs	Sacks
Brannock-Cazzari	Janock	Sysak-Petersen
Boson	Horrigan-Levine	Vandermeys
Brinkmann	McQueen	Walker
Di Mercurio	Pearl	
Fiaschetti	Ross	

Together, these families raised \$109,000, more than doubling the amount raised last year! A very special thank you goes to co-founder Lynn Frohmayer, who raised \$346,000 through the holiday letter campaign. The Fund is tremendously grateful to all those who donated in memory of Amy Winn and to honor the entire Frohmayer family.

We also thank every family who works to raise funds throughout the year to advance FARF's mission. We can't do it without you! This year's holiday campaign will begin the first week of November. Want to get a head start? Call our office at 541-687-4658. ■

FUNDRAISING SCRAPBOOK

On December 11, the Grossman Family was joined by their friends and family to celebrate Jacob's 18th birthday and to give back to the Fanconi Anemia Research Fund. At a restaurant in Skokie, Illinois, they shared a meal and festivities together. The evening was made even more special by two other FA families, the Kalemba's and the Gatzlaff's, who came to celebrate Jacob and support FARF. Altogether, nearly \$20,000 was raised in honor of Jacob. Thank you and happy birthday!

Jacob's Celebration



Faces of FA Calendar

Valeen Gonzales jumped into fundraising for FA research after her now two-year-old son, Teddy, was diagnosed in 2015. After a very successful bowling marathon last spring, she reached out to the Family last fall to collect photos for a new project: the Faces of FA calendar. Her colorful calendar features dozens of FA fighters and was sold for a couple months before the holidays. The calendar was a great success because it involved so many FA families and because it raised nearly \$1,000. Thank you, Valeen & co.!

To honor their daughter's memory, Rachael Alaniz and Kevin Gatzlaff wanted to do something special for what would have been her sixth birthday. They started Aria's Army, Inc. and planned a raffle offering a once in a lifetime experience: an all-expense paid trip to New York and tickets to see Hamilton on Broadway. News of the prize spread quickly and at the time the winner was drawn, \$13,225 had been raised! Thank you to the Alaniz-Gatzlaff Family for such a unique and awesome fundraiser to honor Aria and to support FARF.

Aria's Army Raffle



Music and Marathon

Last fall, the Aggabao family brought their family and friends together for a "Night of Hope Concert" in memory of their daughter, Katrina, and in honor of their son, Jared. Michael Aggabao shared his original songs with the audience and raised over \$2,000 for FARF. Two months later, Jennifer ran the New York Marathon for Katrina, Jared, and all FA angels and warriors. With the support of friends and the FA community, Jennifer raised more than \$3,000. Thank you, Aggabao Family!

On September 24, 2016, people of all ages donned capes and masks and gathered in Renton, Wash. for the FARF Superhero Run. Organized by the Robison and Graham-Anderson families, the run is inspired by their own three young FA superheroes: Blake, Isaac, and Avery. By the end of the day, they had raised nearly \$19,000 for FA research and family support! The event was such a success, they plan to continue the superhero theme in 2017. Mark your calendars for September 23! www.farf5k.com.

2nd FARF Superhero Run



		PATRIOTS									
		0	1	2	3	4	5	6	7	8	9
ATLANTA FALCONS	0										
	1										
	2										
	3										
	4										
	5										
	6										
	7										
	8										
	9										

Super Bowl Squares

At their Super Bowl party this year, the Brinkmann Family invited friends and family to participate in something more than a football game. Two of the Brinkmann children, Sydney and Zach, have Fanconi anemia. Becky Brinkmann, their mom, organized a game of Super Bowl Squares to get people to donate to FARF and have a chance to win (see www.superbowlsquares.org). At the end of the game, the Brinkmann Super Bowl Party group had raised \$1,400!

On the cold and blustery morning of October 23, 2016, Team FARF participated in the Newport Pell Bridge Run in Rhode Island. The team, comprised of 22 Family members, researchers from the University of Rhode Island and loved ones, raised nearly \$10,000 for the Fund! Big thanks to the Eminger, Makowicz and Fiaschetti Families! 2017's race is scheduled for October 22. Registration opens May 1. Visit www.pellbridgerun.com.

Pell Bridge Run



Running to Fight Fanconi

When their son Zach was diagnosed with Fanconi anemia in August 2015, Melissa and Adam Becker approached the life-changing news as best as they could: instead of living in fear, they were determined to fight back. On October 8, Adam ran in the Rock N Roll Brooklyn Half Marathon in New York. As he stated, "with all the doctor appointments, hospital visits, needle sticks, and biopsies that Zach has already endured in his five short years, the least I can do is drag myself through 13 miles!" In the end, Adam raised more than \$10,000 for FARF. Way to go!

FARF GRANTS AWARDED OCTOBER – FEBRUARY



From September 2016 to February 2017, the Fanconi Anemia Research Fund awarded **\$333,600 in research grants to the following projects:**

The FA Research Fund receives numerous applications for grants throughout the year. In order to better manage ongoing proposals and to support our targeted grants program (CFAR), the Fund will now operate on a singular grant cycle (as opposed to twice a year).

<p>Investigator: Blanche P. Alter, MD, MPH National Cancer Institute</p> <p>Title: A Training Grant for Development of a Fanconi Anemia Cohort in Mexico</p> <p>Amount: \$75,500</p>	<p>Investigators: Peter M. Glazer, MD, PhD; Gary M. Kupfer, MD Yale University of Medicine</p> <p>Title: Use of triplex-forming PNAs as a strategy for correction of the FA phenotype</p> <p>Amount: \$112,500</p>
<p>Investigator: Jacob E. Corn, PhD University California-Berkeley</p> <p>Title: Defining tractable approaches for gene editing of Fanconi Anemia hematopoietic stem cells</p> <p>Amount: \$125,600</p>	<p>Investigator: Akiko Shimamura, MD, PhD Harvard Medical School</p> <p>Title: Pilot Study of Metformin for Patients with Fanconi Anemia</p> <p>Amount: \$20,000</p>

The Fund is committed to supporting research to further our mission of finding new treatments and a cure for Fanconi anemia. Over our **28-year history, we have funded **219 research grants** and one service grant to over **110 investigators** worldwide. The total amount of research dollars awarded is over **\$20 million!****

FUND WELCOMES NEW BOARD MEMBERS

The Fanconi Anemia Research Fund is very pleased to welcome new members Rachel Altmann, Nancy Golden, André Hessels, and Bill McCoy to the Board of Directors. The Fund's board is comprised of community leaders and long-time supporters of the organization. The appointment of these four new members will strengthen the work of the board to advance the Fund's mission. Experts in various fields, FARF looks forward to their individual talents and contributions.

Rachel Altmann; Portland, Ore.

Rachel has been active in the Fanconi anemia community for several years, as an FA parent, a contributor to FARF newsletters, and a fundraiser. She has worked in the education sector for many years, as an environmental educator at Hawai'i Nature Center and most recently as Library Outreach Specialist at Multnomah County Library in Portland, Ore. Her daughter, Nina, succumbed to complications from Fanconi anemia in 2006 at age three. Rachel continues to honor Nina's legacy by working to advance FA research and support other families who face FA.



André Hessels; Wayne, Penn.

André became involved with the Fund when his two children, Dylan and Joy, were diagnosed with FA in 2013. Since then, he has participated in numerous runs to raise funds, including the New York, Chicago, and Boston marathons. André is a seasoned corporate financing specialist who currently works as VP Program Manager for Healthcare & Life Sciences at DLL Financial Services in Wayne, Penn. Originally from the Netherlands, André has worked in Europe, China and North America as a relationship banker for Rabobank. He brings a wealth of financial knowledge to the board and is looking forward to expanding his fundraising efforts.



Nancy Golden; Springfield, Ore.

A pillar of the education community in Oregon, Nancy has served in a number of leadership roles, including Superintendent of Springfield School District, Chief Education Officer of the Oregon Education Investment Board, and Education Policy Advisor to Oregon Governor Kitzhaber. She is currently a professor of Practice, Educational Methodology, Policy and Leadership at the University of Oregon. A friend of the Frohmayer family, Nancy has great respect for FARF and is looking forward to advancing the work of the organization.



Bill McCoy; Orlando, Fla.

Bill is a long-time FARF supporter, raising funds over the past decade with his annual event, Your Rope Team Mountain Climb. Bill is Senior Vice President and Chief Information Officer for Universal Parks and Resorts in Florida. Previously, he served as Vice President of Global Infrastructure for IBM. Bill is an avid supporter of local and global causes for children. He has completed 40 marathons, including 15 consecutive Boston Marathons. He is also an accomplished speaker who frequently speaks on the power of teams and is working on a book entitled "Who do you want on your rope team?"



I Want to Raise Funds – Where Do I Start?

We are here to help you fundraise! Visit www.fanconi.org and click "Fundraising" to view the fundraising toolkit, a step-by-step guide to making sure your event – big or small – is a success. Call the office at 541.687.4658 or email info@fanconi.org to request a hard copy sent to you by mail. We look forward to working together!

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Sherri Van Ravenhorst, Communications and Conference Coordinator

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FUND SAYS FAREWELL TO LONG-TIME BOARD MEMBERS

It is rare that an organization experiences such tremendous commitment and service from volunteers. We would like to acknowledge the contributions of two long-time board members, Barry Rubenstein, JD, and Eva Guinan, MD., and thank them for their years of service.

Barry first joined the board in 1998 as secretary. Four years later, he became president, a position he expertly held for 14 years. The FA Research Fund has greatly benefitted from Barry's leadership, fiscal and legal advice, and friendship for 20 years. He skillfully led the Fund through critical moments of growth and transition, working to ensure long-term stability and success. He was a voice of reason and thoughtfulness as the organization transitioned from a locally-focused nonprofit to one with worldwide impact in the fight against Fanconi anemia.

Barry reflected on his time with the Fund: "my tenure on the board was personally rewarding by being a small part of the amazing growth of the organization and its science, and by the opportunity to associate with such a remarkable group of people."

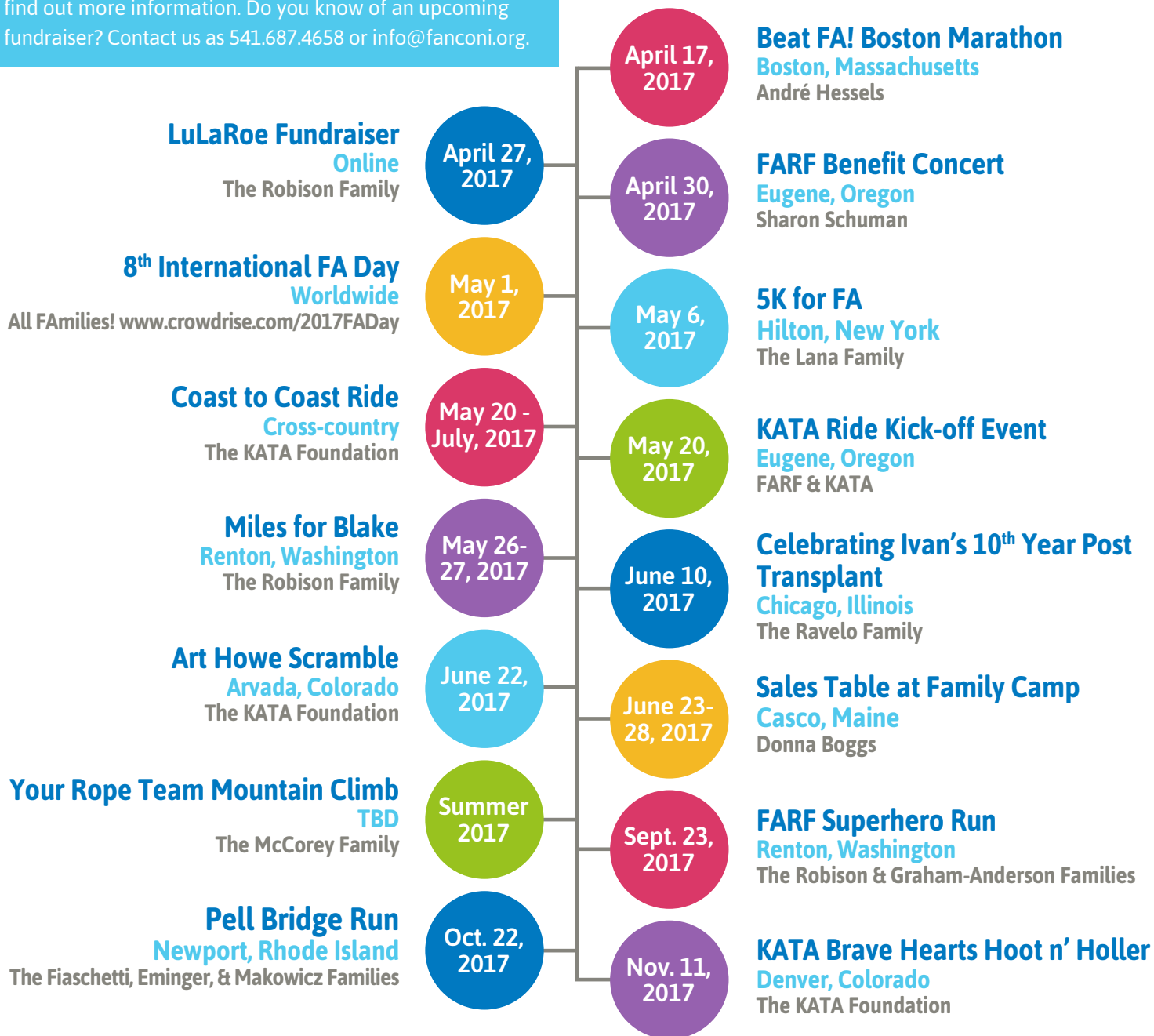
Dr. Guinan served on the Fund's Scientific Advisory Board from 2000 – 2016. She has been a vital voice in helping determine FARF's scientific and clinical priorities. David Frohnmayer, Fund co-founder, presented Dr. Guinan with the Distinguished Service Award in 2009 for her "extraordinary leadership in advancing clinical care for people with FA." Dr. Guinan was chair of the Fund's first Consensus Conference in 1998 to establish standards of clinical care. She went on to chair the process again in 2003, 2008, and 2013, which resulted in the publication of *Fanconi Anemia: Clinical Guidelines for Diagnosis and Management*. She has chaired many sessions at FARF Symposia and has served as a mentor to several young investigators.

"It has been an honor and a pleasure to work with all of you and to participate in advancing the science and clinical care of this disorder. My involvement with patients and families has been an endless source of personal growth. I thank all of you collectively for not only enriching my own life, but also for setting a standard of engagement that has been a real inspiration," said Dr. Guinan.



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.

HOW YOU CAN HELP

Donations Online:

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

Donations by Phone:

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

Donations by Mail:

1801 Willamette St., Suite 200, Eugene, OR 97401

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