FANCONI ANEMIA RESEARCH FUND, INC.

FAMILY NEWSLETTER

#22 A Semi-annual Newsletter on Fanconi Anemia for Families, Physicians and Research Scientists Summer, 1997

Masquerade Party at Camp Sunshine

Seventh Annual Family Meeting Sets Attendance Record

Weather ranged from spring thundershowers to brilliant sunshine. But nothing dampened participation by a record 66 families from eight nations who attended Camp Sunshine. Our seventh annual family meeting was held at Point Sebago, Maine from May 16-20, 1997.

Families enjoyed a full schedule of recreation, family support meetings, presentations by leading scientists and evenings packed with entertainment. A continuing tradition of spontaneous late-night bonfires provided welcome opportunities for companionship.

Among many highlights:

- A special tribute to two long-time volunteers to FA family meetings, the husband/wife team of Andy Eichenfeld, MD and Nancy Cincotta, MSW. Nancy's social work expertise in leading our counselling groups and Andy's expert on-site medical care have made our meetings far richer and more productive.
- A wonderful, enthralling magic show sponsored by Ron and Fredi Norris and dedicated to the memory of their gifted son, Alex.
- Well-organized and informative scientific lectures addressing progress in understanding FA genes, improvement in survival rates for high risk transplants, the challenges of gene therapy, and hope for future AML therapy.
- Dedication of an FA family memorial site for families who have lost precious children, siblings and spouses to this disease.
- Dedication ceremonies for the expansion of Camp Sunshine's permanent facilities.

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HIGHLIGHTS

MEDICAL NEWS

New FDA Cord Blood Regulations Announced

Over the past year, patient families and patient advocacy groups joined Congressional leaders, industry representatives, physicians, and medical professional organizations in a massive effort to change the Food and Drug Administration's intent to regulate umbilical cord blood as a new drug. Many FA families and FARF staff and Board members sent letters to David Kessler, formerly Commissioner of the Food and Drug Administration, and to their local congressional delegates agreeing that safety should be insured, but urging that new regulations do nothing to limit cord blood supplies and patient access to them. We pleaded that new regulations should not restrict a parent's ability to bank an infant's cord blood for possible future therapeutic use.

Our lobbying campaign appears to be succeeding. On his last day as FDA Commissioner, (February 28), Kessler issued guidelines which aim to ensure the safety of nondrug therapies, yet speed the treatments to market. The new model, endorsed by Vice President Gore, proposes a tiered approach that ranges from little or no regulation on cell and tissue products with relatively little health risk, to increased oversight for therapies with greater risks.

Members of the American Association of Blood Banks, such as Viacord in Boston, had feared that their services would be regulated

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Scientific Developments Highlighted

Among the numerous reports of scientific progress at Camp Sunshine were the following:

Understanding FA Proteins

Alan D'Andrea, MD, of the Dana-Farber Cancer Institute, Boston, reported that the normal FA-A and FA-C proteins bind to each other and form a protein complex in the nucleus of the cell. This new discovery suggests possible functions of the FA proteins.

Grover Bagby, MD, Oregon Health Sciences University presented a detailed hypothesis about the function of the normal FA-C protein. Cells lacking this protein are extraordinarily sensitive to a variety of substances produced by the body (such as interferon-gamma and tumor necrosis factor alpha) or in the environment. Bagby's research concludes that if the FA-C protein doesn't work (which is the case in all patients with FA of the C type) progenitor cells and stem cells exposed to certain substances will undergo premature programmed cell death. See article about apoptosis in the last FA Family Newsletter.

Unrelated Donor Stem Cell Transplantation

John Wagner, MD, reported results of 23 FA patients transplanted at the University of Minnesota using marrow or umbilical cord blood from related (non-sibling) or unrelated donors. Three additional patients were treated on this protocol at other centers, with donor bone marrow processed (by T-cell elutriation) at the University of Minnesota. Two of these patients are alive and well 1½-2 years after transplantation; one patient died as a result of graft failure.

Overall survival rate for the Minnesota patients is 48%. This figure is remarkably better than FA unrelated or non-sibling transplant results just a few years ago. Significantly, there has been no experience of life-threatening acute graft vs. host disease with T-cell elutriated transplants.

The greatest cause of mortality in this patient population has been failure to engraft with donor cells (36%). The Minnesota protocol has recently been changed; patients at greatest risk now receive 600 cGy of total body irradiation instead of 450 cGy. The protocol will be altered depending upon outcomes.

Leukemia Prevention and Treatment of Early Stage Leukemia

Robert Arceci, MD of the Cincinnati Children's Hospital reported early results of research in leukemia prevention and treatment of acute myelogenous leukemia while the number of malignant cells is quite small. Since FA patients are at high risk of leukemic transformation, these studies are vital. Arceci's method focuses on stimulation of the body's own immune system to develop an effective anti-leukemic response. Much work remains to be done before this approach could be ready for human clinical trials.

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New Methods for Complementation Analysis, Carrier Detection, and Gene Therapy

We wish to notify FA patients, families and their doctors of the joint effort at Children's Hospital of Boston, Dana-Farber Cancer Institute, and the Oregon Health Sciences University to perform improved (1) FA subtyping analysis (2) FA carrier analysis and (3) FA gene therapy.

With the use of the FA cell repository, begun at OHSU in 1992, we have begun systematically to analyze blood cell lines and skin (fibroblast) cell lines established from FA families. Identification of FA patients as type A or type C is a prerequisite for subsequent gene therapy trials.

Previously, the complementation group of an FA patient had to be determined based on lengthy cell-cell fusion studies or mutation detection. Using cultured skin fibroblasts it is now possible to subtype a family into groups A or C based on functional correction with a retrovirus and/or analysis of FA proteins. These tests are independent from the particular kind of mutation in a family and are therefore more rapid than older techniques.

Protocol Sequence

1) Establish skin (fibroblast) cell lines from all FA patients and blood cell lines from siblings and parents.

2) Subtyping Analysis. Analyze cell lines for:
   a) Presence (or absence) of FA proteins.
   b) Correction by FA-C and FA-A retroviruses.
   c) Specific mutations in FA-C or FA-A genes.

3) Carrier Detection. Once a complementation group has been determined, carrier testing can be performed with genetic linkage studies. If informative FA proteins or gene mutations have been identified, carrier detection is possible for all family members.

4) Confirmed FA-A and FA-C patients become candidates for gene therapy protocols. Gene therapy protocols will be based on the use of retroviral vectors carrying the normal FA-A or FA-C gene. Retroviruses will be used to correct hematopoietic stem cells, followed by autologous transplant with corrected cells.

For questions regarding the status of FA cell lines, FA patient subtypes, or carrier detection, please have your physician contact:

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3181 SW Sam Jackson Pk
Portland, OR 97201
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or

Alan D. D'Andrea, MD
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44 Binney Street, L103
Boston, MA 02115
phone: 617-632-2080
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Scientific Developments

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Gene Therapy

Christopher Walsh, MD, formerly of the NIH and now at the University of North Carolina, reported on the results of a first effort to treat three FA-C patients with gene therapy. Each patient underwent plasmapheresis to capture the early progenitor cells from the blood. These cells were then mixed with a retrovirus modified to carry the FA-C gene, and then were returned to the patient. Patients underwent this procedure four times, once every three months. This study demonstrated that this method was safe and, according to Walsh, "the clinical results were very encouraging." In one patient all three blood counts increased; two patients showed an increase in the number of cells in the bone marrow. Researchers have now developed a retrovirus which carries the FA-A gene into cells. Walsh asked FA patients to supply blood samples for use in testing vectors in preparation for future gene therapy trials.

A number of centers, including Boston, NIH, Oregon Health Sciences University, University of North Carolina, Indiana University and others are working to find improved vectors to target the very earliest stem cells, to infect a much larger number of stem cells with normal FA genes, or both.

For more information about these studies and others, please see the *FA Science Letter.* ◆

Physician and Patient Alert: Regular Blood and Marrow Testing is Vital for FA Patients

by John Wagner, MD

All patients with FA should have a marrow examination with chromosomal analysis once a year, and peripheral blood counts more frequently (e.g., every 3-4 months) as long as the ANC>1500, Hgb>10, and PLT>100,000. Once any of these values decreases below the suggested levels or once a cytogenetic (chromosomal) abnormality is detected, the test should be performed more often. If there is a cytogenetic abnormality, then marrow should be tested every 4 months. The one possible exception to the above relates to FA-C patients with exon 14 or IVS-4 mutations, in whom more rapid hematologic deterioration is predicted according to data presented by Gillio and Auerbach. These patients should have more frequent examinations even when the counts are above the described values.

All females should consider having blood sent for stem cell "clonality studies". The FA Research Fund recommends that all such patients contact Fund staff periodically for an update on the need for blood donations for clonality studies. For patients specifically followed by me at the University of Minnesota, blood will also be evaluated by Dr. Stella Davies at the University of Minnesota. Results will be used to decide if more frequent marrow testing should be performed. We believe that the development of stem cell clonality will predict the development of a clonal cytogenetic abnormality and leukemia. We must emphasize that this has yet to be proven and is part of the reason for the study. However, these data may serve to provide additional motivation to the referring hematologists, who may otherwise be reluctant to perform yearly marrow examinations.

All FA patients whom I follow at the University of Minnesota will have blood sent to Dr. Arleen Auerbach for repeat DEB testing, and complementation group analysis, if not already performed by her laboratory. Mosaicism will be determined. Marrow specimens obtained here or sent in from outside the University of Minnesota, will be divided between various groups performing research in FA. A fraction of all marrow obtained will be stored in a cell repository at the University of Minnesota; marrow obtained from patients with FA-C will be sent to Drs. D'Andrea (Boston), Liu (Bethesda), Walsh (Chapel Hill), and Broxmeyer & Clapp (Indianapolis); marrow obtained from patients with FA-A will be sent to Drs. D'Andrea (Boston), Walsh (Chapel Hill) and Williams (Indianapolis); marrow obtained from patients with FA-A or FA-C with myelodysplastic syndrome or leukemia will be sent to Dr. Bagby (Portland).

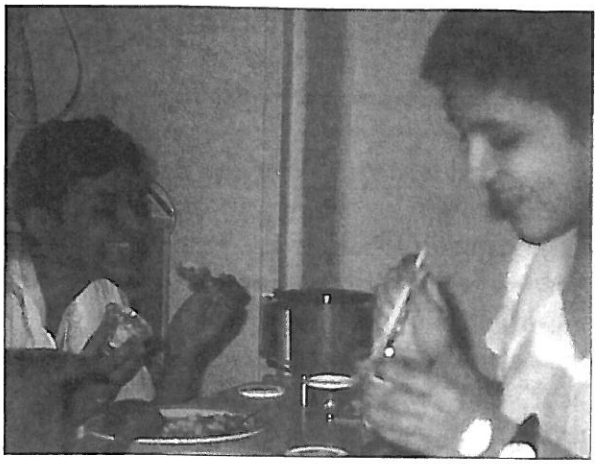
We assure patients and their families that these studies will be done only with their permission. Consent forms are signed only after the research plan is discussed in advance. Even in situations where research specimens are not obtained, every marrow examination should include a detailed evaluation for myelodysplasia and leukemia as well as an assessment of cytogenetic aberrations. When possible, additional samples should be obtained at the same time to help researchers better understand the defect in FA, predict the course of the disease for an individual patient and develop safer methods of treatment. Therefore, when the referring physician asks whether these tests are necessary, there should be a resounding "yes". These tests will teach us more about the disease and how to treat the hematological problems associated with it more effectively and more safely; the tests will also help us to avoid problems in some cases, such as progression to leukemia.

To review: Why the clonality test? To heighten physician monitoring for myelodysplasia and leukemia. Why test for mosaicism? To determine if mosaicism predicts a milder disease course or greater risk of graft rejection after marrow transplantation. Why evaluate the blood and marrow so frequently? To optimize the medical management of patients with FA as well as help laboratory researchers more quickly to find the cure for this disease. Finally, how will the results be communicated back to you? This needs to be arranged when you decide to participate. For patients followed at the University of Minnesota, results of DEB testing, complementation group analysis, mosaicism and clonality will come from Dr. Davies or myself. Results of marrow studies looking to develop gene therapy protocols or understanding the defects responsible for marrow failure or development of myelodysplasia or leukemia will be presented only in composite form by the researchers themselves at future meetings. These latter studies may benefit patients in the future. ◆

FAMILY NEWS

Chronicle of a Bone Marrow Transplant

Imroze Ardeshir wrote a thorough chronicle of the bone marrow transplant she received at Hammersmith Hospital, London. The reader can obtain the full report through the FA Research Fund. The following summary has been edited extensively due to space constraints.



Imroze Ardeshir and her sister Farah (right)

Imroze Ardeshir received her first bone marrow transplant from her sister, Farah, on October 24, 1996. She described at length the protocol used to prepare her for transplant. Following the transplant she had a sore mouth and was on liquids for about 4 days. The doctors were generally pleased with her progress and her counts began to rise satisfactorily.

On November 12, Imroze's blood counts suddenly dropped. She had a high fever and "the situation was tense". She was administered G-CSF and immunoglobulin. These drugs were not effective and her counts did not improve. Steroids were then administered but also proved ineffective. On November 25 the doctors declared that the transplant had failed. They would use another protocol and do another transplant. "This demoralized all of us but the alternative was unthinkable and the second transplant was necessary."

"I had borne all the pain, discomfort, very bravely, co-operated in every way, maintained a positive attitude, kept my spirits up throughout, but this news shattered me completely and I broke down into heavy sobs. I was longing to be out of the hospital soon and this meant at least one and a half months more. Farah will have to donate more marrow but she took it very bravely."

Imroze was again given chemotherapy and for three days she also received "radiotherapy and Anti-Lymphocyte Globulin. Each is painful and has a lot of side effects in itself. But when both were administered the same day one after another, the results were devastating as there was over-reaction of radiotherapy. I was losing a lot of blood and water. I was in agony so was given various drugs in succession."

Farah again donated bone marrow, but this time it was more difficult to obtain as she had previously given only a month and a half before. She had to stay in the hospital an extra day and was very weak.

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Emily Salo Receives Bone Marrow Transplant

We recently received an update from Lori Salo, mother of Emily, age two. We quote in part:

We went to Camp Sunshine again in May. We had a good time despite the lack of sunshine. There were old friends to catch up with and new friends to welcome to our great big "family". Lots of babies to hold and hugs to be had. While we were there, we talked to Emily's bone marrow transplant doctor and he presented us with a sad reality. After assessing her recent blood counts he told us that we needed to go to transplant soon if we did not want her odds of survival to decrease. So here we are, about to leave for the biggest "journey" of our lives.

We are going to Minnesota on June 12. On June 17 we will be checking Emily into the University of Minnesota for her bone marrow transplant. We expect to be there a minimum of two months. Emily is to undergo chemotherapy and radiation treatment before the transplant. I was told she is going to lose all of her gorgeous curls and feel VERY bad for at least two weeks. They liken it to having the flu times 4!

We are frightened, nervous, and terrified. And yet at the same time we are very hopeful that this will

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My Adam, My Teacher

By Darla Patrick

The impact of Adam not being in our lives anymore does not seem to be diminishing with time. It actually seems to be increasing, gaining strength and power, like a snowball that started out baseball-size and by the time it gets to the bottom of the hill, it is boulder-size. The depths of this loss and the accompanying feeling of abandonment, coupled with the bewildering "How did this happen?" question that surges through us all, is indescribable. Even as we stand at his headstone, talking to him, sending balloons up to him in heaven, it's still difficult to comprehend that we won't go home and find him there, like he had played this big ol' practical joke on all of us. If there is any comfort level at all in this horrible ordeal, it is that I came away from this wiser and more fulfilled from this child having touched my life and, more importantly, my heart.

From the moment I met Adam Day, I saw him as an old soul in a young man's body. He had just turned eight years old. Eight going on forty, that is. In so many ways, he was such a child—curious, determined, headstrong, a little overindulged—but with such an uncertain future ahead of him, that was not such a devastating breach of parental control. In other ways, he was such an adult. His perception of everyday life well surpassed my own, as well as many other adults with whom he came in contact. When in conversation with Adam, it was easy to forget that you were talking with a child. His intensity, his humor, his compassion, his strength, his courage—even his anger—were all

Adam Day launching his wish boat

so finely-tuned and so much more developed than that of us so-called grown-ups. You didn't get away with much around Adam. If he didn't like you, there were never any pretensions about it. However, if he loved you, you knew something special had touched your soul.

I defy anyone of sound mind and body not to have fallen at least a little bit in love with Adam Day. He was an affectionate, giving, caring and wise young man. He was thirty-two years younger than I, but I learned so much from him. Even though I had lived many more years than Adam, he taught me about life. Through my many visits to the hospital with him for platelet transfusions, he taught me about patience (and patients). Through living with him every day, he taught me about unconditional love—the kind between a child and a parent/parental figure. I learned about respect from him—sometimes how a child sees things brings you to a harsh reality about your own prejudices. I learned about not taking myself so seriously but taking him very seriously, despite his age (and became a staunch advocate of "Kids Are People, Too"). I learned about bravery and inner-strength from him and, unfortunately, I learned about death from him. Which has taught me so much about living. And I wish I could wrap my arms around him and tell him how grateful I am.

I was not a biological member of his family but he certainly treated me as though I were. He showed me that family is sometimes a state of mind and not necessarily a blood relation. I shared twenty months of his life with him. Twenty years still would not have been enough. If I ever had a hero, it was Adam Day. I want to be just like him when I grow up. ◆

Emily Salo

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help Emily to live many years. Emily has very good odds and we have been given this gift of a chance because of Miranda, her eight-month-old sister. With all of your prayers and good wishes I just don't see how we can lose! I want so much for Emily to feel good and be able to do things with other children without the fear of a virus hurting her.

If you would like to contact us while we are in Minneapolis, you can write to us at:

Emily Salo
c/o Ronald McDonald House
608 Ontario St., SE
Minneapolis, MN 55414

Or e-mail us at: esalo@sprynet.com

For an update on Emily, visit our web site at: http://home.sprynet.com/sprynet/esalo

Your editors visited the Salos' web site in mid-July and learned that the transplant went well and the family was preparing for Emily's discharge from the hospital. How we hope that all continues to go well for Emily! ◆

Starting School and Taking FA With You

By Carol Siniawski

We know all parents are a little nervous the first day of school. But we were really nervous, almost to the point of considering home schooling Jake. We worry about him even when he is in our care. How would we get through the day knowing he is in others' care, with kids older, bigger, stronger and more agile than he is? What if he got seriously hurt at recess? You see, Jake's platelets are around 30,000, his hemoglobin ranges from 7-10 and his absolute neutrophil count goes from 500-2000. The thought of what could happen at school really bothered us.

We finally decided that home schooling would be a disservice to Jake. Not because we couldn't do it nor that he wouldn't learn what he needed. Jake would miss out on all the friendships, camaraderie, bus rides, and yes, even recess. We decided to send him to school just like the other kids, but we would do a few extra things to educate the school environment and help the school staff make the right decisions in the event of an injury.

Once we started looking into this, we received many good recommendations and had a few thoughts of our own. We want to share them with you in an effort to ease your own worries.

1. We had medical alert tags engraved with Jake's name, date of birth, pediatrician's phone number, and "Aplastic Anemia (Fanconi)". Since most medical professionals know what "aplastic" means, we began with that instead of "Fanconi", a word unknown by most. We laced the tags in the shoe strings of his tennis shoes. Jake was seeing more than one doctor, so we called his pediatrician to ensure that right inside Jake's file were the names and numbers of all his doctors.

2. We wrote up a one page "information sheet" for the school. It gave the basic facts, so that a medical team could get 80% of what they needed to know by looking at a single piece of paper. It is not written in paragraph format, but instead uses bullets and a few key words. It explains that FA is characterized by bone marrow failure, gives the names of the drugs Jake takes, his current blood counts, name of his pediatrician, and gives instructions in case an emergency transfusion is ever needed. We used bold type for the most important information. Using a red marker, we colored 1/4 inch of the outside edge of the paper, so it would be easy to find in Jake's school file. The school really liked this information sheet and copied it for each teacher Jake would see during the school week. The sheets are kept in the classrooms. We certainly would be willing to send anyone a copy. Just let me know.

3. We obtained another copy of the FA Handbook and gave it to the school. That, too, is kept in Jake's file for reference.

4. We talked with Jake about his helmet. Earlier that summer he received a soft karate helmet from another FA child, Jerid DeMarco. It was too small for Jerid, so he offered it to Jake. Jake loves it. He is really good about wearing his protective gear when he rides his bike, but this soft helmet would be good for other activities. He wears it when we practice baseball out in the back yard, even though the ball is a "soft" ball, and he wears it when he is playing on his swing set/fort. I don't worry when he's just with family members, but when there is a small crowd of his friends, I worry a little more. School would be more than just a "small crowd." We talked to Jake about that and he was willing to wear the soft helmet on the bus, during recess, and gym class. He is such a sport about all this.

5. Staff at Children's Hospital in Cincinnati go to a cancer patient's school and talk to classmates and teachers about that child. We asked a staff member at Children's to talk about FA at Jake's school. She admitted she didn't know much about our disorder. We sent her the Handbook and the "information sheet" and she was willing to go to Jake's school.

We talked to Jake about this also. He was enthusiastic about having someone come to talk about blood cells, why he wears a helmet and why he doesn't do every activity in gym class. It went very well. The school is relatively small, so even though staff from Children's usually talks to the classroom, a staff member talked to the entire school assembly. The school broke into two groups, because the older kids would ask more detailed questions than the younger group. The Children's Hospital staff also talked

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A Mother/Daughter Perspective on Learning of the Diagnosis of FA

by Lynda J. Moureau, Mother and Skyleigh J. Moureau, Daughter

It all began on August 20, 1996. Skyleigh, my daughter, went to the doctor's office to have a physical examination before her sophomore year in high school. I asked them to do a blood profile which she had never had. One week later, we heard from our doctor. He said Skyleigh needed to see a hematologist at the Medical Center. The doctor mentioned that Skyleigh's blood levels were very abnormal. Red blood cells were low, her MCV (size of RBC) was very high, and her platelets were very low. We waited two weeks to see the hematologist. Skyleigh had a bone marrow biopsy to determine what was going on inside of her body. Her hematologist mentioned to us that she may have Fanconi anemia. We left the hospital wondering what was happening to her.

Skyleigh has always been a very healthy person. But I remember when she was little, she always bruised very easily. When I asked doctors about this, they would always say the same thing. Some children just bruise more easily than other children. Skyleigh has always had upper respiratory problems, as well as chronic fatigue.

We had gone camping over Labor Day weekend. We got home and on our answering machine was a message from our doctor, confirming that Skyleigh indeed had Fanconi anemia. I was very upset at her doctor for leaving her diagnosis on an answering machine. I was unable to reach Skyleigh's doctors, and experienced a tremendous amount of stress. Skyleigh's hematologist saw us Monday afternoon, and he explained what Fanconi anemia is. All I could think of was a terminal illness, that my daughter was going to die. We grieved for weeks. I felt weak and totally helpless. Finally with family support we were able to pick up and move on.

Skyleigh has always had a strong will toward life. She has always been very independent. I had to turn my feelings around about this disease. I started researching about Fanconi anemia. The Ronald McDonald House in Burlington, Vermont sent me lots of material on the disease. Through a neighbor's internet connection, I learned about the FA Research Fund in Eugene, Oregon. I called Leslie Roy, who was so kind and understanding. I no longer felt isolated. Leslie sent me lots of literature on Fanconi anemia. I made more calls and received more information. I spent a lot of time reading.

I felt Skyleigh needed me so much at this point. She was watching me fall apart, and had fallen apart as well. Who wouldn't under the circumstances? This experience has been the hardest thing to deal

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An Update on David Kwon's Bone Marrow Transplant

Sejin Kwon reported on his son's recent bone marrow transplant at Duke University. We quote in part:

As you may know, David was admitted to the Pediatric BMT Unit at Duke on December 26. He failed to engraft his baby brother's cord blood. He had two more transplants and still did not engraft. Nonetheless, he was in good clinical condition through May. We certainly had had crises like pneumonia and numerous line infections. But every time, he managed to recover despite no white cell count for such a long period of time. In early June, he had a T-cell depleted peripheral stem cell transplant from my wife, which was his fourth transplant. [David's mother was a 3/6 mismatch for her son.]

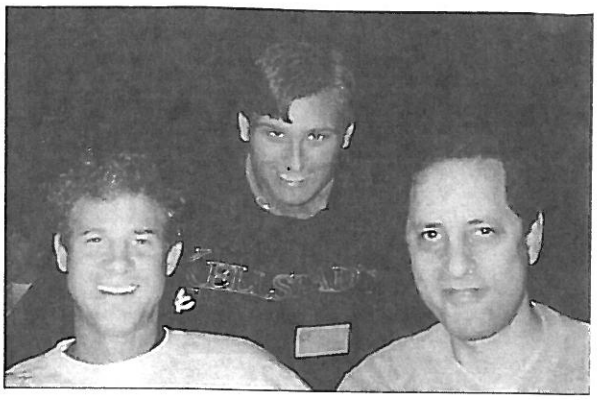
Miraculously, he has been doing well. Currently his WBC is 12.0 with 80% segs (neutrophils), and all his organs are robust. No signs of GVHD yet except a minor skin rash.

The reason we chose Duke is not just that my sister is living there. I searched the articles on FA therapy from the Medline CD-Rom, which contains all the medical papers published in major journals including BMT, Stem Cell, and Blood. I learned that Dr. Joanne Kurzberg has performed close to half of all the cord blood transplants worldwide. We consulted with specialists in Korea and the US, and concluded that Duke was the best choice for David. Heading for Duke, my estimate for success was 50%.

It's still too early to say anything about David's long-term survival. But considering his current clinical condition, I believe he'll do fine.

All of us at the FA Research Fund share the Kwons' hope that David will continue to do well! ◆

Chris Hull



Chris Hull (center) with Tom Massino (L) and Darryl Blecher (R).

I was born without a right thumb and a partially developed left thumb. I had reconstructive thumb surgery as a toddler at Rainbow Babies and Children's Hospital in Cleveland, Ohio. This gave me full use of my left thumb. I was diagnosed with FA when I became ill and my family doctor became concerned with my low blood counts. Ever since then I have had chronic bronchitis, chronic sinusitis, and sores in my mouth, and I was very susceptible to colds.

Recently I had a liver resection. Seventy-five percent of my liver was removed because of a 14 cm tumor. The tumor was hepatocellular carcinoma, most probably caused by my oxymetholone therapy. This is the first time I've really been in serious health danger since I was diagnosed with Fanconi anemia at the age of 8. The tumor came upon me quite quickly. Within a matter of a couple of weeks I became delirious. This was probably due to liver failure. I vaguely remember the last few days before the surgery. My recovery has been slow but positive. From now on I'll undergo bi-monthly CAT scans and MRIs.

Before the onset of cancer, my life was normal. I have been lucky. As anyone with FA, I've had to watch the drinking of alcohol, the blood counts, injuries, and hope that I could continue to stay as healthy as possible, hoping that a cure can be found before it is too late for me, as well as others.

I've been using the oxymetholone for as long as I can remember. I currently take 17 mg/day. In the past I've tried to cut back. I even went off the therapy a couple of times, but each time I did my blood counts dropped dramatically. Currently, I continue with the oxymetholone until another solution can be found. I have not had a BMT.

Two years ago I moved to Scottsdale, Arizona, to live in a drier climate. Fortunately for me it worked. I've had only one bout with bronchitis sinusitis, my headaches have diminished greatly in number and I am in better health than ever (with the exception of the cancer which has been removed for now). About nine months after my relocation my blood counts were as high as ever (RBC-2.77, WBC-4.0, Platelets-130). And now just three months after my surgery, my blood counts are nearly back to those levels again, and my bilirubin level (a measure of liver function) is better as well.

I'm about to turn 31 years of age. My life with FA has changed dramatically. Before the cancer came I never really thought too much about FA. I was always concerned whenever I became ill but chose to live as much as possible without FA changing my lifestyle. To say I wasn't depressed or confused would be a lie. I always kept the thought of not overcoming FA deep in the back of my mind. I was often scared.

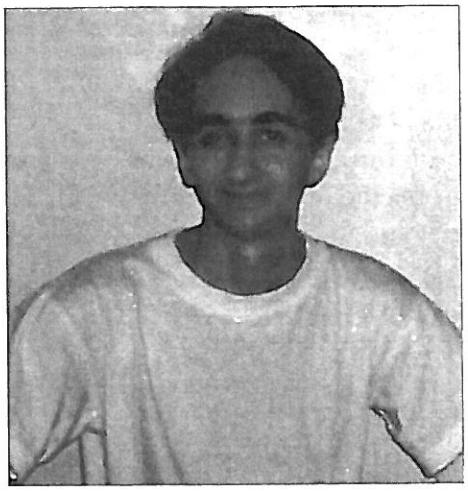
Now my life is completely different. As much as I have tried not to let it happen, cancer has changed my life. It's only been a few months so it's sort of hard to say exactly how I feel besides lucky and thankful for modern medicine, friends and family. To this point I've relied heavily on my friends and family for support. Now it's time to add my own strength, and the support of other FA individuals/families and cancer survivors to my support network.

From now on I keep telling myself to live each day as if it were my last. Sometimes it works, other times not. My attitude couldn't be better. After all, I've lived with FA for 30 years and recently overcame a very aggressive cancer. Chances are, the cancer will return. But next time I'll be a stronger person and I'll be ready for it. I feel that once I get back into mental and physical shape, I'll be ready for anything.

I'd be more than happy to talk to anyone with FA or family members of someone with FA. Actually, joining the adult support group is something that I need at this time, so I may be contacting some of you. Also, if there is anyone in the Phoenix area who would be interested in fundraising for FARF, please let me know.

As for those of you who have recently learned of your diagnosis or a family member's diagnosis, I believe success is as much mental as it is physical. Your mind and soul must be in the fight as well. Keep your chin up. Our FARF researchers are doing all they can to help us, and there are always exceptions to the rules. ◆

Giovanni Pagano wrote the following poem in memory of his son, Antonio. It is written in three parts. The first one is for the bud of promise and hope; it was written a few days before Antonio's death and hearing it gave him one of his last smiles. The second bud is one of grief and darkness. The third goes ahead to the everlasting dream of Olympia, the green leaves of the young who come, have their contests, and pass away. Yet, in spite of destructions, Olympia will never die.



Antonio Pagano

Giovanni writes "It is a special place for me in the memory of my son, as we went there together the last year before we learned of his declining health. This poem helps me get there again in a way with him."

BUDS

Buds of Hope
They're planting limes at Roman Springs
for the oncoming droughts
we shall have shady steps and scents in June
nests and trills.

Buds of Grief
Briar snow falls at Rime Springs
with claws deep nailed in your flesh
wearing out shoots and breath
the scoffing gift of berries will not repair the outrage.

Buds of Peace
Smiling willows at Olympia Springs
from every stone a memory of songs other races
defeated and victors from every laurel on the young heads
the wounded branches will bud again.

Starting School and Taking FA With You

continued from page 7

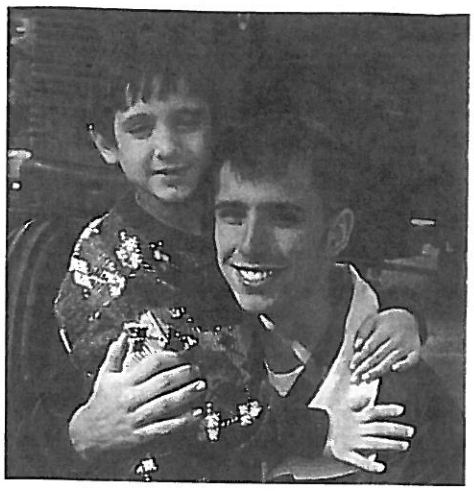
to the teachers separately. Those questions were even more detailed, as you might imagine. The entire set of talks went very well.

Staff from Children's also prepared a letter that went home with every child. The letter stated that there is a child in school with FA and that it's not contagious, but his immune system is compromised. The letter asks that if a child comes down with an illness, like chicken pox, please inform the school, so that we would be alerted. Parents have been great about calling the school about chicken pox, strep throat, and lice. The support has been great. Even the bus driver was informed of the situation and has Jake sit in the front of the bus and keeps a watchful eye on him. We were pleased with the level of questions and Jake was pleased that kids stopped asking him about his helmet, because now they understood.

These efforts have eased our worries somewhat. There have been minor injuries and the staff knew just what to do. When Jake started having nose bleeds, those, too, were handled well. Good decisions were made by the school staff and it is because of all the pre-work. Jake has been doing well in school and loves it. FA does not seem to be subtracting from that wonderful experience. For that, we are grateful. Quality of life is what it's all about. Being "normal" as much as possible is important to him and us. We can only hope that some of these experiences can help a few other families. This is a long journey we are on and helping each other out, any way we can, will ease the load. ◆

Jorge Cabrera Perez

by Jesus Cabrera (father of Jorge), Tenerife, Spain
translated by Pedro Meregotte



Jorge Cabrera Perez and Counselor Jarod

Once upon a time there was a child who got ill after his fifth birthday. At the beginning things seemed simple to take care of, but as time passed, the child who had run and played was in his house now. The doctors advised us to avoid any kind of small accidents. How to have childhood without running, scrapping or hits! But time passed by and everything became worse and worse, until one night, which seemed it would be the last one. Death came to claim, and the child saw it and smiled, and then Death fell dead because it could not resist the smile of the child. The child will survive all the bad prognoses. The doctors did not know what was wrong; they were more and more baffled. Now the child not only suffered from fever and blisters in his mouth that did not let him take food or water. He suffered with the needles that went into him and took pieces of him to look at under a microscope. He suffered with the blood that was taken from him. He suffered being in a hospital, away from his friends and with other children who also were passing their childhood in the hospital.

The parents also suffered. Everybody talked about the rare illness, orphan, little known about it. The blood was sent far, to Germany and from there they confirmed the diagnosis: Fanconi anemia. It's a name that sounds smooth, innocent. How can a tragedy be assigned? How to understand that those who suffer it will not have enough blood, that magic liquid symbol of life?

At the beginning the parents sighed peacefully, when the doctor told them "it is not leukemia". And then they discovered that it was worse than leukemia, because in the world many leukemic patients get well, but this Fanconi anemia does not offer alternatives. Nobody survives without a bone marrow transplant. And in his country, the doctors have never treated this condition. This is the most complex of the transplants, for a child with this illness. There was no hope!

And again, the child smiled. He crossed the Atlantic. He flew to small islands from which years ago his grandparents had immigrated, carrying that blood that, little by little, in him was extinguishing. The parents knocked on the doors, they searched and discovered new news every day. And a new problem evolved. Very few hospitals in the world have performed this transplant in children with that condition and not all have succeeded. Again night covered everything, but this child does not give up. He discovered a star in the sky and got there. Miracles do happen! And he found a friend. His eyes are blue. He has been for years living and dying with the children that suffer this illness! He has saved many and has seen many die. But he does not abandon them. He keeps firm holding hope! ◆

A Mother/Daughter Perspective

continued from page 8

with in my life. However, with the support of family and friends, I began to see the light. Skyleigh has a disease and each day that goes by we thank the Lord she is with us one more day and we are one more day closer to a cure. We are in a cancer support group as well as individual therapy for Skyleigh and us as a family.

Skyleigh is the same person she was before she was diagnosed. This disease is a part of Skyleigh and always has been. It's accepting the disease and moving on. Skyleigh will need a bone marrow biopsy once a year to determine how things are going. She is not on any medication. Her platelets are 62,000. She has a very positive attitude towards life. So at this point we are very grateful.

We plan to attend the summer camp in Maine. We are looking forward to meeting people with FA and their families.

I would like to thank you, Leslie, for helping me through the toughest time of my life.

~ Lynda J. Moureau

Grandma Moureau, Lynda (Mom), Bradley, Skyleigh (with frog shirt), Sacha

My name is Skyleigh and I'm sixteen years old. I've known I have Fanconi anemia for six months now, although I have lived with it all my life. I think this has been the most difficult time in my life. But I'm still here, so I guess I've got a lot more difficult times to look forward to. Being sixteen, just starting my sophomore year and finding out I have a terminal illness makes me think about life so differently. I didn't think anything like this could ever happen to me. I'm not using drugs and I'm a good student. I thought my life was finally on track.

Then one day I went to the doctor for my physical examination before school starts. A week later I went in to find out how everything was. The doctor told me I had to see a hematologist. I didn't really know what to think. My mom works in the medical field, so I knew it had something to do with the blood. But I had no clue what. The hematologist then told me I have Fanconi anemia. Well, I still didn't think a lot about it. I just walked out kind of doubtful of the whole talk.

Currently, I've been seeing a therapist to talk about what's going on and how I can deal with this. It hit me about two months ago. Depression! I was quiet and didn't want anybody's advice. I just wanted to be left alone. I was always crying and was always mad. I had a right to be: my life was miserable. I hated my friends and family. I blamed my parents for giving me this disease. Then, one day I said to myself: what am I doing soaking in my sorrow, when I could be doing everything in the world to make myself happy? I realized that I wasn't really mad at my friends or family. I wasn't mad at myself. I was just going through what I needed to go through.

Today I'm glad my family is here. I don't know what I would do without them. They have been there through thick and thin for me. I thank them so much for that.

The doctor just told me that my disease is mild, compared to some types. I think back on how I acted when there are people out there who are worse off than I am. I feel for them, and say to them that I wish and hope that I can have their strength one day.

I'm only happy that I'm not in the worst stage of this disease. And I know that people are praying for me and that I'm praying for myself. And I'll never give up!

~ Skyleigh J. Moureau

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 sponsoring 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.

An FA Memory Book

As we are all painfully aware, this past year has been extremely difficult for our FA Family Support Group. Because of this we have been inspired to create a special Memory Book for our loved ones taken from us by FA. This book is one way of remembering these special children and adults.

This project is for each and every one of you. Your input and contributions are needed to make it beautiful and thoughtful. The FA Memory Book will be a continual work in progress. We hope to fill it with pictures, poems, stories of favorite memories, and whatever is meaningful to you. You can contribute to your own loved one's page or you can contribute on behalf of your FA friends.

The FA Memory Book will be displayed each year at our Family Meeting. Please feel free to mail your materials throughout the year to Leslie Roy at the Fanconi Anemia Research Fund office. ◆

Chronicle of a Transplant

continued from page 5

"My brave sister said after all that she underwent, she would donate her marrow again if someone needed it."

Imroze's second transplant occurred on December 6. "My taste buds altered, I had mood swings and did not feel like doing anything. It was the spirit of Christmas that started enlivening me. A friend brought a Christmas tree for me and the festive atmosphere in the room made me feel better and I started taking an interest in things." Imroze subsequently developed a urinary tract infection "which kept me in agony for over a week. But a ray of sunshine was seen in the form of my blood counts which were rising very slowly. Everyone, including the doctors, was on tetherhooks, as no one could forget the engraftment period of the first transplant. It took longer than normal for the doctors to proclaim the successful engraftment of the new marrow, as this time they were very cautious.

The engraftment stage was not without its usual side effects. I experienced acute and persistent stomach pains and cramps accompanied by fever. The doctors took regular cultures, swabs, etc. I had fungal infection in my mouth which was treated with amphotericin and vancomycin." Imroze suffered from a persistent fever. A full body scan failed to reveal an infectious source.

Imroze was released from the hospital February 10, after 120 days of hospitalization. "My joy knew no bounds... I was thrilled at the thought of not going back to the hospital. My daddy rushed home on February 15 to save his job which unfortunately he lost.

My joy was short-lived. I began to feel sick at nights and I was readmitted on February 16 as I felt very sick with fever and a sore throat." Doctors found that Imroze was infected with cytomegalovirus and she was put on gancyclovir. She was discharged on March 5, 90 days after her second transplant. Imroze was readmitted on May 5 with a very high fever, given antibiotics and discharged after 6 days. She writes "I am feeling fine now. I am longing to go to the USA for Camp Sunshine.

To the readers of this chronicle I would like to say that there is hope for all if you think positively as my family did. My father plans to start a Bone Marrow Foundation to help others unable to find suitable marrow donors. He has named the foundation F & I BONE MARROW FOUNDATION after my brave sister and me." ◆

Auerbach Receives MERIT Award

As we go to press, we have just learned that prominent FA Researcher, Arleen Auerbach, has received a prestigious MERIT (Method to Extend Research in Time) Award from the National Heart, Lung, and Blood Institute of the NIH. This award "is designed to provide long-term, stable support to investigators whose research competence and productivity are distinctly superior, and who are likely to continue to perform in an outstanding manner." It can be renewed for up to ten years, and the investigator does not need to reapply. Our sincere congratulations!

FUNDRAISING

Family Fundraisers

Here are some of the family fundraisers since January, 1997:

- Chris Hull, one of our newest support group members, wrote letters to family and friends sharing a little about his life with FA and asking for contributions. Donors were very generous, especially one woman who gave $20,000!

- The Moose Lodge of Arcata, California held a spaghetti dinner and auction in honor of Jessica Paulson. Auction items, both new and used, were contributed by members of the community. Local radio DJs presided over the proceedings. Jessica and her grandmother, Marlene Stone, were interviewed over the local TV station. Enough money was raised to sponsor the Paulsons' trip to Camp Sunshine and still send in over $5,000 for research.

- We would like to thank Chris Scaffs' sisters who have informed fellow employees about the opportunity to give through United Way to our Fund. Donations from their local agency, the United Way of Southwest Pennsylvania, amounted to $2,239 during this past six months.

- Dick and Judi Selke, grandparents of Ron Dean, once again entered their losing lottery tickets from the Colorado Lottery into a special drawing for non-profits. This year they won the $10,000 prize (last year $2,000) and the money was sent to our Fund to support research.

- The Losekamp family has had some great help in raising funds. Nikki and Eric Lee's great grandfather organized a Monte Carlo Night at his retirement center and raised over $500. Plus a big thank you to nine year old Nikki, who has been selling ribbons to raise money for research.

- Children from Lauren LeRoux's day care center participated in a hop-a-thon, raised over $900 and had a great time too! Rene LeRoux continues to work hard encouraging fellow employees, friends and family to make contributions through their United Way campaigns. ◆

Tiger Foundation Grants New $75,000 Challenge

Families who attended Camp Sunshine were cheered by the promise of new research dollars from the Tiger Foundation of New York. Tiger will match the next $75,000 dollars raised by FA families between May 7 and December 31, 1997.

We send profound thanks to Gerald Norris and Trustees of the Tiger Foundation for their generous philanthropy and commitment to our mission.The rapid progress of FA research has been hastened by their generous support. When this challenge is reached, the Tiger Foundation will have awarded nearly $600,000 to FA Research over the last six years. An additional $400,000 has been raised in matching funds by families.

As this newsletter went to press, $11,981 had been raised toward the new $75,000 goal. Several families report they have special summer and fall events on the drawing board, which promise to give the campaign more momentum. They discover that donors are even more motivated to give when their gift will double. Simply ask your contributors to write "Tiger Challenge" on the memo line of their checks. ◆

Newsletter Benefactors – Printing by Image Masters, Unisource Paper, Western Paper, Bindery West, and The Edwin and June Cone Fund of the Oregon Community Foundation

Consultants – Tanya Harvey, Wild Iris Graphics and Darren Hunt, E&D Graphic Services

Family Fundraising Efforts

From January 1, 1997 through June 30, 1997, sixty-four families raised funds or made individual contributions for a total of $203,887.93. Miscellaneous contributions donated through United Way and the Combined Federal Campaign totaled $3,825. If your family or friends are giving through either of these agencies, please let us know so that the contributions can be credited to your family. We sincerely appreciate the many donations made in memory of FA children and adults.

We once again thank Mike and Beth Vangel for producing the Fundraising Booklet.

Many families have requested a copy to help plan future fundraising events or letter campaigns. Copies are still available through the FA Research Fund. Our office staff is always willing to assist in any way possible. Remember fundraising needs to be an annual occurrence with the understanding that every effort, big or small, helps.

We appreciate all the hard work that goes into raising funds for research and family support. Without the endeavors of FA families, discoveries like the FA-A and FA-C genes would not have been made. It's only as long as our group is united in setting and reaching our financial goals that progress will continue.

We are deeply grateful to the following families who worked hard to support our research fund over the past six months:

$90,000 - 100,000

Dave, Lynn & Kirsten Frohnmayer

$10,000 - 30,000

Chris Hull
Teddi Matlack, Dick and Judi Selke
Andrew & Vicki Athens

$5,000 - 8,000

Pat & Maria Gleason
Jeff & Beth Janock

$2,000 - $5,000

Ken & Jeanne Atkinson
Day Family
LeRoux Family
Paulson Family
Jeff & Debby Slater
Marc & Sandi Weiner

$1,000 - $2,000

Al & Janeth Acosta
Cheryl Banks
Mark & Linda Baumiller
Chris & Susan Collins
Pat & Bill Danks
Bill & Jackie Lucarell
Stuart Cohen & Deane Marchbein
Jack & Tannis Redekop
Chris Scaff
Bill & Connie Schenone
Alan Goldberg & Laurie Strongin
Richard & Janice Thomas

$500 - $1,000

April Benton
Greg & Diane Hayes
Keleher Family
Eric & Beth Losekamp
Jack & Lisa Nash
Terry & Therese Robertson
Erik & Lori Salo
Mark & Susan Trager

Up to $500

Pastor & Mrs. Charles Barnhart
Ed & Barb Brookover
Ceresa Family
Susan Combs
Josh & Susan DaRosa
Joseph & Tracy DeMarco
Pat & Mary DiMarino
Ed & Janice Duffy
Neil & Iris Frank
Gary & Melody Ganz
Amber Garthus
John & Karilyn Kelson
Robert & Jennifer Kiesel
Kwon Family
Mike & Myra Lewis
Lowrimore Family
Jack & Pam McCarty
Steven & Alison McClay
Cecelia Meloling
Ron & Fredi Norris
Kevin & Lorraine O'Connor
Vicki Phillips
Aaron & Jean Randolph
Dan & Bonnie Rosen
Rick & Lynn Sablosky
Bryan & Karen Siebenthal
Brad Simon
Paul & Debra Sundsvold
Dennis & Sharon Swanson
Mike & Beth Vangel
Lynn Welfare
Reese & Nancy Williams

"The capacity to care is the thing that gives life its deepest meaning and significance."

- Pablo Casals

Camp Sunshine volunteers are super friends!

Family Meeting Sets Record

continued from front page

While the group consoled the numerous families who lost loved ones in recent months and years, we were heartened by the bonds of friendship and outreach that united both first time participants and long-time attendees.

We all express continuing gratitude to Larry and Anna Gould and their dozens of volunteers whose vision and selflessness create large islands of respite and beauty amidst this sea of loss and anxiety. ◆

New Cord Blood Regulations

continued from page 2

heavily, like drugs. However, Cynthia Fischer, Viacord president and founder is pleased with the new approach. "In the case of private banking firms like ours," says Fischer, "the new policy reduces the FDA oversight, since the collected and stored blood is staying within a family."

Under the new guidelines, all blood banking services must follow strict disease-screening requirements to ensure safety. But now they will not need premarket approval to serve families in need, in part, because they are not altering the cord blood.

The FDA has reserved high levels of regulation for those tissues that undergo manipulation to alter their function, such as genetically altered cells injected into patients to supply missing or malfunctioning DNA. This and other therapies would require the same kinds of controlled clinical trials that all new drugs and medical devices go through under the new FDA plan.

Thanks to patient families and treating professionals for whom cord blood is a lifeline, the FDA has a new understanding of the complex and dynamic field of stem cell therapy from which it can draft practical guidelines. Mary Pendergast, deputy commissioner of the FDA, says the FDA will hold public hearings over the next two to three years, after which final rulings will be made. ◆

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Image Masters Printers, Eugene, OR

Support for Distribution
The Edwin & June Cone Fund of the Oregon Community Foundation