



FAMILY Newsletter

Spring 2020

A semi-annual publication of the Fanconi Anemia Research Fund

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TO ALL INDIVIDUALS WITH FANCONI ANEMIA: IT'S TIME TO JOIN THE FA REGISTRY

WHO IS THE FANCONI ANEMIA REGISTRY FOR?



adults with
Fanconi anemia



minors with Fanconi
anemia (parents sign them
up)



anyone with a diagnosis of
Fanconi anemia, anywhere
in the world



the FA research
community

WHY SHOULD I JOIN THE REGISTRY?

Once you are in the registry, you can find out when there is an opportunity for you (or your child) to participate in clinical trials or research opportunities.



You'll have a personal,
private record of your FA
medical history



The more people in the registry, the
better researchers can direct
studies that lead to better
treatments, faster.

HOW TO JOIN THE REGISTRY



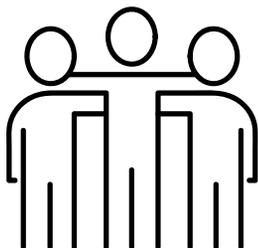
Visit
fanconiregistry.
iamrare.org
to create your private
account



Answer questions about
your personal info and
experience with FA.
Come back to
complete at any time.



Don't have
access to all
of your
medical
records?
We can help.



Fanconi anemia is a rare disease and can be hard to diagnose and treat. Research is the key. And the key to research is you.

If you have been diagnosed with Fanconi anemia, or your child has FA, join the registry today.

JOIN THE FA REGISTRY TODAY: [HTTPS://FANCONIREGISTRY.IAMRARE.ORG](https://fanconiregistry.iamrare.org)

Research Update: Cancer in FA

Fanconi anemia (FA) is a hereditary disorder with a high predisposition to cancer, especially leukemia and squamous cell carcinoma (SCC).

Head and neck squamous cell carcinoma (HNSCC) is the most common SCC diagnosed in people with FA. Diagnosis typically occurs at an earlier age in this population and patients often have poorer outcomes compared to the non-FA population. Many FA researchers are focusing efforts on advancements in early detection, prevention, and genomic analysis of FA tumors and pre-clinical SCC FA models. Researchers presented on these topics the 2019 FARF Scientific Symposium.

Dr. Eunike Velleuer from the University of Düsseldorf and the Children's Hospital Moenchengladbach Neuwerk presented her work on a clinical research study that was initiated in 2006 for early detection of oral lesions in people with FA. The goal of the study was to determine whether visual inspection of the oral cavity, followed by brush biopsy and cytological sampling and DNA analysis of visible oral lesions, has high diagnostic accuracy. The study results show that this approach is indeed a useful technique to detect the presence of cancer. Importantly, this work demonstrates that people with FA with a negative test result would not require additional incisional biopsies for diagnosis. The technique also results in detection of a substantial number of SCC and precursor lesions at a non-invasive or early stage. This study was recently published in the journal *Cancer Cytopathology*. For more on this study, see page 4.

Dr. Parinda Mehta from Cincinnati Children's Hospital reported on outcomes from a phase II clinical chemoprevention trial for squamous SCC in people with

FA. The purpose of this phase II study is to determine whether quercetin (a naturally occurring anti-oxidant) treatment for 2 years will prevent or delay the development of SCC in 45 post-transplant and 10 pre-transplant patients with FA. Clinical and laboratory tests will determine whether quercetin is effective at reducing oxidative stress and DNA damage. The hope is that this will lead to a new prevention strategy for SCC in post-transplant patients with FA that will eliminate or at least delay the development of SCC.

So far, 20 people with FA were enrolled in the study between May 2018 and May 2019. The median age is 17.2 years; 17 people were post-hematopoietic cell transplant, and 18 of the participants have been on quercetin for a median of seven months. All patients are tolerating quercetin well, and in six patients with available results, the frequency of micronuclei (a measure of genomic instability and DNA damage) in oral mucosa brushings decreased by 29% after one month (in 5 patients) and after six months of treatment (in one patient) with quercetin. The interim results demonstrate that quercetin therapy is well tolerated and leads to improvement in surrogate markers of genomic instability/DNA damage in buccal mucosa.

Dr. Josephine Dorsman and Mr. Khashayar Roohollahi from Amsterdam University Medical Center presented their work focused on (1) identifying the vulnerability in FA head and neck squamous cell carcinoma (HNSCC) and on (2) characterization of preclinical models that are used to study the mechanisms of the disease. These are important studies because developing an understanding of

how FA-pathway deficient cancer cells acquire growth advantages is needed to develop tailored and less-toxic therapeutic options.

Dr. Dorsman presented the work of Dr. Govind Pai, a post doc in her group, who is trying to identify which proteins are required to keep FA cancer cells alive. By "knocking down" or eliminating different proteins using a special screen, Dr. Pai hopes to identify drugs that will target these proteins without affecting normal or non-cancerous cells. Dr. Dorsman's research group identified RBBP9 as a potential protein that could be targeted to kill FA cancer cells. RBBP9 has also been shown to play a role in pancreatic cancer cell survival by other research groups. Dr. Dorsman will continue investigating RBBP9 and additional factors that are crucial for tumor cell survival that could be exploited for clinical benefit.

Mr. Roohollahi, a student in Dr. Dorsman's research lab, presented his work on using a range of sequencing methods to characterize the mutation and expression profiles of six cell lines derived from head and neck tumors from people with FA. Researchers also performed this analysis on cell lines derived from non-FA tumors. The results show that the FA-HNSCC cell lines do not differ drastically from non-FA tumor cell lines and that specific genes involved with immune system response, tumor-promoting signaling, and cell cycle regulation were different between these two groups. This work demonstrates the importance of understanding the underlying mechanisms of tumor development and survival as a way to exploit therapies. ■

Can brush biopsies identify pre-malignant or malignant oral lesions? Study results show **yes**



Ralf Dietrich (FA German Family Support Group) takes a sample from Alejandra at the FA Adult Meeting

Over the past 13 years, a team of individuals from the **Fanconi Anemia German Support Group**, plus FA researchers and clinicians from the **University of Düsseldorf**, have conducted a study to determine if oral pre-malignant and malignant lesions can be accurately identified using brush biopsies. These biopsies provide a painless and non-invasive method to obtain oral tissue samples. Researchers collected data from more than 700 participants worldwide. They summarized their results in an article entitled *Diagnostic accuracy of brush biopsy-based cytology for early detection of oral cancer and precursors in Fanconi anemia*, published in the journal *Cancer Cytopathology*, February 2020.

Individuals with Fanconi anemia (FA) are at high risk of developing oral cancer. Unlike cancers found in the general population, these FA cancers cannot be treated with chemotherapy because of the underlying DNA repair problem. Therefore, early detection and surgical intervention are essential.

Oral cancer begins as a precancerous lesion. If a lesion does not heal in four weeks, FA individuals are advised to

consult with an otolaryngologist (ENT physician). If the lesion appears suspicious, the physician often performs a biopsy. Could precancerous lesions be detected much earlier, *before* they become cancerous and without enduring a painful, invasive biopsy?

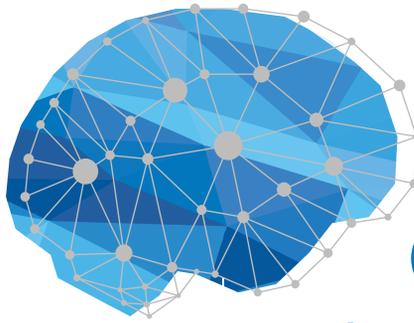
Dr. Schramm, head of the department of Cytopathology from the University of Düsseldorf, noted that routine use of brush biopsies in the general population

is controversial. Dr. Schramm noted: “Especially in a patient cohort like individuals with FA, where changes of the oral mucosa develop frequently, we had to show that this method is accurate.”

The published results show that this method is, indeed, accurate. Using two different methods of analysis, the “sensitivity” of these studies was 100% (ability to correctly identify those **with** pre-malignant or malignant lesions) and the “specificity” (ability to identify those **without** disease) was 92.2%. Equally significant, approximately 63% of squamous cell carcinoma and precursor lesions were detected at a non-invasive or early stage, making surgical removal effective.

This is how brush biopsies work: the lesion is swabbed, and if the brush test is negative, the lesion can be monitored clinically, and an invasive biopsy is not necessary. If the test result is ambiguous or positive, the results need further investigation. Dr. Schramm notes: “We don’t want to avoid invasive biopsies completely. But we hope that with the brush biopsy as a screening tool, we will identify those lesions that should be treated by experts.” Dr. Eunike Velleuer, University of Düsseldorf, hopes that this procedure will improve the basic care for individuals with FA who develop visible

“ If we can identify common risk factors and learn more about the nature of those cancers, we can work on prevention. ”



Central nervous system abnormalities in Fanconi anemia

oral lesions. These lesions are more frequently malignant in FA than in the general population, but only a minority of all visible oral lesions in people with FA is worrisome, which is why it is unnecessary to biopsy all FA oral lesions.

Ralf Dietrich of the German Support Group believes that in the past, many people who developed oral cancer experienced poor survival because they were diagnosed too late. Dietrich is convinced that with routine use of brush biopsies, oral cancer will be diagnosed much earlier and will therefore be more treatable.

Dr. Velleuer sums it up: “Now that we know this methodology is an accurate way to detect early cancers in FA patients, we can look to the future with more hope. We need to make this technology widely accessible so that all individuals with FA can have these screenings performed by their doctors, and learn why people with positive results developed cancer. If we can identify common risk factors and learn more about the nature of those cancers, we can work on prevention.”

Are you interested in participating in the oral screenings study? Team members are available at the FA Family Meeting and Meeting for Adults with FA every year, in addition to numerous gatherings of FA families around the world. The team also visits individuals in their homes to perform these screenings. Please contact Suzanne Planck, FARF Family Services Coordinator, if you're interested in learning more: Suzanne@fanconi.org, 541-687-4658. ■

Fanconi anemia (FA) is associated with bone marrow failure, cancer predisposition, and multiple congenital abnormalities. Brain atrophy and other abnormalities of the brain have also been reported, although the cause of these abnormalities and recommendations for clinical care are not yet defined. At the 2019 FA Scientific Symposium, Dr. Stella Davies from Cincinnati Children's Hospital Medical Center and Dr. Tekin Aksu from Hacettepe University in Ankara, Turkey, presented on their recent work focused on central nervous system (CNS) abnormalities in people with FA.

Dr. Davies discussed case reports of five FA patients with CNS abnormalities. Magnetic resonance imaging (MRI) of the brain showed that all five cases shared similar lesions (abnormalities) and features of inflammation. The identified lesions were often ring-like in appearance, and in all cases, the patient's muscular activity was affected. Four of the five patients with acute lesions received steroids and had a significant response. Interestingly, staining of brain biopsy tissue revealed expression of the JC virus. JC virus is one of three very similar polyoma viruses that affect humans (JC, BK and SV40 virus).

Polyoma viruses are found in many individuals with FA, as well as in the

non-FA population. The majority of people who contract these viruses do so during childhood. In general, these viruses do not cause disease, but can reactivate in people with suppressed immune systems (i.e., people with FA). The reactivation of JC virus in immunosuppressed people causes brain inflammation called progressive multifocal leukoencephalopathy (PML). Dr. Davies' research group hypothesizes that the brain lesions described in the five case studies result from inflammation caused by the JC virus.

Dr. Aksu discussed a recent analysis of 314 FA patients treated at the Hacettepe University from 1976 to 2019. Out of these 314 patients, 33 were evaluated with an MRI of the brain. The study found at least one abnormality in 18 patients of the study group. The abnormalities included density changes and reduced blood flow to the brain that led to brain cell damage. Dr. Aksu's research group hypothesizes that these findings are related to impaired DNA repair mechanisms and/or congenital vascular malformations (irregular formation of blood vessels supplying the brain).

These important studies highlight new clinical features in people with FA. More research is needed to understand how to best treat and prevent the brain abnormalities from occurring. ■

2019 SYMPOSIUM HIGHLIGHTS



One of FARF's guiding principles is to gather researchers and clinicians in the FA community every year to share updates in research and care. In September 2019, we held the 31st Fanconi Anemia Scientific Symposium in Chicago, Ill. Forty-four presenters gave talks and 58 presented posters. Presentations addressed a number of topics relevant to the understanding and treatment of Fanconi anemia. Topics included the FA pathway (p. 8), gene editing and gene therapy, hematopoietic stem cell biology (p. 10), central nervous system abnormalities (p. 5), and cancer in FA (p. 3-4).

A dynamic opening session

Dr. Stella Davies of Cincinnati Children's Hospital Medical Center opened the conference with a session entitled "It is not in the stars to hold our destiny but in ourselves". The appropriately titled panel featured talks from four different stakeholders in the FA community. First, Jack Timperley shared his experiences and challenges as a young man living with FA, including physical differences, dating, and how being different has informed and inspired his career path. Next,

Lindsey Romick-Rosendale, a researcher at Cincinnati Children's Hospital Medical Center, spoke about metabolic abnormalities associated with FA. Dr. Margaret MacMillan from the University of Minnesota then presented on her experiences as an FA clinician. Finally, FA parent Rutger Boerema shared her family's struggles and triumphs with FA, speaking to the experiences of all three of her children – two with FA and one without. Video of this session is available on FARF's website and YouTube channel.

Poster sessions

Fifty-eight posters were presented covering a range of topics, including bone marrow failure, patient registries and natural history studies, cancer in FA, and the FA pathway. Two poster receptions allowed presenters to share their work with other researchers, doctors, and FA family members.

Each year, a panel of expert FA researchers and clinicians determine the best three abstracts/posters. Each winner is presented with an award and a \$500 check. Congratulations to the 2019 abstract/poster award winners:

Best Clinical – Melody Mazon for "Molecular chaperone binding profiles reflect cellular phenotypes of Fanconi anemia missense mutations"

Best Translational – Jose Casado for "NKG2D – ligands expression in hematopoietic stem cells from FA patients: implications in Bone Marrow Failure"

Best Basic – Ivan Rosado for "A novel endogenous source of DNA damage in Fanconi anemia"



FARF Tank

FARF Tank made a splash for the second year! The contest allows up and coming researchers an opportunity to pitch their innovative ideas to the scientific community and win a \$10,000 grant. Eight contestants had only five minutes each to pitch their ideas to the audience and our panel of judges. At the end, the audience voted for the project they thought most deserved the \$10,000 grant. The judges also chose a winner. Congratulations to this year's winners:

Sylvie Van Twest (people's choice): Expanding the Fanconi anemia research and diagnostic toolkit: Developing alpaca nano-antibodies

Adam Nelson (judges' choice): Viral Specific T cells for HPV-associated SCC



Jack Timperley accepts the Winn/Byrd Award for Adults with Fanconi Anemia

Winn/Byrd Award

Jack Timperley, 20, received the third Amy Winn and Christopher T. Byrd Award for Adults with FA. As a member of the Fanconi Anemia Adult Council, Jack serves as a voice for other adults and teens living with FA. He inspired the audience, telling his story and expressing just how crucial their role is in advancing outcomes for Jack and others with FA. To read more about Jack, see page 23.



Alan D'Andrea and his team

Alan D'Andrea Receives Lifetime Achievement Award

The FA Research Fund has existed for more than 30 years, and in that time, few researchers can say they've dedicated their careers to studying Fanconi anemia. Dr. Alan D'Andrea is one of those researchers. He first received funding from FARF in 1998, and went on to receive four more grants from FARF, which led to much larger grants to study FA from the National Institutes of Health. Dr. D'Andrea has continued to make tremendous advances over the last 20+ years and has recently joined FARF's board of scientific advisors. We are immensely grateful for the work of Dr. D'Andrea and his team. ■



Grant Rowe Receives David B. Frohnmayer Award

Dr. Grant Rowe of Boston Children's Hospital became the third recipient of the David B. Frohnmayer Award for Early Investigators. The award was inaugurated in 2015 to honor the memory of FARF co-founder David Frohnmayer, who passed away earlier that year. To honor David's commitment to mentorship and to young investigators, the award is meant to acknowledge an early investigator who has significantly impacted FA science and contributed to the FA community at large.

Is targeting the FA pathway a promising way to treat people with Fanconi anemia?

Mutations in 23 Fanconi anemia (FA) genes cause defects in DNA repair, which leads to chromosome instability, bone marrow failure, malformations, and susceptibility to cancer. The most well-described role of FA genes in DNA repair is known as the “canonical” or typical role. Much is known about how the FA proteins and the central pathway member, FANCD2, orchestrate DNA repair. When DNA is damaged, it starts a cascade of events that require specific interactions between FA proteins. For example, proteins such as FANCA, FANCG, and FANCC, form a complex that is required to monoubiquitinate (or modify) FANCD2 and FANCI. It is this monoubiquitination event that leads to proficient DNA repair. New research has shown, however, that non-canonical (atypical) functions of FA proteins also exist, such as their role in regulating mitochondrial function.

At the 2019 FA Scientific Symposium in Chicago, Ill., investigators shared updates from their current research on the FA pathway and revealed advancements that are reshaping our understanding of the complexity of both canonical and non-canonical functions of the FA genes. These newly identified complexities of the FA pathway demonstrate that understanding how FA proteins function in cells during both DNA repair and other processes may

reveal alternative mechanisms to treat and prevent the complications of FA.

Research presented by Dr. Alexandra Sobeck from the University of Minnesota showed a new mechanism of how FANCD2 works during DNA repair. For example, Dr. Sobeck showed, for the first time, that the ATRX protein is required to work with FANCD2 during certain types of DNA repair, even though it is not an FA gene. Similarly to Dr. Sobeck’s work, Dr. Di Yang from the lab of Dr. Martin Cohn at Oxford, revealed that the new protein, WRNIP1, binds to FANCD2, and is also involved with DNA repair.

Dr. John Tainer from the University of Texas MD Anderson Cancer Center addressed the question of whether specific FA defects could be mitigated by targeting proteins that work upstream (before the FA proteins perform DNA repair functions) during DNA repair. Once such protein, EXO5, works to initiate DNA repair prior to the functions of the FANCA core complex. Dr. Tainer’s work showed that blocking EXO5 led to improved DNA repair in the absence of the FANCA protein. FANCA is mutated in approximately 65% of FA patients; therefore, these results mean that inhibiting EXO5 in people with FANCA mutations might be a plausible way to treat complications of FA. Dr. Tainer also highlighted that EXO5 expression correlates with worse cancer prognosis

and patient survival for cancers in the general population. Dr. Tainer’s group is interested in developing small molecule drugs to target EXO5.

Research on the non-canonical roles of FA proteins was also discussed at the Symposium. Dr. Elizabeth Sierra Potchanant from Indiana University highlighted her work on the role of FANCC in regulating the cell cycle. Her research showed that FANCC regulated cell division by binding to a protein called CDK1, a master regulator of cell division. Other work has shown that this specific binding domain (place where other proteins can bind) on FANCC is often mutated in cancers in the non-FA population, which highlights the importance of this interaction.

Dr. Valeria Naim from the University Paris-Sud, Gustave Roussy and Dr. Jessica Luzwick from the University of Texas MD Anderson Cancer Center discussed additional non-canonical roles. Dr. Naim described a new role for FANCD2 that links DNA fragility during replication to mitochondrial function. Mitochondria are organelles (tiny cellular structures) found in cells that are responsible for energy production. Dr. Luzwick discussed the knowledge that many FANCC proteins localize to the mitochondria, but that their roles were not clearly defined. Research she presented showed that FANCO/RAD51C and other FANCC proteins are required for protecting the mitochondrial DNA replication process from oxidative DNA damage (damage caused by reactive oxygen species produced from normal body processes such as metabolism), thereby preventing inflammation.

“ Knowledge of the mechanisms of how other proteins interact and function in concert with FA proteins could reveal new ways to target the pathway for therapeutic effects. ”

Research presented during the FA Pathway session at the 2019 Symposium highlights that the FA pathway is highly complex and involves proteins that are not considered FA genes. Knowledge of the mechanisms of how other proteins interact and function in concert with FA proteins could reveal new ways to target the pathway for therapeutic effects. Additional research highlighted at the Symposium also revealed non-canonical roles of FA proteins in regulating cellular processes such as cell division and in maintaining mitochondrial function. Future research is needed to develop a deeper understanding of the non-canonical functions of FA proteins. ■

Glossary of scientific vocab:

Canonical—typical and well characterized function

Non-canonical—atypical or different from the well characterized function

Monoubiquitination—addition of a ubiquitin protein to another protein to change the function of that protein

Mitochondria—organelle in all cells that regulates energy production

Mitochondrial DNA—DNA that is only located in mitochondria and is different than nuclear DNA

Nuclear DNA—DNA in the nucleus of all cells

Cell cycle—process that occurs in an ordered sequence of events as a cell prepares for cell division

Oxidative damage—damage to cells caused by reactive oxygen species to molecules such as DNA, RNA and proteins

Reactive oxygen species—an oxygen containing molecule that reacts with other molecules in a cell and can cause damage. They are generated constantly by mitochondria.

Protein complex—a group of proteins that bind together to form a larger complex to perform a specific function that can only be done when they are in a complex

Binding domain—a specific part of a protein that is able to bind to another protein

Upstream—in a cascade of biological events, an event that is upstream of another means that it occurs prior to the other event

Advancements in Aldehyde Research

Aldehydes, including acetaldehyde and formaldehyde, can cause DNA damage to hematopoietic stem cells, leading to bone marrow failure and cancer in people with Fanconi anemia (FA). Exposure to aldehydes is inevitable because our own bodies produce them *and* we encounter them in everyday life through automobile and cooking fumes, as well as dietary sources like alcoholic beverages.

There are currently no available tests to measure aldehydes in people. Such a test would be useful because it would provide an indication of the aldehyde load of people with FA and aid in our understanding of the relationship between aldehyde exposure and FA.

At the 2019 Symposium, Mr. Hyun Shin Park, a graduate student from the laboratory of Dr. Eric Kool from Stanford University, presented on a new way to measure intracellular aldehyde levels in human blood by analyzing leukocytes (blood cells that fight infection) before and after the ingestion of alcohol. Mr. Park used a technology called the DarkZone probe and a laboratory technique called flow cytometry to measure intracellular aldehydes. He found that the technique is reproducible and precise and suggests that intracellular aldehyde concentrations in leukocytes are substantially higher than previous estimates of whole blood aldehyde levels.

More research will be required to determine whether this technology can be used for people with FA, and, importantly, whether knowledge of intracellular aldehyde concentrations can help unlock the mechanisms of bone marrow failure or cancer development in people with FA. ■

Clinical Trial Updates: Bone Marrow Failure

Pilot Study on Metformin

A National Institutes of Health grant on Fanconi anemia (FA) led by Dr. Markus Grompe at Oregon Health & Science University identified that metformin (a drug approved by the FDA to treat type II diabetes) demonstrated improved blood counts and delayed tumor formation in FA mouse models, and also decreased chromosomal breakage in cells from people with FA. Additional research has shown that non-FA patients taking metformin also have a decreased incidence of cancer.

These previous studies led to the initiation of a clinical trial to study metformin for Fanconi anemia in April of 2018 at Boston Children's Hospital. People with FA receive metformin orally for six months followed by a one-month follow-up period. The clinical trial studies the effects of metformin on blood counts, the safety and tolerability of the medication, and the effects on DNA damage pathways and bone marrow health.

Dr. Akiko Shimamura, one of the investigators, presented an update on the study at the 2019 FARF Symposium. So far, the study has enrolled 14 patients, and 13 were eligible to start study treatment. Six patients have completed study treatment and seven patients are currently still on metformin.

With small numbers of patients and short follow-up, the study most commonly observed an increase in neutrophil counts. Trends suggest an increase in the colony-forming capability of hematopoietic cells and a decrease in chromosomal breakage along with reduction in DNA adducts (segments of DNA bound to a cancer-causing chemical). The trial aims to recruit 10 additional participants. Formal analysis of the study results will be performed once the trial is complete.

Quercetin Phase I Trial to Reduce Oxidative Stress in Children with FA

Dr. Parinda Mehta presented results on an FDA-funded phase I study focused on the effects of quercetin (an antioxidant found in many foods) on reactive oxygen species (ROS) and hematopoietic stem cells that is currently being conducted at Cincinnati Children's Hospital. Six patients with FA (median age six years) received quercetin at an optimized weight-adjusted dose (max dose of 4000 mg/day). All patients completed four months of quercetin treatment and five out of six patients received quercetin up to two years on the continuation phase of the study. ROS levels in peripheral blood showed sustained reduction after two years of quercetin treatment compared to baseline pre-treatment levels. ROS levels also decreased in bone marrow and stem cells at two years. Bone marrow colony formation increased at two years compared to baseline and the total number of stem cells also increased. These results supported enrollment of an expansion cohort of an additional 15 patients who are receiving optimal dose quercetin. The study results thus far indicate that quercetin therapy leads to a sustained reduction in oxidative stress in children with FA after two years of treatment, along with continued improvement in marrow function.

Haploidentical transplantation with Post-Transplant Cyclophosphamide (Haplo-PTCg)

Haploidentical transplants, or allogeneic transplants that rely on half-matched donors, are sometimes necessary in countries with limited resources. Dr. Carmen Bonfim from the Federal University of Paraná in Curitiba, Brazil presented on using haploidentical transplantation with post-transplant cyclophosphamide (Haplo-PTCg) in 32 patients with Fanconi anemia (FA) between 2008 and 2019. The median age of the patients was 10 years and 72% were male. For 49 patients, it was their first transplant, and it was the second or third for four of the patients. Three patients rejected the first transplant and received a second one using different family donors. Forty-four patients were in aplastic phase and nine patients had

continued on page 31



Scientist Spotlight

Name: Grant Rowe, MD, PhD

Institution: Boston Children's Hospital

Area of expertise: Stem cells

My work: I am interested in developing new models of FA using human stem cells. I try to make FA blood stem cells in a culture dish to understand the mechanisms of how they fail prematurely. If we can find the genes that drive this process, we can hypothesize how to target or repair them for new treatments. Recently, we have also become interested in how FA blood stem cells transform into cancerous blood stem cells. Understanding how this occurs and revealing the genetic mutations required for this process could impact how we monitor FA patients and treat pre-leukemia and leukemia.

What motivates me to work on FA: As a pediatric hematologist and stem cell transplant specialist, I understand that FA is a very unique and challenging problem. The only curative therapy for the bone marrow failure and leukemia of FA is blood stem cell transplantation. Because the defect in DNA repair that causes Fanconi anemia sensitizes patients to chemotherapy and radiation, stem cell transplantation can be very challenging for FA patients, especially if leukemia is present. FA patients can be especially prone to adverse post-transplant complications. New treatments that boost

blood counts and improve the efficacy and safety of stem cell transplantation are needed. The development of a renewable, robust, human based model system of FA will reduce reliance on precious patient samples as we pursue these aims. Such a model system also has the potential to serve as a platform to better understand and improve stem cell transplantation.

Meeting families affected by FA in my practice and at the annual FARF meeting continues to attach personal stories to these clinical and research challenges. These experiences make the goal of contributing even a small incremental advance in our understanding of FA extremely gratifying.

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

Either practicing with my swim team or relaxing at home.

Anything else you want FA families to know?

Please continue your outreach, advocacy, and engagement with clinical and basic researchers. The FA community is like no other, where families and researchers act as one entity to collaboratively work to achieve better lives for FA patients. ■

THE AMY WINN AND CHRISTOPHER T. BYRD
**AWARD FOR ADULTS
WITH FANCONI ANEMIA**

NOW ACCEPTING APPLICATIONS | DEADLINE 7.12.2020

The advertisement features a blue border and includes portraits of Amy Winn on the left and Christopher T. Byrd on the right. The text is centered and uses a mix of bold and regular fonts.



A Day at the 2019 FAdult Meeting

Multiple sessions on wellbeing & integrative health

One of the important aspects of planning a meeting for adults with FA is making sure the information is relevant to the issues adults are facing now. That's why there was a session about integrative health, to discuss issues around the connection of our minds, bodies, and the everyday steps that can be taken to improve wellbeing. In addition, we held sessions on navigating insurance issues, fertility, a panel with the lead doctors from FA transplant centers, and even a cooking demo with audience participation.

A space for all the feelings

If you've ever been to a FARF meeting, you know that support sessions are always a part of the agenda. This is a place to process and discuss shared experiences, questions, and information that might be helpful for one another. There are groups just for adults with FA, just for parents/caregivers, just for spouses, and for the whole group together.

Time to attend the Scientific Symposium

The meeting for adults takes place the same weekend as the annual FA Scientific Symposium. This allows attendees at the adult meeting to sit in on scientific sessions, go to poster receptions, connect with FA researchers, and participate in the Symposium banquet dinner. Researchers rarely meet people living with the disease they are working on, and how often do you get to be surrounded by dozens of FA experts at once, ready to talk to you?

Downtime to spend together

Some of the most valuable experiences at the adult meeting occur outside of organized sessions. Whether this is time spent in quiet areas around the hotel, or exploring the city, there are many opportunities to connect in authentic and laid-back ways. ■

Each year, the number of adults with Fanconi anemia grows. As this population grows, so too does the annual Meeting for Adults with FA. In 2019, 57 adults and 66 guests attended the three-day meeting in Chicago. The FAdult Council, a committee comprised solely of adults with FA, helped to plan a meaningful meeting that responded to the needs of this population. That's why the 2019 meeting focused on opportunities for connection, learning in a relaxed (and even fun) way, time dedicated to simply talking and listening to others, chances to meet with FA experts, and a dedicated space for attendees and their guests to decompress, relax, play games, or just hang out together. We look forward to welcoming returning and new adults at the 2020 meeting in Austin (September 16-20). Here are some highlights from the last meeting to give you a small glimpse of what it's like.

Wall-to-Wall

To kick things off, the group played the Wall-to-Wall game, answering questions like:

- Are you from the US or abroad?
- Is this your first meeting?
- Do you have FA or love someone with FA?

As they moved through the questions, adults with FA and their partners, parents, care takers, and kids moved from wall to wall, to represent the shared experiences within the community. While experiences vary, there's a beautiful shared connection in identifying a room full of people who GET IT.

FA Jeopardy

Dr. Stella Davies (Cincinnati Children's Hospital) led a fun, informational game

of "Fanconi Anemia Jeopardy", where audience members got to be contestants and show what they know about FA, FARF, and even some Chicago trivia. You can watch the entire game, including the Q&A at the end, on FARF's website and YouTube channel.

Free screening for lesions

Ralf Dietrich & Christine Krieg (German FA Family Support Group) were there throughout the meeting to give screenings for oral lesions. This is part of their research study, "Reducing the Burden of Squamous Cell Carcinoma in Fanconi Anemia" (see more on page 4). It's easy to be part of this study since the German group is always at the meetings, AND there's always chocolate at the end ☺.



Words of encouragement for everyone transitioning from pediatric to adult care



By Mary-Beth Johnson

Earlier this year, as I was coming back from a bone marrow biopsy, I had a lot of thoughts about folks with Fanconi anemia (FA) transitioning from pediatric to adult care. Before I get into that though, I want to acknowledge that for a lot of our population, there will always be a need for long-term or extended care takers. To those in that space: I see you, and I don't want to erase your narrative. Caretakers are warriors in their own right, and they're really often overlooked in our communities. If you're doing the advocating and care follow-up/decisions—THANK YOU. This level of love is special. You deserve to be seen.

For those patients (and parents!) in peds or reaching a transition, here's a little encouragement:

01

Transitioning to adult care with a rare disease like FA is a GIFT. Not everyone with our diagnosis gets to adulthood; enjoy this rite of passage. Push the boundaries of medical providers' knowledge by taking up ALL the space you can in adult care. As you transition, have adult care providers consult with your peds team to fill in any gaps or questions about your care.

02

It's REALLY empowering to know your health really well. Learn early to understand your CBC. Ask the ears off your pediatrician so you know what each of your tests/procedures are before you reach adult care.

03

Make sure your adult care doctors are humble enough to know you are the specialist on your body, and your specific case. You're going to have to self-advocate A LOT. Fire the doctors who don't listen to you/dismiss you for the one slide in medical school they got on your disease, and linger with the ones who let you be your own quarterback.

04

Go ahead and make sure you have a mental health specialist as part of your care team. You're not alone in your anxiety, and having a dedicated safe space to have unfiltered thoughts is REALLY helpful. You owe your mind the same care as your body.

05

Ask for the support you need in the transition. If your medical institution has a patient coordinator, ask for one. Have them help you set up appointments. Get all the phone apps to communicate with your care team. Have your parent come to your first few appointments. Interrupt your doctors when you don't understand. You're allowed to need help as you transition.

06

TRANSITION. FA is a **FORMER** childhood disease. There aren't enough adult care doctors who are used to seeing us. The more of us they see and get familiar with, the more specialized our care becomes as adults with FA.

I'm always cheering for every single one of us, and sending love to you as you navigate life (a full and beautiful one, I hope) alongside this diagnosis. ■



Podcasts to Explore

Did you know there are podcasts out there focused on rare diseases? There's even one specific to Fanconi anemia, hosted by Danish adult with FA, Daniel Kold. We've pulled together a list of some podcasts you may want to check out. Let us know if you have any other suggestions! You can email jordan@fanconi.org. Search online for links, or visit www.fanconi.org for a list of direct links.

FA Specific

Life, Death, & Happiness Podcast

Caregiving

Real Men Podcast - Jacks Caregiving Coalition
Happy, Healthy, Caregiver Podcast

Business, Science, Policy

Rare Perspective Podcast
Rare Disease, Cell & Gene Therapy Weekly Roundup
AI in Drug Discovery
An Arm & a Leg Podcast
Healthcare Policy Podcast

Biopsychosocial

Diverse Perspectives on Health & Illness Podcast
RARE Cast
Rareshare
Openly Rare
Beyond Your DX
Ten Percent Happier
Meditation for Fitness Peeps
Sickboy
Misguided Notions Podcast
Disarming Disability

Patient Stories

Rare in Common

Psychosocial (Kids)

Be Calm Ahway Island Bedtime Stories
Peace Out

AIM Strong: A Resource for Families Going Through Bone Marrow Transplant



By Mary Jo Becerra

Our sunflower story started with our son, Israel. When he was in the hospital after his bone marrow transplant (BMT) in 2015, he told me he wanted to plant sunflowers. He died before we were able to honor his wish. Not long after he died, I became aware of the saying 'I want to be like a sunflower so that on the darkest days, I can stand tall and find the sunlight.' That phrase, 'Find the Sunlight,' resonated with me in so many ways, not just in the context of Fanconi anemia and bone marrow transplant, but in life in general. We all have dark days, and yet, if we can be like the sunflower, we CAN find the sunlight, even if it's just for a moment. In doing so, we can make a difference for ourselves, our family and the people around us.

What is AIM Strong?

This phrase, "find the sunlight," inspired our family to help others, so we founded AIM Strong, Inc. The mission of AIM Strong is to help support and encourage families who have a child undergoing bone marrow transplant. The sunflower in the AIM Strong logo is for our son Israel. We



University of Minnesota Masonic Children's Hospital delivery



Methodist Children's Hospital delivery

provide gift cards to offset the expenses of relocation and extended hospital stay required for bone marrow transplant. We made our first delivery to the University of Minnesota Masonic Children's Hospital in June 2017 when we were in the city for our daughter Mariana's six-month post-transplant appointments.

How can families find support through AIM Strong?

At this point, we are able to provide assistance to families who have a child undergoing bone marrow transplant in the United States. We have a brief application for families to complete that helps us determine how best to help the family. We are currently partnered with the following facilities: The University of Minnesota Masonic Children's Hospital in Minneapolis, Cincinnati Children's Hospital, Seattle Children's Hospital (Seattle Cancer Care Alliance), Texas Children's Hospital in Houston, Children's Medical Center in Dallas, Methodist Children's Hospital in San Antonio, and The Children's Hospital at OU Medical Center in Oklahoma City. We have also helped families with a child undergoing bone marrow transplant at Memorial Sloan Kettering in New York. Families

with a child having their transplant at these facilities can request our application from their social worker. If a child has their transplant at a different facility, then the family must ask their social worker to reach out to AIM Strong, Inc. on their behalf. This must be done due to HIPPA requirements. Social workers can email me for information at maryjo@aimstronginc.org.

As of February 2020, we have helped over 450 bone marrow transplant families.

Our biggest project is spreading the word about AIM Strong, Inc. and raising money to fund our mission. That's why I started the 'Find the Sunlight' campaign. I encourage people to purchase our merchandise and to post and share where they find the sunlight. Your messages and stories are so encouraging to other families, and to our community as a whole.

Please visit our website (aimstronginc.org) or follow us on Facebook or Instagram (@aimstronginc). To request an AIM Strong, Inc. decal, email me at maryjo@aimstronginc.org.

We look forward to helping you stand tall and find the sunlight. ■





Intoxicating Gratitude

By Egil Dennerline

Thirty years ago, I was told that I had an incurable terminal illness called Fanconi anemia (FA). My father was alone when he received the diagnosis and was told not to tell the nurses, as the news would be too devastating for them. This effectively communicated the gravity of the situation while at the same time leaving my father to navigate not only his own feelings in what could only be described as a serious family crisis, but also to protect the feelings of those in charge of his son's treatment.

Growing up, the FA diagnosis seemed like a death sentence. As a teenager, I was anti-social, seemingly isolated in my corpus shell and was in no way ready to accept the weighty facts or face the tough decisions ahead. I stonewalled the whole thing. On the one hand, I was going to live my life to its fullest and continue to move forward, unabated. On the other hand, I was keenly aware that I may not be moving forward for very long and the concept of a future seemed abstract. Back then, my father was told I probably wouldn't live into my 20s.

Traumatic experiences have a way of staying crisp in one's mind, so although

this initial diagnosis now seems like a lifetime ago, I remember it well: the steady stream of blood tests, the bone marrow biopsies, and the tension at home surrounding these uncontrollable events. And yet, I also look at it all with a sense of overwhelming gratitude. I'm grateful for the passionate friendships and powerful experiences I have been afforded in all the extra time I didn't think I had. What we thought would be four years has turned into 30. The irony of how a life-threatening illness can result in strong gratitude is intoxicating in its own funny sort of way. There is so much to be thankful for.

I am thankful for all the learning opportunities I've had, be it through a curiosity about the world, through the throes of life in general, or through the people I've met on my path. Creativity and storytelling are paramount in my world, as a means of underscoring and emphasizing the beauty I believe to be all around us – in good times as well as in the bad – and to express this gratitude. As such, when I was diagnosed with tongue cancer eight years ago and my best friend, keyboard wizard Palle Hjorth, told me I had to survive because we were going to make

an album together, it was something I really held on to. It took seven years to finish, as I battled several additional cancers as well as a host of infections and other medical issues, but we did it. The band is called "Wall to Wall" and our first album, released about a year ago, was appropriately called "Waiting".

Despite all of the infections and viruses I've endured this past fall and winter, I deeply appreciate this life and everyone in it. So, whether it is a whimsical exchange of words or a life-long conversation, thank you for being there for me, and may 2020 be a year of high notes for you all! ■



Egil with his wife and their daughter

A dedicated space for caregivers

In the 30 years that this newsletter has been published, it has featured articles, stories, and updates written for and by a variety of stakeholders in the FA community. In the days before the Internet, the newsletter was the main forum for families to learn about progress in research and how their fundraising dollars were used to advance research and support families. The FA community expanded, and the newsletter became a space for FA family members to share their stories, admit their fears, and express their hopes for the future. As children with FA grew into adults, this newsletter grew to include the unique experiences of adults with FA.

One group of people we often think about but don't often directly acknowledge is caregivers. All of you partners, parents, siblings, grandparents, friends or family members, we want you to know how vital you are to this community. We want to make sure you have a space in this newsletter specifically for you, by you. To accomplish this, we asked our friend Allison Breininger to help us. Allison is mom to Maya and wife to Sean, who lives with FA. She is an accomplished writer who combines her creative talent with her lived experience as a caregiver to create a blog dedicated to others in what she calls "The Negative Space." This is the space around or in between the subject that is often overlooked but is vital to the whole picture, like how a caregiver is vital to the livelihood of a person with FA. To read more of Allison's work, including popular essays like "The Job Description of a Caregiver" and "What Your Struggling Friend Really Wants," visit www.thenegativespace.life.



Maya, Sean, and Allison

Written by Allison Breining

Dear Caregiver,

This is for you.

I know that each time this newsletter arrives, you pore over its contents, hoping that this will be the issue where you'll read about an advancement, a new drug, a potential cure, *something* that will change the statistics you've read, studied, had nightmares about.

I know that you take notes from these newsletters on the featured fundraisers, jotting down tips and tricks so that your next event can funnel even more money towards some answers, some hope.

I know you look at the FA adults who are highlighted here, taking in their ages and their accomplishments, and allowing their longevity to bring with it the fresh air of possibility.

I know you grieve at the "In Loving Memory" section, looking carefully at the birth year of each person lost, comparing it to that of the person you love. I know you mourn the names listed that you had been lucky to befriend and that you have moments of wondering if you have it in you to continue to make new friends that you may very well lose. I know you do all you can not to imagine your person's name, written in that font, in that box.

How can I claim to know these things? My husband, Sean, was diagnosed with Fanconi anemia nine years ago at the age of 32. Since then he has undergone a bone marrow transplant and has been diagnosed with and treated for cancer of the tongue, throat, gums, bladder, lips, and skin. I have been by his side for every appointment and procedure. I have read the research, done the fundraising, gone to the meetings, attended funerals for friends from this community. Like you, I am an FA caregiver.

Because I am, I know that just like your life, this newsletter has been dedicated to the person in your world with Fanconi anemia. But, dear caregiver, I'm delighted to tell you that that is about to change.

The Fanconi Anemia Research Fund knows that alongside every person diagnosed with FA is a caregiver. Whether that title resonates with you or not, if you are reading this newsletter because a person you love has FA, we are talking to you. When describing your role, you likely say, "I just do what any other parent/partner/friend would do..." but let me tell you that I am well aware of what it is you do and there is no "just" about it.

Caregiver, we want you to know that you are seen. We see you at the family and adult meetings taking notes, asking questions, cornering the researchers in the breakfast line. We see the coordination and preparation you take on to get your loved one to and through those meetings. We see you posting questions on the family Facebook page late at night from hospital rooms. We see you rallying around your fellow caregivers at transplant centers, Ronald McDonald Houses, and online, using energy you don't have to hold up others you've barely met but with whom your connection is so powerful. We see you counting everything from platelet levels to days post-transplant to everyday blessings.

Caregiver, we see you, we honor you, we are here to support you.

Moving forward, we will have a space in every newsletter dedicated to caregivers. We are working on ways to honor, include, and support you through the work of the Fanconi Anemia Research Fund all year long.

We realize that so many of the services, supports, and appointments you spend your days on are aimed at the needs of your loved one. But we also see that you, dear caregiver, are living through and with the effects of this disease as well. We want you to have a space that's dedicated to you and to the reality that is being a caregiver for a person with Fanconi anemia.

Caregivers, this is for you. ■

FUNDRAISING SHOUT-OUTS



Grand Canyon Hike for FA

A first-time fundraiser, Wendy Vitiritto decided to turn her annual rim to rim hike of the Grand Canyon into a fundraiser in honor of her son Vinny, an adult with FA. Wendy, her husband Joe, and their oldest son's girlfriend hiked all 25 miles of the Grand Canyon in one day and raised over \$11,000! Incredible job, Vitiritto family! Thank you for reminding us that sometimes the best fundraisers are born from an activity we're already doing.



Running for a Cure

This past November, Julie Williams took on the challenge of running 100 miles on behalf of her 13-year-old daughter, Claudia. On race day, Julie pushed through a knee injury and completed 42 miles before the medical staff decided it was not safe for her to finish the course. Though the race did not go as planned, Julie raised over \$5,000 during her first fundraising campaign! Thank you, Julie – you are truly an inspiration!



Benefit Concert

After their first successful holiday appeal in 2018, the Starnar family decided it was time to try out a fundraising event. In October 2019, the Starnars hosted a benefit concert at their church that featured a local quartet and the inaugural performance of their brand-new church organ. The event raised more than \$7,000 for FA research and support services. Way to go, Starnar family! Starting with an appeal then trying out an event is a great way to take your fundraising to the next level.



Play for FA Golf Tournament

Play for FA does it again! This fall, the Vandermeys family of Moseley, VA tried out a new event and hosted the first ever Play for FA Golf Tournament. The event was a hole-in-one and raised over \$17,000 – incredible! Thank you, Cynthia and Gerard, for continually raising the bar for fundraising, and making the FARF mission a cornerstone of your community.



Olivia's Fight for A Cure

They first started with an online fundraising appeal, and then the Eubank family of West Virginia held their first fundraising event in 2018. In 2019, they hosted their second Olivia's Fight for a Cure fundraiser. The winery event featured a live auction and raised over \$6,000! Way to step it up, Eubank family!



Pell Bridge Run

FARF fundraising veterans, the Fiaschetti family, participated in the annual Pell Bridge Run, a four-mile run/walk over the historic Rhode Island Pell Bridge. Once again, the Fiaschetti family teamed up with Dr. Niall Howlett and lab members from the University of Rhode Island to raise almost \$8,000! Thank you, David, Mary Ann, Dr. Howlett, and the rest of the Pell Bridge Run FA team.



One Day for FA

The Mingo-Ritchie and Branov families of Vancouver, Canada, hosted their first ever fundraising gala this year. One Day for FA was a must-attend event! The Roaring 20s themed party featured everything from live music to an auction, dancing, food, and drinks, and raised over \$60,000! Thank you, Mingo-Ritchie and Branov families! By bringing your two families and communities together, you were able to make an even greater impact.

#GIVING
TUESDAY

2019 Giving Tuesday

In 2019, many of you showed up and gave FARF its best Giving Tuesday ever. Thanks to a generous matching donor, you raised over \$59,000 on Giving Tuesday alone! Thank you to everyone who shared our Facebook posts, sent emails and texts to their friends, and created a personal fundraising page online. Year over year, you've shown that when we all commit to a single day to make an impact, we get it done.

UPDATE FROM HEADQUARTERS



By Mark Quinlan, Executive Director

As FA research advances and our community of FA families grows, our roles in the FARM office also evolve. It's our job to make sure we are pushing our mission forward in the most effective ways possible. To that end, I am excited to report that since the last newsletter, two new staff members have joined the FARM team. In November, Jordan Deines assumed the role of Family Services Director, and Alexander LaVake filled the newly created position of Research Program Manager.

Jordan has a master's degree in social work and a strong passion for helping others, which led her to work with trauma survivors in previous roles. Since her arrival, she has impressed me with her desire to work in partnership with the FA community. Her ability to connect with and empower families during their challenging times is one of the qualities that brings tremendous value to the support services we offer.

Alexander comes to us with a master's degree in public health and extensive experience with grant management in health education settings. This position of Research Program Manager is a new role that will strengthen the research pillar of our organization by providing support to existing staff and to the FA research community. Alexander is working to improve our grant management process and our communication strategies with the research community.

Earlier this year, in February, Alexander and Jordan joined other staff members in Orlando to meet with members of the FARM Board of Directors, the FAdult Council, and the FARM Scientific Advisory Board at our annual gathering of the organization's leadership. This two-day event is a time for presentations, meetings, and opportunities for collaboration. The groups meet separately and together, and by the end, we produce a set of deliverables that include defining priorities and recommendations that will drive our work over the next year.

What impresses me most about this gathering is the energy and dedication of all attendees to brainstorm ideas and deliberate opportunities that will directly impact individuals living with FA. It humbles me that I can facilitate these conversations each winter, and I am filled with hope as I look back year over year at the accomplishments that

have come from ideas developed at these meetings.

At this year's meeting, we discussed improvements to our grants process that will allow us to expand our reach within the FA research community. We strategized how to target specific gaps in FA care, like how to address gynecological cancers. We also identified action steps to strengthen existing programs like our virtual tumor board and clinical registry.

Additionally, we strategized new ways to advance FA research and improve clinical care for people with FA. We believe an effective way to move the field forward would be to develop a consortium for targeting head and neck cancer. The idea behind this consortium is to create – for the first time ever – a collaborative group of cancer experts who will develop preclinical experimental models and a coordinated, centralized system to make clinical data (like tissue samples) accessible to the research community. Resources developed in the consortium would then be used to foster large, multi-institutional research grants that will be led by multi-disciplinary experts who are focused on FA cancer. Collaborative projects that evolve from a wide range of expertise have the capacity to identify therapies faster.

The expertise, knowledge, and passion that is present in these meetings is a result of co-founders Dave and Lynn Frohnmayer's idea, so many years ago, to get people together in a room and talk about the issues this disease presents. It is now incumbent on the FARM staff to take these ideas, fine-tune them, and drive them forward to meet our mission. We embrace this challenge because we know the outcomes will improve the lives of individuals with FA and their families. ■

JACK TIMPERLEY

receives the 2019

Amy Winn & Christopher T. Byrd Award for Adults with Fanconi Anemia



Jack Timperley, 20, lives in Park Ridge, Illinois, where he is a student of philosophy and business administration at Northeastern Illinois University. Before pursuing this degree, Jack attended Oakton Community College, where he was a proven leader in his community, earning himself a Gigi Campbell Student Trustee Excellence Award from the Illinois Community College Trustees Association.

As a member of the Fanconi Anemia Adult Council, Jack serves as a voice for other adults and teens living with FA. He works with the FARF staff and board of directors as an advisor and advocate for the needs of the adult FA population. In this role, he has spoken at several fundraisers all over the country, telling his story and expressing to supporters just how crucial their role is to advancing research.

At the 2019 Fanconi Anemia Research Fund Scientific Symposium, Jack gave

the keynote address to researchers and clinicians entitled “What is your why?” He inspired the audience to find the courage and drive to make a difference, as Amy and Chris did. He reminded those in attendance of the value of connection and the importance of perseverance in the face of adversity.

After he graduates, Jack hopes to pursue a PhD in a philosophy-related discipline and eventually start his own research and technology company. He’s also interested in one day teaching philosophy at the college level. In addition to these career aspirations, Jack hopes to

continue honing his public speaking skills. “I want to talk to people about how to live a life of happiness and of joy. I was born with FA and I’ve had an extensive medical history. But despite all that, I continue to thrive and continue to live life to the fullest every day. I know it sounds cheesy, but it’s true. And my public speaking is related to that. Our lives are so precious. To recognize our own mortality might motivate us to live life and do everything we can today because we may not see tomorrow.”

Watch Jack’s acceptance speech at www.fanconi.org. ■

“ Our lives are so precious. To recognize our own mortality might motivate us to live life and do everything we can today because we may not see tomorrow. ”

ONE MONTH. ONE CAUSE. It's time to make an impact.

**Last year, FA families raised more than \$75,000 for FA Day.
How much will we raise this year?**

Eleven years ago, Peg Padden, FA parent and enthusiastic fundraiser, encouraged FA families to start International Fanconi Anemia Day to raise funds for research and family support services. We know that longer and better lives for people with FA are possible when we all get involved.

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

It starts with you. Whether it's \$50 or \$5,000, the money you raise or give goes to fund our incredible FA researchers, bring our FA families together, and keep our mission moving forward. We celebrate FA Day all throughout May. You can add your page to the campaign website in just a few clicks. We'll have a blurb about the cause pre-loaded for you. Then, you just share your page with your community. We can help you share your story and reach your fundraising goal!

Visit <http://fundraise.fanconi.org/faday2020> to get started today.

#THISISHOWIFA
FIND ANSWERS. FIGHT ALWAYS. FANCONI ANEMIA.

We're stronger together. That's why when you sign up to fundraise, you'll join your regional team.

Step 1: How much do you want to raise?
Step 2: Click "fundraise" and join your regional team!

<p>\$150</p> <p>Supporter \$150</p> <p>A great place to start if you've never fundraised. Ask ten friends to donate \$15! Or ask three family members to give \$50.</p> <p>FUNDRAISE</p>	<p>\$250</p> <p>Fan \$250</p> <p>New to the fundraising game? Ask ten friends to give \$25!</p> <p>FUNDRAISE</p>	<p>\$500</p> <p>Friend \$500</p> <p>Ready to level up? Ask 10 friends for \$50, or ask five family members for \$100.</p> <p>FUNDRAISE</p>	<p>\$1000</p> <p>Leader \$1,000</p> <p>Feeling ready to make a larger impact? Ask 50 friends/family for \$50, or ask 10 close friends for \$100!</p> <p>FUNDRAISE</p>
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Everyone raising funds in your area counts toward your region's overall goal.

West : Washington, Oregon, Idaho, California, Montana, Wyoming, Alaska, Hawaii

Southwest : Texas, Oklahoma, New Mexico, Arizona, Colorado, Utah

Midwest : Ohio, Indiana, Michigan, Illinois, Missouri, Wisconsin, Minnesota, Iowa, Kansas, Nebraska, South Dakota, North Dakota

Southeast : West Virginia, Virginia, Kentucky, Tennessee, North Carolina, South Carolina, Georgia, Alabama, Mississippi, Arkansas, Louisiana, Florida

Northeast : Maine, Massachusetts, Rhode Island, Connecticut, New Hampshire, Vermont, New York, Pennsylvania, New Jersey, Delaware, Maryland

International : all other countries



Which region will raise the most?



2020 Board of Directors (from left to right): Jasmine Bennetsen, Rachel Altmann, Tracy Strimling, Lisa Mingo, Orion Marx, Lynn Frohnmayer, Andre Hessels, Stella Davies, John Connelly, Aileen Carlos.

Welcome, new board members, and thank you, outgoing members

The FA Research Fund is able to thrive thanks in large part to the committed volunteers who serve on the board of directors. This winter, we thanked three outgoing board members: Sharon Schuman, Bill McCOREY and Mark Pearl, and welcomed two new ones: Jasmine Bennetsen and Tracy Strimling.

Thank you, Sharon, Bill & Mark

Sharon Schuman joined the board in 2014, though her service to the organization began many years earlier. A longtime friend to Amy Frohnmayer Winn and the entire Frohnmayer family, Sharon began supporting FARF more than 20 years ago by organizing an annual chamber music benefit concert and auction. She increased her fundraising efforts as a board member, co-chairing the Philanthropy Council and helping to build the fundraising team at FARF. She also served as secretary during her tenure on the board. A gifted writer, Sharon wrote and edited several articles for the FARF newsletter and website, including one of the most popular pieces FARF has ever published: "No Longer an Orphan: the Fanconi Anemia Story." We look forward to Sharon's continued presence at scientific conferences and as a fundraiser and friend to the organization.



Sharon Schuman

Another FA friend-turned-board member is Bill McC Corey, who joined the FA community over a decade ago as a fundraiser, then became a board member in 2017. Bill first learned about FA when his friend Kevin McQueen's son was diagnosed in 2001. Bill is an avid mountain climber and began challenging himself and others to complete climbs to support FA research. Several climbs and hundreds of thousands of dollars later, Bill will make his final 'climb' this year, as he steps off the board. In addition to fundraising, Bill helped strengthen the board of directors by serving on the nominating committee.



Bill McC Corey

Mark Pearl has dedicated his time, skills, and fundraising dollars to strengthening this organization since he joined the board of directors in 2003. In that time, Mark was instrumental in seeing FARF through key periods of growth, including helping to build a strong team and FA adult



Mark Pearl

council. Fundraising is a family affair for the Pearls, who have consistently reached out to their community for support, which in turn has benefited the greater FA population. Mark served on the budget and investment committee as well as the nominating and executive committees. He also filled the role of vice president before serving as president of the board until early 2020.

We are indebted to Mark, Bill, and Sharon for their many years of dedication to building a stronger organization at key moments of opportunity and growth. Thank you!

Welcome, Jasmine & Tracy

Jasmine, a 28-year-old native Floridian, was diagnosed with FA as a teenager and received a bone marrow transplant in August 2019. She has been a leader in the FA adult community and currently serves as co-chair of the FAdult Council, an advisory council that advocates for and represents the growing adult FA population.



Jasmine Bennetsen

An avid photographer, Jasmine put her skills to use working for Apple. She travels around the country to speak at FARF events and inspires many with her enthusiasm and authenticity. In her own words, "I am most excited to have the opportunity to impact that patient and family community for the better. I look forward to bringing the patient perspective to the board, and to being someone my fellow adult community can speak through."

Tracy is a longtime FARF supporter, both as a donor and as a volunteer at the annual benefit concert organized by Sharon Schuman. Tracy served the Eugene community by volunteering in several public schools, before going on to teach English at an alternative school for at-risk students.



Tracy Strimling

In addition to serving on FARF's board, Tracy is also on the board of the Oregon Cancer Foundation, an organization that provides financial assistance, support, and education for cancer patients and survivors. Tracy is enthusiastic about supporting FA research and families, and is particularly interested in the connection between FA and cancer. ■

FAMILY FUNDRAISING LIST

From January through December 2019, FA families raised more than \$2,800,000 for the Fanconi Anemia Research Fund! 233 families raised funds, with 140 raising at least \$500. Each dollar donated advances research and family support, making a difference for all those affected by FA and their families. Sincere thanks to every family and individual who worked so hard to raise funds in honor or memory of loved ones.

\$1,450,000+

Lynn Frohnmayer

\$285,000+

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

\$100,000+

Orion and Lisa Marx
Kevin and Lorraine McQueen

\$50,000- \$99,999

Gerard and Cynthia Vandermeys

\$20,000- \$49,999

John and Kim Connelly
John and Martina Hartmann
Stephen and Jennifer Klimkiewicz
Tim and Mary Ann Lana
Todd and Kristin Levine
Neil and Emily Robison

\$10,000- \$19,999

Brian and Carly Adel
Rachel Altmann and Tyler Morrison
Brian Anderson and Sultana Graham
James and Jennifer Armentrout
Herminia Carvalheira
Mauro and Kerri Cazzari
Mark De Groot and Hanneke Takkenberg
David and Mary Ann Fiaschetti
Brian Horrigan and Amy Levine
Charles and Kathleen Hull
Peggy Padden
Rose and David Pennell

Joe and Wendy Vitiritto
Nigel and Ann Walker

\$5,000- \$9,999

Rachael Alaniz and Kevin Gatzlaff
Amanda Barber
David and Sarah Borden
Ryan and Rebecca Brinkmann
Joeseeph and Nancy Chou
Erin Furr
Zach and Rachel Gratz-Lazarus
Stan and Michelle Kalemba
Susan and Skip Longstaff
Daniel and Angie McMahon
Ian and Tricia Mitchell
Caroline Nguyen
Nancy Nunes
The Pearl Family
Peter and Janice Pless
Paul and Rena Rice
Andrea and Robert Sacks
Bradley and Darlene Starner
Julie and Robert Williams

\$2,000-\$4,999

Michael and Jennifer Aggabao
Adam and Marissa Becker
Jennifer Bland
Randolph and Nancy Bloxom
Richard and Tena Bason
Chris and Jennifer Branov
Jerry and Natalie Christensen
Andrew Coons and Valeen Gonzales
Jonathan and Sharon Drew
James and Crystal Eubank
Justin and Britteny Ferrin
Daryn and Carol Franzen
Andrew and Jennifer Gough

Alan and Rachel Grossman
Owen Hall and Margaret Kasting
Andre Hessels and Rutger Boerema
Jeff and Beth Janock
John and Karilyn Kelson
Dan Klug and Elizabeth Bertrand-Klug
Keith and Jessica Loo
Kory and Julie MacMurray
Lisa Mingo and Mark Ritchie
Ronald and Fredi Norris
David and Stacy Ownby
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In Loving Memory

Taonashe Muzanhamo
 9.21.2013 – 12.9.2019

Lindsey Sablosky
 11.23.1983 – 1.10.2020

Sanjeev Singh Parmar
 12.16.1979 – 9.20.2019

Eric Miller
 10.2.1983 – 10.29.2019

Ronda Walters
 7.21.1963 – 2.1.2020

Amanda Dellapenta
 3.4.1985 – 2.24.2020

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



Clinical Trial Updates: Bone Marrow Failure continued from page 10

myelodysplastic syndrome (MDS)/acute myeloid leukemia (AML). All patients had failed androgen therapy and 93% were transfused.

The preparatory regimen consisted of fludarabine plus total body irradiation with or without cyclophosphamide and with or without r-anti-thymocyte globulin (r-ATG). The results from the study show that Haplo-PTCg should be offered as an option for patients with FA who lack a matched related or unrelated donor and have not responded to androgens. The cohort in this study were high-risk patients transplanted in a country without access to expensive and sophisticated T-cell depletion techniques. Despite these limitations, almost 80% of the patients who received ATG in the preparatory regimen are alive. The incidence of graft-versus-host disease (GvHD) is still high and identifying better techniques to prevent and treat GvHD are urgently needed in order to improve the quality of life of surviving patients. ■

Use of Logo

A reminder to our families: 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.

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

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