FAmily Newsletter

Spring 2021 A semi-annual publication of the Fanconi Anemia Research Fund

issue 69

In this issue

Macy, age 10, Michigan

Update from HQ

The last year has forced us all to reexamine our ways of working, our lifestyles, our priorities and strategies. The pandemic required the Fanconi Anemia Research Fund (FARF) to collaborate and deliver service to the Fanconi anemia (FA) community in new and different ways. The past year shifted our perspective and challenged our old systems. From this uncertainty, new ideas emerged and we discarded some that no longer fit our mission. We are proud of our advancements during these trying times and look ahead with confidence and optimism.

First, we needed to shift to virtual events. If you've ever been to the FA Family Meeting or the Meeting for Adults with FA, you know that these events are hugely impactful. There's something truly special about coming together with people who get it. It's hard not to be able to gather in person in 2020 and 2021, and we are very excited to resume these events in 2022. However, we have seen some benefits to our shift to virtual events. This format provides greater access to the wider FA community and allows individuals to participate in real-time or on their own time by viewing the recordings of the sessions. We will continue this effort in 2021 with our FA Connect series (p. 28) providing education opportunities and psychosocial sessions with trusted consultants.

Our efforts to prevent and treat FA cancers are moving forward in big ways this year. First, we entered a partnership with the widely known organization Stand Up to Cancer to support a $3.25 million grant on head and neck squamous cell carcinoma (HNSCC) research. This is the first time FARF has collaborated with a large cancer-focused organization on such an impactful grant, and we are excited that FARF is leading efforts on HNSCC research. Read more on page 8.

In September, we invested in a project between the University of Washington and The Rockefeller University to study anogenital cancers in people with FA. Researchers will collect and study data about these cancers to help identify the best treatments. This is also a first for FARF – to fund a study exclusively focused on anogenital cancers.

In other funded research news, in 2020, FARF awarded our largest grant to date to our colleagues in Germany. This $732,000 grant will expand the use of brush biopsies to prevent and detect oral cancers in individuals with FA.

The FA Patient Registry continues to grow, with over 100 FA individuals consented and imputing data. This year we have also continued to develop and refine the FA virtual tumor board to assist individuals who are diagnosed with a cancerous tumor.

Another significant achievement includes advancements in the neuroinflammation issues faced by many in the community through collaborative efforts between several institutions. Read more about one individual's experiences with these issues on p. 12.

Finally, earlier in 2021 we released the 5th edition of the Fanconi Anemia Clinical Care Guidelines. This comprehensive manual has served as the go-to resource for families and clinicians for over 20 years. We will spend the next few months transferring this resource onto the FARF website, where it will be updated as new information is shared.

The pandemic will have lasting impacts, but FARF has discovered unexpected strengths and opportunities. If there was an enduring lesson during this time, it is that we are capable of far greater adaptability than we ever previously imagined. We have embraced this adaptability at FARF, and all of us here have seen it as an opportunity to build a revitalized organization and emerge from this crisis collectively stronger.

Mark Quinlan
Executive Director

Scientist Spotlight

Name: **Jordi Minguillón, PhD**

Institution: **Genomic Instability Syndromes and DNA repair Lab (Sant Pau's Hospital Research Institute, IR-Sant Pau), Centro de Investigación Biomédica en Red de Enfermedades Raras (CIBERER-ISCIII)** and **Universitat Autónoma de Barcelona (UAB), Barcelona, Spain.**

Area of expertise: **Drug screening, cancer therapeutics, orphan drugs**

My work:

In Jordi Surrallés' lab, we are focused on finding safer therapies to prevent and/or treat Fanconi anemia's worst clinical manifestations, such as bone marrow failure or solid tumors. To find an effective therapy for solid tumors, especially head and neck cancer, is an enormous challenge for people with FA, as they have an extreme sensitivity to conventional chemotherapy and radiotherapy. In our research group we plan to conduct a clinical trial with afatinib, a non-genotoxic anticancer drug already used in advanced lung cancer. We have preclinical evidence that afatinib can stop tumor growth *in vitro* in Fanconi-derived cell lines, and *in vivo* in Fanconi tumors grown in immunodeficient mice, and shows low toxicity in FA non-tumor cells and mice. We believe afatinib will be effective in FA patients as it has been in the general population. We also know that in the future, different kinds of therapy will be needed to control tumor growth. We are taking the first steps on a long road, and thanks to the great support from FARF, we are paving the way to make cancer a non-terminal disease for FA patients.

What motivates me to work on FA:

For me, it's clear: in the last few years, our research has shown that we can have an impact on the survival of people with FA. And step by step, we are seeing the possibility to improve life span for those with FA, like we've seen in the general population. We need clinical evidence to translate our research into care for patients. And once achieved, we will need additional therapies to improve the previous ones, then preclinical evidence, then clinical evidence, and so on. I'm motivated to extend survival and to overcome this devastating disease, step by step.

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

Spending time with friends, playing sports, trekking, or enjoying important moments with my family. I'm also a great fan of renewable energies and electric mobility. We could have a great and long talk about that! :)

I'd like FA families to know:

I share with you a photo of my close family: my mother and my youngest brother David, with Down Syndrome. David inspired me to study biology, to do a PhD, and finally, to work as a researcher focused on DNA repair, cancer therapeutics and of course, Fanconi anemia. Thanks to him I have learned that small steps can lead to big moves. And that's why I want to translate our research to people, to improve their quality of life, no matter how few they are.

This report is an overview of research shared at the 2020 Fanconi Anemia Research Fund Scientific Symposium covering three main topics: solid tumors, fertility, and bone marrow failure.

RESEARCH UPDATES

SOLID TUMORS

What is the issue and how does it affect people with FA?

Fanconi anemia (FA) is a DNA repair disease that leads to the development of squamous cell carcinoma (SCC). SCC of the head and neck and anogenital regions are the most common for people with FA as they reach adulthood. Current therapies used in the general population are toxic for people with FA; therefore, there is an urgent need for novel interventions that will safely prevent or treat SCC in this population.

What does current research in this field mean for people with FA?

Collectively, research presented in cancer sessions at the 2020 Scientific Symposium demonstrate four key areas that researchers are addressing to benefit people with FA:

1. Understanding the natural history of FA cancer through genome sequencing of tumors and building patient registries focused on cancer;
2. Cancer prevention using oral screening, brush biopsy of oral lesions, and liquid biopsies of plasma and saliva;
3. Chemoprevention using compounds such as the antioxidant quercetin administered systemically, or tocilizumab (an immunosuppressive drug) delivered using Janus nanoparticles; and
4. Targeted therapeutics of the specific pathways (PI3K and EGFR), and radiotherapy.

Overall, the research presented shows that we are beginning to understand the driving mechanisms of FA cancer and are identifying and testing potentially effective and safe therapies.

What are the next steps?

The next steps for FA cancer research are to create preclinical models that mimic the disease so that targeted therapies can be tested. Additionally, further characterization of individual FA tumors is necessary to identify targetable drivers of the disease. Lastly, cancer-focused clinical trials for people with FA are needed to test potential prevention and therapeutic modalities.

Highlighted projects on solid tumors: Genomic characterization of tumors from Fanconi anemia patients

Agata Smogorzewska, The Rockefeller University

We know FA patients are at heightened risk of developing cancer of the head and neck and anogenital regions; however, there are very limited data available on tumors from FA patients and the natural history of the disease. Dr. Smogorzewska described results from a study focused on whole exome and genome sequencing of FA HNSCC tumor samples that were collected from the International Fanconi Anemia Registry. Her results showed that FA cancers are characterized by a complex structural landscape that impacts key genetic regions associated with HNSCC development.

Janus nanoparticles for local delivery of tocilizumab for oral squamous cell carcinoma chemoprevention

Susan Mallery, Ohio State University

Successful chemoprevention of FA HNSCC requires adequate coverage of chemoprevention compounds throughout the oral cavity. At the 2020 Symposium, Dr. Mallery discussed her research focused on janus nanoparticle (JNP) delivery of tocilizumab, a drug that blocks the cytokine, IL-6, which has been shown to have a vital role in the development of oral squamous cell carcinoma.

Janus nanoparticles are formulated to deliver drugs to oral epithelium and provide release of the drug to the appropriate compartments. In this study, results show that different cell types internalized the janus nanoparticles (JNPs) and tocilizumab. Dr. Mallery's lab is currently investigating whether sustained release tocilizumab JNPs are effective in preclinical tumor models.

Proton radiation to maximize head and neck cancer treatment in FA

Mathieu Sertorio, University of Cincinnati

X-ray radiation therapy (XRT) is one of the most frequently used cancer therapies for HNSCC in the general population. Knowledge of the biology and effects of XRT is well known due to its use over many decades. Patients with FA who are treated with traditional XRT experience many adverse side effects. Proton radiation therapy (PRT) is a precise and advanced form of radiation therapy that may have benefit in treating FA head and neck and anogenital cancers. This type of therapy uses a specialized pencil beam that destroys cancer cells while minimizing damage to surrounding healthy tissues and organs. At the 2020 Symposium, Dr. Sertorio discussed his pre-clinical research focused on delineating the differences and efficacy of XRT versus PRT for patients with FA. In his study, he reported that major differences exist when XRT or PRT is administered to cultured FA cell lines. It is too early to tell what these differences mean, but it is encouraging that there is a differential response between the two types of therapy. Future work will focus on modulating PRT therapy to maximize efficacy with minimal toxicity.

FERTILITY

What is the issue and how does it affect people with FA?

In addition to progressive bone marrow failure and the high risk of developing squamous cell carcinoma of the head and neck and anogenital regions, patients with FA also experience reduced fertility. Females with FA may experience a variety of gynecologic issues, including structural abnormalities, delayed puberty, decreased fertility, early menopause, and a high risk of SCC of the lower genital tract. Males with FA may have numerous structural abnormalities of the reproductive system and extremely low sperm count that affect fertility. Compared to other more commonly studied aspects of FA, not much is known about infertility in people with FA. As more individuals with FA reach adulthood, there is a growing need to address the issue of fertility in this population.

What does current research in this field mean for people with FA?

Collectively, research shows that many FA investigators are pursuing fertility-related research on FA. Current research demonstrates the feasibility that fertility restoration may be possible for people with FA. Other studies highlight the functional role of the FA pathway in regulating fertility. Insight into these mechanisms may provide the framework for future therapies that can restore and repair FA-related defects in germ cells.

What are the next steps?

Future research will need to focus more on understanding the molecular role of the FA pathway in fertility and how issues presented by a faulty FA pathway can be treated to restore fertility in FA individuals.

Highlighted project on fertility: Fanconi anemia genes in fertility and reproductive health

Eva Hoffman, University of Copenhagen

Dr. Eva Hoffman discussed an overview of the male and female reproductive lifespan and the role of the FA pathway in fertility. Dr. Hoffman showed that rare mutations in FA genes cause primary ovarian insufficiency and common variants in DNA repair genes are implicated in natural menopause. Infertility and hypogonadism is common in FA male patients, but more variable in females. The good news for FA patients is that researchers are working to make fertility restoration possible. In females the goal would be ovarian tissue cryopreservation prior to gonadotoxic treatments used during bone marrow transplant procedures and in male patients this would be culturing spermatagonia stem cells isolated during infancy and transplanting back into the stem cell niche during adulthood. Dr. Hoffman's team has currently shown that sperm stem cells from male infants can be propagated in vitro and can populate the stem cell niche in nude mice. The goal of future experiments will be to test whether the procedure can be done in humans.

BONE MARROW FAILURE MECHANISMS AND CLINICAL APPLICATIONS

What is the issue and how does it affect people with FA?

Individuals with FA often have progressive bone marrow failure (BMF); however, research has advanced clinical care of BMF and now patients with FA can successfully undergo curative hematopoietic cell transplants (HCT). Despite the success rates

of HCT for FA patients, issues related to chronic graft-versus-host disease and secondary cancers from genotoxic treatments used during HCT need to be addressed. In addition, the progression to myelodysplastic syndrome (MDS) and leukemia has not been well defined.

What does current research in this field mean for people with FA?

Research shared at the Symposium demonstrates efforts focused on preventing, treating, and understanding the progression of bone marrow failure. Drugs that can prevent DNA damage in the hematopoietic compartment have the potential to prevent BMF and circumvent the need for hematopoietic cell transplants. Studies focused on understanding the progression of FA hematopoietic stem cells to MDS or acute myeloid leukemia (AML) can offer potential screening mechanisms for early detection and treatment. Research focused on improving HCTs by using nontoxic conditioning antibodies may lead to less toxic HCT procedures that have less risk of longer-term complications, graft-versus-host disease, and the development of solid tumors.

What are the next steps?

The next steps for bone marrow failure research fall into four categories:

1. Identifying potential drug targets to prevent and treat bone marrow failure;
2. Understanding the progression of MDS and AML;
3. Nontoxic transplants using conditioning antibodies; and
4. Gene therapy and editing.

Identifying potential drug targets will require continued preclinical work to demonstrate the efficacy of drug candidates. Newly identified compounds that pass testing in the preclinical stage will then need to be tested in clinical trials. Studies focused on understanding the genetic changes that lead to the progression of MDS and the development of AML will need to demonstrate efficacy in patient populations before clinical screening can be implemented routinely.

The antibody conditioning field will need to determine efficacy and safety in the FA patient population prior to the widespread implementation of this type of transplant. A clinical trial at Stanford is set to begin in 2021.

Gene therapy and editing are the final frontiers for treating bone marrow failure and the root cause of the genetic defects in FA patients. Gene editing is currently in the preclinical phase of research. Gene therapy clinical trials for people with FA are ongoing in both the United States and Spain, with more sites opening in the US and the United Kingdom.

Highlighted projects on bone marrow failure: JSP191 antibody conditioning for hematopoietic cell transplantation: anticipating new therapy for Fanconi anemia

Agnieszka Czechowicz, Stanford University

Hematopoietic cell transplant outcomes for patients with FA are excellent with newer standard treatments, but the procedure is still imperfect due to the use of toxic chemotherapy or radiation used during the procedure. Long-term complications such as graft-versus-host disease and an increased risk of cancer following transplant are both concerning issues. Improved transplant procedures that do not use toxic reagents to clear diseased bone marrow prior to transplantation with healthy donor cells are desperately needed. Dr. Czechowicz discussed her research focused on non-toxic conditioning using an anti-C-KIT antibody called JSP191. JSP101 targets C-KIT, which is a cell surface protein expressed primarily on hematopoietic stem cells and progenitors. Dr. Czechowicz's preclinical studies using mouse models showed that the antibody eliminates host diseased FA hematopoietic stem cells and is safe. Dr. Czechowicz will be initiating a Phase 1b/2a clinical trial using JSP191 in transplants for patients with FA in 2021.

Preclinical efficacy of ALDH2 activator in protection of HSC from aldehydic injury: rationale for a clinical trial in Fanconi anemia

Kenneth Weinberg, Stanford University

Reactive aldehydes are the major cause of DNA damage in individuals with a faulty FA DNA repair pathway. Dr. Weinberg discussed his work focused on prevention of damage by way of activation of ALDH2 through a small molecule called Alda1. ALDH2 is a member of the aldehyde dehydrogenase (ALDH) family of enzymes, which oxidize aldehydes into less toxic compounds. ALDH2 specifically coverts acetaldehyde to acetic acid, which does not cause DNA damage. Sustained administration of Alda-1 to FA animal models restored hematopoietic stem cells (HSC) and reduced aldehyde-mediated DNA damage with no adverse side effects observed. These preclinical experiments provide compelling evidence that sustained activation of ALDH2 can prevent HSC loss in FA and that the drug could be used as a potential preventative treatment for FA.

https://fanconiregistry.iamrare.org

How you can move research forward

Fanconi anemia (FA) is a rare disease, and it manifests in many different ways, making it challenging to diagnose and treat. Research is the key. And the key to research is you. This is where the FA registry comes in. If you or your child has been diagnosed with Fanconi anemia, join the registry today.

How does the registry help?

The Fanconi Anemia Research Fund (FARF) created the FA Registry to:

- Let people with FA know if they might be eligible for clinical trials
- Help develop new diagnostic and treatment options
- Create a centralized place to view anonymous data of users
- Understand how FA changes over a person's lifetime

How do I register?

Joining the FA registry is free and easy. Visit **https://fanconiregistry.iamrare.org** and click "register" to create your private account.

You will then be asked some questions about yourself or your child. This includes basic personal information, as well as questions about the FA diagnosis, medical history, and experiences with the disease.

Don't have all the information on hand? No problem. You can pause and come back to the survey at any time.

The FARF staff is here to help. Call us at **541-687-4658** or email **registrymanager@fanconi.org** with any questions.

FARF Enters Head & Neck Cancer Collaboration with Stand Up To Cancer

People with Fanconi anemia (FA) are at a very high risk of developing head and neck cancers because of the underlying DNA repair problem in FA. Research is the key to understanding, diagnosing, and treating these cancers. That's why the Fanconi Anemia Research Fund (FARF) has joined Stand Up To Cancer® (SU2C) and three partner organizations to fund $3.25 million in grants to find new treatments for head and neck cancers. This project reflects FARF's commitment to directing, accelerating and investing in cancer research.

The grants include contributions of $1.5 million each from the Fanconi Anemia Research Fund and the Farrah Fawcett Foundation. The American Head and Neck Society and the Head and Neck Cancer Alliance are each providing $125,000 to support the project.

The grants will support new approaches to treating head and neck cancers, especially those associated with FA and human papillomavirus (HPV). The incidence of head and neck squamous cell carcinoma in people with Fanconi anemia is 500- to 700-fold higher than in the general population and treatment options are limited. HPV is a very common virus that can cause cancer, including cancer of the throat; approximately 45,300 people with HPV will get a cancer diagnosis every year in the U.S.

Head and neck cancers associated with FA or HPV have something in common: genetic defects that cause FA, as well as genetic changes resulting from HPV infection, both adversely affect DNA repair systems, which can lead to cancer.

"These grants will collectively benefit people with Fanconi anemia and their families in a meaningful and impactful way," said Mark Quinlan, Executive Director of the Fanconi Anemia Research Fund. "We're excited to join the other funders and Stand Up To Cancer on this project, which will facilitate the collaboration between experts from multiple fields to help tackle some of the biggest challenges associated with head and neck cancers."

The research team will be titled the Stand Up To Cancer–Fanconi Anemia Research Fund– Farrah Fawcett Foundation Head and Neck Cancer Research Team and the researchers will be brought together in an innovative way. After submitting applications, selected scientists will attend a two-day Ideas Lab, scheduled for Spring 2021. The Ideas Lab will be structured to help leading scientists from different disciplines explore how they could work together to make progress on understanding head and neck cancers. At the end of the meeting, the researchers will be invited to write formal proposals and suggest a budget for a three-year grant.

"Bringing together this group of donors provides a unique opportunity to unite researchers who typically take different approaches to similar underlying molecular problems that lead to various forms of head and neck cancers," said Lee Helman, MD, Director of the Osteosarcoma Institute and vice-

FANCONI ANEMIA RESEARCH FUND
Advancing Research. Supporting Families.

Farrah Fawcett Foundation

Head and Neck Cancer ALLIANCE

chair of the SU2C Scientific Advisory Committee. Helman will lead the Head and Neck Cancer Research Team joint scientific advisory committee. "This is the type of collaboration that Stand Up To Cancer fosters and it's wonderful to see how this model leads to new ways to approach questions in cancer biology and treatment."

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>
> — Lee Helman, MD, Director of the Osteosarcoma Institute and vice-chair of the SU2C Scientific Advisory Committee.

About Stand Up To Cancer

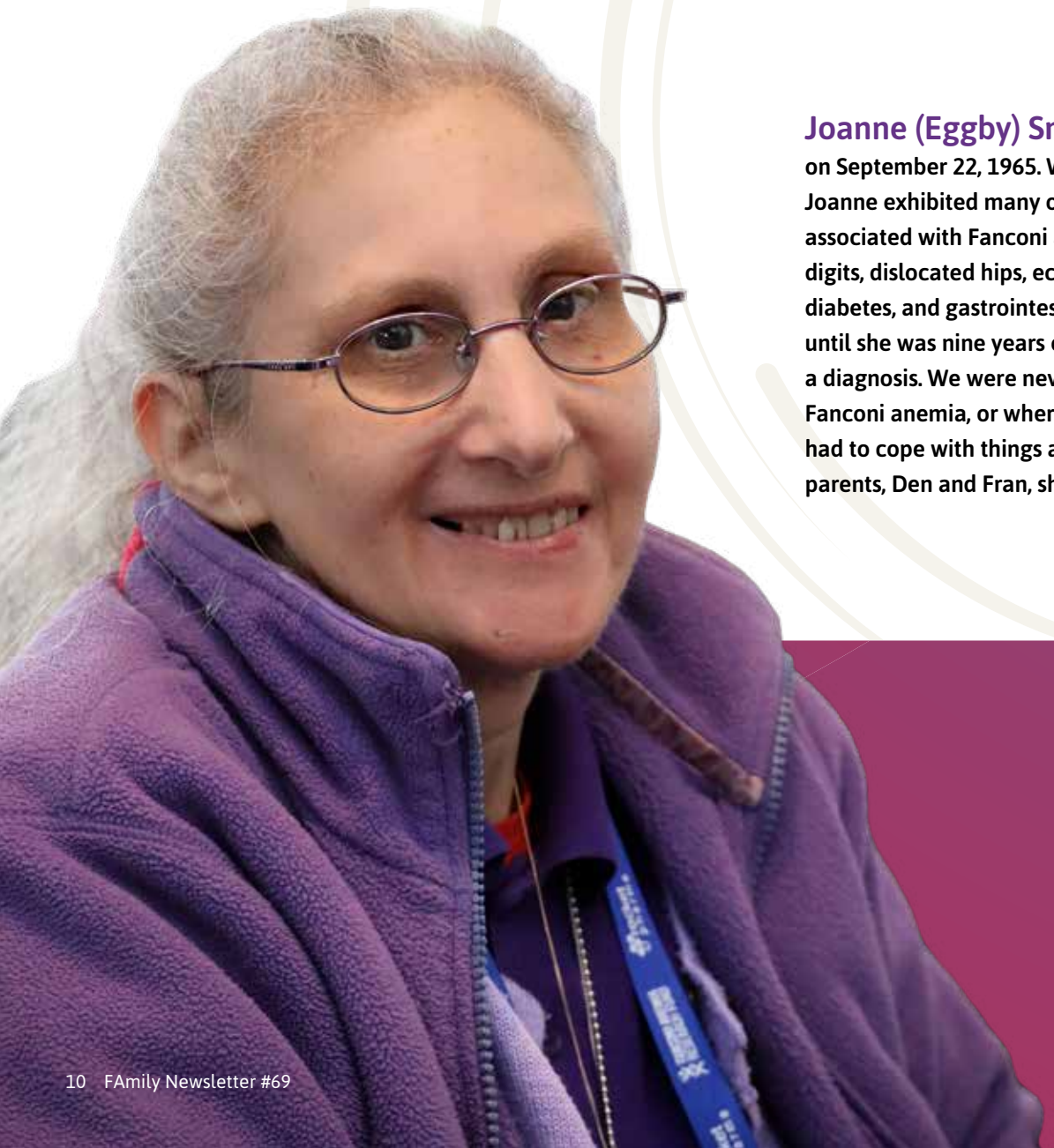
Stand Up To Cancer® (SU2C) raises funds to accelerate the pace of research to get new therapies to patients quickly and save lives now. SU2C, a division of the Entertainment Industry Foundation, a 501(c)(3) charitable organization, was established in 2008 by media and entertainment leaders who utilize these communities' resources to engage the public in supporting a new, collaborative model of cancer research, to increase awareness about cancer prevention, and to highlight progress being made in the fight against the disease. As of August 2020, more than 1,950 scientists representing more than 210 institutions are involved in SU2C-funded research projects.

Under the direction of our Scientific Advisory Committee, led by Nobel laureate Phillip A. Sharp, Ph.D., SU2C operates rigorous competitive review processes to identify the best research proposals to recommend for funding, oversee grants administration, and ensure collaboration across research programs.

Current members of the SU2C Council of Founders and Advisors (CFA) include Katie Couric, Sherry Lansing, Kathleen Lobb, Lisa Paulsen, Rusty Robertson, Sue Schwartz, Pamela Oas Williams, and Ellen Ziffren. The late Laura Ziskin and the late Noreen Fraser are also co-founders. Sung Poblete, Ph.D., R.N., serves as SU2C's CEO. For more information, visit StandUpToCancer.org.

> With the help of FARF, I feel like I have an extended family and great support.

Joanne's Legacy



Joanne (Eggby) Smith was born in England on September 22, 1965. Weighing only 4 lb 7 oz, Joanne exhibited many of the physical anomalies associated with Fanconi anemia including missing digits, dislocated hips, ectopic kidneys, type 1 diabetes, and gastrointestinal abnormalities. "It took until she was nine years old before we were given a diagnosis. We were never told anything about Fanconi anemia, or where to get any help, and just had to cope with things as they cropped up," her parents, Den and Fran, shared.

In spite of her many health obstacles, Joanne grew to be a lively child who loved Elvis Presley and animals. Her older sister, Helen, always admired how easily Joanne made friends. "She was such a good sister and much-loved Auntie to my three children. We always enjoyed our family get togethers both as children and as adults."

In 1983, Joanne met Kevin, the love of her life. The two were married three years later and spent their marriage traveling through 23 countries, attending various athletic championships, and mastering new skills like playing darts. "She even won a darts competition in Germany," Kevin fondly recalled. Joanne and Kevin lived in Munich, Germany for seven years, where she learned German and started porcelain painting.

It wasn't until 2010 that the two noticed that Joanne was not able to do the things that she used to do. Cancer crept into the picture and eventually she had to use a walking stick and wheelchair.

On April 16, 2020, Joanne passed away.

But in true Joanne-fashion, she made two final gifts to the FA community which have been dedicated to advancing the efforts of Fanconi Hope, in the UK, and providing international scholarships for adults living with Fanconi anemia who wish to attend FARF events in the United States.

You see, Joanne had never met another person with Fanconi anemia until she was nearly 50 years old. When reflecting on her first experience at a FARF Adult Meeting in 2012, Joanne said, "this was the first time I had met anyone else with FA, and it was a great inspiration to share our experiences. Even though we are all FA warriors, we all have different characteristics. With the help of FARF, I feel like I have an extended family and great support. Being with FARF reminds me of the famous English soccer song, "You'll Never Walk Alone".

Joanne's life and legacy lives on through her family and her spirit of generosity. She inspires us all to ask, 'what will our legacy be'?

Joanne with her loving husband, Kevin, at a FARF meeting (left) and a Fanconi Hope fundraiser (right).

You'll Never Walk Alone

When you walk through a storm
Hold your head up high
And don't be afraid of the dark
At the end of a storm
There's a golden sky
And the sweet silver song of a lark
Walk on through the wind
Walk on through the rain
Though your dreams be tossed and blown
Walk on, walk on
With hope in your heart
And you'll never walk alone
You'll never walk alone
Walk on, walk on
With hope in your heart
And you'll never walk alone
You'll never walk alone

Tara (center) with her mother and siblings.

Our Journey with FA and Brain Lesions

By Mary Eilleen Cleary

Eternal hope. Of all the characteristics that Fanconi anemia (FA) may or may not bring out in any of us, this is the one that will help us the most. We must always believe medical answers are right around the corner. We must always believe tomorrow will be better than today. Following this isn't easy, but then again, nothing about FA is easy. We know that.

This is the second time I have been asked to write about our journey with FA. The first time was right after my now 19-year-old daughter Tara had successfully, and without many issues, gone through a bone marrow transplant in 2009.

Tara is the youngest of four children and the only one with FA. She had never been ill, and we were shocked to discover she was in bone marrow failure when she was seven. She was born small and stayed small, and sure, she had café au lait spots, but we had never heard of Fanconi anemia. Right after the diagnosis, I couldn't even see the words without feeling sick. I'm sure every parent shares this moment when the knowledge of a demon you didn't know to fear takes over your world.

> **"I also nurture eternal gratitude: for the medical professionals, who are just as gritty and determined as we are to find answers and give hope; for the FA family support group that rallies around any family in crisis; for FARF and the individuals who are part of its history and its future; for family and friends who listen, care and pray."**

Tara was transplanted right after her eighth birthday. Her sister Brenna was the match, and my writing at that time, in retrospect, was obnoxiously optimistic and now seems somewhat naïve. But I defend the need for optimism, especially when FA yields a surprise. It took our best medical detectives to figure out the surprise that began for Tara in 2013.

"Mom, my hand doesn't work," Tara said to me one day.

That was the beginning of the next leg of our FA journey, although linking the issue to FA would take years. We began an odyssey with neurologists, hand experts, and occupational therapists. No one seemed willing to tackle the source of the issue. (The preceding sentence encapsulates what would take me a book to explain). What good is trying to treat a hand that is losing strength and coordination if one has no idea what is causing the problem?

My frustration was growing after visiting multiple doctors and sites, and when I started seeing signs of trouble in Tara's

walking in 2014, my frustration turned to absolute panic. The internet is often not the answer when panic has you by the throat, but in this case, it led me to a care team second to none. Enter Dr. Stella Davies and her colleagues at Cincinnati Children's Hospital and Medical Center (CCHMC), most notably Dr. Greg Wallace and nurse care manager Leann Mount. After seeing what Dr. Davies did for us upon our first visit, I quickly nicknamed her "The British Dynamo."

I searched the web for top neurology programs and CCHMC was near the top, and I knew it had an FA center. I made an appointment with neurology and then decided I should let the FA team know what was going on and that we would be visiting. That was all it took. We could have left the next day – they were that eager to help. We arrived for clinic appointments but by the end of the day, Tara was checked into the hospital so they could get a myriad of tests completed quickly. Stella Davies took control and worked with neurology and infectious disease to get as much data as possible. The MRI showed lesions on the brain that puzzled the experts and were not known at that time to be associated with FA. They are now.

Since 2014, we have tried plasmapheresis, steroids and a series of medicines and infusions. One step forward, three steps back. We added visits to Mayo and the University of Michigan. Suddenly in 2019, there was a breakthrough that linked the brain lesions to FA.

Dr. Davies became aware of at least two other people with FA who had similar-looking brain lesions. Those two had brain biopsies, which Tara has never had. In each, they found evidence of the JC Virus, something the majority of us have within our bodies and manage without incident. It seems the immune systems in some people with FA damage brain tissue while fighting the virus.

So now what? A huge question with no immediate answer. Dr. Davies and others are searching, writing and presenting papers. Scientists and clinicians around the world are now aware of the issue, and we can hope and pray that combined knowledge and curiosity result in answers for those we love – in my case, my sweet, determined, gutsy Tara.

Since Tara was 13, she's had to watch her abilities whittle away. She used to play volleyball and soccer. She used to be able to sign her name with ease. Now, she has to think about each step and struggles with balance, her vision and her memory. She's had seizures and in November spent two nights in the PICU when a seizure came with an unexplained fever. She got out of the hospital on a Wednesday night and that Friday was answering questions during a Zoom job interview. She got the job. She just finished another class at the community college. Tara dreams of following in her sisters' and brother's footsteps – graduating from college, getting her own place and driving. Boy, does she want to drive. I want her to drive, too. I want it all for her. The path is not apparent now, but we remain hopeful.

I nurture eternal hope because the alternative is unimaginable. I also nurture eternal gratitude: for the medical professionals, who are just as gritty and determined as we are to find answers and give hope; for the FA family support group that rallies around any family in crisis; for FARF and the individuals who are part of its history and its future; for family and friends who listen, care and pray.

When you deal with a critical illness, eternity seems the proper measure.

Research on central nervous system abnormalities

FA clinicians Stella Davies (Cincinnati Children's Hospital) and Eunike Velleuer (University of Düsseldorf) are collaborating with experts from the University of California San Francisco (UCSF) who specialize in infections and autoimmune diseases of the central nervous system.

The UCSF team has started to evaluate FA patients as part of a research study. Their group also is establishing a collaboration with a care team on the east coast so that patients in that region can be seen. FARF recently received a letter of intent (LOI) from this group to request funding to study CNS abnormalities in a clinical study. This is the first LOI we've received on the topic and first non-FA specific research team to get involved with the CNS issue. This is an important advancement to understanding these complications and finding therapies.

If you or your loved one with FA may be experiencing central nervous system issues like those shared in this article, please contact FARF (info@fanconi.org or 541-687-4658) and we can put you in contact with those working on this.

MY JOEL

Natalia and Joel on their wedding day, surrounded with love from nurses, doctors, and staff at Memorial Sloan Kettering Cancer Center.

By Natalia Gimenez

I remember the day well. My husband Joel had his annual check-up in New York on a random April day in 2017. In May, shortly after his appointment, Joel was diagnosed with Fanconi anemia (FA). At first, we thought it was just regular anemia, but then the doctor informed us that he might need a bone marrow transplant and we began to take the situation more seriously. The news was devastating and left us feeling hopeless.

I prayed for help and found the Fanconi Anemia Research Fund (FARF). Immediately, I received support from FARF. Staff members recommended that I make an appointment with Dr. Farid Boulad at Memorial Sloan-Kettering Cancer Center (MSKCC) in New York City. I spoke to FARF Family Services Director that morning, and by the afternoon I

> When I think back on this moment, it is so clear that quality of life is not only determined by one's health, but that it is often determined by the joy and love that you let into your heart.

received a call from Dr. Boulad. He and Dr. Maria Cancio restored our hope. After speaking to them, we decided to go to MSKCC to treat Joel's condition.

Joel received his first bone marrow transplant in December of that year. He responded very well to treatment and didn't experience any negative side effects. My brother Federico flew all of the way from Paraguay to assist me with caregiving. The three of us (Federico, Joel, and I) truly made an amazing team. Humor, love and faith was what kept us strong and positive, even though ultimately, Joel's long-term prognosis was poor.

Right after his transplant, and before Joel was discharged from MSKCC, we received the news that he had another condition called myeloproliferative neoplasm (MPN). At that time, we weren't worried about it. We were just so overwhelmed by the love and care that we received from hospital staff.

In May of 2018, Joel received the news that he could proceed with another bone marrow transplant, or that he could continue to proceed with his current treatment regimen. The decision was difficult for him, but ultimately, he decided to take the calculated risk of having a second transplant. He was matched 10/10 with an international donor and proceeded with the transplant. We were encouraged by the fact that the cells were one hundred percent engrafted, however, shortly after, we received the awful news that Joel had now developed leukemia. The thought of a third transplant was almost unbearable. We learned that Joel's body would be unable to withstand another transplant or any other strong treatment for leukemia.

In November of 2018, Joel asked me to marry him. We were married that December, and family from Paraguay, Canada, and Maryland were able to celebrate that most unforgettable moment in our lives. Nurses, doctors, assistants, volunteers, friends, staff members on the floor joined us during the ceremony as well, which was held in the chapel at MSKCC. Our flower girl was a patient from the same unit as Joel, and was the sweetest little person that Joel met at the hospital. We couldn't have asked for more in that moment!

That day, Joel was beyond blessed and the happiest he'd ever been. He received an abundance of love, so much more love than he could have imagined or asked for. He called each person who celebrated with us and thanked them for being angels in his life.

When I think back on this moment, it is so clear that quality of life is not only determined by one's health, but that it is often determined by the joy and love that you let into your heart. It's determined by the joy and love that you give freely and receive freely. Joel had this. He had this along his entire FA journey, even to the very moment he received his own pair of angel wings in February of 2019.

His mission was to connect people and we try to keep this mission alive. Nowadays, I remain in close contact with the people who took care of Joel at MSKCC, including Dr. Cancio, whom I consider a close friend. She was and still is an angel in my life with Joel and along this FA journey.

CAREGIVER SPOTLIGHT:

LOVING SOMEONE RARE

By Allison Breininger

Originally published February 28, 2021 at www.thenegativespace.info

My husband is one of the 30 million Americans living with a rare disease.

Fanconi anemia (FA) has no color recognizable by the general public. You can't buy cereal or water bottles or winter hats at your local Target to support its cause. Professional athletes don't wear colorful sneakers to raise awareness on its behalf.

Upon hearing about Fanconi anemia for the first time, well-meaning people searching for something with which to connect, pounce on the word "anemia," making comments about how iron pills should probably help and how their uncle was once anemic, but is now doing just fine.

Even medical professionals are often unaware of the disease, though some remember the one question on their board exams that mentioned it.

Ten years into being a caregiver for a person with Fanconi anemia, I can say without a doubt that I am grateful that this disease is rare. The effects of FA are devastating, and I don't want the numbers of people who live with it to be any larger than they currently are.

Yet there are ways in which caregiving, an already incredibly challenging role, is made even more difficult when your person's disease is rare.

All those cereal boxes, water bottles, and NBA sneakers raise funds. Funds for research and resources so that treatments can be improved upon, medical centers can be well-equipped, and family support can be provided.

By contrast, in the rare disease community there is more pressure on the families to organize fundraisers if they want to move the needle towards a cure. Holiday letter appeals are written, 5ks and bake sales are organized — all on top of caregiving for the person living with the disease. Even when beautifully executed, these grass roots appeals can fall on deaf ears due to lack of awareness of the disease. I mean, how expensive can a few iron pills be after all?

In addition to funds, it can be hard to gather support or even a level of sympathy when the people in your life have no reference point for the disease with which your family is living. If I was to say to a new

How friends, family members, and coworkers can show up for caregivers of someone with FA:

Ask me:

"That's not something I've heard of before. Can you tell me more?"

"What does having that disease look like for him on a daily basis?"

"What does that diagnosis mean for you, his caregiver?"

"Is there a place where I can make a donation to support research?"

Finally, as you see appeals from caregiver friends to take part in a walk, buy a t-shirt, or order some frozen pizzas, know that what they are actually asking for in that moment is recognition that they are caring for someone who is rare, for help in the work they are doing to raise awareness, for financial support to get closer to a cure, and to be less alone on the rare disease path than they are right now. As you are able, buy a shirt, help spread the word, offer to bring snacks to the walk, commend them for this and all they do, and tell them that you see and honor them.

Our disease is rare, but compassion for our loved ones *and* ourselves doesn't have to be.

colleague or neighbor that my husband has pancreatic cancer, there would be an immediate level of understanding about the severity of the illness, what treatment may entail, and what needs we may have, because the listener would likely have some experience with that disease. On the contrary, when I say, "My husband has a rare, genetic disease called Fanconi anemia," the listener's brain searches in its schema files for some sort of reference point. Finding none, iron pill comments are made or the subject is changed.

Fundraisers and awkward conversations, while hard, are a mere nuisance in comparison to being treated by a medical team that has little to no knowledge or experience with the disease from which your loved one suffers. As a caregiver, this means I — someone who is not medically trained — need to stay current with research, protocols, signs and symptoms for which to watch, and I need to educate his team and to advocate on his behalf. Most families living with FA and other rare diseases must travel great distances to be seen and treated by teams who are equipped to help. But even when working with those who are experts in the field, there are often no clear paths, since being rare often means being the first of your kind. With no trails blazed before us, significant treatment decisions are left to us to research, coordinate, and decide upon using the few, if any statistics that exist.

Our family – and yours too, if you're reading this newsletter – is fortunate to be connected with the Fanconi Anemia Research Fund (FARF), whose team works with dedication, passion, and compassion to move that needle towards a cure, to support families in educating their medical teams about the ins and outs of the disease, to coordinate experts in making decisions about treatments, and to intentionally recognize and support caregivers.

Our disease is rare, but compassion for our loved ones *and* ourselves doesn't have to be.

ONE MONTH.

ONE CAUSE.

It's time to make an impact.

Twelve years ago, FA families started an International Fanconi Anemia Day, now known as FA Month, to raise funds for research and family support services. Last year, together we raised nearly $110,000. This year, we hope to raise another $110,000!

Through the years, the theme of FA Month has been #ThisIsHowIFA, #FindAnswers, and #FightAlways. This year's theme is #FamilyAroundTheWorld. People with FA, family members, scientists, doctors, volunteers, friends, and staff members are all part of this FA family that spans the globe.

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 Month 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!

#FamilyAro

We're stronger together. That's why this year's theme is

#FAMILYAROUNDTHEWORLD

Together we can make a huge impact!
Go to **https://fundraise.fanconi.org/FAmonth** to add your page and see families like yours around the world who are raising funds this FA month.

undTheWorld

All about FANCONI ANEMIA

What is FA?

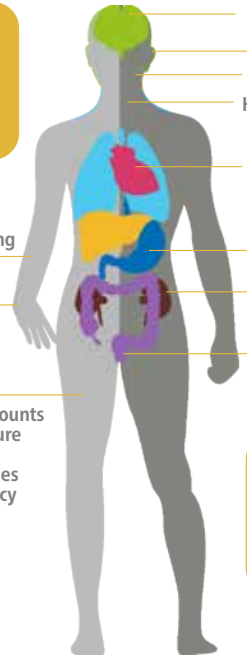
Fanconi anemia (FA) is a genetic DNA repair disorder that may lead to bone marrow failure, leukemia, and/or solid tumors (cancer). It is caused by one of at least 23 genes. FA can affect all systems of the body. It is a complex and chronic disease that is psychologically demanding.

1 in 131,000

FA occurs almost equally in males and females and is found in all ethnic groups. The likelihood of a child being born with FA is about 1 in 131,000 in the U.S., with approximately 31 babies born each year.

If both parents carry a mutation in the same FA gene, they have a 25% chance of having a child with FA.

How does FA affect the body?



People with FA may experience any combination of these symptoms, ranging from one, to many.

People are living longer

More than half of the FA population registered with FARF is age 18 or over!

Here's why you should care

It's simple: children and adults with Fanconi anemia need research to live. Without research, they won't get the advances in treatment that they need to survive. Here's another big reason you should care: FA research benefits the rest of the population, too. Bone marrow transplants have become much safer & more effective because of studies with FA patients. At least five FA genes are also breast cancer susceptibility genes, meaning therapies developed for FA patients would benefit breast cancer patients, too. And, Fanconi anemia research is in the process of unlocking the mysteries of DNA repair problems, which are at the root not only of FA, but of cancer.

Here's how FARF helps

Research has added years to the lives of people with FA. Decades ago, children rarely survived to adulthood. Now, there are adults with FA that live into their 30s, 40s and beyond.

Thanks to research, the rate of successful bone marrow transplants has gone from 20% in the 1990s to over 90% today.

FARF provides support to individuals with FA and their families by way of educational resources, support groups, and family meetings.

FANCONI ANEMIA RESEARCH FUND
Advancing Research. Supporting Families.

Here's what needs to happen next

- ☐ Fund clinical trials aimed at treating and preventing cancers in people with FA
- ☐ Develop treatment plans for all stages of FA cancers, recognizing that chemotherapy and radiation can be toxic to FA cells
- ☐ Continue to fund research to make bone marrow transplants even safer, or, to prevent bone marrow failure altogether

We need you.

Covid-19 Vaccine & Fanconi Anemia

This article is based on information available as of March 25, 2021. Visit Fanconi.org/news for the latest updates.

This statement was written with the input of the National Organization for Rare Diseases and clinicians specialized in treating Fanconi anemia patients.

In December 2020, two COVID-19 vaccines were approved by the FDA in the United States. Another was approved in early 2021. What does this mean for people with Fanconi anemia and their families?

About the vaccine

As of March 2021, the three vaccines approved for use in the United States are from Pfizer, Moderna, and Johnson & Johnson.

The Pfizer vaccine is approved for people 16 years and older (administered in 2 doses, 3 weeks apart), and the Moderna vaccine is approved for individuals 18 years and older (administered in 2 doses, 4 weeks apart). Both the Pfizer vaccine and Moderna vaccines are mRNA-based vaccines. These vaccines do not contain the virus or even a piece of the virus and therefore cannot cause COVID-19 infection. Both vaccines are generally well tolerated with no serious safety concern and have greater than 95 percent efficacy at preventing COVID-19 symptoms.

The Johnson & Johnson vaccine is administered in a single dose, opposed to two doses like both the Pfizer and Moderna vaccines. The vaccine is currently recommended for people aged 18 years and older. The J&J vaccine does not contain SARS-CoV-2 and cannot give you COVID-19, and differs from the Pfizer and Moderna vaccines, in that it is not an mRNA vaccine but is a viral vector vaccine. For additional information on the differences between an mRNA and viral vector vaccines, please visit these CDC website (cdc.gov/coronavirus).

As more vaccines become available, you can find the most up-to-date information on the CDC website.

Should individuals with FA get the vaccine?

The best and safest way to protect yourself from the virus is by getting vaccinated. Individuals, with or without FA, should get the vaccine as soon as it is available to them, unless there is a strong reason not to, as reviewed with their own provider. This may include:

- If a patient has allergies to a vaccine component
- If a patient is currently undergoing transplant or had a transplant and has not begun the reimmunization process (timing to be determined by your transplant clinicians). According to the FA Clinical Care Guidelines (2020), "Patients with FA should be screened for immune reconstitution one year after transplant. The primary care physician should discuss the exact timing of immunizations with the patient's transplant physician" (Chapter 13, p. 230).

We strongly advise you and your family members to speak with your primary health care providers about whether and when to be vaccinated against COVID-19. Just like everything else with Fanconi anemia, the risks and benefits of any medical decision should be weighed on a case-by-case basis and discussed with the patient's treating physician.

When can I get the vaccine?

Review the CDC website to learn more about considerations for people with underlying medical conditions. While the CDC makes recommendations for those who should be offered the COVID-19 vaccines first, each state has its own plan for deciding who will be vaccinated first and how they can receive the vaccines. Contact your local health department for more information on COVID-19 vaccination rollout plans in your area.

According to the US government, all Americans age 16+ will be eligible to receive the vaccine as of May 1, 2021.

> **"The best and safest way to protect yourself from the virus is by getting vaccinated."**

FAMILY FUNDRAISING SHOUTOUTS

Benefit Boards

On New Year's Eve, the Borden family from Wisconsin partnered with a local fundraising organization to sell charcuterie boards to benefit FARF. The Benefit Boards featured various items including cheese, crackers, popcorn, and chocolate. Talk about a delicious way to help raise money for FA research and support services! We love seeing the creative ways families are raising funds in a virtual space. Thank you, Borden family!

Holiday Appeals

During the holiday season, more than 20 families sent out fundraising letters or emails to family and friends around the world. Over half of FARF's income is raised between the months of November and December and it's all because of dedicated families like yours, who continue to raise funds for grants and support programs. Thank you to *everyone* in the FA community who participated in 2020's holiday appeal season.

Shining A Light on Fanconi Anemia

For his senior capstone project, Alex Vandermeys, a 17-year-old young adult with FA, decided to focus on raising money and spreading awareness. Alex interviewed individuals in the FA community and shared their experiences and stories on his FARF fundraising page, where viewers could learn and donate. By the end of his capstone project, he raised nearly $3,000 for FA research and support services. Thank you, Alex, for shining a light on FA!

One Man Race Against Cancer

Every fall, the Fiaschetti Family from Rhode Island participates in a four-mile Pell Bridge Run that raises funds for FA research and support services. This past fall it was canceled, but that didn't stop David and Mary Ann from rallying their community to get involved. David took on the challenge and ran the race on his own. Together, the Fiaschetti community raised over $7,500. Thank you, David and Mary Ann, for continuing this tradition and supporting research.

100 Miles For A Cure

In October, the McQueen family and some determined friends completed a 100-mile bike ride to show that they were #SeanStrong. Sean McQueen, the inspiration for the run, is a college student in Virginia who lives with FA. In the last couple of years, Sean has experienced similar issues as Tara (pg. 12). This has prompted his family and community to raise funds for research into central nervous system abnormalities in FA. Thank you to all the riders, donors, and support crew that tackled this endeavor. Together, the McQueen's raised nearly $44,000! Way to go, McQueen family!

#GIVING TUESDAY

Giving Tuesday

On December 1, 2020, donors from around the world united for a day of giving back to the causes that matter to them. It's a way to balance out #BlackFriday and #CyberMonday. This year, thanks to a generous matching donor, FA families raised over $68,000 to support FA research and support services *in a single day*! Thank you to everyone who donated, fundraised, shared their story on social media, and sent emails to friends and family. This is your impact and we celebrate you for continuing to show up.

amazonsmile

DONATE WHILE YOU SHOP ON AMAZON

We are all shopping more online these days. When you buy on Amazon, you can designate FARF as your charity of choice, and we will receive a portion of the sales. Visit smile.amazon.com, select Fanconi Anemia Research Fund as your charity, and start shopping. That's it!

FAMILY FUNDRAISING LIST

From January through December 2020, FA families raised more than $2,600,000 for the Fanconi Anemia Research Fund! A record number of 253 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.

$1,400,000+
Lynn Frohnmayer

$260,000+
The Kendall and Taylor Atkinson Foundation with the Nash and Griggs Families

$200,000+
Orion and Lisa Marx

$50,000-78,000
Kevin and Lorraine McQueen
Talmage and Jean Rogers

$25,000-49,999
Gerard and Cynthia Vandermeys
Nigel and Ann Walker

$15,000-24,999
Mauro and Kerrie Cazzari
Mark De Groot and Hanneke Takkenberg
David and Mary Ann Fiaschetti
Rachel and Zachary Gratz-Lazarus
John and Martina Hartmann
André Hessels and Rutger Boerema
Brian Horrigan and Amy Levine
Rose and David Pennell

$10,000- 14,999
Brian and Carly Adel
Ryan and Rebecca Brinkmann
Herminia Carvalheira
Kevin Gatzlaff
Anonymous
Ian and Tricia Mitchell
Tyler Morrison and Rachel Altmann
Peggy Padden
Mark and Diane Pearl
Neil and Emily Robison
Andrea and Robert Sacks
Kevin Smith
Jessica and Jonathan Young

$5,000-9,999
James and Jennifer Armentrout
Joeseph and Nancy Chou
James and Crystal Eubank
Alan and Rachel Grossman
Charles and Kathleen Hull
Keith and Jessica Loo
Kristina Mack
Sheila Meehan
Nancy Nunes
David and Stacy Ownby
Chris and Mel Payne
Mark Ritchie and Lisa Mingo
William and Connie Schenone
Robert and Julie Williams

$2,000-4,999
Michael and Jennifer Aggabao
Brian Anderson and Sultana Graham
Amanda Barber
Adam and Marissa Becker
John and Francene Berglund
David and Sarah Borden
Chris and Jennifer Branov
Mary Eilleen Cleary and Gleaves Whitney
John and Kim Connelly
Britteny Ferrin
Patrick and Maria Gleason
Owen Hall and Margaret Kasting
Jeff and Beth Janock
Stan and Michelle Kalemba
John and Karilyn Kelson
Timothy and Mary Ann Lana
Daniel and Angie McMahon
Peter and Janice Pless
Paul and Rena Rice
Craig and Alisha Rushing
Richard and Marilyn Sablosky
Ron and Alice Schaefer
Bryan and Karen Siebenthal
Joseph and Natalie Vitrano

$1,000-1,999
Glen and Teresa Alessandri
Jennifer Bland
Sean and Allison Breininger
David and Kim Chew
Andrew Coons and Valeen Gonzales
Donna DellaRatta
Egil Dennerline and Nanna Storm
Ezat and Laila Faizyar
Vera Friberg
Erin Furr
Judith Hoffman
Erik Kjos-Hanssen and Turid Frislid
Stephen and Jennifer Klimkiewicz
Chad and Lauren Kriner
Robert and Anna Langtry
Col. Gregory & Lt. Col. Lynnette Lowrimore
Kelly and Gerald Mlachak
Kate and Daniel Montgomery
Lisa and Jack Nash
Ronald and Fredi Norris
Susan Ortiz
Pedro and Marina Ravelo
George and Kathryn Reardon
Les and Nancy Ross
Colleen Satterlee
Bradley and Darlene Starner
Janice and Kenneth Sysak
Alejandra Tabar Concha and Elvin Estevez Lopez
Devon and Charles Tessier
Bruce and Loreen Timperley
Emily and Gail Webster
Jason and Joan Woodle

Up to $999
Peter and Donna Abramov
Jennifer and Bryan Aitkens
Assila Al-Marshoudi
Jeff and Susan Amestoy
Dr. Vicki Anton-Athens
Marzban and Daisy Ardeshir
Ronald and Juanita Arroyo
Yavin Atzmon and Sharon Harari
Charles Balow and Xandra Towndrow
Cherie Bank
Faith Barbe and Shane Estelle
Mark and Linda Baumiller
Israel and Mary Jo Becerra
Conrad and Joan Bender
Jasmine Bennetsen
John and Elaine Beyer
Domenico Bertolucci and Federica Bonati
Tracy Biby
Rodrigo and Junia Bires
Randolph and Nancy Bloxom
Jeffrey and Donna Boggs
Richard and Tena Boson
David Boudreaux
Preziosa Briga
Manuel and Ann Brito
Nathalie Britt
Donald and Danielle Burkin
Elizabeth and Richard Butts
Flavio and Aliza Canonica
Robert and Barbara Capone
Lynn Check
Stuart Cohen and Deane Marchbein
Brian and Margaret Curtis
Darrel and Kalani DeHaan
Jeremy and Michelle DellaValle
Robert and Dawn Desmond
James and Carol Dillon
Marie Di Mercurio
Cleonice DiSandro
Brian and Jennifer Dorman
Delbert and Linda Dotson
Oscar Duque and Yanira Ramirez
Chloe Eminger
Billy Joe and Debra Estep
Curt and Crystal Fales
Scott and Windy Farmer
Carole Felmy
Daryn and Carol Franzen
Fabio and Sune Frontani
Brian and Cindy Fuller
Emmanuel and Dana Gallegos

Melody Ganz
Hilary Gosztonyi
Andrew and Jennifer Gough
Eugenio Grassi and Brittany Miller
Gary and Heidi Grassi
Madeline and Patrick Gregg
Dr. Michael Greenberg
David Guidara
Shawn and Doreen Gummoe
Rachel and Kristian Guttulsrud
Gary Haftek
Eric and Elisabeth Haroldsen
Robert and Victoria Hathcock
Barbara and Chris Hawkshaw
Patricia and Michael Hilbert
Mark and Carol Hill
Shane and Colleen Irvin
Nancy Jansen
Mary-Beth and Ben Johnson
Angela Keaton
Caroline Keenum
Lila Keleher
Dan Klug and Elizabeth Bertrandt-Klug
Christopher and Dana Lamb
Eugene and Renee Lemmon
Mayra Lemus
William and Nancy Lenahan
Peg LeRoux
Todd and Kristin Levine
Larry and Gayle Licari
Robert and Darla Lindenmayer
Tanner and Jessica Lindsay
Judy Lopez
William and Jacquelyn Lucarell
Kory and Julie MacMurray
Aaron and Nicki Marsters
Daniel and Nicole McCarthy
Peggy McDaniel
Kevin and Barbara McKee
Esther and Jamison McKellar
John and Barbara Miller
James and Holly Mirenda
John and Betty Mozisek
Des Murnane and Mai Byrne
Kenny and Lisa Myhan
Tony and Lina Nahas
Louis and Virginia Napoles
Jack and Tammy Neal
Philip Nelson and Candy Lindsey
Caroline Nguyen
Alice Nicholson
Robert and Mary Nori
Lorraine O'Connor
Alissa and Patrick O'Toole
Set Parelman
Arianna Pederson and Robert Bright
Michael and Joanna Peros
Tim and Ashleigh Pinion
John and Dianne Ploetz
Lynn and Shirley Quilici
Kelsey Robinson
Sean and Angela Ross
Jennifer and Brian Sadlowe
Ty Sanders
Sharon Saunders
William and Marisela Schaecher
Colleen Scholl
Beatrice Score
Sean and TaLisa Sebourn
Thomas and Brenda Seiford
Sylvette Silverston
Jim Siniawski
Julia, Wilhelm, and Lorelei Skakandy
Jamie Slappo
Karin Staab
Lea Ann and Jeff Stiller
Sharla and Josh Strickland
Greg and Brandi Stuart
Paul and Debra Sundsvold
Sharon Swanson
Mary Tanner
Esther Thompson
Mark and Tressa Timmer
Mark and Susan Trager
Thomas and Cathy Uno
Lucian Valor
Michael and Beth Vangel
Theresa and Louis Viola
Joe and Wendy Vitiritto
Ira and Terry Walker
Anthony Walsh
Marc Weiner
Jessica and Ezekiel Werden
David and Erica Williams
Michael and Kimberly Williams
Troy and Debra Williams
Alex Winn
David and Marivel Winn
Gerald and Elizabeth Wisz
Chad and Dawn Wood
Kyle and Madison Wright
Wesley and Susan Wycoff
Sean and Kristin Young
Thomas and Marjorie Zaborney

In Loving Memory

Finlay Clanton
8.13.2020 – 11.22.2020

Nashe Nhari
6.29.2012 – 12.9.2020

Shaun Vona
3.23.1989 – 1.13.2021

Hunsley Strickland
5.9.2014 – 1.20.2021

Jacob Grossman
12.1.1998 – 2.12.2021

FAdult Council News

Mary-Beth Johnson

Thank you, Mary-Beth

Mary-Beth Johnson joined the Fanconi Anemia Adult Council in 2019 as an inaugural member and served until February 2021.

She brought an authentic and passionate perspective to the FAdult Council by imploring both the council and FARF to examine the world through a multicultural and trauma-informed lens. Mary-Beth is a champion for diversity, equity, and inclusion of people of color, LGBTQIA+, those with differing abilities, and those of lower socioeconomic backgrounds in the FA community.

In addition to graciously sharing her personal FA and fertility stories *and* helping to develop meeting programming, Mary-Beth offered her culinary expertise at the FA Adult Meeting by demonstrating the preparation of a three course, nourishing meal.

Mary-Beth is a dedicated leader whose efforts on the council will have a long-lasting impact on this community, on FARF staff, and on the organization as a whole. We deeply appreciate every ounce of energy and passion that she shared with us during her time on the council. And while she will be very missed on the council, we know she will always be an incredible champion for the FA community.

Michelle Watkins

Daniel Kold

Amy Vangel

Welcome, Michelle, Amy and Daniel

Michelle Watkins is 44-years-old and lives in Pittsburgh, Penn. with her husband and son. During her battle with cancer at age 32, she was diagnosed with FA. Since her FA diagnosis, Michelle has been very involved in the FA community and openly shares her story in the hope it may help others. She currently works as a medical assistant and manages a neurology practice and infusion center. She is passionate about volunteering at health fairs and is an FA and cancer advocate. In her free time, Michelle enjoys traveling, fishing and camping with her family. She is also a diehard Pittsburgh Steelers fan.

Daniel lives with his wife and son in a small town close to Copenhagen, Denmark. He is 45-years-old and works as a pedagogical assistant at a nursery. Daniel received a bone marrow transplant in 1985 and had one oral cancer in 2014, the same year he attended his first FA Adult Meeting. Daniel strives to be a positive role model and to show younger people with FA and their parents that there are plenty of reasons to plan for a better future for yourself or your child. FA is serious, but Daniel sees the power of using humor to make life a little bit easier. A self-proclaimed "big child", gaming is a big passion of his. He worked in the gaming industry for years and also used it to raise funds though livestreaming online games. He launched a podcast called "Life, Death, and Happiness" that featured the stories of those living with rare diseases (many guests have FA). Next on his creative endeavors is releasing a Danish rap album in May 2021. Daniel is

committed to living life to the fullest and serving as an inspiration on how to live with FA in a positive way.

Amy is 30-years-old and lives with her fiancé, Matt in Weymouth, Mass., where she currently works as a nanny for two young boys. She was diagnosed with FA in 1995 and underwent an unrelated stem cell transplant in 2002. Recently, she was diagnosed with squamous cell carcinoma and was successfully treated. Amy has been a guest speaker at Dr. Alan D'Andrea's class at Harvard Medical School, where she shared her experiences and perspective as an FA patient. Amy is open about sharing her story, her knowledge about growing up with FA and how it impacts everyday life. Described as positive and spunky, Amy is looking forward to bringing her perspective of living with FA to help bridge connections between patients and scientists. Amy hopes to provide her personal insight for those who are dealing with FA to know that they and their families are not alone and can live happy lives with this diagnosis.

"Are you an adult with FA? Connect with us at info@fanconi.org to gain access to a private support group and upcoming events only for adults with FA."

5th Edition of the Clinical Care Guidelines now available!

The newest edition of the Fanconi Anemia Clinical Care Guidelines (fifth edition) is now available. You can download it free on the FARF website or request a copy for yourself and your doctor.

When you registered with FARF Family Services, you received a handbook about managing Fanconi anemia. This was likely the 4th edition, published in 2014, or maybe one of the previous editions.

The fifth edition is a revision of the fourth edition published in 2014. The contributing authors are physicians or clinical care providers with expertise in treating patients with FA. The fifth edition provides evidence-based recommendations and is geared toward clinical providers as the primary intended audience. Patients and families who wish to secure optimal treatment by improving their understanding of FA may also benefit from this edition.

The *Guidelines* start with a brief summary of the molecular mechanisms of the FA DNA repair pathway and the diagnostic testing process for FA. Subsequent chapters examine more specific health issues faced by people with FA, including hematologic issues, squamous cell carcinoma, oral and dental care, gynecologic care, dermatologic care, gastrointestinal issues, endocrine disorders, hearing and ear issues, and skeletal abnormalities. Finally, the guidelines conclude with a summary of clinical care recommendations for patients with FA.

Learn more at **www.fanconi.org/explore/clinical-care-guidelines**

2021 EVENTS FOR Individuals with FA, Caregivers, and Families

FA CONNECT
LOG ON | LEARN | SHARE

Visit the FARF website for more information on the FA Connect Series: https://www.fanconi.org/calendar

Events have long been a cornerstone for connection and learning for the Fanconi anemia community. Getting together is of huge value to our community. Whether it's adults with FA, families and caregivers, researchers and clinicians, or donors – coming together in support of a shared mission is what makes us so connected and strong. After much deliberation and feedback from stakeholders across the FA community, FARF made the decision to continue hosting virtual events in 2021.

Meet the FA Connect Series: a combination of standalone virtual events for individuals with FA, families and caregivers to learn and receive support from.

What types of events are hosted?

Educational Sessions (60-minute Q&A sessions with expert clinicians or researchers)

Support Groups (90-minute psychosocial support groups or **FA adult meetups**)

FA Connect events are free and hosted through Zoom meeting format on a rolling annual basis. Educational sessions are recorded and posted online in case you cannot attend at that specific time. Live Spanish interpretation is also available during all FA Connect educational sessions. *Note: Support sessions and adult meetups are not recorded or reshared.*

Where can I find the event schedule?

All upcoming events have been posted on the FARF website's event calendar for easy browsing and sign-up. Interested attendees will be able to sign-up ahead of time to prompt a calendar reminder or can easily log in on the meeting date by clicking the posted zoom link.

2021 FA CONNECT EVENT SCHEDULE (MAY – DECEMBER)

MAY

FA Connect Support:
You're Part of the Story – Caregiver Support Group
May 8, 2021 | 9:30 am PT
Allison Breininger, FA Caregiver

FA Connect Support:
FA Resource Swap
May 13, 2021 | 10:00 am PT
Suzanne Planck, Fanconi Anemia Research Fund

FA Connect Support:
Bereavement Workshop: Living with Loss and Grief
May 26, 2021 | 10:00 am PT
Nancy Cincotta, Camp Sunshine at Sebago Lake

JUNE

FA Connect FAdult Meetup:
Adapting to Change
June 18, 2021 | 10:00 am PT
Jack Timperley, FAdult Council Member

FA Connect Support:
Sibling Moments
June 29, 2021 | 10:00 am PT
Nancy Cincotta, Camp Sunshine at Sebago Lake

JULY

FA Connect Q&A:
Nutrition & Metabolism in FA: Tracing nutrients from consumption to energy production
July 13, 2021 | 10:00 am PT
Lindsey E. Romick-Rosendale, Cincinnati Children's Hospital Medical Center

FA Connect Support:
Adults with FA: Life Decisions – Careers, Families, and Medical Care
July 20, 2021 | 10:00 am PT
Nancy Cincotta, Camp Sunshine at Sebago Lake

AUGUST

FA Connect Q&A:
Taking Charge of Your Survivorship
August 12, 2021 | 10:00 am PT
Megan Voss, University of Minnesota

FA Connect Support:
Reflections on Growing up with FA
August 19, 2021 | 10:00 am PT
Nancy Cincotta, Camp Sunshine at Sebago Lake

FA Connect Support:
Bereavement Support for Parents & Caregivers
August 28, 2021 | 10:00 am PT
Rachel Altmann, FARF Board Member; FA Parent

SEPTEMBER

FA Connect Support:
FA Adult Connection Day
September 4, 2021 | TIME TBD

FA Connect Support:
Newly Diagnosed Families & Support within the FA Community
September 20, 2021 | 10:00 am PT
Nancy Cincotta, Camp Sunshine at Sebago Lake

OCTOBER

FA Connect Q&A:
Coloring Outside the Lines: Creative Solutions to Educational Barriers for Children with Chronic Illness
October 7, 2021 | 10:00 am PT
Sarah Steinke and Valerie Theile, Cincinnati Children's Hospital Medical Center

NOVEMBER

FA Connect Q&A:
Talking to Children about FA
November 10, 2021 | 10:00 am PT
Pennie Grubbs and Rebekah Doshi, Cincinnati Children's Hospital Medical Center

FA Connect Support:
Supporting the Others – Involving and Caring for the Children in Your Family Who Don't Have FA
November 17, 2021 | 4:30 pm PT
Allison Breininger, FA Caregiver

DECEMBER

FA Connect Support:
Sharing Talents!
December 6, 2021 | 4:00 pm PT
Nancy Cincotta, Camp Sunshine at Sebago Lake

FA Connect FAdult Meetup:
Topic TBD
December 10, 2021 | Time TBD
Group Facilitator TBD

New space for those dealing with loss

How does one grieve the loss of someone loved so deeply? It's not something we're taught to do. It's not something most people even think about until life is turned upside-down with the loss of a loved one. Whether anticipated or completely unexpected, grief of the death of a loved one is all-consuming and life altering. There is no right or wrong way to grieve; it's a natural, instinctive and universal experience. Even when one might attempt to push out, squash, ignore, and fight grief, it remains, messy and complicated.

Time, community, grace, and love can all help move one through grief to a place that doesn't always feel so dark, desperate and detached. FARF strives to be present for and provide community for the grieving FAmily. We have recently opened a new, private FAmily Grief Support group on Facebook. We hope that if you are anywhere in your grief journey, you'll join us in hearing, supporting and caring for one another as we share stories, remember and honor loved ones, and give and receive support through connection and resources. The group is open to parents, significant others, siblings, and primary caregivers age 18+. International members are welcome.

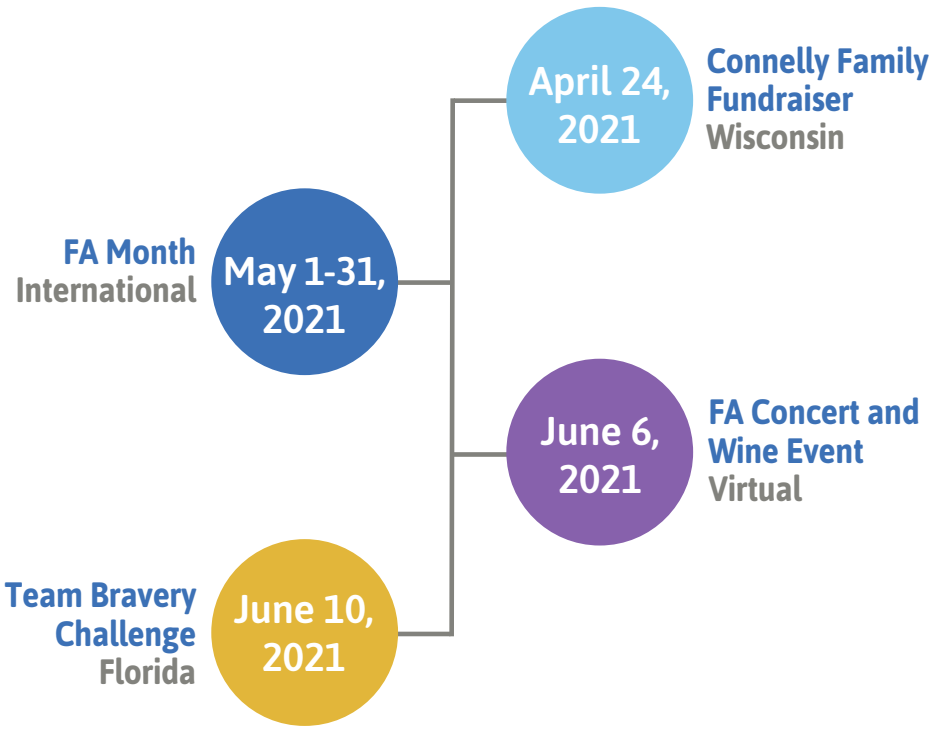
To join the group, go this web address and click "join".

https://www.facebook.com/groups/157783602615192

Questions? Email **suzanne@fanconi.org**

UPCOMING FUNDRAISERS

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 for more information. Do you know of an upcoming fundraiser? Contact us as 541.687.4658 or **info@fanconi.org**.



USE OF LOGO

A reminder to our families with FA: Please use our logo or letterhead only after you have consulted staff at the Fanconi Anemia Research Fund and received approval. This step is necessary to be sure our messages are accurate and consistent, and it helps avoid legal complications. We are happy to collaborate on fundraisers and mailings.

EDITORS' NOTE AND DISCLAIMER

Statements and opinions expressed in this newsletter are those of the authors and not necessarily those of the editors or the Fanconi Anemia Research Fund. Information provided in this newsletter about medications, treatments or products should not be construed as medical instruction or scientific endorsement. *Always consult your physician before taking any action based on this information.*

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RETURN SERVICE REQUESTED

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

HOW YOU CAN HELP

Donations Online:
Donate via the Fund's website (www.fanconi.org)

Donations by Phone:
Call us at 541-687-4658 or toll free at 888-FANCONI (USA only)

Donations by Mail:
360 E. 10th Ave., Suite 201, Eugene, OR 97401

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