



# FAMILY NEWSLETTER

#30 A Semi-annual Newsletter on Fanconi Anemia for Families, Physicians, and Research Scientists Fall 2001



## Families Learn, Share Experience, and Have Fun at Annual Family Meeting

Families from all over the world arrived eagerly in Williams Bay, Wisconsin, for the 11th annual FA Family Meeting. This meeting, held on the campus of Aurora University on the shores of Lake Geneva, brought together FA families and eminent FA researchers and clinicians for a three and one-half day meeting filled with science, practical advice, new friends, and fun.

Thirty-seven FA families with a total of 41 children from Germany, Norway, Argentina, Brazil, Canada, and the United States attended this event. Old-timers were awed by the number of new families at the meeting. This year, the Fund was able to offer eight scholarships to new and old families alike, so many more families were able to attend. The new families

clearly added to the strength of the meeting.

Again this year, Rob Sawyer and the members of his teen group from Rotary International in Trenton, Michigan, provided childcare and a recreation program for the children who attended. This group of exceptional teenagers raised money via events throughout the year to finance their trip to Lake Geneva. Rob also brought a number of wonderful and energetic adult volunteers this year. *Our sincere thanks to our Trenton, Michigan, friends.*

The speakers at this meeting were outstanding. You are strongly encouraged to read the *Science Letter* and Medical News section of this *Newsletter* for detailed information on all medical and scientific presentations.

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## Medical Controversy Emerges at Family Meeting

An FA transplant expert sparked controversy and intense discussion by recommending that FA patients with matched, unrelated donors should, in most cases, go to transplant without undergoing androgen treatment. This subject dominated discussion among parents and treating physicians at the summer meeting and did not produce a clear consensus among those in attendance.

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## Harris Presents Guidelines for Treatment of FA

At our August 2001 Family Meeting, Richard Harris, Children's Hospital, Cincinnati, presented an overview of different treatment options for FA patients, including the benefits and risks of each option (see *Science Letter* for details).

Harris urges patients with matched sibling donors to go to transplant when platelets fall below 50,000. Combining statistics from all transplant centers, between 65% and 85% of patients are now alive two years or more following a matched sibling donor (MSD) transplant. Cincinnati has now performed 30 of these transplants. Ten patients received prior androgen therapy. Twenty-five are currently alive, for an overall survival rate of 83%.

The options are more complicated for the 75% of patients who lack a sibling donor. Harris discussed transfusions, androgens and cytokines, outlining the advantages and drawbacks of each option. For those choosing transfusions, he urged the use of designated donors and blood filters, to

minimize the risk of patient sensitization to blood products. Harris stated that it is the number of different donors and not the number of transfusions that can lead to complications at the time of transplant.

Harris noted that androgens often work for years and there is a low risk of early death. The disadvantages are that androgens can cause serious side effects (adenomas, adenocarcinoma, and lakes of dilated blood vessels in the liver) and reduce the chance of a good outcome of a later transplant. For most patients, Harris is no longer recommending use of androgens prior to a matched unrelated transplant.

Cincinnati has now performed eight unrelated donor transplants using the new fludarabine protocol. Five patients received bone marrow; three received peripheral blood stem cells. Harris does not recommend cord blood for unrelated transplants. Of these eight patients, six are alive from three to twenty-five months post-transplant. One death was due to chronic



graft-versus-host disease (GVHD) in combination with an aspergillus infection; one was due to pulmonary failure. Three patients experienced significant GVHD. Harris estimates that between 60% and 70% will now survive an unrelated donor transplant. The high risk of dying within the first three months (estimated at around 30%) is obviously the greatest drawback to an unrelated transplant. The primary advantage is that a successful transplant is curative of the bone marrow complications of FA. ♦

## Reducing Toxicity – Transplant Results in Germany

Wolfram Ebell of Children's Charité Hospital, Berlin, reported on 17 FA transplants at our recent Family Meeting. By eliminating radiation and cyclophosphamide, Ebell hopes to reduce secondary tumors and toxicity. Engraftment is achieved by using fludarabine, high stem cell doses and T cell antibodies.

Six patients received stem cells from matched sibling donors; eleven had alternate donors. Eight patients had severe aplastic anemia, six patients had myelodysplastic syndrome and three

had leukemia at the time of transplant.

All six patients with matched sibling donors, and seven of eleven with alternate donors survive. Of the four who died, three died of infection and one of leukemia. Toxicity was very limited. Five patients suffered graft failure after the first transplant. Four of these patients engrafted after the second transplant, and one after the third. Ebell has modified his protocol in an effort to prevent graft failures. Mean follow up post-transplant is 18 months. See *Science Letter* for details.

Ebell believes that it is still too early to recommend an unrelated transplant before a trial of androgens. He notes that many patients respond well to these hormones for a long time. Almost all of the patients he has transplanted have been on androgens for extensive periods of time. Ebell has not seen serious complications related to androgen usage during transplantation, and does not consider androgens a risk factor for transplantation. ♦

## Wagner Advocates Earlier Transplantation For Patients with HLA Matched, Unrelated Donors

At our August, 2001 Family Meeting, John Wagner, University of Minnesota, discussed the use of androgens in the treatment of bone marrow failure, and recent survival outcomes at his center following the use of the conditioning agent fludarabine. Based on his latest data analysis, he now recommends that most patients with HLA matched, unrelated donors proceed to transplant without first undergoing a trial of androgens.

Androgens are successful in increasing blood production in approximately 50% of FA patients. Red cells, white cells and platelets can all be affected. Many patients can taper the dosage of androgens and still maintain stable counts, but eventually these drugs are no longer effective. Wagner outlined the many side effects of androgen therapy (see *Science Letter*), and his concern that androgens may be associated with lower rates of survival following bone marrow transplantation. Androgens delay the time to transplant, increasing the likelihood of infections and other complications. In some cases, androgens have been implicated in organ toxicity during transplantation. Several studies cite androgens as a risk factor in transplantation outcomes.

In the past, graft failure was the major cause of death in FA patients undergoing an unrelated bone marrow transplant. (In 1998, Wagner reported that approximately 39% failed to engraft). Between April 1999 and July 2001, 23 patients at the University of Minnesota were conditioned with a new protocol, which included the drug fludarabine. Primary neutrophil engraftment was achieved in 20/20 evaluable patients. Probability of one-year survival was 46%. However, when survival outcomes were analyzed according to several risk factors,

outcomes were strikingly different between patients classified as “standard risk” and those who were deemed to be “high risk” patients. Standard risk patients had aplastic anemia with or without abnormal clones, or early signs of myelodysplasia, and no history of major infections. High risk patients had infections, myelodysplasia or leukemia at the time of transplant. Of 14 standard risk patients, 12 survive. Of nine poor risk patients, only two survive.

For the first time, survival rates comparable to those of matched sibling donor recipients can be achieved, at least short term, in FA patients who come to transplant without infections, myelodysplasia or leukemia. Because of this, and considering the adverse effects of androgen therapy, Wagner advocates earlier transplants for most patients, before initiating a trial of androgens.

Wagner believes that androgens should be considered for patients at higher risk for problems with unrelated donor BMT. These patients include adults, those with HLA mismatched donors, and those with pre-existing significant organ dysfunction. Androgens may also be used to delay transplantation in families attempting *in vitro* fertilization and pre-implantation genetic diagnosis in an effort to have a healthy, HLA matched sibling.

Because of the poor survival rate in high risk patients, Wagner is changing his protocol for this population. He will eliminate total body irradiation and use busulfan, fludarabine, cyclophosphamide and ATG in an effort to improve outcomes. ♦

## Alter Questions Changing Guidelines

At our recent Family Meeting, Blanch Alter, National Cancer Institute, questioned the wisdom of changing the guidelines established at the 1998 Consensus Conference concerning androgen therapy for patients with matched, unrelated donors. As transplant technology improves, a change in direction may well be justified. At the present time, however, Alter believes that there is insufficient evidence to make this change in policy.

The Conference recommended that patients lacking a matched sibling donor begin androgen therapy when one of the following is present: Hb <8g/dl or symptoms from anemia; platelets <30,000/mm<sup>3</sup>, and neutrophils <500/mm<sup>3</sup>. Alter stated that the hemoglobin usually improves in >50% of patients within one to three months, and platelets in a smaller proportion by 4 to 6 months. Neutrophils may or may not respond, and improve more slowly than the other cell lines. Alter stated that there are patients who have been treated with androgens for up to 40 years, as well as patients who have taken androgens for 5 to 10 years, after which they no longer needed androgens to maintain blood counts.

The side effects of androgens are potentially extensive, although many are reversible when the treatment is discontinued (see *Science Letter*). Alter addressed the concern that androgens are implicated in poor transplant outcomes. While androgens are often used by patients with low neutrophils, they are not the cause of infections. And inadequate monitoring can lead to higher dosages than necessary, increasing the likelihood of liver toxicities. While acknowledging the worrisome side effects of androgen therapy, Alter believes that this drug still plays an important role in the management of FA. ♦

## Five Patients Transplanted with Mismatched Donors Do Well

Farid Boulad, MD, Memorial Sloan-Kettering, reports that he has now transplanted five FA patients using related (4 patients) or unrelated (1 patient) mismatched donors. Two were mismatched at two antigens; three were mismatched at one antigen. For the first patient, this was a second transplant; for the subsequent four patients, it was a first transplant. Dates of transplant were 5/27/98, 3/31/99, 1/20/00, 10/26/00 and 4/5/01. Except for the patient receiving a second transplant, patients received total body irradiation. Protocols included fludarabine and cyclophosphamide. Four patients received G-CSF-mobilized peripheral blood stem cells and one patient received bone marrow stem cells. All stem cells were T cell depleted to prevent graft-versus-host disease (GVHD).

All five patients engrafted early (between days 9 and 11 post-transplant). Immune reconstitution occurred at 6-8 months for three patients and 12 months for one patient. The

fifth patient cannot yet be evaluated. All five are alive and well; the first three patients are alive three years, two years and one year post-transplant, while the last two are alive 10 months and 5 months post transplant. No patient has yet experienced acute or chronic graft-versus-host disease. The third patient had post-transplant complications (CMV infection and EBV lymphoma), which have been resolved.

Most of these five patients were considered very high risk. The first patient had aplastic anemia. Patients 2, 3 and 4 had myelodysplastic syndrome (MDS) with abnormal morphology, increased blasts and cytogenetic abnormalities. Patient 5 had morphological evidence of MDS only. She had no cytogenetic abnormalities and no increase in blasts. The fifth patient was age 21 at the time of transplant. Her donor was a one antigen mismatched unrelated donor, and she engrafted with no GVHD. She has had candida and herpes infections;

Boulad writes that it has been “very tough” getting her through these complications.

The number transplanted under this protocol at Memorial Sloan-Kettering is very small, but the outcomes to date are encouraging. Boulad continues to be optimistic about these very promising results. ♦

### 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.

**The Thirteenth  
Annual**

## **International FA Scientific Symposium**

**NOVEMBER 14-17,  
2001**

**Hilton Hotel  
Portland, Oregon**

## Centers Eliminate Radiation Hoping to Reduce Later Malignancies

Researchers and treating physicians have long suspected that transplant patients have a high risk of later cancers, especially of the head and neck. Among the risk factors are the use of radiation and the development of GVHD. In an effort to reduce the risk of later malignancies, several transplant centers, including Hadassah University Hospital, Israel, University of Minnesota, Cincinnati Children's Hospital, and University Hospital Charité, Berlin have eliminated radiation for some or all of their FA transplants.

For FA patients with matched, related donors, the University of Minnesota has replaced radiation with fludara-

bine and ATG. Between November 1996 and June 2001, 12 FA patients were transplanted with this fludarabine regimen. All engrafted and no patient developed severe acute GVHD or chronic GVHD. Two patients later lost their grafts, but were successfully transplanted a second time. One patient died from a malignancy two years after transplantation. The remaining eleven patients are alive and well. Long-term follow-up is needed to determine if this approach reduces the risk of later malignancies. ♦

## Preimplantation Genetic Diagnosis

Allen Horwitz, MD, PhD, the Director of Medical Genetics at the Reproductive Genetics Institute (RGI) in Chicago, spoke at the Family Meeting regarding Preimplantation Genetic Diagnosis (PGD). The Institute was in the news last year with the birth of Adam Nash, whose cord blood was used to transplant his sister Molly, who has FA. In PGD, unaffected embryos can be tested before implantation in association with *in vitro* fertilization.

Dr. Horwitz emphasized that FA families do not have to travel to the RGI in Chicago frequently for this procedure. The RGI will work with *in vitro* fertilization clinics around the country, making it unnecessary to travel to Chicago until the last week of the process.

Although it is preferable that the specific DNA mutation be known prior to undergoing PGD, for FA-A it may be possible to perform linkage analysis on parents and affected and unaffected children to help in the FA diagnosis part of the PGD. At the present time the DNA mutation needs to be known for the other FA complementation groups.

The time required for PGD, not including the 40 weeks for a pregnancy, varies greatly dependent on the individual, but is between 18 and 36 weeks. This includes 12 to 24 weeks to custom develop the DNA testing required to select an embryo free of FA and to select an embryo that will be an HLA match for the FA-affected child. This also includes 6 to 12 weeks for the stimulation of the mother's ovaries for the harvest of the ova used in this process.

Dr. Horwitz reported that there is about an 18% chance of an unaffected match in a given batch of fertilized eggs, and 30% chance of a pregnancy in a cycle via PGD if two unaffected

matched embryos are available. The average cost for the first cycle is \$18,000. Subsequent cycles cost \$12,000 - \$14,000 each. In the experience of RGI, many insurance companies will pay for the IVF part of the cycle only if the couple is infertile. With rare exceptions, insurance will not pay for the genetic testing.

According to Dr. Horwitz, the risks of PGD are the risks that are related to the *in vitro* fertilization procedure because it is a medical/surgical procedure. In addition, the *in vitro* process could result in multiple pregnancies. A third risk factor is that a PGD misdiagnosis could result in the birth of a

child with FA and/or one who is not an HLA-match for the FA-affected sibling. Dr. Horwitz reported that RGI has a 98% accuracy rate in selecting an embryo via PGD. He reported that the unknown effects of biopsies are the fourth risk of PGD.

For inquiries about PGD, contact can be made through hematologists and geneticists or directly by contacting:

Christina Masciangelo, MS, Genetic Counselor  
or Dr. Horwitz at:  
(773) 472-4900  
fax (773) 871-5221  
e-mail: rgi@flash.net ♦

## Gene Therapy Trial Produces First Encouraging Results

Christopher Walsh, University of North Carolina Gene Therapy Center, reported on results from an ongoing FA gene therapy trial at his center (see *Science Letter*). Four FA-A patients, ranging in age from 11 to 48 years, have entered this trial. Three of the four patients had severe pancytopenia requiring blood transfusions and/or androgen support.

Early blood stem cells, called CD34+ cells, were mobilized, either from the patient's peripheral blood or from a bone marrow harvest. Bone marrow produced far more cells than the peripheral blood, but the number of cells collected was only a fraction of the number expected from a person with normal bone marrow (from 1/10 to 1/100). In the laboratory, cells were then transduced with the normal *FANCA* gene, using a retroviral vector, and returned to the patient.

All four patients tolerated the procedure without adverse complications.

Analysis of blood and bone marrow samples revealed that two of the four patients had significant long-term gene transfer.

A year and a half after gene transfer, one patient shows evidence of a clinical response to gene therapy. This response has become increasingly impressive with time. At the time of gene transfer therapy, this patient had 2,000 white cells, hemoglobin of 9, and 120,000 platelets. Today, this patient has 5,000 white cells, hemoglobin of 12, and 240,000 platelets. This patient has had a significant increase over time in the number of peripheral blood cells carrying the *FANCA* gene. This result is extremely encouraging.

Walsh has now developed new vectors based on the lentiviral virus system and believes these will be superior to the vector he is using in his clinical trial. He is requesting patient bone marrow samples, and anticipates future clinical trials using the new vectors. ♦

## Oral Cancer Precautions

At the FA Regional Meeting in Columbia, Maryland, on June 16, Stephen Engroff, MD, DDS, Chief Resident at the University of Maryland Cancer Center, Oral and Maxillofacial Surgery, presented information on behalf of Robert Ord, MD, DDS, from that department. Ord has extensive experience with oral cancer in FA patients and recently co-authored a landmark article in the *Journal of Oral and Maxillofacial Surgery* entitled "Squamous Cell Carcinoma of the Tongue After Bone Marrow Transplantation in a Patient with Fanconi's Anemia."

Dr. Engroff reported that:

- Over 90% of oral cancers are squamous cell carcinoma.
- There are five types of premalignant lesions in oral cancer. Three of these (submucous fibrosis; lichen planus; and discoid lupus) are unlikely to occur in FA patients. The remaining two, leukoplakia and erythroplakia, are implicated in oral cancers of FA patients.
- Leukoplakia is the most common pre-malignant lesion. Dr. Engroff defined leukoplakia as "a white patch or plaque of oral mucosa that cannot be rubbed off and cannot be characterized as any other disease." Other white patches that may occur in the mouth, such as thrush or candida, can be rubbed off.
- Leukoplakia is painless.
- In the general population, there is up to a 17% risk of conversion of leukoplakia to squamous cell carcinoma per year. This may be higher in the FA population.
- If leukoplakia is identified, the dentist or oral surgeon can apply a solution of toluidene blue to the oral cavity to help identify areas that contain cells undergoing abnormal growth. Toluidene blue will stain premalignant or malignant cells, thereby helping the oral surgeon determine the area of biopsy.
- Erythroplakia is a red patch of oral mucosa. With erythroplakia, more so than leukoplakia, the changes that have occurred in the tissues of the mouth are more frequently dysplasia, carcinoma in-situ or frank carcinoma. Erythroplakia that has formed in a leukoplakia is particularly worrisome. Engroff recommends a biopsy if erythroplakia is suspected.
- In the general population, oral cancers commonly occur on the floor of the mouth, on the sides of the tongue, and the lower lip. These are areas where saliva pools, or, in the case of the lower lip, areas exposed to the sun. However, in FA patients, oral cancers also occur on the top of the tongue and elsewhere in the oral cavity.
- Treatment varies depending on the location and extent of the lesion, but often involves surgical or laser excision of the lesion. Laser excision does less damage to the non-cancerous tissue surrounding the lesion. Studies on other treatments, such as mouthwashes that may be effective and on photodynamic therapy are currently in progress.

Based on this information, Dr. Engroff recommended that FA patients take the following precautions regarding oral cancer:

- In the absence of any danger signs, have a complete dental examination at least once every six months, starting very early in the child's life. Dental examinations must occur more often if there are any suspicious lesions.

- Make certain that the dentist is aware that the patient has FA and that FA patients are at extremely high risk for oral cancer.
- Ask the dentist to examine the entire oral cavity, not just the teeth or the gums surrounding the teeth.
- If a suspicious lesion is identified, consultation by the dentist or referral to an oral surgeon with expertise in FA would be extremely helpful.

Dr. Engroff emphasized that, if premalignant lesions are identified early, treatment may prevent the progression to cancer. If carcinoma does develop, cure rates are much improved when lesions are treated at an early stage. Thus, frequent and thorough check-ups by a dentist well versed in the high risk of FA patients for oral cancer are critical. ♦

### More Information in Science Letter

Families frequently seek guidelines concerning how and how often to monitor liver function while a patient is on androgens. The reader is referred to the article by John Wagner on page 6 in the *Science Letter* for guidelines on this subject.

Blanche Alter presented helpful guidelines to families on how and when to screen for cancer in FA patients at our recent Family Meeting. She also discussed androgens and screening for liver dysfunction. The reader is referred to her articles on these subjects in the *Science Letter*.

## Oral Health and Bone Marrow Transplantation – Some Precautions

Dr. Mark Schubert, Director of Oral Medicine, Seattle Cancer Care Alliance, is quoted at length in a July 2001 article in *Center News*, a publication of the Fred Hutchinson Cancer Research Institute. He discusses the importance of oral health as it relates to bone marrow transplantation.

Chemotherapy and radiation given prior to transplantation can lead to oral mucositis, an inflammation and ulceration of the moist tissue lining the oral cavity. Mucositis can be worsened by preexisting dental disease and conditions such as trauma or irritation. Schubert strongly recommends a thorough oral examination and treating oral problems prior to transplantation, in the hope of lessening the

severity of mucositis.

Chemotherapy and radiation can leave a patient without a healthy immune system. This sets the stage for bacterial, fungal and viral infections. Schubert notes that we all have bacteria that live inside our mouths. If there is a way to gain entry into the blood, microbes usually considered harmless can cause serious problems. Particularly worrisome is the potential systemic spread of infection to various organs. Severe mucositis can make the spread of infection more likely.

Schubert also recommends that patients not get their teeth cleaned for six to nine months after transplant. “With the immune system still recovering, the spraying and splashing



involved with routine dental treatment can result in patients inhaling bacteria and debris that can significantly increase the risk of pneumonia. If patients have graft-versus-host disease, routine dental treatment often needs to be delayed longer.” ♦

## Grant to Fund International FA Transcriptome Consortium

The FA Research Fund has just awarded a two-year grant to Grover Bagby, Jr., MD, to establish the International FA Transcriptome Consortium. Bagby is Director of the Cancer Institute, Oregon Health Sciences University, Portland. The Transcriptome Consortium builds on the recent sequencing of the human genome and on the collaboration of several bone marrow transplant centers as a result of the Bone Marrow Transplant Conference (sponsored by the FA Research Fund) in Chicago in April 2001.

Five transplant centers are participating in the Transcriptome Consortium: Curitiba, Brazil; Capetown, South Africa; University of Minnesota; Children’s Hospital, Cincinnati; and St. Jude’s Hospital, Memphis, TN.

The purpose of this project is to study the causes of bone marrow failure and leukemia in FA. This will be

done by studying the pattern of gene expression in normal bone marrow cells and in cells from FA patients in various complementation groups. The pattern of gene expression in marrow cells can be measured by the “**transcriptome**” (see below). Differences in the transcriptomes of bone marrow cells from children with FA and with both FA and myelodysplasia may reveal clues to the molecular cause of leukemia in patients with FA.

Each human cell contains about 30,000-50,000 genes, but only some of these genes are active (expressed or turned on) at any one time in any cell type. An active gene is one that is producing messenger RNA (mRNA). The production of mRNA by a gene is called **transcription**. The array of all the various mRNAs produced by the cell at one time is called a **transcriptome**. The mRNAs carry the genetic

blueprints from the genes to the part of the cell that translates the blueprints into proteins. It is the proteins that carry out many of the essential functions of the cells.

Using modern “DNA chip” technology, researchers will place on a single small silicon chip the DNA sequences that code for all normal human mRNAs. They will then add mRNA from normal bone marrow, as well as cells collected from FA patients in different complementation groups, including those with bone marrow failure, myelodysplasia, and AML. Researchers will be able to visualize and compare which genes are turned “on” and “off” in normal and FA cells. By comparing gene expression in different types of cells, researchers hope to reveal important pathways for targeted therapy. ♦



# 20th Anniversary of the International Fanconi Anemia Registry (IFAR)

by Arleen D. Auerbach, Ph.D.

In 2002 The International Fanconi Anemia Registry (IFAR) will have its 20th anniversary. For this occasion, we are attempting to get a complete clinical update on all patients in the Registry. When my laboratory at The Rockefeller University founded the IFAR in 1982, we didn't yet know anything about the genetic complexity of the disease (at least 8 complementation groups and many different mutations within groups). The IFAR was started with the goal of identifying a large number of patients with this rare disorder in order to study the clinical and genetic features in FA. We hoped to better define this heterogeneous disorder and differentiate it from other syndromes with overlapping features.

Questions relating to diagnosis, natural history of the disease, prognosis, treatment, and cancer incidence in FA are being addressed by the IFAR studies. Information regarding genotype-phenotype correlation is being obtained; we hope this will help to determine the physiologic roles of the cloned genes. We have recently updated our laboratory website, and information on the IFAR can be found at <http://www.rockefeller.edu/lab-heads/auerbach/auerbach.html>.

To date, the IFAR has registered 755 patients from North America affected with FA, as well as many patients from Brazil and other countries. Diagnosis was confirmed by hypersensitivity to the clastogenic effect of DEB in all cases. We have determined the complementation group for 341 of the North American patients. The distribution of IFAR patients is FA-A: 207; FA-D2: 2; FA-C: 78; FA-F: 8; FA-G: 46. We are actively determining complementation groups for additional patients by functional complementation with retroviral vectors

and by mutation screening. Mutations are recorded in the Fanconi Anemia Mutation Database: <http://www.rockefeller.edu/fanconi/mutate/>. It would be helpful for this research project for any family in the IFAR (or willing to be registered in the IFAR) to contact me by e-mail, mail, or by phone (ask for Kathy Mah, our genetics counselor). We are able to work with families in the IFAR to help them obtain information they need regarding preimplantation genetic diagnosis (PGD)/*in-vitro* fertilization and other prenatal testing, as well as carrier testing, where appropriate.

As part of the IFAR investigation, two new studies have recently been initiated. The Grandparent Study, in collaboration with Marianne Berwick, PhD, an epidemiologist at Memorial Sloan-Kettering Cancer Center (MSKCC) in New York, investigates whether carriers of FA have an increased risk to develop cancer. For this study, we are asking grandparents of individuals diagnosed with FA to participate. Participation involves sending a blood sample to our lab for carrier testing and filling out a health survey and consent forms. If you would like more information about this study, please contact Katherine Mah in Dr. Auerbach's office.

Dr. David Kutler and Dr. Bhuvanesh Singh (Laboratory of Epithelial Cancer Biology, MSKCC) are collaborating with the IFAR to investigate a growing body of evidence that suggests a relationship between FA and cancer, particularly squamous cell carcinomas. Many patients with FA who survive into adulthood develop multiple oral cavity and vaginal squamous cell carcinomas at a relatively young age and have a poor prognosis. Dr. Kutler is working with the IFAR in

order to investigate the incidence, prevalence and the risk of squamous cell carcinoma in patients with FA. Applying the knowledge gained from this initial clinical project, we will begin to investigate the genetic and molecular changes that cause this increased incidence of cancer. Our hope is to develop human and mouse models to explore the genetic relationship between this disease and epithelial cancer. If you are interested in participating in this study or have questions concerning Fanconi-specific screening and treatment of cancer, contact Dr. Kutler.

## Contacts

To register in the IFAR:

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(ask for Kathy Mah)

For information about the Grandparent study:

Katherine Mah, genetics counselor  
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212-327-8862  
[mahk@mskcc.org](mailto:mahk@mskcc.org)

For information about the Cancer Study:

Dr. David Kutler  
[kutlerd@mskcc.org](mailto:kutlerd@mskcc.org)



## Bone Marrow Transplant Conference Held in Chicago



The FA Research Fund sponsored a one-day meeting of bone marrow transplant physicians at the O'Hare Hilton in Chicago on April 28, 2001. Physicians from eight countries and five continents attended this landmark meeting, which allowed physicians who perform FA transplants to share their protocols, learn from one another, and collaborate in the future.

Although participants did not agree on a uniform transplant protocol for FA patients, the meeting was distinguished by a high level of collaboration and the willingness of all to share their experience, listen, and, in some cases, modify a protocol because of the information presented at the meeting. A number of transplanters at this meeting will collaborate in the immediate future on a project entitled the International Fanconi Anemia Transcriptome Consortium sponsored by the FA Research Fund (see page 7). ♦

The participants at this meeting, which was also attended by several members of the FARF Board of Directors, Ralf Dietrich from the German FA group, and Lorne Shelson from FA Canada, were:

Grover Bagby, Jr, MD, Oregon Cancer Institute, OHSU, Portland;  
Farid Boulad, MD, Memorial Sloan-Kettering, New York;  
Martin Champagne, MD, Hôpital St. Justine, Montreal;  
Joachim Deeg, MD, Fred Hutchinson Cancer Center, Seattle;  
Ygal Dror, MD, Hospital for Sick Children, Toronto;  
Wolfram Ebell, MD, Children's Charité Hospital, Berlin;  
Alfred P. Gillio, MD, Hackensack University Medical Center; New Jersey;  
Eliane Gluckman, MD, Hôpital St. Louis, Paris;  
Helmut Hanenberg, MD, Heinrich Heine University, Düsseldorf;  
Richard Harris, MD, Children's Hospital Medical Center, Cincinnati;  
Mary Horowitz, MD, IBMTR/ABMTR, Milwaukee;  
Peter Jacobs, MD, Constantiaberg Medi-Clinic, Capetown, South Africa;  
Hans-Peter Kiem, MD, Fred Hutchinson Cancer Research Center, Seattle;  
Leslie Lehmann, MD, Dana-Farber Cancer Institute, Boston;  
Alexei Maschan, MD, Children's Hospital, University of Moscow, Russia;  
Jose Zanis Neto, MD, Federal University of Parana, Curitiba, Brazil;  
Shimon Slavin, MD, Hadassah University, Jerusalem, Israel;  
Franklin Smith, MD, Indiana University School of Medicine;  
Joanne Van Burik, MD, University of Minnesota Medical School;  
John Wagner, MD, University of Minnesota Medical School, Minneapolis;  
Donna Wall, MD, Cardinal Glennon Children's Hospital, St. Louis.

## Medical Controversy Emerges at Family Meeting

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The *Standards for Clinical Care* handbook, developed at a May 1998 medical conference, provided guidelines concerning the timing of bone marrow transplantation for Fanconi anemia patients. Survival outcomes for patients having matched sibling donors had improved steadily over the years. When the *Standards* were published in 1999, the survival rate for patients transplanted after 1997 was 83%. Patients with severe aplastic anemia and a matched sibling donor were advised to go to transplant after the onset of bone marrow failure, and before attempting other therapies.

However, transplantation from an alternate (i.e., mismatched related or unrelated) donor then was associated with relatively poor survival (~30%). For this reason, transplanters recommended that other treatment options, such as androgens or hematopoietic growth factor therapy, should be attempted first. Transplantation was recommended after the failure of these therapies, or after evidence of myelodysplasia or leukemia.

At our August 2001 Family Meeting, John Wagner, University of Minnesota, proposed a change to these guidelines. He now believes that most patients with bone marrow failure and an HLA-matched unrelated donor should go to transplant prior to the use of androgen therapy. He cited greatly improved transplant outcomes in "standard risk" patients and concerns that androgens may be an added risk factor. (See Wagner article, page 3). Richard Harris, Children's Hospital, Cincinnati, later concurred with Wagner's recommendation.

Wolfram Ebell, Children's Charité Hospital, Berlin, stated that milder conditioning regimens should mini-

mize toxicity related to prior androgen therapy, and recommends transplantation only after the failure of androgens. Blanche Alter, National Cancer Institute, stated that androgens may not always be the cause of transplant problems, but may be suffering from "guilt by association." Androgens do not cause infections, but may be used by patients with severe neutropenia who are at greater risk of infection. While not denying the importance of androgen-induced liver problems in transplant, Alter suggests that the judicious use of androgens has a definite role in the longevity of many patients.

Many parents whose children's blood counts have been stabilized on androgen therapy expressed anxiety and confusion following the presentations. Androgens might buy time, and time could bring less toxic transplant preparatory regimens and better outcomes. Greatly improved statistics are no guarantee that any one child will

survive transplantation, and children doing well at present risk losing everything. Unrelated FA transplant outcomes are much better, but the total number of children transplanted is small and the time post-transplant is still short.

Some parents were concerned that a child's prior use of androgens would jeopardize the transplant outcome. It should be noted, however, that almost all of the children transplanted to date with alternate donors had taken androgens, some for many years. Many of these patients appear to be long-term transplant survivors.

Your editors advise readers who are contemplating transplantation with alternate donors to read articles related to this subject in this *Newsletter* and in the *Science Letter*, and to discuss these issues with your treating physician. Remember that positive news—improved transplantation outcomes—has led to a close examination of this subject. ♦

## Cincinnati Children's Opens FA Center

Drs. Richard Harris and David Williams have announced the opening of the Fanconi Anemia Comprehensive Care Center at Children's Hospital Medical Center in Cincinnati. Harris, the Medical Director of the BMT Outpatient Clinic, has extensive experience in the transplantation of FA patients. Williams, Director of Experimental Hematology, has extensively researched blood stem cell biology and gene therapy.

Experts in the following areas of specialty will design individual treatment regimens for patients: bone marrow transplantation; cardiology; endocrinology; genetics; hematology; nephrology and urology; orthopedics and hand surgery; and psychosocial support.

The Center will offer a thorough evaluation, including confirmation of diagnosis and genetic typing, HLA typing, and multi-system evaluation. Ongoing care can be provided on site or through consultation with the referring physician. The Center will also continue to research and evaluate new FA therapies, and FA patients are invited to participate in the clinical trials conducted by the Center.

For more information, call the FA Care Coordinator at (513) 636-3218 or write

CHMC

3333 Burnet Avenue

Cincinnati, OH 45229-3039. ♦

# REGIONAL MEETINGS

## Regional Meetings

The FA Research Fund has held regional meetings in Chapel Hill, NC, Waltham, MA, and Columbia, MD, since the first of the year. These meetings, funded through a grant from the Meyer Memorial Trust in Portland, Oregon, are designed to bring FA families together to hear the latest research and clinical information on FA and to encourage families to raise funds for FA research and services.

### University of North Carolina at Chapel Hill

This meeting was held on Saturday, March 31, at The Carolina Inn on the campus of UNC, Chapel Hill. Six families, including twelve family members, attended. Christopher Walsh, MD, PhD, of UNC, Chapel Hill hosted this meeting and made a presentation on the results of his FA-A gene therapy trial to date. He also gave the group a tour of his laboratory. On this Saturday, many of his staff were diligently working in the lab, and FA family members commented that it was heartening to see researchers so hard at work on FA research. Back at The Carolina Inn, the group heard an excellent presentation on Hematology 101 by Andrew Eichenfield, MD, from Mt. Sinai, and a presentation on fundraising for FA research by Kevin and Lorraine McQueen, FA parents from Richmond, Virginia. At the end of the day, Nancy Cincotta, MSW, led the group in a networking and debriefing discussion.

### Dana-Farber Cancer Institute, Harvard University

Alan D'Andrea, MD, from Dana-Farber was featured at this meeting on



*Family meeting in Columbia, Maryland*

Saturday, May 19, at the Best Western in Waltham, Massachusetts. Ten families, including 28 family members, attended. D'Andrea discussed his recent landmark research regarding the gene pathway of FA, and described the services available at the Fanconi Anemia Center that he has established at Dana-Farber. In addition, Eichenfield made a helpful presentation on understanding blood counts, and Nancy Cincotta led the group session at the end of the day. Mike Vangel, an FA parent and a member of the FARF Board of Directors, made an excellent presentation on fundraising for FA research.

### National Cancer Institute/Johns Hopkins/University of Maryland

This regional meeting, held on June 16 at the Sheraton in Columbia, Maryland, featured Blanche Alter, MD, MPH, from the National Cancer Institute (NIH). Alter, well-known to the FA community, made a presentation on FA 101 for the newcomers in the group, and Gregory Kato, MD, from Johns Hopkins followed with an excellent presentation on Hematology 101.

In the afternoon, Alter presented the results thus far of her epidemiological studies on FA and cancer and on prevention and early detection of cancer. Alter reinforced the fact that FA patients are affected by cancer at a much earlier age and with significantly greater frequency than the general population. Steven Engroff, MD, DDS, chief resident of the Oral and Maxillofacial Surgery at the University of Maryland Medical School, discussed oral cancer in FA patients. As in Chapel Hill, Kevin and Lorraine McQueen made an excellent presentation on fundraising. The McQueens are new to FA, in that their son, Sean, is just two years old. However, they have already established an impressive fundraising effort for FA research in Richmond, through a "casino night" that they held last year and which they have scheduled again for this October. Once again, Nancy Cincotta generously volunteered to lead the networking/debriefing discussion. A total 13 FA families, including 29 family members, attended this highly informative regional meeting. ♦

## Spain Holds National Symposium on Fanconi Anemia

Spain held its First National Symposium on FA on September 10, 2001 in Madrid. The purpose of this meeting was to optimize efforts directed towards the study and therapy of FA in Spain.

Eighty participants attended this meeting. Scientists studying the molecular and cellular biology of FA, treating physicians, families of FA patients and representatives of the Ministry of Health and Consumption actively participated in the symposium.

Manuel Buchwald, PhD, Hospital for Sick Children, Toronto, discussed the biology of FA, and Irene Garcia-Higuera, Dana-Farber Cancer Research Institute, Boston, reviewed her studies on the functional interaction of FA proteins. Twelve speakers from Spain presented studies currently being conducted on the biology, diagnosis, and clinical management of FA patients.

The Spanish FA registry includes 54 patients. Thirty-two families have been studied for complementation group, and mutations have been identified for a few families. This meeting promoted extensive collaboration between clinicians and groups involved in the molecular diagnosis of this disease.

Participants, including FA families, are very enthusiastic about creating a National Working Group on FA. Carmen La Huerta, mother of an FA patient, volunteered to interact with other FA families worldwide and with specialists working on this disease. Speakers and family members agreed to summarize and distribute the conclusions of the symposium. ♦



*Attendees at the UK FA Family Meeting at Hammersmith Hospital in London, May 2001*

## First FA Family Meeting held in United Kingdom

FA families in the United Kingdom (UK) held their first family meeting on May 19, 2001 at the Hammersmith Hospital, London. Thirty participants attended this event.

Chris Mathew, PhD, Guys Hospital, and Marc Tischkowitz, MD, Guys Hospital, described their research efforts. Inderjeet Dokal, MD, Hammersmith Hospital, gave an overview on FA and current therapies. Marcus

Carr, medical student and FA patient, agreed to produce a fact-sheet on FA for new families.

Leslie Roy led a discussion group on coping with FA. A website at [www.fanconi-anaemia.co.uk](http://www.fanconi-anaemia.co.uk) will provide British families with information, news, and links to other websites.

Thanks to FA parent Gail Richardson for organizing this successful event! ♦

## Family Support Coordinator Joins Staff

In April 2001, Suzanne Lauck became our new Family Support Coordinator. Suzanne has a BS degree in Nutritional Science and a BS degree in Nursing. She has her RN license in both California and Oregon. From 1988 to 1993, Suzanne worked for the Jenny Craig Weight Loss Centers in the San Francisco area, as a trainer and manager for three centers. She then received her nursing degree and worked from 1995–2000 as a nurse in the Adult Medical Unit, the Newborn Nursery, and the Neonatal Intensive Care Unit of El Camino Hospital in Mountain View, California. Besides skills in nutrition and nursing, Suzanne



has experience working with and speaking to groups. We are delighted that Suzanne has joined our staff! ♦

## Surviving a Difficult Transplant

by Debby Slater

Our son Nick was diagnosed with FA at the age of 5, and eventually was put on oxymetholone. The medication raised his counts for 7 years, and then suddenly stopped working. He became transfusion dependent for platelets and red cells within 2 months.

When Nicholas was 14, we were told that he needed a transplant. Our lives were shattered. We were scared.

Our hematologist told us about Dr. Farid Boulad at Memorial Sloan-Kettering Cancer Center in Manhattan. Before we even met with Dr. Boulad, he ordered extensive blood work on our oldest son Jason, who turned out to be a 5 of 6 match for Nick. When we finally met Dr. Boulad, we immediately liked him. He is a very warm, caring person, which comes through

in his work as a physician. Dr. Boulad always spoke with confidence that Nick would be fine. A transplant date was set for January 20, 2000.

To say the least, we were scared and so was Nick. I tried to make every day special, but it was hard. Dr. Boulad always answered our questions, and we were told to call him anytime, day or night. He also helped Jason, who was scared.

Nick was in the hospital for 7 weeks from day one when his central line went in, until the day we were released to go to the Ronald McDonald House. Nick had daily CBCs and IV meds at the clinic. Nick had the usual side effects from radiation and chemotherapy: diarrhea, nausea, painful mucositis, and high blood pressure. Every-

thing was controlled by medication, but he didn't eat or drink for weeks because of the mucositis. Nick returned to the hospital 3 times, but for only 4 days each time.

By +80 days post-transplant, however, Nick still was not feeling well. We learned that he had Epstein-Barr virus (EBV) lymphoma. Some of the engraftment was lost, and he was in the hospital for 5 weeks, on transfusions and antibiotics. I was not doing well. The doctors found a psychiatrist who gave me meds to help me sleep. Nick was also having a hard time and got meds to help with his anxiety.

Dr. George is a transplant doctor and her specialty is EBV, so she was instrumental in Nick's care. Nick was

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*Amy Frohnmayer at the Family Meeting*

## Is There An Effective Alternative to Oxymetholone?

by Lynn Frohnmayer

For many years, our daughter Amy's blood counts were declining gradually but noticeably. By the summer of 2000, they were approaching levels where treating physicians often recommend therapeutic intervention. Given our concern about masculinizing effects, we dreaded putting Amy on oxymetholone.

At our 2000 Family Meeting, we spoke at length with the Sablosky family. Their daughter, Lindsey, had been first on oxymetholone and later on danazol (or Danocrine), an attenuated androgen. The Sabloskys stated that Lindsay's blood counts responded

equally well to both drugs. However, she experienced far fewer masculinizing effects on danazol (see *Family Newsletter #28*). After consulting with several families and physicians, we decided to attempt a trial of danazol.

On 10/13/00, Amy's blood counts were: Hgb 8.0; WBC 1.8; and platelets 26,000. We began with a dosage of 100mg/day. Believing that Amy was tolerating this drug extremely well and hoping for greater stimulation of her bone marrow, Amy's physician gradually increased the dosage of danazol to 200mg/day over a four-month period.

*continued on page 17*

# Forrest Lee Engel

1952-2001

by Frederic B. Engel

Forrest, our first child, was born 8 July 1952 in a dark, dismal, creaky, old hospital in Deadwood, South Dakota. Although his entry weight was four pounds four ounces, he was only a few days short of his estimated time of arrival. His birth was normal except for a loop of umbilical cord draped unceremoniously about his neck. During the ensuing weeks his weight dropped to three pounds fifteen ounces. Then, ever so slowly, his weight began to increase.

His mother, Elsie, was discharged from the hospital after a few days convalescence. Our young, sincere but inexperienced obstetrician decided that it would be best if Forrest remained with the hospital staff until he reached at least five pounds. Three weeks later with his weight hovering at about four pounds six ounces the perplexed, discouraged, and embarrassed staff suggested that we take our son home and try our luck at fattening him up. At that time Forrest fit comfortably between the palm of my hand and my inner elbow. And so I, with the confidence of the mentally myopic, and Elsie, trembling with dread and apprehension, brought Forrest home to our tiny two bedroom share-the-bathroom apartment in Sturgis, South Dakota.

Without the rudimentary know-how that comes with babysitting the diaper-clad set, obviously we had a lot to learn. The following months were filled with constipation, diarrhea, colic, congestion, rashes, and idiopathic disorders which apparently had not been included in the medical curriculum of our two local doctors.

While Elsie and Forrest sat at home or in the doctor's waiting room practicing crying duets, I performed my

duties as a Manual Arts Therapist at Veterans Administration Neuropsychiatric Hospital down the road at Fort Meade, South Dakota. I often wondered which part of my day was frying pan and which was fire. I lightheartedly told a coworker that I strongly suspected that Forrest had acquired a copy of Doctor Spock's child rearing book, which he cunningly used to find age appropriate afflictions to spring on us. Or perhaps he surreptitiously joined a malady-of-the-month club, which, at regular intervals, shipped him succulent lozenges inoculated with the causative agents for every disorder listed in the Center for Disease Control logs. My attempts at humor seem terribly out of place in light of all that awaited us.

Nineteen fifty three ended with Forrest slowly putting on ounces when he should have been gaining pounds; my accepting a position with the Department of the Army at Camp Atterbury, Indiana; and Elsie, poor little Elsie, doing her best to adapt to Forrest's medical machinations. During the Christmas holidays we moved to tiny Trafalgar, Indiana. (Try to find that on the map).

We thought Forrest reached his first birthday exhibiting all the physical and mental calisthenics of a one-year-old. In retrospect, it is difficult to imagine that we did not see the impending disaster or disasters through our too optimistic eyes. Or why friends, family, nurses, and doctors didn't notice Forrest's lagging maturation. We were all deaf and dumb to the warning signs. A case in point: I was sitting on our steps entertaining Forrest, it was probably more that he was entertaining me, while Elsie, now pregnant with our second child, was at our doctor's for a routine maternity checkup. A young



*Forrest (right) with friend Ethan*

lady of 16 years, the wife of a 60-year-old gentleman from the hills of Kentucky, approached us. She silently studied Forrest for a few moments then looked at me and gravely said, "Thars something wrong with yur youngin. His eyes is too small." She turned and continued on her way. I had to agree with her observation; his eyes were small. I foolishly reasoned his eyes as well as all his dimensions were proportional to his diminutive size. Relieved and comforted by my faulty rationalization, I buried the girl's observation under the weight of life's more urgent duties and responsibilities.

Several weeks later, a flare up of one of Forrest's many resident bugs occasioned another visit to the family doctor. I sat "patiently" and waited. Elsie came out of the exam room and through a flood tide of tears told me that the doctor wanted to talk to me. The moment I entered his office, the doctor informed me that Forrest was not developing properly at all. His weight was the least of our problems. He was making an appointment for us with a specialist and, "Goodbye and good luck." The large billboards planted at either end of our village adver-

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## From a New Mom

by Audrey Barrow

Last week at my brother's block party, there was a neighbor, Harold, whom I couldn't wait to see. I wondered what I would say—would he really remember us? I shook my head and thought how different this meeting would be compared to the last time we spoke, when he told us about an illness, Fanconi anemia.

Dr. Harold Duroseau is the hematologist who cared for Kelsey when she was admitted to the hospital for abnormal blood counts. After Dr. Duroseau told us about Fanconi anemia, I remember whispering, "is this 'LT' (life threatening)?" I couldn't bring myself to say the words. With a tear he nodded his head. He referred us to Dr. Lipton, whom he felt had the experience necessary to treat this disease.

One year after diagnosis, thank God and every other saint and relative in heaven, Kelsey is doing great. Dr. Lipton has been able to give Kelsey a life she can enjoy. We opted for G-CSF therapy, and Kelsey's RBCs and Hb have been raised from almost nothing, into the normal range. The G-CSF shots were very difficult at first, mostly from fear of the needle. But when we started using EMLA cream 45 minutes before the injections, and a lollipop while giving the injection, that problem was solved.

Kelsey hates to end her summer vacation, but looks forward to starting the soccer season; she has her goalie gloves all ready. One year after diagnosis, I have learned that although so much has changed forever, one thing has not. Kelsey is "just" a child and looks at me as "just" her mom. She doesn't care how much I know about hematology or stem cell research. She wants me to make her feel safe, not

frightened with all of the changes going on in her body. Our daughter has taught me how important it is to her, for me to see this illness through her eyes. Sure, she knows this anemia is part of her life, but it isn't her whole life. Playing and enjoying the moment are what children live for. She can give me her worries, and I give them to God, so we can enjoy our lives together.

A year after diagnosis, I am not afraid to talk about the future—what she'll be when she grows up, college, and marriage. How dare I not talk to her about her future? This is a mistake that no one should make. As we all know, no one knows what the next day holds. ♦

## A First Step Towards Cancer Surveillance

by Lynn Frohnmayer

At our recent Family Meeting, Blanche Alter, MD, recommended that cancer surveillance of the head and neck begin at age ten, or within the first year after a transplant. Heeding her advice, we took our daughter, Amy, to an otolaryngologist for an examination.

The doctor conducted a thorough examination of Amy's mouth. She then inserted a flexible scope into Amy's nostril and down her throat. She took several pictures of the mouth and throat for later comparison. Amy described the procedure as uncomfortable but not at all painful. At present, all appears to be normal. We will repeat this simple procedure on an annual basis. ♦

## Lynn Welfare's Music Available on CD

Many of you had the pleasure of knowing Lynn Welfare. For those of you who did not, she was an adult FA patient who touched the lives of many with her indomitable spirit. Lynn had a deep love of music and, before she passed away in July 2000, she fulfilled one of her dreams, that of recording a compilation of her own music on a CD entitled "Flyin' High."

The Fanconi Anemia Research Fund has just received a wonderful donation of the remaining CDs recorded by Lynn. Lynn's brother John and her mother Susan have expressed their desire to fulfill an important wish that was held by Lynn—to have her music heard. Lynn's CD is available through the Fanconi Anemia Research Fund and is free of charge. The CD might make a special and unique addition to fundraisers.

### 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.



# Cancer Surveillance Crucial Following Bone Marrow Transplantation

by Richard Briga

*Richard Briga, age 44, was diagnosed with Fanconi anemia when he was 39. His successful bone marrow transplant at the University of Minnesota in 1998, diagnosis with throat cancer in 1999, and subsequent surgery and radiation therapy are described in our Family Newsletter, #27. Richard has contributed valuable information and important advice to our e-mail group, which we include below:*

Before age 39, I felt great, and was a health fanatic. I used to compete in mountain bike races up to 100 miles in length, run 20 miles in one afternoon, and backpack in the mountains for days, carrying a 55-pound backpack in sub-zero conditions. My resting heart rate was in the forties. I ate no animal products for years and still eat very little. How many people do you know who didn't eat a candy bar for over two years? I was totally obsessed with fitness. I always tried new techniques, food and potions to increase speed and strength.

I tell you this because when you're not having problems, you might feel immune, and it's easy to let your guard down. I speak from experience. You can do very well after transplant and can be cured of your bone marrow problems. You can feel that you have conquered FA, can feel invincible, and believe the worst is over. This may sound harsh, but I must speak the truth for the benefit of everyone. The worst may be yet to come. FA is totally relentless and does not let up, so neither can we.

In my opinion, the solid tumor issue is even more frightening than the blood disease aspect of FA. In most cases, treatment for blood diseases is not disfiguring and the side effects not

as severe. Head and neck malignancies can be extremely painful and lead to a host of permanent complications. My bone marrow transplant was a picnic in the park compared to the treatment I endured for cancer.

I strongly recommend regular surveillance, early detection and treatment for cancer. I was followed closely after transplant and my doctors examined my mouth every 2 weeks to check for GVHD, but my tumor was too low to be detected without endoscopy. **My advice to adults with FA would be to see an ENT doctor at regular intervals for endoscopy.**

I believe that FA has made my skin more sensitive to sunlight. I have had over a dozen lesions removed from my face and arms that were caused by solar damage, about half before transplant. One of them was cancerous. It appeared as a small red crusty spot that never cleared up. A biopsy confirmed the diagnosis. I don't trust sunscreen because I've used it on my arms and they still get solar damage. It's better to cover up and use hats and long sleeves. Never assume a lesion is harmless and see a doctor if anything questionable appears. Because I had total body irradiation prior to transplant, and because I have had numerous skin lesions, my hematologist recommends total body evaluations by a dermatologist every six months.



*Richard on his solo backpacking trip*

I see a dentist every three months. I don't produce adequate saliva so I am more susceptible to tooth decay and gum disease. I get frequent lumps and sores inside my mouth; my mouth tissue always seems to be in the process of healing. I fear that someday a lesion won't heal properly and will become cancerous.

It is a mistake to believe that a bone marrow transplant cures FA. Nothing could be further from the truth. Patients are at extreme risk of cancers and need cancer screening and close follow-up. To neglect this is an invitation to disaster. ♦

## Is There An Effective Alternative to Oxymetholone?

*continued from page 13*

Nine months after initiating therapy and believing the response was adequate, he then decreased the dosage to 150mg/day, then to 100mg/day.

On 9/22/01, Amy's blood counts were: Hgb 10.9; WBC 3.1; platelets 47,000. She has experienced some side effects from this drug. Her voice is slightly lower, and she has experienced depression, which she relates to taking danazol. She has had no facial hair, no acne, or any other masculinizing effects from this drug. Liver ultrasound is normal; liver enzymes are in the normal range.

We realize that very few FA patients have been treated with danazol. Apart from a few anecdotal cases, we do not

know how effective this drug is in increasing blood counts of FA patients. The few patients on danazol report very limited masculinizing effects. Since this drug has never been evaluated in a clinical trial, these results are only anecdotal. We have no information on its toxicity in FA patients.

We do not regret our choice for Amy. In general, she has tolerated danazol extremely well. In her case, we believe it has substantially increased her blood production. We hope the lower dose will eliminate her depression. We will continue to monitor her blood counts, bone marrow, liver and liver enzymes on a regular basis. ♦

## E-Mail Group Discusses EMLA Cream, Blood Draws

*by Suzanne Lauck*

Families have posted several messages to the e-group regarding the usefulness of EMLA anesthetic cream prior to blood draws. EMLA is a combination of lidocaine 2.5% and prilocaine 2.5%, which are both local anesthetics. The cream is applied to clean, dry skin and then covered with an occlusive dressing (one that does not permit air to enter) for at least 60 minutes prior to a procedure.

Several studies have been performed that show effective pain reduction with the use of EMLA cream before needle sticks. FA patients using EMLA cream have experienced pain relief when undergoing inoculations. However, many parents noted vasoconstriction (narrowing) of the blood vessels when EMLA is used before venipuncture, making the procedure more difficult. An FA parent/physician agreed that EMLA cream does cause vasoconstriction.

When EMLA cream is applied, initially there is a blanching or whitening of the skin, which may be due to vasoconstriction. However, when the cream is left in place for more than two hours, erythema or reddening of the skin occurs, which may be due to vasodilation. This raises the question of whether the amount of time the cream is left on is related to the difficulty with drawing blood. Families using EMLA prior to blood draws might wish to raise this question with a physician or phlebotomist.

Parents agreed that the experience and finesse of the individual performing the procedure had a significant effect on the comfort level of the child undergoing venipuncture. Several stated that it is advisable and perfectly acceptable to request a specific individual who is capable of drawing blood with the least amount of pain and trauma. ♦

## Surviving a Difficult Transplant

*continued from page 13*

given 2 doses of Jason's T cells, which had been inoculated with the EBV. Within 2 weeks of receiving the T cells, Nick showed signs of improvement, and a biopsy showed that the T cells were doing the job we had prayed for.

We stayed in Manhattan for almost 6 months, which was fine with me because I felt safe being close to the hospital and the doctors. When we finally came home (which is 3 hours driving time from Sloan-Kettering) I had Dr. Boulad's long-range beeper number and used it often. Dr. Boulad was wonderful with answering my endless questions. We initially saw Dr. Boulad every other week, and then once a month. Now, at home, Nick has a CBC drawn once a month.

When Nick got home we found a psychiatrist for him to see, and he has been able to get off the meds for anxiety and depression. When Dr. Boulad gave us the O.K., we found a 2-year-old golden lab for Nick. The two are inseparable.

Nick is doing well. For the first time in two years, he is back in school full time. The days are longer than he is used to, but this doesn't seem to affect him.

It was my instinct as a mother that led me to choose Sloan-Kettering with Dr. Boulad for the transplant. I knew that if we went someplace else, we would not have brought Nick home. With the EBV I know in my heart that this is true. I will always be grateful that God sent us in the direction of Dr. Boulad. The transplant team was wonderful. They were always there when I needed them for a hug or a talk. ♦

## Forrest Lee Engel

*continued from page 14*

tising the doctor's flourishing real estate business should have tipped us to his cavalier attitude toward his Hippocratic oath.

The Indianapolis appointment was made and kept. Ushered into the specialist studio with little formality, our alleged expert stripped Forrest to his diapers. After a cursory perusal he announced, "Your son is what we in the profession call a Constitutional Defective. He cannot be educated. The state hospital will not accept him until he is five years old. Don't waste your time searching for miracle cures." He pointed at Elsie's bulging stomach and added, "You're doing the only thing you can do. Have another child. It will take your mind off this boy." He closed with, "Your chances of having another defective child are 1000 to 1."

Our return to Trafalgar seemed like a one vehicle funeral procession in our well-worn 1942 Nash Ambassador. I remember interrupting our sorrowed silence only twice. Once to assure Elsie that because of my degree in education and my training and experience at Fort Meade, Forrest would NEVER be institutionalized and, later, to state my belief that if God selected parents for a new-born he could not have made a better choice for Forrest.

The following months were a study in quiet desperation. Elsie worried herself into a painful peptic ulcer. Camp Atterbury was being decommissioned. I accepted an offer to accompany the 31<sup>st</sup> Infantry to Camp Carson, Colorado. In the meantime, our second child was born with a horror of fatal flaws. He was rushed to John Whitcomb Riley Hospital in Indianapolis where he expired within 18 hours of his arrival. We were told at that time that the odds of having a normal child were now reversed. It was really 1 in 1000

that we'd have a normal child. From then on we held Forrest closer still. I was sure that I would never laugh again.

Our move to Colorado was uneventful. Our mission at Fort Carson was so fouled up by my predecessor that there was no possibility of my making things worse which lessened the stress of the new job. We found a neat little apartment between Manitou Springs and Colorado Springs. To fulfill my promise to Elsie about looking after Forrest, I immediately set search for an isolated, wooded home site where Forrest would not be subjected to the taunts, jeers, ridicule and embarrassing stares of the ill mannered.

Because of the 6,600-foot elevation and dry, unpolluted air, Forrest's chronic respiratory congestion disappeared within weeks of our arrival. Due to this healthful atmosphere, the moderate seasons, and the lure of the many recreation activities available, Colorado Springs was filled with more professional services than one could imagine. Things were looking up!

When it was time for Forrest's booster shots we chose a pediatrician associated with Colorado Springs Medical Center. With what I can only attribute to divine intervention, we selected Dr. John Kanas, a young, intense, light-hearted, jovial genius. At Forrest's first visit, Dr. Kanas administered the required immunizations. He then carefully examined Forrest from stem to stern. The doctor's first words to us were delivered with a slight degree of annoyance, "What are you doing for this boy?" We outlined Forrest's medical history and described the diagnosis/prognosis of the Indianapolis specialist. Dr. Kanas responded, "Why that (deleted)!" I was shocked to hear a physician use such expletives to describe a fellow member of his sacrosanct profession. He thought that much could be done for Forrest. He



*Forrest and his dog Patches*

thought Forrest had Fanconi Aplastic Anemia. Forrest's deformed thumbs, his many café-au-lait spots and the unusual separation at the base of his toes pointed to this conclusion. He asked if he might have Forrest admitted to a local hospital for a few days so an extensive medical workup could be done. With that report in hand, we could take Forrest to Children's Hospital in Denver.

Dr. Kanas unquestionably saved Forrest's life at that time and went on to extend it to an age, 48, that, even now, is among the record breakers for FA. Many years later, while Forrest was a guest at Rockefeller University Hospital in New York City, an old doctor told us that it was absolutely remarkable that in 1954 a young doctor out there in the wilds of Colorado would recognize the signs and symptoms of a disease as rare and obscure as FA.

In the early morning hours of the following week, Forrest was admitted to Children's Hospital and was whisked away into its labyrinthine corridors. Late that afternoon Forrest reappeared. We joined members of the hospital

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## Forrest Lee Engel

*continued from page 18*

staff in a small classroom. The spokesman confirmed Dr. Kanas' diagnosis of FA. They'd found the medical literature concerning FA very scarce. There was mention of a research physician at Harvard Medical School having some success stimulating the bone marrow with hormones. It appeared that there were only a few incidents of the condition on record and that, unfortunately, it appeared the life expectancy with this anemia was nine years at best.

At our next appointment with Dr. Kanas, he insisted that a psychiatrist evaluate Forrest. The exam was not for his benefit nor for Forrest's but rather to assure us that Forrest was a mentally fit two-year-old. During the meeting with the psychiatrist, Forrest wandered off to look at books in the office. The doctor ended the interview right there. He pronounced Dr. Kanas right again. There was nothing wrong with Forrest's ability to learn.

We built a home on eight wooded acres at the southern base of Cheyenne Mountain. We attempted to adopt a child so Forrest would have the enjoyment of a sibling relationship, but we were turned down because we had an "unusual child" in our home.

We celebrated Forrest's birthday and other holidays as if they would be his last, though for the most part we attempted to give him a normal childhood. He owned mini-bikes, pocketknives, a telescope, and too many books. He rode the rural school bus. He graduated from high school on schedule in 1971. He worked at the main library at Ft. Carson for five years. He owned three VW bugs over the years. Since he was four foot five inches, the brake, clutch and gas pedals were extended. Driving allowed him to meet his many friends and admirers

for lunch in town. I was always very impressed to be in the company of a real celebrity. Anyone who met Forrest remembered him.

After living with FA for years, one day in 1989 Forrest heard something about his disorder on Dan Rather's evening news program. A few days and many phone calls later, I was speaking to David Frohnmayer. He could hardly believe that Forrest had reached his 37th year. I couldn't believe that after 37 years Forrest, Elsie and I were not alone. There was even A FOUNDATION to support the kids and their families.

I end this towering babble knowing very well that I need not relate more of Forrest's experience because the readers have lived or are living the rest of the story. I compare Forrest's struggle with FA: the terrible side-effects of his alleged allies, the drugs he daily ingested, each doctor's appointment, surgery, emergency room visit, CAT scan, MRI, EKG, hematological blood letting, X-raying, and medical specialist's report, to heart-pounding combat patrols. Would this sortie be the one that pronounced the unthinkable? Or would there be that exhilarating sigh of relief that came with the knowledge that he had made it; dodged the bullet once again. There would be another brief stand down before the next skirmish with death.

My dearly beloved son suffered his miserable unwarranted fate without a whimper, a whine or a discouraged word. He was a hero, unsung because I lack the eloquence to disseminate the thoughts that arise in me. Like me, I'm sure you believe that your loved one deserves a Medal of Honor and that the citation that accompanies this prestigious recognition should contain the words bravery, courage, and valor in the face of overwhelming obstacles. ♦



## *In Loving Memory*

**Forrest Engel**  
7/8/52-5/24/01

**Christopher Siebenthal**  
1/13/89-7/20/01

**Jake Siniawski**  
6/25/90-3/17/01

**Meshael Richardson**  
6/20/86-6/19/01

**Mark Quilici**  
11/5/71-7/28/01

**Kyle Simpson**  
9/24/90-4/17/01

**Sarah Ninja Dietrich**  
1/23/80-9/15/01

**Joshuah Abraham**  
5/7/92-9/16/01

**Dacoda Millard**  
5/10/96-5/26/01



*FA Family Meeting founder, Vicki Athens, was honored at the recent Family meeting.*

## FA Family Meeting Founder Honored

Dave Frohnmayer, Vice-President of the Board of Directors of the FA Research Fund, and Mary Ellen Eiler, Executive Director, presented Vicki Athens, DPM, with a Certificate of Appreciation in honor of her founding of the FA Family Meeting in 1991. Vicki planned and organized the first family meeting, which was held in a hotel in Orlando, Florida. One hundred people attended that meeting. Our Fund has held successful annual family meetings ever since.

Dave and Mary Ellen thanked Vicki for her role in recruiting Rob Sawyer and the members of the youth group of the Trenton, Michigan Rotary to provide the child care program at Lake Geneva. They also recognized Vicki for her ongoing dedication to raising funds for FA research. Vicki is the spark behind numerous fundraising events each year in Michigan that have raised over \$100,000 for the Fund in the last four years alone. In addition, Vicki serves with distinction on the FA Board of Directors. Our thanks to Vicki for facing this disease head-on and for doing all that she can to face it down. ♦

## Welcome To Our New FA Families

Mike Brannock

303 Snow Camp Dr.  
Apex, NC 27502  
home: (919) 367-9096  
work: (919) 678-8423  
mbrannock@financialdirections.com

Kerrie Brannock

105 Queens Knoll Ct.  
Cary, NC 27513  
home (919) 461-0068  
work (919) 531-6637  
kerrie.brannock@sas.com  
**Jason~DOB: 8/5/90**  
J.D.-DOB: 3/15/86  
Daniel-DOB: 8/5/94

Randal and Stephanie Race

1305 Legault Blvd.  
Ortonville, MI 48462  
(248) 969-6097  
raceptg@aol.com  
**Haley~DOB: 7/20/00**

Jeffrey and Donna Boggs

11824 Lyons Fork Road  
Coeburn, VA 24230  
(540) 395-5712  
dboggs62@hotmail.com  
**Nicholas~DOB: 9/10/98**

Mike and Catherine Sanders

4409 Camberly Street  
Port St. John, FL 32927  
(321) 632-6032  
cat.net@juno.com  
mike.sanders@airgas.com  
**Ty~DOB: 12/30/91**  
Pierce~DOB: 6/25/93

Bruce and Kerry Timperley

P.O. Box 22  
East Greenwich, RI 02818  
(401) 364-9462  
btimperley@aol.com  
**John ("Jack")~DOB: 2/5/99**

Jerome and Blenda Dahlin

Box 153  
820 2nd Avenue NE  
Kenmare, ND 58746  
(701) 385-4975  
blender@restel.net  
**Dillan~DOB: 5/11/92**  
Dustin~DOB: 3/2/96

Donny and Danielle Burkin

278 Queensland Blvd.  
Perrysburg, OH 43551  
(419) 874-0795  
dburkin21@netzero.net  
**Hope~DOB: 5/9/00**

Karina Villegas and Jorge Daniel Rana

Canale 2461  
Jose Marmol  
Pcia. De Bs. As. (1846)  
Argentina  
kvillegas@arnet.com.ar  
**Guillermina~DOB: 7/16/00**

### New Guide

A Fanconi Anemia Medical Resource Guide will be included in the 2002 International Family Directory. It will include contact information for transplant centers, testing labs and other FA-related services.

# FUNDRAISING



Pat, Maria, Brenna & Amanda Gleason, Marie & Fabrizio DiMercurio

## Beef and Beer Benefit Raises Over \$10,000

by Pat & Maria Gleason

On April 21, 2001 our family held a fundraiser for FARE. We published flyers that I took to work, passed around the neighborhood and distributed at St. Timothy's Parochial School which Amanda attends. The flyers announced "Beef 'N Beer Benefit – Help Amanda Gleason Fight Fanconi Anemia." Over 280 people attended the affair, which was held in St. Timothy's gymnasium/hall. We provided soda, beer, hot roast beef sandwiches, sausage and meatball sandwiches, salads, chips, pretzels, coffee and tea. A DJ played music for listening and dancing. There were door prizes, 50/50 raffles, baskets of cheer raffles, and a Chinese auction. We gave away prizes for over an hour. All the prizes were donated. We had foursomes for golf, weekend getaways, jewelry and much more. A good time was had by all. Many people have asked about

next year, which is already being planned.

Not only did we raise over \$10,000 for FARE, we regained a small sense of control (and therefore mental health), which Amanda's diagnosis took away from us. Having seen how far science has come since Amanda's diagnosis gives us the knowledge that every penny raised is helping in a big way. This allows us the luxury of feeling as if we are helping Amanda the best way we can to a brighter future. What parent would not want to have a college fund for his child? Maybe this \$10,000 will be just the ticket needed that will allow medicine to progress and help Amanda to live long enough to go to college. I recommend that every family who can, *do a fundraiser*. Not only for their affected child's benefit, but *for their own sanity*. It has helped us tremendously. ♦

## Canadian Fundraiser

by Annette Waxberg & Lorne Shelton

On May 27 we held a fundraiser for Fanconi Canada at the Comedywood Comedy Club in Thornhill, Ontario called "Some Entrancing Evening." The event was a huge success. We sold over 180 tickets and raised over \$17,000 for Fanconi anemia research!! Six FA families attended the event, including our honorary FA Canadian family, the Blechers from Pittsburgh.

With proceeds from this event, plus \$21,000 raised by the Cronin family during a golf tournament two weeks ago, and the fundraising done by families across Canada, we now have the means to continue our three-year partnership with the Canadian Government for a Fanconi Anemia Post-Doctoral Fellowship. We recently awarded this fellowship to Dr. Sylvie Pasco of The Hospital for Sick Children, who works in the laboratory of Dr. Madeleine Carreau. We are also considering a grant for another Canadian Research Project.

Since 1995, our family has been trying to fundraise for FA. Our first effort raised over \$200, and we thought we were doing great. Things have snowballed since then. If you had told us then that we could have raised \$17,000 in one fundraiser, we would have told you that you were crazy. The point is that whatever you do to raise funds helps, and eventually your events can really blossom. Thanks to everyone who helped make this event a success!





## Family Fundraising Efforts for the Past Six Months

From January 1 to June 30, families raised \$1,302,484. The Fund also received \$7,107 from the United Way. Our special thanks to all of you who have worked so hard to raise needed research dollars.

### \$1,100,000+

Dave & Lynn Frohnmayer

### \$30,000+

Randy & Christie Kelley

### \$20,000 - \$29,999

Pat & Mary DiMarino

Deane Marchbein & Stuart Cohen

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

Andrew & Vicki Athens

Pat & Maria Gleason

Allen Goldberg & Laurie Strongin

Jeff & Judy Hoffman

Bill & Connie Schenone

Michael & Beth Vangel

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

Eric & Beth Losekamp

Robert & Mary Nori

Jim & Carol Siniawski

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

Mark & Linda Baumiller

John & Audrey Barrow

Randy & Nancy Bloxom

Chris & Susan Collins

Marie DiMercurio

Ed & Janice Duffy

Charles & Katy Hull

Jeff & Beth Janock

John & Dianne Ploetz

Jack & Tannis Redekop

Robert & Andrea Sacks

Bryan & Karen Siebenthal

### Up to \$1,000

Ken & Jeanne Atkinson

Joaquim & Joelle Carvalho

Joseph Chou

Brian & Jennifer Dormans

Paige Ellis

Frederic Engel

The Epps Family

Ezat & Laila Faizyar

Andrew & Jennifer Gough

Dave & Paula Guidara

Mitchell & Tirzah Haik

Roger & Eleanor Herman

Brian Horrigan & Amy Levine

Leardon Keleher

John & Karilyn Kelson

Erik Kjos-Hanssen & Turid Frislad

Marc & Michelle Lauzier

Eugene & Renee Lemmon

Peg LeRoux

Gayle Licari

Dennis & Sharon Lower

Col. Gregory & Lt. Col. Lynette

Lowrimore

Bill & Jackie Lucarell

Tom & Marilyn Massino

Jack & Pam McCarty

Steve & Allison McClay

Kevin & Lorraine McQueen

Griff & Cecilia Morgan

Andrea Morris

Sheila Muhlen

Des Murnane & Mai Byrne

Kenny & Lisa Myhan

Louis & Virginia Napoles

Jack & Lisa Nash

Ron & Freddi Norris

Robin Paulson

Jeff & Arianne Pederson

Lynn & Shirley Quilici

The Russo Family

Rick & Lynn Sablosky

Erik & Lori Salo

Birgit Schmidt

Tommy & Brenda Seiford

Matt & Diane Senatore

Bryan & Connie Simpson

Jeff & Debby Slater

The Stichler/Primmer Family

Mark & Susan Trager

Nanette Vannostran

Marc & Sandi Weiner

The Welfare Family

Gerald & Elizabeth Wisz



*Friends together at the Family Meeting*





*Families relaxing at Karaoke night!*

### **Families Learn at Family Meeting**

*continued from page 1*

Speakers stayed after their presentations and gave families the opportunity to talk with them in informal settings.

A great deal of discussion and controversy erupted this year after John Wagner's talk on the use of androgens. Parents were understandably concerned and anxious after hearing his revised recommendations concerning the timing of unrelated, matched donor transplants, and his concerns that androgens might jeopardize transplant outcomes. Near the end of the meeting, Blanche Alter was kind enough to give an unscheduled talk regarding the history and use of androgens, and Nancy Cincotta graciously yielded some of her time to Rick Harris for further discussions. Clearly, androgens will be much debated in the future as parents wrestle with the very difficult decisions regarding the timing of unrelated donor transplants and the appropriate use of androgen therapy.

As always, we were privileged that Nancy Cincotta, MSW, participated in the meeting. She held a number of "coping" sessions, worked closely with the FA teens, and helped us all absorb

the information that was packed into the meeting. Once again, Andy Eichenfield, MD, donated his services as our very competent and well-loved Camp Doctor.

There were campfires, out of season "trick or treating" for the little ones, many talented karaoke performances, and a magic show that brought big smiles to the faces of the children and adults alike. This magic show was a gift of the Norrises, in memory of their son Alex, who was, among many other things, an accomplished magician himself. ♦

The comments of the families in their evaluations of the Family Meeting attest to its success:

*"Enjoyed being able to network with other parents."*

*"The volunteers with the children were totally awesome! Good job, Rob."*

*"Very nice and beautiful place. I just loved being able to talk to the other families. We feel like we are not the only ones faced with this terrible disease."*

*"As always, the meeting was well organized and very informative."*



*Fabrizio DiMercurio with Marleen and Simon Schmitt*



*Families gather on the docks for the Memorial Balloon Release.*

## We Honor Our Grantors and Corporate Benefactors January 2000 ~ August 2001

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*We also extend our deepest gratitude to the many thousands of caring families, friends, and scientists who contribute to our mission around the world.*



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