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FA Family Newsletter

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Science News from the 2010 Symposium

FA Researchers, Clinicians and Families Gather at Annual Scientific Symposium

Approximately 200 Fanconi anemia researchers, clinicians, board and family members attended the 22nd Annual Scientific Symposium in Minneapolis, Minn., Oct. 21 to Oct. 24, 2010.

The Fanconi Anemia Research Fund hosts this annual meeting to bring together many of the world's top FA researchers and clinicians to assess the current state of FA research and treatment and to plan next steps. Forty oral abstracts were presented on topics including new FA genes, gene repair, stem cells, germ cells and signaling, molecular diagnostics, nuclear crosslink metabolism, transplantation, and cancer.

Four special sessions, featuring talks about radiation mitigation, gene therapy, homologous recombination, dyskeratosis congenita, induced pluripotent stem cells, and RNA interference were spaced throughout the four-day conference. A panel of transplant physicians from FA transplant centers in the U.S., Brazil and Europe rounded out the agenda.

Of particular interest were discussions about the role of HPV in FA patients and questions about vaccinations. Family Meeting attendees heard from several physician presenters at Camp Sunshine that they should consider vaccinating boys and girls as early

A highlight of the Symposium was the dinner program at which Dave and Lynn Frohnmayer, co-founders of, and the driving force behind, the Fanconi Anemia Research Fund, received the Sui Generis *Award.* Sui Generis *is Latin for "in a class of its own," and from the prolonged standing ovation, heartfelt words of congratulations and renewed commitments to FA research, it was evident that this one-time-only award was well deserved. Dave and Lynn's dedication has led to advances in FA research that have improved and prolonged the lives of FA patients.*

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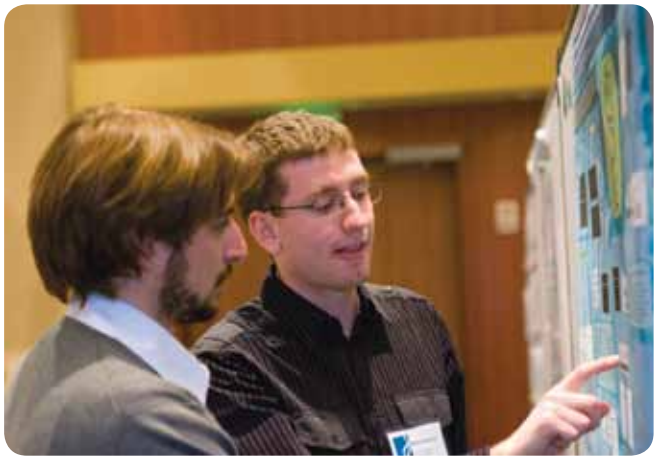


Remi Buisson, right, Laval University, Québec, Canada, points out his research findings to Luca Colnaghi, New York University School of Medicine.

as 6 years of age. Discussions at the symposium suggest more data are needed.

Also of note were presentations about the potential value of induced pluripotent stem cells for FA research and treatment. Although FA cells are generally resistant to reprogramming and much work remains, the rapidly evolving technology for the production of clinically useful, patient-specific cells is promising.

> "Advances happen when scientists challenge each other."
>
> *—An assessment of the symposium shared by a participant*

The symposium was preceded by an exciting first meeting of researchers from Harvard, the University of Oregon and Oregon Health & Science University. This consortium received a $10.7 million grant from the U.S. National Institutes of Health—after receiving initial funding from the Fund— to test thousands of new and existing drugs that could potentially prevent complications associated with FA.

In addition to the awards given to Lynn and Dave Frohnmayer and to Ralf Dietrich (see related photos and text), Detlev Schindler, MD, Christopher Mathew, PhD, Helmut Hanenberg, MD, and their respective lab partners received the Fund's Discovery Award for their collective work to identify the 14th Fanconi anemia gene, RAD51C.

The Fund will hold its 23rd Annual Scientific Symposium in Barcelona, Spain, in October 2011 because of the country's very active FA research and family support communities. The last year the Fund held its annual meeting outside the U.S. was in 2005 when the meeting was in Geneva, Switzerland. ■

Ralf Dietrich, Executive Director of the FA German Family Support Group, received the Fund's Distinguished Service Award, presented by Dave Frohnmayer, for his tireless devotion to and advocacy for FA patients. Ralf's fundraising efforts have generated close to $1 million for FA research in Europe. In addition, Ralf has visited more than 1,000 FA patients, collected blood count histories from dozens of FA patients, videotaped interviews with FA families, treating physicians and researchers from more than 30 countries, and attended more than 100 FA family meetings and scientific workshops. Ralf is only the seventh individual in the Fund's 22-year history to be recognized in this category.

Optimizing Alternative Donor Transplants At the University of Minnesota

Margaret MacMillan, MD, University of Minnesota, reported on the outcomes of 94 Fanconi anemia patients who underwent alternative donor transplants at her institution from 1999-2010, using one of three consecutive trials. With each trial, one change was made from the previous trial in order to evaluate the optimal approach for FA patients.

In Trial 1 (1999-2003), fludarabine was added to the preparatory regimen (Cytoxan, ATG, total body irradiation [TBI] 450 rads) to improve engraftment; in Trial 2 (2003-2006), shielding of the thymus gland during TBI was added to hasten immune recovery; and in Trial 3 (2006-2010), TBI dose de-escalation was initiated to determine the lowest dose required for engraftment. Patient outcomes have improved with each change in the transplant regimen.

Based on analysis of patient outcomes, several findings emerge:

- Fludarabine promotes excellent sustained engraftment.
- TBI 300 is sufficient for engraftment, but TBI 150 is not.
- GvHD rates are low with T-cell depletion (probability of grade II-IV acute GvHD 23%; grade III-IV 10%; probability of chronic GvHD 13%).
- Patients should be transplanted before receiving **any** transfusions.
- Patients transplanted before age 10 have better survival.

> Patient outcomes have improved with each change in the transplant regimen.

Thirty-eight patients were transplanted on Trial 3 (Cytoxan, fludarabine, ATG, TBI 300 rads plus thymic shielding). Probability of survival at six months post-transplant (median follow-up of 24 months) is 88%.

The University of Minnesota also transplanted seven high-risk patients, six with leukemia. This trial used busulfan instead of TBI. Four patients survive. Based on patient toxicity, busulfan will not be used in the future for FA patients at this center. ■

Work Continues on Recommendations Regarding Clonal Aberrations

Leading cytogeneticists gathered prior to the Symposium to consider specific recommendations for screening Fanconi anemia patients for clonal aberrations, and to try to reach consensus on the relevance of specific clonal abnormalities (or their progression) for clinical care.

Interest for this meeting arose from data presented at previous scientific and family meetings by Heidemarie Neitzel, MD, Charité Medical University, Berlin. Dr. Neitzel concluded that certain clonal aberrations, especially gains of 3q and/or deletion of chromosome 7, are the primary cause for progression to MDS/AML. She also maintained that patients may wait too long before going to transplant once a clone appears, or that the clones are missed altogether because patients are not screened correctly or often enough. Betsy Hirsch, PhD, from the University of Minnesota, shared similar data and conclusions in a 2010 scientific journal article.

At the meeting's conclusion, participants agreed to continue to work together on this complex issue to develop clear guidelines for FA parents and physicians. In the meantime, families and adult patients are encouraged to refer to *Fanconi Anemia: Guidelines for Diagnosis and Management* for information on clonal aberrations. ■

Alternative Donor Transplant Outcomes At Memorial Sloan-Kettering

Farid Boulad, MD, Memorial Sloan-Kettering Cancer Center, New York, described alternative donor transplant outcomes at his center. His long-term goals are to eliminate GvHD, eliminate radiation and improve transplant outcomes.

Dr. Boulad reported on 26 alternative donor transplants at his center using total body irradiation (TBI) 450 rads, cytoxan, fludarabine and T-cell depletion. He described these 26 patients as a "tough crowd." Sixteen had been transfused; 17 were on androgens; 15 had prior infections and 15 had advanced disease (seven had leukemia at the time of transplant). Four patients were ages 20-25, and two were older than 30. Total survival at five years post-transplant is 65%, with 57% experiencing disease-free survival. For those younger than age 18, five-year survival is 72% and disease-free survival is 67%. Only 9.6% experienced GvHD. ■

Recent Alternative Donor Transplant Outcomes Are Very Promising at Charité Hospital, Berlin

Wolfram Ebell, MD, Charité Hospital, Berlin, reported on alternative donor transplants at his center using two separate protocols, GEFA 02 and GEFA 03. Although it is too early and the numbers of patients transplanted are too small to draw definite conclusions, results from the current protocol (GEFA 03) are most promising.

The GEFA 02 protocol included fludarabine, busulfan, ATG and OKT3. Eighteen patients ages 2-25, average age 11, participated. Patient characteristics suggest that many were high risk: 56% had MDS/AML, 61% had cytogenetic abnormalities; 85% were on androgens; and 61% were multiply transfused. Eleven of 18 are alive, at an average time post-transplant of 98 months.

The GEFA 03 protocol included fludarabine, busulfan, cyclophosphamide and MabCampath. More than half of these patients had T-cell depletion, compared to 6% in the 02 protocol. Nine patients, ages 7-19, average age 13, were on the GEFA 03 protocol. Patient characteristics are very similar to those in GEFA 02: more than half had MDS/AML; 44% had cytogenetic abnormalities; 78% were on androgens; and 67% were multiply transfused.

All nine patients survive. They are from one to 64 months post transplant (average time 31 months). Both acute and chronic GvHD were reduced (no patient had more than grade 1 GvHD).

> Alternate donor transplants are improving and can be successful without radiation.

In patients who had developed a 3q abnormal clone without progressing yet to myelodysplasia or leukemia, this clone was not a risk factor in either protocol. This suggests a "window of opportunity" to proceed to transplant.

Dr. Ebell said that alternative donor transplants are improving and can be successful without radiation. He concluded that results from his center are "very promising." ■

Recovery of Gonadal Function After Stem Cell Transplant

Teens and young adults with Fanconi anemia have reduced fertility, and only a few cases of successful pregnancy after stem cell transplant have been reported. To prevent irreversible gonadal dysfunction, Hiromasa Yabe, MD, Tokai University School of Medicine, Isehara, Japan, has used ovary-shielded thoracoabdominal irradiation (TAI) since 1991. Initially he used TAI 450 rads; in 2002 radiation was reduced to TAI 300 rads.

Dr. Yabe reported on the outcome of a study to determine if gonadal function can be recovered post-transplant by shielding the ovaries during irradiation. Seventeen female patients transplanted between 1996 and 2009 at Tokai University Hospital were enrolled in the study. Median age at time of transplant was 12 (range 5-28). The preparatory regimen varied over this period of time, and donors ranged from matched siblings to alternative donors.

Most female FA patients receiving ovary-shielding irradiation recovered gonadal function post-transplant.

All patients are surviving five to 164 months post-transplantation. Nine of the 11 patients transplanted before puberty achieved normal puberty at a median age of 12 years; two patients did not complete normal puberty. Five of the six post-pubertal patients had return of menstrual cycles at four to 32 months post-transplant (one of these patients received hormone-replacement therapy). The menstrual cycle did not return in one patient with AML who received intensive chemotherapy before transplant.

In conclusion, most female FA patients receiving ovary-shielded irradiation recovered gonadal function post-transplant. ■

Enhancements Made in International Fanconi Anemia Registry

Arleen Auerbach, PhD, The Rockefeller University, New York, reported on progress in developing an enhanced International Fanconi Anemia Registry (IFAR). IFAR was established in 1982 to collect FA patient and familial data. In 1998 Dr. Auerbach established the Fanconi Anemia Mutation Database to accelerate availability of information on specific FA mutations. IFAR contains data on more than 1,500 patients including demographic, cytogenetic, medical history, mutation, treatment and transplantation data.

Information in the new IFAR will be extremely comprehensive, based, in part, on in-depth information obtained through new patient history questionnaires. The Cancer Genetics Branch, National Human Genome Research Institute, is currently sequencing patient tissue samples, and these data will be included in the new IFAR.

The new database will allow researchers to obtain information on subsets of patients, better to inform treatment decisions. For example, data on how much radiation can be tolerated post-surgery for head and neck cancer patients or how to define transplantation protocols for subsets of FA patients will eventually be available. Researchers can contribute their own information to this database, resulting in better data sharing. Eventually, researchers worldwide will be able to access anonymous IFAR data. ■

Beneficial Effects of Quercetin, a Natural Antioxidant

Studies show that Fanconi anemia patients are prone to diabetes, but the mechanism of this complication is unknown, according to Jie Li, PhD, Cincinnati Children's Hospital Medical Center. Studies also show that cells from FA patients are hypersensitive to oxidative stress, which contributes to bone marrow failure and cancer. In a study performed at her center, Dr. Li investigated the role of FA proteins in insulin signaling when FA mice are challenged with oxidative stress.

Dr. Li reported that mice deficient for two FA genes (Fanca or Fancc), when under inflammation-induced oxidative stress, show impaired insulin signaling and symptoms characteristic of diabetes. The natural antioxidant quercetin restored insulin signaling *in vitro* and *in vivo* and eradicated the progression of diabetes in FA mice. Dr. Li concluded that natural antioxidants such as quercetin might be beneficial therapeutically for FA patients. ■

> ...natural antioxidants such as quercetin might be beneficial... for FA patients.

Improving Engraftment of Gene-Corrected Stem Cells in FA Mice

Three published studies document the failure of gene therapy to correct the bone marrow of Fanconi anemia patients. These disappointing results reflect, in part, both the difficulty in harvesting a sufficient number of patient stem cells and the low repopulation capacity of corrected cells.

In preparation for opening a gene therapy clinical trial of their own within the year, researchers at the Fred Hutchinson Cancer Research Center in Seattle are conducting studies in mice to see if new methods can improve engraftment of corrected cells. Jennifer Adair, PhD, a clinical research associate in the laboratory of Dr. Hans-Peter Kiem, MD, reported on their efforts at the October Scientific Symposium.

Dr. Adair and colleagues use a lentiviral vector, which transduces stem cells effectively and more safely than other retroviral vectors. Still, these mouse studies showed a low level of engraftment. The investigators hypothesized that selective killing of FA uncorrected cells might enhance engraftment. To determine the most effective drug to improve engraftment of corrected cells, they conducted studies using either cyclophosphamide, cytarabine alone, or cytarabine combined with fludarabine, and compared engraftment to results in mice not treated with any preparative regimen.

> ...a preparatory regimen using cyclophosphamide can facilitate engraftment and may be necessary for successful gene therapy...

The most effective engraftment was achieved with cyclophosphamide. No cytogenetic abnormalities were observed in cyclophosphamide-treated mice.

These studies suggest that a preparatory regimen using cyclophosphamide can facilitate engraftment and may be necessary for successful gene therapy in FA patients. ■

Brush Sampling to Detect Pre-Cancerous And Cancerous Lesions: Filling a Gap

Dr. Eunike Velleuer, University Children's Hospital, Düsseldorf, Germany, reported her observation that in spite of the high risk of head and neck cancer in FA patients, patients do not undergo the regular oral cavity screening that is strongly recommended in the 2008 consensus handbook, *FA: Guidelines for Diagnosis and Management.* She and her collaborator, Ralf Dietrich, Executive Director of the Fanconi anemia support group in Germany, embarked on an alternative education and screening strategy focused on patient acceptance and clinical applicability.

Dr. Velleuer has now examined and taken brush samples of six high-risk locations and all visible lesions in the oral cavities of 370 FA patients. She reported on 264 patients over the age of 10, because FA guidelines recommend oral screening twice a year beginning at that age.

Dr. Velleuer detected and photographed 277 visible oral lesions in 84 patients. Brush samples from lesions were examined for genetic changes. Twelve of 277 visible lesions in 10 patients were found to be malignant by cytological evaluation. Dr. Velleuer reported that in this population, no cancer had occurred without the prior appearance of clinically visible lesions.

In spite of FA guidelines, fully 85% of patients interviewed by Ralf and Dr. Velleuer admitted that they were not screened regularly. Patient reluctance and procrastination might account for this non-compliance.

> FA patients are *strongly* recommended to undergo regular oral cavity screening.

Brush screening is well accepted by all patients and might eliminate the need to take an invasive biopsy of every visible lesion. Dr. Velleuer emphasizes that of 277 visible lesions, only 12 were cancer. She states that biopsying every visible lesion "would mean a lot of invasive diagnosis for 'only' 12 cancers and would not increase compliance for regular screening!" ■

Researcher Targets FA Cancer Stem Cells

Cancer stem cells (CSCs) characterize a variety of tumors, including the head and neck cancers that affect Fanconi anemia patients. According to Ian Mackenzie, DDS, PhD, Blizard Institute, Queen Mary University of London, these cells are self-renewing, give rise to other cancer cells, and are particularly resistant to conventional therapies that target malignancies. Because they resist cell death, CSCs are thought to be responsible for recurrence of cancer following remission.

FA cell lines contain cells with CSC properties. These stem cells show strong expression of CD44 and other stem cell markers. They look different than other cancer cells, appearing as colonies of small, tightly-packed cells and forming spheres in suspension cultures.

Radiation and chemotherapy are particularly toxic to FA patients, and often do not succeed in eliminating cancer stem cells. Dr. Mackenzie says that agents targeting the maintenance of CSCs, rather than DNA-damaging therapies, hold particular promise for FA patients. Consequently, the effects of drugs such as Erbitux and Tarceva, that antagonize stem cell signaling pathways, are of particular interest. ■

RAD51C: The 14th FA Gene (FANCO) Or an FA-Like Gene?

The lead researchers to discover the 14th gene related to FA are (left to right) Helmut Hanenberg, MD, Chris Mathew, PhD, and Detlev Schindler, MD.

A group of European researchers recently discovered that siblings with characteristics typical of Fanconi anemia had mutations in the gene *RAD51C*. Since this discovery involved only one family, scientists are in disagreement over whether *RAD51C* is the 14th FA gene *(FANCO)* or should be designated an "FA-like" gene, pending assignment of other patients to this complementation group.

Helmut Hanenberg, MD, Christopher Mathew, PhD, and Detlev Schindler, MD, collaborated on the discovery (published in *Nature Genetics*) that three children born to first cousins of Pakistani origin (and a fourth pregnancy resulting in a miscarriage) had a biallelic disease-causing mutation in the known DNA repair gene *RAD51C*.

The affected children with defects in the *RAD51C* gene had a wide variety of severe congenital abnormalities that are typically associated with FA. Their cells also demonstrated an increased hypersensitivity to mitomycin C, consistent with a diagnosis of FA.

In a second publication in *Nature Genetics*, these researchers also established that the presence of a single germ-line mutation in *RAD51C* is associated with a high risk for breast and ovarian cancer, similar to the *BRCA1* and *BRCA2* genes. ■

Choosing a Donor in the Event of Primary or Secondary Graft Failure

If a Fanconi anemia patient fails to engraft following a stem cell transplant, or if the patient engrafts but subsequently loses the graft, should the same or a different donor be used for a subsequent transplant? Transplant experts Carlo Dufour, MD, Genoa, Italy, and Carmem Bonfim, MD, Curitiba, Brazil, combined data from the European Blood and Marrow Transplantation database and transplant data from Curitiba, Brazil, to answer this question. Their survey, which examined data on 106 FA patients transplanted between 1980 and December 2009, revealed the following:

- Overall, patient survival following a second transplant was approximately 30% seven years post-transplant (but transplant outcomes have greatly improved in recent years).
- If a patient fails to engraft following the first transplant (primary graft failure), using a different donor for the second transplant suggests better survival.
- If the patient engrafts but subsequently loses the graft (secondary graft failure), changing the donor for the second transplant does not affect survival.
- An interval greater than 80 days between the first and second transplant significantly improves survival.

This study was the largest cooperative effort ever to examine this important question. ■

Sweet Syndrome and Fanconi Anemia: A Manifestation of Hematologic Disease Progression

Sweet syndrome is a skin disorder characterized by painful red plaques. In Fanconi anemia patients, these often appear as raised red patches on the head, arms or legs, but can also appear in other organs such as the lungs. Researchers at four centers* discussed the incidence and implications of Sweet syndrome in FA patients in a poster presented at the Scientific Symposium.

The poster described seven patients with FA and Sweet syndrome diagnosed between 2000 and 2010 at three institutions. The incidence of Sweet syndrome in FA at one institution was estimated at 12%. All seven patients were diagnosed between ages 18-36. Two patients had poor and delayed wound healing after skin biopsy; one required extensive skin grafting. Six of seven cases had progression of hematologic disease at the time of diagnosis of Sweet syndrome (MDS in two cases and AML in four patients).

The researchers conclude that clinicians should take caution with obtaining skin biopsies due to significant complications from poor wound healing in some patients. All patients with FA and suspicion of Sweet syndrome need hematologic re-evaluation and, preferably, should proceed to treatment without skin biopsy.

This report was accepted for publication in the *British Journal of Haematology*. ■

Editors' note: *We urge special attention to this report. Our middle daughter, Katie, developed this baffling syndrome at age 8, her first symptom of FA. Prior to this, her appearance and blood counts had been normal. The syndrome can be misdiagnosed as an infection, but it is not. It can be a signal of a serious and ominous underlying bone marrow evolution.*

—Dave & Lynn Frohnmayer

* Memorial Sloan-Kettering, New York; Dana-Farber Cancer Institute, Boston; Hackensack University Medical Center, Hackensack, NJ; and Robert Wood Johnson Medical School, New Brunswick, NJ

Discovery of 15th FA Gene Reported by Two Major Laboratories

Chantal Stoepker, photo left. Yonghwan Kim (center) with collaborators Agata Smogorzewska and Francis Lach.

Chantal Stoepker, MSc, Vrije Universiteit Medical Center, Amsterdam, and Yonghwan Kim, PhD, The Rockefeller University, New York, reported that researchers at their respective centers have independently discovered that a gene previously known as *SLX4* is mutated in Fanconi anemia patients from four families.

The new FA gene, designated *FANCP*, functions downstream of the FA core complex. Six patients from four families have now been assigned to this complementation group.

Patient characteristics seem to resemble core complex patients, more than patients in the downstream complementation groups. *SLX4* is a multifunctional protein and regulates different types of DNA repair.

> The 15th FA gene is designated *FANCP*.

Different patient mutations may give different phenotypes, depending on what function of *SLX4* is affected. All six patients in FA-P have bone marrow failure.

Recent gene discoveries have involved only a small number of FA patients. Approximately 50 FA patients have yet to be assigned to a complementation group. This suggests that a certain number of FA genes have not yet been identified. ■

Multi-Center Transplant Protocol is Launched

Farid Boulad, MD, Memorial Sloan-Kettering Cancer Center, New York, reported on efforts to establish a multi-center Fanconi anemia transplant protocol, substituting busulfan for TBI in the conditioning regimen. Four centers are participating in this effort: Boston Children's Hospital, Cincinnati Children's Hospital, Children's Hospital of Wisconsin, and Memorial Sloan-Kettering, New York. Centers plan to enroll 25 FA patients. To date, eight patients have been transplanted using the busulfan protocol. All but one patient tolerated this regimen well; one patient experienced considerable toxicity. Results so far are "acceptable" and not different from TBI-based conditioning regimens. ■

New Lab Tests Can Potentially Identify Beneficial Cancer Therapy

The Knight Diagnostic Laboratories at Oregon Health & Science University have recently made available new tests designed to identify potential treatment targets in cancer and to predict the likelihood of benefit for patients treated with the latest therapeutics.

The testing is based on an approach called "mass spectroscopy," which supports the readout of hundreds of DNA assays in a very short period of time. These assays, combined into two new panels, allow screening for DNA mutations that are known to play a role in cancer growth and progression. One of the panels is designed for solid tumors; the other panel is for leukemia and related blood disorders.

The Knight Cancer Institute has partnered with the Fanconi Anemia Research Fund to make this new testing available at cost for FA patients who develop a malignancy. The Fund will pay the $300 fee for any FA patient. Testing can be requested by the patient's oncologist using a simple order form. In approximately six months, the form will be available on a new website; in the meantime, either the patient or the oncologist can contact Dr. Christopher Corless (see box) or Teresa Kennedy at the Fund for the form. ***The form, together with the tumor material to be tested and a copy of the original pathology report, can be shipped to the address below.*** A copy of the report will be faxed to the oncologist in 10 to 15 workdays. The testing can be done on almost any tumor material, including paraffin-embedded tissue, blood or bone marrow.

To request a form and/or to send samples:

Dr. Christopher Corless
OHSU Dept. of Pathology (mailcode L113)
3181 SW Sam Jackson Park Road
Portland, Oregon 97239
Phone: (503) 494-6834
Email: corlessc@ohsu.edu

Paraffin-embedded tissue, routinely generated on all surgical and biopsy specimens, is very stable and can be tested months to years after the procedure. Chris Corless or Teresa Kennedy may be contacted at any time before or after the surgery to request a form.

While the new mutation panels represent a novel approach in cancer diagnostics, there is no guarantee that any submitted tumor will yield a mutation that can be targeted by drugs that are currently available or are being tested in clinical trials. Regardless, screening FA malignancies by using the panels will help researchers develop a more complete picture of which mutations are, or are not, contributing to the growth and progression of these cancers.

Finally, please note that this new tissue testing option does not preclude our work with the National Disease Research Interchange (NDRI). The Fund strongly encourages FA patients to donate tissue to NDRI so that FA researchers around the globe can study the tissue samples and potentially learn new information that could lead to better treatments and a cure. ■

Donating Tissue to NDRI: A Researcher's View

By Susanne Wells, PhD
Experimental Hematology and Cancer Biology, Cincinnati Children's Hospital Medical Center

Fanconi anemia is a complex multisystem disorder. Disease development varies from person to person, influenced in part by an individual's particular FA gene mutation.

To study the correlation between the gene mutation and development of symptoms, scientists use laboratory models. A broad spectrum of models aids our quest to understand FA and design new approaches to treatment. Unfortunately, models bear artificial elements and do not reflect all that goes on in the human body or demonstrate all FA-related features. Therefore, **scientific hypotheses and conclusions must ultimately be tested in affected individuals, and your donated blood and tissue specimens are the most important key to the success of this effort.**

My lab aims to understand why and how FA mutations stimulate the development of anogenital and head and neck cancers. These tumors originate in skin cells called keratinocytes; some of these cancers are caused by human papillomavirus (HPV). Whenever possible, we use skin samples from patients who graciously provide them for research. These samples can be genetically manipulated to correct their FA, enticed to form tumors or immortalized to become a renewable cell system that can be used for more research.

HPV can also be introduced in such systems. Studies of these cells have shown that, in the lab, FA mutations can stimulate oncogenic effects of HPV. The question now is if HPV infection causes tumor development in FA. To answer this, we must determine whether and how often HPV is present in tumor samples from FA patients. This will clarify our understanding about development of these tumors and influence clinical approaches to prevent and/or cure these tumors in FA patients.

The Fanconi Anemia Research Fund has partnered with the National Disease Research Interchange (NDRI) to collect, preserve and distribute donated tissues to scientists. This effort helps the scientific community address some of the important research questions related to FA. With your cooperation, we strive to make advancements in the understanding of FA and, consequently, in the ability to help those who are affected. ■

Donating Tissue to NDRI: A Patient's View

By Kari Doctor

In May 2007 I found a large lump on my breast. After getting scans that suggested cancer, I was told to get a biopsy. I knew that if there was a chance of donating some of the tissue, I needed to complete some forms for the National Disease Research Interchange (NDRI). I called the Family Support Director at the Fanconi Anemia Research Fund, and she made it all very easy. If there was a chance that I could help another FAmily, it would be worth it.

After the confirmation that I did have breast cancer, I had surgery at the Oregon Health & Science University (OHSU). The tumor was large enough to donate to cancer researchers at OHSU and to NDRI.

Donating to FA research is so easy, and knowing I can help scientists find a cure is very rewarding. NDRI collects not only solid tumors, but blood samples and skin biopsies as well.

When I learned that my cancer had spread to my liver in November 2010, I didn't hesitate to donate this tumor, too. Through NDRI, this sample was sent all over the world, making a cure for FA even more possible through the many excellent scientists we have working on this horrible disease. ■

Adults with FA Meeting Earns Positive Response

Eighteen adults with Fanconi anemia, together with their guests, gathered in Portland, Ore., in mid-February to learn from clinicians and researchers in the field, interact in hosted group sessions, and socialize during a dinner cruise on the Willamette River. The 2½ day Adults with FA Meeting was held in Portland to coincide with the Seventh Annual Valentine Fanconi Anemia 5K/8K/12K Run/Walk on Feb. 13, a fundraiser organized by Peg Padden, an FA parent and Fund board member.

> *" It's amazing that this support is out there. I'm very lucky to have the FA family—it's great."*
>
> *—A meeting participant*

The meeting participants, ranging in age from 18 to 51, traveled from Australia, France, the US and Canada for this opportunity to meet other adults with FA and to learn more about living and coping with the disease. In addition to clinical talks on cancer, endocrine issues, adult stem cell transplantation and gynecologic issues, a highlight of the meeting was a presentation by Rebecca Block, MSW, LCSW, PhD, Oregon Health & Science University, Portland, Ore., on living with a life-challenging illness. See the related article in this issue.

Bob Nicholson (FA, age 51) attended the Adults with FA Meeting for the first time. Bob hails from Canberra, Australia. He found the meeting "very worthwhile in every way," noting it allowed him the unique opportunity to meet and talk with other adults living with FA.

Some meeting participants connect with fellow adult with FA, John Hanna, via computer.

Amy Frohnmayer's presentation, *Challenges, Coping and Quality of Life Issues in Adults with FA*, was also very well received. Amy summarized study findings from her psychology master's thesis while at Stanford University last year. Amy's survey data findings taken from 96 adults showed prominent use of acceptance and active coping strategies to deal with obstacles related to FA. Analysis of her qualitative data from 18 interviews of adults with FA supported frequent use of positive reframing, psychosocial benefits of interacting with other affected adults, and the importance of hope as an internal resource. Amy will report these and other findings at the upcoming Family Meeting in June.

> *" I cannot thank you enough for providing my daughter this trip to Oregon. For the first time in her life, she told me, 'I actually fit in somewhere.'"*
>
> *—Mother of meeting participant*

Participant feedback about the meeting was very positive, and a suggestion to get together again "sooner rather than later" was roundly endorsed.

Watch the Fund's website for copies of the presentations. They will be made available as received. ■

Living with a Life-Challenging Illness

By Rebecca G. Block, PhD

Let's be honest, living with a life-challenging illness is tough. Following are 11 tools that may come in handy:

1. **Look carefully and closely.** After living with an illness for a while, it's difficult to see what is really going on with your health and emotions. Take time to stop and assess your situation carefully and thoroughly.
2. **Try a new twist.** Look at your illness, treatment, life, day in a different way. This will help you appreciate the smooth times and prepare for the bumps.
3. **Bring pieces together.** Coping with an illness like Fanconi anemia is not unlike having a fulltime management position at work. You may need to identify helpers such as a communicator and a scheduler. Use technology to connect your "staff." Keep a detailed notebook with all relevant information clearly recorded.
4. **Size it up.** Know what you're up against. Break down the issues and decide how much each one needs and deserves. Sometimes fear and uncertainty can become exaggerated in your mind. Take time to find out the risks and then measure the uncertainty.
5. **Be heard.** Having your voice heard accomplishes two goals: (1) your opinion, choice, perspective or desire is known; and (2) it makes you feel valued.
6. **Safe storage.** People need a place where they feel safe and where they can figuratively or literally leave things to address later. Keep a box for memories, both happy and sad. Create empty boxes in your head where you can store feelings that you want to address a little at a time.
7. **Control the flow.** Remember that you can control the flow of emotions. Sometimes it's difficult and it often takes practice. The way to manage difficult feelings is to identify them, then address them one by one. But you don't have to do it alone. Ask a friend to listen.
8. **Maintain perspective.** Living with an illness means holding opposites at the same time. Together the opposites create the whole picture. For example, feeling hopeful requires the recognition of loss and the experience of grief. Another example is in appearances: Although you may look like a healthy young person, you may be suffering from significant limitations. Or the opposite: You may look different than others, but inside you feel just like everyone else.
9. **Know where and who you are.** Your disease changes over time and you change with it. Take time to stay up-to-date on who you are, what you want, need, think, feel. Change can happen slowly or very quickly. Keep track of the changes. Grieve the things you miss; celebrate the things you don't.
10. **Stay connected.** It's true you are the only one inside your body, but that doesn't mean you have to go through the disease process alone. You don't. So don't. Find people to walk with you, celebrate with you, listen to you, talk to you, guide you, follow you and just be with you.
11. **Express yourself.** Write it, draw it, sing it, say it, blog it, post it, flip it. Just get it out. Whatever it is. ■

Rebecca Block, MSW, LCSW, PhD, Oregon Health & Science University, presented at the Adults with FA Meeting in Portland, Ore., on Feb. 11.

Clinical Trial to Test Head and Neck Cancer Drug

Fanconi anemia patients with head and neck cancer who are candidates for surgery may be eligible to participate in a clinical trial for a chemotherapy drug called erlotinib (brand name: Tarceva). William N. William Jr., MD, from the University of Texas MD Anderson Cancer Center, spoke about the trial at the recent Adults with FA meeting. Currently, erlotinib is used to treat non-small cell lung cancer that has spread to nearby tissues or to other parts of the body in patients who have already been treated with at least one other chemotherapy medication and have not gotten better. Erlotinib works by blocking the action of an abnormal protein that signals cancer cells to multiply. This helps slow or stop the spread of cancer cells and potentially shrinks the tumor. This trial is designed to test the efficacy of the drug pre-operatively in combating head and neck cancer.

FA patients with head and neck cancer may be eligible to participate in a clinical trial.

Please note that the Fanconi Anemia Research Fund has a clinical trials scholarship fund to help defray the expenses of participation in approved clinical trials for FA patients who might not otherwise be able to participate. Contact Teresa Kennedy, the Fund's Family Support Director, for more information. ■

An Important Message about Oral Cavity Screening

Aggressive surveillance of the oral cavity is critical because FA patients are at significantly increased risk of head and neck cancer. Any questionable areas should be examined immediately. The third edition of *Fanconi Anemia: Guidelines for Diagnosis and Management* states, "Surveillance should begin by the ages of 10 – 12 years …on a semiannual basis by an experienced professional; *i.e.*, an ear, nose and throat specialist, an oral surgeon or other doctor experienced in head and neck cancer detection and treatment."

Just as important is a reminder that drinking alcohol and using tobacco products can significantly increase the risk of oral cancers.

Finally, all physicians and dentists who care for FA patients should receive a copy of the Fund's clinical guidelines book (referenced above). Contact the Fund for copies, available in both English and Spanish. ■

Since 1989, the Fanconi Anemia Research Fund has awarded 171 research grants to 89 researchers at 48 institutions totaling $13.2 million. For details, go to www.fanconi.org.

Nicholas: Our Angel on Earth

By Donna Boggs

After 16 years of marriage, Nicholas was born into our family in September of 1998. God had blessed us with a special baby boy with an infectious smile that would melt your heart in a millisecond. He wrapped you around his little finger at first glance. He made you take a second look: at him, your life, your blessings, and all the things around you that you take for granted. He drew us closer to God and made us do lots of soul searching, including trying to find a better way to live. He brightened our home in every possible way. He lit up your world and any room he was in. He radiated a pure, simple God-given love and just loved everyone

> He inspired poems, songs, careers, namesakes, but most of all, he inspired love.

he met. Nicholas always found the good in you and encouraged you to exercise it without judgment. He loved life and marveled at the simplest of gifts. He loved Camp Sunshine and all his "girls"! He inspired all he met in every facet of their lives. He inspired poems, songs, careers, namesakes, but most of all, he inspired love.

When Nicholas was 2, we brought home another gift from God named Spencer. When Spencer cried, it would break Nicholas' heart and he would want me to get him a bottle as soon as possible. Nicholas loved his brother and wouldn't do anything that would make him sad or cry. He was humble and always gave Spencer his way. He just wanted him happy.

Nicholas loved to laugh and sing. He loved his family, and loved to snuggle. He wanted to please. He was dynamite—power wrapped in a small package. He was a prankster and an imp with glinting eyes and a huge smile. He loved his Mamaw's and aunt's stories. He had a huge imagination and loved playing and acting out pretend games. He loved cowboy boots and hats. Red was his favorite color. His Dad's grilled steak was his favorite food, even though he couldn't really eat by mouth. He loved his Mamaw's salmon patties, Sissy Bear's chicken 'n' dumplings and cherry pie, his uncles'

> We will always miss our sunshine.

guessing games, Aunt Glenda's birthday cakes, and fussing with his Papaw over his cat, Baby Black Girl. He loved movies, old cartoons, *Batman* and *Ben 10*. He would be mischievous and his eyes would just twinkle. He was thoughtful and always wrote us love notes. He loved Jesus and believed his prayers would be answered, and they were. He was precious! He persevered through the difficult times and never complained. He was a survivor, a noble fighter, and fought to the end of his 12 years on earth.

When I asked Nicholas what he'd want if he could have anything, he said, "Time. Time with my family." Time is the gift without regrets. Truly, he was an angel on earth. We will always miss our sunshine. He was a true gift from God. He is our hero. ■

Search for Eli's Diagnosis Leads to FARF

By Tena Boson

The clock on the computer monitor said 2 a.m. I had spent hours searching for a diagnosis for my son's anomalies. Several tests for rare diseases had been negative. Our next appointment with the geneticist was two years out. I felt lonely and isolated as a mother. What had caused my baby all of these problems? My search led to Matt and Alex Pearl's website. Even though Eli hadn't had problems with his blood, the other issues the Pearls faced sounded like Eli's. After a sleepless night, I decided that eliminating this horrible disease would give me comfort. As I called our geneticist, I asked God for a sign. The receptionist said, "You won't believe this, but there is a note on his file with the words Fanconi anemia and a question mark attached." My sign was that Post-It note. While waiting for test results, two doctors expressed their doubts about Fanconi anemia, saying that Eli's wasn't a classic case of the disease. I prayed that their doubts would be right.

Eli joined our family, five weeks prematurely, in February 2009 after a complicated pregnancy. He had only one working kidney, which was scary. I will never forget my husband's look when he told me that our baby didn't have thumbs or ears. Eli was on life support and getting worse. I couldn't process what the doctors were telling me. Little things told me how serious this was: a private room in a full NICU, a free room to stay in at the hospital, and a life therapist for my healthy 6-year-old daughter, Sophia. We had planned a dedication for Eli the next day, but when a nurse said to make the dedication within two hours, I understood that our baby (whom I had yet to hold) wasn't expected to live. That was Valentine's Day. Eli's lungs were not working and the pressure to get him to breathe was so great that we risked blowing his lungs up. As his vital signs went down, the nurse cried and told me that she was sorry. But with my whole heart I believed that Eli would make it. By the grace of God, his lungs started to improve over the next week, but his kidney function was low. My heart was so heavy with all of this that it's impossible to describe. Eli slowly began healing, with many setbacks. When we left NICU, four nurses told us our child was a miracle.

Eli's life has been full of doctors, hospital stays, therapists, surgeries, chronic daily vomiting and love. He is a very determined, smart and independent boy. He is loving, energetic, and strong. As he nears his second birthday, he is learning to eat a few things like pizza, Cheetos and popsicles. Eli loves to wash dishes, play with electronics and chase his big sister. He is signing and talking, and every day we dance to music. Eli has been stable, growing and doing well. Every day, we thank God we have been blessed to love this amazing child.

When I received the call last fall that Eli was positive for Fanconi anemia, I was shocked and numb. I had commented on our last hospital visit that I never wanted to visit the oncology wing of our children's hospital, and now my child might have to. Thank you to the Pearl family for their website leading us to the Fanconi Anemia Research Fund that told us to test him. We are relieved that we have someone to turn to for advice, a listening ear and support. Thank you to many of you who have reached out to us. We are also grateful to the families that have put in their time raising money for research. Immediately, I felt that the FAmily had paid it forward for our son with the treatments that are available currently.

For so long I felt helpless not knowing what was wrong with Eli. Now that I know the Fanconi anemia diagnosis, I know I can make a difference in my son's health by raising funds for support and science. Even though I was timid about fundraising, I realized that Eli needs this research funded for his future and we need to "pay it forward," too. I sent out our first appeal newsletter a few days before Christmas. (Seeing that bake sales, lemonade stands and yard sale money is valued, too, gave me no excuse not to.) I am excited that the Delta Chi Fraternity at the University of Idaho is stepping forward to help us with a basketball tournament and more. I am looking for ways to have my daughter, Sophia, age 8, raise money, too.

Each day I am blessed to have a miracle to hug and kiss. Hearing his laughter and seeing him be a typical 2-year-old is a wonderful gift. ■

Living Life to Its Fullest Despite FA

By Belinda Gayle Angel

Belinda with husband, Shannon, and children, Corbin and Emma

If the world were a perfect place, there'd be no homelessness, starvation or disease. Diseases like Fanconi anemia. Unfortunately, we live in a world that is not perfect. Still, I have lived a full life for 29 years despite FA.

Let's go back 24 years. I'm 5 years old, bruising horribly and sick constantly. Deeply concerned, my parents began our journey, and I was diagnosed with FA in 1986. My father was diligent at finding the best care for me. He researched numerous treatment possibilities, care plans and hospitals. My parents knew that a bone marrow transplant was the only true option to save my life. There was only one thing missing: a donor. My brother, Billy, was born in 1987. He would hold the key to my survival since he was a 100% match.

I remember waiting for Billy to grow. He was, at the time, the youngest bone marrow donor on record, only 6 months old. My transplant was successfully performed in 1988 at the Fred Hutchinson Cancer Research Center in Seattle, Wash. I remember just about every day spent in that room locked behind that wall of plastic. But, I also remember the day I began the rest of my life. I had won my first battle with FA and was discharged from the hospital.

My late childhood was filled with many great memories—gymnastics, cheerleading, piano lessons. I was a typical child with a full life ahead of me. My adolescence was rocky. Like many teenagers, I participated in various questionable activities which led to a downward spiral of low self-esteem. I became severely depressed, overweight and finished high school with a bleak outlook on my future. But something inside me knew I was special. I knew I was here for a reason and that my life needed to change.

I enrolled in a few college courses. Then my wonderful son, Corbin, was born in 2002. Four years later, Corbin and I moved to Tennessee where I met Shannon Fred Angel who is now my loving and supportive husband. Shannon and I were blessed with Miss Emma who just turned 4 in March. After staying at home with Emma for a few months, I enrolled at Tennessee Technological

> I'm a strong woman who has overcome many adversities...

University in 2007. I graduated last December with my bachelor's degree in elementary education.

Our world may not be a perfect place, but I'm proud of where I've been and who I've become. I'm a strong woman who has overcome many adversities, and Fanconi anemia just happens to be a part of my proud journey. God bless. ■

FANCONI ANEMIA EDUCATION AND SUPPORT MEETING
SEPTEMBER 2011
LOS ANGELES CALIFORNIA

Please watch our website www.fanconi.org for more details.

Argentine FA Patient Plans Documentary

By Paula de Diego

I live in Buenos Aires, Argentina. I am 33 years old, and seven years ago I was diagnosed with Fanconi anemia. My only sister, Julie, died four years ago at the age of 26. She was diagnosed with FA at age 6 and lived a normal life until the age of 21, but then gradually things got complicated and she needed weekly blood transfusions and platelet transfusions. When the situation got much worse, she was hospitalized for four months and then died.

My case is different. I still do not need any strong medication and I remain stable, but my blood counts are low.

After my sister died, I thought about FA and how so many doctors have never heard of FA before. Even the head of the hematology department at a major hospital in Buenos Aires was unsure of how to effectively manage and treat my sister's medical care.

Since I work in the film industry, I've been considering the possibility of making a documentary about FA, primarily for patients and their families as a guide to find information and assistance. But the film should also help physicians and the general public to learn about FA.

So for three years I've been planning and working with this idea, and looking for funding in order to begin filming. In a way, the project is a tribute to my sister, who was a great artist. Not a single day passes that I don't think about her. I miss her terribly.

While researching FA, I came across the Fanconi Anemia Research Fund's website. I appreciate the information, the support, and the encouragement that I've received from FARF, as well as the encouragement and assistance to attend a patient support meeting.

I'm planning to attend the annual Fanconi Anemia Family Meeting in June in Casco, Maine, and am looking forward to meeting you all in person. You can visit my blog at **http://docanemiafanconi.blogspot.com/** ■

Congratulations to:

Lisa Doyle (FA, age 21) and Terry Kinsella on their engagement!

Karly Ross (FA, age 15) on her driver's permit!

Ashley Crow (FA, age 23) and Dan Spalding on their engagement!

Photo credit: Nic MacInnes

Ben Murnane (FA, age 26) on his second book, Feather Silence, Poems by Ben Murnane!

My Sister, My Hero

By Victoria Pless

Julia and Victoria Pless

I've always been told that if you were confronted with a challenge, it was because you were strong enough to handle it. How do you explain that to a 12-year-old after she finds out her little sister has a rare blood disorder called Fanconi anemia? When my sister Julia was diagnosed, I quickly realized that my life was falling apart. My family was suddenly thrown into a world full of uncertainty and fear, and I didn't know where this new road was leading us.

> Julia is my hero and the reason I push myself so hard in everything I do.

When Julia was diagnosed with FA, I hardly knew anything about cancer, let alone a rare blood disorder. What was it? How was I supposed to help? My sister was only 6 when she was diagnosed, which hardly seemed fair. She was just a child! Julia was already in bone marrow failure, and every single day was another day in which we would have to peek around life's corners to see what was coming next.

Fortunately for Julia, she was blessed to be matched with an unrelated donor for a bone marrow transplant just nine months after her diagnosis. During transplant, it was beyond difficult for me to watch Julia as she suffered. I was told that her health would improve, but what I saw seemed to go against what I was told. Normal daily activities were difficult for her. When nurses asked how much pain she was in, I couldn't watch as she pointed to the face on a scale that grimaced the most to show extreme agony.

My heart would break seeing Julia lying in her hospital bed. Surely this couldn't be my little sister, not my Julia. The beaming smile on her small, round face was now replaced with a painful frown and the straight black hair I loved was gone. I wanted to be the one in the hospital bed, to be the one who had the pain, to show her that as her older sister I would be there for her, to protect her from harm.

They say time heals all things, and I believe that to be entirely true. Julia was able to return home from transplant on her 100th day, which also happened to be her birthday. Julia is my hero and the reason I push myself so hard in everything I do. She overcame so much pain and so many struggles, and that makes anything that I must do look miniscule in comparison. People will come and go throughout her life, but may never truly understand her story. As for me, I'll stick by her until the end, because I've seen firsthand what she has gone through to get where she is now. I will never abandon my little sister.

Julia, I will always love you. You're my angel. ■

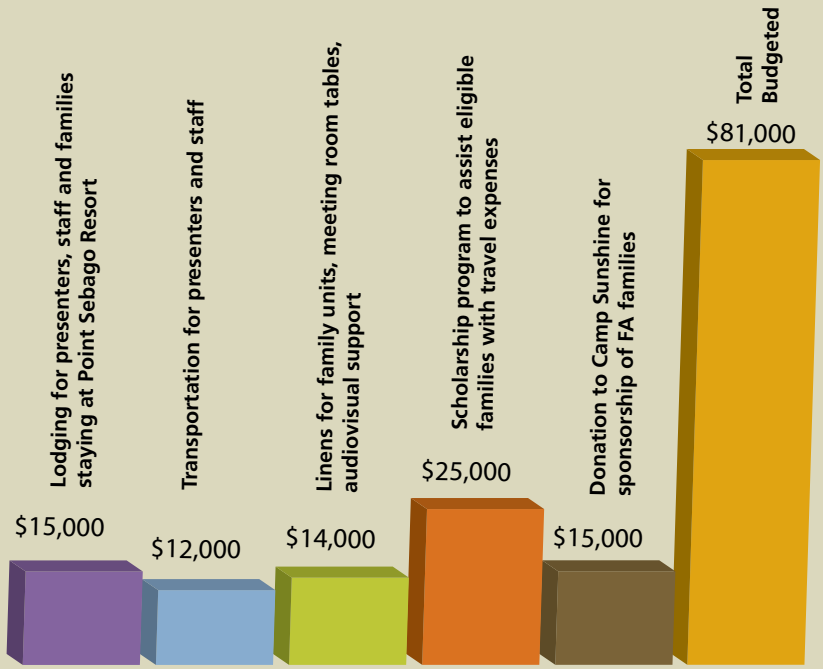
facebook

Find us on Facebook at
www.facebook.com
fanconianemiaresearchfund

Plans Shaping Up for 20th Annual FA Family Meeting at Camp Sunshine

We're busy gearing up for our 20th Annual Fanconi Anemia Family Meeting, June 24 to 28, 2011. A few logistical details have been crossed off of the list such as ordering linens for families and reserving lodging for our speakers and overflow families who stay at the resort. We're continuing to work on inviting FA researchers and new presenters, and developing new topics for the parent education program. For example, this year we hope to address the psychological considerations of going to transplant, quality of life and coping skills for adults with FA, and fertility preservation options for males and females with FA. Please check our website throughout the spring for updates on confirmed speakers and topics. We hope to see you in June! ■

2011 Budget For the Family Meeting



In Loving Memory

"For some moments in life there are no words."

Nicholas Boggs ..... 9/10/98 - 10/16/10

Kelly Turner ..... 3/22/83 - 10/17/10

Jeremy Bresette ..... 10/11/80 - 10/20/10

Saadia Usman ..... 11/29/01 - 11/5/10

Joshua Lytle ..... 10/31/06 - 11/25/10

Paula Ceresa-Guidara .... 7/25/54 - 12/14/10

Richard Briga ..... 9/7/57 - 12/18/10

Second Annual International Fanconi Anemia Day Invites Grassroots Fundraising

By Peg Padden

We're excited to announce the Second Annual International FA Day coming up May 1! Last year, 17 FA families raised more than $153,000 for the Fanconi Anemia Research Fund. Truly amazing! Just imagine what we can raise if we ALL get involved. You can do something small (think penny drive, bake sale), something bigger (yard sale, BBQ) or something bigger yet (auction, run/walk, bike ride) but please do something! It doesn't matter what you do or how much you raise… it all adds up. Remember: *Together,* we will find a cure! ■

FA Families Can Help Fundraising By Approaching Local Foundations

Each year the Fanconi Anemia Research Fund scours lists of private foundations and other granting sources to identify those that might be receptive to an application to help us fund our mission. What we've learned is that the foundations that are most likely to donate are those that have an FA family connection; for example, a foundation in Columbus, Ohio, might consider a donation because of the McCarthy family, or a foundation in New Jersey might donate because a trustee of that foundation knows Kevin and Katie Rogers and has attended one of their fundraisers. Relationships matter. People give to people they know.

In 2010, the Fund received more than $255,000 from about 30 foundations, in amounts ranging from $100 to $80,000, that were directly linked to an FA family.

If there are private foundations in your community, please consider personally informing them of the Fund and how it helps families like yours. We can even help you locate foundations in your area. Additionally, we'd be happy to put together a packet of information for you that tells the Fund's story and how the foundation's help would support our efforts.

Thanks for your consideration of another way to help us fundraise. ■

How FARF Can Help You Fundraise

More than 80% of the Fanconi Anemia Research Fund's annual budget comes from family fundraisers. We're here to help make your events a success. We can:

- Provide sample fundraising letters and help you edit your letter
- Use your photos to personalize your letter, event invitation or brochure
- Use your mailing list to send your letter or invitation from our office
- Provide ideas, information and display materials for events
- List your event on our website
- Send a thank you letter and tax receipt to your donors

We ask that you cover fundraising events with liability insurance. Insurance for a one-time event is often available through a family's homeowner's insurance policy as a relatively inexpensive insurance rider. Please contact the Fund if you need assistance obtaining or paying for this required insurance.

Please ask your donors to write their donation checks to the "Fanconi Anemia Research Fund." When a donation is received, we'll send a letter of thanks from the Fund with a tax receipt, and we'll notify you that a donation has been made in your name.

We appreciate all your efforts to raise funds to combat FA. You're making a difference! ■

Family Fundraising Efforts in 2010 Contribute to Banner Year

In the 12-month period beginning Jan. 1, 2010, Fanconi anemia families and their friends and supporters raised $1,705,920. That amount is the direct result of your efforts – letters you wrote, special events you planned and other personal appeals you made. Funds were also donated in loving memory or in honor of the life of a special child or young adult. Family fundraising was augmented by funds donated by the United Way ($3,500), the Combined Federal Campaign ($5,564), and a first-ever campaign aimed at FA researchers and clinicians ($6,215) for a grand total of $1,721,199, one of the top fundraising years in the last decade!

Thank you!

$600,000 and up
Dave, Lynn and Amy Frohnmayer

$150,000 - $199,999
Kevin and Katie Rogers/My Best Friend

$100,000 - $149,999
Kendall and Taylor Atkinson Foundation with the Nash and Atkinson Families

$50,000 - $99,999
Kevin and Lorraine McQueen
Paul and Rena Rice
Glen Shearer and Peg Padden

$20,000 - $49,999
Kerrie Brannock
Chris and Susan Collins
Peter and Tara Himmelreich
Brian Horrigan and Amy Levine
Steve and Jennifer Klimkiewicz/One for Wyatt
Todd and Kristin Levine
Dan and Nikki McCarthy
Mark and Diane Pearl

$10,000 - $19,999
Mark De Groot and Hanneke Takkenberg
Ed and Janice Duffy
Susan Gannon-Longstaff
Michael Glas and Carol Felmy
Charles and Katy Hull
Pedro and Marina Ravelo
Mike and Beth Vangel

$5,000 - $9,999
Jeffrey and Donna Boggs
Chris and Jennifer Branov
Joseph and Nancy Chou
John and Kim Connelly
David and Mary Ann Fiaschetti
Alan and Rachel Grossman
John and Raquel Hanna
Tanner and Jessica Lindsay
Deane Marchbein and Stuart Cohen
Jeremy and Stacey Mefford
Jim and Holly Mirenda
Jack and Lisa Nash
Bob and Andrea Sacks

$1,000 - $4,999
John and Audrey Barrow
Mark and Linda Baumiller
James and Tracy Biby
Donald and Danielle Burkin
David and Kim Chew
Tyler and Teresa Clifton
Lindsay and Sandra Dunn
Curt and Crystal Fales
Justin and Britteny Ferrin
Allen Goldberg and Laurie Strongin
David and Paula Guidara
Owen Hall and Margaret Kasting
Jeff Hoffman
Jeff and Beth Janock
John and Karilyn Kelson
Erik Kjos-Hanssen and Turid Frislid
Sejin Kwon and Jee-Ai Kim
Gregory and Lynnette Lowrimore
Tue Marker and Kirstine la Cour Rasmussen
Steve and Alison McClay
Sheila Meehan
Charles and Cecelia Meloling
Ian and Tricia Mitchell
Tyler Morrison and Rachel Altmann
Tony and Lina Nahas
Robert and Mary Nori
Peter and Janice Pless
John and Dianne Ploetz
Mark Ritchie and Lisa Mingo
Maureen Russo
Rick and Lynn Sablosky
Ron and Elesa Schaefer
Bill and Connie Schenone
Matt and Diane Senatore
Bryan and Karen Siebenthal
Mitzi Speelman
Charles and Jennifer Sumrall
Mark and Susan Trager
William and Mary Underriner
Marc Weiner
Michael and Kim Williams
Troy and Debra Williams
Wesley and Sue Wycoff
Sean and Kristin Young

Up to $999
Peter and Donna Abramov
Chris and Ellen Allums
Paul and Ann Anderson
Mohamed and Asma Alyammahi
Andrew and Vicki Athens
Yavin Atzmon and Sharon Harari
Keith and Lily Baggett
Gerald and Julie Barbier
Israel and Mary Jo Becerra
Conrad and Joan Bender
Darryl Blecher and Diana Fitch
Randy and Nancy Bloxom
Tena Boson
Roel and Diane Brand
Richard Briga
Robert and Barbara Capone
Lezlie Chesler
John and Launa Clanton
Jeanette Clark
Floyd and Susan Clark
Lauri Cohen
Daniel Conde
Natalie Curry
Brian and Margaret Curtis
Bill and Pat Danks
Richard Day
Tony and Phyllis Dellapenta
Donna DellaRatta
Wendy Delzell
Ginger Eggers
Kim and Stephanie Dillow
Pat and Mary DiMarino
Antonino and Marie DiMercurio
Brian and Jennifer Dorman
Delbert and Linda Dotson
David and Kelly Dunnock
Gene and Lynn Eddy
Sharon Ellis
Doreen Flynn
Bret and Nanette Foster
Gary and Melody Ganz
Stan Gilbert and Chris Brunner
Brian and Lisa Gillott
Pat and Maria Gleason
Andrew and Jennifer Gough
Mitchell and Tirzah Haik
Ousama and Souha Halteh
Bob and Victoria Hathcock
Sean and Helen Healey
Roger and Eleanor Herman
Alan Howard-Jones
Bonnie Hutchins
Lester and Nancy Jansen
Lila Keleher
Randy and Christie Kelley
Matthew and Evelyn Keyes
Keith and Krisstina King
Kayla Lackey
Marcus Lafore and Brianna Smith
Ernie and Nancy Landwehr
David and Shawn Leonardson
Peg LeRoux
Anne Llewellyn
Eric and Beth Losekamp
Bill and Jackie Lucarell
Ryan and Rita Lytle
Gil and Peggy McDaniel
Catherine McKeon
Gianna and Lauren Megna
Adam and Olivia Mindle
Sydney and Betsy Moore
Matt and Andrea Morris
John and Betty Mozisek
Des Murnane and Mai Byrne
Kenny and Lisa Myhan
Jack and Tammy Neal
Bob and Alice Nicholson
Fred and Nancy Nunes
Lorraine O'Connor
Michael and Katharine Ormond
Joshua and Crystal Pepper
Steve Perkins and Karen Magrath
Lori Petersen
Michael and Kay Proctor
Lynn and Shirley Quilici
Mario and Yolanda Ramirez
Gail Richardson
Leonard and Jan Riley
Richard and Dolores Satterlee
Sharon Saunders
Severt and Beatrice Score
Thomas and Brenda Seiford
Lorne Shelson and Annette Waxberg
Jim and Carol Siniawski
Tamara Stephens
Vicki Stephens
Greg and Brandi Stuart
Steve and Melissa Turner
Al and Michelle Valenzuela
Xiaoqing Wang and Ning Liu
Norman and Michelle Wilson
Gerald and Elizabeth Wisz
Jian Yang and Jing Nie
Tom and Marge Zaborney

For the fundraising events calendar and current FARF-funded research, visit www.fanconi.org

Going the Distance

26.2 miles. That's the distance logged by Ken Atkinson and each of four board members and friends of the Kendall and Taylor Atkinson Foundation (KATA) in the New York City Marathon in November. The KATA Foundation was founded by Ken and wife, Jeanne, in 2006 to support research through the Fanconi Anemia Research Fund in memory of their children, Kendall and Taylor, both deceased from complications of FA.

Ken and friends raised more than $30,000 for FA research through their long-distance effort.

"Running long distances does not come naturally for me. I am not fast, but I am determined," says Ken. "I have been blessed with the gift of good health, a gift that was denied Kendall and Taylor. I run in their memory because I can. I run so that other children with Fanconi anemia will live."

FA Fundraising Goes to School

Whenever there is talk of FA fundraisers in the Boggs household, Spencer, 10, brother of Nicholas, who died of FA last October, always wants to help, saying "I can sell those at school!" Spencer did just that last May with The Hand Project, selling paper hands for $1 imprinted with "I had a hand in a cure for FA!"

Spencer launched The Hand Project with a speech made over his school's PA system. Teachers got into the act, and students loved seeing "their" hand posted in the hallway for all to see. As word got out, the fundraiser grew. An aunt sold hands at her school, other FA families sold them at their fundraising events, and even local businesses and a prison joined in. The Hand Project raised more than $1,300, and Spencer is organizing it again this spring. Nicholas was so proud of his brother and Spencer is very happy—confident that he truly has a hand in a cure for FA.

Knit 1, Purl 2

friends & fibers for sofia

funding research for Fanconi anemia—one stitch at a time.

Barbara Smith took up her knitting needles when granddaughter, Sofia, now 5, was diagnosed with FA. Barbara and a knitting friend started Friends and Fibers for Sofia with the motto, "Funding research for Fanconi anemia – one stitch at a time." The Friends raise funds by selling their beautiful and unique hand-knit items at art fairs in the greater Washington DC and Baltimore areas. Even the price tags, designed by Sofia's mom, Holly, are a work of art, featuring a tangled ball of yarn that eventually spells out "thank you" along with Sofia's photo and story. Barbara is excited by the response to Friends and Fibers for Sofia—and the more than $6,400 raised for FA research!

A Capitol Achievement

Matt with State Rep. Tim Jones

Matt Pearl, 14, received the Glory of Missouri Award at the state capitol in Jefferson City, Mo., in February. The award is presented by the state's House of Representatives to students who exemplify one of 14 virtues chiseled in the capitol's rotunda. Matt was chosen to exemplify "progress," as a student who promotes positive changes in the school or community.

Matt's exemplification of this virtue was evident last spring, when his extraordinary efforts raised an astounding $22,500 through his fundraiser, Kick FA. Matt and his sister, Alex, both have Fanconi anemia. Matt says, "Thanks so much to all who make a difference every day for FA. I hope to have your support once again for the Second Annual Kick FA fundraiser so we can exceed last year's totals. I simply ask again, WWYD (what would you do)? I want to live!"

Caddy For A Cure

Christian and Calen Collins with PGA golfer, Phil Mickelson

Christian Collins, who has FA, was the impetus of Caddy For A Cure, created by family friend and professional golf caddy, Russ Holden. Caddy For A Cure raises awareness and funds by offering "inside-the-ropes" PGA tour caddy experiences to the public. One hundred percent of the proceeds benefit five charities, one of which is the Fanconi Anemia Research Fund. Since 2005, the Fund has received more than $60,000 in donations from Caddy For A Cure.

And Christian? Now 17, he is the national spokesperson for Caddy For A Cure, often joining the PGA tour where he has made "quite a few friends in some very high places!"

John Hanna Honored

John Hanna, an FA patient, was awarded the 2010 Legacy Cup by his coworkers at the Hilti Corporation. The company's highest employee award, the Legacy Cup recognizes a team member who is "a leader and leaving a legacy to build a better future." Hilti matched employee contributions to company fundraising drives to benefit a charity of John's choice. The FA Research Fund received $7,000 raised on John's behalf.

John's leadership in the FA community may be best summed up in his first message to families nearly 10 years ago: "I hope that I can be of some help to youngsters with FA who may be going through a rough time. I want them to know that it's not necessarily hopeless and that there are FA patients out there who are healthy. They can't give up hope. They can't give up."

John, ever inspiring and encouraging, is truly deserving of this honor.

A Big Sister Gives Back

Caren Hoffman, 17, found her purpose when her brother, Sam, had a bone marrow transplant four years ago. While staying at Ronald McDonald House, Caren started making art with hospitalized children which inspired her to become a leader-in-training at the Hole in the Wall Gang Camp. As a high school sophomore, she started a student club to raise money for families with sick children. The club has raised thousands of dollars for Relay for Life and local families. "Our town raised a lot of money for us when we went for Sam's transplant," Caren recalls. "I wanted to do the same for others."

As a result of her efforts, Caren was named a WebMD 2010 Health Hero, celebrating "visionary Americans who overcame daunting health challenges to give back to others in truly inspiring ways," and was featured in *WebMD the Magazine*. Caren donated her $2,500 award to the FA Research Fund in honor of Sam. Giving back, Caren says, "is the best decision I've ever made."

Run4Fanconi

Jason Brannock, 16, and friend Karly Ross, 15, are all smiles at the Brannock family's Second Annual Run4Fanconi. The September fundraiser included a 5K race and a kids' fun run on a scenic cross country course outside Raleigh, N.C. A drawing for runners who donated at least $20 on top of their registration fee awarded the winner a vacation cruise! This year's Run4Fanconi raised more than $7,400 for FA research and family support. Way to go, Brannocks!

Chess Piece

Benjamin, right, at Chess for FA

Benjamin Morrison, 14, held his first Chess for FA Benefit Tournament last May. Benjamin, whose younger sister, Nina, died of FA in 2006, recently wrapped up the second Chess for FA Benefit Tournament. His resounding success is evidenced by the numbers: participants in the tournament increased from eight last year to 19 this year, the number of schools involved increased from one last year to nine this year, and the number of sponsors and in-kind donors increased from three to seven! Benjamin also surpassed his fundraising goal of $1,000 by raising more than $1,300. Benjamin is on his way to becoming a grandmaster of fundraising!

Swing & Soirée

Bill Walton with his wife and brother

The Second Annual Swing & Soirée was held in San Diego last September. The event is hosted by the Lucky Duck Foundation, founded by Pat and Stephanie Kilkenny, friends to two families affected by FA. "Swing" included lunch and an afternoon golf tournament, leading to the evening's "Soirée" highlighted by boutique shopping, live and silent auctions, and great entertainment. Fabulous auction items included the chance to join San Diego Padres announcers in the broadcast booth, a behind-the-scenes tour of SeaWorld, and tickets to a Los Angeles Lakers game accompanied by basketball superstar Bill Walton. With a matching donation from the Kilkennys, the Second Annual Swing & Soirée raised $150,000 towards a cure for FA.

Dave Frohnmayer at the Hoot 'n' Holler

Hootin' and Hollerin'

Excitement was in the air at the Fourth Annual Brave Hearts Hoot 'n' Holler. More than 350 attendees and volunteers made it a night to remember and raised a record $154,000. The event is held by the Kendall and Taylor Atkinson Foundation and the Atkinson and Nash families. Dave Frohnmayer shared his insight as an FA parent and provided information and reason to hope. An auctioneer made giving fun, and a comedian had everyone in stitches. Guests bid on silent auction items and purchased squares in the Udderly Crazy Virtual Cow Plop(!). Another highlight was the Pony Up Corral—stick horses carrying a prize package of valuable goodies sell out quickly at $50 a pony. Casino games and dancing rounded out the successful evening.

They sure put on a FUN-draiser in Denver!

Some Enchanted Evening

Evan's Enchanted Evening

Kim and John Connelly just organized their sixth Evan's Enchanted Evening. The much anticipated event is an elegant and magical night of socializing, dinner and dancing, complete with live and silent auctions. Two of the Connelly's six children are affected with FA. Kim and John fundraise because, in their words, "We never want to look back on our children's lives and wish we could have done more." Although the proceeds from the sixth Evan's Enchanted Evening have not been finalized, the five previous Enchanted Evenings have raised more than $300,000 for FA research and family support. Now that's some magic!

The Fund Welcomes New Board Members

Richard Gelinas

Mary Ellen Eiler

Brian Matthews

Lynn Frohnmayer

Four new members joined the Fanconi Anemia Research Fund's Board of Directors recently, each for a renewable three-year term. They are:

Richard Gelinas, PhD, Senior Research Scientist at the Institute for Systems Biology, Seattle, Wash., and long-time member of the Fund's Scientific Advisory Board.

Mary Ellen Eiler, former Executive Director of the Fanconi Anemia Research Fund.

Brian Matthews, PhD, Emeritus Professor of Chemistry, Institute of Molecular Biology, University of Oregon, and former Howard Hughes Fellow.

Lynn Frohnmayer, MSW, Co-Founder of the Fund, FA parent, and former advisor to the board since its inception in 1989.

As we welcomed these new members, we said good-bye and thank you to **Peter von Hippel, PhD,** as his term ended on Dec. 31. Pete was on the Fund's board… forever! He brought a depth of scientific knowledge and a passion for the cause. He will be missed.

Dave Frohnmayer, JD, Co-Founder of the Fund, FA parent, and President Emeritus and Professor of Law at the University of Oregon, was designated an advisor to the Board. Dave had been a voting member of the board since 1989.

Thanks to all our board members for their time and dedication. For a full list of our Board of Directors please see the back page of this newsletter. ■

23rd ANNUAL

Fanconi Anemia Research Fund

SCIENTIFIC SYMPOSIUM

October 20-23, 2011

Princesa Sofia Hotel

Barcelona, Spain

FARF by the Numbers

1	Bottle of champagne to toast retiring board member Pete von Hippel at his last Board of Directors meeting
48	New FA patients/families registered in 2010
53	Number of countries from which people have visited the Fund's website
149	*Anemia de Fanconi: Lineamientos para diagnóstico y manejo, Tercera edición* books distributed
262	Total attendance at [three] science meetings held by the Fund in 2010
385	Registered members on the family support e-group
534	FA patients around the world currently registered with the Fund
596	People who "like" our Facebook page
4,413	Family fundraising letters mailed by Fund staff during the 2010 holiday season
4,681	*Fanconi Anemia: Guidelines for Diagnosis and Management, Third Edition* books distributed
9,008	Emails received by family support in 2010
20,000	Messages posted on the e-group since its inception in 1999
51,048	Dollar amount of postage to mail FA-related materials around the world in 2010

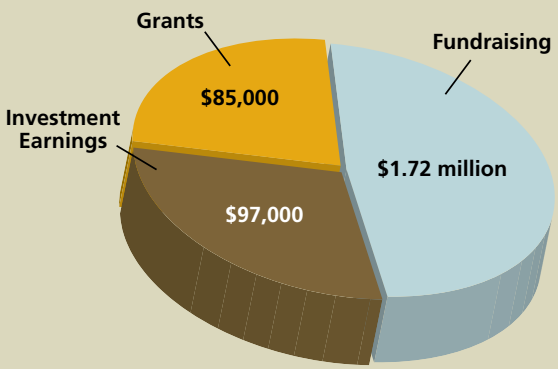
2010 Income and Expenses

The Fanconi Anemia Research Fund had a financially solid year in 2010. Together, families raised more than $1.7 million. The strength in fundraising helped mitigate the effects of a still-struggling economy. On the expense side, we spent $1 million funding research grants and scientific meetings and $230,000 on family support activities such as newsletters and the annual FA Family Meeting at Camp Sunshine.

Less than 10 cents of every dollar raised in 2010 went to administration costs.

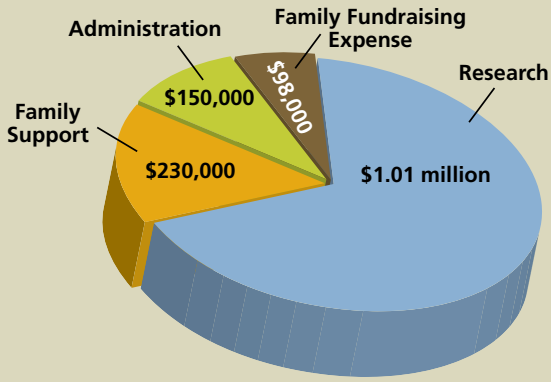
Thanks for helping us meet our growing needs! ■

2010 INCOME



Total Income: $1.90 million

2010 EXPENSES



Total Expenses: $1.48 million

Fanconi Anemia
RESEARCH FUND, INC.

1801 Willamette St, Suite 200
Eugene, Oregon 97401

REMINDER

Use of Logo

A reminder to our families: please use our logo or letterhead only after you have consulted staff at the Fanconi Anemia Research Fund and received approval. This step is necessary to be sure our messages are accurate and consistent and it helps avoid legal complications.

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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HOW YOU CAN HELP

Donations Online:	You can 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

Please go to www.fanconi.org to learn about other ways to donate.

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