



# FA FAMILY NEWSLETTER

#34

A Semi-annual Publication of the Fanconi Anemia Research Fund, Inc.

Fall 2003



## Fun, Science, and Optimism at the Annual Family Meeting

FA families and FA adults from North America, Asia, and Europe returned to Camp Sunshine in Maine for four days in August for the Annual FA Family Meeting. Once again, the participants were overwhelmed by the beautiful location and the friendliness and support of the Camp Sunshine staff and volunteers. The kids were immediately swept up by the volunteers for non-stop fun. The Masquerade Ball, the talent show, miniature golf, volleyball, shuffleboard, the puppet show, arts and crafts, the magic show, the bonfire, swimming, archery, the climbing wall, the visit from the Harley Club, and stories and singing for the little

ones provided a wonderful overload!

The kids who had never before met someone else with a missing thumb or other physical manifestation of FA suddenly were able to make friends with other kids just like themselves. The kids who had met each other at Camp Sunshine in past years got to renew old acquaintances. Parents and staff were heartened by how quickly these friendships were renewed and the strength of the bond between these youngsters. And, parents who have been isolated dealing with this disease in their home communities suddenly got to meet—and become fast friends with—other

*continued on page 14*

## New FA Protein Discovered; Function Described

A group of FA scientists has just published a paper in *Nature Genetics* (October 2003) describing the isolation of a new FA protein, an enzyme called a ubiquitin ligase, which plays a crucial role in DNA repair. Although several other FA proteins have been isolated, and are known to form a complex within the cell, this is the first FA protein shown to function as an enzyme, a protein which promotes a chemical reaction. It occurs in the cell as part of the FA protein complex. It activates the

*continued on page 20*

### HIGHLIGHTS

Radiation Eliminated in HLA-Matched Related Donor Transplants: Results Outstanding...	2
Risk Level Helps Predict Outcome in Minnesota Unrelated Donor Transplants.....	2
Memorial Sloan-Kettering Achieves Exceptional Results with High Risk Transplants .....	3
Head and Neck Cancer Prevention Trials Open .....	6, 8
Family News .....	10
Fundraising .....	17



*Anders Kolb, MD; John Wagner, MD; Richard Harris, MD; Wolfram Ebell, MD participate in panel discussion at FA Family Meeting.*

## Risk Level Helps Predict Outcome in Minnesota Unrelated Donor Transplants

At the University of Minnesota Bone Marrow Transplant Program, a patient's risk level prior to an unrelated donor transplant is highly predictive of survival. Patients deemed "standard risk" do well, with a probability of survival at two years post-transplant of 76%. However, survival for "high risk" patients is only 34%. Several factors can place a patient in the high risk category: age over 18; presence of advanced myelodysplastic disease (MDS) or leukemia; history of gram negative or fungal infection before transplant; recipients of unrelated donor HLA-mismatched bone marrow; or two antigen HLA-mismatched umbilical cord blood.

Several other factors also appear to be associated with a less favorable outcome. These include greater than 20 transfusions; the presence of abnormal clones; and prior androgen therapy (perhaps a measure of waiting too long). T-cell mosaicism is no longer considered a risk factor, since nearly all patients are now engrafting

with fludarabine. The likelihood that several of these risk factors will develop with increased age has prompted transplanters in Minnesota to urge patients to undergo transplant earlier.

Between April 1999 and July 2003, 42 FA patients received unrelated donor stem cells at the University of Minnesota. Thirty-seven received bone marrow; 6 cord blood. The transplant regimen consisted of total body irradiation, fludarabine, cyclophosphamide and anti-thymocyte globulin. Bone marrow was T-cell depleted. All but one patient engrafted. Incidences of grade 2-4 acute GVHD and chronic GVHD were 19% and 16%, respectively.

Minnesota is beginning to enroll high-risk patients in experimental protocols (*e.g.*, new preparative therapies without irradiation and infusion of T-cells containing a "suicide gene" so that T-cells can be destroyed if they cause GVHD), in the hopes of improving present survival rates. ♦

## Radiation Eliminated in HLA-Matched Related Donor Transplants: Results Outstanding

Hoping to reduce complications such as later malignancy, chronic graft-versus-host disease (GVHD), hormonal disorders, and infertility in post-transplant FA patients with matched related donors, transplanters in Minnesota have eliminated irradiation from the preparatory protocol. The new regimen uses fludarabine, cyclophosphamide and anti-thymocyte globulin followed by the infusion of T-cell depleted bone marrow or umbilical cord blood which has not been T-cell depleted.

Between April 2000 and February 2003, 11 patients underwent transplant using this regimen. All patients engrafted; none experienced severe toxicity. No patient developed GVHD. One patient with myelodysplastic syndrome relapsed, and one patient with a maternal donor rejected the graft; both were successfully transplanted a second time. With a median follow-up of 15 months, the probability of survival at 2 years post-transplant is 100%.

These phenomenal results suggest that irradiation can be eliminated safely from the pre-transplant protocol in FA patients with matched related donors. ♦

## Memorial Sloan-Kettering Achieves Exceptional Results with High Risk Transplants

Anders Kolb, MD, Memorial Sloan-Kettering, discussed transplant outcomes at his center from 1998 to the present. Eleven FA patients, ten of whom Kolb considered high risk according to standards developed in Minnesota, received peripheral blood stem cells (8 patients) or bone marrow (3 patients) from unrelated (6 patients) or mismatched related (5 patients) donors. Ages ranged from 5 to 24 years. Four patients had aplastic anemia, 5 had advanced forms of myelodysplasia, and 2 patients had AML. Nine had been multiply transfused, 8 had been on androgens and 7 had a significant history of infections.

All patients engrafted. Of the 11 patients, 9 (80%) are currently alive and disease-free, 6 months to 5 years post-transplant. (One of these patients relapsed with AML, was

given a second transplant, and is now alive disease-free). No patient experienced GVHD. The two patients who died were being treated for reactive airway disease prior to transplant and succumbed to lung infection post-transplant.

Kolb believes that several factors contribute to these outstanding results. He praises Farid Boulad, MD, head of the transplant program, and the entire transplant team for the intense and skilled care they provide. All grafts were aggressively T-cell depleted. This center prefers peripheral blood stem cells to bone marrow because the patient can be given significantly larger doses of stem cells (2 to 20 fold higher). Memorial Sloan-Kettering uses FK506 instead of cyclosporine, finding that this drug is highly effective in reducing GVHD and graft rejection and leads



*Anders Kolb, MD*

to fewer long-term complications.

These numbers are still extremely small. Nonetheless, the impressive results suggest that the protocol used at Memorial Sloan-Kettering may offer a good outcome for high-risk FA patients in need of transplant. Kolb also believes that going to transplant earlier would improve these results. ♦

## The FA Pathway Described

Akiko Shimamura, MD, Dana Farber Cancer Institute, Boston, discussed the FA protein pathway and its implications for rapid diagnosis. Each FA gene encodes (or provides the directions to make) an FA protein. There are at least eight different FA genes identified, each designated by a different letter of the alphabet. A group of FA proteins, including A, B, C, E, F, and G, is required to activate a “downstream” FA protein, D2. If any one of these upstream proteins is not present or not functioning correctly, D2 is not activated.

It is now possible to use a simple molecular test to determine if D2 has been activated. Activation of D2 suggests that all the upstream proteins

are functioning correctly, and that an individual does not have FA. A very small number of FA patients fail to produce a protein downstream of D2 and would not be picked up by this test. FA patients with lymphocyte mosaicism might also be missed; in such patients, D2 testing on fibroblasts would be necessary.

This new technology can be used to screen large groups of people for FA and can separate FA from other very similar chromosome fragility syndromes. It can also be used, along with other technology, to place FA patients in their appropriate complementation group. This D2 test is not yet clinically approved, so the clinical standard currently remains the chromosomal breakage test.



*Akiko Shimamura, MD*

Studies of the FA pathway have led to novel insights into cancer development. These discoveries stand to benefit both FA and non-FA patients. Thus, research into a rare disease has turned out to have broad implications for our general understanding of cancer. ♦

## Alter Helps Families Understand FA, Hematology, and Incidence of Cancer in FA Patients

Blanche Alter, MD, MPH, National Cancer Institute, presented information on three important topics at the August, 2003 Family Meeting. For families new to FA, Alter gave a comprehensive overview of this disease (see *Science Letter*, "FA 101"). Responding to the need of many families to understand the basics of hematology, Alter explained important terms, the function and characteristics of different blood cells, and gave guidance on monitoring the bone marrow (see *Science Letter*, "Hematology 101"). Alter also addressed the troublesome subject of solid tumors in FA patients and gave specific recommendations on monitoring and screening for these tumors (see *Science Letter*, "Cancer Epidemiology").

Families attending Camp Sunshine greatly appreciated Alter's willingness, once again, to stay for the duration of the Camp, answer questions, and provide consultation to patients and parents alike. Expert (and free!) consultation is a huge benefit of attending the summer Family Meeting; our heartfelt thanks, Dr. Alter! ♦

### Use of Logo

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

## Cincinnati's Matched Sibling Donor Transplant Results

### Low Dose Irradiation Included in Protocol

Richard Harris, MD, Cincinnati, reported on 34 FA patients with matched sibling donors transplanted at his center over the past 15 years. Twenty-nine of these 34 patients (85%) survive, from 3 months to 15.6 years post-transplant. His protocol includes low dose thoraco-abdominal irradiation, low dose cyclophosphamide, anti-thymocyte globulin and cyclosporine post-transplant. Bone marrow grafts were not T-cell depleted.

Organ toxicity and infectious complications from the preparative therapy were minimal. Three patients developed Grades I-II graft-versus-host disease (GVHD); two experienced chronic GVHD.

Causes of death included lack of engraftment and infection (one patient); pneumonia (one patient), and multi-organ failure (one patient).



*Fabrizio DiMercurio with Richard Harris, MD*

Two patients developed solid tumor malignancies 6 and 15 years post-transplant; one died of this complication. One patient relapsed with leukemia that was present prior to transplant; another died of AML 12 years post-transplant. This last patient had a pre-transplant monosomy 7 clone, but without overt MDS or leukemia. ♦

## New Strategies for Treating FA Patients with Leukemia and Advanced Myelodysplastic Syndrome (MDS)

Cincinnati Children's Hospital and the University of Minnesota Transplant Program have developed new strategies in the hope of improving survival for FA patients with leukemia or advanced myelodysplastic syndrome (MDS). Traditionally, survival rates for this population have been poor.

Cincinnati first administers a mild course of chemotherapy to get patients into remission. Two to three weeks later, patients begin preparative therapy for a bone marrow transplant. Four patients have recently been enrolled in this protocol. The

chemotherapy was well tolerated by all patients; three patients are presently alive and disease-free from one month to five months post-transplant.

Minnesota has replaced total body irradiation with busulfan in its pre-transplant protocol. Two patients enrolled in this protocol are doing well; the time post-transplant is one month and seven months. In both centers, the numbers are too small and the time post-transplant insufficient to predict effectiveness of the new protocols. ♦

## Prevention, Surveillance, and Treatment of Head and Neck Cancer

The difficult subject of managing head and neck cancers in the FA population was the topic of a presentation by Bhuvanesh Singh, MD, Memorial Sloan-Kettering Cancer Center. Singh stated that FA patients are 500 times more likely to develop these cancers than those in the general population; by age 40, the incidence is 21%. The site of these cancers is primarily the oral cavity, but can range from the mouth through the esophagus.

Singh stressed the importance of prevention. FA patients should abstain from the use of tobacco and alcohol; avoid second-hand smoke (no smoking in the house or car!); avoid mouthwashes that contain

alcohol; maintain good oral hygiene; and follow a regimen of aggressive monitoring.

Routine screening should begin at ages 10-12 and should be done twice a year. A physician with experience in treating head and neck cancers, not the patient or parents, should do the monitoring. The mouth is the key area, but the physician also needs to monitor the throat by inserting a fiber optic instrument through the nose down to the voice box.

Any suspicious areas should be biopsied. Precancerous lesions should be removed, if possible. With the discovery of a precancerous lesion, monitoring should increase to every 2-3 months.



*Bhuvanesh Singh, MD*

If cancer is diagnosed, treatment should occur at a major center with experience in mouth cancers. Surgical removal is the best treatment option. FA patients are very sensitive to chemotherapy and radiation; side effects of these therapies can be extremely harsh and need to be evaluated carefully.

After a cancerous lesion is removed, there is a high risk of a second malignancy, often in the same area. Aggressive monitoring is an absolute must and should be performed by the surgeon removing the lesion.

Head and neck cancer in the FA population is different than in the general population. Researchers are beginning to study the genetic changes that take place in FA patients, so that these cancers can be appropriately targeted. ♦



*Scott Kozin, MD, discusses the chapter on hand and arm anomalies.*

## Physicians Update Standards for Clinical Care

Twenty-eight physicians expert in the care of Fanconi anemia patients volunteered their time to meet for three days in Chicago in March to update the standards for clinical care for FA patients. The results of their work will be published in a second edition of the handbook *Fanconi Anemia: Standards for Clinical Care*. Eva Guinan, MD, Dana-Farber Cancer Institute, did a superb job moderating this conference, as she did in 1998 at the consensus conference for the first edition of the handbook.

We'd like to express our profound gratitude to all the physicians who participated in developing the consensus standards for the handbook. ♦

## Managing Arm and Hand Abnormalities

Scott H. Kozin, MD, Shriners Hospital for Children, Philadelphia, described the arm and thumb anomalies that affect many FA patients and discussed therapies that can improve the function of the hands and arms. See "Hand and Arm Differences in FA," *Science Letter*.

## An Update on Preimplantation Genetic Diagnosis

Preimplantation Genetic Diagnosis (PGD) provides an option to couples who wish to conceive a healthy child who is perhaps also an HLA match for an FA sibling, according to Mark Hughes, MD, Wayne State University. PGD makes it possible to diagnose a genetic disease and HLA status *before* pregnancy occurs.

A mother's ovaries are first stimulated to produce multiple eggs, which are removed and fertilized by the father's sperm *in vitro*. When the pre-embryo (blastomere) reaches eight cells, one cell is removed and tested for disease and/or HLA status. A blastomere free from disease (and

often, chosen because there is an HLA match with an FA-affected child) is implanted in the mother. If all goes well, a healthy baby is born nine months later.

For this to succeed, parents must be highly motivated. A family's specific gene mutations or appropriate genetic markers must be known. The family needs to work with a local reproductive center as well as experts in PGD. The costs can be extremely high and vary greatly from one reproductive center to another. Some insurance companies will cover the costs, others will not. (Editors' note: One family from New York stated that each attempt cost them almost \$20,000).

Each blastomere has a 3/4 chance of being disease-free, and a 1/4 chance of being an HLA match for an affected child, so there is a 3/16 chance that any one blastomere will



Mark Hughes, MD

be disease-free and an HLA match. Hughes does not recommend this technology for couples nearing age 40. The average number of attempts needed to achieve a desired pregnancy is 2.8 if HLA matching is included. One FA family attempted PGD 9 times and did not achieve a pregnancy. For families who have succeeded, the benefits have been enormous. ♦

## Management of Patients with Fanconi Anemia

Parents faced with providing the best possible care for their children with FA are often overwhelmed. In response, Margaret MacMillan, MD, Blood and Marrow Transplantation Program and Stem Cell Institute, University of Minnesota, made a comprehensive presentation at the FA Family Meeting on the management of patients with FA (see *Science Letter*, "Management of Patients with Fanconi Anemia"). MacMillan documented the steps to take, on an individualized basis, to manage the various aspects of Fanconi anemia, including diagnosis, organ dysfunction, feeding problems, immunizations, endocrine issues, bone marrow failure, reproductive issues, and malignancies. After her presentation, Dr. MacMillan, along with Blanche Alter, MD, and Richard Harris, MD, answered questions from the participants in a panel discussion. ♦

## Clinical Trial Targets Patients with Premalignant Lesions of the Oral Cavity and Pharynx

Researchers at the University of Texas M.D. Anderson Cancer Center have opened a clinical trial to study the effectiveness of gene therapy in preventing cancer in patients who have premalignancies of the oral cavity or pharynx. This trial uses an adenoviral vector carrying the tumor suppressor gene p53 into premalignant tissues, hoping to create an anti-cancer effect. The product administered is called Advexin. Advexin, will be delivered both as an intramuscular injection (on day one) and as a mouthwash (days 1-5) every 28 days for 6 courses.

Inclusion criteria include adequate bone marrow function (absolute neutrophil count of 2,000; platelet count of 100,000); adequate liver function (bilirubin less than or equal to 1.0 mg/dl); and adequate renal function (creatinine less than or equal to 1.5 mg/dl).

Contact Shirley Taylor at 713-745-1773 or e-mail her at [sataylor@mdanderson.org](mailto:sataylor@mdanderson.org) for more information or to enroll in this trial. The web page <http://clinicaltrials.gov/ct/show/NCT00064103?order=2> has additional information. ♦

## Regional Meeting for Adult FA Patients

Seven adults and one teenager with Fanconi anemia, many with friends or family members, gathered in Herndon, VA, on June 7 for the first Regional Meeting specifically for adults. They came from as far away as California and Canada, and many were able to attend through the scholarship program provided by the Fund. For some, it was “old home week,” as adult patients reunited with FA friends they have communicated with by e-mail or phone or met at past FA Family Meetings.

Presentations were made by Blanche Alter, MD, MPH, National Cancer Institute, (Cancer Epidemiology and Fanconi Anemia); Susan Rose, MD, Cincinnati Children’s Hospital (Endocrine Issues); Pam Stratton, MD, NIH, (Gynecological Issues); and Carter van Waes, MD, PhD, NIH, (Squamous Cell Carcinoma of the Head and Neck). The speakers provided ample time for



*Blanche Alter, MD, speaks with FA adults about cancer epidemiology.*

questions on the issues that are of such importance to adult FA patients.

The participants had time, after the medical presentations, to debrief with Nancy Cincotta, MSW, Mount Sinai Hospital, New York. Nancy also facilitated a session on coping with FA. Four of the seven adult FA patients, who ranged in age from twenty-one to forty-nine, have

survived one or more episodes of head and neck and/or gynecological cancer. All remain vigilant for the possibility of cancer. The participants ran the gamut from a very active college student and a teacher to FA adults who are parents. They all very much valued this time to talk with others who are facing the same challenges. ♦

### 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.*

## German Transplanter Presents Results, Discusses Complications of FA Transplants

Wolfram Ebell, MD, Humboldt University, Berlin, presented data on 22 patients recently transplanted in Germany. Hoping to decrease the later incidence of solid tumors, Ebell does not use irradiation, but rather a protocol consisting of fludarabine, busulfan, and pre/post immunosuppression. He does not T-cell deplete bone marrow or peripheral blood.

Stem cell donors were as follows: matched sibling (8); mismatched relative (1); and unrelated donor (13). Overall survival was 64%. Of patients receiving unrelated donor stem cells, 61% survive.

Ebell noted the following trends: patients with aplastic anemia only

did better than those with myelodysplasia; those with leukemia did the worst of all. Patients under the age of 10 did better than those over 10. Prior androgen therapy was a risk factor for survival, perhaps because these patients are older. Transfusion history was not a factor in overall survival.

Ebell noted that the biggest problem post-transplant was infection, primarily viral. Seventy-five percent of those receiving unrelated donor marrow had one viral infection; 50% had two separate viral infections. Finding a better way to manage viral infections is key to improving transplant outcomes. ♦

## FA Regional Meeting in Minneapolis

FA parents from the United States and Canada converged on Minneapolis on Saturday, May 17, to attend the FA Regional Meeting held at the University of Minnesota Blood and Marrow Transplantation Program and Stem Cell Institute. Twenty FA families were represented at the meeting, for a total attendance of forty.

John Wagner, MD, made arrangements for a tour of the National Marrow Donor Program and the Ronald McDonald House for those FA parents who arrived a day earlier. Both tours were very helpful, especially for those pre-transplant families in attendance.

Wagner developed an excellent program for the meeting, which included presentations by Betsy



*John Wagner, MD, presents to FA parents.*

Hirsch, MD (Interpreting the Cytogenetic Report); Phuong Nguyen, MD (The Marrow Aspirate in FA); Christine Ternand, MD (Diabetes

and Growth Failure); Sarah Jane Schwarzenberg, MD (Feeding Intolerance); Frank Ondrey, MD (Head and Neck Cancer); Margaret MacMillan, MD (Unrelated Donor Bone Marrow Transplantation); and John Wagner, MD (Pre-Implantation Genetic Diagnosis). Janet Ziegler, MSW, Stacy Stickney Ferguson, MSW, and Julia Copeland, MSW, made presentations on dealing with chronic illness. A panel of post-transplant FA families shared their transplant journeys. Families were awed by the quality of the presentations, as well as by the extra effort that the Minnesota staff made on their behalf.

## Head and Neck Cancer Prevention Trial Opens for Patients

Researchers at the Dana-Farber Cancer Center and the National Cancer Institute have opened a clinical trial to test the effectiveness of celecoxib in preventing cancer in patients who have head and neck precancerous lesions. Patients will undergo a biopsy to confirm a premalignant condition, then will receive oral celecoxib twice daily for 3 months. After 3 months, patients will undergo a repeat biopsy. Patients with a positive response will receive celecoxib for an additional 9 months. Patients are followed every 3-6 months for a year.

Inclusion criteria include adequate hematopoietic function (platelet count of at least 100,000 and absolute neutrophil count greater than 1,500), and adequate liver and kidney function.

This is a multi-center study, occurring at three sites in Boston. Location and contact information are as follows:

- Brigham and Women's Hospital, Boston  
James Mueller, MD: 617-732-5984
- Dana-Farber Cancer Institute, Boston  
Lori J. Wirth, MD: 617-632-3090  
E-mail address: [Lori\\_\\_Wirth@dfci.harvard.edu](mailto:Lori__Wirth@dfci.harvard.edu)
- Massachusetts General Hospital Cancer Center, Boston  
John Ross Clark, MD: 617-726-8748  
E-mail address: [irclark@partners.org](mailto:irclark@partners.org)

Through the sponsorship of Fairview Health Services, participants were treated that evening to an excellent dinner at the Science Museum of Minnesota as well as to a presentation by Wagner on the future of stem cell therapy. Families also had a chance to tour the exhibits at the Museum, which included an excellent exhibit by Wagner on Fanconi anemia. This relaxing evening was an excellent end to a wonderful day. We extend our sincere thanks to Dr. Wagner and his colleagues for this outstanding program. ♦

## Head and Neck Cancer in FA Patients

At the Adult Regional Meeting in Herndon, VA, Carter van Waes, MD, Chief, Head and Neck Surgery, National Institute on Deafness and Other Communication Disorders (NIDCR), discussed the importance of early detection of signs of head and neck cancer in FA patients. Because FA patients are at such extremely high risk for oral cancer, he brought with him a pamphlet entitled *Detecting Oral Cancer: A Guide for Health Care Professionals* published by the National Institute of Dental and Craniofacial Research, NIH, which may be helpful to FA patients in doing self-exams. This booklet can be viewed on the NIDCR website at

<http://www.nohic.nidcr.nih.gov/pubs/detect/pages/contents.html>. If you do not have a computer and would like a hard copy, contact Suzanne Lauck, RN, at the Fanconi Anemia Research Fund.

Although the exact mechanism by which FA patients are at such high risk for head and neck cancer is not yet known, van Waes suggested FA patients may be able to reduce their risk by minimizing diagnostic radiation such as routine dental x-rays or CT scans and by minimizing dental trauma, such as biting, hard brushing, ill-fitting braces or dentures. He noted that, in the general population as well as in the FA population,



*Carter van Waes, MD, speaks to FA adults about head and neck cancer.*

## Hormones and Fanconi Anemia

The identification and treatment of endocrine problems are crucial to the health and development of FA patients according to Susan Rose, Professor of Endocrinology, Cincinnati Children's Hospital Medical Center. Endocrinology is the study of how hormone messages interact in the body. Hormones affect growth, development, and general health.

During the past 18 months, physicians at Cincinnati Children's Hospital Medical Center have evaluated 25 children with FA. Short stature was identified in 64%.

Of this small group of children, nearly every child had an endocrine deficiency. Seventy-eight percent had glucose intolerance or blood sugar high enough to be considered diabetic. Abnormal thyroid function was found in 60%; 47% had low growth hormone levels. Puberty started at a normal age, but sometimes occurred at a very short height. In 76% of boys, testes were small for age.



*Susan Rose, MD*

Rose emphasized that thyroid hormone and insulin are important in childhood growth and health. FA children should undergo a comprehensive baseline endocrine evaluation, with follow-up testing annually, when appropriate. Early administration of thyroid hormone or insulin therapy could optimize the patient's growth potential. ♦

alcohol and tobacco are well-established causes of squamous cell carcinoma and must be avoided.

[Editor's Note: A mouthwash that does not contain alcohol is *Tom's of Maine*, which is available in many supermarkets and health food stores. You can learn more about the product and where to purchase it at [www.tomsofmaine.com](http://www.tomsofmaine.com).]

Van Waes noted that signs and symptoms of head and neck cancer include the following:

### Mouth

- leukoplakia (a white patch in the mouth or throat that does not scrape off)
- redness in the mouth or throat
- a painless or painful ulcer, sore or raised area that has lasted for over two weeks
- loose teeth
- bleeding or pain

### Throat (oropharynx and esophagus)

- difficult or painful swallowing (including pills sticking when trying to swallow)
- foreign body sensation in the throat

*continued on page 18*

## My First Impression of the Fanconi Anemia Family Meeting

by Lim Poh Seng, Singapore

I had traveled alone 10,500 miles from the sunny island republic of Singapore to Camp Sunshine in Casco, Maine, to attend the Fanconi Anemia Family Meeting with the encouragement from my wife. My mission was to find out as much as possible about this terrible disease which afflicts our second son, Samuel, aged five. Before his diagnosis, to tell the truth, I was not aware of this disease. This trip proved to be an experience that I'll never forget.

I was made to feel very welcome at the FA Family Meeting at Camp Sunshine. The staff and volunteers were very friendly and helpful. Many times they went out of their way to make me feel comfortable, and this certainly made my stay at the camp very enjoyable.

The whole Camp felt like family.

Everyone seemed to know everyone else, and there was no barrier or superficiality. Some families, such as the Frohnmayers, had attended every single FA Family Meeting since their inception. Each and every one of the FA families there had been touched by the disease in one way or other. Some families had tragic stories to tell. Even though my own family suffered from having a son with FA, I could tell that some families had suffered more grievously than mine. We all looked forward to the scientific advances that would help those who suffer from this terrible disease have a better chance of survival and a better quality of life.

The medical and scientific presentations by the various physicians and researchers were well-delivered. The Q&A format at the end of each



*Lim Seng asks a question regarding a medical presentation.*

presentation gave the participants a chance to query the physicians and researchers in order to understand the issues better. But I was most struck by how approachable the physicians and researchers were. Here at the camp, they mingled freely with the FA families, and there did not appear to be any condescension or

*continued on page 20*

## Newly Diagnosed Adult FA Patient

by Maria Godwin, St. Mary's, GA

I am thirty-three years old and live in St. Mary's, Georgia, with my husband Josh. After undergoing surgery and radiation treatment for squamous cell carcinoma of the head and neck, I was just recently diagnosed with Fanconi anemia. In fact, I received my diagnosis just days before the Fanconi Anemia Research Fund's Adult FA Patient Regional Meeting in June 2003. I went to the Adult Meeting with a lot of anxiety and questions regarding my illness.

Upon completing the meeting, I was not only very overwhelmed, but I had a better understanding of what

FA is and how it can affect me as an adult. The doctors and researchers were great on keeping up with all the new facts.

I also recently had the opportunity to attend Camp Sunshine in Maine for FA patients and their families. At first I was reluctant to go, and my first few days there were the most difficult of my life. However, after getting the chance to be with the kids and their families and hear what they have gone through and what lies ahead on their journey to cure FA, I realized that Camp Sunshine gave me another FAMILY—a



*Maria and Josh Godwin*

family that I now treasure with all my heart. Thank you, Camp Sunshine, and thank you to all the researchers who help us to LIVE! ♦

# Sylvie's Transplant Journey

by Michelle Lauzier, Thunder Bay, Ontario

September 7, 1996 brought us many joys! Our beautiful daughter, Sylvie, was born in Timmins, Ontario, weighing 5 lbs., 4 oz. A few hours after delivery, our happiness was shattered. We were informed that Sylvie was missing both thumbs. Later that evening, a pediatrician informed us that she suspected FA. Finally, in December, Sylvie's diagnosis was confirmed. It was hard to understand, since she was a very good and healthy baby.

Sylvie's blood counts were terrific at birth. As the years came, we gradually saw these counts decrease. In February 2002, Sylvie received her first red blood cell transfusion. At our yearly visit to the Hospital for Sick Children in Toronto, we were faced with a very important decision: 1) try androgen therapy; 2) rely on transfusions; or 3) go to transplant. At that point, we were reminded of the heated issue from the previous summer's FA Family Meeting: Bring your children to transplant early, BEFORE androgen therapy! For the past six years, we had been told that transplant was our last option since we had no matched sibling donor but, in the last six months, recommendations had changed. We knew we had to take her to transplant, but I had concerns regarding the suggested protocol and the lack of FA experience of our transplant center.

On July 18, 2002, Sylvie and I embarked on our Transplant Journey. Sylvie was admitted to the hospital, where she underwent a series of tests and scans and then began the transplant preparative therapy, which was a 10-day protocol consisting of fludarabine, cyclophosphamide, Mesna and ATGAM. No radiation was used in the protocol. On August 2, Sylvie received her gift of life, an unrelated 6/6 marrow that was not T-cell

depleted. She experienced the usual side effects: loss of hair, nausea, vomiting, high blood pressure, and mucositis. For days, she wouldn't eat, drink or talk because of the mouth sores. On day +10, her marrow engrafted. On day +20, we took her home to the Ronald McDonald House. The transplant had gone much smoother than I had expected. Things were going well. On day +28, the prednisone was discontinued. It was wonderful! We were enjoying the last few days of the summer.

Sylvie had to be re-admitted on day +31 due to morning low-grade fevers and complaints of a sore belly. She had an ultrasound, a chest X-ray, a CT scan, which were all inconclusive. She was discharged on day +36, on her 6th birthday. We had a little party both at the hospital and back at the Ronald McDonald House. Unfortunately, she was readmitted the next morning. The fever was back! After more testing, the doctors still couldn't pinpoint any problems. She was discharged again on day +41. The next day, her EBV (Epstein-Barr virus) levels were reported to be slightly elevated and she was treated with Acyclovir. By day +54, the levels were still elevated so the treatment was changed to Gancyclovir and Cytogam (IV infusions). At the first infusion, Sylvie's temperature rose and she had to be re-admitted to the hospital once again.

This time, she really took a downfall. The fevers were now morning and evening, but fine in the afternoon and through the night. She developed a more pronounced rash. Once again, she had ultrasounds, x-rays, CT-scan, MRI, lumbar puncture, sinus wash; nothing was revealed. She stopped eating and drinking and had to be put back on



Top to bottom: Michelle, Pierre and Sylvie Lauzier

TPN. Sylvie was emotionally drained; her smile disappeared! We got really worried. The doctors finally thought that it might be GVHD, not knowing what else could be the cause. She was put on a treating dose of prednisone. Within a week she was finally doing better. Sylvie was discharged on day +80. It had been a very long 4 weeks.

Sylvie was finally doing well. She gained approximately 15 lbs within three weeks because of the prednisone. On November 15, day +105, we returned home to Thunder Bay. We left behind many transplant friends who were a great support to us. Life would have been unbearable without them. The night before, Sylvie took the time to prepare her brother Pierre. She told him that he would not recognize her since she had no hair and was very big, but that inside she was still the same little girl. Our journey away from home was finally over!

Coming home was a lot more difficult than I anticipated. Sylvie required a lot of my time. She had 25 doses of meds to take every day. That

*continued on page 13*

# Our Bone Marrow Transplant Experience

by Mike Vangel, Hingham, MA

On September 6, 2002 Amy received her bone marrow transplant from an unrelated, mismatched, anonymous donor. In her act of unselfish kindness, the donor has given our daughter the gift of life. We are very thankful to her and to the Pediatric Bone Marrow Transplant team at Fairview University Medical Center in Minnesota. We would not be here without them.

For us, Amy's bone marrow transplant experience started long before September 6, 2002 and, in many ways, will extend well beyond one year later. As Amy's blood counts continued to fail, we became progressively more concerned. Although the bone marrow registry has over 4.5 million potential donors, none had proved a "perfect HLA match" for Amy.

We didn't consider ourselves viable candidates for Preimplantation Genetic Diagnosis (PGD). In the best-case scenario PGD would involve at least 11 months—and that was assuming success after the first cycle. We knew of several couples that had undergone many cycles without success.

As Amy's platelet counts continued to decline (to the point where they were consistently below 20,000), we went to Fairview University Medical Center in March of 2002 for a consultation. We were quite surprised when Dr. (Margaret) MacMillan stated that, for the best possible outcome, Amy should go to transplant within three to six months—and preferably closer to three months.

We knew the inherent risks associated with an unrelated, mismatched transplant for an FA patient. Although the preparative regimen had been improved greatly with the introduction of the drug,



*Amy Vangel at the "Hole In The Wall Gang" camp in Ashford, CT, July 2003, almost one year after undergoing an unrelated mismatched 5/6 bone marrow transplant.*

fludarabine, and the reduction in the amount of radiation administered, it was still a risky, potentially life-threatening proposition. In general, the children who were the healthiest and strongest going into transplant seemed to fare the best.

Amy had been receiving injections of G-CSF and Epogen, with a relatively good response, every other day for the past seven years to stimulate her blood counts and had been transfusion and androgen-free. However, the cytokines were never meant for chronic use and eventually her "turbo-charged" marrow would fail.

As parents, we had to make our decision about whether or not to go to transplant based on the best information available to us at the time. If the strongest children had the best outcomes for unrelated mismatched transplants, would we be lessening her chances of successful outcome by placing her on androgens, hoping to buy more time to get a better match?

Or hope for success in pursuing PGD? We had to consider whether we would be putting her at greater risk by allowing her immune system to deteriorate further as the disease ultimately progressed. Either way, we could not procrastinate. We decided to proceed to transplant with the hope that a suitable HLA match would be found.

We have seen firsthand that each family finds a way to deal with the enormity of a bone marrow transplant. There are no "right" answers, but simply the best choices you can make at the time. Somehow we found the strength to do what we needed to do.

The first milestone for us was the period prior to transplant when we were mentally, emotionally, and financially preparing for transplant. Then we arrived in Minnesota for the two-week period of preliminary work-up, which was followed by

*continued on page 18*

## The Ardeshir Family from India Attends the Family Meeting

by Marzban Ardeshir, Bombay, India

I, along with my wife Daisy and daughter Imroze, attended Camp Sunshine this year. This was my second time at Camp Sunshine, the first being soon after Imroze's transplant in 1997. Imroze and I had good memories of Camp, as Imroze was most pampered by her volunteers and I had the first time experience of such facility.

The Camp is now at a very picturesque site, and the amenities are excellent. Arrangements were made for us to be picked up. Since we came all the way from India, we were tired after nearly 36 hours of airport waits and air travel. Our volunteer, Walter, was so good as to notice that, so he called the Camp to keep our dinner, as we would be arriving after dinner time. We were well received at the Camp, and after dinner we discovered that our luggage had found its way to our room. This surely did surpass the five star treatment of any

American hotel. Thanks again to the efforts of dear Walter. Our room was spacious and well equipped. The climate kept us cozy as we, being from a tropical country, cannot tolerate the cold.

The lectures by the distinguished and eminent medical personalities were engrossing, informative and very educational. We obtained a lot of knowledge for our daughter Imroze, especially the "do's and don'ts" of which we were much in the dark. We could interact with the faculties freely and, as Mr. Dave Frohnmayer put it, "no question is a silly one." We learnt much from the questions asked. The hot and regular replenishing of coffee kept me alert.

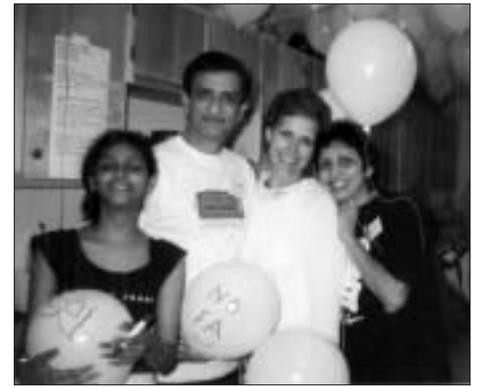
Imroze was very excited about the activities of the Camp. She enjoyed the company of the other children, and we found it a Herculean task to get her to bed. All children seemed to be having a roaring time. The

### Sylvie's Transplant Journey

*continued from page 11*

was a huge challenge. I also had to monitor her blood pressure, bathe her, apply cream which she hated, care for her central line, feed her, monitor her food and fluid intake, watch for signs of GVHD, and run to the clinic. Pierre, who hadn't seen me in four months, was very demanding. Lots of temper tantrums! I tried to give him as much attention as possible, but it was never enough to satisfy him. My husband, Marc, works away from home so I was on my own a lot. Little tasks required so much energy! Sylvie was in isolation so we couldn't visit anyone. We had very few visitors at the house. I had

cabin fever. We have no family in Thunder Bay; therefore, I had very little physical support. We have friends, but they all work full-time and have children with busy schedules. When Marc came home, I would crash down. This went on and on for months. I was frustrated with myself because I had a hard time. I was supposed to be Super Mom and handle everything. In the spring, Marc got a promotion at work. I should have been thrilled. Instead, I was jealous because his career was flourishing and I had a hard time holding my life together. I was angry with myself for feeling this way. Sylvie was doing well—I should have been happy. She didn't require as much time anymore. Why was it



*The Ardeshir Family with Suzanne Lauck of the Fund*

balloon ceremony, the boat floats, the Karaoke, the talent show, all lent finesse to the Camp arrangements. We made many new friends too.

Our special thanks to Laura and Suzanne, who made our arrangements for the Camp and answered all our e-mails patiently. Our thanks also go out to the other staff of Camp Sunshine, who worked unseen. The highlight of Camp Sunshine was the dedication of the volunteers and staff. God bless them all. ♦

taking me so long to recuperate? It was really rough!

Finally, in July, I decided that the kids and I would go camping. We packed up the trailer and headed out. A change of scenery! That was the best thing I could have done! I managed to find time for myself and enjoy time with my precious little ones. We went swimming, biking, and walking. We played ball, went to the park, and sang by the fire. The tranquility of the forest led me on a road to recovery. I still have the odd days where life is more difficult, but I manage to survive. As Sylvie would sing: "The sun will come out, tomorrow!" ♦

## Fun, Science, and Optimism at the Annual Family Meeting

*continued from page 1*

parents who are dealing with the same difficult disease.

Parents spent most of their time attending the medical and scientific presentations and meeting with Nancy Cincotta, MSW, who held a number of much-valued sessions on coping with FA. Once again, we all were honored by the presence at the meeting of the physicians who gave up their weekends to travel to Maine to meet with us. The presentations are summarized in the *Medical News* section of this newsletter. The complete text of the presentations can be found in the *Science Letter*. Families were overwhelmed by the amount of medical and scientific information they received and were extremely appreciative of the generosity shown by these physicians in taking precious time from their very busy schedules to meet with them. Parents were also buoyed by progress that has been made in the clinical care of FA patients and, particularly, in the results of bone marrow transplantation for FA patients.



*The Gleason kids are welcomed through the stairstep doors of Camp Sunshine.*

Parents also had fun! Camp Sunshine prepared a wonderful banquet for the adults on Saturday night, complete with tablecloths, candlelight, and prime rib! After the delicious meal, parents—with no urging at all—showed off their impressive karaoke talents. Whether they could carry a tune or not, all shone in their time in the spotlight, and everyone had a wonderful time.

Most notably, this year's meeting had a definite "different" feel to it. Despite wanting to reunite with other families, FA families who regularly attend the annual meeting are sometimes apprehensive because the treatment options for FA patients have not always seemed to progress as rapidly as hoped. Parents often have departed at the end of the Family Meeting quite concerned by medical news they've learned at the meeting. Adding to that, patients who have had successful bone marrow transplants in the past often stopped attending subsequent Family Meetings, so most attendees at the Family Meeting were families whose youngsters had no bone marrow donors or who were still awaiting transplant.

In large part due to the growing recognition by FA parents that FA does not stop at the conclusion of a successful transplant, this year saw the return of four children who have survived transplants in the last year at three separate hospitals. To everyone's great delight, all are doing well! Seeing kids who looked pretty sick last year come through the welcoming



*FA families listen to medical presentations.*

*continued on next page*

doors of Camp Sunshine with a healthy color to their cheeks, big smiles, and energy to burn was a treat for all and a source of positive energy for the entire meeting.

Parents this year heard a wonderful panel presentation by five FA teens who have attended Camp together for years. These remarkable and articulate teens assured their parents that they did not blame them for their FA. Most had come to the conclusion that having FA has been a positive experience for them, even a “gift.” The wisdom and strength of these teens put the final touches on a wonderful Family Meeting! ♦



*FA parents enjoying banquet dinner*



*Kids dressed up for Masquerade Ball*



*Amy Frohnmayer and Amanda Gleason at Camp*



*Camping out at Camp Sunshine*

## Camp Sunshine Volunteers' Expressions of Gratitude

by Sadie P. Hutson, Jamie Rubin, and Tracy Downing

I don't think any of us will forget volunteering at Camp Sunshine in August 2003 during a week devoted to children with Fanconi anemia and their families. The adventure began with a meeting of approximately seventy-five amazing volunteers from varied backgrounds to discuss camp policies, routines, and responsibilities. What no one told us was how much we would learn from the FA families and how the experience would change us forever.

The three of us came together, not having met one another, and joined forces with others to volunteer in the teen group at camp. From the very first activity, we watched kids who already knew each other rekindle friendships and simultaneously take in the newcomers with open arms. Parents did the same. As one camper described, "There is an unspoken friendship between each and every one of us here." This demonstration of genuine warmth was amazing to watch and truly indescribable. We all got the immediate

sense that this camp is able to provide an experience that all kids and families deserve, a place where they can celebrate life, have fun, and most importantly, just be themselves.

For those volunteers who are familiar with FA and the devastation it causes, it was incredible to watch families come together, especially given the rarity of this disease. It was enlightening to hear kids with FA tell us how having the disease has been a positive experience, something that they would not trade or wish upon anyone else. Listening to parents describe the impact of their child's diagnosis was also powerful. The week was full of times when we wanted to cry tears of sadness and joy all at once.

It is for all of these reasons and many more that we wanted to write to thank the families for the wonderful reward and privilege they have



*Volunteer Sadie Hutson with Ruthie Saunders from Israel.*

given us by allowing us to share in this special week at Camp Sunshine. You will forever remain in our hearts. Whether by boat, by balloon, or by the hand of a higher power, may all of your wishes be granted. ♦

## In Loving Memory

Jacob Ashurst  
7/16/01 – 6/13/03

Elma Dueck  
9/2/61 – 3/22/03

Cameron Goodrich  
10/3/95 – 3/9/03

Brandon Knupp  
7/27/91 – 8/30/03

Diamond Ashley Persson  
10/16/01 – 3/29/03

Salmaa Sajee  
4/24/95 – 10/19/02



## Fighting FA by Fundraising

by Fred Nunes, Cumberland, RI

Some time after the initial shock and denial wore off, I promised Duncan as he lay sleeping that I would try to do something every day to improve his life with FA. After attending the FA Regional Meeting in Minneapolis where we met several active FA families, I was inspired to take our first step in FA fundraising, sending a letter requesting donations for the Fund.

I set a deadline of July, the one-year anniversary of Duncan's diagnosis. In order to jumpstart the letter writing process, I asked Suzanne Lauck at the Fund for sample letters, which she quickly forwarded. Suzanne also offered the fundraising guide for FA families, which I gladly accepted. The sample letters and template in the fundraising guide were a great help. We told our story, but also stole some sentences directly from the sample letters in the guide.

It was emotionally difficult to write the letter, to try to convey what a serious disease this is, and to move people to make generous donations. We sent the letter with stamped FAREF-addressed envelopes to family, friends, neighbors, and co-workers. I thought that, once we sent the letter, it would be easy and the work would be done. But people not only sent donations, they also contacted us with letters offering support and offers to run additional fundraisers. The offers are heart-warming and gratifying, but the letters and conversations immerse me in the whole emotional whirlpool again. It is so intense. But our discomfort is a small price to pay for the hope of effective and more gentle treatments for our child.

It was difficult taking the step of asking for help, even though we were not asking for help which would directly benefit our family. But I made a promise to Duncan that I want to keep. I want to be able to say to Duncan that we did, and continue to do, everything we possibly can to make things better for him. ♦



Duncan Nunes

## Your FA Research Dollars at Work in 2003

During 2003, the Fanconi Anemia Research Fund has awarded \$838,336 in research grants to the following projects:

Investigator: Ruud Brakenhoff, PhD, Hans Joenje, PhD, and Vincent van Beusechem, PhD, Free University, Amsterdam

Title: *Development of Virotherapy Using Retargeted Adenoviruses to Eradicate Preneoplastic Lesions for the Oral Cavity and Oropharynx*

Amount: \$116,000

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

Title: *The Fanconi Anemia Transcriptome Consortium (addendum for genotyping)*

Amount: \$51,920

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

Title: *Non-Viral Gene Therapy for Fanconi Anemia*

Amount: \$213,017

Investigator: K. J. Patel, PhD, MRCP, MRC Laboratory of Molecular Biology, Gonville and Caius Colleges, Cambridge University, Cambridge, UK

Title: *Biochemical Analysis and Structural Determination of FA E and D2 Proteins*

Amount: \$88,172

Investigator: Inder Verma, PhD, Salk Institute for Biological Studies, La Jolla

Title: *Gene Therapy for Fanconi Anemia Using Lentiviral Vectors*

Amount: \$77,227

Investigator: Margaret MacMillan, MD, University of Minnesota, Minneapolis

Title: *Immune Reconstitution and Opportunistic Infections after Unrelated Donor Bone Marrow Transplantation*

Amount: \$90,000

Investigator: Weidong Wang, PhD, National Institute on Aging, NIH, Bethesda

Title: *Identify New FA Genes and Understand the Disease Mechanism through Protein Association*

Amount: \$192,000

## Our Bone Marrow Transplant Experience

*continued from page 12*

chemotherapy and radiation. Next came the transplant itself. As the engraftment for Amy became evident by day 10 or so, the ravages of the chemotherapy and radiation became more apparent too. The steroids administered to Amy made her face swell incredibly and gave her considerable mood swings. And, of course, her beautiful, thick blond hair by now was falling out by the handful.

After Day +21 some transplant patients are discharged from the hospital to convalesce at their apartments. Unfortunately, such was not the case with Amy. She was admitted into the hospital in late August and didn't get out until late October. We tried our best to keep Amy's spirits up, but it wasn't easy. While we were on the transplant floor, we witnessed BMT patients, some with Fanconi's, who succumbed from complications. It was heartwrenching to see these kids and their parents go through all of this. We learned true courage, nobility, and grace by getting to know them. We learned a deeper respect for the compassion and knowledge of the nurses and doctors who help fight this ongoing battle everyday.

### Donations

Donations may be made to the Fanconi Anemia Research Fund, a 501(c)(3) organization, as follows:

**Online:** Look for the Donations link on our home page ([www.fanconi.org](http://www.fanconi.org)).

**Telephone:** Call us at (541) 687-4658 or toll free (800) 828-4891.

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

Amy engrafted well, but ran into many complications that continued to keep her in the hospital until October 20th, her birthday. Finally on that day, almost two months from when she was first admitted, she was discharged. She then spiked a fever the following day and was readmitted. She stayed in the hospital for the next few weeks. She was beginning to think she would never get out.

Fortunately, Amy eventually improved enough to return to our apartment. She was incredibly weak, nauseous, and uncomfortable. She was taking over thirty oral meds and was on an intravenous pump. With frequent vomiting, diarrhea, and the general lack of adequate hydration, we hoped that she was able to absorb

the correct amount of medication. Over time she slowly improved.

On December 20, Dr. Macmillan gave Amy (and us) the green light to return home. Amy still had a very long way to go on the path to recovery, but had also come so very far to get to this point. We arrived home on Christmas Eve. It took another five months of semi-isolated recuperation at home until Amy would be feeling close to what would normally be considered "well."

After missing a year of school, Amy has just rejoined her classmates to commence 7th grade, something we were not sure we would be able to witness. We are very lucky and grateful. ♦

### Head and Neck Cancer in FA Patients

*continued from page 9*

- sore throat on one side
- enlargement or pain in the tonsil on one side
- ear pain on one side

### Nose, Sinuses and Upper Throat (nasopharynx), especially signs and symptoms affecting one side

- nasal blockage
- bleeding
- stuffy ear
- headaches
- visual changes

### Voice Box (larynx)

- hoarseness
- cough
- bloody phlegm

### Neck

- a painless or painful swelling in the neck that is not related to a cold

For routine screening, Van Waes recommended that patients do a monthly examination of their mouth and neck. He also recommended a dental and an oral examination every 6 - 12 months by a dentist who is familiar with the high risk of FA patients for head and neck cancer, and an examination and endoscopy by an otolaryngologist (one associated with a university system or major cancer center) once a year. If leukoplakia, dysplasia or other signs of pre-cancer or cancer are detected, van Waes recommends a vastly increased regimen of screening and surveillance, including biopsy, again by a physician highly experienced in the treatment of head and neck cancer and of FA. ♦

## Family Fundraising Efforts

From January 1 through June 30, 2003, FA families raised \$337,672 for Fanconi anemia research. The Fund also received donations of \$5,128 through the United Way and \$3,824 through the Combined Federal Campaign. Our thanks to all of you who have worked so hard to raise critically-needed research dollars.

### \$120,000 and up

Dave & Lynn Frohnmayer

### \$60,000 and up

Audrey & John Barrow

### \$20,000 - \$60,000

Christopher Scaff

### \$10,000 - \$19,999

Laurie Strongin & Allan Goldberg

Christie & Randy Kelley

Deane Marchbein & Stuart Cohen

Mark & Diane Pearl

Jeff & Arianne Pederson

### \$5,000 - \$9,999

Beth & Jeff Janock

Jack & Lisa Nash

Andrea & Bob Sacks

### \$1,000 - \$4,999

Mark & Linda Baumiller

Darryl Blecher & Diana Fitch

Randy & Nancy Bloxom

Marie & Antonino DiMercurio

Ed & Janice Duffy

Gene & Lynn Eddy

Andrew & Jennifer Gough

Jeff & Judy Hoffman

Lorraine & Kevin McQueen

Leighsa & Stephen Perlish

Dianne & John Ploetz

Jack & Tannis Redekop

Lynn & Rick Sablosky

Brenda Seiford

Karen Steingarten

Greg & Brandi Stuart

Michael & Beth Vangel

Kim & Michael Williams

### Up to \$999

Vicki & Andrew Anton-Athens

Ken & Jeanne Atkinson

Cherie Bank

Barbara Bedoya

Eric & Jennifer Bray

Richard Briga

Donald & Danielle Burkin

Joelle & Joachim Carvahlo

Joseph Chou & Frances Wang

John & Kim Connelly

Richard Day

Charles & Dahne Deeks

Pat & Mary DiMarino

Nathan & Ann Eckstadt

Kay Eubanks

Ezat & Laila Faizyar

Susan Gannon

Gary & Melody Ganz

Pat & Maria Gleason

Alan & Rachel Grossman

Mitchell & Tirzah Haik

Helen Healey

Brian Horrigan & Amy Levine

Charles & Katy Hull

Leardon Keleher

John & Karilyn Kelson

Shaid & Melvina Khan

Erik Kjos-Hanssen

Ayala Laufer

Eugene & Renee Lemmon

Peg LeRoux

Dylan Lewis

Gayle Licari

Eric & Beth Losekamp

Gregory & Lynette Lowrimore

Steve & Allison McClay

Cecelia Meloling

Griff & Cecilia Morgan

Sheila Muhlen

Kenny & Lisa Myhan

Robert & Mary Nori

Robin Paulson

Pedro & Marina Ravelo

Marcia Reardon

Les & Nancy Ross

Erik & Lori Salo

Mike & Catherine Sanders

Ron & Elesa Schaefer

Bill & Connie Schenone

Bryan & Karen Siebenthal

Connie Simpson

Jim & Carol Siniawski

Jeff & Debby Slater

Mark & Susan Traeger

Marc & Sandi Weiner

Nancy & Reese Williams

Barry Wood ♦

## Fundraising Assistance

Did you know that 85 percent of the donations to the FA Research Fund are raised by FA families?

The staff of the Fund stands 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. This insurance for a one-time event is often available through a family's homeowners insurance as a relatively inexpensive insurance rider. Please contact the Fund if you need assistance obtaining or paying for this required insurance.

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

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

## New FA Protein Discovered

*continued from 1*

FANCD2 protein by attaching a molecule of the tiny protein, ubiquitin, to it.

The newly-discovered ubiquitin ligase is encoded by a new FA gene, *FANCL*. The discovery of this enzyme should further enhance our understanding of FA, DNA repair, cancer, and even the aging process.

Scientists from the laboratories of Weidong Wang, PhD (National Institute of Aging), Maureen Hoatlin, PhD (Oregon Health Sciences University), Hans Joenje (Free University of Amsterdam), and Colin E. Bishop, PhD (Baylor College of Medicine) collaborated in this important discovery.

Grants from the FA Research Fund supported this research, confirming the outstanding record of the Fund in facilitating important discoveries in FA science. ♦

## My First Impression of the Fanconi Anemia Family Meeting

*continued from page 10*

patronizing on their part. In fact, I could clearly see that the physicians and researchers were genuinely interested in the welfare of the FA families.

Finally, I would like to thank FARF and, in particular, Suzanne Lauck for diligently arranging and making it possible for me to attend the FA Family Meeting. In Singapore (population: 4 million), Fanconi anemia is so rare that no more than five families are afflicted by it. As such, there is no patient education on this disease available there. I feel fortunate to be able to attend the FA Family Meeting and, God willing, I look forward to coming back to the FA Family Meeting in the coming years, this time with my family. It would be like meeting with family and old friends again. ♦



# Fanconi Anemia

RESEARCH FUND, INC.

1801 Willamette St., #200  
Eugene, OR 97401

phone: (541) 687-4658

(800) 828-4891 (USA only)

FAX: (541) 687-0548

e-mail: [info@fanconi.org](mailto:info@fanconi.org)

web site: [www.fanconi.org](http://www.fanconi.org)

### Newsletter Editors

Lynn & Dave Frohnmayer

Mary Ellen Eiler

Joyce Owen, PhD

### Layout and Design

Tanya Harvey, Wild Iris Design

### Staff

*Executive Director:*

Mary Ellen Eiler

*Family Support Coordinator:*

Suzanne Lauck

### Board of Directors

Barry Rubenstein, JD, President

David Frohnmayer, JD, Vice President

Ruby Brockett, Secretary/Treasurer

Deane Marchbein, MD

Kevin McQueen

Mark Pearl

Robert D. Sacks

Michael L. Vangel

Peter von Hippel, PhD

Joyce Owen, PhD, Director Emeritus

Lynn Frohnmayer, Advisor to the Board

### Scientific Advisory Board

Grover C. Bagby, Jr., MD, Chair

Manuel Buchwald, PhD, OC

Richard Gelinis, PhD

Eva Guinan, MD

Hans Joenje, PhD

Christopher Mathew, PhD

Stephen Meyn, MD, PhD

Raymond J. Monnat, Jr., MD

Maria Pallavicini, PhD

Leona D. Samson, PhD

Kevin M. Shannon, MD

Neal Young, MD

### Printing

Image Masters Printers

# The Sixteenth Annual International FA Scientific Symposium

## OCTOBER 14-17, 2004

Hyatt Regency Hotel  
Cambridge, Massachusetts