



# FAMILY NEWSLETTER

#27 A Semi-annual Newsletter on Fanconi Anemia for Families, Physicians, and Research Scientists Spring 2000

## Mark Your Calendars for Our 9th Annual Family Meeting

We look forward to returning to Aurora University's George Williams Lake Geneva campus in Williams Bay, Wisconsin from August 11-16, 2000 for our annual family meeting. This will be our third year at Lake Geneva with its picturesque lake-front. Besides hearing informative talks from several doctors and researchers, families can enjoy swimming and boating, an eighteen-hole golf course, tennis, shuffleboard, nature trails and campfire sites. In addition, all families have free access to the university's computer lab with internet services. Lake Geneva is an easy, less than two-hour drive from Chicago's O'Hare airport or forty-five

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*Manuel Buchwald, PhD, introducing James Hejna, PhD, at the FA Scientific Symposium.*

## Fanconi Anemia Scientific Symposium

One hundred and thirty-five researchers, treating physicians, and a good representation of FA family members from fourteen countries met in New Orleans for the Eleventh Annual FA Scientific Symposium, December 1-2, 1999. Forty-four scientists and treating physicians, many of whom had not spoken previously at FA Scientific Meetings, gave formal presentations. There were 18 posters, open to all attendees at four different times throughout the meeting. Uniformly positive evaluations assure us that this was our best scientific meeting to date.

Research topics covered seven areas: Gene Discovery and Gene Regulation; Diagnosis and Murine (mouse) Models; FA Protein Com-

plexes & Traffic; FA Protein Function; Mosaicism, Gene Therapy & Transplantation; AML, MDS & Cancer; and Function of Fanconi Gene Products. Some highlights:

Of most immediate therapeutic relevance were findings presented on bone marrow transplantation. In two transplant centers, the addition of fludarabine has allowed transplanters to reduce the toxicity of the conditioning regimen with matched sibling donors. In five centers, fludarabine as part of different protocols has enabled engraftment to occur in fifteen out of fifteen patients, with only mild toxicity in most patients. Thirteen of these patients had unrelated or mismatched donors; fourteen

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## A Comprehensive Approach to Improving Transplant Outcomes at the University of Minnesota

by Margaret L. MacMillan, MD and John E. Wagner, MD

There are four major obstacles to successful bone marrow transplantation (BMT) for FA, regardless of the donor type (matched sibling versus unrelated donor): graft failure (the failure of the transplanted bone marrow to establish itself in the patient, and grow new marrow and blood cells), graft-versus-host disease (GVHD, a condition in which the donor's blood cells attack the tissues of the patient), infections, and late effects, like cancers of the head and neck. To improve the survival and quality of life of all FA patients and particularly those undergoing BMT, we are systematically investigating each of these issues at the University of Minnesota.

### Graft rejection

The high rate of graft failure after BMT in FA patients is due to an inability to suppress the patient's immune system sufficiently. Cyclophosphamide (Cytosan®) and radiation have been the mainstays of therapy because of their potent immunosuppressive properties. Unfortunately, FA patients often do not tolerate conventional doses of this therapy due to their extreme sensitivity to these agents.

Crucial studies done in the early 1980s by Eliane Gluckman and co-workers demonstrated that FA patients with sibling donors could tolerate *low doses* of cyclophosphamide and *limited* irradiation, and

that such therapy would suppress the patient's immune system enough to allow engraftment of marrow transplanted from a matched sibling. While graft failure still occurred in about 10-15% of FA patients (as compared to <1% in non-FA patients with leukemia receiving high dose therapy), survival approached 65-85%, depending upon the age of the patient and prior therapy.

Unfortunately, such low dose therapy is insufficient for those with unrelated donors. Even when doses of cyclophosphamide and radiation were increased to the highest tolerable levels possible for FA patients, graft failure remained 35%. In February

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## FA Transplantation Results, New Approaches for Unrelated Donor Transplants, and Upcoming Gene Therapy Trials at the Fred Hutchinson Cancer Research Institute (FHCRC), Seattle, WA

by Peter Kurre, MD, Ann Woolfrey, MD, Mary Flowers, MD, Rainer Storb, MD, and Hans-Peter Kiem, MD

It has been 26 years since the first patient with FA was successfully transplanted with stem cells from a matched sibling donor in Seattle. Bone marrow transplantation has since become accepted as the only curative approach for the progressive bone marrow failure of FA. A large number of centers around the world now routinely perform transplants for FA patients.

For patients with related sibling donors, a major reduction in the overall toxicity of conditioning has been achieved. Total body irradiation

has been eliminated from the conditioning regimen, without compromising engraftment. More recently, collaborative efforts between FHCRC and the University Hospital in Curitiba, Brazil have demonstrated that substantial dose reduction of cyclophosphamide (Cy) is possible without a significant increase in risk of graft failure and with improved survival. Recently published results show a 3-year actuarial survival of 88% for patients with total Cy dose of 100 mg/kg. We are continuing to evaluate the effect of dose reduction

and have thus far not seen an increase in graft failure with current accrual at the 60 mg/kg dose level. Reduction in total Cy dose has resulted in reduced incidence and severity of toxicities such as mucositis and cystitis, compared to prior patient groups treated at total Cy doses of 140 mg/kg or higher.

Unlike most other patient groups, transplantation of FA patients from unrelated donors has been a formidable challenge. Conventional myeloablative regimens have, in the past,

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## **Trial of Interleukin 11 (IL-11) Not Successful in Raising Platelet Counts**

*By Jim Croop, MD, University of Indiana*

We have now used IL-11 on three patients with FA, and I know of a fourth. Unfortunately, none had a sufficient response to consider the use of IL-11 to be successful. We were hoping for a response rate of at least 40% of the patients. Our statisticians believe that with our current results, there is at best a 3% chance of this occurring. Hence, we have decided not to enroll any more patients with FA, as the likelihood of response is not worth the potential side effects. I wish we had more to offer. ♦

## **Squamous Cell Cancers of the Head and Neck**

*Frank Ondrey, MD, University of Minnesota*

Squamous cell aerodigestive cancers refer to malignant tumor formation of the linings of the lips, gums, oral cavity, and a variety of structures of the throat, including the larynx (voice box) and entrance to the esophagus. These cancers affect 40,000 people in the United States yearly, with most patients being older than 45 years of age and with men affected about twice as often as women. This disease is most common in individuals who use tobacco excessively through smoking or chewing and is also more common in individuals who drink alcohol to excess. FA patients are unusually susceptible to these cancers whether or not they drink or smoke, and at much earlier ages than the general population. Once a person has been diagnosed with a head and neck squamous cancer, he or she is more susceptible to getting additional tumors of the aerodigestive tract (throat, lung and esophagus).

Squamous cell cancers start out as small ulcers, irritated areas, or whitish or reddish plaques with a sandpaper-like texture. These lesions are slow growing, and many individuals with these tumors do not notice them until they become painful or interfere with eating and drinking.

Because these lesions are most curable at early stages and because they are recognizable by Ear, Nose and Throat surgeons, they can be effectively screened and usually treated on an outpatient basis if they are less than the size of a quarter. However, many lesions go unnoticed because of hesitation to seek attention or a variety of other social and psychological factors, and the tumors may go on to affect speech, breathing, and eating. These cancers can progress in size and spread to the lymph glands of the neck and the lung. Large tumors or tumors that spread to the neck usually require extensive treatments, involving surgery, radiation, and sometimes chemotherapy. In spite of great advances in treatment, survival for these more advanced stage lesions is less than 50% at 5 years. This rate of cure has not essentially changed for over 25 years. Furthermore, there is considerable morbidity and rehabilitation after the successful removal of these tumors since these tumors affect organs of communication and eating. It is possible that treatment of these tumors could involve complete removal of the voice box or removal of significant portions of the roof of the mouth or the tongue. Interfering with the function of these organs

## **New Gene Therapy Trial Begins**

Lynn Welfare Mendenhall, age 46, underwent gene therapy at the University of North Carolina, Chapel Hill Medical Center, under the direction of Chris Walsh, MD. The procedure took place on February 14, 2000. In the report carried by CBS news, Dr. Walsh stated that results could not be expected for several months. Additional patients in complementation group A will be admitted to this trial. We salute Lynn's pioneering heroism.

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often requires a difficult rehabilitative process.

After successful treatment of these malignancies, head and neck cancer patients require extremely close follow-up with immediate attention to any abnormalities of the throat and lungs. Any symptom referable to these organ systems (e.g., hoarseness, chronic cough, coughing up blood) may indicate that another tumor is forming.

A variety of factors may contribute to the growth and spread of these tumors, but it has been well recognized that defects in immunity of head and neck cancer patients exist and are associated with decreased success in treating these lesions. It is not known whether the immune defects noted in patients with head and neck cancer are secondary to the tumor growth itself, a lack of good nutrition, or some other factor. It is known, however, that certain immune-compromised patient groups are more susceptible to developing head and neck cancer whether they smoke or not. There is considerable evidence that patients who undergo transplantation of organs,

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# Toluidene Blue as a Screening for Oral Cancer in FA

by R.A. Ord, DDS, MD, Dept. OMS, University of Maryland

As more patients with FA have survived into adulthood with the use of bone marrow transplant, an increase in certain cancers has become apparent. Among these is squamous carcinoma of the oral cavity, particularly the tongue. This cancer is most common in FA patients in their mid-twenties (mean 26.8 years) and has a higher incidence in females. The origin of this cancer may be related to defective DNA repair, immunodeficiencies or immune suppression during bone marrow transplant.

Oral cancer frequently develops in premalignant lesions which are either white (leukoplakia) or red (erythroplakia). When these mucosal changes are widespread, as may occur in FA, it can be very difficult for the clinician

to know where best to biopsy. Oral cancer has its best prognosis when diagnosed early, particularly prior to metastasis to the lymph nodes in the neck. Usually, premalignant lesions will develop varying degrees of dysplasia prior to becoming an invasive carcinoma, and this is the ideal time to diagnose these lesions.

In order for the clinician to biopsy the specific area of a large red or white lesion that is most likely to show dysplasia or early carcinoma, screening with toluidene blue has been advocated. This vital dye stains cells with increased DNA synthesis and will stain early carcinomas. Dysplastic lesions show variable staining, with more severely dysplastic lesions more likely to show positive staining.

The toluidene blue test can be

performed as a rinse or applied topically to the lesion in question. Mashberg and Samit recommend decolorizing with 1% acetic acid following application, to clear excess stain. The entire test takes less than 5 minutes. In order to assess the need for biopsy in the diffuse red and white lesions of FA and to minimize sampling errors, toluidene blue staining can be used as a screening test. In one review of 6 large studies with 1,071 patients with oral lesions, sensitivity was 96% - 100%, and specificity was 87% - 100%.

(Study reference: Clayman, L. "Management of Mucosal Premalignant Lesions, *Oral and Maxillofacial Surgery Clinics of North America*, Vol. 6, No. 3, 431-444, August, 1994). ♦

## FA Transplantation Results

*continued from page 2*

resulted in high rates of graft failure, excessive regimen-related toxicity, significant morbidity, and mortality from graft-versus-host disease, with poor overall outcome. While the immunologic barrier between unrelated donor and host requires more intense immunosuppression, complete ablation of the patient's marrow, such as used in most conditioning regimens, may not be necessary. During the past several years, researchers at FHCRC have pioneered the use of low dose non-myeloablative conditioning regimens for a variety of malignant and non-malignant conditions. Results from more than 60 non-FA patients treated with this non-myeloablative regimen over the past 2 years have demonstrated that marrow grafts from HLA-matched related and unrelated donors can be

established with substantially reduced toxicity. Based on these encouraging results, we have developed a non-myeloablative treatment protocol for FA patients who have an HLA-matched unrelated donor (Fig. 1).

Along with other groups, we continue to make progress in the field of retroviral vector-based gene replacement, and this approach can now be considered for those FA patients without a stem cell donor. A newly

formed International Fanconi Anemia Consortium between FHCRC, Indiana University, St. Jude Children's Research Hospital, and the University of Düsseldorf, Germany will conduct pilot trials to investigate the safety and efficacy of gene replacement therapy. We will focus initially on patients with FA complementation group C and later plan to extend our efforts to patients belonging to groups A and G. ♦

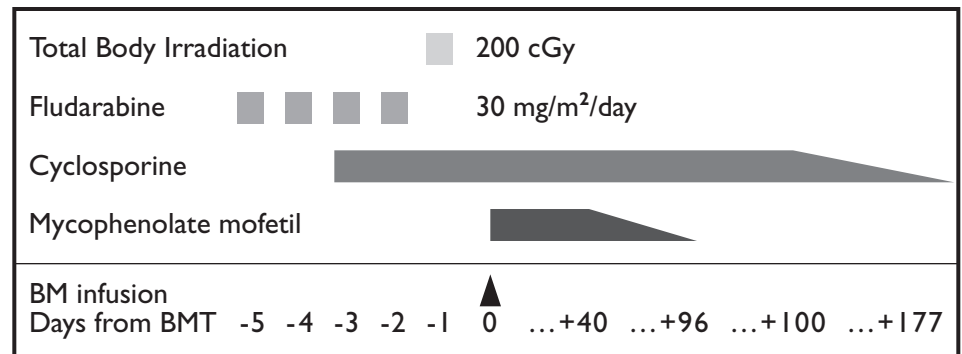


Figure 1: A non-myeloablative treatment protocol for FA patients who have an HLA-matched unrelated donor

## A Comprehensive Approach

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1999, we developed a new preparative regimen. Fludarabine is a potent immunosuppressive agent which does not cross-link DNA, making it a good choice for use in patients with FA. To date, nine FA patients have been enrolled on the protocol combining fludarabine with cyclophosphamide and total body irradiation at the University of Minnesota. Of the seven patients who had their unrelated transplant at least one month ago, all seven have successfully engrafted. These excellent results with fludarabine suggest that we may have overcome the barrier of graft failure, a major obstacle to the success of unrelated donor BMT.

### Graft-versus-host disease

In general, GVHD is a major complication after BMT, particularly with unrelated donors. While there are several approaches for reducing the risk of GVHD, T cell depletion of the marrow prior to transplant is the most successful method. Eliminating GVHD has the additional potential advantage in FA patients of reducing the risk of later head and neck cancer. Although the association has not been proven, late cancers in FA patients have occurred primarily in those who had chronic GVHD.

To date, none of the FA patients enrolled on the fludarabine, cyclophosphamide, and total body irradiation protocol and transplanted with T cell depleted unrelated marrow at the University of Minnesota has developed GVHD. One recipient of HLA 2 antigen mismatched umbilical cord blood developed moderate (grade 2) GVHD. None of the patients has thus far developed proven chronic GVHD, but follow-up is too short. These excellent results

with T cell depletion suggest that we may have overcome the barrier of GVHD, which had been a major obstacle to the success of unrelated donor BMT.

### Infections

All patients are susceptible to infection after BMT. However, FA patients may have a higher risk of infection. Possible reasons for this increased risk may be 1) increased breakdown of the normal mucosal barriers (i.e., lining of the gut) with chemotherapy and radiation, 2) prior immunosuppressive therapy (e.g., prednisone, cyclosporin A, anti-thymocyte globulin), 3) prolonged period of neutropenia (i.e., absolute neutrophil count of less than 1,000 for months or years prior to BMT).

In an effort to reduce the risk of infection, we have instituted a new evaluation and treatment program prior to BMT. Notably, we have already observed a high rate of infections not obvious by routine screening (occult infections) that we previously would not have detected. More patients are delayed from going to BMT in an attempt to minimize the risks of infectious complications. With closer monitoring by an infectious disease team interested specifically in FA, it is hoped that the obstacle of infection, particularly fungal infections, will also be overcome. New approaches for speeding immune recovery, such as infusing genetically modified lymphocytes, will be investigated clinically in the near future.

### Late effects

While BMT can cure the blood abnormalities in FA patients, these patients, unfortunately, remain at risk for cancers, especially of the head and neck, and cervix in females. In transplanted patients, two factors appear to be associated with risk of



*Margaret L. MacMillan, MD  
addressing FA Scientific Symposium*

late malignancy: 1) the use of irradiation, and 2) development of chronic GVHD. In an attempt to decrease the risk of malignancy, we have developed a regimen that does not include radiation for patients receiving matched sibling donor transplantation, and that uses T cell depletion to reduce the risk of GVHD, even in patients with HLA matched sibling donors.

We do not know if BMT changes the risk of cancer in FA patients. It is well known that non-transplanted FA patients are at high risk of squamous cell carcinoma of the skin, cervix, head and neck. Under the direction of Frank Ondrey, MD, an Ear, Nose and Throat surgeon at the University of Minnesota (see Ondrey's article on page 3), we plan to initiate clinical prevention and treatment trials for FA patients at high risk of carcinomas of the head and neck.

In summary, BMT is the only treatment that can "cure" the blood problems of FA. Fludarabine promotes engraftment, and T cell depletion reduces the risk of GVHD. Research is now directed toward development of new approaches for reducing the risks of infection and cancer after BMT. The University of Minnesota is also initiating an FA gene therapy program. ♦

## Words of Wisdom and Advice From Our E-Mail Group

### Bob Sacks:

Our 25-year-old daughter, Dani, is diabetic. Her blood sugar levels have been extremely resistant to insulin shots, even at high doses and various combinations. I asked the endocrinologist at Cornell Hospital in New York City if any of the newest oral diabetic medicines might help to control her levels. He recommended Actos as probably having the least potential side effects for an FA patient. Within two weeks of single daily doses of Actos, Dani now has had several days of perfectly normal blood sugars for the first time, and we have been able to reduce her insulin doses, which were dangerously high.

Arleen Auerbach believes there is a much greater than normal risk for FA patients to develop insulin-resistant diabetes. This is especially true after transplant or chemotherapy. FA teens need periodically to have their blood sugar levels checked, especially if they show clinical signs such as frequent urination and lethargy. Each patient is different, and your endocrinologist and hematologist should advise you.

### Lynn Welfare Mendenhall:

Don't mess around with a persistent mouth sore. My doctor told me not to worry about my sore, as did the doctor of another adult FA friend. As it turns out, we both had cancer! The only way to find out is to biopsy the suspicious area.

### John Wagner, MD:

In response to a teen's concern that she would inherit a BMT donor's likes and dislikes, John Wagner writes: "Feelings, likes and dislikes come from you, not the bone marrow or cord blood that is transplanted. Be assured that the sexual orientation, race, religion, etc., of the donor in no way will influence you. The fact that someone (your donor) cared enough to give a part of him or herself to you, looking for nothing in return, however, should change you in some way. Can you imagine someone risking his or her life to save you without even knowing who you are!"

### Dave Frohnmayer:

Let me echo the Hoffman's response on chicken pox. It is a very serious viral infection and a common and often dangerous bone marrow suppressant for FA patients. Vaccinate against it if you can. See Dr. Shahidi's comments on this subject in the FA Handbook! In our experience, the counts can recover (in some patients they do not), but it may (and usually does) take many months.

### Beth Vangel:

Beth Vangel had described her daughter Amy's blood count decreases following a bout with chicken pox and the subsequent recovery of her counts. She adds "I know when Amy gets a bad cold, stomach flu or minor illness her white count drops, but does rebound (thank God)." ♦

patients were alive and well from one month to over two years post-transplant. Richard Harris, MD, Cincinnati, wrote a comprehensive report on these transplants, with a graph indicating specific protocols used. See last paragraph of this article on how to obtain a copy of his article and graph.

Researchers in the laboratory of Hans Joenje, The Netherlands, have cloned and characterized the gene for complementation group F. That is the fourth FA gene isolated to date.

There is strong evidence that a fifth FA gene has been identified. Further details will follow.

Much information was presented on the function of the Fanconi anemia proteins, how these proteins interact, and how they are regulated. It was apparent to all in attendance that tremendous progress has been made in this area. The FA proteins remain a crucial subject for further investigation.

Presentations focused on improved and novel vectors for gene therapy, correlations between specific genes (and mutations) and patient symptoms and outcome; future strategies for treating leukemia; and the effect of mosaicism on BMT outcomes.

Your editors include an outline of the symposium, giving the name of each presenter and the title of his or her presentation. Given the tremendous volume of this material and its highly technical nature, we have not prepared a *Science Letter*. If you wish a copy of one or more of these abstracts, please indicate your request on the enclosed form and return it to the FA Research Fund office. ♦

# From Participants' Evaluations of the Scientific Symposium



*"Congratulations! Your work is generating a lot of information about this challenging disease. Keep up with it."*

*"This was a good meeting. I came away with a feeling that a lot has been accomplished in the last year!"*

*"Best meeting to date."*

*"This meeting was excellent. The rate of progress in terms of knowledge (genes, protein function, correlation between genes and phenotype) has been really impressive when compared to what was known at the Denver meeting."*

*"I have been intermittently attending these meetings since 1990. I believe they have gone from strength to strength. This meeting for me so far has been the best yet."*

*"The Fanconi anemia symposium continues to be a very interesting and motivating meeting. Please keep up the good work!!! Bringing together all the scientists for lively discussions is very informative!!!"*



*"Of most therapeutic relevance were the talks on FA transplants using fludarabine (although the numbers are small presently). Fludarabine seems to improve stem cell transplantation success remarkably."*

*"Overall the meeting was excellent. The data exchange that occurs every year is helpful to everyone regardless of field."*

*"I find the combination of scientific rigor and the presence of families to be unique. It is a fantastic meeting."*

*"Deepest thanks and sincere praise to the families who suffer from Fanconi anemia."*



# FAMILY NEWS

## Sharing Hope

by Judy & Jeff Hoffman

We remember the day our daughter was born. We remember the day our son was born. We remember the day our son was diagnosed with Fanconi anemia. These are days that are etched in our minds for eternity.

We have just hit the one-year mark when Sam was diagnosed with FA. He will be turning two in February. Sam was born with right thumb duplication and with one kidney. With the exception of thumb surgery at six months, his daily routine has been remarkably "normal." On the other hand, my wife and I are just starting to learn to breathe again.

We have grown up incredibly in the last year. We have become better

people since the day our son was diagnosed. We have become better parents. Facing this adversity has caused us to look at everything completely differently. It is amazing how our perspective and our views about issues in life have changed so dramatically from one day to the next. Things we used to think were important no longer have the same meaning. Priorities change.

We have learned so much during this past year. Whether it is from the e-group or through personal dialogue, we have learned to see all of the fear, anxiety, and hope that lie in each parent of an FA child. We are tremendously grateful to all of you who have



Sam and Jeff Hoffman

shared these feelings with us and taught us how to deal with this crisis. We hope we can teach others in the

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## Struggling with Fanconi Anemia

by Richard Briga

My name is Richard Briga, age 42, and I am afflicted with FA.

I always lived a healthy lifestyle (eating right, exercising) and because of this I thought I'd be the last person to ever get cancer. I have no physical anomalies of FA. There is no previous history of cancer or FA in my family. I have a brother and sister, and neither tested positive for FA. Except for frequent colds, I have been healthy and rarely needed medical care.

In the summer of 1997, I began to develop declining blood counts following a brief flu-like illness. I was treated with B<sub>12</sub> injections and iron supplements by my primary care physician, but my counts continued to plummet. I was not informed that

my blood was low on all counts (Hgb, WBC, platelets) and was being treated for simple anemia. I was constantly sick with colds, respiratory infections, and finally facial erysipelas (strep infection of the skin). After nine months of suffering, I was referred to a hematologist for a long overdue bone marrow biopsy. In May 1998, I was diagnosed with myelodysplastic syndrome (MDS). In July 1998, I became dependent on blood transfusions. Since my brother, David, was a perfect match, an immediate bone marrow transplant was given as my only hope.

Seeking a second opinion, I was referred to Dr. Steven Shuster at the University of Pennsylvania Hospital, Philadelphia. Determined to find a

cause for my MDS, he recommended an FA test prior to my scheduled bone marrow transplant. A short time later, he called to tell me the test was positive. He suggested I have my BMT at the University of Minnesota under the guidance of Dr. John Wagner, because of their expertise with FA patients. Dr. Shuster's astuteness in pursuing the Fanconi anemia test probably saved my life, since this test is not usually performed before BMT. FA patients require a greatly reduced dose of chemotherapy and radiation.

Confused and knowing nothing about this disorder, I contacted the Fanconi Anemia Research Fund (FARF) the next day and learned the

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# My Bone Marrow Transplant

by Christopher Byrd

My name is Christopher Byrd, I am 17 years old, and I am on day 71 of my unrelated bone marrow transplant in Minnesota.

For the past year, my counts had continually fallen. The Anadrol (oxymetholone) and the GM-CSF (a colony-stimulating factor) that I had been on for seven years seemed to be losing their effectiveness.

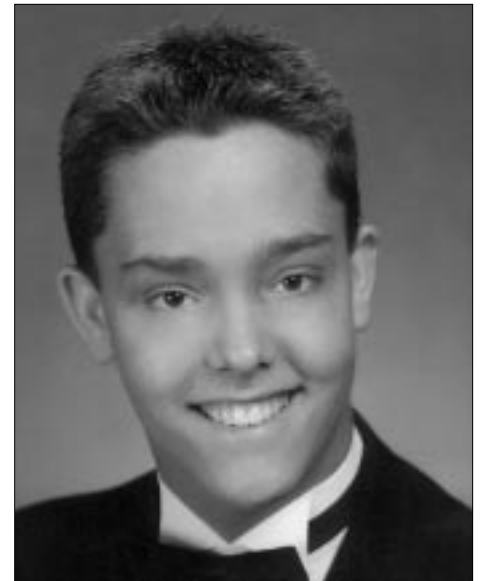
I felt somewhat tricked this summer when my Mom asked me to go to the FA camp in Wisconsin and meet with some doctors. I thought sure, I could use a little vacation! The next thing I know, I am having dinner with Dr. John Wagner, who happens to be one of the finest bone marrow transplant doctors in the country. He gave it to me straight, as he always does. I needed to go to transplant as soon as we could find a donor. My ANC (absolute neutrophil count) was dangerously low, and I was transfusion-dependent. I sat in on all of the meetings and, ultimately, the choice was mine. Recent success using the new drug fludarabine and the fact that I had two 6/6 matches were very encouraging. As you can see, I decided to go for it and, as it turns out, not a moment too soon. During my pre-transplant work-up, my chromosome studies came back with two clones, one being monosomy 7.

Going through transplant is much like going on a roller-coaster ride. You never know what is around the corner. I tolerated my chemotherapy and radiation phase very well. The mouth sores from the chemo usually develop a couple of days after transplant. My mucositis (mouth sores) was very tolerable (3 on a scale of 1-10). But when the pain became too much, they put me

on morphine. I really don't remember much about those two weeks at all. The time flew by. I engrafted on day 14 and, just when I thought I'd be discharged from the hospital, I spiked a fever. It turned out to be *Candida krusei*, a very nasty fungus. The doctors decided to take the line out of my chest and replace it with a PIC line in my arm. That hurt!! They treated me with amphotericin B, and the candida was gone in five days. Very lucky! They put a new central line in my chest, and everything was going great until CMV (cytomegalovirus) appeared in my lungs. Another major bummer. Thanks to an extensive and thorough line of treatment, my lungs are now clear and I am feeling great. Some other problems I've had include high blood pressure, which we have under control now, and diabetes. I needed insulin for only a few weeks, and now they check my glucose level regularly.

I was discharged from the hospital on December 30, 1999, just in time for the new year. Now I go to the clinic almost every day for counts, transfusions, etc. The only thing I've had to battle lately is my hemorrhagic cystitis. It is a major pain but not life-threatening. This is an after-effect from the chemotherapy. It seems it takes the bladder longer to recover. This is especially true in my case!

Dr. Wagner told me yesterday that I am now "just doing time" until my 100th day when I return to my family, friends, and fun in the Florida sun, with my hat, SPF 30 sun block, and a long-sleeve T-shirt! Even though there were some tough times along the way, I feel that I made the right decision. I urge you not to think of bone marrow transplant as a



last resort but as a possible cure of the bone marrow. The medical staff here in Minneapolis has been like a family away from home, and I cannot thank them enough for their hard work, dedication, and love. I know it's not over yet, and that it will be a long year, but it has been and will be a small price to pay for many healthy years to come. ♦

## 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.

## A Loving Tribute and Remembrance of Chris Hull

by Jeff Hull

When we started this whole transplant process, Chris called me just before he entered the hospital to have his CV lines surgically inserted. It was a Wednesday and I was still in Montana. I would fly to Boston the following day, the Thursday of Thanksgiving. Chris called around ten in the morning. He was a little nervous when he told me he was heading over to the hospital, but nervous the way you are on game day—just ready to get on with it. “I’m going over there to start a new life,” Chris said. “A new life?” I asked. “One way or another,” Chris said, “today is the beginning of a new life for all of us.”

As of this morning, December 16, 1999, we will all have to start living that new life without Chris to help us, without him to show us how you do it with courage and grace. Chris has left this life. At 11:55 EST Christopher Stuart Hull died in the ICU of Children’s Hospital in Boston. Sometime during the late afternoon yesterday, his kidneys ceased working. Ironically, the bone marrow graft between Sarah, our sister, and Chris worked. When he died, Sarah’s marrow had successfully grafted into his body. But it was too little, too late. There were just too many other obstacles for his body to overcome.

It’s important to remember that Chris was very sick when he walked into this hospital, that he had liver cancer and would have died without trying what he and his doctors in Boston tried. He knew the dangers. Nobody was more aware of his chances than Chris. That he died during his last effort to survive is only

final evidence of the fierceness with which he lived his life.

It is a rare thing to be granted the privilege of knowing a spirit like Chris’. I hope that, in the days and years to come, when all of you think of Chris and of his life, what you will notice is how much those memories resonate with love and admiration. I hope remembering Chris helps us all realize how precious is the time we have with those we love, how those shared moments are the moments from which memories are born. Ultimately, memory is the only difference between life and death.

As we move into our new lives, our lives without Chris, it would be a terrible waste if we did not apply what we could learn from his long struggle. Each of us saw how relentlessly Chris lived, and we saw what



he chose to focus upon so intently: family and friendship, yes, but fun, too. Chris loved to have fun. I hope we all try to live some of the rest of our days like Chris lived his. When we remember our old lives, the ones with Chris in them showing us how to be brave and strong, let’s please learn the courage to love with whole hearts, and to find a voice for that love, to say it out loud to the people who inspire it.

The last thing Chris ever said to us, to me and my father and mother, he said while he sat in his room on the Transplant Unit. Chris had already made the decision to go on the ventilator, and he was scared, his legs jittering and his focus scattered. But he sat down with us and pulled himself together to look at us all clearly one last time.

“I want you to know how much I appreciate everything you’ve done for me in my life,” Chris said to us, before the nurses and doctors wheeled him away to start his new life, “and I want you all to know I love you very much.”

Given the opportunity, I think that was something he would have said to each of you, too.

Peace,  
Jeff Hull

*The Hull Family*  
invites you to join us in  
**“The Celebration of the Life”  
of Christopher Hull**

*May 6, 1966 - December 16, 1999*

**on Saturday, May 3, 2000  
at First Presbyterian Church  
Fremont, Ohio**

*Lunch following at the Fremont  
Country Club*

*Anyone interested in attending can get  
directions and other details from*

*Dr. & Mrs Charles Hull  
1421 Birchard Ave.  
Fremont, OH 43420  
phone (419) 332-6618  
fax (419) 332-9099*



## *In Loving Memory*

**Kyle Burzynski**  
11/11/96 – 1/26/00

**Thomas Fitzgerald**  
1/5/96 – 1/26/00

**Bubba Gray**  
3/24/94 - 1/26/00

**Chris Hull**  
5/9/66 - 12/16/99

**Joshua Sundsvold**  
8/26/94 – 5/20/99



*Danielle DiMarino*

### **I Only Wanted You**

*They say memories are golden,  
Well maybe that is true,  
I never wanted memories,  
I only wanted you.*

*A million times I needed you,  
A million times I cried,  
If love alone could have saved you,  
You never would have died.*

*In life I loved you dearly,  
In death I love you still,  
In my heart you hold a place,  
No one could ever fill.*

*If tears could build a stairway,  
And heartache makes a lane,  
I'd walk the path to heaven,  
And bring you back again.*

*Our family chain is broken,  
And nothing seems the same,  
But as God calls us one by one,  
The chain will link again.*

*These poems were written by Jennifer DiMarino, sister of Danielle. She read them at Danielle's memorial service.*

### **When I Must Leave You**

*When I must leave you  
Please don't say that I gave up  
Just say that I gave in.  
Don't say I lost the battle  
For it was God's war to lose or win.  
Please don't say how good I was  
But that I did my best.  
Just say I tried to do what's right  
To give the most I could, not less  
Please don't give me wings or halos  
That's for God to do.  
I want no more than I deserve  
No extras, just my due.  
Please don't give me flowers  
Or talk in real hushed tones.  
Don't be concerned about me now  
I'm well with God  
I've made it home.  
Don't talk about my illness  
It's over and done.  
Just see to all my family needs  
Especially the little ones.  
When you draw a picture of me  
Don't draw me a saint  
I've done some good, I've done some  
wrong.  
So use all your paint  
Not just bright and light tones  
Use some gray and dark.  
In fact don't put me down on canvas  
Paint me on your heart.  
Forgive me for the wrongs I've done  
And with love that's left  
Thank God for my soul's resting  
Thank God for all who loved me  
Praise God who loved me best.*

## Struggling with Fanconi Anemia

*continued from page 8*

seriousness of this diagnosis. Things just seemed to be going from bad to worse, and it became apparent that, without a successful transplant, I would not survive much longer. By this time, my hemoglobin had dropped to 6, and my white count was so low I was riddled with infections.

My mother, brother David (donor), and I departed immediately for Minneapolis, Minnesota. I underwent BMT on November 17, 1998, and things went incredibly well. My transplant was without complication and I was allowed to return home to Pennsylvania on day 50. Transfusions have been unnecessary since my discharge from the hospital. I developed some mild chronic graft-versus-host disease (GVHD) of my skin and eyes while on a cyclosporine taper at about Day 150. Basically, I developed dry eyes and skin with slightly blurred vision. My cyclosporine was then increased to 225 mgs., and my symptoms disappeared. Everything seemed to be better than I ever expected. By the summer of 1999, I resumed my conditioning regimen of hiking and cycling. Life finally seemed to be getting back to normal, and I was getting into amazingly good condition again. I began to plan on returning to work in the fall. Things couldn't have been better.

In September, I was brushing my teeth one morning, and I felt like I had something lodged in my throat. I called my doctor immediately and was referred to an Ear, Nose and Throat (ENT) specialist. An endoscopy revealed a tumor the size of a quarter in the supraglottic region of my throat adjacent to my epiglottis (a small flap of cartilage that seals off the airway while swallowing). I was followed closely after transplant

and my doctors examined my mouth every 2 weeks to check for GVHD, but this tumor was too low to be detected without endoscopy. Knowing that adults with FA are at high risk for oral cancer, I expected the worst. A biopsy a week later revealed a malignant squamous cell carcinoma, and to be honest I wasn't surprised. This disease is relentless, unpredictable, and exceedingly cruel. Just when I was doing so well something like this had to happen. Now what would I do?

My ENT doctor recommended a partial laryngectomy with possibly a total, depending on how far the cancer had spread. The thought of being disfigured was devastating to me, and at this point, I felt worse than when I was dying of MDS. But I knew I had to make a decision fast because this cancer was spreading. I decided to return to Minnesota because my doctors there know my case best. I underwent a partial laryngectomy on October 5, 1999. The surgeon, Dr. George Adams, excised the area above the vocal cords. My voice was preserved, but I had to learn to swallow in a different way to compensate for the missing epiglottis. I have not taken cyclosporine since the surgery and, fortunately, my GVHD has not recurred.

Since the cancer spread into my neck, Dr. Adams also performed a neck dissection and excised the left side of my throat. I was hospitalized 11 days and the pain and misery from this surgery were much worse than my bone marrow transplant. Dr. Adams removed 28 lymph nodes, and 6 were positive for cancer. The cancer was therefore likely to recur, so radiation treatment was necessary to eliminate any residual cells. However, radiation is complicated and risky for FA patients.

Presently, I am in Minnesota



undergoing radiation treatments, and my dose is slightly reduced because of FA. I had my 8th treatment this week and, aside from a dry mouth and a sore throat, I am feeling quite well. My blood has not been affected. My counts were Hgb 13.5, WBC 5.3, and platelets 288,000. My doctors feel that the cyclosporine had "something to do" with my developing throat cancer, but it may have been there before my BMT. **Regardless of the cause, my advice to adults with FA would be to see an ENT doctor at regular intervals for endoscopy.** This type of cancer presents no early symptoms but, if detected while small, radical surgery may be unnecessary.

I thought after my BMT that my problems with FA would be over, but this disease seems to be tenacious. Looking over the past 18 months, FA has turned my life into a nightmare and seems to make everything more complicated, more painful, and more expensive. Mentally and physically I'm holding up surprisingly well, but it isn't easy. The moral support I receive from my family, many friends, co-workers, my doctors, and FARF has made an unpleasant situation bearable and, above all, hopeful.

*Editors' note: Richard is now at home and recovering. ♦*

## Sharing Hope

*continued from page 8*

future some of the wonderful “tips” that have been passed on to us.

The one “tip” my family would like to pass on to all those reading this article is **you must never give up hope**. For it is in hope that a cure for this disease exists. It is in hope that God lives and allows us to love our children in the way only a parent of a child with FA can understand. And it is in hope that we are able to share a common bond with people we do not even know. Hope allows us to breathe again. It allows us to understand science that a few years ago many of us did not even think about. It is hope that has given us the strength to deal with this disease that haunts us every day and night. Look into your children’s eyes and see the hope that exists. Let this be your driving force and your source of strength. Most importantly, let them look into your eyes and see your hope.

Sam has plenty to be hopeful for. Currently, all of his counts are normal. He has a wonderful sister (Caren who is six years old) who adores him, and a grandmother who

lives with us. Sam also has parents who really get to “experience” all of him. Most of all, Sam has love from all of us. We were fortunate that Sam had the advantage of being diagnosed with FA when he was young. It has allowed us to get educated. It has allowed us to get proper counselling for us as well as counselling on issues regarding Caren and her feelings. Thanks to the Fanconi Anemia Research Fund, we are eleven years further into research on FA. Transplant success rates are getting better. Medical science is advancing rapidly. People are rallying their support around our family.

Though I wish things were different for Sam and our family, we firmly believe that this is happening for a reason. We accept our fate and our place in this world. We will love Sam for all that he is and all that he will be. We will love Caren for all that she is and for all that she will be. Most importantly, we will love each other as a family. We will hope and pray together that a cure is found which will help all of us. Thanks to everyone for helping us through this first year. We will share our hope with you when we see or speak to you. God bless all of our children. ♦

## Tracing the Origin of the IVS4 Mutation in Ashkenazi Jews

I am Phyllis Goldberg, grandmother of an FA child. I have a request for members of the FA community with the *FANCC* IVS4 mutation or who are carriers of this mutation. This is the mutation most commonly found in Ashkenazi Jews, who originated in Eastern Europe. This mutation may be unique; that is, it may have occurred once, in a common ancestor of all those who have it. I am interested in tracing its roots. If your family carries this mutation, and if you know the towns or general area where your ancestors came from, could you please contact me?

Strand Drive #12,  
North Bethesda, MD 20852.  
phone: 301-468-3896  
e-mail: stranddr@aol.com

## Squamous Cell Cancers

*continued from page 3*

like a kidney, seem to get more squamous cell cancers. Often these tumors occur in skin and the aerodigestive tract. Squamous cell cancer in transplant patients also seems to be a more virulent form of the disease. There appears to be decreased survival of transplant patients who contract squamous cell cancer.

It has long been recognized that individuals with FA are predisposed to developing squamous cell cancers, particularly of the aerodigestive tract, skin, and cervix. These cancers represent one of the most common malignancies affecting FA patients. Unfortunately, there are no good markers for predicting who may develop squamous cell aerodigestive cancer that can be used for screening purposes in the FA population. At the present time, the following are recommended for individuals with FA:

- Decrease or eliminate smoking and alcohol consumption.
- Have periodic head and neck examinations by an Ear, Nose and Throat surgeon, primary care physician or dentist.
- Seek evaluation of any new abnormalities in speech or swallowing or abnormal appearing areas of the lining tissues of the mouth and throat.

We are interested in studying any biopsy material in patients being evaluated for potential squamous cell aerodigestive cancers. Prior to surgery, please notify Dr. John Wagner or Dr. Margaret MacMillan so that arrangements can be made to obtain part of the biopsy for research studies. Clinical trials, including radiation therapy as well as novel preventative agents, are open or are being planned for FA patients. ♦

# One Family's Journey With Fanconi Anemia

by Andrea & Bob Sacks

We were appalled by some physicians' lack of knowledge about FA when our daughter, Danielle, nearly died at age 5 on the operating table. Her pediatrician said there was nothing wrong with her despite her failure to grow normally, her missing thumbs and her missing radial bones. Her hand surgeon had no clue that her bone marrow was failing. During that near fatal hand surgery, neither the surgeon nor the anesthesiologist looked at a blood count, which would have revealed a platelet count of 8,000. Naturally, we stopped using those providers and took her to a geneticist at Children's Hospital, Washington, D.C., who immediately recognized her condition as textbook FA.

Dani had a successful bone marrow transplant at Johns Hopkins Medical School in 1984 at age nine. Several years ago, we asked a course coordinator there if we could address a medical student class on FA and patient care for orphan diseases. To our surprise, the medical school created an annual lecture for first year medical students on FA. A Hopkins hematologist teaches the first half of the class on medical aspects of this disorder. Then our entire family talks about Dani's symptoms, and our trials and tribulations in navigating appropriate care for her.

A regular portion of our talk describes the occasionally insensitive encounters we have had with hospital residents. We give two examples. Sean, our 12-year-old son, is minutes away from being wheeled down the hall to undergo general anesthesia for harvesting his bone marrow. An unthinking anesthesiology resident tells him, "Now one of the complications of anesthesia is death." Obviously, I expressed my displeasure to

the resident. Later, my wife and I are helping Dani get to the bathroom every few minutes, due to the intravenous flushing of her system with Lasix, needed because of lethal doses of chemotherapy given in preparation for transplant. Just outside her room, in loud conversational tones, a resident is informing a nurse: "Push Lasix through the Sacks kid until she goes into heart failure." That conversation nearly pushed Andrea and me into heart failure. As I caught up with the resident, whose name surprisingly was not Dr. Kevorkian, I asked him to explain his heart failure remark. He said Lasix was necessary to keep Dani's internal organs from becoming dysfunctional due to the chemotherapy. I suggested that patients and parents, unaccustomed to hearing doctors prescribe heart failure procedures on patients, might be upset by his insensitive words, spoken in easy earshot of patients and parents. By the way, the good news for current transplant recipients is that the high dose chemotherapy used on Dani in 1984 is no longer utilized because it is too toxic for FA patients.

Too many doctors fail to practice good medicine due to their own egos. It is gratifying to us that, following the lecture, medical students communicate a high interest in FA and our concerns about patient care. We share resources from FARF with those who are interested. I wouldn't be surprised if a current or future graduate from Johns Hopkins is now more sensitized to the heartaches of FA families. I urge you to try setting up similar lectures at medical schools near your home. Maybe a future FA researcher will have been stimulated by just such a lecture.

As I describe Dani's medical histo-



ry, remember that FA produces a wide range of complications. Many FA children do not exhibit the short stature, missing thumbs, and early bone marrow failure that Dani exhibited before age five. Also, the course of your child's treatment can affect the type of outcome you experience. We don't know how many of Dani's current complications are a result of the high dose chemotherapy she received prior to her bone marrow transplant. Drs. Alter and Auerbach have confirmed that many of her complications have occurred in other FA patients without this conditioning.

Dani's complications include: acquired hydrocephalus which was relieved by a ventricular-peritoneal shunt placed at age 12 (ask the doctor to consider whether the medicine, Theo-Dur, commonly used on asthmatics to help them breathe under general anesthesia, can cause grand mal seizures following brain surgery, as it did on Dani); insulin-dependent and resistant diabetes at age 20; eye cataracts at age 20; macular degeneration of her eyes at age 23; endocrine system failure from birth (supplemented by hormone replacement therapy); hyperlipidemia (extremely high cholesterol levels) from early childhood; abnormal liver enzymes (confirmed as a benign fatty liver via liver biopsy at age 20); cellular abnormalities which are often precancerous

in oral and gynecological membranes (abnormal pap smears in her late teens, with gynecological surgery at age 22), and tongue cancer (removal in April 1999); esophageal abnormality (needs stretching periodically during an endoscopy); and gastrointestinal abnormalities (handled with various anti-ulcer and anti-acid medicines).

During her hospitalization for the transplant, Dani experienced a common, severe fungal infection, treated by amphotericin. To minimize the risk of infection following the destruction of her immune system (Dani received high dose chemotherapy but no radiation), we decided to isolate Dani from children the first year post-transplant. We did home tutoring for a year and a half. Although she had virtually no graft-versus-host disease, her immune system took a long time to regenerate and remained severely suppressed for more than a year.

I would encourage FA families to err on the side of caution and have the FA patient see appropriate specialists on a regular basis. We have followed the advice of Drs. Alter and Auerbach and have Dani regularly examined by her oral-maxillofacial surgeon (monthly or more often if her leukoplakia looks particularly menacing), her gynecologist-oncologist (every six months), her gastroenterologist (every year), her eye specialists (every three to six months), and her endocrinologist (irregularly throughout the year). The doctors also recommend regular endoscopies, since the risk of esophageal cancers is high with FA older children and adults. We also follow Dr. Shahidi's high dose vitamin recommendation of ACES (vitamins A, C and E) which may or may not have any effect on preventing chromosomal mutations, but don't seem to harm her, either.

We all take flu shots every year.

Dani is the subject of a research paper at the University of Maryland Dental School by her oral-maxillofacial surgeon on their experiences with her FA as it relates to leukoplakia and tongue cancer (see article by Dr. Ord, page 4, this issue). She is currently being evaluated for a 3-month NIH study on Cox 1 and Cox 2 inhibitors taken to improve oral leukoplakia (whitish areas in the mouth and on the tongue) and prevent it from becoming cancerous. Doctors are first trying to eliminate a fungal infection in her mouth by using gyne-lotrimin as a mouth lozenge.

Throughout all of this ordeal, Dani manages to lead a fulfilling life, with its ups and downs. She is an

extremely happy, up-beat person to be around, both at work (when she is not seeing a doctor) and at home. Her quality of life is excellent, and her positive attitude has a lot to do with that.

Dr. Chris Walsh, a FARF-funded researcher, told us at the 1999 Family Meeting that he believes FA patients are ideal candidates for gene therapy. Our affected children may one day be the source from which come the cures, not only for FA, but for all cellular-based irregularities of the human body, which is just about every other disease. Since geneticists tell us that every human body has at least five fatal genetic mutations, that should encourage not only FA families, but every one else as well, to support our FARF with a vengeance. ♦

## **We Welcome New Families Who Have Joined Our Support Group**

Brian Horrigan and Amy Levine  
1068 Lincoln Ave.  
St. Paul, MN 55105  
(651) 228-1842  
brian.horrigan@mnhs.org  
amy-l@ospa.umn.edu  
Delia ~ DOB 12/31/87

Tim and Melissa Stichler  
2510 Eastcleft Drive  
Columbus, OH 43221  
(614) 459-8163  
tim1mel@aol.com  
Erika Primmer ~ DOB 8/28/89  
Chelsea Primmer ~ DOB 5/8/87

Anne-Marie Thorstensson and  
Martin Persson  
Betesvagen 9  
903 55 UMEA  
Sweden  
011 90 138556  
am.thorstensson@usa.net  
Josefin Persson ~ DOB 4/27/93

Noreen and John O'Donoghue  
69 Ballinderry Park, Mayfield  
Cork, Ireland  
011 87 20072019  
Lisa ~ DOB 11/17/89  
Jason ~ DOB 2/17/95

Lorraine and Kevin McQueen  
2214 Floyd Ave.  
Richmond, VA 23220  
(804) 359-3841  
Sean ~ DOB 10/25/99

## **Change of Address**

Joe and Susan Leslie  
9901 N. Oracle Rd. #6101  
Oro Valley, AZ 85737

Mike and Pam McCoury  
1826 Keogh St.  
Burlington, NC 27215-1936

# FUNDRAISING

## A Wake-Up Call

by Mary Ellen Eiler, President, FARF Board of Directors

Many of you know that on October 22, Dave Frohnmayer suffered cardiac arrest while attending a conference at the NIH in Maryland. Fortunately, he was in the company of many medical personnel and lived through the crisis. What many of you may not know is that Dave very nearly died. Lynn was told that he would not survive. In fact, his attending physician considers that it was literally a miracle that Dave lived. Most people who suffer cardiac arrest due to ventricular fibrillation, in fact, do die. My point in writing this is not to elicit sympathy for Dave or his family. My point is to elicit grave concern about the ongoing health of the Fanconi Anemia Research Fund.

As Dave's friend of thirty years, I was devastated upon hearing the news of his illness. I knew that it was highly unlikely that he would survive such a medical catastrophe. And, as the president of the Fanconi Anemia Research Fund Board of Directors, I knew that it was equally highly unlikely that the Research Fund would survive if Dave did not survive. I remain extremely concerned about what the loss of Dave would mean to FA patients who desperately need the services of FARF. The harsh fact is that the Frohnmayers provide close to 90 percent of the funding for FARF through their ongoing fundraising efforts. We simply would not be able to continue the Research Fund if the Frohnmayers, for whatever reason, were unable to continue that work.

Leslie has written an article in this edition of the newsletter commend-

ing the increase in FA families who have participated in fundraising projects this year. I enthusiastically add my voice to hers. However, I also must add the cautionary note that both the number of families engaged in fundraising projects and the amount of money they raise are far too small. I urge you to consider it a personal challenge to increase both.

I know that Dave and Lynn, because of Dave's public position, have greater ability than others to raise funds. Yet, while we all cannot raise vast quantities of money, we all can write a fundraising letter to our friends during the holidays. We all

*continued on page 18*

## National Golf Tournament

Chris Collins of Delray Beach, Florida, is working on an ambitious and highly promising fundraising program to benefit both the National Marrow Donor Program and the FA Research Fund. It is a national golf tournament modeled after the successful Oldsmobile Scramble. This tournament would be the only national golf event with benefits going exclusively to charity. The event has already received the full endorsement of the C.M.A.A. (Club Managers Association of America) and has enlisted the support of several celebrities. If you would like to support this event with ideas or by enlisting more support from celebrities and sponsors, contact Chris at 561-637-0853. ♦



## Vicki Athens Honored

Dr. Vicki Athens was selected the 1999 Soroptimist Woman of the Year by Soroptimist International of Wyandotte, Southgate, and Taylor. Athens was honored for her extensive work on behalf of the FA Research Fund. She planned and organized our first Annual Family Meeting, has been a tireless fundraiser for research dollars, and has assisted in organizing medical symposia. As a Board Member, Athens has represented our organization repeatedly at numerous scientific and regional family meetings.

Congratulations, Vicki, on this highly deserved honor! ♦

## FA Handbook Available Soon

The third edition of *Fanconi Anemia: A Handbook for Families and Their Physicians* will be ready for distribution soon. Please contact the FARF office if you would like one or more copies.



# Family Fundraising Efforts

by Leslie Roy

Fundraising is the blood of our organization. Without money coming in, there is no output: no office, no staff, no family support, no publications, no research support, and most important of all, no CURE.

I'm a firm believer that there is strength in numbers. This year 32 families have raised a 1999 total of \$580,292. In 1998, eight families raised a total of \$512,806. These amounts exclude \$2 million given by Phil and Penny Knight in response to the Frohnmayers' letter campaign during 1998 and 1999. It is most encouraging that there was a four-fold increase in the number of families who decided to conduct events or letter writing campaigns during 1999. Families are seeing value in allowing others to help in our crusade to find a cure.

We need to keep the momentum going in 2000. Several families are planning community dinners, bike-a-thons, and races. Our office staff is always available to offer fundraising advice, to send out copies of our fundraising handbook, and to assist families in preparing their letter writing campaigns. Families need to be aware of foundations within their communities and companies. In past years, we have received significant amounts from foundations offering our families matching grants.

During our FA Board's annual planning meeting in January, we discussed fundraising at some length. We asked ourselves, "What are reasonable fundraising goals for FARF?", "How can we increase the amount of money raised by families?", and "Are we managing the funds we have in the most effective ways?" We are determined to be fiscally responsible to each member of

our support group as we seek better treatments and ultimately a cure. **But we need the help of each and every family.**

Thanks to all who made a special effort to raise funds during 1999. We report below on the last six months of 1999 only. Including the Knight gift, we raised \$1,362,634 during this period. This includes memorial donations of \$28,117 and United Way/Combined Federal Campaign contributions of \$7,084.

Funds raised from July 1 through December 31, 1999, were attributed to the following families:

## \$1,200,000

Lynn & Dave Frohnmayer

## \$25,000 - 27,000

Mike & Beth Vangel

## \$21,000 - 25,000

Laurie Strongin & Allen Goldberg

## \$11,000 - 12,000

Chris & Susan Collins

## \$8,000 - 9,000

Chuck & Katy Hull  
Deane Marchbein & Stuart Cohen  
Bob & Andrea Sacks

## \$5,000

Joseph Chou  
Eric & Beth Losekamp

## \$2,000 - 4,000

Andrew & Vicki Athens  
Jeff & Beth Janock  
Steve & Melissa Turner

## \$1,000 - 2,000

Mark & Linda Baumiller  
Diana Fitch & Darryl Blecher  
Antonino & Marie DiMercurio  
Ezat & Laila Faizyar

Gary & Melody Ganz  
Pat & Maria Gleason  
Learnon Keleher  
Robert & Jennifer Kiesel  
Gil & Peggy McDaniel  
Lynn Mendenhall  
Jack & Lisa Nash  
Jack & Tannis Redekop  
Bill & Connie Schenone  
Mark & Susan Trager  
Marc & Sandy Weiner

## Up to \$1,000

Ken & Jeanne Atkinson  
Bedoya Family  
Carl & Stephanie Benshoff  
Gilbert Bodier  
Shirley Carvalho  
Bob & Carole Cavanaugh  
Brian & Margaret Curtis  
James & Carol Dillon  
Ed & Janice Duffy  
Dave & Paula Guidara  
Jackie Hardy  
Brian Horrigan & Amy Levine  
John & Irene Kalman  
John & Karilyn Kelson  
Eugene & Renee Lemmon  
Dennis & Sharon Lower  
Greg & Lynette Lowrimore  
Jack & Pam McCarty  
Mike & Pam McCoury  
Cecelia Meloling  
John & Barbara Miller  
Griff & Cecilia Morgan  
Sheila Muhlen  
Bob & Alice Nicholson  
Ron & Fredi Norris  
Shirley Quilici  
George & Kathryn Reardon  
Glen & Maureen Russo  
Rick & Lynn Sablosky  
Mark Salo  
Jeff & Debby Slater  
Karen Steingarten  
Robert & Lynn Tharp  
Reese & Nancy Williams

*continued on page 18*

## Fundraising

*continued from page 17*

Contributions were received in memory of the following FA patients:

Adam Day  
Danielle DiMarino  
Paige Ellis  
Elizabeth Healey  
Chris Hull  
Carrie Janock  
Lauren Kelson  
DeeDee DePersis-Doutt  
Caleb Cohen  
Michael Elzinga  
Sean Meloling  
Alex Norris  
David Russo  
Amber Garthus  
Marti Turner  
Avi Weiner  
Donna Williams ♦

*“Pray as though everything depends upon God, but work as though everything depends upon you.”*  
- Greg & Gail King

### Use of Logo

This is just 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 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.

## A Wake-Up Call

*continued from page 16*

can ask one relative or friend to write such a letter to their friends. We can take advantage of FARF's offer to help us write the letter. We can keep our ears open to the creative fundraising ideas that other FA families are doing and replicate one of those ideas in our community. We can pay attention to organizations in our local community that donate funds to organizations like FARF, and we can determine with FARF staff the best way to contact them. The bottom line is that all FARF families collectively can raise at least as much money as Dave and Lynn do individually.

I know all too well from my friendship with Lynn and Dave how emotionally and physically exhausting it can be to care for FA-affected family members. I know that there are many days when they would rather do anything than write another fundraising letter or dial the phone one more time. This is a terribly difficult and demanding disease for them, and I know that it is for you too. I wish that no FA family had to worry about fundraising or about anything associated with this disease. That is not the reality, however.

We clearly dodged a Fund-ending bullet last October 22nd. We were the beneficiaries of a miracle, not, incidentally, the first we have received. The first miracle was that the Frohnmayers raised funds for the creation of the Fanconi Anemia Research Fund as a resource for FA-affected families in the first place. The second miracle was that Dave lived through his cardiac arrest to continue to provide leadership to this organization. We cannot afford to count on a third miracle.

The FA Research Fund is heeding

this wake-up call and is going to focus this year on moving the Fund into a financial position that will ensure its survival for all FA families, regardless of the Frohnmayers' participation. We will need your help like never before.

Finally, I'd like to make a special plea to those FA families who have successfully gone through a bone marrow transplant. The increased success of bone marrow transplantation has been an enormous blessing to our FA community. Yet, because FA patients are now living longer due to better medical treatment and to successful bone marrow transplants, we are now realizing that successful bone marrow transplants are not the end of the struggle for FA patients. Instead, older FA patients are extremely likely to get cancers, such as head or neck cancers, as they move into young adulthood. And, because of the underlying disease of Fanconi anemia, these patients are far less likely than others to survive such a cancer. While it is tempting after a successful bone marrow transplant to breathe a huge sigh of relief and get back to "normal life," the reality is that Fanconi anemia has a "one-two punch," the second one being these solid tumors. FARF is facing this reality and is seeking to fund research in this area. We need your help in this effort.

To all FA families, the FARF staff stands ready to provide whatever assistance you may need in individually fundraising for FARF. I feel confident that, together, we can meet the challenge.

*“The act of doing fundraising can be healing for families.”*

- Manuel Buchwald

## Family Regional Meetings

by Leslie Roy

The Meyer Memorial Trust in Oregon has given us a grant to establish regional networks to offer family support and to work together to improve the outcome for FA patients. Many families are unable to attend our annual family meetings due to scheduling, health, and financial problems. Regional mini-educational and support meetings allow us to support the needs of many more families.

Meetings consist of a presentation on treatment, by video or by a treating physician, a session on regional fundraising issues, psycho/social issues, and time for families to meet and share experiences. Meetings are on Saturdays over an eight-hour period.

FA is a rare disease and many families never meet another FA family. I remember Lynn Welfare Mendenhall telling me she thought she was the only FA patient until she found our support group at age 42. She knew then she couldn't give up the struggle. She had much to share with other families and much to gain from knowing them. She learned there was a group of scientists working on

behalf of FA patients around the world, and she volunteered to give blood, skin, and tumor samples. As she became educated, she became empowered.

Education enables families to fight this disease. Educated parents and patients can make knowledgeable treatment decisions with their doctors and not feel powerless.

The success of these meetings depends on the participation of families. If you have suggestions concerning content of a meeting or you would like to assist the FA staff in preparing a meeting in your region, contact the FA office.

### Meetings scheduled over the next few months are:

- The Southeastern region on February 26 in Tampa, Florida at the Holiday Inn City Centre.
- The Midwest region in April in the Detroit area. Specific date and location to be announced.
- The Mountain region on June 3 in the Denver area.
- All regions at our annual family meeting in Wisconsin in August ♦

## Mark Your Calendars

*continued from page 1*

minute drive from the airport in Milwaukee, Wisconsin.

We will follow last year's format of having two days of science presentations held on the weekend, followed by two days of psycho/social sessions. Families can attend either session or stay for the entire meeting.

The science meeting will begin with dinner on Friday, August 11, and end early afternoon on Sunday, August 13. Researchers and treating physicians will address topics such as FA 101 (a session for those families new to this disease), post-transplant issues, guidelines concerning treatment, gene therapy, unrelated transplant protocols, and the return of a popular session from last year: gastrointestinal problems with FA patients.

The psycho/social portion of the meeting will begin Sunday afternoon, August 13, and continue through Wednesday morning, August 16. Leslie Roy and Nancy Cincotta will lead discussions relevant to coping and living with FA, sibling issues, and bereavement issues. Small group discussions will be scheduled for teens, older FA patients, and children with FA.

A comprehensive, age-related children's program will be offered during all presentations for the duration of the entire meeting. Evening activities will include a magic show, karaoke, and nightly bonfires. The meetings have proven to be highly educational, emotionally supportive, and fun!

Families have received information related to cost and a pre-registration form. Contact the FA office for additional information or with any questions. We hope to see all of you in Wisconsin this coming August! ♦

### **The Twelfth Annual International FA Scientific Symposium**

## **OCTOBER 27-29, 2000**

### **Novotel Hotel, Amsterdam, The Netherlands**

We thank researchers at the Free University, Amsterdam, for their generous invitation. This location will strengthen our outreach into the international research community, and make it possible for a larger number of European researchers to attend. Contact Joachim or Merrie at the FARF office for details.

## New Staff

### Jan English

During the last year, we experienced some turnover among our staff. We are happy to announce that Jan English joined us on a temporary basis in November 1999, as half-time office assistant. Her presence has made a big difference, and we are pleased that she has made an open-ended commitment to staying with us.



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### Merrie Garoutte

For seven years, Merrie Garoutte was the Executive Director of a local Big Brother Big Sister program. As of January 1, 2000, she joined our staff as half-time administrative assistant. We are delighted to be benefitting from her extensive experience. She also brings with her considerable experience in the field of grant writing.

### Susan Castillo

Susan Castillo joined us in January 2000 as Director of Development. She will devote an average of 20 hours per week in a pilot project to explore new fundraising venues by building on her extensive experience as a TV journalist and Senator in the Oregon State Legislature. We are thrilled to have her on board. ♦

