

FA FAMILY NEWSLETTER

#47

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Spring 2010

Long-time Donors Pass the Million Dollar Mark

Pat and Stephanie Kilkenny were recognized by Fanconi Anemia Research Fund founders Lynn and Dave Frohnmayer for contributing an extraordinary cumulative amount to the Fund through Dec. 31, 2009: \$1,064,950.

Fittingly, the informal recognition took place at a University of Oregon football game in Eugene, Ore., where Dave and Lynn presented the Kilkennys with a commemorative crystal vase. Pat, a former Athletic Director at the University of Oregon, is a die-hard Ducks fan. He and Stephanie rarely miss a game.

“Stephanie and Pat Kilkenny’s generosity is truly inspiring,” says Dave. “Fanconi anemia patients

continued on page 19



Lynn and Dave Frohnmayer (center) with Stephanie and Pat Kilkenny

21st Annual Scientific Symposium

Nearly 200 members of the worldwide FA scientific community met in Baltimore, Md., for the 21st Annual Fanconi Anemia Research Fund Scientific Symposium last October.

Once again, the caliber, openness and dedication of the participants—traveling to Baltimore from 14 different countries—contributed to an exceptional three days of scientific collaboration and learning. The rapid advances made in understanding and treating Fanconi anemia are clearly fueled by this meeting.

continued on page 7

Gene Therapy Necessary before Creation of iPS Cells

Researchers from Spain demonstrated for the first time that skin cells taken from FA patients, following gene correction, could be induced to become pluripotent stem (iPS) cells. These cells can be cultured to become blood-producing cells, opening new treatment possibilities for FA patients.

Juan Bueren, PhD, CIEMAT, Madrid, and Jordi Surrallés, PhD, Universitat Autònoma de Barcelona, reported on the findings of their collaborative work with Angel

continued on page 7

HIGHLIGHTS

| | |
|--|----|
| Two Studies Suggest Strong Link Between HPV and Some FA Cancers..... | 2 |
| Eva Guinan, MD, Receives Distinguished Service Award | 5 |
| Family News..... | 8 |
| Fundraising..... | 15 |
| Research Funded in 2009 | 21 |

Two Studies Suggest Strong Link Between HPV and Some FA Cancers

High-risk human papillomavirus (HPV) subtypes are known to cause cancer in the general population, including oral squamous cell carcinoma (SCC) and anogenital cancers. FA patients are at extremely high risk for developing these same types of cancer. However, the role of HPV in FA SCCs remains controversial. Two studies are underway that help to explore this link.

Susanne Wells, PhD, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio, is exploring whether FA deficiency makes patients more susceptible to HPV-induced malignancies. High-risk HPVs carry the cancer-causing genes *E6* and *E7*, known to promote tumor growth and malignant transformation. Wells' group took cultures of FA skin cells called keratinocytes (some gene-corrected, some not) and inserted the

E6 and *E7* genes. The respective cell populations were then injected into immunodeficient mice, and tumor development was studied over time. FA-deficient keratinocytes rapidly formed malignant tumors similar to human SCCs. When *E7* was studied in isolation, FA gene deficiency increased protein expression of *E7*, thus revealing a possible mechanism by which FA could stimulate HPV-associated cancers. Dr. Wells is now studying oral cells from FA patients to determine if HPV is over-represented in these patients.

Jung Wook Park, a graduate student at the University of Wisconsin, provided additional evidence that the FA defect increases the likelihood that HPV infection might lead to certain cancers in FA patients. The lab of Dr. Paul Lambert created two types of mice: both carried



Susanne Wells, PhD

the HPV-*E7* cancer-causing gene but one group of mice had a normal *Fancd2* gene and the other group did not. Both groups were then given a drug that induces head and neck cancer. Mice in both groups developed malignancies, but the rates were

continued on page 11

Making Induced Pluripotent Technology Safer for Possible Therapies



Arleen Auerbach, PhD

In this same newsletter, Drs. Bueren and Surrallés from Spain describe their pioneering efforts to turn FA patient skin cells into induced pluripotent stem (iPS) cells. Their method relies on viral vectors to transport four different genes into FA skin cells. Arleen Auerbach, PhD, The Rockefeller University, noted that viral vectors cannot be removed from iPS cells or from blood-producing cells derived from these cells. This method creates genetic changes that are known to cause tumors in mice. She concluded that human trials based on this approach would not be safe.

Dr. Auerbach described a novel “ePiggyBac” system that does not use

viral vectors and is highly efficient in delivering genes. This technology, developed by her Rockefeller colleague Ali Brivanlou, PhD, has the added advantage that genes delivered to create iPS cells can be removed from the genome when their activity is no longer needed.

Dr. Auerbach and her colleagues have successfully used this approach on skin cells derived from an FA-C patient. They were able to combine correction of the FA defect with iPS reprogramming, while providing the possibility of removing tumor-causing genes. She described this as a “major breakthrough” toward clinical application of patient-derived iPS cells. ♦

Two Studies Agree on Major Findings: Clonal Aberrations are Persistent and Correlate with Poor Outcomes

Study #1: Heidemarie Neitzel, MD

In a 2003 article in *Blood*, Dr. Neitzel, Institute of Human Genetics, Charité Hospital, Berlin, Germany, described the relationship between bone marrow clonal abnormalities and hematopoietic disease progression in FA patients. She reported on her update of that study at our recent Scientific Symposium.

Of 177 patients studied, 57 had clonal aberrations in the bone marrow. Thirty-nine of these 57 progressed to myelodysplastic syndrome (MDS) or acute myelogenous leukemia (AML). Of the 120 patients without clones, only six developed MDS or AML.

Almost all (93%) of the FA clones were abnormalities of chromosomes 3 (3q-gain), 7 (complete or partial monosomy), or chromosome 1. Dr. Neitzel made the following observations:

- Clonal aberrations, mainly 3q, are the main cause for progression to MDS/AML. Of the 18 patients with a 3q-gain, only five are alive. All five had a bone marrow transplant. Of the 35 patients without this clone, 26 are living.
- Progression to AML occurs because patients wait too long after appearance of a clone, or are not screened often enough.
- Half of the patients with a 3q-gain subsequently develop a monosomy 7.
- The worst combination is a 3q-gain plus monosomy 7. These clones expand very aggressively within a few weeks or months.
- The clinical significance of 1q-gains is not clear.
- It is a myth that abnormal clones come and go in FA patients. Of the 57 patients with abnormal clones, only one experienced the disappearance of a clone. (An abnormality on chromosome 1 can sometimes disappear, but is replaced by an abnormal clone on 3 or 7).
- Conventional cytogenetic screening for abnormal clones is not sufficient to detect 3q-gains. Molecular cytogenetic techniques or I-FISH are needed.
- Decreasing blood counts and/or severe infections can suggest a clonal abnormality.
- Screening of peripheral blood with I-FISH is 95% effective in detecting these clones (compared to 100% with I-FISH screening of bone marrow).

Dr. Neitzel emphasized the tremendous importance of regular bone marrow or peripheral blood screening to monitor for abnormal clones.

Study #2: Betsy Hirsch, PhD

Dr. Hirsch, University of Minnesota, and her collaborators reported on the relationship between myelodysplastic syndrome (MDS) and abnormal clones in the January 2010 issue of the *American Journal of Clinical Pathology*. Bone marrow specimens from a total of 119 patients with FA were examined independently by a pathologist (to look for abnormal cells) and by a cytogeneticist (to look for abnormal chromosomes or “clones”). Among their findings were the following:

- The presence of an abnormal clone was associated with a 73-fold increase in the risk of myelodysplastic syndrome (MDS).
- Clonal abnormalities were present in 36 patients (32%). Of the patients with abnormal clones, 78% had abnormalities that included gains of material from the long arm of chromosome 1 (1q-gain), gains of material from the long arm of chromosome 3 (3q-gain), and/or loss of part or all of chromosome 7. More than one of these abnormalities frequently occurred together in the same clone.
- The 3q-gain is often not obvious and special techniques (such as FISH or CGH) are required to confirm it. Previously, therefore, these abnormalities were probably unrecognized.
- Patients with 3q and an additional abnormality all had MDS. When the cells have more than one chromosome abnormality, it appears that the 3q-gain comes first, and then the other chromosome abnormalities develop later.
- Multiple bone marrow samples from 8 patients were studied over time (one to five years). In no case did a clone identified at initial presentation disappear over time.

Dr. Hirsch kindly offered to be a resource to any parent or patient who has questions about this subject. Her email address is: hirsch003@umn.edu.

Factors That Correlate with Relative Longevity of Adult FA Patients

What are the characteristics of FA patients who live to adulthood? Holger Hoehn, MD and Detlev Schindler, MD, University of Würzburg, Würzburg, Germany are attempting to answer this question. With help from FARE, they recently completed a two-year preliminary study of 134 FA patients (92 from their own cohort; 42 from the literature) who reached ages 20 and older. They will continue their analysis of the original population and are expanding this study to 200 adult FA patients.

Among the 134 patients, 74 were ages 20-29; 46 were ages 30-39; eight patients were in the 40-49 year old group and 6 were older than 50. As expected, the majority of adult FA patients belong to complementation group FA-A, and all patients older than 50 were in this subgroup.

At present, the precise reason for prolonged survival has not been fully resolved in 53% of the adult patients. Early findings suggest that 26% survive because of stem cell transplant, 17% due to development of somatic mosaicism, and 4% because they have mild mutations.

Drs. Hoehn and Schindler next seek to clarify the role of androgen therapy in survival to adulthood. The researchers conclude that FA is in the process of evolving from primarily a childhood to an adult disease. ♦



Detlev Schindler, MD

High Incidence of Cancer in FA Adults Found in Longevity Study

A German study examining reasons for relative longevity in FA adult patients reveals an extremely high risk of squamous cell carcinoma

(SCC) in these patients. Researchers Holger Hoehn, MD and Detlev Schindler, MD, University of Würzburg, Würzburg, Germany completed a preliminary analysis of 134 FA adults over the age of 20. The prevalence of SCC was 40% in the 20-29 year-old cohort, over 50% in those ages 30-39, 60% in the 40-49 age group and 80% in the six patients older than 50. While bone marrow transplantation is known to increase the risk of malignancy, **an alarming 63% of the never-transplanted adult population developed SCC.**

Drs. Hoehn and Schindler cite studies that support the conclusion that FA cells are uniquely sensitive to oxidative stress. The SCCs that affect FA patients are mostly found in the oral cavity and anogenital region. These researchers speculate that contact with oxygen in the ambient air present at these sites may contribute to the distribution of these tumors.

Drs. Hoehn and Schindler will submit their final conclusions to the FA Research Fund upon the completion of this study. ♦



Holger Hoehn, MD

Editors' Note and Disclaimer

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Resveratrol Shows Promise in Partially Correcting FA Defect in Mice

Researchers in the laboratory of Markus Grompe, MD, Oregon Health & Science University, Portland, Ore., have been using FA mouse models to test various small compounds that might prevent or delay bone marrow failure and cancer in FA patients. Qingshuo Zhang, PhD, reported on recent findings using the drug resveratrol on mice bred to be defective in *Fancd2* (*Fancd2*^{-/-} mice). *Fancd2*^{-/-} mice have early defects in hematopoiesis such as significantly lower blood colony-forming abilities, so are appropriate models for testing potential therapies.

Researchers chose the compound resveratrol because it is a strong antioxidant and mimics the activity of “Sirt1,” which has been shown to enhance the capacity to repair damaged DNA. Resveratrol significantly increased the numbers of early colony-forming cells in these mice. The number of colonies doubled in most mice, and was in a normal range in one-third of the mice.

Preliminary data also suggest that resveratrol was able to delay tumor formation in *Fancd2*^{-/-} mice bred to develop epithelial cancers. **Dr. Zhang concludes that resveratrol is a promising compound that both delays tumor formation and improves hematopoiesis in FA mice.** ♦



Qingshuo Zhang, PhD

Eva Guinan, MD, Receives *Distinguished Service Award*



Dave Frohnmayer presents Eva Guinan, MD, with her award

Citing her extraordinary leadership in advancing clinical care for FA patients, Fanconi Anemia Research Fund Co-Founder, Dave Frohnmayer, presented Eva Guinan, MD, with the Fund’s *Distinguished Service Award* at the 21st Scientific Symposium on Oct. 2, 2009 in Baltimore, Md.

Dave noted Dr. Guinan’s role as chair of the Fund’s first Consensus Conference in 1998 to establish standards of clinical care, a difficult task given the variety of strong opinions on the subject. “Because of Eva’s expertise, professional reputation, tact, and profound belief in the importance of the end product, the first *FA: Standards of Clinical Care* was published to great acclaim and of great value to patients and their physicians,” said Dave. Dave added that Eva chaired the process again in 2003 and 2008. The 2008 Consensus Conference resulted in the publication of *FA: Guidelines for Diagnosis and Management*, a lifesaving guide for FA patients worldwide.

Dr. Guinan, a Harvard-trained pediatric oncologist, is also a member of the Fund’s Scientific Advisory Board, a group of dedicated researchers and clinicians who help guide the Fund’s research and clinical activities.

Dr. Guinan’s personal areas of research currently include problems related to mismatch in transplantation and the mitigation of regimen-related toxicity. In addition to maintaining a private practice, she is the Associate Director of the Center for Clinical and Translational Research at the Dana-Farber Cancer Institute in Boston.

FA patients, families and researchers alike know that they have a brilliant and caring friend in Eva Guinan. ♦

Tumor Necrosis Factor Contributes to FA-C Bone Marrow Failure and Can Be Suppressed by Pharmacological Agents

Grover Bagby, MD, Knight Cancer Institute at Oregon Health & Science University, Portland, Ore., has long asserted that FA genes have important functions independent of their roles in protecting DNA. Years ago he observed that the cytokine Tumor Necrosis Factor (TNF-alpha) is especially damaging to the cells of FA patients.

Dr. Bagby and his collaborators at the Indiana School of Medicine and at the Children's Hospital Medical Center in Cincinnati have established that TNF-alpha contributes to the aplastic anemia characteristic of patients in FA-C and can even play a role in the onset of leukemia. Not only are FA cells hypersensitive to TNF-alpha, they *overproduce* TNF-alpha. Bagby sought to identify the mechanism by which FA cells produce so much of this cytokine, and elaborates on his findings in the Dec. 17, 2009 issue of *Blood*.

Dr. Bagby has screened 120 compounds to determine which ones reduce the abnormal production of TNF-alpha by FA cells. He has identified two families of compounds (including FDA-approved drugs and drugs in clinical trials) that serve this function. Dr. Bagby will determine if his findings are relevant to all complementation groups. He is currently conducting experiments in FA knockout mice and expects that his work will lead to clinical trials of the most promising compounds. ♦



Grover Bagby, MD



Ian Mackenzie, DDS, PhD

The Challenge of Targeting and Destroying Stem Cells that Cause Cancer

Ian Mackenzie, DDS, PhD, Blizard Institute, Queen Mary University of London, noted that every tissue in the body, including malignant tumor tissue, has a stem cell component capable of self-renewal. Cancer therapies often rely on cancer cell death induced by DNA-damaging agents but cancer **stem** cells (CSCs) resist death, which can lead to recurrence of cancers. FA patients are at high risk for cancer recurrence and, as all of their cells are unusually sensitive to DNA damage, they could particularly benefit from agents that target and kill CSCs in other ways.

The small population of cancer stem cells present in both FA cancers and cancers from non-FA patients depends on certain signaling pathways that can be blocked to cause loss of CSC ability to maintain the cancer. Unfortunately, this strategy for targeting CSCs can also kill normal stem cells in the surrounding tissue. The challenge is to find CSC survival mechanisms that are not shared with normal stem cells, or are less important for their maintenance. Dr. Mackenzie believes this is possible, and reports growing interest by biomed companies in the development of such approaches. ♦



Amy Frohnmayer addresses participants at the Symposium dinner.

21st Annual Scientific Symposium *continued from page 1*

Forty-five researchers gave oral presentations and an additional 41 researchers gave poster presentations on numerous aspects of Fanconi anemia. Ann Gillenwater, MD, Director of the Oral Cancer Prevention Clinic at the University of Texas MD Anderson Cancer Center, gave a keynote address on her research involving novel technologies for the detection and diagnosis of oral cancer and pre-cancer, giving us hope that we may better address these life-threatening complications. M. William Lensch, PhD, from Harvard Medical School in Boston, Mass., gave a keynote address on the potential of iPS cells to cure the diseased bone marrow of FA patients—a significant breakthrough and exciting new avenue of research.

Three researchers received awards for their poster presentations:

Best Basic Science Poster: Judith Langenick, PhD, MRC Laboratory of Molecular Biology, Cambridge, UK.

Best Clinical Poster: Melissa Araujo, PhD, University of Sao Paulo, Curitiba, Brazil.

Best Translational Poster: Holger Tönnies, PhD, University Clinic Schleswig-Holstein, Kiel, Germany.

Fanconi anemia patient and Stanford University graduate student, Amy Frohnmayer, spoke at the Symposium's dinner. Amy thanked the researchers at the event for their efforts and remarked on her future plans for continued studies and for life's possibilities.

At the meeting's conclusion,

Grover Bagby, MD, chair of the Fund's Scientific Advisory Board, reported that he found the 2009 Symposium "open, friendly and collegial." Dr. Bagby noted that there was a good balance between basic science presentations and presentations that were more "translational," that is, with potential for clinical application.

Fund staff is at work planning the 22nd Annual FARF Scientific Symposium which will take place in Minneapolis, Minn., Oct. 21 to 24, 2010. ♦

Following is a sampling of quotes taken from the Symposium participant evaluations:

For me, the power of the meeting is the bringing together of all angles...which generates lots of new ideas and potential collaborations.

Every talk and poster is something new, contributing to both medical and molecular understanding of the disease.

We are following some interesting new leads and hopefully will have some interesting data to present next year.

Gene Therapy Necessary before Creation of iPS Cells

continued from page 1

Raya, PhD, and Juan Carlos Izpisua-Belmonte, PhD, both from the Center for Regenerative Medicine, Barcelona, at the October FA Scientific Symposium. Researchers were successful in obtaining iPS cells from three of six FA patients. A crucial step in their strategy was the genetic correction of the disease in the skin cells by gene therapy. They report that 12 out of 28 reprogramming attempts were successful when using genetically corrected skin cells versus none out of 28 attempts with uncorrected cells. Additional studies to improve the efficacy and safety of iPS cells and derived blood progenitors are required before this approach can enter human clinical trials. ♦



Jordi Surrallés, PhD

FAMILY NEWS

Blake & Sydney

by Rena Rice

Our first child, Blake, was born March 26, 2000.

As first time parents, we were excited to learn the gender of our baby so we paid for an ultrasound and a sneak peak. This is when we learned that something wasn't right. I had excessive amniotic fluid, which meant that our baby was not taking in enough nutrition for proper growth. We were told that this condition is sometimes caused by a birth defect where the esophagus is not connected to the stomach. Thanks to the ultrasound, our physician was able to monitor the pregnancy closely and scheduled a C-section to reduce delivery risk.

When Blake was born, we felt like our worst fears were realized when we learned that, in fact, his esophagus was not connected to his stomach. Immediate surgery was necessary to correct the problem. In addition, Blake was born with low birth weight (4lbs, 11oz), an absent right thumb, and an underdeveloped left thumb. Although Blake's surgery went very well, he was in the hospital for the first 42 days of life. After various tests Blake was diagnosed with VATER (or VACTERL) because of the abnormalities of his trachea, esophagus, and hands. Because his seemingly random physical anomalies appeared to fit this diagnosis, we didn't question it further.

From the beginning, Blake faced feeding and eating challenges. He had difficulty consuming even the smallest amounts of milk and had little desire to eat. Blake had two feeding tubes before the age of two. It took years for him to overcome



Blake and Sydney Rice

different food textures and to get to the point where he was willing to try new foods. Although we continue to struggle with his calorie intake, Blake has made great strides.

By the time Blake was a year old, he had fully recovered from his esophagus surgery and we decided it was time to address his hands. A hand surgeon evaluated Blake and recommended surgery to create functional thumbs. Knowing that this procedure was "elective" in nature made it a tough decision, but after some research we both felt that this was the best course for our son. This procedure would allow Blake to perform anything that required a thumb/finger pinch grasp, such as cutting, writing, buttoning clothes,

zipping and grasping a baseball bat. Blake's surgeries were very successful; no physical therapy was required—his play was his therapy. It has been beautiful to watch the evolution of his new thumbs and we've never regretted our decision. Interestingly, our hand surgeon did question us about Blake's condition. We were surprised and a little offended when he wondered if there had been drug involvement during pregnancy. In retrospect it's clear his questions should actually have suggested another diagnosis that could have explained Blake's hand anomalies.

During the fall of 2005, we were blessed with our beautiful daughter, Sydney. We were thrilled that our

continued on page 13

Lily Lou's Amazing Year

by the De Groot Takkenberg Family

It is Feb. 13, 2010 and we are all in the pool of our local town in the Netherlands to cheer on Lily Lou (age 7) trying to pass her swimming exams almost 11 months after her bone marrow transplant. She has so much fun in the pool. At the end of the exam, her sister Jasmin and her best friend Mirthe jump into the pool with their clothes on to join in the excitement. Lily Lou receives a beautiful award and a gold medal from her sister. We are very proud of her!

Lily has been doing great since she got her new bone marrow from an unrelated donor. Our next check-up will be in six months.

Lily Lou's story starts in Massachusetts on Jan. 19, 2008, when she was 5 years old. During her sister Michelle's basketball game, she got hit in the nose by an errant ball. We couldn't stop the bleeding and rushed her to Children's Hospital in Boston, where we found out she had Fanconi anemia with bone marrow failure.

After consultations with Drs. Colin Sieff and David Williams in Boston, and a blitz visit to Paris, France to see Prof. Eliane Gluckman, we decided to have Lily Lou treated in the Netherlands. Being Dutch expats in the U.S., it felt best to be among family and friends while going through the inevitable stem cell transplant. Given Lily's common HLA type, we were assured that finding a suitable donor shouldn't be a problem. We met with Dr. Marc Bierings, head of the pediatric stem cell transplant unit at Wilhelmina Children's Hospital in Utrecht, the Netherlands, at the beginning of the summer of 2008. Utrecht provided us with the great option of staying with Lily Lou in isolation 24 hours a day, which was really

important to her. Also, the modified stem cell transplant protocol for FA patients—no radiation therapy and mild chemotherapy—appealed to us.

After moving back to the Netherlands, we began to prepare ourselves for the transplant as Lily's levels were dropping quickly. Lily received several transfusions to keep her going while waiting for a suitable donor. In the beginning of 2009, we got the fantastic news that a 10/10 match was found. Lily was admitted on March 4 and received her new bone marrow on March 18. Although the transplantation process was challenging, we were proud of Lily Lou for being brave and always keeping her thumbs up. On April Fools' Day, Lily Lou's new bone marrow showed the first signs of engraftment and she was discharged two and a half weeks later.

After being home for three weeks, Lily Lou and her sisters got a little baby sister, which helped the family to focus on something else. 2009 has been an amazing year for us all. Lily is back in school and doing all the things normal 7 year olds are doing, with a high level of energy. We feel that together with the fantastic support of family, friends, neighbors, school, work, and faith in the continuous progress of research and medicine, we have changed our lives for the better. We cherish every day that we are together and have grown closer as a family. We are fully aware that Lily Lou still has a lot of challenges ahead of her but we will worry about these things when the time comes. It is so great to live in the moment and to see her walking around with her gold medal. ♦



Sisters Michelle (11), Rosaluna (10 months), Lily Lou (7), and Jasmin (14)



Participants at the 2009 Brazil Family Meeting

First Annual Brazil Family Meeting a Success

The first Fanconi Anemia Family Support Meeting in Brazil was held Nov. 20 and 21, 2009 in Curitiba, Brazil. The event was organized by Carmem Bonfim, MD, Director of the Hospital de Clinicas Pediatric Stem Cell Transplant Program and Coordinator of the FA Outpatient Clinic. Dr. Bonfim reported that the meeting was a great success—almost 200 people, including 79 FA patients, attended from all over Brazil. The event was so well received, in fact, that many attendees slept on floors as there were not enough rooms to house them. Dr. Bonfim noted that everyone was very happy to attend, meet each other, and eager to learn more about FA.

Patients and families attended clinical presentations by Ricardo Pasquini, MD and other physicians, asked questions, and discussed treatment and psycho-social issues. Eunique Velleuer, MD and Ralf Dietrich traveled from Germany and collected

oral samples for squamous cell carcinoma screenings from almost all of the patients. In sum, Dr. Bonfim wrote that the meeting “was great,

worth every single minute.” A newly formed organizing committee is already making plans for the next meeting. ♦

In Loving Memory

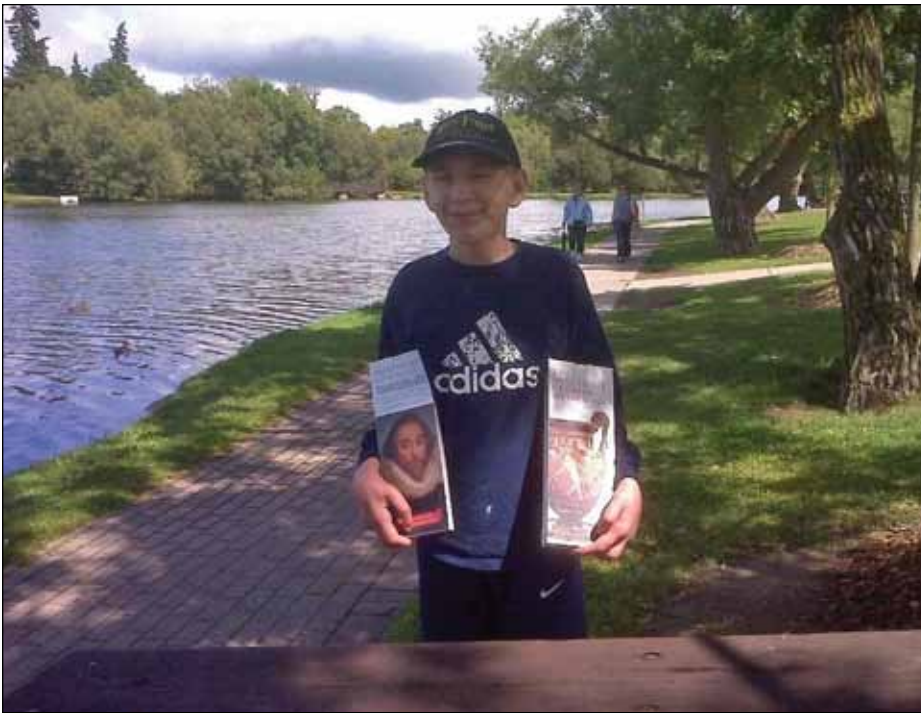


Samantha McCarthy
3/15/98 ~ 10/15/09

Charisse Howard-Jones
5/17/78 ~ 12/3/09

Valerie Comfort
9/24/64 ~ 1/11/10

Anne Llewellyn
12/12/69 ~ 1/30/10



Aaron Shelson age 16, 7 years post-transplant

7th Re-Birthday Promises Exciting Experiences for the Future

by Lorne Shelson

This past weekend, Aaron Shelson celebrated his 7th re-birthday since having an unrelated transplant in Minneapolis. He was discharged from the hospital on Day 20, readmitted briefly around day 25, discharged on day 28. Around day 100, we were headed home to Toronto.

Since transplant, he has visited Newfoundland, gotten a Dutch sheepdog (big mistake!), eaten caribou, flown to Iceland, walked through the New Hampshire woods, seen whales, met his donor, eaten blood pudding (if you don't know what it is, you don't want to), climbed a volcano, read all the Harry Potter books, played with a crab in a tidal pool in New Brunswick, ridden Splash Mountain (but didn't enjoy the fast part), hung out with Bill Cosby (they go way back), seen the original geyser

(called Geysir), been up close and personal with a moose, had cataract surgery, gone swimming, seen *Billy Elliot* on Broadway, memorized all the songs in *Sweeney Todd* (the stage musical and the movie), started riding the subway home from school and much, much, much more.

Aaron is now a grade 11 high school student. He has been in several school drama productions and studies improvisation at Second City's training center here. He plans on becoming an actor. He will be a great one. ♦

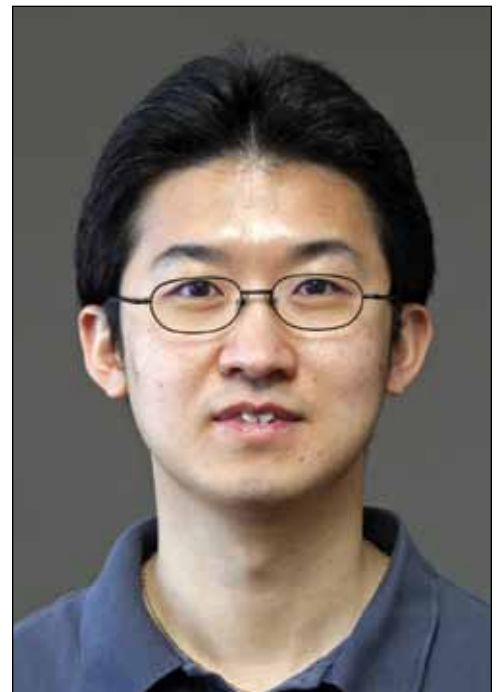
The FDA approved the HPV vaccine for boys ages 9–26 in October 2009.

Two Studies Suggest Strong Link Between HPV and Some FA Cancers

continued from page 2

significantly different: 20% of mice with a normal *Fancd2* gene developed cancer, compared to 52% of mice with a deficient *Fancd2* gene. Park then tested FA normal and FA affected mice that were not expressing HPV-E7. Neither the FA normal nor the FA deficient mice developed cancer after treatment with a cancer-causing drug.

These studies support the belief that FA patients are predisposed to HPV-associated head and neck cancers. **This research suggests that HPV vaccines may help prevent these cancers in FA patients.** ♦



Jung Wook Park, Graduate Student, University of Wisconsin

The Clifton Family vs. Fanconi Anemia

by Teresa & Tyler Clifton

Seven years ago, we welcomed our little Emma into the world. She was an attention-seeker from the beginning and needed open-heart surgery (TAPVR) when she was five days old. She also had many other abnormalities and we had no idea why or what had caused them. We imagined it was a fluke and that after she was finished with the heart surgery ordeal, we'd meld back into life and continue on with our beautiful baby girl.

It was quite a shock when she was 11 days old, and the geneticists sat down with us and told us that our little Emma had Fanconi anemia. We'd never heard of it, much less knew that anything like this existed. The words "bone marrow failure", "leukemia", and "cancer" seemed to echo through our minds as we reeled at the news we were given. It felt like the world stopped and was shocked with us.

But life continued, and we learned more and more about FA. It didn't bring a lot of comfort by understanding it better. We learned how aggressive and life-threatening the disease is. We connected with other families and watched as many of them lost their children to the disease, but we also saw many children survive transplants. Regardless, it wasn't an easy reality to accept.

We felt like we had a choice—we could become overwhelmed by FA and the fear of losing our little girl, or we could control what we could control and that was to give Emma a happy, fulfilled life. We chose the latter.

When Emma was 3 years old, we began seriously considering having another baby. It's a hard decision to make, but we made it and felt confident with our decision. We trusted in the Lord to bless us with the baby

that we were meant to have, FA or not. When Emma turned 4 years old, her baby brother, Tyler, was born. He was not a match for Emma, but he was healthy, and they've become the best of friends.

A year later, we felt it was time to have another baby. We did, and our little Violet was born. She was also healthy and was also not a match for Emma. She's been such a wonderful little addition to our family.

We are currently pregnant with baby #4. We just found out that it's another little girl, and according to ultrasound, she looks completely healthy. We'll know for sure once she's born, and we'll also find out if she's a match for Emma.

For us it was important to continue on with life. We couldn't let FA decide whether or not we continued our family. We feel blessed to be parents of such wonderful children. We are so thankful for our little Emma and every moment we get with her. We would choose time and time again to have her in our family and feel the same applies to any of our children, FA or not.

A couple months ago, Emma got sick with what seemed like a couple of minor illnesses. Over a six week period, her blood counts plummeted to transfusion levels. On New Year's Eve, she received her first red blood cell and platelet transfusions.



The Clifton Family

Two weeks later, she had another red blood cell transfusion and needs platelets whenever she bleeds. We're waiting and hoping and trying androgens to see if we can get her blood counts back up. So far not a lot seems to be changing, and she'll likely need another transfusion in a few days.

This FA journey is bittersweet and even terrifying at times. It brings with it a new set of emotions. Life can feel so uncertain and so threatening. But even with all of that, and while it really doesn't make any sense, we have never been as happy as we are now. ♦

Blake & Sydney

continued from page 8

second child was the picture of good health. And Blake was thrilled to have a little sister.

Throughout Blake's first 7 years, he was prone to respiratory issues and once developed pneumonia. These problems seemed to escalate during his eighth year, with two episodes of pneumonia in a six month period. Blake frequently seemed tired, and his energy levels weren't what they used to be. It was certainly noticeable, but there was always something to blame, such as, "he's just getting over being sick."

When you see someone every day, you don't always see a slow decline in health. When Blake was 8, his dentist casually mentioned that he looked a little pale. I dismissed it, thinking his coloring has never quite looked like mine (or my husband's for that matter). We assumed paleness was most likely due to his recent pneumonia.

A month later, we returned to the dentist for his last cleaning before braces. The dentist once again mentioned that Blake appeared pale. I immediately thought I had better check this out. Blake's pediatrician ordered a blood count. The initial results came back so low that the nurse said the machine must be broken. The doctor sent us to the main lab. Hours passed and the kids and I were on our way to a Halloween store, when I got a call from my husband, saying we needed to get home now—they had a bed ready for him at the hospital. Tears filled my eyes, and I instantly felt nauseous. We had no idea what was wrong but were thrown into an instant state of panic. We headed straight to the hospital where they did another blood and a bone marrow test and gave him a transfusion.

After a battery of tests and a thorough review of his health history, the



Alissa, Tyler, and Patrick O'Toole

Congratulations!

We would like to extend our best wishes to Alissa O'Toole (FA, 33) and her husband Patrick who recently welcomed a new son, Tyler, to their home through adoption. ♦

hematologist told us that Blake didn't have leukemia but rather he suspected Fanconi anemia. Within a week his suspicions were confirmed. We started researching FA and concluded that working with Minnesota was our best option. We entered survival mode.

At our appointment with Dr. MacMillan at the University of Minnesota we learned all about the treatment options. Our doctor also recommended that our 3-year-old daughter Sydney be tested for FA. We were surprised because we assumed that Sydney was in perfect health. Furthermore, our local hematologist who had diagnosed Blake had said in a matter of fact way that she didn't need to be tested because she had no signs of FA. The weekend

before Thanksgiving, we received a call from our doctor in Minnesota, updating us on Blake's donor information. As I was taking in everything she was saying, I suddenly heard the

continued on page 24

Use of Logo

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

This One Goes Out to Sam!

by Nikki McCarthy

From the day she was born, Sam was full of personality. At a young age she had a very extensive vocabulary, just like her little brother, Jack. She had dark hair and a cute chubby round face, just like her brother, Joe, with whom she also shares a very giving and loving heart. She was also a sensitive music and animal lover, just like Finn, our oldest son.

So Sam is right here with Dan and me every day. We see her very clearly. Her presence goes beyond the fact that her room remains the way it was when she left for the hospital the last time, and beyond the fact that there are pictures and reminders of her all over our house. “*She made her mark but was just getting started. It wasn't long enough together, but it was long enough to last Forever.*” Those are the words of a Rascall Flatts’ song, one of Sam’s favorite groups.

So many of my memories of Sam come with a soundtrack. Although I’m not a musician or musically gifted in any way, I can remember the words to most songs after hearing them just once or twice. Sam was the same way. We love music in our home and play a lot of it. Our enjoyment of music seemed to rub off on our kids. Sam was turning into a great piano player and she spent a lot of her free time listening to music. She liked the typical 10-year-old girl stuff like Miley Cyrus, Demi and most of Radio Disney, but eventually she found new and different music that Dan or I liked. Two things that were always tucked in her bed next to her (other than Baby Baby and Carrot) were her phone and iPod, access to the world—and to her own world—when she needed to escape.

The soundtrack of memories is full of love songs, but not the really sappy, slow kind. Most songs are about loving and then losing



Finn, Nikki, Joe, Sam, Dan, & Jack McCarthy (left to right), summer of 2008

someone, or a really tough time that someone has overcome—that would be us! It can be a song we loved and sang together, a song that played the day we came home from the hospital, an artist that she and I loved, or one that I know she would have liked. She knew that we were “*Better Together*,” a song by Jack Johnson, one of her favorite artists.

Together continues to be how our family is dealing with Sam’s absence. We find that love is the only thing that gets us through each day. We have a strong love for each other and for family and friends who picked us up and followed us “*On and On*” (a song by Mat Kearney) throughout the last 17 months or so. While that journey seemed to go on and on, we decided the best thing we could do was to go on as a loving family. We will not let FA or any other sadness take away what we have built

together as a family. We are fortunate and lucky in so many ways. It just so happens that as blessed as we are, we have experienced an equal amount of pain with this loss.

Although we “*Don’t Know Why*” this happened (a beautiful song by Shawn Colvin), Sam will always remain the love of our lives—all five of us in different ways. She is our “*Hey Soul Sister*” which is our favorite song (by Train) that makes the boys and me think of Sam. When it comes on, we turn it up loud! ♦

FUNDRAISING

6th Annual Valentine 5K Unites Patients and Families from Across the US

by Peg Padden

The 6th Annual Valentine Fanconi Anemia 5K and *first time ever* 8K run/walk was held (fittingly enough) on Feb. 14 in Portland, Ore.

Eight hundred and fifty four people registered for the run; 58 people volunteered and together we raised \$33,000 for research. In addition, 35 people were voluntarily added to the bone marrow registry via on-the-spot cheek swabs. Wow!

What made things extra special this year was the larger-than-ever involvement from the Fanconi anemia community. FA individuals, families, researchers and staff from FARE were all a part of the event, each doing his/her part to make a difference.

Spencer Shearer was on the route cheering people on and pointing them in the right direction; Amy Frohnmayer and Roy Proctor ran the

course; Josie Proctor Keyes walked along with her husband, (very cute) baby, Clover, and her parents, Kay and Mike; Nikki McCarthy ran the race in memory and honor of her daughter, Sam; Rachel Altmann and son Benjamin passed out T-shirts in memory and honor of Nina; and Mark Pearl's cousin, Candice Behm, also participated. Rhode Islanders, Mary Ann Fiaschetti and her son Peter, going through transplant, registered online as virtual walkers. Next year, they'll walk it in person.

FA researchers from OHSU and many of their family members were there as well. Bev Mayhew and Teresa Kennedy from FARE drove two hours from Eugene to volunteer, participate and be a part of it all.

Next year will be quite exciting as the (2nd ever) Adult FA Meeting will be held in Portland the same

weekend as this event. FA adults may run, walk or volunteer, widening the FA community involvement.

Why does this matter? What does it all mean? What it means is that we're not alone. Fanconi anemia can be isolating and sometimes seem hopeless, but it isn't when this many people band together and work as one. We're not alone. Whether we're affected with FA, have a loved one affected, are a researcher or work for FARE, we're all in this together with the exact same goal: Find a cure! Alone we can't reach our goal. Together we can. And we won't quit until we do. ♦

1st International FA Day Connects Families around the Globe

by Peg Padden

The First International FA Day is on May 1, 2010. FA individuals, FA families and friends and relatives of FA families from around the world will hold events to raise money for research. Some of the events planned include car washes, garage sales, an author talk by an FA mom, T-shirt and candle sales, and a chess tournament. We are very excited to have the FA community from all over come together at this one time to work toward our common goal: Research for better treatments and some day... a cure!

If you would like to be a part of this special day, either for this year or next, contact Peg Padden at pegpadd-pad@hotmail.com ♦



Amy Frohnmayer (front center) with Roy, Kay, and Mike Proctor

America's Giving Challenge

This year, the *Parade* magazine's 2nd Annual America's Giving Challenge proved to be a successful fundraiser for the Fund. Over the course of 30 days between October and November, the Fund received 631 donations through Facebook totaling an impressive \$12,749! Included in the total was a \$1,000 bonus for having the largest number of donations on Oct. 16. Thank you all for your help in spreading the word about the Challenge and for your generous donations! ♦

A Generous Gift from Across the Atlantic

Fanconi Hope, a UK-based charitable trust, donated \$50,000 to the Fanconi Anemia Research Fund in December. The generous donation—the first from a charitable organization outside the US—will support an FA research project led by Majlinda Lako, PhD, in Newcastle, UK. Dr. Lako's research will look at the use of stem cell technology in conjunction with gene therapy to cure Fanconi anemia. She will utilize iPSC technology to understand early hematopoietic development in Fanconi anemia patients.

"Every donation moves us closer to finding a cure, and Fanconi Hope's generosity provides a meaningful leap forward," Barry Rubenstein, the Fund's Board President, wrote in a thank you letter to Fanconi Hope's principals, Thomas Carroll, MD, Chairman and Bob Dalgleish, Secretary.

Although founded less than two years ago, Fanconi Hope has made impressive gains. To learn more about Fanconi Hope, visit its web site at www.fanconi.org.uk. ♦

How You Can Help

Your donations have helped move this fatal disease from an orphaned status in 1989 to a disease with treatments that now buy precious time for FA patients. As the genetic basis of Fanconi anemia continues to be deciphered, your donations are also having an impact on the lives of millions in the general population. We continue to move to the mainstream of scientific interest. To help us in this fight, consider these ways to donate:

Gifts to celebrate an occasion: If you are celebrating a birth, a birthday, an anniversary, a graduation, a marriage or other event, consider asking that donations be made to the Fund in lieu of a gift.

Gifts to commemorate a loved one: Families who have lost a loved one may ask that a donation to the FA Research Fund be made in their memory. The Fund has received many thousands of dollars from caring people who have commemorated loved ones in this way.

Bequests: If you are preparing or reviewing your *Last Will and Testament*, consider making a bequest to the Fund.

Matching Gifts: Many employers match the charitable gift of an employee. Ask if employers have taken this initiative to encourage philanthropy. This is an excellent way to double your donation.

Gifts of Appreciated Property: Donors who have property that has gained greatly in value (stock, vacation homes, art items, etc.) can avoid tax liabilities and provide enormous support by donating this property to the Fund. Please contact us for advice and suggestions.

Sales on eBay or Purchases through iGive: If you sell an item on eBay, you can designate that all or a portion of the proceeds be given to the Fund through their non-profit MissionFish program [see www.missionfish.org]. You can also donate to the Fund by shopping online through iGive [www.igive.com].

United Way or Combined Federal Campaign: If you work for an organization that participates in either of these campaigns, consider making a donation and asking your colleagues to do the same.

Donations Online: You can donate via the *PayPal* button in the *Donations* section of our web page (www.fanconi.org) or through www.networkforgood.org.

Donations by Telephone: Call us at (541) 687-4658 or toll free at 888-FANCONI.

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

One Stitch at a Time

by Barbara Smith

My beautiful granddaughter, Sofia Miranda, was born in May 2005 with FA. As her grandmother, living many states away from her, I felt at a loss for what I could do to help.

NOW, I have found my way.

With a suggestion from a friend and fellow knitter, we started *Friends and Fibers for Sofia* in November 2008—knitting and felting purses, hats, mittens and other items. Our plan was to sell our work and donate all proceeds to FARE.

We had no idea how our knitted and felted items would be received. They were an absolute hit right from the start. We started selling our hand-knitted items at a local Christmas Bazaar. We knit bags of every size, and often attach a piece of old jewelry to make the finished product a complete original!

In the 15 months since we started, I have sent nearly \$3,000 to the FA Research Fund. I feel that I am now able to do something to help Sofia and further FARE's important work.

I have met so many incredible people during this short journey: relatives of FA patients, people at the FA Research Fund, yarn shop owners, and other individuals. People who listen to our story and want to help. As long as I can knit, I will be committed to helping fund research for Fanconi anemia—one stitch at a time.

Send Your Old Jewelry and Yarn!

I'm always on the lookout for great yarn and costume jewelry. Tag sales, garage sales, estate sales, church bazaars—who knows where I'll find the next bag of great "jewels" or stash of yarn! We are always looking for donations of yarn and old jewelry.

Knitters are Always Welcome!

If you knit and would like to take on a project or two, email me at smith.babby@gmail.com for more information. ♦



Teddy and Sofia in hats knitted by Fibers and Friends for Sofia

Fundraising Assistance

Did you know that 85 percent of the donations to the FA Research Fund are raised by FA families? Obviously, we need the efforts of everyone who reads this newsletter!

FA Family Fundraising Teams now exist on a regional level to assist our families with fundraising. If you are unsure how to contact your team leader, contact the FA Research Fund.

The staff of the Fund is ready to assist you with your fundraising efforts. We'll help you write or edit your fundraising letter; photocopy it; provide the postage; and mail it from our office, using your mailing list. If you're going to hold a fundraising event, we'll provide similar help.

The FA Research Fund asks FA parents to make certain that any event they hold is covered by liability insurance. Insurance for a one-time event is often available through a family's homeowners 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 check to the "Fanconi Anemia Research Fund." When a donation is received, we will generate 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.

Our sincere thanks to all of you for your efforts to raise funds to combat this devastating disease.

Family Fundraising Efforts

FA families raised \$1,693,921 for the Fanconi Anemia Research Fund in 2009 by once again asking friends, families and acquaintances to donate directly, pledge to a future giving amount, or participate in events.

Although down from the previous year, we are nonetheless very thankful for every dollar raised. Our researchers made great headway last year and we're confident that 2010 will yield more discoveries that will help patients.

One hundred and fifty-five FA families raised funds this year; of those, 82 families raised \$500 or more and four raised more than \$50,000. Thanks so much for your support. We couldn't do it without you!

\$500,000 and Up

Dave and Lynn Frohnmayer

\$100,000 to \$499,999

Ken and Jeanne Atkinson

Chris and Susan Collins

Glen Shearer and Peggy Padden

\$50,000 to \$99,999

Fanconi Hope Charitable Trust

\$25,000 to \$49,999

Kerrie Brannock

Mike and Tracy Brannock

Peter and Tara Himmelreich

Dan and Nikki McCarthy

Kevin and Lorraine McQueen

\$20,000 to \$24,999

Brian Horrigan and Amy Levine

Todd and Kristin Levine

\$15,000 to \$19,999

John and Kim Connelly

Andrew and Jennifer Gough

Steve and Jennifer Klimkiewicz

\$10,000 to \$14,999

Ed and Janice Duffy

Charles and Katy Hull

Deane Marchbein and Stuart Cohen

My Best Friend/Kevin and Katie

Rogers

\$5,000 to \$9,999

Yavin Atzmon and Sharon Harari

Chris and Jennifer Branov

Joseph and Nancy Chou

David and Mary Ann Fiaschetti

Alan and Rachel Grossman

Adam and Olivia Mindle

Tyler Morrison and Rachel Altmann

Bob and Andrea Sacks

\$1,000 to \$4,999

Andrew and Vicki Athens

Mark and Linda Baumiller

Randy and Nancy Bloxom

Donald and Danielle Burkin

David and Kim Chew

Lauri Cohen

Donna DellaRatta

Pat and Mary DiMarino

Doreen Flynn

Nanette Vannostran Foster

Owen Hall and Margaret Kasting

Beth and Jeff Janock

Christie and Randy Kelley

John and Karilyn Kelson

Shaid and Melvina Khan

Erik Kjos-Hanssen and Turid Frislid

Joseph Konikowski

Sejin Kwon and Jee-ai Kim

Lynette Lowrimore

Jeremy and Stacey Mefford

Jim and Holly Miranda

Jack and Lisa Nash

Robert and Mary Nori

Ron and Fredi Norris

Fred and Nancy Nunes

Kevin and Lorraine O'Connor

Derek and Ginger Persson

Peter and Janice Pless

John and Dianne Ploetz

Pedro and Marina Ravelo

Paul and Rena Rice

Lynn and Rick Sablosky

Ron and Elesa Schaefer

Bryan and Karen Siebenthal

Mark and Susan Trager

William and Mary Underriner

Mike and Beth Vangel

Marc and Sandi Weiner

Kim and Michael Williams

Up to \$999

Peter and Donna Abramov

Gerald and Julie Barbier

John and Audrey Barrow

Tyren and Kelly Bennett

John and Francene Berglund

James and Tracy Biby

Jeffrey and Donna Boggs

Matt and Lisa Buglehall

Patti Carter

David and Paula Ceresa

Jeanette Clark

Tyler and Teresa Clifton

Daniel Conde

Brad Curry

Brian and Margaret Curtis

Dottie Day

Richard Day

Mark De Groot and Hanneke

Takkenberg

Charles and Dahne Deeks

Wendy Delzell

Mike Dennis and Ginger Eggers

James and Carol Dillon

Antonino and Marie DiMercurio

Brian and Jennifer Dorman

Sandra and Lindsay Dunn

David and Kelly Dunnock

Gene and Lynn Eddy

Sharon Ellis

Billy Jo and Debbie Estep

Ezat and Laila Faizyar

Curt and Crystal Fales

Carol Felmy and Michael Glas

Justin and Britteny Ferrin

Susan Gannon

Gary and Melody Ganz

Lisa and Brian Gillott

Pat and Maria Gleason

Maria and Josh Godwin

Allan and Lori Goldberg

Beatriz Goris

Mitchell and Tirzah Haik
 Roger and Eleanor Herman
 Jeff and Judy Hoffman
 Alan and Charisse Howard-Jones
 Shane and Colleen Irvin
 Lila Keleher
 Kristina King
 Kayla Lackey
 Eugene and Renee Lemmon
 Peg LeRoux
 Tanner and Jessica Lindsay
 Eric and Beth Losekamp
 Bill and Jackie Lucarell
 Steve and Alison McClay
 Kevin and Barbara McKee
 Catherine McKeon
 Charles and Cecelia Meloling
 Sydney and Betsy Moore
 Griff and Cecilia Morgan
 John and Betty Mozisek
 Tony and Lina Nahas
 Jack and Tammy Neal
 Bob and Alice Nicholson
 Lori Peterson
 Michael and Kay Proctor
 Lynn and Shirley Quilici
 Mario and Yolanda Ramirez
 Lester and Phylis Resh
 Gail Richardson
 Jan and Leonard Riley
 Mark Ritchie and Lisa Mingo
 Les and Nancy Ross
 Steph and Chantel Rossouw
 Erik and Lori Salo
 Richard and Dolores Satterlee
 Sharon Saunders
 Bill and Connie Schenone
 Severt and Beatrice Score
 Thomas and Brenda Seiford
 Matt and Diane Senatore
 Lorne Shelson and Annette Waxberg
 Jim and Carol Siniawski
 Tamara Stephens
 LeaAnn Stiller
 Greg and Brandi Stuart
 Daniel Terryah
 Steve and Melissa Turner
 Tom and Kathy Uno
 Joe and Wendy Vitiritto
 Sean and Kristin Young
 Marge Zaborney ♦

Mark Your Calendar

Annual FA Family Meeting

**Camp Sunshine
Sebago Lake, Casco, Maine**

Friday, June 25–Tuesday, June 29, 2010

All FA families and adult FA patients are welcome! Camp Sunshine applications are available at <http://campsunshine.org/programs> or by telephone at 207-655-3800. Acceptance to the meeting is on a first-come, first-served basis of completed, accepted applications. Limited scholarships are available through the Fund (application deadline is April 1). Contact Teresa Kennedy at teresa@fanconi.org or at 1-888-FANCONI.

Adult FA Meeting

Portland, Oregon

Friday, February 11–Sunday, February 13, 2011

All adult FA patients are welcome! More details will be available soon. Please contact Teresa Kennedy at teresa@fanconi.org or 1-888-FANCONI if you are interested in attending.

Long-time Donors Pass the Million Dollar Mark

continued from page 1

worldwide benefit from the scientific advances made possible by their personal donations and fundraising efforts on behalf of the Fund.”

In addition to annual personal donations to the Fund, the Kilkennys’ Lucky Duck Foundation hosts an annual benefit event to raise funds for Fanconi anemia research. Last year’s event, the San Diego Swing & Soiree, included a golf tournament and boutique shopping. (Mark your calendar for the 2nd Annual San Diego Swing & Soiree on Sept. 27, 2010 at Rancho Santa Fe Golf Club. For more information, go to www.luckyduckfoundation.org.)

“Pat and I are moved to support the Fanconi Anemia Research Fund by our connection to two families who are touched by Fanconi anemia,” said Stephanie. “Both the Frohnmayer and the Shearer/Padden families have lost children to FA and each continues to battle for the life of another child,” Stephanie continued. “It’s our sincere wish that our contributions—combined with the contributions of others—will make a significant difference in the fight for a cure.”

We guarantee that a difference has already been made. Our profound gratitude to Pat and Stephanie. ♦

Generous Bequest Surprises and Inspires Collins Family

The Fanconi Anemia Research Fund received an early—and unexpected—holiday gift in December 2009: a \$360,000 bequest from the estate of Marguerite Sherwin in honor of the Collins family. The Collins have two children with Fanconi anemia, Christian, 16, and Calen, 11. (You might know Christian as the national spokesperson for Caddy for a Cure, a PGA tour fundraising initiative spearheaded by Russ Holden.)

Mrs. Sherwin had made several donations to the Fund ranging from \$100 to \$2,000 since 1998, but the Collins family was caught by surprise by the generosity of Mrs. Sherwin in her will.

Susan Collins explains, “my husband Chris was Mrs. Sherwin’s advisor when we lived in Florida over eight years ago. While Mrs. Sherwin had continued giving faithfully to the Fund we had no idea that she had also included FARF as one of her beneficiaries. I think this amazing gift is a testament not only to Mrs. Sherwin’s enormous generosity, but also to the deep and lasting impact that FA children have on the lives of the people they meet—often in ways that we may not even see or know about. The challenge comes to us, the parents, to also try to have as much of a positive impact on those around us as our children do.”

The gift boosted the Fund’s 2009 family fundraising total to \$1,693,921—an impressive amount in any year but especially in one in which the recession lingered and unemployment grew. ♦



Charles Chiappone (right) and a fellow rider take a break

Riding for a Cause

By Charles Chiappone

We all have “Get Healthy” goals for ourselves, but moving beyond the words into action isn’t easy. Stating the goal wasn’t enough motivation to actually get going. I’d tried that for years! Then I met a vibrant 10-year-old, Sean McQueen, who was playing with my children in our neighborhood. Shortly thereafter I learned that he has Fanconi anemia. I thought of how blessed we all are to experience parenthood. With all of the good, bad, and ugly, we are here for our children!

I had found my “Get Healthy” motivation.

At that moment, I decided to “Ride for FA.” Friends, family members, and business associates happily sponsored my bike ride with a lump sum donation or a per-mile amount. They generously let me accumulate miles throughout the summer. By

the end of August, I had completed a 100-mile ride that wasn’t easy for me. However, the motivation I needed was swirling around in my head: Ride for the kids. Ride in thanks for my children. Ride for the cause that is so important to many others.

With the successful completion of the 100-miler, I clocked 859 miles and raised \$3,900 for FA. A BIG thank you goes out to Sean and his family. Their inspiration and dedication to this cause gave me the drive to push beyond my “out of shape” limits. As a result, I am in much better health and know that the money raised goes to improving the health of children with FA. Together, our small contributions can lead to something huge!

Ride on,
Charlie



Your FA Research Dollars at Work in 2009

In 2009, the Fanconi Anemia Research Fund awarded \$1,234,007 in research grants to the following projects:

Investigator: Grover Bagby, Jr., MD, Oregon Health & Science University, Portland, Ore.

Title: *Preclinical Evaluation of Small Molecules as Potential Therapeutic Agents in Fanconi Anemia*

Amount: \$49,000

Investigator: Phillip Connell, MD, University of Chicago, Chicago, Ill.

Title: *Restoration of Homologous Recombination in Fanconi Anemia*

Amount: \$208,138

Investigator: Neelam Giri, MD, MBBS, National Cancer Institute, Rockville, Md.

Title: *Studies of Immune Function in Patients with Fanconi Anemia*

Amount: \$157,388

Investigator: Markus Grompe, MD, Oregon Health & Science University, Portland, Ore.

Title: *iPS Cells from Fanconi Anemia Fibroblasts*

Amount: \$53,082

Investigator: Nick Lakin, PhD and Annette Medhurst, PhD, St. Peter's College, Oxford University, Oxford, UK

Title: *Defining the Molecular Function of FA Proteins during S-phase (Year 2)*

Amount: \$100,149

Investigator: Majlinda Lako, PhD, Lyle Armstrong, PhD, and Christopher Mathew, PhD, Institute of Human Genetics, Newcastle University, Newcastle upon Tyne, UK

Title: *Using iPSC Technology to Understand Early Haematopoietic Development in FA Patients*

Amount: \$47,000

Investigator: Susan Mallery, DDS, PhD, Ohio State University, Columbus, Ohio

Title: *Mucoadhesive Patch Delivery of Fenretinide and Berry Anthocyanins for Oral Cancer Chemoprevention*

Amount: \$100,000

Investigator: Susan Olson, PhD, Oregon Health & Science University, Portland, Ore.

Title: *Pathophysiology and Treatment of Fanconi Anemia*

Amount: \$55,000

Investigator: Jakub Tolar, MD, PhD, University of Minnesota, Minneapolis, Minn.

Title: *Correction of Human Fanconi Anemia Induced Pluripotent Cells by Homologous Recombination*

Amount: \$125,000

Investigator: Susanne Wells, PhD, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio

Title: *HPV Replication and Transformation in FA Squamous Cell Carcinomas*

Amount: \$200,000

Investigator: Stephen C. West, PhD and Andrew Deans, PhD, Cancer Research UK, South Mimms, UK

Title: *Coordination of the Fanconi Anemia and Bloom's Syndrome Complexes by FANCM*

Amount: \$139,250

Upcoming Fundraisers for FA Research

2010

May 1: 1st Annual International FA Day. For information, please visit fanconianemiaday.blogspot.com or email fanconianemiaday@gmail.com.

May 1: My Best Friend's Fiesta for FA, Garden City, N.Y. Vibrant Cinco de Mayo themed party. Contact Katie Rogers at KRogersK@aol.com.

May 1: 2010 Chess for FA Benefit Tournament, Corbett, OR. Chess Tournament at Corbett School Multi-Purpose Building. Pre-registration required; suggested donation \$10. For more information, email ChessForFA@gmail.com or visit chessforfa.wordpress.com.

May 2: Fundraiser for Fanconi Canada, Vaughn, Ontario. Dinner and silent auction for Fanconi Canada at the Terrace Banquet Centre. Three course dinner with outstanding entertainment. Special guest speaker Amy Frohnmayr. More info at www.curefanconi.org.

June 25: 6th Annual Coley's Cause Golf Tournament, Lakeville, Mass. Poquoy Brook Country Club. For more information, visit www.coleyscause.com.

July 10: 7th Annual Fanconi Anemia Golf Scramble, Brush Prairie, Wash. Cedars on Salmon Creek Golf Course. Golf scramble, silent auction, BBQ, and raffle. For more information, contact Peg Padden at pegpadpad@hotmail.com.

Ongoing

Maddie's Angels Cookbooks: These cookbooks are for sale to support FA research for \$15 each. For more info, please contact Nancy and Ernie Landwehr at maddiesmom50@yahoo.com or ernie_landwehr@msn.com.

Kaps for Kendall: Donate \$25 to FARF through Kaps for Kendall and a knitted hat will be donated to a patient who has lost his or her hair from chemo and radiation treatments. If you are a knitter, you can help by supplying a hat. Contact Allison and Whitney Atkinson at www.kapsforkendall.com.

Kendall and Taylor Atkinson Foundation (KATA): The Kendall and Taylor Atkinson Foundation raises money for Fanconi anemia research. Visit www.katafoundation.org for more information, to donate, or to purchase from their online store.

Caddy For A Cure: Caddy For A Cure, Inc, raises funds for designated charitable organizations while offering the opportunity to be "inside the ropes" as a caddy for a Tour player at a PGA Tour event. This perfect gift for a golf fanatic offers a one-of-a-kind professional sports fantasy while contributing to genetic disease research. 25% of the proceeds from Caddy For A Cure are donated to the Fanconi Anemia Research Fund. Contact Russ Holden at www.CaddyForACure.com.

Scholarship Fund for Adult Meeting Launched

Lynn Frohnmayer had a special relationship with Alex Norris, a young man with FA who died in 1994 at age 16 from complications of Fanconi anemia. "Alex was a wonderful kid," Lynn said. "He was smart, generous, and very kind."

In honor of Alex, Lynn and Dave Frohnmayer have established a scholarship fund for the Adult FA Meeting, a new meeting created by the FA Research Fund specifically to address the medical and psychosocial needs of adults living with FA. The inaugural meeting was held in July 2009 in Denver, Colo., with 15 patients and nine family members and friends participating; the next meeting is tentatively planned for February 2011 in Portland, Ore.

Lynn said that if Alex and her daughter, Kirsten, were alive today, they would be actively involved in the adult FA group. "They were both go-getters and knew instinctively how to draw people together and create something positive." Kirsten Frohnmayer died of FA in 1997.

If you would like to donate to the scholarship fund for the Adult FA Meeting, please note the designation on your donation to the Fanconi Anemia Research Fund. Or, if you would like to fundraise specifically for this cause, please contact our office so that we may assist your efforts. ♦



Alex Norris, age 16



Participants at the 6th Annual Golf Scramble

6th Annual Golf Scramble Brings out Generous Friends and Family

by Peg Padden

The 6th Annual Fanconi Anemia Golf Scramble, Silent Auction, BBQ and Raffle was held at Cedars on Salmon Creek Golf Course in Brush Prairie, Wash., on July 18, 2009. More than 100 loyal and caring family and friends who—for the 6th year in a row—showed up to golf, bid on silent auction items, make donations, buy raffle tickets

for sporting items AND buy more raffle tickets for a trip to Greece! Did I mention they even paid for Mulligans? Because of these generous, good people, we raised \$55,000 for important FA research.

We are so fortunate and grateful to have these people in our lives. The many volunteers and participants have given their time and

money again and again. In memory of our wonderful son Jake, in honor of our equally wonderful son Spencer, and for everyone else affected with FA, we can't thank them enough.

We look forward to the 7th annual event scheduled for July 10, 2010. ♦

The Twenty-second Annual Fanconi Anemia Scientific Symposium

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Marriott City Center Hotel
Minneapolis, Minnesota



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Blake & Sydney

continued from page 13

words, “Rena, she has it too.” I was being told that my daughter, Sydney, also had Fanconi anemia. My heart sank, and gut wrenching pain set in. I could hardly take in that our son needed a bone marrow transplant—and now our daughter had the same disease.

The months following diagnosis were a blur of activity and emotions as we prepared for transplant. Coming off what we felt was a string of bad luck, we couldn’t help but feel extremely fortunate when we learned that Blake’s dad was a match and could be the donor. On Jan. 26, 2009, Blake received the precious life saving bone marrow, and 8 days later he started to engraft. We definitely had some rough days in the hospital, but as transplants go, we

were fortunate. Blake was out of the hospital on day 21 and we were able to come home May 6, 2009.

Today, both kids are doing very well. Blake is a year post transplant and recently returned to school and Sydney is a typical happy, playful, four year old. Currently, doctor’s orders are for both kids to receive quarterly blood tests and an annual bone marrow biopsy/aspiration. Last month’s biopsy results for Sydney showed that her bone marrow cellularity had dropped, as had her platelets. Despite this worrisome trend, her counts are still good, and she’s otherwise doing very well. The doctor said Sydney could go for years at this level. Although the challenges ahead of our family are enormous, we’re enjoying our lives together and remain optimistic and hopeful for the futures of our children. ♦