FANCONI ANEMIA RESEARCH FUND, INC

FAMILY NEWSLETTER

#20 A Semi-annual Newsletter on Fanconi Anemia for Families, Physicians and Research Scientists Summer, 1996

Families Share News and Enjoy Camp Sunshine Meeting

Fifty-six families from nine countries gathered under fair spring skies during our FA Family Meeting at Camp Sunshine, May 16-21. For the fourth straight year, Larry and Anna Gould and their dedicated staff and volunteers hosted us at Lake Sebago, Maine. Exciting news on FA research, a strong family support program, and full youth recreation created one of our best meetings ever.

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HIGHLIGHTS

Hans Joenje Receives FARF's Award of Merit

The Fanconi Anemia Research Fund presented its Award of Merit to Amsterdam scientist Hans Joenje at Camp Sunshine. Here are excerpts from the letter written by President Joyce Owen to Joenje, notifying him of the award.

Dear Hans,

It is with great pleasure that I inform you that the Board of Directors of the Fanconi Anemia Research Fund, Inc. has voted to bestow on you our Award of Merit....

In the seven years that our Fund has been in existence, we have bestowed the Award of Merit only once before. That was to Manuel Buchwald, for cloning the FA-C gene. Your award is not for a single accomplishment, but for several crucially important contributions, which have made possible major advances in FA research, both in Europe and in North America.

You were, and still are, the driving force behind EUFAR, a consortium of European FA research teams....

The most important outcome so far has been the establishment

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MEDICAL NEWS

Study Tests Alternative Transplant Protocol

FA patients and their physicians from a variety of hospitals nationwide are preparing to test a promising new transplant protocol for patients who do not have a suitably matched sibling donor.

The protocol, initiated in the U.S. by Dr. John Wagner of the University of Minnesota, is being tested as part of a multi-institutional clinical trial (see *FA Family Newsletter 19* and its *Scientific Supplement* for preliminary results and patient eligibility requirements). At least 25 FA patients under the age of 55 will be served prior to publication of the results.

The preliminary findings which led to this trial were based on the outcomes of 218 of a total 400 unrelated donor transplants completed at Minnesota between May 1985 and 1994; seven of these patients were being treated for Fanconi anemia, and the rest for other hematologic disorders such as leukemia, aplastic anemia, and immune difficiency disorder.

Wagner reports that 17 FA patients are now ready to participate in the new study at the University of Minnesota Hospital and Clinic. Two are scheduled for transplant this summer, and 15 await results of a complete donor search. Since this study's inception in September 1995, at least 13 institutions have indicated a serious interest in using the protocol and in submitting follow-up data to Minnesota. Several centers have asked the Wagner team to complete just the bone marrow processing portion for patients who now are scheduled for transplant at their local facility. Dr. Wagner has asked for FDA approval that would allow him and the requesting physician to ship marrow freely across state boundaries. In such cases, the University of Minnesota must recover its costs as part of a patient's insurance agreement with his or her primary transplant facility.

The protocol requires the use of counterflow elutriation to remove donor T cells, which are the cause of graft vs host disease in the patient. Secondly, in order to recover as many donor stem cells as possible, the lymphocyte-containing fractions are passed over a CellPro CEPRATE SC system. The FA patients are treated before transplant with a specified combination of Cytoxan, total body irradiation, and antithymocyte globulin (ATG).

"Due to the relatively large number of patients involved, we should know fairly soon if this protocol is going to work," says Wagner. He urges patients who are not in immediate need of a transplant to await the results of this trial. ◆

New Procedure for Prenatal Diagnosis

A procedure called Preimplantation Genetic Diagnosis may offer new hope to couples who are carriers of a genetic disease. The mother is treated with a hormone to stimulate the production of several eggs. The eggs are removed from the surface of her ovary, and are fertilized with the father's sperm *in vitro*. At the eight-cell stage, a single cell is removed from each "pre-embryo", and tested for the presence of the specific mutation(s) carried by the parents. The same cell can also be tested for HLA type. Disease-free pre-embryos can then be transferred to the mother's uterus. This technology can be used only in families in which the specific mutations have been identified. For now that means FA-C mutations. But very soon, FA-A mutations will be identified as well. For more information, see John Wagner's article in the *Scientific Supplement* to this newsletter. ◆

Volunteer Marrow Donors Give Twice

The following summarizes an article in The Marrow Messenger, *Volume IV, 1995, a publication of the National Donor Program:*

Until recently a patient whose leukemia recurred following an unrelated donor transplant had few options for therapy. The use of chemotherapy to treat recurrent leukemia, especially those relapses occurring soon after transplant, was frequently associated with unacceptable side-effects. Now a new therapy based on the concept known as the "graft versus leukemia" effect is being used with success in this group of patients.

As a group, patients experiencing graft versus host disease (GVHD) have a lower likelihood of recurrent leukemia after transplant. While GVHD is generally considered a complication to be avoided, data also suggest some beneficial effect in eradicating remaining leukemic cells. Cells in the donor bone marrow known as T cells cause GVHD, by attacking the patient's cells. It was suspected that similar, if not identical cells might kill residual leukemic cells.

During the past several years, marrow transplant physicians in Europe and the United States have taken advantage of this "graft versus leukemia" effect to treat post-transplant leukemic relapse. Bone marrow donors have been asked to go through an apheresis process. Some of the donor's T lymphocytes are removed and saved; the rest of the blood is returned to the donor. The donor's lymphocytes are then infused into the patient.

More than 30 National Marrow Donor Program donors have been asked to undergo apheresis so that their lymphocytes could be used to treat relapsed leukemia in their recipients. It is too early to analyze results. But a group of transplant centers in Europe presented results indicating that for patients with relapsed chronic myelogenous leukemia, the new infusion of T cells in 54 of 84 individuals cleared all evidence of malignant cells.

[Editors' note: This therapy has been much less effective in patients with relapsed *acute* myelogenous leukemia (AML), the type of leukemia which often afflicts FA patients.] ◆

Eighth Annual FA Scientific Symposium to Honor Dr. Nasrollah Shahidi

The Eighth Annual Fanconi Anemia Scientific Symposium will be held from October 10 - 12, 1996 in Madison, Wisconsin. At least 18 leading FA laboratory and clinical researchers will present their latest findings in the fields of hematology, oncology, genetics, and molecular biology.

Special tribute will be paid to FA researcher and physician, Dr. Nasrollah T. Shahidi, Professor of Pediatrics at the University of Wisconsin Children's Hospital. Dr. Shahidi will be honored by the University and the FA Research Fund for his 30-year contribution to hematological research, and his long-standing commitment to early and accurate diagnosis and treatment of FA and its complications.

Several new and critically important collaborations are launched each year as a result of discussion among scientists during the FA Scientific Symposium. The schedule provides time for questions or discussion after each speaker and at the end of each session. There is also time for informal discussions between sessions.

Parents and adult patients are welcome to attend, although the presentations are quite technical. Special conference lodging rates are available until September 10 through the FA Research Fund office. Registration materials may be obtained and lodging reservations may be made by phone, FAX, or e-mail to the office. ◆

Dr. Nasrollah T. Shahidi

Hans Joenje Accepts the FARF Award of Merit

I am very happy indeed to be honored with this precious award. The Frohnmayers and the FA Research Fund have gained a lot of respect and prestige throughout the world, and being next in line after Manuel Buchwald to receive this award certainly is a milestone in my career. Even though you have made this into a personal award, you must be well aware that the credit is to be extended to all the people in our department who have actually done the work, and without whom it would have been impossible to achieve any of the things that the award was meant for.

The FA Research Fund started to support our research three and a half years ago, after I had tried to get funding from the Dutch Cancer Society for our planned studies on complementation analysis in Fanconi anemia. The Cancer Society found the plan interesting, but of low priority. This is the usual polite way of saying "sorry, we don't like your project too much, so please go somewhere else to find funding." We did go to the FA Research Fund and they concluded that for them this was high priority research that should be carried out. I think this illustrates quite convincingly that a patient support group can have a very strong impact on research of a rare disease like FA.

Research on Fanconi anemia has been very rewarding for me from at least two perspectives. First: as a scientist, the disease always has been a great challenge, because we still don't understand precisely what goes wrong in a cell that is affected by FA mutations. I've been working on FA for nearly 20 years now and the challenge is getting stronger every day. As of today, I am more than ever motivated to devote the rest of my career to the elucidation of this disease. I have still some 15 years to go before my retirement. By then I am sure that many of the major questions about the disease will be answered, and I am very grateful to be part of it and contribute.

Second: although scientific curiosity and ambition are sufficient as a driving force to keep me going, my job as a scientist has gained an extra dimension by getting to know you: the people who are actually suffering from this terrible illness.

The person who changed my life in this respect was Ralf Dietrich. I met Ralf for the first time in 1991, between Christmas and New Year's Eve, so that was about four and one-half years ago. That was a turning point in my career. Collaborating with him, a father of two affected children and coordinator of the support group in Germany, has intensified my research efforts tremendously. FA was no longer a mutation in a cell culture, but it had dramatic consequences for people in the real world. Not only has Ralf added the human component to my scientific ambition, he has actually made our successes possible by his endless efforts to motivate FA families to take part in our studies. His efforts have also greatly helped to give me the motivation for trying to organize collaboration (rather than competition)

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Hans Joenje [right] enjoys his award with Dave Frohnmayer (Oregon) [left] and Ralf Dietrich (Germany) [center].

FAMILY NEWS

Sophie-Lauren Butler

by Karen O'Driscoll, Chorley, UK

Our daughter Sophie-Lauren is now 19 months old. She was born at 38 weeks gestation by normal delivery. She spent the first three weeks of her life in the special care baby unit because of her low birth weight, 3 lbs, 12 oz. No other problems were encountered at that time. She slept and fed reasonably well.

We had been advised to terminate the pregnancy, but neither I nor Sophie-Lauren's dad (Philip) felt that this option was the right choice to make, as Sophie-Lauren was our baby and she deserved every chance for life.

Sophie-Lauren is very sociable, chatty and cheerful, as you can see from her photograph. She has radial club hands and is short in stature. These two problems which are physical haven't delayed her from getting on with life. She is due to undergo surgery on her hands on June 3rd at Alder Hey Children's Hospital in England. She has to undergo a series of four operations in order to straighten the bones in her arms and to move both index fingers into the thumb position.

Sophie-Lauren attends a child development center on a weekly basis where her skills are observed and developed. She receives therapy there by a number of fully trained staff, which include physiotherapists, occupational therapists, nursery skill trainers and many others who are involved in her development.

The outlook for Sophie-Lauren at the present time continues to be optimistic. She is in good health and is monitored closely by the Hematology Department at Pendlebury Children's Hospital in England. She also is seen by her pediatric consultant, Dr. Campbell,

at 6-month intervals.

As Sophie's parents we hope that all her operations go well and that she remains in good health for the years to come. We love our little girl and we would not change her for anything. She's so special to us and everyone who meets her finds her so adorable and lovable. To her dad and me she's our special little girl; we would do anything for her as we love her so much! ◆

Toby Carr Receives Gene Therapy

by Mike Carr, Ruckland, UK

Toby and I learned a lot from our first visit to the NIH and a lot about the logistics of the travel from this tiny village in the middle of England. We were referred to the trial through the Hammersmith Hospital (the Royal Post-Graduate Medical School) which is one of London's foremost teaching hospitals. It is also not too far from Heathrow, so for this second trip we gave the samples to be sent to NIH, started the G-CSF treatment and then flew off to New York in the evening.

We weren't expecting snow storms and a biting cold wind, or a sudden drop in the Stock Exchange, so New York was quieter than we had imagined and nobody was rude to us! Nevertheless it was a great experience staying in Midtown, seeing Central Park in the snow, the Metropolitan Museum of Art, Manhattan, and the World Trade Center.

We were exhausted when we arrived at the NIH in Bethesda on Sunday, but we went straight for catheterization to be ready for apheresis the next day.

We were very pleased to be able to stay at the Children's Inn again because it is very close to the Clinical Center, and I could easily fit into the physicians' routines and get to know them better. Gene transfer is not easy to understand

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In Memory of Michael Stefanowski

by Jeanne Stefanowski, Arlington Heights, Illinois

By relating the story of Michael's passing I want every FA family to know how critical it is to attend to issues in medical care—wherever one goes—that could have life or death consequences.

My son Michael was born on February 12, 1990 weighing 4 lbs. 11 oz. He grew into a happy, energetic and bright little boy but was small for his age and a picky eater. Michael had chicken pox in February 1995. During his five year physical in April his blood was discovered to be anemic. Through further testing, including the DEB chromosome breakage test, in May 1995 Michael was diagnosed with Fanconi anemia. At that time his platelets were 65,000. He had a normal white count and his hemoglobin was 10.3.

Michael's two sisters were tested. Both were negative for FA and both perfect bone marrow matches. In talking with several hematologists in the Chicago-Milwaukee area, every doctor concurred that with two sibling matches, the transplant was the only option and the sooner the better before Michael became transfusion dependent. During the summer Michael's platelets dropped to the low 30,000's.

In early August we traveled to Cincinnati to the Children's Hospital Medical Center for pre-transplant testing. I chose that hospital because of its excellent success rate with bone marrow transplants with Fanconi patients.

Even though Michael's platelet count had increased to just under 50,000 I decided to go for the transplant and he was admitted to Children's Hospital Medical Center on August 16, 1995. Based on the conversations with the transplant team, I thought he would breeze through the transplant. Michael was under 10 years of age, extremely healthy, no physical anomalies, no blood transfusions and a sibling match. We would be home in 6-8 weeks.

In the hospital Michael received Cytoxan 20 mg/kg, thoraco-abdominal irradiation 400 cgy, ATG before and after transplant, and cyclosporin. On August 22, 1995 he received bone marrow from his youngest sister, Margaret. All was going smoothly. The new bone marrow was producing more white blood cells every day. In early September Dr. Harris had talked about possibly releasing him from the hospital in 7-10 days. We were ecstatic. Unfortunately one week later Michael's new bone marrow was in trouble. The white blood count was falling and he had terrible fevers. By mid-September Dr. Harris performed a bone marrow biopsy to look for female (donor) cells in Michael's marrow. There were very few female cells around. We were told that Michael had rejected the first transplant. Possibly not enough of his lymphocytes were killed with the low dose Cytoxan. The lymphocytes multiplied and destroyed Margaret's marrow. We and the doctors were very surprised. No FA child transplanted at Children's Hospital Medical Center had ever rejected marrow from a matched sibling donor.

One month after the first transplant, preparation started for a second transplant with Michael's older sister Beth donating her marrow. He received twice the amount of Cytoxan the second time, ATG over a longer duration and no radiation. On September 27, 1995 the second transplant took place. More marrow was also given than the first time to help ensure a better outcome.

Michael had a rough time due to the higher dose of Cytoxan, including more mouth sores, pancreatitis and temporary damage to liver and kidneys. In mid-October he had a seizure related to high blood pressure and then was taken off cyclosporin and put on methylprednisolone (a steroid) to guard against graft vs host disease (GVHD). Bone marrow transplant patients often take cyclosporin for

Living with Fanconi Anemia

by Lynn Spach Welfare, Thomasville, NC

I am a 42 year old woman with Fanconi anemia. When Leslie Roy asked me to write an article about living with FA I thought, "Where to begin? I could write a book." I was diagnosed with FA at age 6 after having a profuse nosebleed in my first-grade class. Just this year I discovered the FA Research Fund, Camp Sunshine, and the tremendous support of many of you. So, I will start with today.

My blood counts have been like a roller coaster and still are. I have taken Anadrol (oxymetholone) intermittently as a maintenance dose and as a treatment for 6 months, or 1-2 years and then tapering off. It is used to help my red blood cell count, but it has also helped my platelets. When I first started using oxymetholone, it took six months to show any effect. Today my platelets are at 6,000. I am taking 200 mg of oxymetholone in the morning and 200 mg in the evening. This will continue until my counts return to "my normal stable level" of 30,000 platelets. Then, I will taper off again for about 3-6 months to give my body a break.

The lab counts for my liver are borderline high, so I do not want to take Anadrol any longer than I have to. Side effects can be numerous with it, i.e. cancer, liver tumors, breast cancer, hirsutism (hair growth on the face, even for girls), etc. Yes, I have to shave every morning like a guy, and sometimes in the evening. I may use electrolysis eventually, but to my knowledge this procedure hasn't been tried on anyone taking Anadrol and no one knows if it will work.

I was born without eardrums, with pin size ear canals narrowing into nothing, and fused ear bones. When I was in the 12th grade with hearing of 40%, Dr. Michael Glasscock (Nashville, Tennessee) miraculously fashioned ear drums from the skin of my left hip (right ear) and my right upper arm (left ear). This microscopic surgery took 5 hours and corrected my hearing to 90-95%. Noise pollution presently has it down to 78%. I continue to be blessed by the wonderful care of such nice people who work so hard in the medical field. Input has come from doctors at places like Baptist Hospital where I was born, Duke University Hospital, formerly Durwood Medical Clinic in Charlotte, North Carolina, and Boston Children's Hospital.

Coping with FA. I could go on forever about how I have dealt with FA. I exercise, swim, play, sing, and dance. I live life, enjoy life, family and friends. Whatever I can get involved in, I do. When the going gets tough I literally make myself get in line with "get yourself together, get your butt in gear, enough of this self-pity, and get on with it." If I don't, I stay stuck. Sure, I've been depressed, but I also have had so many great times. It all depends on which direction I look and choose. One of the biggest obstacles I had to overcome was accepting that I had FA. I found it was much easier for me to deal with things after I quit exhausting my energy denying FA, or for that matter anything that was negative in my life.

I overcome FA by bringing myself to acceptance, then moving on. It's a lot easier, I promise! This way I can be more objective about my care and able to think more clearly. I am challenged by it, and do not want to give in to it. I want to beat the odds.

With the help of my family, friends, and doctors I have been able to remain an informed part of my medical team. Importantly, I have continued to get regular complete blood counts (CBCs). Equally important, I have learned to read the results of these as well as many other tests, such as that used to measure my electrolytes (SMAC). This medical teamwork helped a great deal when a lump

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Toby Carr

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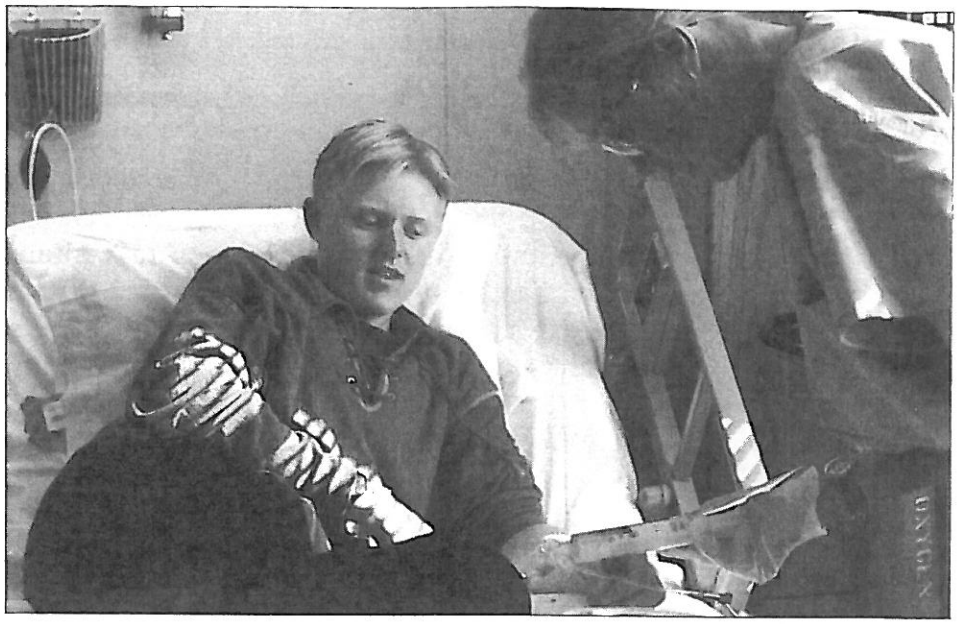
and lots of difficult questions from a parent need a lot of patience from a physician even to a quasi scientist (the newsletters and our London physicians keep me pretty up-to-date). I even had time to give blood and platelets myself.

Toby bled a lot during the apheresis and subsequently through his nose, despite a high platelet count, and this kept him in the hospital all week. No sight-seeing until Saturday!! He formed very good relationships with the nursing staff in the Department of Transfusion Medicine, and with their help the transduced cells were re-introduced (see photograph) and topped up with platelets, and we flew off to New Orleans for St. Patrick's night (rarely celebrated in England).

We packed in two sunny days relaxing with plenty of food and jazz and returned to the U.K. via Chicago mid-week, for more platelets in London and some jet-lag at home.



Toby Carr & Senior Nurse Joy —receiving transduced cells at the end of a week in NIH Clinical Centre.

There are five in our family, including my wife, who had a serious brain injury eight years ago through herpes encephalitis, and now has no memory. She can't get involved in all this and stays in the hospital while I am away. My daughter Katie is 19, about to leave for University, and thankfully fit as a fiddle. She's not even an FA carrier!

My son Marcus is 17, just starting pre-University. He wants to be a Medic! He was diagnosed as having FA at five, following the

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Living with Fanconi Anemia

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was removed from my right breast in April 1996. It was not what we expected. It was tissue that somehow had been damaged, then had pooled in a possible fibrotic mass and hardened. Platelets were given to me prior to surgery and I recovered nicely.

In all, I choose to deal with today's problems and worry about tomorrow when it comes. My family has always been great about letting me live as normal a life as possible. This also included discipline as a child. My parents were strict and it didn't hurt me a bit. I was treated as a person with a special problem, but my two brothers had their special problems, too. To anyone just finding out that you have FA, there could not be a better time. You have at your fingertips support groups, people you can talk with, options, great opportunities to participate in research and findings of your own with physicians and scientists. Medical advancements are taking place so rapidly these days. I will continue to look for a bone marrow match in case I need to have a transplant. It's good to know these things. So far no one in the family matches. Yet I may not need a transplant at all. Radiation can cause secondary malignancies. So if I can go another route I may choose to do so. Who knows what the future holds?

Thank you Lynn and Dave Frohnmayer for creating the FA Research Fund. Thank you Leslie and Linda for your friendship, for being so persistent and for allowing me to share my story with individuals and our support group. For those I have not met and those who have lost a loved one, I hope to meet you next year at Camp Sunshine. I am eager to reunite with the many friends I made for the first time this year. I need you as I am sure so many others need you. We must press on! ◆

CAMP SUNSHINE '96

Camp Sunshine
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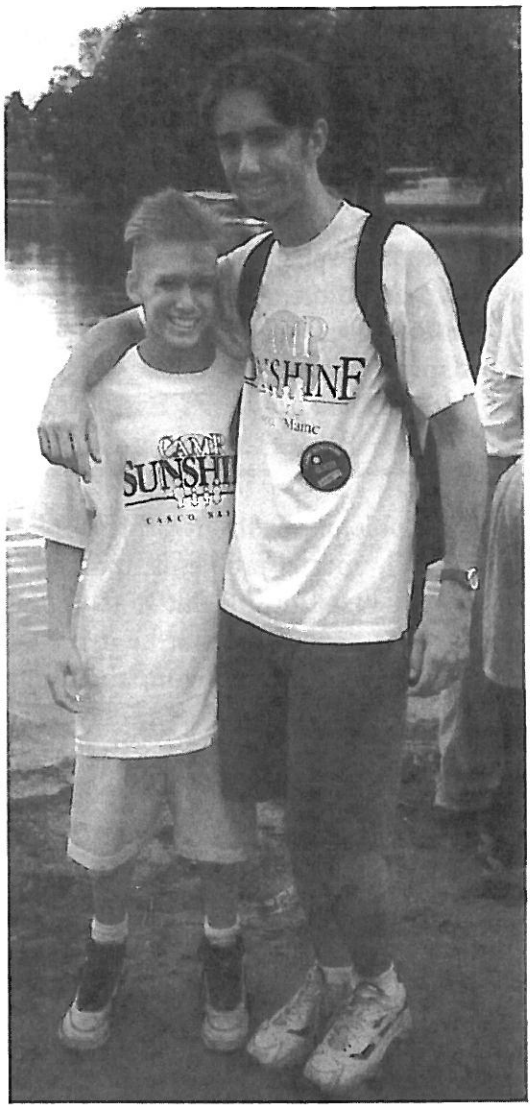
Ten leading researchers and clinicians presented their latest findings in basic FA science and therapy. Many of them arrived early or lingered after their talks, giving parents and patients a chance to ask questions and to express concerns. "All speakers were up front and honest about the way it is," offered one father.

New FA families from Argentina, Cuba, Mexico, and South Africa, frustrated by lack of FA medical information and treatment at home, talked at length with physicians and transplant specialists. A record number of families and patients contributed blood cells for research in the laboratories of Alan D'Andrea, Markus Grompe, and Hans Joenje.

The latest results in research labs brought new hope to many families at Camp Sunshine (see *Scientific Supplement*). A rousing tribute to Dr. Hans Joenje of Amsterdam, recipient of the Award of Merit, brought all participants to their feet (see related articles on pages 1 and 4.)

Parent support sessions were led expertly throughout the action-packed program by Nancy Cincotta of Mt. Sinai Medical Center in New York. "The unity of the families is special," reflected one parent. Others reported "The fellowship is great—it helps everyone in the family to be able to talk and share about living with FA." "I liked the chance to talk about adjustment issues." "Let's have more time with Nancy, she's great!" Many ideas about easy ways to raise funds for research, with and without using your family name, were shared during closing discussion. Chris and Susan Collins, publishers of the *FA Express*, were honored for their generous gifts of time and financial support.

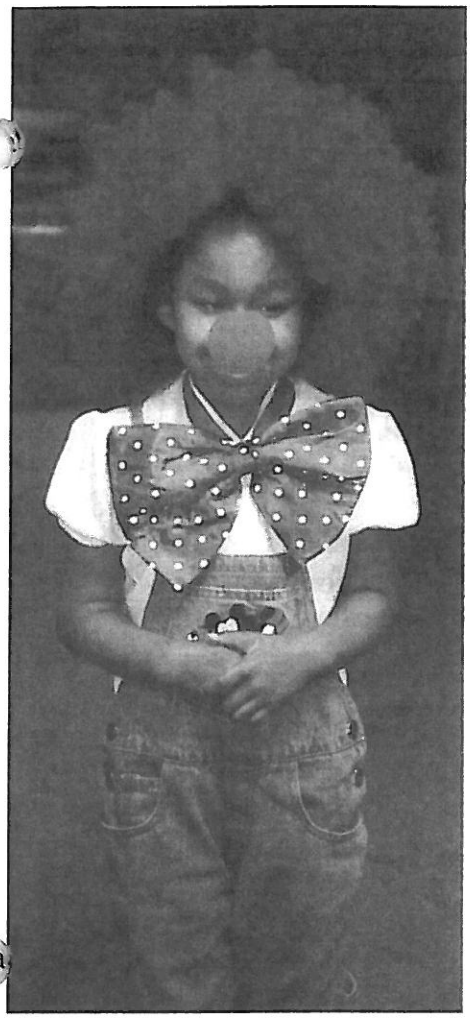


Derek DaRosa and Camp Counselor Jarrett Krosoczka

Camp Sunshine's executive director Wayne Wagner, program administrator Tim Rabine, volunteer coordinator Joe Pappalardo, and our Fund's staff built a program that gave everyone time for productive conversation and family recreation. Several evaluations declared, "The youth program is great. Kids are really bonding to each other." "We could really relax, and make new friends." and "I learned a lot—some I wish I didn't know—but it was helpful."

Excellent meals, an opening magic show (sponsored by Ron and Fredi Norris in honor of their son, Alex), clowns and comedians, the infamous FA Family Talent Show, arts and crafts, bass fishing lessons, a youth sleep-out, an adults-only gourmet dinner and dance, a masquerade party, and many campfires, filled the meeting with joy.

Thanks again, Larry and Anna, and everyone present, for creating Camp Sunshine. It remains a beautiful place where, in a few short days, we gain new strength through knowledge and lasting friendships. ◆



Angel Atwood clowns around

L-R: Mary Beth Salguero, Dany Salguero, Jorge Cabrera

We all had a wonderful time at camp. The experience of being with all the other families was incredible. It really helps us to face this next year.

We were also impressed with the doctors and all of you from the FA Fund. We genuinely felt the love and concern from everyone at camp.

Thank you again for the memorable camp.

~ Sharon Atwood, Waddell AZ

Masquerade party revelers. Can you guess who these revelers are?

We Welcome New Families Who Have Joined Our Support Group

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Sophie-Lauren ~ DOB 10/25/94

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Emily ~ DOB 11/25/91

Adrianna & Santiago Gomez
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Gayle Licari
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Kevin & Robin Moroney
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Metuchen, NJ 08840

Robin & David Paulson
6885 Hemlock St.
Redding, CA 96001
(916) 244-6616

We no longer have a correct address for **Marion Folkes** of Toronto. If anyone has a current address for Marion please contact the FA Research Fund office. ◆

In Loving Memory

Leah Delcamp
9/27/95

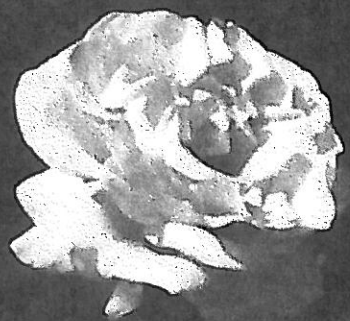
Julie Fena-Lavulo
7/10/96

Christine Gonzalez
7/27/95

Scott Graybill
12/17/95

Jenny Hamilton
3/24/96

Hikmet Topcuoglu
4/19/96



Hans Joenje Receives Award

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of a Cell Repository in Amsterdam and the achievement of doing fine diagnostics of European FA families by complementation analysis on a relatively large scale. You took complementation analysis, ... an important step forward, by adding more patients to each of the 4 groups that had been previously identified by Manuel among 7 unrelated patients, and adding at least one more complementation group (E). The E group is now likely to comprise even more groups, indicating an unexpected high degree of genetic heterogeneity in FA.

You have determined that the FA-A complementation group is by far the most common among European FA patients. Your classification of patients into complementation groups has made possible fine-tuned linkage analysis, pinpointing the FA-A gene to a small region of chromosome 16q24.3.

You have freely and generously sent your cell lines, assigned to specific complementation groups, to investigators all over the world. At our 1995 Scientific Symposium, many of the scientists thanked you in their presentations. Your work has provided the foundation for many other research projects.

Your discovery of a "strong candidate c-DNA" for FA-A will soon lead to diagnostic tests based on identification of specific mutations. Your discovery also offers the possibility of gene therapy for the many patients in the FA-A group.

On behalf of the Board of Directors and our many FA families, I wish to thank you for your important contributions to FA research! ◆

Toby Carr

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investigation of a small skeletal abnormality of his right wrist, complicated by low platelets. With the help of oxymetholone he keeps on the right side of the line with platelets about 26, Hb at 12 and WBC at 6. Also Katie is a perfect match for a BMT should he need it which means chances of success are really high.

Toby however, who is 14, is well below the line. He doesn't have a donor and for the last two years has survived with twice weekly HLA-matched platelet transfusions, coupled with blood transfusions after serious nosebleeds. He was diagnosed at six and proceeded like Marcus until two years ago. His platelets pre-transfusion are usually less than 10, his Hb drops from 9 to 6 regularly and his WBC, steadily decreasing, is around 2.5. Thankfully the cell investigations showed him to be FA-C.

The excitement of traveling to the US keeps his spirit up, and I usually try to add something special each time. We managed to be with friends for Thanksgiving; maybe next time we'll hit Independence Day!

So far we know that transduced cells are present in his marrow, but have yet to see any increase in peripheral blood counts. We are still confident and thankful to be in on this trailblazing gene therapy trial. ◆

Hans Joenje Accepts Award

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among European research groups. This culminated in the research consortium EUFAR, which has been officially recognized and sponsored by the Commission of the European Union.

Through the FA Research Fund we have received substantial amounts of money for our studies. We are well aware of the fact that it takes a lot of your energy to raise this money and that all your fund-raising efforts are made with only one objective: improving the outlook for your children. You can be sure that we fully realize this and that we are spending every one of your dollars with this objective in mind.

It has been a most stimulating experience to work with the people who are running the Fanconi Anemia Research Fund on a day-to-day basis. I am now specifically thinking of Linda DeSpain, Leslie Roy and Joyce Owen, with whom I have been in touch by e-mail almost every other day, and also the members of the Board of Directors and the Scientific Advisory Board. I continue to be impressed by the vigor, enthusiasm and professionalism that characterizes the way they do their job. I am very grateful to be in a position to work with all of you. Let's continue with this spirit of collaboration, which in my mind is the best guarantee that we will meet our objectives as soon as is humanly possible.

Thank you very much. ◆

FUNDRAISING

Collins Fundraiser a Huge Success

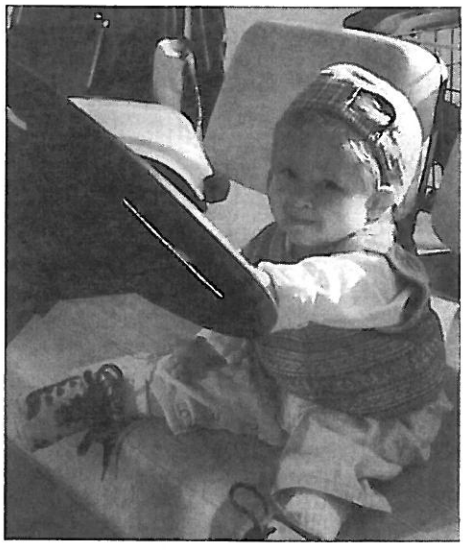
by Leslie Roy

Springtime in Florida means beautiful days, sunny skies, lush greens and golfing. Golf tournaments are a dime a dozen. Chris and Susan Collins wanted to hold a tournament "to raise funds for research which is giving our family and so many others hope. We need to keep pushing for financial support. A lot of kids are counting on us." What could Chris and Susan do to make their tournament different or more attractive?

With much speculation and planning and some apprehension, the First Annual Christian Thomas Collins Golf Tournament to benefit the Fanconi Anemia Research Fund was held April 18th. The entry fee of $250 included an instructional clinic with top golf professional and former Masters champion Bernhard Langer. Afterwards, there was food and drink, a silent auction, and a live auction. There were also great gifts and prizes for the participants. Chris and Susan invited local celebrities to make it more fun for the other players, and were fortunate to have a well-known local TV anchor as host of the live auction.

Chris's many contacts and the help of a few close friends gave them the opportunity to find the participants needed to make the tournament a success. Two newspaper articles about Christian's life with FA and the need for funds helped to increase public awareness prior to the event. Susan handled the large mailing, and contacted people to donate auction items and/or sponsor a hole for the tournament.

There were 84 participants in the tournament. Susan said, "It was a great day, people were very kind and Christian had a good time running around at the golf course and being the center of attention. It was nice to have Christian there so that our supporters could see exactly why we are fighting so hard against FA. I think all of our players left the course that day feeling very good about the contribution they had made." The tournament, auction and letter campaign brought in $34,000. Susan and Chris are already planning for the Second Annual tournament and would be more than happy to help any other families who are interested in putting a fund-raising event together. ◆



Christian Collins heads out onto the green.

Tiger Foundation Grants New $75,000 Challenge

The Tiger Foundation of New York has presented FA families, friends, and relatives with a $75,000 challenge. Starting May 1, 1996, the Foundation will match every dollar raised in the name of the "Tiger Challenge," up to a total of $75,000.

We send our deepest gratitude to Gerald Norris and Trustees of the Tiger Foundation for their generous philanthropy and commitment to our mission. When this challenge is reached, the Foundation will have awarded over $500,000 to the FA Research Fund over the last five years. An additional $300,000 has been raised in matching funds by families. The rapid progress of FA research has been made possible by this support.

Contributing to research is rewarding when you know your gift will double. Simply write "Tiger" on the memo line of your check. For fundraising ideas by other families, consult the latest editions of the *FA Family Bulletin* (May, 1996) and the *FA Family Newsletter 19* (Winter 1996) or talk with Leslie Roy or Linda DeSpain at the Fund office. Brochures, labels, custom envelopes and stationery, postage reimbursement, and donor contribution lists are available to help make your campaign run smoothly. All named donors receive a letter of thanks as their receipt. ◆

Family Fundraising Efforts

From January 31, 1996 through June 14, 1996, forty-two families helped to raise a total of $90,178. Our Fund received an additional $3,625 from the Combined Federal Campaign and/or United Way Campaigns that we were not able to attribute to particular families.

Five research proposals were funded at our May Board of Directors' meeting, and three more are out for review. If these are approved, the eight proposals will cost our Fund $465,545. Our ability to continue to support quality scientific research depends largely on families' fundraising efforts. The new $75,000 Tiger Challenge Grant comes at an excellent time to help motivate giving during the next six months.

Seven of the forty-two families listed below raised funds by either letter-writing campaigns or holding an event: a golf tournament, garage sale, or benefit dance. Many of the forty-two families made special efforts to inform family and friends of the opportunity to give to our Fund by way of payroll deductions through the Combined Federal Campaign and/or local United Way chapters. We are deeply grateful to each and every family for the time they have invested.

Our support group now has over 450 members. Not all families are able to do fundraising, because of health problems, but for those of you who are, please consider what you can do to help. Every dollar raised helps support research seeking life-prolonging treatments, undiscovered genes, and ultimately, a cure for FA!

The amounts reported below were received at our office by June 14, 1996. Funds received after that date will be reported in the Winter newsletter.

Over $40,000

Laurie Strongin & Allen Goldberg

$10,000 - 40,000

Chris & Susan Collins

$5,000 - 10,000

Lynn, Dave & Kirsten Frohnmayer
Jackie & Bill Lucarell

$2,000 - 5,000

Vicki & Andrew Athens
Lori & Erik Salo
Chris Scaff

$1,000 - 2,000

Fredi & Ron Norris
Connie & Bill Schenone
Susan & Mark Trager
Therese & Terry Robertson

$500 - 1,000

Alison & Steve McClay
Myra & Mike Lewis
Deane Marchbein & Stuart Cohen
Beth & Mike Vangel
Virginia & Louis Napoles
Debby & Jeff Slater
Sandi & Marc Weiner

Up to $500

Janeth & Al Acosta
Sarah Baker
Linda & Mark Baumiller
Pam Baxter
Susan Combs
Carol & James Dillon
Iris & Neil Frank
Diane & Greg Hayes
Karilyn & John Kelson
Leardon Keleher
Jeanne & Tim Kucera
Peg & Rene LeRoux
Beth & Eric Losekamp
Lynnette & Gregory Lowrimore
Pam & Jack McCarty
Karen & Gene McDaniel
Lisa & Jack Nash
Debbie & David Risher
Andrea & Bob Sacks
Robin Paulson & Marlene Stone
Lynn & Rick Sablosky
Myra & Carlos Salguero
Karen & Bryan Siebenthal
Jeanne Stefanowski

Contributions were received in memory of Caleb Marchbein-Cohen, John McLaughlin, Alex Norris and Michael Stefanowski. ✦

Dessert Reception Fundraiser

by Laurie Strongin

Upon learning of our son Henry's diagnosis of FA, many of our friends and family have asked us how they can help. Already having contributed more than their share of love and support, my sister Abby Cherner and her husband Andy came up with an idea well worth sharing.

On Wednesday, June 26, Abby and Andy invited their close friends and family to their home for a dessert reception to benefit the Fanconi Anemia Research Fund. The evening was a great success, not only in the money they raised, but in the growing number of people who are now familiar with Fanconi anemia and committed to finding a cure.

Abby was able to keep the costs to a minimum: she baked all the desserts herself (a delicious labor of love); arranged for the printing

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Website Update

In our Winter issue, *FA Family Newsletter 19*, we reported that a member of our Family Support Group, Jeff Janock, had set up a Website. Jeff took on the tremendous job of converting our Handbook to Web language (HTML), and has posted the entire book, including the Appendices (*Fanconi Anemia: A Handbook for Families and Their Physicians*). Point your browser to **http://bc.cybernex.net/~jj/fa.**

Another family member, Allen Goldberg, has set up a Website for his son Henry. He has posted Linda DeSpain's editorial about cord blood banking, which appeared in the *Wall Street Journal*. Check it out at **http://www.nab.org/henry/**.

Later this summer we will have our own official Website! Our editor has already converted *FA Family Newsletter 19* and its *Scientific Supplement* to (HTML). We plan to post all subsequent newsletters there, as well as information about FA, registration forms, grant application guidelines, and links to related sites. We will notify all of you for whom we have e-mail addresses as soon as we are up and running. I'm sure Jeff and Allen will have links to our Website as well. ◆

Eugene Gets New Area Code

Please note that the area code for the FA Research Fund is now 541. Please change this on your phone and FAX lists. The old area code will no longer work. The 800 number remains the same.

Dessert Reception

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of the invitation to be donated; got a price break on the photos of Henry that accompanied the invitations; and obtained a discount on the cost of some rental items from a local caterer.

In addition to raising money for the Fund, Abby and Andy wanted their friends to learn about Fanconi anemia. Working through the Fund, they invited Dr. Alan D'Andrea, a hematology/oncology researcher from Dana Farber in Boston to speak. Dr. D'Andrea was very honest about the severity of this disease, which caused a great deal of sadness, yet at the same time instilled hope by explaining that much progress has already been made and the money donated would help him and others continue their search for improved treatment and an eventual cure.

The event also provided an opportunity to inform the guests about the proposed FDA regulations that would limit cord blood supplies and transplants. We handed out information and talking points to include in a letter to Dr. Kessler, Senator Kassebaum and Jane Williams, Health Policy Advisor, Committee on Labor and Human Relations. We have been told that more than a dozen letters have already been sent (and those are just the ones we have been told about.)

All in all, 97 families contributed a total of $13,090.

If anyone has any questions about the event, please feel free to call Abby Cherner at (202) 965-7060. ◆

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