FAmily Newsletter

Fall 2017 A semi-annual publication of the Fanconi Anemia Research Fund

issue 62

The Breakdown: Gene Therapy for FA

Gene therapy is all over the news, and rightfully so! Last year the European Medicines Agency (EMA) approved the first successful gene therapy for inherited immunodeficiency, this year the US Food and Drug Administration (FDA) approved the first ever gene-based therapies for cancer, and more approvals are sure to follow in 2018.

Building on these exciting successes, research groups in Spain (CIEMAT, Madrid) and the US (Fred Hutchinson Cancer Center, Seattle) are working hard to create gene therapies for Fanconi anemia. Although still relatively early in development, FA gene therapy could be a significant therapeutic advance, potentially eliminating the need for a bone marrow transplant.

However, there are many technical challenges. FA gene therapy is still investigational (phase 1 and phase 2 trials), has not yet been tested for clinical efficacy, and does not replace current state-of-the-art transplant procedures that are highly effective in FA.

In this article, we briefly describe gene therapy and why this treatment holds promise for FA.

What is gene therapy?

We know that FA is caused by defects in any one of 22 different genes called "FANC" genes. Thus, if we could somehow repair these defective FANC genes, we could treat or prevent FA disease.

Gene therapy is an investigational approach to do just that. The overall idea is to augment or replace defective FA genes with fully functional normal genes. Gene therapy addresses the root cause of inherited diseases like FA by "fixing" or "repairing" the disease-causing gene, enabling cells to produce normal FA proteins that are necessary for health. This should restore normal function to diseased stem cells and prevent bone marrow failure.

continued on page 2

Go, Camp Sunshine!, see story on page 12

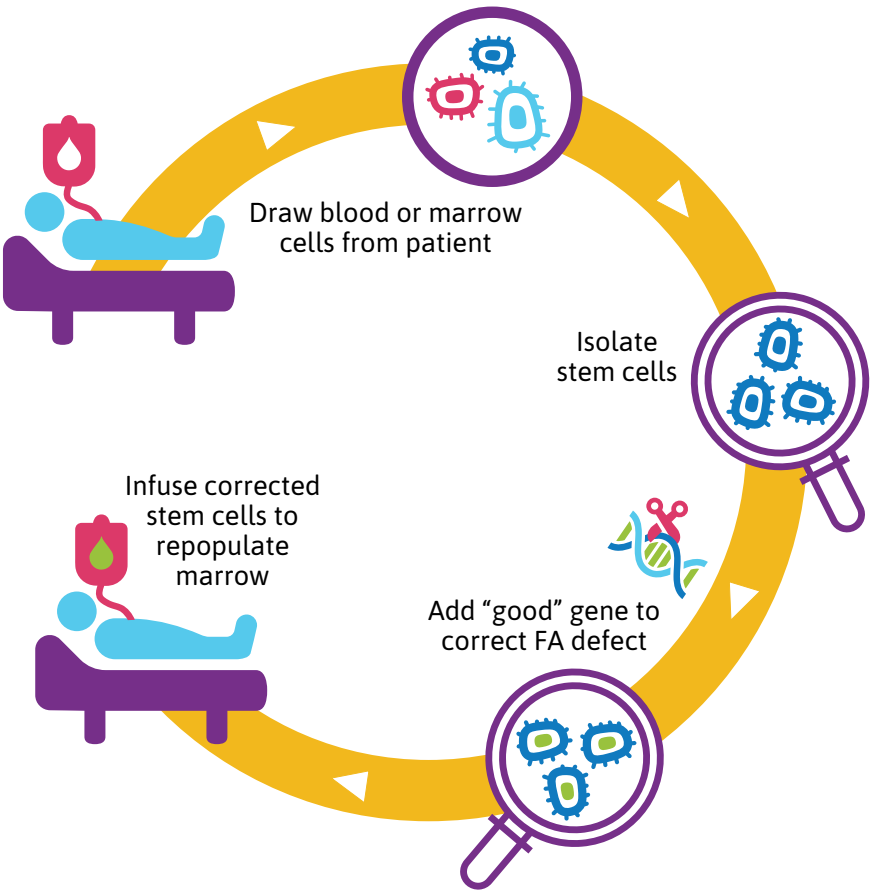
Sarah, Stella, and Calix with a Camp Sunshine volunteer

Gene therapy step-by-step

To deliver a fully functional normal gene into a cell, you have to first decide which cells need therapy. FA usually presents first with bone marrow failure, so we want to fix bone marrow cells to prevent failure. But more specifically, we want to fix a unique type of cell in the bone marrow called a "stem cell". Stem cells have the ability to generate all other types of blood cells in the body: white cells, red cells, platelets, everything! If we can fix the FA defect in stem cells, then all blood cells that come from a stem cell are also fixed.

To achieve this (see figure), blood or bone marrow cells are first drawn from a relatively healthy FA patient. Blood and bone marrow contain all types of cells, including rare stem cells. Stem cells are separated from other blood cells using a fancy stem-cell purification device.

A fully functional normal FA gene ("good" gene) is then added to create essentially normal, non-disease cells. These cells are then infused into the patient as soon as possible because when they are outside the body they risk degradation. After infusion, the repaired stem cells repopulate the patient's bone marrow, where they can generate normal blood cells and thereby prevent or delay bone marrow failure.



This sounds like it should work! Why aren't we using FA gene therapy now?

Well, it's complicated. Science is hard and doesn't always work in practice the same way it works in theory. For one, the stem cell population is reduced in FA, which creates challenges isolating enough stem cells to grow and manipulate in the laboratory. Another challenge is that stem cells are relatively fragile when removed from a patient, and FA stem cells appear particularly difficult to grow in the laboratory. And there is always the challenge of "engraftment"; can the stem cells with repaired FA genes repopulate the bone marrow and outgrow the diseased FA cells that are already there?

The Madrid and Seattle teams are making significant headway to overcome these challenges. Through collaborations, a potential biotech partnership, and open data exchange fostered by the International Fanconi Anemia Gene Therapy Working Group, key advances are happening. This is an area we all need to follow with hope and anticipation. The very best international scientists are working on gene therapy. Breakthroughs are certain, and FA scientists and physicians are at the forefront, well positioned to quickly implement these breakthroughs to improve the health of individuals with FA. ■

Current CLINICAL TRIALS and RESEARCH OPPORTUNITIES

New treatments and therapies for people with FA are not possible without research. Listed below are current clinical trials and available research opportunities. Visit the links to learn more about eligibility and protocol descriptions. If you're interested in participating in a clinical trial, scholarships are available from FARF in order to help offset the cost of transportation and housing. Please contact Marie Sweeten, Family Services Director: marie@fanconi.org or 541-687-4658. The FA Research Fund is not officially affiliated with any of the following opportunities, but encourages families to consider participation.

National Disease Research Interchange (NDRI)

FA researchers are working hard to find effective treatments and a cure for Fanconi anemia, but they can't do it alone. They need you. Researchers need samples to study, such as tumor samples and biopsied tissue. Please consider donating research material. All it takes is a phone call to FARF and completion of paperwork for the National Disease Research Interchange (NDRI).

Contact: Marie Sweeten, FARF
541-687-4658 | marie@fanconi.org

Study of Pembrolizumab (MK-3475) for High Risk Oral Intra-Epithelial Neoplasias

M.D. Anderson Cancer Center, Houston, TX | currently recruiting participants

The goal of this clinical research study is to compare pembrolizumab to standard of care observation (no treatment) in controlling oral pre-malignant lesions. Pembrolizumab is FDA approved and commercially available for the treatment of certain types of melanoma and non-small cell lung cancer. It is currently being used for research purposes in head and neck cancer. FA patients who have not had a transplant and who have a history of oral lesions may be eligible for this trial.

https://clinicaltrials.gov/ct2/show/NCT02882282
Contact: Renata Ferrarotto
713-792-6363 | CR_Study_Registration@mdanderson.org

Eltrombopag for People with Fanconi Anemia

National Heart, Lung, and Blood Institute (NHLBI), Bethesda, MD
Almost open

Objective: To find out if a new drug, eltrombopag, is effective in people with Fanconi anemia and to know how long the drug needs to be given to improve blood counts.

https://clinicaltrials.gov/ct2/show/NCT03206086
Contact: Sophia Grasmeder
301-827-0367 | grasmeders@mail.nih.gov

A Study of Prexasertib in Patients with Solid Tumors with Replicative Stress or Homologous Repair Deficiency

Dana-Farber Cancer Institute, Boston, MA | currently recruiting participants

This is a research study of a checkpoint kinase 1 (CHK1) inhibitor as a possible treatment for advanced solid tumors that harbor genetic alterations in the homologous repair (HR) pathway or with genetic alterations that indicate replication stress.

https://clinicaltrials.gov/ct2/show/NCT02873975
Contact: Geoffrey Shapiro | 617-632-4942
Geoffrey_Shapiro@dfci.harvard.edu

Quercetin in Children with Fanconi Anemia; a Pilot Study

Children's Hospital Medical Center, Cincinnati, OH | currently recruiting participants

This is a pilot study to assess feasibility, toxicity and pharmacokinetics of oral quercetin (a dietary supplement) therapy in patients with FA and is a first step towards a clinical study of the efficacy of quercetin therapy in delaying progression of bone marrow failure in FA.

https://clinicaltrials.gov/ct2/show/NCT01720147
Contact: Stephanie Edwards
513-636-9292 | stephanieL.edwards@cchmc.org

Gene Therapy for Fanconi anemia

Fred Hutchinson Cancer Research Center, Seattle, WA | currently recruiting participants

This pilot clinical trial will access the toxicity and efficacy of infusion of gene modified cells for patients with Fanconi anemia (FA). Infusion of autologous patient blood stem cells that have been corrected in the laboratory by introduction of the normal gene may improve blood counts in patients with FA.

continued on page 5

New Approaches in Stem Cell Transplantation

"Go to a major Fanconi anemia transplant center!" was the strong advice from Margaret MacMillan, MD, University of Minnesota — and not just when it's time for a stem cell transplant. Shortly after the FA diagnosis, when transplant is needed and throughout adulthood, it is crucial to maintain regular contact with transplant experts in FA. These specialists have extensive experience with this disorder and can help guide the overall treatment plan with the help of a team of physicians, surgeons and other health care providers at their center.

Some individuals with FA never need a transplant, so timing of transplant should not be dictated by age but rather by blood counts or early signs of myelodysplastic syndrome or leukemia. Patient outcomes are better before transfusions, infections and before the onset of leukemia.

The University of Minnesota has transplanted over 250 individuals with FA, including 180 with alternative (non-matched sibling) donors. Through a series of different protocols designed to improve outcomes, survival has improved greatly over the years. Five-year survival following alternative donor transplants for low-risk patients is now 94%, but increased age (as with any type of bone marrow transplant), a history of one or more transfusions and life-threatening infections greatly decrease survival.

The University of Minnesota transplants patients of all ages. Thirteen adults have undergone stem cell transplants at the University of Minnesota using their current alternative donor protocol. Survival is approximately 65%. One reason for decreased survival compared to children is that some of these adult patients had inadequate specialized Fanconi anemia care, leading to poor health prior to transplant. Many patients developed leukemia, infections or required multiple transfusions. These results stress the importance of early diagnosis and life-long follow-up with an FA specialist to optimize all aspects of medical care.

A new transplant trial aimed at shortening the time to cord blood engraftment and hastening immune recovery is planned for early 2018 at the University of Minnesota. Cord blood cells will undergo expansion prior to transplant, resulting in many times the number of cells obtained through a single cord blood donation. Time to engraftment could decrease from an average of 17.5 days to six days, greatly hastening immune recovery and perhaps even decreasing the risk of later cancers.

According to Stella Davies, MD, Cincinnati Children's Hospital Medical Center, it is now possible to identify a stem cell donor for everyone needing a transplant. Even transplants using parents who are only half matches for their children have been successful at her center.

Dr. Davies described excellent outcomes using busulfan instead of radiation in the preparatory

> "Some individuals with FA never need a transplant, so timing of transplant should not be dictated by age but rather by blood counts or early signs of myelodysplastic syndrome or leukemia."

regimen. Cincinnati Children's uses a "personalized" regimen, where the amount of conditioning given is based on the age and disease status of the patient. Of 22 FA individuals recently transplanted at her center, 21 survive.

Viral infections can be a threat to survival post-transplant. Cincinnati Children's is now collecting additional T-cells from donors prior to transplant, and training these cells in the laboratory to fight various viral infections. Her institution is growing, freezing and storing these cells in the event they are needed later. To date, this center has successfully used these special cells on one individual with FA.

Dr. Davies described the fertility preservation program at Cincinnati Children's, established in 2009. Fifty-four females getting bone marrow transplants have participated to date. This program includes cryopreservation of embryos, eggs, ovarian tissue and testicular tissue.

Of tremendous interest to Dr. Davies is her ongoing FA trial using quercetin, an antioxidant, in an effort to prevent additional DNA damage and stabilize blood counts. The question she now wants to explore is whether quercetin could possibly prevent head and neck cancer in individuals with FA. Twelve FA patients, ages 4-21, have enrolled in her dose escalation quercetin trial. There have been no negative side effects. The initial dose of 750 mg/day has escalated to 4000 mg/day. All of the children taking the high dosage experienced a reduction in oxidants in their blood. **In the next 6-9 months, Dr. Davies will initiate a patient study to determine if long-term quercetin use can prevent cancer in FA individuals. There will be no age limit to this study.**

continued from page 3

https://clinicaltrials.gov/ct2/show/NCT01331018
Contact: Pamela S. Becker 206-288-7234 | pbecker @u.washington.edu

Lentiviral-mediated Gene Therapy of Fanconi Anemia Patients Subtype A (FANCOLEN-1)

Hospital Infantil Universitario Niño Jesús, Madrid, Spain & Hospital Vall d'Hebron, Barcelona, Spain | currently recruiting participants

This is an open clinical trial to evaluate the safety and efficacy of a hematopoietic gene therapy procedure with an orphan drug consisting of a lentiviral vector carrying the FANCA gene for patients with Fanconi anemia of subtype A.

https://clinicaltrials.gov/ct2/show/NCT03157804
Contact: Julian Sevilla | +34 915035938 | julian.sevilla @salud.madrid.org

Cancer in Inherited Bone Marrow Failure Syndromes

National Cancer Institute (NCI), Bethesda, MD | currently recruiting participants

This is a study to provide information regarding cancer rates and types in inherited bone marrow failure syndromes (IBMFS), including Fanconi anemia. It is a natural history study, with questionnaires, clinical evaluations, clinical and research laboratory tests, review of medical records, and cancer surveillance.

https://clinicaltrials.gov/ct2/show/NCT00027274
Contact: Blanche P. Alter 240-276-7239 | alterb@mail.nih.gov

TRIALS vs. TREATMENT

It is critical to understand the difference between medical treatments and clinical trials. A medical treatment is a regimen specific to an individual patient and his/her condition, administered by doctors. A trial tests a potential drug, procedure, or medical device in people. Participants in trials play an integral role in determining the safety and efficacy of drugs or procedures. It is important to remember that clinical trials are meant for research, not to administer proven medical care.

Even though a medication may be approved for one condition or disease, it must be tested in the new population it is meant to help. It's vital to conduct research in many people, because people may respond differently to the same treatment. Self-treatment with medications that have not been approved for a specific population/condition can be harmful to the individual; it may also hinder knowledge of the appropriate therapeutic use and benefit(s) of the medication. Always consult your physician before taking any action regarding medications or treatments.

Transplant: Frequently Asked Questions

Always consult your physician before taking any action based on the information presented on this page.

What is a Hematopoietic Stem Cell Transplant (HSCT)?

An HSCT is a medical procedure that destroys the stem cells in a patient's marrow and replaces them with stem cells from a matched or partially matched related or unrelated donor. The closer the match, the less likely that the new stem cells will recognize the patient's cells as foreign and attack them, a complication known as graft-versus-host disease (GvHD). At the present time, stem cell transplantation is the only long-term cure for the blood defects in FA. Stem cells can be taken from a donor's bone marrow or peripheral blood, or can be obtained from cord blood harvested at the time of a baby's birth. To prepare for transplant, the patient's own bone marrow is destroyed, making space for the new, healthy stem cells to engraft.

Box A

Pre-transplant laboratory tests

- Confirmatory diagnostic testing for FA (DEB or MMC most commonly)
- Confirmatory HLA typing
- Bone marrow aspirate and biopsy with cytogenetic evaluation
- Infectious disease assessments
  - Prior exposures (cytomegalovirus; hepatitis A, B and C; HIV; HTLV1/2; EBV; syphilis)
  - Presence of active infections (CT scan of sinuses, chest, and abdomen; dental evaluation)
- Organ function assessments
  - Lung (pulmonary function tests, oxygen saturation)
  - Heart (EKG, echocardiogram)
  - Liver (liver enzymes, ultrasound)
  - Kidney (chemistries, nuclear medicine studies such as glomerular filtration rate or GFR, ultrasound)

How do we know when/if it's time to go to transplant?

It is important to note that not every person with FA will need a transplant. Although likely, it is not a certainty. Someone needs a transplant when his/her counts are low enough to require it*. If a patient with FA appears to be a good candidate for transplant based on history and physical examination, a number of routine tests should be performed immediately prior to transplant to verify eligibility and to determine if any adjustments are needed in the treatment. For example, poor kidney function could result in important drug dose adjustments or an anomaly on chest CT might result in additional evaluations, antibiotics, or delay in transplant until resolved. See Box A for a list of the types of tests performed at most transplant centers.

*More info? For specific information on counts and indications that it's time for transplant, see pgs. 223-224 of the Fanconi Anemia Guidelines for Diagnosis and Management.

Are there alternatives to transplant?

While transplant is generally recommended as first-line therapy for bone marrow failure, MDS or leukemia in patients with FA, there are a couple of alternatives for delaying transplant or for patients considered too 'high risk' to undergo transplant therapy. These alternatives may be androgens or hematopoietic growth factors. Gene therapy trials (for FANCA) are also open on two continents to determine if the patient's marrow may be corrected by this method. The patient and family should discuss the risks and benefits of alternative approaches with the hematologists at an FA comprehensive care center.

See p. 238 in the Guidelines book for more info on HSCT alternatives.

Where should we go for transplant?

Because of the unique complications associated with HSCT and the late effects associated with FA itself, it is recommended that whenever possible, patients be cared for at selected centers with comprehensive care clinics specific to FA. Though only a few of these specialized centers exist worldwide, patients who travel to these centers help advance

FA research as much as they themselves benefit from the centers' comprehensive care. The dramatic improvements in transplantation for patients with FA over the past decades, for example, would not have been possible without research that benefited from the concentration of patients at a few centers. Treating patients at selected centers may also help clinicians and researchers improve the management of FA-associated conditions that develop later in life, particularly cancer. See Box B for some useful questions to consider when choosing where to seek treatment. More info? See pgs. 224-229 in the Guidelines book for more information.

What are the success rates of transplant?

Many factors influence the likelihood that a transplant will be successful. Younger children with marrow aplasia (no myelodysplastic syndrome [MDS] or leukemia) do very well, with a better than 90% chance of a good outcome. Transplants are harder in older people and people with MDS or acute myeloid leukemia (AML), or people who have abnormal organ function or past infections. Your doctor will be able to tell you what the chances for success would be in your individual case.

How long will transplant take?

The actual transfer of cells usually takes less than 10 minutes. Most transplant centers will expect the patient to remain near the facility for a minimum of 100 days. While complications can occur after this period, the first 100 days are considered the highest risk period associated with HSCT. During the initial hospitalization for the transplant procedure, patients are isolated (to a room) to reduce exposure to infectious agents. Once the marrow has recovered sufficiently, patients are allowed out of their hospital rooms unless intervening problems prevent this.

Factors that influence the length of time spent away from home include the number of transplant complications such as GvHD and infections, access to a BMT facility closer to the patient's home, the comfort of the referring physician, and evidence of immune recovery. These factors should be discussed on a case-by-case basis.

Ok, it's time to go to transplant. What about logistics, like insurance and a place to stay while getting treatment?

If the insurance company is associated with a transplant center that has limited or no expertise in FA, the insurance company will often give approval for the patient to travel to an experienced FA center once the insurance company understands the differences in the centers' experience and the importance of experience in patient survival. Insurance denials or less than complete coverage for transplant at an FA-experienced transplant center (because they are "out-of-network") can often be contested successfully. FA centers can help with getting insurance approval.

Other FA families are excellent resources when it comes to navigating insurance issues as well as other practicalities like lodging during transplant. If you have a specific question, put it out there on the Family Support Group on Facebook for suggestions from others. Marie Sweeten, FARF Family Services Director, is also available to help answer questions.

Box B

Questions to help assess a transplant center's experience with FA.

- What is the total number of transplants that the center has performed specifically in patients with FA?
- How many FA transplants have been performed each year for the past 5 years? How many of those patients are still alive?
- What treatment regimen do you propose? Please tell me the exact doses of each drug and the radiation dose (if applicable). How many patients have been treated with this regimen at this center? How many are still alive?
- What is the risk of acute and chronic GvHD in FA patients using this regimen? How do you plan to prevent GvHD?
- How long will you follow the patient (me/my child/my spouse)? Who will follow the patient (me/my child/my spouse) long term?

How can I learn more?

Of course, speaking with your doctor(s) is the best way to educate yourself and make decisions. The Fanconi Anemia: Guidelines for Diagnosis and Management are also an excellent resource. The complete Guidelines are available at www.fanconi.org or can be mailed to you. Request your copy(ies) at info@fanconi.org. Finally, updates in stem cell transplantation are provided every year at the Family Meeting at Camp Sunshine in June (see p. 12 to learn more, including how to apply). ■

Research

The path to a cure for FA is a long and winding one, with many different routes and detours. It all starts with research. Each idea or concept builds on the others, taking us closer and closer to better treatments and a cure. Many of today's treatment protocols began 25 years ago as new research ideas funded by FARF.

1 Discovery or idea

2 Fundraising for research

3 Development of drug or protocol

4 Research is 'translated' for preclinical testing

5 Clinical trial

6 New treatments for people with FA!

Dr. KJ Patel, FARF researcher

Treatments

The Breakdown: Endocrine Issues in FA

What is the endocrine system?

The endocrine system produces hormones that allow our bodies to develop and function. This system consists of glands in the head, neck and abdomen that release many different types of hormones into the bloodstream. These hormones perform a variety of functions in the body, from regulating blood sugar levels to triggering physical changes during puberty. Think about it this way: endocrine cells make a hormone, or message. These "messages" are carried in the bloodstream to other cells, telling the body to grow taller, for example, or to go through puberty.

How is the endocrine system affected in FA?

The DNA damage caused by FA leads to death of some endocrine cells. This loss of endocrine cells results in lower hormone levels. About 80% of children and adults with FA have an endocrine abnormality. These abnormalities can affect the body in a variety of ways:

- Short stature
- Poor weight gain, or overweight status
- Abnormal glucose with low insulin secretion (can contribute to poor weight gain or diabetes)
- Hypothyroidism (low levels of thyroid hormone, causing poor height growth, delayed puberty, irregular periods, difficulty becoming pregnant)
- Early or late puberty, underactive testes or ovaries, infertility
- Growth hormone (GH) deficiency
- Low bone mineral density

How can these issues be managed?

In FA we talk a lot about comprehensive care, and that is because FA is a disease that affects multiple systems in the body. For this reason, it is important to involve an endocrinologist or pediatric endocrinologist early, a dietician, and for females a gynecologist or reproductive endocrinologist. This team should work in close collaboration with other FA specialists to provide comprehensive care.

According to Dr. Susan Rose, MD, endocrinologist and FA specialist (Cincinnati Children's Hospital Medical Center/University of Cincinnati), people with FA should have **annual growth screenings** to track height and weight, identify and treat any causes for poor growth, assess the thyroid and treat any deficiencies of thyroid hormone, vitamin D, GH, or pubertal hormone.

Maintaining a healthy diet and exercise is important and a **dietician should assess the person's nutritional intake**. If a child with FA is short, s/he should be **screened for growth hormone deficiency** (GHD). Growth hormone therapy may be used to treat GHD, though there is no current consensus on safety of this therapy in FA. After age 10 years (or younger if puberty started early), an **annual puberty screening** is recommended to assess hormone levels and to then determine whether treatment is needed for either early or delayed puberty. Finally, after bone marrow transplant or after age 16 years (whichever comes sooner), an annual bone screening to check bone mineral density is recommended. ■

For a full, detailed list of endocrine screening recommendations for patients with FA, see p. 149 in the Guidelines for Diagnosis and Management. Chapter 7 is this book is dedicated entirely to the endocrine system in FA. Also, check out the 2014 Fall Family Newsletter (p. 6) for another detailed summary on endocrine issues (available on our website: www.fanconi.org).

Sun Protection for People with FA

(and Everyone Else, Too!)

Slop

Slip

Some background on dermatologic issues in FA

Skin abnormalities (such as altered skin pigmentation either overall or in spots) can suggest Fanconi anemia, and other skin abnormalities may emerge as children with FA become adults. Skin problems may suggest Sweet's syndrome, which presents as red sores, but is often caused by a serious underlying bone marrow syndrome called myelodysplastic syndrome (MDS). It is essential to learn about sun protection and cancer prevention because some skin abnormalities are linked to sun exposure. These include basal or squamous cell carcinoma, actinic keratosis (scaly patches), and melanoma.

The sun is not your friend

At the Family Meeting in June, Dr. Christina Boull, MD, University of Minnesota, reminded families that "the sun is not your friend!" It causes premature aging and skin cancer (in anybody). What about Vitamin D? It *is* an essential nutrient, but because of the harmful UV radiation associated with the sun, Vitamin D should be obtained through dietary sources and supplementation rather than from sunlight.

You may think, "Between work and home, I don't actually spend that much time in the sun." Today many of us get the bulk of our sun exposure riding in our cars, which has become the 'new outdoors.' How much time do you spend in a car? Dr. Boull reiterated that skin cancer is the most common type of cancer and that more sun exposure in early childhood can dramatically increase the risk of skin cancer in adulthood.

> **"Most of us get the bulk of our sun exposure riding in our cars."**

What about in FA?

The risk for skin cancer for people with FA is suspected to be higher, since known risk factors include inability to repair DNA and complications related to bone marrow transplant, including radiation, chemotherapy, graft-versus-host disease, and immune suppression. A handful of people with FA have reported skin cancer, which suggests the importance of prevention and developing good sun and skincare habits from a young age.

How can I protect my/my child/ my spouse's skin?

Wear sunscreen

- Dr. Boull recommends using a sunscreen that includes ingredients like zinc oxide or titanium dioxide. Zinc and titanium are inert minerals that just sit on the outside of the skin. They do not have carcinogenic properties and tend to cause less irritation and skin allergies than the chemical sunscreens. They are safe in all ages including infants.
- Spray-on sunscreen doesn't get as good of coverage as a cream or lotion. There is an unknown inhalation risk, so it's better to go for the cream.
- Use sunscreen with an SPF (sun protection factor) of 30 or higher. Remember that higher is not always better; some companies market sunscreens with SPF 100+, but really, those are not much more effective than SPF 30 because they only block about 2% more of the sun's rays.
- Reapply approximately every two hours, even on cloudy days.

- If swimming or sweating, sunscreen needs to be applied more often.

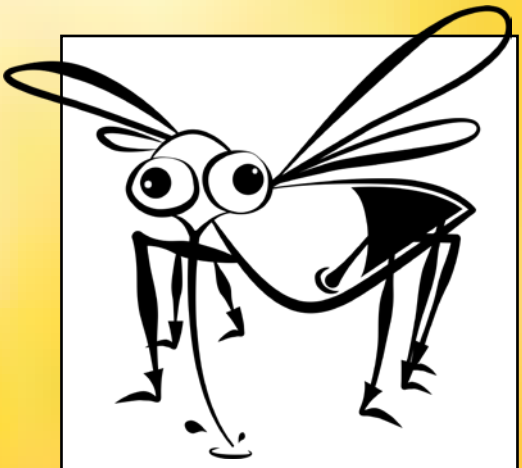
Wear protective clothing

- Look for UV protection factor (UPF) of 50 or higher when buying swim shirts and other sun protective clothing.
- Remember SLIP, SLOP, SLAP, SEEK, and SLIDE. **Slip** on protective clothing, **slop** on some sunscreen, **slap** on a hat (wide-brimmed, at least two inches all the way around), **seek** shade (check the ultra violet index on your weather app. If it's above three, it's better to stay out of the sun) and **slide** on sunglasses (make them big).

Get a skin check

- *How often?* According to Dr. Boull, skin should be examined annually if the person with FA is post bone marrow transplant.
- *At what age?* Dr. Boull suggests starting at a young age, like 6-8, because it builds good habits. Every case is different, so it is important to consult with your/your child's doctor for individual recommendations. ■

Chapter 9 in the *Guidelines for Diagnosis and Management* book is dedicated entirely to dermatologic issues in FA.



What about insect repellent?

It is important to protect against numerous diseases transmitted by insects. In her presentation, Dr. Boull asserted that DEET is most effective, and according to the Environmental Protection Agency, DEET is **not** a carcinogen**.** It is a neurotoxin when used in high doses, so combination sunscreen/insect repellant products should not be used. The requirement of frequent application for the sunscreen component would result in excess exposure to DEET.

Go, Camp Sunshine!

FARF brings FA families together at 26th annual meeting in Casco, Maine

In July of 1991, more than 100 FA parents and children attended the very first "FA Family Symposium" in Washington, D.C. This gathering marked the beginning of what would become a cornerstone of the FA community: an annual meeting of individuals with FA, their families, and physicians and researchers with the purpose of forming connections and improving the lives of those affected by FA.

Twenty-six years later, in June 2017, nearly 230 people with FA, family members, and FA experts gathered at Camp Sunshine in Casco, Maine for five days of educational sessions, support groups, and irreplaceable bonding opportunities. Fifty-two FA families from four countries attended. Among this group were 53 individuals with FA (including 14 adults) ranging in age from one year to 38 years. In addition, more than 100 wonderful Camp Sunshine volunteers and staff worked hard to ensure that the FA Family Meeting ran smoothly and successfully.

Each year, expert scientists and clinicians share findings from their research and updates in treatment protocols. This year, 14 speakers gave updates on transplantation (p. 4), endocrine issues (p. 9), nutrition, gene therapy (p. 1), TGFß inhibitors in FA treatment, dermatological care (p. 10), head and neck cancer treatment, fundraising, and currently funded FARF research and clinical trials (p. 3).

Additionally, those with FA were invited to participate in a number of research opportunities throughout the week, including oral cancer screenings, dermatological screenings, and the

> "Words cannot describe our experience. It's what we look forward to every year."

52
families

4
countries

14
speakers

53
people with FA

> **“Inspiring, and a fantastic place for making life-long friends and connections. I always leave wishing I had just one more day at the camp with these inspiring families and absolutely incredible volunteers.”**

Fun fact: 8% of this year's FA campers are named Eli!

National Institutes of Health's Inherited Bone Marrow Failure Syndromes Study. All of this is made possible by fundraising dollars. Following the meeting, one parent remarked: "It is *so* awesome knowing that our family fundraising efforts are translating into progress toward a cure!"

Apart from scientific sessions, attendees are invited to participate in a number of support sessions with Camp Sunshine Psychosocial Director Nancy Cincotta, MSW, MPhil. These support sessions are geared to specific groups such as adults with FA, spouses, children with FA, siblings, and parents (including a special group for bereaved parents).

Of course, the Family Meeting would not be complete without the many social activities such as swimming, mini golf, the ropes course, and rock climbing wall, as well as special events like karaoke night, the talent show, masquerade ball, and sleep out.

The FA Research Fund would like to thank the Camp Sunshine team for their years of excellent coordination and tremendous job hosting Fanconi Anemia week. In the words of one attendee, "it's better than Disneyland! We wouldn't miss it and hopefully we'll never have to!"

What is the Family Meeting?

The Family Meeting is a unique event that allows families registered with FARF to attend presentations by researchers and physicians who are active in the research and treatment of Fanconi anemia (FA), to attend support groups for help in coping with the disease, to voluntarily participate in FA research projects, and to connect with other families affected by FA. Camp Sunshine volunteers provide a fun-filled program for children with FA and their siblings.

What is Camp Sunshine?

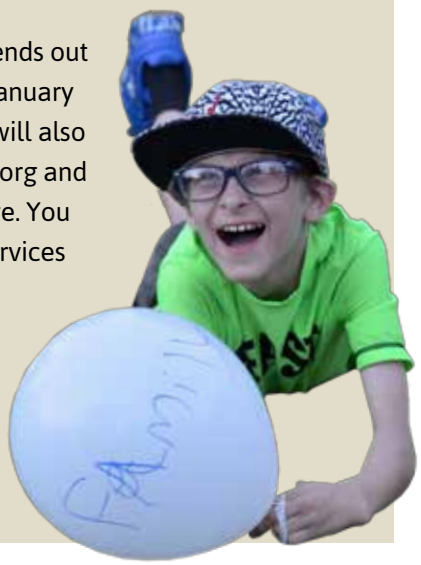
Camp Sunshine provides retreats combining respite, recreation and support, while enabling hope and promoting joy, for children with life-threatening illnesses and their families through the various stages of a child's illness. The program is free of charge to families and includes on-site medical and psychosocial support. Bereavement sessions are also offered for families who have experienced the death of a child. Learn more at www.campsunshine.org.

Who can go?

Anyone with FA and his/her family are welcome and invited to apply. There is no age limit. People from all countries are welcome. Newly diagnosed families are especially encouraged to apply. There is no cost to attend other than travel expenses. Travel scholarships are available through FARF to assist with these expenses.

How do I apply?

The FA Research Fund sends out an invitation via email in January of each year. Information will also be posted at www.fanconi.org and on the FARF Facebook page. You can also contact Family Services Director Marie Sweeten (marie@fanconi.org) with any questions. The FA session usually takes place in late June. ■



Chloe, Sarah, Eli and David Borden (l to r)

From Fear to FAmily

By David Borden

I will never forget May 15, 2017. It was the day our geneticist called to tell me that my son had Fanconi anemia and what that meant for him and for our family. I was in shock. This couldn't be real. A few months earlier, our geneticist told us this test was to *rule out* FA – that she didn't think our son had this disorder. The gene came up on an exome sequencing for "unknown significance." She didn't even spend much time explaining what it was during the last appointment. She said it was a terrible disease that she didn't think Eli had, but that it would be smart to rule it out. We almost didn't take the expensive DEB test (diepoxybutane, which tests for FA).

We had spent years working through various medical issues and finally thought most of it was behind us. Our local children's hospital seemed to be baffled by Eli for years, which left us feeling terrified about our future. My mind raced as the geneticist started rattling off outdated information and figures...90% fatality by age 18 is the only number I remember. Then she instructed me to call the local hematology department and make an appointment as soon as possible. I had never felt so alone and terrified for my wife to come home from work, as I would soon share with her what I had learned.

After the call, I started frantically researching. Wherever I went searching for FA, I found the Fanconi Anemia Research Fund (FARF). Later that same evening, FARF Family Services Director Marie Sweeten returned my emails with answers to **ALL** of my questions. She provided a list of doctors who knew about FA and told us about a camp we should attend just a few weeks later. Suddenly we were not so lost. We had a plan. We were moving forward.

I tried to call our local children's hospital to set up an appointment with the doctor on the list FARF gave me, but I was told that doctor only sees patients in bone marrow failure. I told them we just were diagnosed with Fanconi anemia and that we wanted to see the FA specialist. We ended up with an appointment with a regular hematologist. This frustrated me, so we cancelled the appointment. A few weeks later, we found ourselves in Minnesota for a full FA workup to find out where we stood. Another few weeks after that, we were traveling to Camp Sunshine in Maine, our first ever family road trip.

It was at Camp Sunshine where we found the courage to face FA head on. While at times we felt very overwhelmed, we never felt alone. The outpouring of love and support from the other families made us feel welcome, gave us strength, and filled us with courage. It was at camp that we realized that this terrible, ugly, dark disease has a silver lining, if you can believe that. It comes along with some of the strongest and most caring parents and children we have ever met. You know when you meet someone like this because that person is electrifying to be around. It's one thing to meet one person like this, but we got to go to a place filled with people like this. What an incredible experience we will not soon forget! We miss our new FAmily and already look forward to next year.

> "The outpouring of love and support from the other families made us feel welcome, gave us strength, and filled us with courage."

The keystone to all of this was FARF. Without FARF, none of this would have been possible. We would still be lost and filled with sadness and despair. We will never forget what FARF has done for our family and the other FA families. Thank you from the bottom of our hearts!

Mary-Beth Johnson

Heart of Celebration

By Mary-Beth Johnson

My name is Mary-Beth Johnson. I'm 25, and I have Fanconi anemia. I climb mountains, make avocado roses, and I spend a lot of money at Target when I'm stressed. I don't generally identify as someone who is sick, because I'm healthy for the most part. I had an older brother, Danny, who died of FA over 20 years ago. Ever since then, I've balanced a great isolating act when it comes to the FA community.

If I'm being completely honest, I was peer pressured to attend the adult meeting this year (looking at you, Marie Sweeten). I went in silently, kicking and screaming, convinced that I could remain stoic and detached...that I was there for the science, and to learn how to boss my doctors around better.

But here's the thing: there's a magnetic quality to other people who share parts of your story. The anxiety, fear, the memorized CBC numbers. The feeling that you need to squeeze as much life into your years as possible. The necessity to love, love, love and rise, rise, rise and continually chase after joy in the face of uncertainty.

I won't lie to you: I had a lot of feelings about the adult meeting. I called my husband every night and started our conversations with, "I've had 37 meltdowns, but at least half of them were good." And here's why: there is no better antidote for isolation than to find your heart in other people. Maybe that sounds melodramatic (my brain operates at a base level soap opera), but when you're an adult with FA it can be hard to explain the crossroads where you live.

As an adult with FA, I want a lot of things for my life. I want to have a successful career. I want to be generous, hopeful, and life-giving for the people around me. I want a family. I want to travel the world with the love of my life. I want old age. GOSH, I want old age. Give me the wrinkles, age spots, the Velcro sneakers; I crave sassy grandma status.

And yet, as an adult with FA, I also keep a list of my symptoms. I message my doctors every other week. I force them to check and recheck for cancers. I pay way too much money just to be sure I'm still healthy. I lose sleep sometimes, imagining what it will be like when my waiting is over and I finally get a diagnosis.

So you see, when I walked into a room with 41 other adults who are thriving in their present, dreaming and working for their futures, and claiming control over their health, I felt a new part of my heart.

I am indebted to the doctors, researchers, and scientists who are fighting for us. Indebted to the parents and caretakers—lay people who became medical experts—who have fought for and championed our health. Indebted to the spouses and partners, who stepped into our stories of FA and forever changed the way we understand unconditional love. Indebted to the FARF staff and board, for taking our orphan disease and giving us hope.

But mostly, and forever, I will be indebted to the adults with FA. For creating a community of generosity and hope. For sharing the pit of feelings. And for finding the resilience to climb out of the pit, with arms reaching down to pull others up, and sit around a pool late into the night sharing life, laughing at morbid jokes, and squashing gigantic Georgia cockroaches. ■

Check out Mary-Beth's incredible food blog at **www.heartofcelebration.com** and find her on Instagram: **@heartofcelebration**

Mariana and Mary Jo Becerra

Finding the Sunlight

By Mary Jo Becerra

Our life as FAmily began in 2010 with the diagnosis of our son, Israel. This news came two months after Israel's cousin Abid, who also had Fanconi anemia, passed away due to complications from his bone marrow transplant (BMT). In one day, my boy went from being a child who was small for his age and had only one kidney and some developmental delays, to having a life-threatening disease that just claimed the life of his cousin.

Once we came to grips with his diagnosis, we connected with the Fanconi Anemia Research Fund (FARF) and started making plans for him to see the FA specialists at the University of Minnesota. Over the next three and a half years, we watched his counts slowly decline. Also during this time, our family grew. Despite knowing that we had a 25% chance of having another child with FA, I felt strongly that Israel should have a sibling. In 2011, we welcomed Isaac and learned not long after he was born that he did not have FA. My precocious Israel kept insisting he wanted a sister and he got his wish in September 2012. By October, she wasn't growing well and an ultrasound showed an extra thumb on her right hand so they concluded she likely had FA. Mariana was born at 37 weeks weighing just 4 pounds, 5 ounces. Her official FA diagnosis, just two weeks after her birth, brought sadness, but not regret.

In July 2014, I received the call I had been dreading for four years...Israel was in complete bone marrow failure and it was time for his transplant. We all moved to Minneapolis and he had his transplant on October 28, 2014. Israel's transplant was a success in that he engrafted, but he was plagued by engraftment syndrome, hemorrhagic cystitis, graft-versus-host-disease, and pneumonia. On June 28, 2015, the eight month anniversary of his transplant, I held Israel as he took his last breath.

When I think about all Israel endured, I recall a little boy with more courage than I have ever seen displayed in my life. He didn't always like what he had to do for his various treatments, but he did it. I witnessed this same courage and resilience in my then three-year-old daughter Mariana during her transplant.

Mariana's counts precipitously declined in the year following Israel's death and in August 2016 I again received the dreaded call informing us that it was time for her transplant. I felt ill-prepared to return to the BMT unit so soon after being there with Israel. Her doctor was hopeful and assured me her transplant course could be different from Israel's because her brother Isaac was her donor.

Mariana received her transplant on December 14, 2016. Her post-transplant course was uncomplicated and we returned home as scheduled after she reached Day +100. She was cleared to start school this fall and is thriving in her pre-K class. My heart is so very grateful for how well she is doing, but I continue to worry about her. Overcoming transplant is just the beginning for her.

Despite my worries, I hold on to my faith. Prior to Israel's transplant, a friend shared with me the verse Joshua 1:9—*Be Strong and Courageous, do not be afraid, do not be discouraged, for the Lord your God is with you wherever you go*. At times, it is very hard to be strong and courageous, but my faith that God is with me wherever I go

helped me during my children's transplants and continues to sustain me to this day.

Our family was loved, supported and encouraged throughout the transplant journeys of both Israel and Mariana. I now feel called to do the same for other families like ours. Before Israel died, he asked me if he could plant sunflowers, but we were never able to honor his wish. I believe this request was a gift from him to me, for now sunflowers represent him in my life. It is my heart's mission to honor Israel and his memory by helping BMT families be like sunflowers so that even on the hardest days, the darkest days, they can stand tall and find the sunlight.

Our primary mission is to support and encourage families who have a child undergoing bone marrow transplant by providing gift cards to assist in off-setting the expenses of relocation and the extended hospital stay. Our secondary mission is to support research efforts into finding effective treatments and a cure for Fanconi anemia, so 10% of all the funds we raise goes go to the Fanconi Anemia Research Fund. Visit Fb.me/aimstronginc to learn how you can help!

The Amy Winn and Christopher T. Byrd Award for Adults with Fanconi Anemia is Presented to Matthew Pearl

In early 2017, the FARF Board of Directors created the Amy Winn and Christopher T. Byrd Award for Adults with Fanconi Anemia. This award honors former board members Chris and Amy, who set high goals, devoted their time and energy to making a positive difference, and lived their lives enthusiastically in spite of the challenge of FA. Matthew Pearl, a motivated and inspiring young leader, received the honor and the $5,000 award. Matt has embraced the challenges of FA and turned them into lessons to guide his life positively and meaningfully. He explains that FA has shaped the way he lives and has led him to identify some guiding rules to a purposeful life. See those lessons below. FARF Executive Director Mark Quinlan presented the award to Matt at the Annual Family Meeting in Casco, Maine on June 26, 2017. On behalf of the FARF board and staff, congratulations, Matt! To view Matt's inspirational submission video, visit http://bit.ly/2u7dsXl

The Building of a Community

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

What does it mean to grow up with Fanconi anemia? Is it about growing up with FA or growing into it?

In 2017, someone with Fanconi anemia may still not meet anyone else with FA before coming to a FARF meeting. At the meeting for adults with FA in Atlanta this year, there were several such situations. Each first encounter evokes how lonely the journey can be prior to connecting with the FARF and finding friendship in the FA community.

> **"Adults with FA are trailblazers, finding their way in relationships, making day-to-day decisions, and taking responsibility for themselves and their medical care."**

When a group of people with FA and their families come together, it feels more like a community than a place of casual connection. Since the disease is so rare, to meet others on the same journey (for the first time) generates an automatic, existential connection. Some young adults with FA, who have grown up with each other at Camp Sunshine, have formed unique bonds. The mutual support of the community formed in these spaces mitigates against the uncertainty of the future. Friendships begin at a different level when FA is the common denominator. Researchers, families, FARF staff, board members, and other professionals add to the sense of a unique, embedded community. Choosing to deal with the challenge of FA proactively enables the foundation upon which this cohort is built and unified.

For teens and younger children with FA and their families, it is important to know that the meeting in Atlanta had 42 adults in attendance and that the sense of promise was unequalled. The ease with which people connected with one another, their camaraderie, and their inquisitiveness can help younger generations to understand that they are a part of a growing movement filled with hope, strength, and support.

There is a new reality on the horizon. Searching for a cure comes with the caveat of finding the best quality of life until there is a cure, which is truly learning to live with FA. Adults with FA are trailblazers, finding their way in relationships, making day-to-day decisions, and taking responsibility for themselves and their medical care. All of it is routine, yet all of it is ground-breaking, which in turn, is exceptional.

I had the privilege of meeting with everyone attending the Atlanta meeting, at first all together, and then separately with

adults with FA, their partners, and their parents. Adults expressed their own issues and concerns for their partners and their parents. Hearing and learning about FA from the vantage point of a partner was not just interesting, but inspiring. The dialogue, which begins with "when and how" to tell a potential partner about FA, ultimately gets to negotiating the complexities between parents and partners as "caregivers" surrounding the medical journey. The whole concept of "partnership" is a welcome challenge filled with an extraordinary sense of love.

Towards the end of the meeting, one young woman noted that she was usually frightened when going for scans, but after this meeting she did not feel frightened. The support, honesty, and openness when talking about fears, aspirations, and the true FA narrative is another hallmark of this community.

Reflections from some volunteers who attended the summer family meeting spoke to a perception of the remarkable resilience seen in this cohort, and the lessons that FA families embody about humanity and humility. Can resilience be built? It seems as though the support, information, ongoing generation of knowledge, and the ability to change science are all having an impact on people living with FA. It is that resilience, the ability to endure, and the hope for the future that are forming the culture of a truly remarkable community. ■

> **"The ease with which [adults with FA] connected with one another, their camaraderie, and their inquisitiveness can help younger generations understand that they are a part of a growing movement filled with hope, strength, and support."**

In Loving Memory

Alcide Costard .....3.23.10 – 4.16.17

Josh Seibert.....11.26.79 – 5.4.17

Yan Keok .....9.21.81 – 5.15.17

Hope Burkin..... 5.9.00 – 6.7.17

Carson Vitrano .....7.22.03 – 7.7.17

Driscol Pennell.....2.17.14 – 7.15.17

Anthony Miller.....2.6.96 – 9.21.17

FANCONI ANEMIA RESEARCH FUND
Advancing Research. Supporting Families.

Dear FARF FAmily,

I am truly in awe at the devotion of the many people who have embraced the mission of the Fanconi Anemia Research Fund and made it their own. Researchers, clinicians, FA families, staff, and volunteers are all working toward the same vision: finding a cure for Fanconi anemia.

2017 has been a very exciting year for our community. We awarded several research grants to world-class institutions, including the Dana Farber Cancer Institute, Stanford University, Boston's Children's Hospital, Ohio State University, and Yale University. We held another meaningful Family Meeting at Camp Sunshine in June, bringing together more than 50 families for educational and support sessions. In September, for the first time, we held our Scientific Symposium and our Meeting for Adults with FA over the same weekend, allowing the two groups to glimpse into the other's world in a significant way.

As members of the FA community, we are working toward the same goal – to fulfill FARF's vision – so it makes sense that we should all be moving in the same direction. As we set our sights toward the upcoming year, I want to take this opportunity to share how the FA Research Fund is evolving to better carry out our mission.

In June, the FARF Board of Directors began the process of developing a new strategic plan. This plan will be completed early next year and will help set the organization's priorities and direction for the next three years. Also early next year, FARF will hire its first Philanthropy Director. This position will professionalize the fundraising strategy at FARF and allow us to reach untapped resources in the development arena. These two efforts are practical strategies that will bring us closer to achieving our vision. In marshalling these resources, we'll be able to better address the specific needs of the FA community.

Now is an optimal time for *you* to help FARF by reaching out to your friends, family, coworkers and neighbors with an appeal letter. You can make an impact by sharing your compelling story, discussing the advances made in the FA fight, and inviting your friends to help. Our staff is here to assist you at every step. We can edit, customize, print, pay for postage, and mail your letters. We can even provide templates for your letter. Take a look at some of our highlights over the past year, listed on the next page. Please contact our office at 541-687-4658 or info@fanconi.org and let's work together!

The success of FARF is enhanced by *your* efforts. Together, let's support families, conduct innovative and leading research, and most importantly, change lives.

Thank you so much for your support and all you do for FARF.

Mark Quinlan
Executive Director

a look back

FARF awarded funds to two new gene therapy projects that use cutting edge technologies to remove or replace a segment of DNA, allowing gene correction, in a process called "gene editing". One team recently pioneered a method to repair sickle cell genes and will attempt the same with FA. The other team will explore the use of nanoparticles to deliver the repaired genes to FA patients by simple injection into the blood stream.

Another research project will address the great need to prevent oral cancer in people with FA. Specifically, researchers will investigate whether it is possible deliver cancer preventing agents to the oral cavity without negative side effects.

In June, nearly 230 people with FA, family members, and FA experts gathered at Camp Sunshine in Casco, Maine for five days of educational sessions, support groups, and bonding opportunities. Fifty-two FA families from four countries attended. Among this group were 53 individuals with FA (including 14 adults)!

FARF funded the first stage of a clinical trial to prevent or delay bone marrow failure and leukemia in FA. This will be the first clinical trial that repurposes an established, safe drug – metformin, used for diabetes – to treat FA.

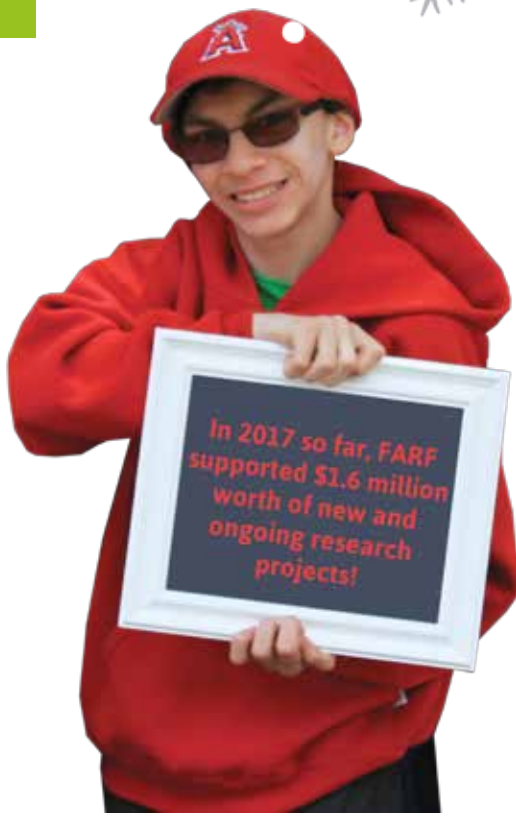
2017 HIGHLIGHTS

The Fund hosted the 29th Annual Scientific Symposium in Atlanta, Ga., bringing together more than 200 FA experts and stakeholders to share updates in research and treatments. This year's conference included new sessions on rare disease drug development and opportunities for industry partnership.

One of this year's grant recipients will investigate the possibility of reprogramming human (FA) skin cells into blood forming stem cells. This has the potential to provide a powerful tool for understanding the defects in human FA blood formation, and may provide a system for testing new drugs.

The 8th International Gene Therapy Meeting will take place this November in Heidelberg, Germany. This group of dedicated gene therapy experts meet every year to share data and results and update protocols. There are now FA gene therapy trials open on two continents.

Last year, FARF's database of people with FA showed for the first time that there were more adults than children living with FA. Subsequently, this past September, FARF held the largest meeting for adults with FA since the meeting began eight years ago. Forty-two adults gathered for four days of medical and research updates, support sessions, and quality time together. Because of the growing adult population, this meeting will now be annual instead of every 18 months. ■



4,000 Miles, $90,000, Two Bikes, One Mission

By Steve Rice, President, KATA Foundation

Over 10 years ago, my good friends Ken and Jeanne Atkinson lost two children to Fanconi anemia. Despite their tremendous grief, they were inspired to create the Kendall And Taylor Atkinson Foundation (KATA) to raise money to continue the fight against Fanconi anemia and enhance the lives of children. In April 2016, Ken was senselessly killed outside his own home as he saved the life of a neighbor who was a victim of domestic violence. While the loss of our great friend was devastating, we know he would want us to continue the fight in his honor.

This summer, my close friend Dave Kummer joined me on a life-changing journey: a Coast to Coast bike ride from Oregon to Maine. We covered 4,132 miles (6,650 kilometers) in 56 days, and traveled across 12 US states and one province in Canada. Our purpose was to connect with FA families across the country, increase awareness of FA, and raise funds for research.

We began our journey on May 20, riding from the Pacific shore into Eugene, Ore., home of FARF headquarters. Fifty-six days later, we dipped our tires into the Atlantic Ocean in Maine. We are thrilled to report that we raised more than $90,000! Each day of our journey was dedicated to a different individual with FA. Their stories gave us the motivation and strength needed to get through some especially tough rides.

Since the end of the ride, I've had some time to catch up on life and reflect on this adventure. My overall takeaway is that I am incredibly blessed. I've had the opportunity to bike through miles of extraordinarily beautiful landscape. I have ridden in the footsteps of Lewis and Clark. I was able to visit historic landmarks in both the United States and Canada, including Mount Rushmore and Fort Ticonderoga. I've chased notable waterways such as the Erie Canal, dipped my toes into many of the Great Lakes, and dipped my bike tires into two expansive oceans. For all of these experiences, I am grateful.

That said, my fondest memories of this journey will always be the FA fighters and families I met along the way. It began on day 1 when we met Alissa and Mary-Beth at the FARF Kick-Off Party and asked them to sign our jerseys. On day 8, as we endured a difficult and long 11-mile climb up White Bird Hill, we looked up to see young Eli Boson sticking his head out the sunroof of his family car, clapping and cheering for us. We spent Canada Day in Canada, at a picnic with the Bland family, and celebrated the 4th of July at Niagara Falls with the Collings, Blake, and Bentley families, allowing us to acquire a new appreciation for the hospitality of our neighbors to the North. We met another Eli – Eli Lana – who stole the show during our interview for the Rochester morning news show in New York. Many families opened their homes to us, fed us, or otherwise went out of their way to connect with us along the way. We had our jerseys signed by 22 Fanconi fighters (or family members), rode part of our cross-country journey with four of them – Bella, Lily, Eli Lana, and Tiernan – and met 26 FA families during our ride. Each one was a blessing that kept us inspired.

Thank you to each one of you who helped us along the way, whether it was by making a donation to the KATA ride, donating in honor or memory of a Fanconi fighter, encouraging us as we rode, or covering us with your prayers throughout the ride. I truly felt loved and supported every moment. I am still astounded at the love and generosity of this community. *Thank you.*

KATA Ride Checklist

- ~~4,132 miles~~
- ~~12 US States & Canada~~
- ~~Raise awareness for Fanconi anemia~~
- ~~Pacific Ocean~~
- ~~Atlantic Ocean~~
- ~~56 Days~~
- ~~$80,000+ for FA Research!!!~~

Do Something Epic

By Orion Marx

Dream. Challenge yourself. Go for it. Do something no one thinks you can do. Set an audacious goal. Fix our DNA. Cure Fanconi anemia. Cure cancer. This is what all FA families request from the doctors, scientists, researchers and bio/pharma companies. **We** can help, too!

As families with FA, we all feel the challenges facing our families every day. As parents, we wish we could solve FA, find the cure, and fix all that is afflicting our children. I wish I could swap places with my daughter, Avery; I would take on FA and let her deal with life's normal challenges without FA. **Although we cannot solve FA directly ourselves or swap places with those who have it, there are epic ways we can all help.**

We can apply our ability, passion, encouragement and energy into raising funds to help the FA Research Fund carry out its mission and find that cure. When we, as parents and family members, are able, it is imperative that we do all we can to support our children with FA. Hopefully we do so with the grace, strength and discipline that is required to manage the FA diagnosis and all it entails.

Our family started Team BrAvery in 2010, shortly after Avery's diagnosis. Prior to that, we were unknowingly training for future Team BrAvery events. My father-in-law Charlie Scott, my brother-in-law Zar Toolan, and I previously completed challenges like hiking the Rim to Rim to Rim in the Grand Canyon. We did them for fun and because we could. We also wanted to accomplish something special: see and experience nature in a way that few are able to experience.

Now, Team BrAvery completes challenges for an even more important reason. Someone may ask "Why are you biking the 500 mile Natchez Trace without support?" This is a great opportunity to tell them all about FA, our fundraising efforts, the importance of registering with The National Marrow Donor Program (www.marrow.org) and most importantly, this is an opportunity to encourage them to donate! "Why are you running a marathon on Saturday in Alabama and another one the next day in New Orleans?" Great question! The team's challenges are a fantastic opportunity for team bonding, to accomplish the seemingly impossible, as well as raise funds and awareness for FARF. In other words, do something epic.

Team BrAvery has very high expectations throughout all phases of challenge research, preparation, training and results. We know the challenges won't be easy, and they won't always be fun. There are times when we are not sure that we will make it, yet we always do and we always will. We use what God gave us: passion, patience, skill, stamina, humor, and the desire to succeed. None of these tough challenges equates to the difficulty that children and adults with FA face just to survive. Whether we are trail running, kayaking or biking, we regularly remind each other that whatever we're facing pales in comparison to what families with FA endure each day. Keeping FAmily in the forefront of our minds helps us stop our whining, keep on going, and accomplish our goal.

Doctors, researchers and scientists face challenges, too. Often, results may not be what they expected. Yet they continue to endure, they set audacious goals and they work to solve a medical mystery no one currently understands. Together with all FA families, Team BrAvery walks alongside this team of experts. We provide encouragement and support throughout their challenges, all the while knowing they will use their passion, preparation, research, and training to deliver results. The results are amazing. The results will be epic. **Do something epic.** ■

Learn more about Team BrAvery's next crazy adventure at **www.goteambravery.com** and follow them on Facebook **@goteambravery**.

FAMILY FUNDRAISING EFFORTS

From January 1 through August 31, 2017, 154 Fanconi anemia families raised an amazing $1,013,003 for the Fanconi Anemia Research Fund. Eighty-five cents of every dollar donated goes directly to research and family support to make a difference in the lives of individuals and families affected by FA. Thank you for your outstanding fundraising efforts so far this year!

Over $1 million raised!

$254,000
The Kendall & Taylor Atkinson Foundation with the Nash, Griggs, and Atkinson Families

$127,000
Lynn Frohnmayer

$68,000 - $78,000
John and Kim Connelly
Kevin and Lorraine McQueen

$48,000 - $54,000
Orion and Lisa Marx
Gerard and Cynthia Vandermeys

$21,000 - $30,000
Mary Ann and Tim Lana
Todd and Kristin Levine
Pedro and Marina Ravelo
Nigel and Ann Walker

$10,000 – $20,000
Jimmy and Jenny Armentrout
Adam and Marissa Becker
Kerrie Cazzari
Kevin Gatzlaff and Rachael Alaniz
Andre Hessels and Rutger Boerema
Charles and Katy Hull
Steve and Jennifer Klimkiewicz
Ian and Tricia Mitchell
Tyler Morrison and Rachel Altmann
Peg Padden
Emily and Neil Robison
Rick and Lynn Sablosky

$5,000 – $9,999
Donald and Danielle Burkin
David and Mary Ann Fiaschetti
John and Martina Hartmann
Peter and Janice Pless

$1,000 – $4,999
Israel and Mary Jo Becerra
Jeffrey and Donna Boggs
Richard and Tena Boson
Chris and Jennifer Branov
Ryan and Becky Brinkmann
Robert and Barbara Capone
David and Kim Chew
Colin and Ashley Chorneyko
Ana Concha
James and Crystal Eubank
Ezat and Laila Faizyar
Daryn and Carol Franzen
Gary and Melody Ganz
Laurie Gerhardt
Heidi and Gary Grassi
Alan and Rachel Grossman
Owen Hall and Margaret Kasting
Brian Horrigan and Amy Levine
Randy and Lisa Jones
Daniel Kold
Sheila Meehan
Jack and Lisa Nash
Ron and Fredi Norris
Mark and Diane Pearl
Rose and David Pennell
Paul and Rena Rice
Bob and Andrea Sacks
Brian and Jennifer Sadlowe
Bryan and Karen Siebenthal
Jan and Ken Sysak
Anthony and Elisa Walsh
Marc Weiner

Up to $999
Peter and Donna Abramov
Al and Janeth Acosta
Michael and Jennifer Aggabao
Juanita and Ron Arroyo
Andrew and Vicki Athens
Jasmine Bennetsen
John and Francene Berglund
Tracy Biby
Randy and Nancy Bloxom
Sean and Allison Breininger
Anita Casani
Tom and Mary Eilleen Cleary
Daniel and Melinda Coleman
Chris and Heidi Collings
Andrew Coons and Valeen Gonzales
Michael and Kim Curvey
Darrel and Kalani DeHaan
Donna DellaRatta
Jeremy and Michelle DellaValle
Wendy Delzell
Antonino and Marie Di Mercurio
Alex Eddy
Sharon Ellis
Chloe Eminger
Billy Jo and Debbie Estep
Curt and Crystal Fales
Justin and Britteny Ferrin
Nancy Finnegan and Scott Finnegan
Liz Funk
Mitch and Erin Furr
Emmanuel and Dana Gallegos
Manuel Gaytan
Ben and Stephanie Griggs
David Guidara
Patti and Mike Hilbert
Tammy and Ben Hilton
Jeff Hoffman
Frances Hutchins-Huff

PayPal Giving Fund

Do you want a simple way to increase giving to the Fanconi Anemia Research Fund? Welcome to the PayPal Giving Fund.

EBay sellers are encouraged to give a percentage of their proceeds to a nonprofit certified by PayPal Giving Fund each time they list an item for sale. EBay members can also choose to make an online gift with PayPal. The Donate Now tab lets anyone with a PayPal account donate. For more information, see www.paypalgivingfund.org.

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Joseph and Natalie Vitrano
Jian Yang and Jing Nie
Sean and Kristin Young
Cecelia Zurhellen

A note to our fundraisers: we greatly appreciate your efforts to raise money for FARF, and we want to recognize you all accordingly and with 100% accuracy. If we have inadvertently made an error, please let us know by emailing info@fanconi.org. Thank you.

DONATE WHILE YOU SHOP ON AMAZON

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

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Stick It To FA: 8th International FA Day Raises Nearly $180,000

May 1 is International Fanconi Anemia Day, when families and individuals affected by FA raise funds and awareness to advance research and support services. This past spring, more than 30 families from 15 states and 4 countries held fundraisers large and small, from art and yard sales to video game marathons, banquet dinners, concerts, runs, and more. Together, they raised nearly $180,000! The Fanconi Anemia Research Fund would not be able to work toward its mission without the efforts and dedication of these families. A major THANK YOU goes to all of the families and individuals who helped Stick It To FA this year!

To date, FA Day fundraisers have raised nearly $880,000 to support the Fund's mission. ■

FUNDRAISING SCRAPBOOK

Concert and Wine Benefit for FARF

On April 30, FARF board member and longtime supporter Sharon Schuman gave her 18th annual concert and FARF benefit at the McMorran House in Eugene, Ore. More than 100 guests enjoyed wine, hors d'oeuvres and a silent auction along with beautiful music. They also heard from FARF scientists Rich Gelinas, Brad Preston, and Grover Bagby about the strides FARF is making in research and treatment. This year, Sharon exceeded her goal and raised more than $20,000! Way to go, Sharon!

Beat FA!

In April 2016, then 8-year-old Dylan Hessels went through a bone marrow transplant in New York. One year later, Dylan and his family made a video to share his story and to raise funds. That same spring, Dylan's dad and FARF board member André ran the Boston Marathon to raise funds for FARF. Between the video and the marathon, the Hessels family raised more than $10,000! Thank you for getting us closer to a cure.

Fiesta for FA

On June 10, the Ravelo Family held the "Fiesta for FA" in Chicago to celebrate the 10-year anniversary of FA fighter Ivan's cord blood transplant. Friends, family and community members gathered to enjoy great food, music, dancing and a raffle. By the end of the night, more than $25,000 was raised for FA research and family support! Thank you, Ravelo FAmily, and happy rebirth anniversary, Ivan!

5K for FA

The Lana family held their third annual 5K for FA on May 7 in Hilton, NY. Despite a last minute rainstorm that flooded their original route, the community came together to help the Lanas pull off another incredible and successful event. In addition to the trail run/walk and kids fun run, participants enjoyed Zumba, raffles, music, and food. The run, which has raised more each year, brought in more than $25,000 for FARF! Visit www.5kforfa.com to learn about next year's race.

Eat for a Cure

Inspired and encouraged by other FA families, the Walsh Family wanted to make a difference and set out this past summer to raise funds for FARF. They reached out to restaurants in their local community of Columbus, Ohio to share their story and work together to raise funds. On three consecutive weekends in August, the Walsh family held fundraisers with Johnson's Real Ice Cream, TAT Ristorante Di Famiglia, and Jersey Mike's Subs. The community rallied alongside them and they were even joined by other local FA families. By the end of the month, the Walsh family raised $2,500 for FARF. Thank you!

Dream Getaway to Orlando

In July, the Gatzlaff/Alaniz family held an online raffle for an Orlando Disney Getaway through their organization, Aria's Army, named for their daughter, who died from complications of FA in 2015. Inspired by Aria and their family's fond and special memories together at Disney World, Aria's Army put the word out. The raffle brought in over $12,500, and one lucky winner out of an incredible 850 entries won a vacation for four to Disney World and Universal Orlando! Congrats, winners, and thank you, Aria's Army! Learn more at www.ariasarmy.com.

ARIA'S Army
www.ariasarmy.com

FARF GRANTS AWARDED FEBRUARY – OCTOBER

From February 2017 to October 2017, the Fanconi Anemia Research Fund awarded $663,508 to the following projects:

Investigators:
Grant Rowe, MD, PhD; George Daly, MD, PhD
Boston Children's Hospital

Title:
Engineering Fanconi Anemia Hematopoietic Stem Cells from Human iPS Cells

Amount: $175,000

Investigators:
Kenneth Weinberg, MD, PhD; Mochly-Rosen, Daria, PhD
Stanford University

Title:
The prevention of DNA damage in Fanconi anemia HSC by ALDH activators

Amount: $190,940

Investigator:
Alan D'Andrea, MD
Dana-Farber Cancer Institute

Title:
TGF-ß pathway inhibitors for the treatment of bone marrow failure in Fanconi anemia

Amount: $175,000

Investigators:
Susan R. Mallery, DDS, PhD; Joerg Lahann, PhD
Ohio State University

Title:
Field-Coverage Oral Cancer Chemoprevention via Janus Nanoparticles

Amount: $122,568

FARF is committed to supporting research to further our mission of finding new treatments and a cure for Fanconi anemia. Over our 28-year history, we have funded 223 research grants and one service grant to 112 investigators worldwide. The total amount of research dollars awarded is over $20 million!

STELLA DAVIES JOINS FARF BOARD

The FA Research Fund is excited to welcome Dr. Stella Davies to the Board of Directors.

As director of the Division of Bone Marrow Transplantation and Immune Deficiency at the Cincinnati Children's Hospital, she has helped pioneer significant advances in bone marrow transplantation to improve survival rates and reduce toxicity. Dr. Davies has led efforts to sustain and improve an already-strong clinical center of excellence for the care of Fanconi patients and their families. She has also played a key role at Cincinnati in fostering and supporting strong clinical and basic science focused on Fanconi anemia. She is an active and vocal participant in the FA scientific community and the Fund is fortunate to have her amazing intellect, boundless energy, and infectious enthusiasm in the fight against Fanconi anemia.

JOEL WALKER SCIENTIFIC MEETING SERIES ESTABLISHED

Joel Walker, an adult with FA, sadly passed away from complications of head and neck cancer in November 2016, at the age of 33. In honor of their son, Joel's parents, Nigel and Ann Walker, created the Joel Walker Scientific Meeting Series. A generous bequest from Joel's estate and on-going support from the Walker family make this important series of scientific meetings possible.

"We are extremely grateful for the gift left by Joel and thrilled to honor his memory with this effort," said Mark Quinlan, FARF Executive Director. This series will support focused scientific meetings on a variety of topics, beginning with head and neck cancer prevention and treatment in FA. The first meeting is planned to take place in 2018.

UPDATE: NAC TRIAL

In 2014, FARF approved funding for a prospective phase I/II trial to evaluate the drug N-acetylcysteine (NAC) in patients with Fanconi anemia.

Due to the multicenter and international nature of the trial, the primary institute was unable to secure necessary permissions and approval to move forward with the trial. For this reason, FARF discontinued funding to the project in spring 2017. Should the project status change, FARF will provide an update.

FARF BEGINS WEBSITE REMODEL PROJECT

The FA Research Fund is thrilled to announce the website remodel project, officially in action as of fall 2017.

The FARF website, www.fanconi.org is getting a complete makeover! It will be more user-friendly, informative, compelling and vibrant. The new site will feature many more news stories, expanded family services (such as an online directory and separate section for adults with FA), a robust research center including a directory of all FARF-funded grants and researchers, and create-your-own peer-to-peer fundraising pages, to name a few improvements. The expected launch date of the new site is early spring 2018. Stay tuned!

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STAFF

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Sandra Hilliard, Finance Coordinator
Suzanne Planck, Family Services Coordinator
Bradley Preston, PhD, Scientific Director
Marie Sweeten, Family Services Director
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I Want to Raise Funds – Where Do I Start?

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

UPCOMING FUNDRAISERS

Fundraising events are a great way to get involved and make a difference in the FA community. And, they're fun! 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.

Jewelry for Jacob: Holiday Boutique Jewelry Sales
Ongoing
Worldwide (online)
The Grossman Family

KATA Hoot n' Holler
Nov. 18, 2017
Denver, Colorado
The KATA Foundation

Family Holiday Appeals
Holiday Season
Worldwide
All Families

LA Marathon
March 18, 2018
Los Angles, CA
The Aggabao Family

Band, Brew & BBQ
April 20, 2018
Richmond, Virginia
The McQueen & Vandermeys Families

9th International FA Day
May 2018
Worldwide
All FAmilies

5K for FA
May 5, 2018
Hilton, New York
The Lana Family

Ontario Fundraiser for Fanconi Canada
May 6, 2018
Toronto, Canada
Fanconi Canada

FARF Benefit Concert
Spring 2018
Eugene, Oregon
Sharon Schuman

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.*

FANCONI ANEMIA
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Advancing Research. Supporting Families.

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

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

Donations by Mail:
1801 Willamette St., Suite 200, Eugene, OR 97401

Donations of appreciated stock:
Please contact our office at 541-687-4658 or email info@fanconi.org.

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