



FA FAMILY NEWSLETTER

#43

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Spring 2008



Hans Joenje and Dave Frohnmayer

Hans Joenje Receives Distinguished Service Award

On behalf of the Board of Directors of the Fanconi Anemia Research Fund, Dave Frohnmayer presented Hans Joenje, PhD, Vrije Universiteit, Amsterdam, with the Fund's *Distinguished Service Award* at the Symposium. Dr. Joenje is the only person to receive both the *Award of Merit* and the *Distinguished Service Award*, an accomplishment magnified further by his receipt of numerous *Discovery Awards*.

In making this award, board members noted Joenje's tireless and expert contributions to the Fund through his long service on the Fund's Scientific Advisory Board. Additionally, they recognized his seminal contributions to FA science

through gene discovery and pathway elucidation, contributions which are, quite simply, legendary. Joenje's eleven FA gene discoveries are the present foundation of FA science, upon which other advances have been or will be built.

Of equal importance, Dr. Joenje has consistently combined great compassion for FA families with his brilliance in research, by attending the FA Family Meeting to explain the scientific basis of FA to families and by working hard to help them determine their particular FA gene. FA families know Dr. Joenje as a caring individual who sees beyond the test tube to their need for answers and hope. ♦

The Fund Wins \$50,000 in America's Giving Challenge!

Peggy Padden, FA parent from Portland, OR, whose son Jake died of FA in 2003 and whose son Spencer also has FA, seized on the opportunity for the Fund to compete for \$50,000 through the *America's Giving Challenge* sponsored by *Parade Magazine* and the Case Foundation. The *Challenge* pledged to award \$50,000 to each of four charities that secured the most individual donations during the contest period.

The focus of the *America's Giving Challenge* was to encourage charitable use of social networking and the internet to empower non-profits

continued on page 7

HIGHLIGHTS

Reducing Radiation Using Matched or Partially Matched Unrelated Donors.....	3
Results of Alternate Donor High-Risk Transplants at Memorial Sloan-Kettering Hospital	3
FA Carriers and Cancer Risk	8
Family News.....	12
Fundraising.....	18
Research Funded in 2007.....	25

FA Scientific Symposium

The 19th Annual Fanconi Anemia Scientific Symposium was held in Chicago, IL, from October 8–11, 2007. One hundred eighty-nine scientists and clinicians from eighteen countries attended this exceptionally outstanding meeting. Keynote presentations by Lee Niswander, PhD, University of Colorado Health Sciences Center, on developmental defects; by Laura Niedernhofer, MD, PhD, University of Pittsburgh Cancer Institute, on endocrine dysfunction; and by Gary Clayman, DMD, MD, FACS, University of Texas MD Anderson Cancer Center, on cancer were cited by many participants as among the highlights of the conference. The reprise of *FA 101* by Ray Monnat, Jr., MD, and Akiko Shimamura, MD, PhD, both from the University of Washington, again earned rave reviews.

Participants commented that the quality of science presented at this meeting, by the oral and poster presenters alike, was simply first-rate. The value of those presentations was magnified by the high level of questions and participation from the audience and the leadership of the session chairs. Attendees completed evaluations at the end of the conference, and the comments were overwhelming helpful and positive.

At the Symposium Dinner, Dave Frohnmayer, co-founder of the Fund, presented the following researchers, all from Cincinnati Children's Hospital Medical Center, with awards for their posters: Best Basic Science Poster: Xiaoling Zhang, *Defective Adhesion, Migration and Homing Associated with Altered Rho GTPase Activity in Cells from FA Patients*; Best Clinical (Trial) Poster: Susan Rose, MD, *Thyroid Hormone in Children with Fanconi Anemia*; and Best Translational Poster: Abdullah Ali, *An Unusual Presentation of FANCL: Making the Case for Complementation Analysis in All FA Patients*. ♦



Abdullah Ali receives Best Translational Poster Award from Dave Frohnmayer.

Haploidentical Donor Transplants for FA Patients: A Collaborative Effort

by Monica S. Thakar, MD, Fred Hutchinson Cancer Research Center, Seattle

Patients who need hematopoietic cell transplantation (HCT) but cannot find HLA-matched sibling or unrelated donors are in a dire situation. In order to provide HCT to all patients, the Seattle-Curitiba, Brazil collaborative group has recently opened a clinical trial investigating the use of HLA-haploidentical family donors. Such family members, whose HLA tissue type is matched by at least 50% to the patient, allow either parent to be a donor, and assures that virtually every FA patient has a suitable donor.

Incorporating their extensive experience using cyclophosphamide safely and effectively in the HLA-matched sibling setting, this group is extending the use of cyclophosphamide both before, and now also after, HCT in order to overcome the strong immunological barriers that lead to graft rejection and graft-versus-host disease. They are actively accruing patients and expect between 7–35 enrolled between both sites. ♦



Monica S. Thakar, MD

Reducing Radiation Using Matched or Partially Matched Unrelated Donors: Update from Minnesota

Over the past decade, there has been a major change in the chance of survival after unrelated donor transplantation. Two goals of transplanters at the University of Minnesota are to reduce the risk of infection, and to find ways to improve quality of life in the rapidly growing number of long-term survivors. Margaret MacMillan, MD, states that reducing use of irradiation is one strategy to achieve those goals.

Prior to 2006, the standard of care at the University of Minnesota was total body irradiation (TBI) 450 cGy (centigray or rads), with thymic shielding in combination with cyclophosphamide, fludarabine and ATG. In July 2006, the transplanters initiated a TBI dose de-escalation trial.

The first cohort of 11 patients received TBI 300 cGy. Ten of these patients engrafted long-term; one lost the graft after six months and died

of infection. Having achieved the goal of maintaining an engraftment rate of greater than 90%, they next treated two patients with TBI 150 cGy. While both patients engrafted initially, both lost their grafts within the first 100 days. They received a second dose of TBI 150 cGy, and both demonstrated marrow recovery.

All subsequent patients have received a single dose of TBI 300 cGy. As of February 2008, 15 patients have been treated with this protocol. The incidence of primary engraftment is 100%, with one patient (above) developing late graft failure six months post-transplant. Fourteen of 15 patients survive. Two patients have had grade II acute graft-versus-host disease (GVHD). Both responded to steroid treatment, and no patient has had chronic GVHD.

MacMillan states that, thus far,



Margaret MacMillan, MD

high rates of engraftment and survival support the continued use of TBI 300 cGy. It is hoped that the lower dose of irradiation will improve quality of life long-term, but longer follow-up is needed to see the impact of this approach on hormone levels, fertility and risk of cancer. ♦

Results of Alternate Donor High-Risk Transplants at Memorial Sloan-Kettering Hospital, New York City



Farid Boulad, MD

Farid Boulad, MD, Memorial Sloan-Kettering Hospital, New York City, has now transplanted 22 FA patients who had alternate (non-sibling) stem cell donors. Patients ranged in age from 5.5 to 35 years, including 6 patients older than 18 years. Ten patients had aplastic anemia, five had developed myelodysplastic syndrome, and 7 had acute myeloid leukemia. All patients had been transfused, 16 had taken androgens, and 15 had a history of infection.

The protocol included total body irradiation (450 rads), fludarabine and cyclophosphamide. Immunosuppression consisted of antithymocyte globulin and tacrolimus. Nineteen patients received T-cell depleted peripheral blood stem cells and 3 received T-cell depleted bone marrow.

All 22 patients engrafted and maintained their grafts. Two patients had acute graft-versus-host disease (GVHD) grade III–IV, and one patient had acute GVHD grade II.

With a median follow-up of 4.5 years, 15 of 22 patients are alive and disease-free. Immune reconstitution was achieved at approximately 6 months post-transplant for most patients. Causes of mortality were leukemic relapse (2 patients), infection (2 patients) and severe GVHD (2 patients). One patient died five years post transplant of cervical squamous cell carcinoma.

These results are excellent, given the risk factors that characterized the vast majority of this group of patients. ♦

Related Donor Transplants without Radiation or Fludarabine: Results from Brazil and Seattle

Carmem Bonfim, MD, Federal University of Paraná, Curitiba, Brazil, reported on the results of 43 FA bone marrow transplants using HLA-matched relatives, performed between July 1999 and October 2006. This study was a collaborative effort between Curitiba and the Fred Hutchinson Cancer Research Center in Seattle, Washington.

Most of these patients had matched sibling donors (37); six patients had non-sibling HLA-identical relative donors. Patients received cyclophosphamide 60 mg/kg, no irradiation and no fludarabine. Drugs were administered to prevent or reduce graft-versus-host disease (GVHD).

With a median follow-up of 3.7 years, 40 of the 43 patients are alive. One was excluded from analysis due to early death. Forty-one of 42

engrafted, but 5 experienced graft failure. Four of these were late graft failures, occurring between 156 and 365 days post-transplant. Two of these late graft failure patients were rescued after a second transplant, one after a third. Most patients experiencing graft failures either received transplants from non-sibling related donors or had cytogenetic abnormalities.

Seven patients developed acute GVHD grade II–III; ten had extensive chronic GVHD and two experienced limited chronic GVHD. Two additional patients developed severe GVHD: one after the third transplant and the other after a second transplant.

Three patients died, two from graft rejection and one from regimen-related toxicity. One patient developed squamous cell carcinoma



Carmem Bonfim, MD

of the tongue five years post-transplant, at the age of 12. This patient had also experienced extensive chronic GVHD.

Researchers concluded that this regimen was basically well tolerated and that overall survival was excellent. ♦

A Pilot Study Using Etanercept in FA Patients with Early Bone Marrow Failure

Stella Davies, MD, Cincinnati Children's Hospital Medical Center, Cincinnati, Ohio, is conducting a research study to evaluate the safety and efficacy of etanercept (Enbrel) in treating early bone marrow failure in FA patients. Studies show that FA patients are very sensitive to tumor necrosis factor alpha (TNF- α), a protein that causes bone marrow cells to die. FA patients have unusually high levels of this protein in their bodies. Etanercept blocks the action of TNF- α . Researchers hope that this drug will delay or prevent the progressive bone marrow failure associated with FA.

To be considered for this study, patients must be at least four years of age and have evidence of early bone marrow failure, as defined by two blood counts drawn one month

apart which show at least one of the following: platelet count less than 100,000, hemoglobin less than 9, and an absolute neutrophil count of less than 1000. A negative tuberculosis skin test is also required. The patient or caregiver must be able to administer injections given just under the skin.

The first three patients have tolerated this drug well, without major complications or severe side effects reported. It is too early to present any conclusions about the effectiveness of this drug in preserving stem cells in FA patients with early marrow failure.

For further information, contact:
Robin Mueller, RN
Nurse Coordinator
1-800-344-2462, ext. 3218
robin.mueller@cchmc.org ♦

Transplantation Results from Minnesota, Using Matched Sibling Donors.

In 2000, Margaret MacMillan, MD, and John Wagner, MD, University of Minnesota Fanconi Anemia Comprehensive Care Program, initiated a trial to determine whether FA patients with HLA-matched sibling donors could be successfully transplanted without the use of irradiation. Patients receive cyclophosphamide, fludarabine and ATG. To date, 16 FA patients under 18 years of age with aplastic anemia have been treated using this approach. All patients engrafted. No one has developed acute or chronic GVHD. The probability of survival 2 years after transplant is 100%. ♦

Targeting Adenoviruses to Pre-cancerous Cells in the Oral Cavity

Hester van Zeeburg, Graduate Student, Department of Otolaryngology/Head and Neck Surgery, Vrije Universiteit Medical Center, Amsterdam, stated that FA patients frequently develop oral squamous cell carcinomas (OSCC). These cancers are preceded by pre-cancerous fields of genetically altered cells that clinically are often not visible. These fields can be diagnosed using genetic markers, but the treatment options are very limited. Efforts to eliminate these pre-cancerous fields (such as the ONYX015 mouthwash therapy) gave some benefit to patients but the long-term effectiveness was limited.

Van Zeeburg described the efforts of her laboratory to develop a more effective adenovirus that will target and eliminate pre-cancerous tissues of the oral cavity but will not affect normal tissues. She tested nine different cancer-specific adenoviruses on 5 head and neck cancer cell lines, including 3 cell lines from sporadic (non-FA) tumors and 2 cell lines from FA tumors, and compared them to two non-malignant cell lines. She noted that the sporadic and FA tumors were sensitive to the same adenoviral vectors. Her laboratory identified the most effective adenovirus (Survivin-driven E1A), that would bind to and replicate in precancerous tissues, but would selectively spare normal cells.

Much work needs to be done before this research will be in clinical trials. ♦



Hester van Zeeburg

Screening for Pre-Cancerous Lesions: A Non-Invasive Method

Ruud Brakenhoff, PhD, Vrije Universiteit Medical Center, Amsterdam reminded attendees at the Scientific Symposium that FA patients are at high risk for developing squamous cell carcinomas (SCC). Specific risk factors for these tumors are bone marrow transplantation (BMT) and graft-versus-host disease. SCCs of the oral cavity are preceded by large fields of pre-cancerous tissue. Eighty percent of these are invisible to the naked eye. They can be detected by biopsy, which is painful, and by careful examination of tissues for genetic alterations.

There is at present no treatment for invisible precursor lesions, except for increasing surveillance and removing all abnormal looking mucosa, especially when genetic changes have been detected in a certain area. Brakenhoff has developed a non-invasive method for screening for

these lesions. Using a small brush, he removes superficial cells from seven regions of the mouth, and from any visible lesion. He then screens the cells for four specific chromosomal alterations associated with cancer: those involving 3p, 9p, 17p and 11q.

Brakenhoff analyzed 20 normal controls; none had genetically altered cells. In 25 non-FA patients with leukoplakia (white patches in the mouth), 11 had chromosomal alterations. Biopsy of leukoplakia tissue revealed the same abnormalities as brush samples from the same patient, indicating the value of this approach.

Brakenhoff has now analyzed brush samples from 70 FA patients who had not undergone BMT. He identified chromosomal alterations in 20 of these patients.

Brakenhoff's screening method was not informative for FA patients who had undergone BMT. Samples



Ruud Brakenhoff, PhD

revealed complex genetic patterns, caused by the presence of donor DNA in the brushed samples. The percentage of donor DNA varied from 0% to >90%. Researchers are now investigating whether the mucosal epithelium in post-BMT patients consists of a mixture of

continued on page 23

Delaying Onset of Epithelial Tumors in FA Mice

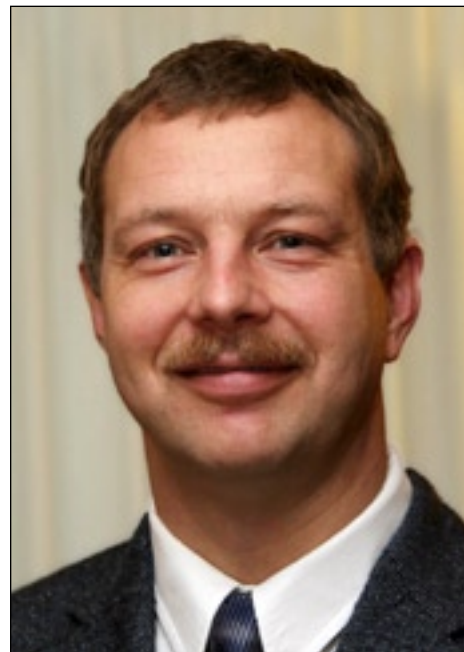
Bone marrow failure and malignancies greatly shorten the life span of FA patients. Bone marrow transplantation is currently the only curative therapy for the bone marrow problems that affect FA patients, but all FA patients are still at risk for cancer. Qingshuo Zhang, PhD and his colleagues from the Markus Grompe lab at Oregon Health & Science University are using a mouse model of FA to test various therapeutic interventions that may reduce malignancies or delay the onset of these tumors.

Using a strain of FA mice that usually develop epithelial tumors at about one year of age, Zhang tested whether tempol, an oxygen radical scavenger, might delay tumor formation. Half of the FA mice were

treated with tempol; half were given a placebo. Tempol treatment significantly delayed the onset of epithelial tumors and increased the mean epithelial tumor-free survival time by 38% (from 312 days to 432 days).

Researchers further discovered that tempol did not have any negative effect on the repopulation capability of FA hematopoietic (blood) stem cells. In addition, analysis of urine and DNA from multiple tissues revealed that tempol treatment reduced oxidative DNA damage in both FA and wild-type (normal) mice. This finding suggests that tempol may have an antioxidant function.

The Grompe lab concluded that tempol might be a good candidate for an FA clinical trial aimed at the



Markus Grompe, MD

prevention of solid tumors in FA patients. ♦

New Insights into Characteristics of FA Cancer Stem Cells

Ian Mackenzie, DDS, PhD, FDSRCS, Institute of Cell and Molecular Science, Queen Mary University of London, believes that a small population of cancer stem cells (CSCs) is responsible for the generation, growth and spread of malignant tumors. Current therapies can often eliminate the bulk of the tumor but are less effective in killing cancer stem cells, a phenomenon which may explain the recurrence of many cancers. Studying CSCs *in vivo* (in the body) would be a daunting task. Mackenzie stated that oral cancer cell lines, generated from patient tumors, contain stem cells which show unusual properties. These stem cells can therefore be used to identify special CSC characteristics that may allow selective therapeutic targeting. Mackenzie compared stem cells in normal cell populations with those in cell lines derived from either sporadic (non-FA) or FA-related head and neck squamous cell carcinomas.

Normal cells are much more apoptotic (subject to cell death) than cancer cells. However, both normal and malignant stem cells were 10 times more resistant to death than non-stem cells. Treating cell lines with DNA damaging agents (to mimic what happens during therapy) killed non stem cells but not stem cells. Instead of dying, stem cells remained for a long time in the “G2” phase of the cell cycle, the phase during which cells try to repair DNA damage.

Mackenzie studied two cancer cell lines from FA-A patients. The CSCs from these patients showed high resistance to cell death and exhibited a major block in the G2 phase of the cell cycle, much like sporadic (non-FA) cancer stem cells. However, the one cell line studied from an FA-C patient showed significantly higher rates of cell death and less blockage in G2, and was therefore more similar to the



Ian Mackenzie, DDS, PhD

non-malignant cell population.

It is not yet clear how FA genes influence stem cell survival, but such differences could suggest new therapy strategies. ♦



Laura Hays, PhD

OHSU Compares Squamous Cell Carcinomas in FA and in Wild-Type Mice

Are there significant differences between FA head and neck squamous cell carcinomas (HNSCCs) and those same tumors that afflict the general population? In an effort to address that question, Laura Hays, PhD, Oregon Health & Science University Cancer Institute, induced HNSCC in FA and normal (wild-type) mice by exposing the mice to a carcinogen known to cause this cancer in laboratory animals.

After treatment with a carcinogen, both FA and wild-type mice were similar in tumor onset rates, in type of cancer induced (squamous cell carcinomas) and in location of primary tumors. Hays was unable to determine whether FA tumors of this type spread more rapidly than non-FA tumors, however, because mice had to be euthanized early after appearance of the primary tumor.

Analysis of tumors from FA mice did reveal that FA cells had significantly higher levels of chromosomal breakage than wild-type tumors. Future experiments will determine if FA tumor cells that are re-injected into mice spread more quickly than tumor cells from wild-type mice. ♦

Consensus Conference to Update FA: Standards for Clinical Care

On April 10 and 11, 2008, physicians expert in the care of Fanconi anemia patients will meet in Chicago, IL for a conference to arrive at consensus on the optimal standards for the medical care of FA patients. This conference will result in the publication of the third edition of the handbook, *Fanconi Anemia: Standards for Clinical Care*. The first edition (in 1999) and the second edition (in 2003) were prepared through similar consensus conferences in Portland, OR and Chicago, IL respectively. Eva Guinan, MD, Dana-Farber Cancer Institute, expertly chaired the past two conferences and will serve again as chair.

As a product of the April conference, chapters in the 2003 edition will be updated. New chapters entitled *Ear and Hearing Problems in FA Patients*; *Post-transplant Care and Late Effects*; and *Clinical Management Checklist* will be included in the 2008 edition. The updated edition will be available for all FA patients and their physicians worldwide by the end of the year, with editions printed in English and in Spanish. ♦

The Fund Wins \$50,000

continued from page 1

to raise funds. To that end, charities entered in the contest received a point for each unique online donation of at least \$10 (only one donation per individual was allowed). While many FA families were doubtful that a non-profit for a disease with as few patients as FA could win this contest, Peggy's enthusiasm and certainty that this was within the grasp of our group soon became contagious.

Families rallied as never before, asking every friend, family member, and acquaintance they could possibly recall to vote by donating for the Fanconi anemia entry. Each day brought more and more donations and greater and greater hope that we could succeed in this contest. Amazingly, on February 21, after all the votes were vetted, the Fund officially was one of the four winners, in a strong second place! In addition to the \$50,000 prize, the donations to the Fund made through our entry totaled over \$65,000! These funds

will be put to excellent use in advancing FA research.

We cannot thank Peggy enough for her leadership in this effort. She has been an inspirational leader in fundraising since the diagnosis of her two sons. In addition to other fundraising efforts for FA research, she annually organizes a Valentine Fanconi Anemia 5K Run/Walk in Portland and a golf tournament in Battle Ground, WA. A huge thank you as well to all the other FA families who joined with her as members of this unstoppable team! Great job to all! ♦

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.



Laura Haneline, MD

A Small Molecule Compound Might Enhance FA Bone Marrow Function

Several researchers have now demonstrated that, in the laboratory, exposure to oxygen or to tumor necrosis factor (TNF) leads to loss of hematopoietic (blood) stem cells from FA patients. The laboratory of Laura Haneline, MD, Indiana University School of Medicine, Indianapolis, Indiana, has further demonstrated that increased levels of oxygen and TNF activate certain proteins, including the p38 enzyme, which operate in an apoptotic (causing cell death) pathway. Inhibiting p38 enhances the function of FA mouse cells in culture.

Haneline cultured FA mouse cells with a p38 inhibitor (SB203580), then transplanted these cells into FA mice. Twelve months after transplant, these mice showed normal bone marrow function and produced all blood lineages. There was no sign of myelodysplastic syndrome or leukemia in the marrow of these mice. Haneline demonstrated that bone marrow cells from these mice were self-renewing following secondary transplants.

Haneline will next test this inhibitor on FA patient cells in culture, and plans to treat mice *in vivo* (in the body) with this compound. She believes that certain pathways are inappropriately activated in FA patients. A small molecule compound, such as a p38 inhibitor, might correct this deficiency. ♦

FA Carriers and Cancer Risk

Researchers and FA families have long wondered if FA carriers are at increased risk for cancer. The discovery that one of the FA genes (*FANCD1*) is also the breast cancer susceptibility gene *BRCA2* intensified this concern. In addition, two FA genes that interact with *FANCD1/BRCA2* are breast cancer susceptibility genes (*FANCF* and *FANCG*), although they pose less risk than *BRCA2*.

Arleen Auerbach, PhD, The Rockefeller University, and collaborators conducted the largest carrier study to date, to determine if FA genes apart from *FANCD1/BRCA2* and its interacting partners give carriers a higher likelihood of developing cancer. Her study appeared in *Cancer Research*, October 1, 2007.

Relatives of FA patients (784 grandparents and 160 other relatives) participated in this study. There was no increase in overall cancer incidence in this population of carriers

and noncarriers. However, Auerbach detected a significantly higher rate of breast cancer than expected among carrier grandmothers. Among carriers of the common FA genes (*FANCA*, *FANCC* and *FANCG*) only carriers of *FANCC* mutations were found to be at increased risk.

An analysis of all 154 carrier grandmothers revealed 18 breast cancers, significantly higher than the 10.7 expected. This increase was not observed among the 161 noncarrier grandmothers. There were 6 breast cancers among the 33 grandmothers who were carriers for a *FANCC* mutation, compared with 2.5 expected. When all 47 female carriers of FA-C (33 grandmothers and 14 other female relatives) were analyzed, the same trend was apparent. Auerbach found three *FANCC* mutations among the eight *FANCC* carriers who developed breast cancer: p.L554P (n = 1), IVS4 (n = 5) and c.322delG (n = 2).



Arleen Auerbach, PhD

Auerbach concluded that overall, there is no increased risk for cancer among FA carriers. However, in addition to the known risk of breast cancer in carriers of *FANCD1/BRCA2*, *FANCF* and *FANCG*, there is some evidence that *FANCC* mutations increase the risk of breast cancer. ♦

In Vivo Gene Delivery Using Unique Method

Alena Chekmasova, PhD, Thomas Jefferson University, Philadelphia, described a unique method to deliver genes into rabbit bone marrow by direct injection of recombinant SV40-derived vectors (rSV40) into the rabbits' femurs.

Chekmasova reviewed the theoretical advantages of *in vivo* (in the body) gene delivery in the context of FA. Most studies on gene therapy to correct FA bone marrow have involved *ex vivo* (outside of the body) modification. Stem cells are removed from the blood or bone marrow of the patient, corrected in the laboratory, and reintroduced into the same patient. These methods are problematic for FA patients, who have few hematopoietic stem cells (HSCs), and whose cells are easily damaged by laboratory manipulation. Past efforts to correct FA patients' bone marrow through gene therapy have failed due to insufficient numbers of stem cells.

Chekmasova stated that rSV40 vectors have several advantages over other vectors used for gene delivery purposes. They can transduce both resting and dividing cells very efficiently. They are not attacked by the immune system so can be administered repeatedly. And they integrate into cellular DNA, providing moderate but stable gene expression over a long period of time.

Chekmasova's results demonstrate that a single injection of rSV40 vectors carrying marker genes into the femoral bone marrow cavity of rabbits results in 30-35% transgene expression levels in different blood lineages. Expression was maintained until the study ended after 13 months. Thirty-five percent of bone marrow cells in the femur expressed these marker genes, as well as 31% in the tibia and 45% in the iliac crest. In addition, almost 74% of cells in the femur that are used as markers



Alena Chekmasova, PhD

for HSCs expressed the marker genes 13 months after injection into the femur.

Researchers hypothesize that this strategy of gene delivery could be an effective way to deliver normal genes to the bone marrow of FA patients. ♦

Curing the Bone Marrow of Mice with *In Vivo* Gene Therapy

Madeleine Carreau, PhD, Laval University, Québec, Canada, outlined some major obstacles presently faced in human gene therapy trials. FA patients have a greatly reduced number of hematopoietic (blood) stem cells. Removing and treating these cells *ex vivo* (outside of the body) can cause these fragile cells to undergo premature cell death. This process leaves even fewer corrected cells to populate the marrow. In addition, *ex vivo* manipulation can cause chromosomal abnormalities, adding risk to this procedure.

Carreau described the successful efforts of her laboratory to insert genetic material directly into the femur bone of mice. This direct injection process is called *in vivo* (in the body) gene therapy. Using lentiviral particles containing the normal

FANCC gene, Carreau's laboratory was able to correct mouse bone marrow. The corrected marrow could then produce all blood cell lineages. Additional tests established that these cells had self-renewal capability and were resistant to the chromosomal damage caused by the cross-linking agents used to test for FA.

The *in vivo* gene therapy approach was then tested on mice that had been pre-treated with a DNA-damaging agent to reduce their bone marrow cellularity to 30%, mimicking the aplasia characteristic of FA patients. Gene transfer corrected the remaining bone marrow cells and successfully reconstituted the marrow.

Carreau suggested that this strategy might be an alternative gene therapy approach for FA patients. ♦



Madeleine Carreau, PhD

Researchers Receive Discovery Awards

Researchers were honored during the Symposium Dinner in Chicago for their work in the discovery of *FANCI*, which was published in *Cellular Oncology*; *Nature Structural & Molecular Biology*; and *Cell*. Additionally, researchers received Discovery Awards for their work in the discovery of *FANCN*, which was published in *Nature Genetics*.

On behalf of the Board of Directors, Dave Frohnmayer presented Discovery Awards to the following researchers:

Honoring Initiative in FA Research: *FANCI* Discovery

Identification of the FA Complementation Group I Gene, FANCI, Cellular Oncology:

- Josephine Dorsman, PhD, Marieke Levitus, MSc, Davy Rockx, Martin Rooimans, Anneke Oostra, Anneke Haitjema, Sietske Bakker, MSc, Jürgen Steltenpool, PhD, Fré Arwert, PhD, Gerard Pals, PhD, Quinten Waisfisz, PhD, Johan de Winter, PhD, Hans Joenje, PhD, **Free University, Amsterdam**
- Dezsö Schuler, MD, **National Institute of Child Health, Budapest**
- Sheila Mohan, MBBS, DCH, PhD, **Apollo Specialty Hospital, Chennai, India**
- Detlev Schindler, MD, **University of Würzburg, Würzburg**
- Christopher Mathew, PhD, **King's College School of Medicine, London**

FANCI is a Second Monoubiquitinated Member of the FA Pathway, Nature Structural & Molecular Biology:

- Ashley Sims, Robert Sims III, Adriana Arita, Tony Huang, PhD, **New York University School of Medicine, New York**
- Elizabeth Spiteri, PhD, Francis Lach, Thomas Landers, Arleen Auerbach, PhD, **The Rockefeller University, New York**
- Melanie Wurm, Marcel Freund, Helmut Hanenberg, MD, **Children's Hospital, Heinrich Heine University, Düsseldorf**
- Kornelia Neveling, MSc, **University of Würzburg, Würzburg**

Identification of the FANCI Protein, a Monoubiquitinated FANCD2 Paralog Required for DNA Repair, Cell:

- Agata Smogorzewska, MD, PhD, Shuhei Matsuoka, PhD, E. Robert McDonald III, PhD, Kristen Hurov, PhD, Ji Luo, PhD, Bryan Ballif, PhD, Steven Gygi, PhD, Stephen Elledge, PhD, **Harvard Medical School, Boston**
- Patrizia Vinciguerra, PhD, Alan D'Andrea, MD, **Dana-Farber Cancer Institute, Boston**
- Kay Hofmann, **Miltenyi Biotec GmbH, Köln, Germany**



Authors of Cellular Oncology article on FANCI

Honoring Initiative in FA Research: *FANCN* Discovery

Fanconi Anemia is Associated with a Defect in the BRCA2 Partner PALB2, Nature Genetics:

- Bing Xia, PhD, Qing Shenbg, PhD, David Livingston, MD, **Dana-Farber Cancer Institute, Boston**
- Josephine Dorsman, PhD, Najim Ameziane, MSc, Yne de Vries, MSc, Martin Rooimans, Gerard Pals, PhD, Hans Joenje, PhD, Johan de Winter, PhD, **Free University, Amsterdam**
- Abdellatif Errami, PhD, **MRC-Holland BV, Amsterdam**
- Eliane Gluckman, MD, **Hôpital Saint-Louis, Paris;**
- Julian Llera, MD, **Hospital Italiano de Buenos Aires, Buenos Aires**
- Weidong Wang, PhD, **National Institute on Aging, Baltimore**



Authors of one of the Nature Genetics articles on FANCN

Biallelic Mutations in PALB2 Cause Fanconi Anemia Subtype FA-N and Predispose to Childhood Cancer, Nature Genetics:

- Sarah Reid, Karen Barker, Sandra Hanks, Patrick Kelly, Sheila Seal, Nazneen Rahman, MD, PhD, **Institute of Cancer Research, Sutton, United Kingdom**
- Detlev Schindler, MD, Reinhard Kalb, MSc, Kornelia Neveling, MSc, **University of Würzburg, Würzburg**
- Helmut Hanenberg, MD, Marcel Freund, Melanie Wurm, **Heinrich Heine University, Düsseldorf**
- Sat Dev Batish, PhD, Francis Lach, Arleen Auerbach, PhD, **The Rockefeller University, New York**
- Sevgi Yetgin, MD, **Hacettepe University, Ankara, Turkey**
- Heidemarie Neitzel, MD, **Charité Universitätsmedizin, Berlin**
- Hany Ariffin, MRCP, **University of Malaya Medical Centre, Kuala Lumpur, Malaysia**
- Marc Tischkowitz, MD, PhD, **McGill University, Montreal**
- Christopher Mathew, PhD, **King's College School of Medicine, London** ♦



Researchers discuss posters at the Symposium.

The Brannock Brothers

by Jason and Daniel (Boone) Brannock

Editor's Note: Jason Brannock wrote "Welcome Home, Jason" to describe what he had learned from a significant life event. His article is followed by his brother Boone's short essay that he used for a college application. Boone will be attending the University of North Carolina, Chapel Hill this fall.

Welcome Home, Jason

by Jason Brannock

In the spring of 2004, I was in the Children's Hospital in Cincinnati, OH for a bone marrow transplant. The donor of my new bone marrow was my older brother, Daniel (Boone). With dozens of pricks, doctors sucked out about a pint of bone marrow from my brother. At the same time, they needed to completely obliterate my old bone marrow with radiation and other chemicals, so getting even a little bit sick could literally kill me. Finally, the doctors put the new marrow into my blood, and it eventually spread throughout my bone marrow. But, it's not the bone marrow transplant I want to tell you about; it's the relationship I had with my fellow second grade classmates and my family during my stay in the hospital.

I never thought I'd be loved so greatly—even by my friends from the second grade, who cheered me by sending a box full of heartwarming gifts: a lot of handmade cards, a video of them all saying "hi" and wishing I'd get better soon, and a giant quilt with a drawing from each classmate. "Hi Jason! I hope you feel better and get back soon," one of them exclaimed. I felt much more reassured, and a little teary-eyed when I read some of the cards, and then thought that, whatever happened, no matter what it took, I had to get home as soon as possible.

The transplant took about three months. It wasn't as bad as I thought it would be. Even while I was isolated due to my immune deficiency, I still had family visitors come and stay for about a week long each. I felt really appreciated because they all came from a long way away.

Brothers fascinate me. No matter how many times you fight with them, they love you anyway. That's exactly how it was with my brother Daniel. He would always



JD, Jason, and Boone Brannock

cheer me up. He would play almost any game I wanted to play. The best part was, he did it all on his own free will, so he wasn't forced to do it.

"Well now, hold on a second. That was just a practice round. This is the real thing now," argued my granddaddy. "Granddaddy, that was not a practice round. I won the game fair and square," I replied. My grandfather would always say that it was just a practice round every time I won a game. It used to really bug me a lot, but now I realize that he was just a very smart man. It may sound weird, but he knew I needed another kid with me all the time. He was that kid for me. I needed someone to fuss with, argue, and play with. He was with me the entire time I was in Cincinnati. That's saying a lot.

Although the bone marrow transplant was hard, it was nice to see how much my family and friends really cared for me. When I got back home, my mom drove me to school. I was not allowed to be in direct contact with any children at that time, but they showed me how much they missed me all the same. The whole second grade class was standing on the sidewalk holding up signs saying, "Welcome home, Jason!", "We missed you!", and "You did it!" It had been a long journey but, with the help of my family and friends, I made it back home. Really back home.

continued on page 16

The Proctor Family's Journey with FA

by Kay Proctor, with a postscript from Roy

Mike and I were married in 1978 and have lived in Eastern Oregon since. When our first child, Josie, was born in 1980, she was cute as a button, happy and healthy. By the age of two years, her growth rate had slowed. With several female relatives hovering around the 5' mark, no one was too concerned.

But by age seven, Josie's height was well below normal. After many tests, we learned that her pituitary gland was abnormally located and that her thyroid and growth hormone levels were low. No primary diagnosis was made, and the pediatric endocrinologist told us that our daughter was a bit of a mystery to him.

At age eight, Josie began a low dose of thyroid replacement medication. At age 10½ years old, she was just 3'11" tall and weighed 54 lbs, so she began daily injections of human growth hormones (HGH). By age 13½, she had reached 4'11½" and weighed 98 lbs. She chose to quit the daily injections then and just be "normal." Josie continued to grow on her own, eventually reaching 5'1". Our family continued with our typical American lives, keeping up with our children's activities, such as 4-H, Future Farmers of America, and sports.

Then, at a routine doctor visit in January, 2000 for our youngest child, Roy, age 12, the doctor thought that Roy looked pale and ordered a CBC. The doctor called us within an hour of the test with the news of terribly low blood counts and asked how sick Roy was. I replied, "Not at all. In the past 24 hours, he had three friends at our house for a sleepover, had a double 4-H Cooking Club meeting, bowled three games for his bowling team, and is on his way to spend the night with a friend."

Those plans—and our lives—quickly changed. By April Roy was diagnosed with Fanconi anemia, as was Josie, which solved her earlier medical mystery. Our middle child, Julie, was thankfully left out of the FA diagnosis.

Besides a few small *café au lait* marks on his trunk and no fat pads at the base of his thumbs, Roy had no other outward signs of FA. He said he felt tired, but he thought everyone felt that way. His growth rate slowed. However, HGH treatment options for Roy were soon not an option as his growth plates had fused and he was out of puberty. He reached 5' 3½" several years ago.

Roy's immunity failure began to result in hospitalizations: a week in 2004 for high fever from an unknown virus; four days in 2005 for a viral infection; and five weeks in 2006 for pneumonia, which resulted in the surgical removal of Roy's lower left lung and nearly losing him from post-op complications. Somehow, he fought his way through. Although we knew that Roy was in imminent need of a bone marrow transplant, for us it was best to postpone a transplant while research continued to improve survival rates for high-risk candidates such as Roy. But, when he was diagnosed with Sweet's Syndrome in the fall of 2006 while at college, we all knew that it was time for transplant. With no related matches, an unrelated donor was found—an eight out of ten match but, nonetheless, Roy's lifesaving donor! With our family's background of Russian, Irish, German, Cherokee, and English, the donor search was not easy.

April 2007 found Roy and me in New York City at Memorial Sloan-Kettering Cancer Center's (MSKCC) Pediatric Hematology Transplant Center in the care of Farid Boulad,



Roy Proctor

MD, and his medical team. The scariest moment during the whole process? That would be Roy's first night as an in-patient on the Transplant Floor. His system reacted to the total body irradiation with a high fever and plunging blood pressure, and he was transferred to Intensive Care. The best moment during the transplant was when Roy received his donor's precious stem cells on May 17th. It took only a few ticks on the clock to inject those life-giving cells into Roy to begin their miracle. At the time, I wanted to do cartwheels all the way down the hospital's hallway—it was an incredible feeling!

There were difficulties during our stay in NY. A lot of time and energy was spent dealing with Roy's stomach pain. Yet his biggest challenges were the diet restrictions during his neutropenic stage and avoiding crowds. During most of his recovery in NY, he felt better than he had ever remembered feeling and at age 19, it was impossible to hold him back from living "*la vida loca*."

MSKCC believes in treating the mind as well as the body, so their transplant teams include a

continued on page 17

In Memory of Dani Sacks

by Bob Sacks

My wife, Andrea, and I have spent a lot of time grieving over the loss of our daughter, Dani, who died from complications related to Fanconi anemia. Every day she remained alive became another blessing for all of us, and she eventually became the oldest living patient with the Ashkenazi Jewish mutation of the FA-C gene. We hope and pray that those who follow Dani go on to easily break that record.

We and those who knew Dani well were truly blessed by her presence. Her best friend and adopted second mother, Lynn Green, described her as a Superwoman, cat-like in her abilities to survive many deadly encounters with the medical system, while portraying a stoic, courageous and happy attitude toward all who were lucky to spend time with her. She was our family hero, along with our son, Sean, whose bone marrow rescued her from certain death at age nine. He has continued to save lives as a volunteer paramedic every week for the Howard County Fire Department.

Andrea and I have been tormented by the meaning of Dani's death, which is both spiritually and physically taxing on our beliefs. Dani loved life and willingly did combat with death whenever necessary. Knowing that, we had considerable disdain for comments from medical providers who wanted Dani to make the decision to withhold medical care and let nature take its course in lieu of her courageous losing battle with cancer. We knew that was not her choice and certainly not ours. Without Dani and her indomitable spirit, medical science would not have advanced as quickly. Many of the medical mistakes she encountered are no longer practiced at the hospitals where she was treated. Without Dani, we all would have missed that giggle, smile and loving embrace of a child toward a parent or friend. We would not have enjoyed her too short life with us.



Marie, Adrianna, and Antonino DiMercurio with Andrea (top) and Dani Sacks

While Dani was alive, there was a sublime, incredibly loving connection between her and those who knew and admired her. And, when she was taken away from Andrea and me, a bleak numbness crept into our souls that had us doubting ourselves. It troubles me greatly that, had we known of Dani's genetic disease before she was born and all the physical traumas and premature death it would eventually cause, we might have made a choice to terminate Andrea's pregnancy. So perhaps it was a blessing that the disease was too rare for many in the medical profession to diagnose or understand at the time she was born. She went undiagnosed by orthopedic specialists and pediatricians for her first five years of life despite having the classic, physical anomalies of the disorder.

God instills in us a capacity to love one another, but it takes a Fanconi anemia child like Danielle to realize the depth of a spiritually, everlasting love which is God's essence. So while we grieve selfishly for the loss of our angel, spiritually she remains within us. ♦

In Loving Memory

YiChen Cheng
8/4/03 ~ 12/12/07

Clay Eubanks
11/7/75 ~ 2/02/08

Michelle Humboldt
4/19/68 ~ 4/7/07

Jack Simbulan
6/11/01 ~ 10/29/07



Noah Felmy

by Carole Felmy

Our second son, Noah, was born on Christmas Eve 2003 in Melbourne, Australia. He was tiny, only 2 kg [5 pounds], and he was beautiful. Michael and I were very proud, of course.

Noah was diagnosed with FA a few weeks later, while he was recovering from a surgery done on his duodenum. The professor who talked to us then was blunt, but within a year Noah had grown into a happy, healthy and active little boy, and I got carried away with hope of a cure being found before anything bad happened.

On his second birthday, we shared a big chocolate mousse with friends and family. Noah loved chocolate and would have been happy living on a diet of milk and chocolate. For a while all seemed to be going well: check-ups at the hospital were getting further apart; his blood counts were always very good; he was growing slowly but surely; barely caught any cold; was as affectionate as he was cheeky; and had endless energy to run around and follow his big brother everywhere.



Noah Felmy

Then, about a month before his third birthday, he grew more and more tired, lost his appetite, and had recurring gastrointestinal problems. On his birthday and Christmas, he managed a few smiles but mostly cuddled in my arms and wasn't much interested in even opening presents.

We brought him to the Children's Hospital on December 26. After two days and many tests and x-rays, the doctors didn't understand what was going on and still only suspected something on the digestive level. We came back home for a night as Noah seemed to keep a bit of food down that day. But the next morning we drove back to the hospital emergency department. Things quickly deteriorated until, eventually, Noah had a seizure. Doctors and nurses were over him within a second and gave him all the medication needed so he would not feel pain. He was put into

a coma and a brain scan was done.

It revealed a tumor in the left lobe of the brain. Neurosurgeons were called immediately, and then the final diagnostic fell.

The tumor was just too big. It had spread to many places and even down to the spine. Nothing at all could be done. We were offered the possibility of waking him to say goodbye, but this meant that Noah would have felt pain. So we didn't. We just kept holding him, telling him our love again and again through our tears and, then, on New Year's Day 2007, he passed away.

Noah was three years and eight days old. We miss him every day, every hour. I wish I could share more. It may come later, but now it's been over a year and the grief is still crippling.

But I keep hope for the rest of your children. May a cure be found.



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.*



Karly with her Science Fair exhibit

My Science Fair Project

by Karly Ross

For our Bronze Award, my Girl Scout Troop 2563 held a Science Fair. For my project, I decided to learn about bone marrow transplants, because I wanted to know more about what I went through during my transplant on March 26, 2003 when I was only seven years old. I'm glad I learned more about the transplant and my disease, Fanconi anemia. What I went through was scary, but I'm better now.

Eighteen projects were entered, and over 80 people came to our Fair. The people who came to the Science Fair were interested in learning about Fanconi anemia after they saw my display. Nobody had heard of it! But now they know it exists and why I needed a transplant. They learned about how the bone marrow works and how the transplant replaced my failed marrow with a Marine's "super marrow" that works very well. My troop got our picture in the newspaper, and we earned our Bronze Award, the highest award a Junior Girl Scout can earn! ♦

The Brannock Brothers

continued from page 12

1996

by Boone Brannock

1996, for me, was the year that everything began. 1996, for me, was the year that my brother became abnormal.

Although 1996 is the year that will notoriously stand out in my mind as the time in which Jason became different, it is not so for the rest of my family. For them, it began in 1994, the year that Jason was born and experienced his first of several surgeries. Only when I turned six and he received his second tracheotomy did my little brother seem any different to me than all the other little boys. From then on, conditions did not improve for Jason and, several surgeries later, he found himself trapped in the hospital again—only this time, I was with him. Jason had been diagnosed with Fanconi anemia, a blood disease so rare that it becomes underlined in red as a typo the moment I finish typing it. As a result of FA, in 2002 Jason began to develop the earliest signs of leukemia and needed a bone marrow transplant. Through the will of God (and this is one of the chief occasions on which I base my faith), I am a perfect donor match for Jason. That is why he lives today and why he has my marrow in his bones.

Due to this early introduction to genetic disorders, I have always been fascinated with genes and how they are passed down from parent to child, particularly genes which cause illness. More than anything else, I have dreamt of researching how to prevent families from being put through the same stress that faced, and continues to face, my family. ♦

The Proctor Family's Journey with FA

continued from page 13

psychiatrist with whom Roy and I talked to face the years of mental challenges and stress from dealing with FA. It is important to own a positive attitude and to keep strong. I urge FA families to find their source of strength and go to it often to renew their energy and resolve. My strength is my faith. Roy found strength in meditation, and my husband's strength is visiting the outdoors of Eastern Oregon: the Columbia River or the Blue Mountains.

Health insurance and medical bills have been a concern the last eight years. Even though Mike and I have group health insurance through our employers, our out-of-pocket medical expenses have reached over \$110,000 in the past eight years.

Roy and I returned to Oregon in October. Dr. Peter Kurre at Doernbecher Children's Hospital in Portland follows him locally. Roy is currently dealing with a post-transplant challenge of shingles on the skin of

his trunk and arms. However, with his new and improved immunity, he is dealing with it.

We do not know what is in our future, but we will deal with that as it comes. Roy has already adjusted to believing he now has a future. He is currently visiting a dear and special friend in Toronto, Ontario and taking online college courses. Josie is married and in good general health. She is physically active and works with children. Julie has worked hard over the years helping everyone—she is a very giving person. She will finish college soon. My husband is lighter in spirit since our return from New York. Except for a short visit in July, Mike stayed in Oregon to work, which was hard for him to do. As for myself, it has taken awhile to settle in. I do find myself shaking my head when I hear others complaining about trivial matters or creating their own problems. I pray that they realize what really matters in their lives and count their blessings every day. My family and other FA families have learned just how precious each and every day of life is.

Roy has sent this message:

To those who face transplant:

I am trying to think of why I constantly pushed and ignored the doctor's guidelines and still came out of transplant with few complications. That answer I do not know, but I know this: do not be afraid. There is nothing in this procedure that will hurt you more than you most likely have already been hurt. It is going to be a chapter of your life's book that is all to itself; yet, the great hope regarding this book is that you get to have such an impact on how it is written.

Your story will go on, weaving through paths of wonder and discovery; through turmoil and doubt; through hope and love. We all have our challenges in life; we all must meet those in either victory or failure. Such is life, and it is what it is. You must go through what you must. There is work ahead.



*The woods are lovely, dark and deep
But I have promises to keep
And miles to go before I sleep
And miles to go before I sleep.*

-Robert Frost



For more information about Roy's stem cell transplant, visit royproctor.caringbridge.org. ♦



Roy and snowy friends

Our Journey with Fanconi Anemia

by Olivia Mindle

Our journey with FA began while we were expecting our third child, Sara. During a routine ultrasound, our perinatologist told us that our baby had hydrocephalus (abnormal accumulation of fluid in the ventricles of the brain) and that, at best, she would be born with significant brain damage and, at worst, she would die either in *utero* or shortly after birth. He recommended that we promptly terminate the pregnancy.

Petrified and deeply saddened by the diagnosis and recommended course of action, we consulted with a top pediatric neurosurgeon at Children's Hospital in Los Angeles. He told us that, while these horrible, unimaginable things could happen to our daughter, it could also be true that none of those things would happen. We left his office with hope in our hearts and prayers. We also found a new perinatologist.

As the pregnancy progressed, our new perinatologist determined that our baby had duodenal atresia (a condition in which the small bowel has not developed properly). Additionally, the ultrasound showed that she had clenched fists, a possible sign of some level of brain damage. At one point, our pediatric neurosurgeon suggested that we perform surgery to correct the hydrocephalus in *utero*. Despite the mounting medical evidence suggesting that our baby would have severe brain damage, we hoped and prayed for a miracle.

On April 24, 2002, our beautiful daughter Sara was delivered three weeks early by a C-section. Although she was only 4 pounds at birth, she



Adam and Olivia Mindle with children Sara, Michael and Joshua

looked perfect. We could not imagine that such a beautiful baby could have the terrible prognoses constantly forecast by our doctors.

As a result of the duodenal atresia and Sara's premie status, she was kept in the neonatal intensive care unit (NICU) for six weeks. While there, she had surgery to correct the atresia and attempted to avoid the many medical challenges that seemed continually to arise. As Sara fought for her little life, the doctors performed an MRI of her brain to determine if surgery would be required to relieve the pressure on her brain caused by the hydrocephalus. The results of the MRI were miraculous: no evidence of hydrocephalus—just big ventricles, no different than people who have big eyes or teeth! An enormous weight was lifted from our hearts, and we felt that we could breathe a sigh of relief and put this horrible chapter of our lives behind

us. Unfortunately, we were wrong.

After the MRI, a geneticist contacted us and informed us that Sara's hypoplastic (undeveloped) thumbs and the duodenal atresia could be markers of a rare genetic disease called Fanconi anemia. Needless to say, we were shocked and angry. After Sara proved all of the doctors wrong, how could this doctor give us this horrible, devastating news? Sara was fine; she was perfect. We had no family history that suggested any connection to FA. We were convinced that the geneticist had to be wrong. Unfortunately, a week after Sara was tested, the diagnosis was confirmed. Our beautiful, sweet daughter had FA.

Thus began the most frightening journey of our lives. While Sara was still in the NICU, her two older brothers were tested for FA. To our great relief, they tested negative. All of us were also tested to determine

if we were bone marrow matches for Sara. Thankfully, our oldest son, Joshua, was a perfect match. When the time came, Joshua would be able to donate his marrow to save Sara's life.

After Sara came home from the NICU, we desperately tried to find our way back to a normal life. Over time, we came to realize that we would never have a "normal" life. We had to learn to live with the reality of life with FA, and we had to find a new definition of normalcy for our family. Believe it or not, it was our Sara who healed us. Her smiles brightened our spirits, and her warm hugs and constant laughter carried us through. The child whom the doctors said would be a burden was actually a blessing.

Two and a half years later, one of our worst fears came to fruition: Sara's blood counts began to drop. We faced the biggest decision of our lives: do we go to transplant and endanger Sara's life while she's still healthy? After carefully weighing all of the factors, consulting with all of the doctors and praying for guidance, we decided to go to transplant in March of 2005 at City of Hope Comprehensive Cancer Center in Duarte, California. This was a very daunting, emotional, and exhausting time; one of us was either with Sara or our boys at all times. Although it was painful at times and extraordinarily difficult to keep a two-year-old in one room for weeks on end, we were blessed with a relatively smooth transplant experience. Five long weeks later, we were elated and grateful to bring Sara home. One week later, we celebrated Sara's third birthday.

Today, we are thankful that Sara's counts continue to climb steadily. She started school last year and, outside of a few short hospitalizations (for pneumonia and urinary tract infections) and some bad colds, we've been blessed that Sara's been doing great.

With Sara home and "healthy," we decided to focus our attention on the one thing that could possibly save her life other than our prayers: raising funds to find a cure. We sent out our first fundraising letter two years ago. It was very difficult and painful to disclose Sara's plight to others. Even though we had been living with the reality of FA on a daily basis, somehow, putting it down on paper with Sara's name on it made it seem even more real. However, we were overjoyed with the generous donations that we received. The response to our letters again this year was even greater. It is our sincere belief that we have the ability to find a cure to this horrific disease if we work together to raise the money required to fund the research.

The pain of that first day when we found out that Sara had FA has never truly gone away. Usually, it sits in the core of our hearts—a dull ache that attaches itself to every moment of everyday. Other times, the pain floods in and overtakes all of our being. We once saw a psychologist who told us, on those days, to imag-

ine that we are in a big file room with many file cabinets. When the pain is overbearing, we open up one of the file drawers, put the pain and all the thoughts that go with it in the drawer and shut it. We file the pain and heartache away for a time when we feel strong enough to cope with it.

We've learned that our faith and our children continually provide the source of our strength to go on. Their laughter, innocence, hope, joy and unconditional love force us to avoid despair and look forward to helping find a cure. We look at Sara as an example of how to live. After all of the pain and adversity that she has endured during her five short years, she has come out smiling and happy. She has a smile for and is loving and caring to everyone she meets: doctors, nurses, teachers and classmates—and even her older brothers! If she can do it, then so can we. Sara continues to inspire us to live and do the very best that we can.

Thank you for letting us share our story with you. We pray for a cure and in the meantime, we pray for health and for strength for us all. ♦

Mark Your Calendar

Annual FA Family Meeting

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

Friday, August 8–Tuesday, August 12, 2008

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. Scholarships are available through the Fund (application deadline is July 1). Contact Suzanne Planck at suzanne@fanconi.org or at 1-888-FANCONI.

The Best Insurance for the Future

by Lynnette Lowrimore

My younger brother Mark Lamer died in 1969 at age nine of Fanconi anemia. He is buried in Arlington Cemetery underneath a big maple tree with a breathtaking view of Washington, D.C. Mark was the middle of five children born to my military officer father and nurse mother. He was always a sickly child, and when he was four, my parents got the FA diagnosis from the doctors in Boston. Off and on, for his remaining five years of life, he had long stints in the hospital and/or visits to the doctor for examinations. All of us kids were really too young to process how very sick he was, and my parents worked hard to keep it that way. I can't imagine how hard that must have been for them—especially because there was so little information about the disease in the 1960s.

Even though Mark was an undersized, sickly kid, he had a huge heart and an uncanny way of connecting with even perfect strangers. The church where his funeral was held was filled to overflowing, and more people waited in silence out on the lawn. I'll never forget how many people felt strongly enough about him to come mourn his death and celebrate his brief life.

I'm not really sure when or where I read about the Fanconi Anemia Research Fund, but I immediately knew I wanted to contribute to it on a regular basis. By that point, I was a mother myself, with two healthy daughters. In the back of my mind, I knew that FA is genetically inherited, so the possibility existed that the seeds of the disease were somewhere in the DNA I passed on to my daughters. Thus, my contributions to the Fund are like paying an insurance premium: someone in my future family tree could benefit from the money I send in each month. It's

like the movie about good deeds and how you need to "pay it forward." I treat my bimonthly contributions to FARF as a "bill." In the military, I get paid twice a month, so I have 24 envelopes pre-addressed to take me through the year.

My children are adults now and still blessed with good health. My oldest made us grandparents of a vivacious, healthy, little red-haired darling 15 months ago and will give us grandchild #2 in the summer. The years of contributions previously made to the Fund for their research represent a nest egg—one I hope never to collect on, but there gathering interest nevertheless. Other families, not so lucky, are benefiting from the cutting-edge research, family camps, and scientific seminars that my parents didn't have available when their world seemed so bleak

after Mark's diagnosis, illness, and death. There was no internet website filled with valuable information to fill in the huge gap in knowledge and no support staff to walk them through the available avenues of doctors and treatment options. They did not have professionals armed with knowledge AND empathy to make them feel they weren't carrying the heavy burden alone.

I marvel at the medical advances when I get the newsletters, but I am especially uplifted by the varied ways that the families are supported. There is power in community—even if it is one that you would prefer not to have to be a part of. Even though my family's experience was long, long ago, I feel part of that community and will continue to support the Fund financially as long as I am able. ♦



The Lamer Children: Matthew, Michelle, LeeAnne, Lynnette, and Mark

Fourth Annual Fanconi Anemia Valentine 5K Walk/Run

FA parent Peggy Padden organized the FA Valentine 5K Walk/Run in Portland, OR, again this year to benefit FA research. This event, on Sunday, February 10, attracted 600 entrants and raised \$10,000 more than last year's run!

The FA Valentine 5K benefits greatly from the support and participation of many doctors and researchers at Oregon Health & Science University who research FA. Peter Kurre, MD, the OHSU hematologist for Peggy's son Spencer Shearer, noted that "The 5K walk illustrates the grassroots support that fuels FA research. This is a chance to raise awareness and demonstrate our commitment to the research that gives hope to patients like Spencer and their families."

In addition to the 5K, participants were offered the opportunity to join the National Marrow Donor Program (through a simple mouth swab). Sixty-seven runner/walkers took advantage of the opportunity and are now entered as potential donors of bone marrow for lifesaving transplants! ♦



Spencer Shearer, Andrea Schleter, Katie Watson and Katie Anderson having fun at the Run!

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.

Brave Hearts 2007: Hoot 'N' Holler

by Jeanne Atkinson

“Brave Hearts,” the name for the fundraising event we sponsored with Lisa and Jack Nash and through our foundation, the Kendall and Taylor Atkinson Foundation, was chosen to reflect the courage, strength and bravery that it takes to live everyday in the shadow of FA. One must have a brave heart to survive the journey and to fight the best fight. It is a bravery born in the heart, not the head—a bravery of love.

In 2006, at the age of 56 and not having ever run much over six miles, my husband Ken ran the NYC Marathon in memory of our children Kendall and Taylor. We raised over \$75,000 for the Fund through a letter asking for support for Ken’s run, and he contributed by finishing the race—and later undergoing knee surgery!

Then, we felt that we wanted to give something back to our fabulous donors by creating an evening that would be fun for all who came. Most of our donors have supported the fight against FA for years and have supported our family in ways too countless to name. Yes, those



Suzanne Miller, Will and Lisa Freyschlag, Julie Colby, and Mary Ellen and Tim Haynes enjoying the Hoot 'N' Holler

invited would be asked to donate for FA research, but we wanted them to come away feeling that they sure had a lot of fun in the process. Hoot 'N' Holler seemed to conjure up images of “fun” and be the perfect name for the 2007 Brave Hearts event. We contacted Denver FA parents Jack and Lisa Nash to team up with us.

Their daughter Molly, who has FA, attended the event and worked her way into the hearts of all present. Without this Atkinson/Nash team effort, the event would not have been nearly as successful as it was.

And, it was very much a success! Our total net profit was over

continued on page 23

As Hillel said,

*If I am not for myself,
who will be for me?*

*If I am not for others,
what am I?*

And if not now, when?



Molly Nash, Ricky Zinter, Sarah Robbins, and David Fiaschetti at the Blackjack table

Upcoming Fundraisers for FA Research

2008

March 1: Pine Island Rock and Roast for Fanconi Anemia, Pine Island, FL. All day live music, live and silent auctions and raffles. Contact: Joe Feidner, 239-464-6736.

March 8: Evan's Enchanted Evening, Green Bay, WI. Dinner, oral and silent auctions, dance. Contact: Kim Connelly at Kimjohnconns@aol.com.

April 5: Crop'In for a Cure, Green Bay, WI. All day scrapfest with meals, contest, giveaways, raffle and prizes. Contact: Kim Connelly at Kimjohnconns@aol.com.

April 27: Annual Toronto Fundraiser to Support Fanconi Canada, Toronto, ON. Fabulous three-course dinner, silent auction, and music by the Ron David Quartet.
Details at <http://www.fanconicanada.org/en/inf71.php>

May 18: Eighth Annual Concert and Wine Auction, Eugene, OR.
Contact: Sharon Schuman, sschuman@uoregon.edu.

June 20: Fourth Annual Coley's Cause Memorial Golf Tournament, Lakeville, MA. Golf tournament, silent auction, dinner and raffle. Details at www.coleyscause.com.

July 12: 5th Annual FA Golf Tournament, Silent Auction and BBQ, Cedars on Salmon Creek Golf Course, Battle Ground, WA. Contact: Peg Padden at pegpadpad@hotmail.com.

November 15: Brave Hearts 2008 Hoot 'N' Holler, Denver, CO. Food, drinks, silent and live auctions, entertainment, and Texas Hold 'Em. Contact: Ken & Jeanne Atkinson at 303-349-1309 and Jack & Lisa Nash at 303-773-6228.

November [exact date TBA]: Play for FA, Bookbinder's Grill, Midlothian, VA. Dinner. Contact: Lorraine McQueen at lmcqueen01@comcast.net.

Ongoing

Kaps for Kendall: In memory of Kendall Atkinson, donate to the Fund by sponsoring a volunteer to knit hats for children and adults who lose their hair to chemotherapy and radiation. Contact: Allison and Whitney Atkinson at www.kapsforkendall.org.

Caddy for a Cure: Caddy for a Cure, Inc, generates charitable funds for designated 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.

Brave Hearts 2007: Hoot 'N' Holler

continued from page 22

\$130,000, and everyone there had a great time. Most wore jeans or casual Western clothes. Food was served at food stations, rather than through a sit-down dinner. We sold "Cowboy Coffee" (Kahlua) in boot-shaped shot glasses for a chance to win a diamond. Stick horses made from grocery bags and yardsticks were sold for \$50 and were associated with a prize bag worth at least that much. Kendall and Taylor's kindergarten teacher made them exactly like the ones they used when they were in kindergarten during "Rodeo Week." One of the highlights of the silent auction was the "Faces of FA" table, which displayed pictures and brief descriptions of FA children. We were very grateful to the FA families who took the time to send information about their children.

When all was said and done, "Brave Hearts" was a lot of work and a lot of fun—and "Brave Hearts 2008...Hoot 'N' Holler" is on for November 15! ♦

Screening for Pre-Cancerous Lesions

continued from page 5

patient and donor cells.

Brakenhoff concluded that multiple genetic alterations in the oral mucosa of FA patients are found in a high proportion, which is in line with the increased risk for SCC. As treatment options for SCC in FA are very limited, early diagnosis and immediate surgical intervention are of paramount importance. Brakenhoff's laboratory will focus on the prognostic value of his findings, and develop strategies for BMT patients. ♦

Third Annual AGIA Foundation Golf Classic

The La Costa Resort & Spa in Carlsbad, CA, was the scene of the Third Annual AGIA Foundation Golf Classic on November 13, which benefited research into Fanconi anemia. Over 180 golfers participated in this event, which featured 18 holes of golf on both the North and South golf courses at La Costa. After the golf round, 250 participants attended a dinner and live and silent auction at the Yellow Coyote Tortilla Factory. Amy Frohnmayr, the daughter of the Fund's co-founders Dave and Lynn Frohnmayr, spoke to the group about living with Fanconi anemia and the urgent need for research into the disease. One of the attendees wrote later that "the talk by Amy was very moving and inspiring. Makes me pause and appreciate my family's incredible blessings." For her part, Amy was awed and appreciative of the support shown by the Foundation and its supporters.

The Golf Classic, which raised over \$200,000 for the Fund, was spearheaded by Stephanie and Patrick Kilkenny as part of their continuing commitment to help find a cure for FA. The Kilkennys are aware of the urgent need for research into Fanconi anemia because of their personal friendship with Glen and Peggy Padden Shearer and Lynn and Dave Frohnmayr, parents of children with FA.

If you would like to attend the Classic in 2008, send a note to PO Box 7271, Rancho Santa Fe, CA 92067 or email Jerid Keefer at jkeefer@arrowheadgrp.com. ♦



Stephanie Kilkenny and Marianne Harmon, members of the AGIA Event Planning Committee



Foursome Jimmy Dimatteo, Steve Harmon, Jerry Gammieri, and Jeff Denning, a member of the Planning Committee

Your FA Research Dollars at Work in 2007

In 2007, the Fanconi Anemia Research Fund awarded \$1,188,197 in research grants to the following projects:

Investigator: Simon J. Boulton, PhD, and Spencer Collis, PhD, London Research Institute, Cancer Research UK, London, UK

Title: *Elucidating the Role of HCLK2 in the Fanconi Anemia Network*

Amount: \$137,778

Investigator: Robert Brosh, Jr., PhD, National Institute on Aging, NIH, Baltimore, MD

Title: *Molecular and Cellular Investigation of the FANCI Helicase Defective in FA*

Amount: \$50,000

Investigator: Laura E. Hays, PhD, Oregon Health & Science University Cancer Institute, Portland, OR

Title: *Comparative Genetic and Metastatic Potential Analyses of Head and Neck Squamous Cell Carcinomas from Wild-type and Fancd1-deficient Mice*

Amount: \$55,000

Investigator: Maureen Hoatlin, PhD, Oregon Health & Science University, Portland, OR

Title: *DNA Structure-specific Activation of the FA Proteins FANCD1 and FANCD2*

Amount: \$30,539

Investigator: Holger Hoehn, MD, and Detlev Schindler, MD, University of Wuerzburg, Wuerzburg, Germany

Title: *Revertant Mosaicism in FA: Causes and Consequences*

Amount: \$120,000

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*

Amount: \$119,360

Investigator: Ian C. Mackenzie, DDS, PhD, FDSRCS, Institute of Cell and Molecular Science, University of London, London, UK

Title: *Influences of Stem Cell Behavior in Head and Neck Cancers in Fanconi Anemia Patients*

Amount: \$192,345

Investigator: Ruhikanta Meetei, PhD, Cincinnati Children's Hospital Medical Center, Cincinnati, OH

Title: *Defining the Fanconi Anemia-DNA Repair Pathway by Protein Association Analysis*

Amount: \$150,000

Investigator: Amy Skinner, PhD, and Peter Kurre, MD, Oregon Health & Science University, Portland, OR

Title: *Systemic In Situ Delivery of Lentivector to Hematopoietic Stem Cells to Reverse Fanconi Anemia-associated Bone Marrow Failure*

Amount: \$51,389

Investigator: Toshiyasu Taniguchi, MD, PhD, Fred Hutchinson Cancer Research Center, Seattle, WA

Title: *Identification of MicroRNAs Regulating the Fanconi Anemia-BRCA Pathway*

Amount: \$200,000

Investigator: Monica Thakar, MD, and Hans-Peter Kiem, MD, Fred Hutchinson Cancer Research Center, Seattle, WA

Title: *Non-myeloablative Hematopoietic Cell Transplantation for Patients with FA using Related, HLA-Haploidentical Donors: A Phase I/II Dose-Finding Study*

Amount: \$81,786

Family Fundraising Efforts

In 2007, FA families raised an astonishing \$1,816,526 for the Fanconi Anemia Research Fund, exceeding our past annual fundraising efforts. Families accomplished this by sending fundraising letters to their families, friends, and acquaintances and by holding a variety of creative fundraising events, a number of which are memorialized in this newsletter.

One hundred forty-two FA families raised funds this year and, of those, eighty-three families raised \$500 or over and eight raised over \$50,000. Of great importance, the Fund continues to be less dependent on the fundraising efforts of the founders of the Fund, Lynn and Dave Frohnmayer. While they again exceeded their fundraising goal this year, all other FA families combined raised 68% of the total funds raised, compared to 32% for the Frohnmayers. We are grateful to all families who worked so hard to raise funds so that the urgent work of the Fund can continue to move forward at a fast pace.

FA families who raised funds in 2007 are the following:

Over \$500,000

Dave and Lynn Frohnmayer

\$190,000 to \$499,999

Kevin and Katie Rogers
Glen Shearer and Peggy Padden

\$100,000 to \$190,000

Kevin and Lorraine McQueen

\$50,000 to \$99,000

Ken and Jeanne Atkinson
Audrey and John Barrow
Dan and Nikki McCarthy
Steve and Jennifer Klimkiewicz

\$25,000 to \$49,999

John and Kim Connelly
Todd and Kristin Levine
Jack and Lisa Nash

\$15,000 to \$24,999

Michael Glas and Carol Felmy
Peter and Tara Himmelreich
William and Carol Kuell
Tanner and Jessica Lindsay

\$10,000 to \$14,999

Donald and Danielle Burkin
Stuart Cohen and Deane Marchbein
Chris and Susan Collins
Andrew and Jennifer Gough
Brian Horrigan and Amy Levine
Charles and Katy Hull

\$5,000 to \$9,999

Joseph and Nancy Chou
John and Martina Hartmann
Jeff and Beth Janock
Adam and Olivia Mindle
Jim and Holly Miranda
Tyler Morrison and Rachel Altmann
Bob and Andrea Sacks
Mike and Beth Vangel
Joe and Wendy Vitiritto
Michael and Kim Williams

\$1,000 to \$4,999

Andrew and Vicki Athens
Mark and Linda Baumiller
Randy and Nancy Bloxom
Jeffrey and Donna Boggs
Mike and Kerrie Brannock
David and Kim Chew
Tyler and Teresa Clifton
Antonino and Marie DiMercurio
Ed and Janice Duffy
Sandra and Lindsay Dunn
David and MaryAnn Fiaschetti
Stephen and Doreen Flynn
Susan Gannon
Alan and Rachel Grossman
Owen Hall and Margaret Kasting
Milton and Jackie Hardy
Bob and Victoria Hathcock
Saundra Jackson
Lila Keleher
John and Karilyn Kelson
Robert and Jennifer Kiesel
Erik Kjos-Hanssen and Turid Frislid
Gregory and Lynette Lowrimore
Charles and Cecelia Meloling
Matt and Andrea Morris

Tony and Lina Nahas
Robert and Mary Nori
Fred and Nancy Nunes
Derek and Ginger Persson
Peter and Janice Pless
John and Dianne Ploetz
Marcia Reardon
Les and Nancy Ross
Rick and Lynn Sablosky
Ron and Elisa Schaefer
Bill and Connie Schenone
Matt and Diane Senatore
Bryan and Karen Siebenthal
William and Mary Underriner
Marc and Sandi Weiner
Sean and Kristin Young

Up to \$999

Cherie Bank
James and Tracy Biby
Joachim and Joelle Carvahlo
Jaclyn Catlett
Bob and Carole Cavanaugh
Leslie Chesler
Jeanette Clark
Laurie Cohen
Brian and Margaret Curtis
Bill and Pat Danks
Donna DellaRatta
James and Carol Dillon
Pat and Mary DiMarino
Frank and Susan Dixon
Brian and Jennifer Dorman
Gene and Lynn Eddy
Sharon Ellis
Clay and Kay Eubanks
Curt and Crystal Fales
Justin and Britteny Ferrin

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 for 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.

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The Twentieth Annual International FA Scientific Symposium

OCTOBER 4-7, 2008

Hilton Hotel and Conference Center
Eugene, Oregon

Note to U.S. and international air travelers: Eugene is served daily by direct flights from Seattle, Portland, Denver, Salt Lake City, Phoenix, Los Angeles, and San Francisco. Eugene is an easy two-hour drive on Interstate 5 from Portland International Airport.



Fanconi
Anemia

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