



FAMILY NEWSLETTER

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



Families Learn from Experts, Share Experiences at Family Meeting

Twenty-nine families from the United States, Canada, Norway, and Germany met at Aurora University at Lake Geneva, Wisconsin, for our 10th annual FA Family Meeting. Evaluations were extremely positive concerning the content of the scientific sessions, the value of the group discussions led by Nancy Cincotta, and the unique opportunity for friendship and sharing with other affected families. A parent new to this disease stated he had learned more about FA in one day than in six months since diagnosis. Another commented that the family meeting provided a chance to consult with and learn from the best experts from around the country, in just one weekend. And a seasoned FA father declared that if he could make

only one trip a year, it would always be to this Family Meeting, for he always learns so much that helps in treating and planning for his children.

Our numbers were smaller this year. This past year we have lost children and young adults who were extremely active in our group. Many families are in transplant, or recovering from transplant. And some families find the cost of our Family Meetings beyond their reach. Our Fund will brainstorm on ways to assist those who want to come, but cannot afford the cost. Family camp provides a learning opportunity that is unparalleled, and an opportunity to share feelings and insights with others that cannot be duplicated in any other way. All who

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FA and Cancer

FA families know that bone marrow failure affects almost all FA patients, and is frequently the cause of death in this population. We have worked to develop life-extending therapies, and have devoted resources to improving bone marrow transplant outcomes, especially for patients lacking matched sibling donors. As a result, FA patients are living longer and transplant statistics are steadily improving.

Increasingly, however, FA families have been alarmed by the large number of patients, transplanted or not, who have developed solid tumor malignancies. These concerns have been reported in previous issues of the newsletters, and we have stressed the importance of early detection.

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Cloning of the Fanconi Anemia Group D Gene

In 1995, a group of researchers in Portland, OR, led by Markus Grompe, MD, mapped the Fanconi anemia group D gene to chromosome 3p. This area was then narrowed to three possible genes. Very recently, mutations from two patients' cells were found in one of these genes, proving that the *FANCD* gene had been cloned. There are four unique characteristics concerning this important discovery:

1) Not all complementation group D patients have mutations in this gene. For example, the cell line used to define FA-D does not have mutations in the new gene. It is therefore likely that FA-D represents at least two distinct genes. Because another cell line is the reference cell line for group D, the new gene is called *FANCD2*.

2) Many other multicellular organisms including plants, fruit flies, and worms, have a *FANCD2* homologue.

This is in contrast to other FA genes isolated to date, which seem to exist only in vertebrates. This is important for the future study of the FA pathway, since these other organisms are comparatively easy to study.

3) The FANCD protein resides in the nucleus of cells and exists in two forms, termed FANCD2-S (short) and FANCD2-L (long). Cells from over 95% of all FA patients contain only the short form of the protein. Normal cells contain both the short and long form of the protein.

4) FA can be diagnosed by a rapid antibody test which determines whether both forms of the protein are present or not. If only the short form of the protein is present, the patient has FA. See related story on "New Diagnostic Method for Fanconi Anemia" below. ♦

New Diagnostic Method for Fanconi Anemia

The DEB test has long been the "gold standard" for diagnosing Fanconi anemia. But this test does have its drawbacks. It can generate false negatives in the case of FA mosaicism, requires a high degree of technical expertise, and it cannot detect carriers of FA. Researchers at the Dana Farber Cancer Institute and Oregon Health Sciences University have developed a rapid, improved diagnostic test for FA, which exploits recent advances in the molecular understanding of the FA pathway.

Alan D'Andrea states that the inability to produce normal proteins

by any one of the FA genes which function upstream of *FANCD2* (e.g., *FANCA*, *FANCB*, *FANCC*, *FANCE*, *FANCF*, and *FANCG*) blocks the ability of the FANCD2 protein to be modified from the short form of its protein to the long form. Absence of the long form of this protein, therefore, indicates that a patient belongs to one of the upstream complementation groups and has Fanconi anemia. FA-D2 patients have no FANCD2 protein. Researchers have used a combination of retroviral gene transfer and *FANCD2* analysis to assign patients to specific complementation groups. ♦

Six FA Genes Have Been Cloned!

We now know that there are at least seven FA complementation groups (A-G). Six FA genes have now been cloned. The first FA gene (*FANCC*) was cloned in 1992 by Manual Buchwald, PhD, Hospital for Sick Children, Canada. Markus Grompe, MD, Oregon Health Sciences University, has recently cloned *FANCD2*. The other four genes (*FANCA*, *FANCE*, *FANCF* and *FANCG*) were all cloned in the laboratory of Hans Joenje, The Netherlands. Laboratories supported by FARF have made astounding progress during the past eight years! ♦

Clonal Abnormalities in FA

John Wagner, MD, Minnesota, believes that all FA children should have yearly bone marrow examinations with cytogenetics, to evaluate the possibility that the marrow is progressing towards myelodysplastic syndrome (MDS) or acute myelogenous leukemia (AML). The presence of a clone (two or more cells with the same chromosomal abnormality) frequently but not always suggests such a progression and should be closely monitored. According to Wagner, once a clonal abnormality is identified, more frequent examinations are indicated (every 3 to 4 months). See the *Science Letter* for additional information on clonal abnormalities in FA patients. ♦

Two Patients with 4/6 HLA-Matched Donors Do Well

Farid Boulad, MD, Memorial Sloan-Kettering, reports that the two FA patients he transplanted with HLA 4/6 matched, related donors are alive and well, one more than two years post-transplant, the second 17 months post-transplant. His protocols, which include fludarabine and T-cell depleted peripheral blood stem cells, are described in the *FA Family Newsletter* 26. Both patients engrafted very early (on days +10 and +11) and had a fully reconstituted immune system in eight months. A third patient, receiving an unrelated 6/6 bone marrow transplant, is also doing well, but experienced more infectious complications than the two previous patients. Boulad wondered if the use of peripheral blood explained the rapid immune system recovery. Since the first two patients are of Korean ancestry (where minor HLA antigens are more likely to match), it is impossible to know yet what accounts for these outstanding results. ♦

Hans Joenje, PhD, The Netherlands, confirmed earlier work of Drs. Hanenberg of Germany and D'Andrea of Boston that the cell line for complementation group H really belongs in complementation group A. Therefore, that cell line does not define a separate complementation group.

Matched Related Donor Transplants

One year ago, this newsletter reported on two successful bone marrow transplants performed in Israel under the direction of Dr. Shimon Slavin, using matched related donors, fludarabine, and no irradiation. The transplant protocols are detailed in the *FA Family Newsletter* 26. Slavin has now performed five such transplants using this protocol. One transplant is too recent to be evaluated, but all five patients are alive and well. The University of Minnesota and University

Children's Hospital, Charité, Berlin have each done three matched related transplants, using fludarabine. Five of these patients had no irradiation. One Minnesota patient who received maternal bone marrow required a second bone marrow infusion after limited field irradiation. All six patients are alive and well. Please read articles by MacMillan and Ebell in the *Science Letter* for specific details on the protocols utilized. ♦

Preimplantation Genetic Diagnosis Yields Healthy Matched Sibling

On August 29, 2000, Lisa and Jack Nash became the proud parents of Adam Nash. Adam has made history as the first baby born to FA carriers following preimplantation genetic diagnosis. Because of this procedure, Adam's status as a healthy baby and perfect HLA match for his FA-affected sister, Molly, were known before Lisa became pregnant.

Preimplantation Genetic Diagnosis (PGD) was developed for couples at high risk for conceiving children with genetic diseases. PGD involves stimulating a woman's ovaries with hormones so that she will produce several eggs. The eggs are removed and fertilized with the husband's sperm. When the embryos contain eight cells, one cell is removed. This cell can be HLA tissue-typed and tested for the absence of a genetic disease. In Lisa's case, the successful pregnancy took place on the fifth attempt. Sixteen eggs were fertilized, and only one embryo was both disease-free and a perfect match for Molly. This embryo was transferred to Lisa, and a pregnancy developed.

For this procedure to be successful, it is necessary to know not just

one's complementation group, but at least one specific disease mutation. According to Dr. Charles Strom who spoke at our Family Meeting, the procedure is expensive and usually is not covered by insurance. Cost depends upon location. In New York City, each attempt costs \$16,000; in Chicago the cost is \$8,000; and in Los Angeles the price tag is \$4,000.

Strom stated that approximately 60% of all couples get pregnant after three cycles. One hundred and fifteen (all healthy) babies have been born following this procedure, mostly to parents carrying genes for cystic fibrosis.

Parents who have attempted PGD report that the process is extremely stressful, perhaps due to the combination of hormonal stimulation and the failed attempts that can result. One FA family underwent nine failed attempts at PGD before opting for a matched, unrelated transplant. Nonetheless, when it works, families experience the intense joy of carrying a healthy child and minimizing the risks of transplant. We are all delighted for the Nash family! See Dr. Strom's article in the *Science Letter*. ♦

Alternate Donor Transplants for FA

Gradually, over the past decade, bone marrow transplant results for patients lacking matched sibling donors have shown steady improvement. The recent addition of fludarabine to the preparatory regimen has improved rates of engraftment and overall outcomes. We are aware of recent transplant results in four major centers: University of Minnesota; University Children's Hospital, Charité, Berlin; Memorial Sloan-Kettering in New York City; and Children's Hospital, Cincinnati.

A total of 22 patients with alternate donors have been transplanted at these four centers using fludarabine. Patient characteristics and protocols have differed considerably from one center to another (see articles by MacMillan and Ebell in the *Science Letter*.) For example, Dr. Ebell from Germany has eliminated Cytoxan and irradiation, but uses marrow which has not been T-cell depleted, and busulfan. It is too early to know which approach will ultimately be the most successful for which patient. Yet it is encouraging to note that well over 50% of these patients are presently alive. The numbers are very small and the time post-transplant is very short for some patients. In spite of that disclaimer, patients are surviv-

ing at higher rates, and families have renewed reason for optimism.

In her presentation at our Family Meeting, Dr. MacMillan cited infection as the major cause of death in this population. She believes patients often enter transplant with incipient infections, which become unmanageable after immunosuppression and the lengthy period of time needed to reconstitute an immune system. She

suggests a trial of androgens prior to transplant. But at the first sign of falling counts, and before the absolute neutrophil count (ANC) is consistently below 500, families should start planning for transplant.

MacMillan also expressed concern that 40% of post-transplant FA patients develop a solid tumor malignancy within 15 to 20 years post-transplant. ♦

Gene Therapy Trial Begins at Chapel Hill, North Carolina

Christopher Walsh, MD, PhD, reported on four FA-A patients, ages 11-48, who enrolled in an FA gene therapy trial at the University of North Carolina Gene Therapy Center. All four had severe bone marrow failure. Walsh collected stem cells from the peripheral blood of two patients, and stem cells from the bone marrow of two patients. The two bone marrow harvests contained more CD34 cells than the peripheral blood harvests, but amounts were still extremely low. After transferring the normal gene into the harvested cells, Walsh returned corrected cells to the patients. He noted that he was able to return to the

patients only a small fraction of what one would give to someone with normal blood counts.

One month after gene therapy, Walsh believed that results from one patient were encouraging, in that the blood counts were slightly increased. In two patients there was no early response. Walsh believes that it is too early to see if this approach will be effective in these patients. A fourth patient died of head and neck cancer, unrelated to gene therapy.

Walsh will continue to explore different target cells, new gene transfer vectors, and possible ways of expanding cells. Based on a recent trial in France, he believes gene therapy can be successful. In this trial, four patients with Severe Combined Immunodeficiency Disease (SCID) were given gene corrected cells. Their blood counts improved, and have stayed at higher levels for over a year and a half.

Walsh added that the best candidates for FA gene therapy might well be patients who still have good bone marrow cellularity. Treating patients with severe aplastic anemia may not yield a sufficient number of correctable cells. ♦

The Twelfth Annual International FA Scientific Symposium

OCTOBER 26-29, 2000

Novotel Hotel, Amsterdam, The Netherlands

We thank researchers at the Free University, Amsterdam, for their generous invitation. This location strengthens our outreach into the international research community, and made it possible for a larger number of European researchers to attend.

FA Patients and Endocrine Disorders

Michael Wajnrajch, MD, Cornell Medical Center, reported on a joint project he is undertaking with Arleen Auerbach, PhD, of The Rockefeller University, to evaluate FA patients for endocrine disorders. To date, 70 patients have been evaluated. Endocrine disorders are a common finding in FA patients and may manifest as short stature, growth hormone (GH) deficiency, hypothyroidism, and abnormal glucose/insulin metabolism, leading to impaired glucose tolerance or diabetes mellitus. Wajnrajch recommends a thorough endocrine evaluation of FA patients.

At our Family Meeting, Wajnrajch discussed the question of treating short FA patients with GH. According to Wajnrajch, 38 FA patients throughout the world have been treated with GH. Of these, three died: one from leukemia, one from a severe infection, and a third from an undetermined cause. None of these deaths was directly attributed to the GH treatment. Two GH-treated patients reported problems with glucose intolerance. Wajnrajch concludes that GH appears to be a relatively safe treatment in FA, but there are only minimal data on which to make this assumption. ♦

Reducing Toxicity - Early Transplant Results in Germany

Wolfram Ebell of Humboldt University, Berlin, reported on eight FA transplants at our Family Meeting (see *Science Letter*). Ebell sought to reduce toxicity and secondary tumors by avoiding high dose chemotherapy and especially radiation. He tried to achieve engraftment by the use of fludarabine, high stem cell doses and T-cell antibodies. Most of these patients would be considered high risk for a variety of reasons, including myelodysplastic syndrome (MDS) with multiple cytogenetic abnormalities in three patients and acute myelogenous leukemia (AML) in two patients.

Six of the eight were surviving with functioning grafts between one and 18 months post-transplant. Of the six survivors, three had matched sibling donors; three had alternate donors. The two who did not survive had alternate donors. One died from progression of AML in spite of three transplant attempts. One patient died due to transplant related complications.

Two of the survivors required a second transplant.

Based on the results of this early trial, the transplant protocol has been altered, in the hope of further reducing toxicity and increasing sustained engraftment. ♦

Editors' Note and Disclaimer

Statements and opinions expressed in this Newsletter are those of the authors and not necessarily those of the editors or the Fanconi Anemia Research Fund. Information provided in this Newsletter about medications, treatments or products should not be construed as medical instruction or scientific endorsement. Always consult your physician before taking any action based on this information.

FA and Cancer

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In response to our need for more reliable data, Dr. Blanche Alter agreed to do a survey of FA patients in the United States to determine the prevalence of cancer in this population, and the risk of developing malignancies in the future. The results of her preliminary study are sobering.

In January 2000, Alter sent a survey to 284 FA individuals whose families belong to FARE. By July 2000, 127 responses were received (45%). The absolute prevalence of cancer was ~20%, with leukemia accounting for one-third of the malignancies, and cancer of the head and neck accounting for another third. Males were as likely as females to develop cancer. The cumulative risk of leukemia was ~25% and leveled off at age 25, while the risk of solid tumors did not level off, and was >90% by age 45.

With or without FA, our entire population is cancer prone. However, a much larger percentage of FA patients will develop malignancies, and these cancers appear much earlier than in the general population. For example, the median age for developing acute myelogenous leukemia (AML) in the general population is 67, but in FA patients, the median age is 14. For cancers of the head and neck, median age in the general population is 64, compared to 25 in FA patients.

Data from the above study were self-reported and not validated by a physician. The numbers are very small. Alter plans to continue this study, and we urge all families to help by returning her questionnaire. Based on the above, we must conclude that FA patients are at very high risk of cancer. Our organization must devote resources to understanding and treating this devastating complication of FA. ♦

Amifostine for the Treatment of Fanconi Anemia

by Eva Guinan, MD, Dana Farber Cancer Institute, Boston, MA

At the Children's Hospital in Boston, we are conducting a study of a possible new treatment for Fanconi anemia.

Why is this study being done?

In this research study of up to 21 patients, we are testing to see if treatment with a drug called Amifostine can help stimulate the bone marrow to make more red blood cells, platelets and white cells. Amifostine is a drug which has been used before to help prevent side effects of cancer chemotherapy. It has also been used to stimulate the production of blood cells in other bone marrow failure syndromes. However, it is currently not FDA approved for use in treating bone marrow failure.

Who will participate in this trial?

Patients with Fanconi anemia who are aged 2 and over with bone marrow failure and who have not had a bone marrow transplant are eligible for this trial. Patients may not be treated with growth factors or androgens (steroids) while they are on the trial.

What is the procedure?

The drug will be given into the vein over 3-5 minutes three times a week for three weeks. Patients would need to come into the General Clinical Research Center at Children's Hospital, Boston, for about 3-4 hours each time so that we can examine you/your child each time before the dose and monitor patients after the dose is given. Patients will also have a bone marrow test at the start and at the end of the three-week period. We will also draw 1-5 teaspoons of blood for tests at the start of the study, every week during the study, and for three weeks afterwards.

What are the potential benefits?

We hope that the treatment with Amifostine will increase the levels of red blood cells, platelets and white cells in your or your child's blood. This may reduce or delay the need for transfusions and may reduce the risk of infections. However, it is also possible that there may be no direct benefit from treatment with Amifostine.

What are the potential risks?

Some patients who have received Amifostine have experienced low blood pressure, low levels of calcium in the blood, nausea and vomiting. We expect these side effects to be rare because we are using low doses of Amifostine. However, it is also possible that other side effects, which have pre-

viously not been seen, may occur. Patients will be monitored closely after the medicine is given and in the event of any side effects will be treated appropriately.

Whom should I contact to learn more about this trial?

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Dr. Eva C. Guinan
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Is Cancer in Non-FA Adults Related to Acquired Problems in FA Genes or Their Proteins?

Hans Joenje has observed that acute myelogenous leukemia (AML) in FA patients is similar to that same disease in non-FA adults. He wondered if non-FA patients with AML had acquired a defect in an FA protein. He studied 10 AML cell lines from non-FA adults and found abnormal FA protein patterns in 5 of these 10 cell lines. He then studied 15 fresh AMLs from non-FA patients, and 11 of these had abnormal FA protein patterns. This suggests the strong possibility that a disturbance in the FA pathway can lead to AML in otherwise healthy adults.

Joenje also noted that FA oral tumors are very similar to non-FA oral tumors. He is now looking for abnor-

mal FA protein patterns in non-FA patients with oral squamous cell carcinoma.

In non-FA patients, environmental factors such as cigarette smoking are clearly linked to oral cancers. It is therefore possible that environmental factors also contribute to oral cancers in FA patients. Joenje stresses the importance of a diet rich in fresh fruit and vegetables, and noted that some scientists believe that tomatoes and tomato-based products are particularly effective for the general population. There are no current data suggesting that these products help FA patients, but nonetheless one cannot discount the possibility that a benefit may be achieved. ♦

FAMILY NEWS

A Time of Heartache and Hope

by Mrs. Shaikh, Plaiston, London

This past year has been so difficult. Nine years after Mariam's death in 1991, we wanted to try once more for a baby. We knew that we would terminate the pregnancy at any stage if an abnormality was found. I had scans every two weeks, each lasted 2-3 hours and everything was checked. A long list of every part of the baby's body was marked normal. We wanted to have a chorionic villus sampling (CVS) test, but it was considered too risky. However, an amniocentesis was normal.

It was a lovely pregnancy. I was able to work up to my eighth month and I felt great. A doctor at Guy's Hospital, London monitored the pregnancy. We really thought that we were lucky this time. A girl or boy would be lovely, as long as the baby was healthy.

I was to have a Caesarean section, so a date was set. On December 4, 1998, I had an epidural so that I could see the baby as soon as it was born. My husband was with me, and a baby boy was delivered. I gave him a quick kiss and he was taken for his check-up.

We were so happy.

But this was not for long. When the doctor came in, his face said everything. At this point, I started to hemorrhage badly, so was put under general anesthesia. My husband started to panic. The baby was taken to the special baby care unit, and I went to the surgery room. My husband was told that if they could not stop the bleeding, they would need to perform an emergency hysterectomy.

The doctor said our baby had only 48 hours to live. We could not believe this nightmare.

Murad Shaikh was born with no anus, multicystic kidneys, and a short radius in his right arm. They performed a colostomy when he was one day old. He was then found to have a duodenal web, and was operated on a week later. In spite of the odds, he survived. We lived in the hospital for three months. He came home on Mothers' Day, 1999.

Murad is now 21 months old and is such a happy little boy. He dances to



music and sings; he loves the teletubbies and his nanny. He plays well with his sister Annie. He has such a beautiful face. He has developed well, considering his bad start. He is on a lot of medication. We go to the hospital once a month for blood counts and developmental checks.

All our plans for the future are on hold. Once again, we fear the future. We are doing our best to carry on. Each day is a blessing. We try to think positively, and we will give our precious little boy the best. We have had

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Paula Ceresa Battles Precancerous Mouth Condition with ONYX-015

Approximately five years ago, a dental hygienist noticed a white area on the rear palate of Paula Ceresa's mouth, and notified the dentist. Over the next three years, three different doctors observed the area, and none was concerned. Hearing this story, Ralf Dietrich, Germany, urged Paula to get another opinion. An oral surgeon performed a biopsy, and the diagnosis was microinvasive squamous cell carcinoma of the palate. Areas on the gum and

cheek were diagnosed as moderate and severe epithelial dysplasia. The oral surgeon wanted to freeze these areas, but Paula had heard this was not advised for FA patients and sought another opinion. Another oral surgeon opted to excise these areas. The diagnosis came back as cancer, not severe dysplasia.

Things remained stable until October, 1999, when another mouth area was diagnosed as moderate to severe dysplasia, and was removed. Biopsy of

the tongue showed hyperplasia.

Last spring, Paula learned of an experimental drug called ONYX-015, used to treat precancerous lesions of the mouth. This drug is an adenovirus which has been altered so that it cannot grow in most cells, and should not cause damage to normal cells. However, this virus can grow in cells with mutations in the p53 tumor suppressor gene, and can kill these cells. The

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Fanconi Anemia is for Life

by Jan Turner, New Zealand

Recently our daughter Kelly (17 years old) was found to have a malignancy on her tongue. Kelly had a successful unrelated bone marrow transplant in Paris in 1992. I can remember thinking that as long as Kelly survived the transplant that the worst would be over, we could cope with whatever else was thrown at us. I guess it was easy to slip into complacency. Life has been pretty good and easy over the last 6 or so years.

In a way it was like slipping into denial. OK, my daughter has survived a very difficult transplant, we've achieved it. We've saved her life. Life will be sweet from now on. My wishful thinking gave me a respite for a length of time. It was wonderful going along to checkups with the many specialists whom Kelly sees as each time there were no major problems, life was great.

Even when the growth was found on her tongue it was expected to be non-malignant. The ENT specialist was so sure. But oh the shock when the histology results came back. It was malignant and a large portion of her tongue would have to be removed. Shock, horror, etc. Why has this happened? It can't be happening. Kelly survived the bone marrow transplant, she's been through enough. Why is this happening? Well, it doesn't matter how much I may have "bargained," hoped or prayed. Kelly still has Fanconi anemia.

I consider myself a fairly intelligent person. So how is it that even though I have saturated myself with everything I can learn about Fanconi anemia that I managed to slip into denial? The relief of getting Kelly through the bone marrow transplant had a lot to do with it. I can remember driving myself through that time, promising

myself if I could only get Kelly through it alive I could cope with anything else. It was probably necessary to do it that way. Maybe it gave me a strength I needed. But who am I to say, "Thanks for the test, but don't send me anymore, please."

Now reality has hit like a Mack truck. Fanconi anemia is for life. Yes, it is important to deal with the bone marrow failure and it is a major problem. Years ago there was no hope for FA. Bone marrow transplants were not an option or were too experimental. Now our FA children are surviving, they are being given chances. But cancer is a real threat. We know they are at risk. Please remember this: be care-

ful not to let complacency take over. We must watch our kids very carefully for any signs of malignancies and deal with them as early as possible. FA is not a short term problem. It is life-long. Let's continue learning all we can and fight this together.

I am so glad to hear that Blanche Alter has accepted a position at the National Cancer Institute and will be focusing her work on FA and subsequent cancers. How lucky we are to have such special people on our side. It gives me so much comfort to know so many others around the world are able to make a difference for all FA families. ♦

Early Transplant Results Promising

by Laurie Strongin, Washington, D.C.

Henry Strongin Goldberg, age 5, had a 5/6 unrelated bone marrow transplant at The University of Minnesota Medical Center (Fairview) on July 6, 2000. Our decision to move forward with the transplant was strongly influenced by his week-long hospitalization in February due to pneumonia. At that time, he had been on androgen therapy for two years and his platelets had been stable at about 30,000; white count around 2.5; and hemoglobin around 9. After the pneumonia, Henry needed two platelet and two red cell transfusions within a month's time. We became concerned about his becoming transfusion dependent and decided to give him the best chance at a long life. We would go to transplant before his bone marrow further deteriorated.

Henry's white cells came in at Day 11. His Day 21 bone marrow biopsy indicated that he had 100% donor cells with a cellularity of 10 percent, very encouraging information. Today is Day 67 and we just returned from his second post-transplant biopsy and aspiration. We will get results later in the week. Today Henry's platelets are at 55,000, his white count is 4800, and his hemoglobin is 9. He has not had any transfusions for a month and hasn't had a fever since Day 6. There have been no significant complications and no rehospitalizations.

So far, we feel blessed with his progress. His attitude amazes us. He and brother Jack are having so much fun being back together after a 35 day separation. If things continue to go well, we will head home at the end of October. If anyone has any questions, please e-mail me at lauriestrongin@hotmail.com. ♦

An Alternative to Oxymetholone: One Family's Experience

by Lynn & Rick Sablosky, Winsted, CT

Our daughter was diagnosed with FA at the age of 10 years. At age 12, her counts were: Hgb 7.2, WBC 2.2, and platelets 34K. She began taking 50 mg/day of oxymetholone. Her counts increased gradually over the next 3 months and averaged: Hgb 12.0, WBC 2.5-3.0, and platelets 60K-70K. We discontinued the oxymetholone after 9 months due to the development of a clone in her bone marrow, which is a marker for AML in the general population. She also was experiencing the side effects typical of oxymetholone use: deepening of the voice, excess facial hair, weight gain, acne, and emotional swings (the most difficult effect for her to deal with). Five months later the clone was no longer detectable and has not reappeared in five years. Her Hgb gradually fell over the next 12 months to 8.2, and we again needed to consider androgen therapy. During that year her WBC remained in the 2.5 to 3.0 range, and her platelets averaged about 50K.

At this point our daughter was 15 1/2. Due to the extreme emotional lability induced by the oxymetholone, neither she nor we were willing to use this drug again. Therefore, we chose to try danazol, an attenuated androgen, to stimulate her bone marrow. Our doctor's first choice would have been oxymetholone, but we were resistant to subjecting our daughter to its side effects again. We all agreed we would give danazol a 3 month trial period and would transfuse if necessary during that 3 month time frame. We would return to oxymetholone if there were no response on danazol.

Danazol/Danocrine is a drug historically used to treat women with endometriosis and fibrocystic breast

disease. The therapeutic dose is 100 mg to 400 mg in two divided doses per day, depending on the disease and the patient response. Many of the side effects that are listed on the drug insert are the same as for oxymetholone: i.e., weight gain, acne, edema, voice change (which may take the form of hoarseness, sore throat, or instability or deepening of pitch), mild facial hair, and emotional lability. Enlargement of the clitoris is rare. As with oxymetholone, patients must be monitored for liver dysfunction. Abnormalities in laboratory tests (such as the glucose tolerance test) may occur during therapy with danazol. Consult your physician concerning the meaning of laboratory test results while taking this drug. Typically, drug inserts list potential side effects and reactions too numerous to mention here. However, an increase in red cell and platelet counts was cited as a possible side effect. A causal relationship between blood counts and use of danazol has not been confirmed nor refuted.

Fortunately, our daughter responded nicely to a dose of 400 mg of danazol a day (200 mg twice a day). Her Hgb and platelets gradually climbed over a 4-month period to a high of 12.0 for Hgb and 60-70K for platelets. Her WBC remained in the 2.5-3.0 range. We also gradually reduced the dose from 400 mg a day to 200 mg a day, then to 200 mg every other day. Her counts remained stable at 200 mg every other day. After taking danazol for 7 months and maintaining her counts at the relatively low dose mentioned, we discontinued the drug because of the emotional lability side effect (specifically, anger management). This was the only side effect that she experienced. Her voice

had returned to normal after discontinuing the oxymetholone. She has been plagued by acne, but that condition has been a problem with and without androgen therapy. Over the next 6 months she maintained decent blood counts, but with a gradual decrease of her Hgb from a high of 12.0 to 9.5, and platelets from a high of 70K to 50K.

Because she is symptomatic with a Hgb below 10, we decided to begin danazol therapy again with a dosage of 200 mg a day. Two months after beginning the danocrine, her Hgb climbed from 9.5 to 10.6. Her platelets are 75K. Her WBC is unchanged at 2.6. Every other time she has been on androgen therapy, her counts have reached their maximum after 3 to 4 months.

We are aware that danazol is not the first drug of choice for Fanconi anemia. However, after our daughter's experience with both oxymetholone and danazol, we and she certainly prefer danazol based on the minimal side effects she experienced as compared to oxymetholone. As with any drug therapy for any condition, we are diligent about reading all the information that is available concerning side effects and then proceed to make our best decision. We hope this account is beneficial to other parents (particularly of daughters) when considering androgen therapy. Confer with your own doctor, of course. ♦

Luke's Story

by Jacqueline Jones, Wales, United Kingdom

Luke was born on 6/17/89 at Heath General Hospital, Wales, U.K., weighing 5 lbs 12 oz and measuring 36 cm. Apart from jaundice which resolved in three days, he appeared very healthy. He came home and met his sister, Sharlene, now age 14.

Three months later, Luke was found to have a painful hernia in his groin, which required surgical repair. Then Luke developed an eating problem. He would take in very small amounts of food, and began losing weight. A dietician placed Luke on a powder known as Duocal, meant to be odorless and tasteless when added to other foods. Luke's weight continued to decline. When our daughter accidentally ate cereal containing Duocal, she became sick because of the foul taste. I had been adding this to Luke's food on a daily basis, with no idea that it tasted so horrible.

I next noticed that Luke wasn't grasping things properly. Luke was sent to a plastic surgeon and I learned that he had a hyperplastic left thumb. Luke spent his second birthday in the hospital recovering from an operation to rebuild a thumb. On one doctor visit, we were told that Luke had hyperthyroidism which explained his small size. Luke also began to see an Ear, Nose and Throat (ENT) specialist for suspected hearing loss. This specialist felt it was difficult to diagnose this problem at such an early age, so monitored him every few months. By the time Luke was three, he was under the care of a pediatrician, an ENT doctor, and a top plastic surgeon, but no answers were found.

Luke's real test began when he started school at age four. He was always being treated as a baby by children younger than he was. Little girls would

pick him up and give him swings, or play house and use him as the baby. In addition, Luke was really struggling in school. The teachers felt he just wasn't trying but I found this hard to believe.

He would come home from school and complete in an hour the work which had taken him the whole day at school. There were many confrontations between me and the school staff. When I next met with the ENT specialist, I explained Luke's school problems and the fact that at home, the TV and music were always on high. Luke was hospitalized for diagnostic tests, and I learned that he had a missing left ear canal. I was so angry to discover that among everything else that was happening to him, he was treated very unfairly at school. The problem was not a naughty child's destructiveness but a confused and sad little boy who had a hearing problem. Luke was fitted for a hearing aid.

We began working with a hearing specialist, who proved a Godsend. She spoke to the school staff and explained the difficulties of hearing impaired children. I learned about Catig, a school with a hearing impaired unit. This unit had nine children and two teachers. Everything was designed for the needs of hearing impaired children. When the move took place, Luke was happier than he had ever been. He discovered he was not the only child with a hearing aid. Some of his peers were deaf and dumb; some wore two hearing aids. Luke had only one and he could speak. His confidence grew as did his love for his school. For once, it seemed that things were going right for Luke. He began eating better,



Luke, Jacqueline & Sharlene

looked better, and overall was a contented little boy.

Sadly, there was more bad news to come. A blood test showed a continuing drop in platelets. Luke was admitted to the hospital for growth hormone tests, which were followed by a bone marrow test. I was told Luke would be sedated. At that time, I didn't know that Luke could have been put to sleep. I will never forget that day. I was told that Luke would be fine and to go and have a cup of tea. I was halfway down the corridor when I heard Luke screaming. I charged through the doors where nurses and doctors held Luke down in the hope of getting bone marrow. I remember running down the corridor with Luke in my arms. I dressed him and took him home. I told the doctor that the trust that had built up over the years between him and Luke was gone.

The following week, I received a letter saying that Luke could have the bone marrow repeated using a general anesthetic. The results of both the bone marrow and the growth hormone test were fine. I believed that whatever was causing Luke's problems, it wasn't serious.

The blood tests continued and then came the next blow for Luke. He

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Luke's Story

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started having nose bleeds. Over time, these became more frequent and more serious. They never occurred around the doctors so they thought I was exaggerating. Then one day Luke came in from playing in the yard covered in blood. I took him to my nearby clinic. I was told that Luke had hay fever and he was given antihistamine. But the nose bleeds became even more frequent.

When Luke was nine, I was visited by a genetic nurse who explained that a new doctor had a diagnosis for Luke. I heard the diagnosis and remember saying, "Thank God." The word "anemia" brought thoughts of iron medicine and simple treatments. Slowly, I discovered how far I was from the truth.

In December 1999, Luke was placed on oxymetholone, 25 mg. In February, 2000, he was found to have a missing left kidney, but both kidneys are present on the right side. I am now awaiting kidney function test results.

After all is said and done, I am so proud of Luke. He is a kind and loving little boy and never complains. Fanconi anemia has robbed him of many things, but hasn't taken his personality. Luke is doing well on oxymetholone and is pleased that he is going to get bigger.

For Luke

This time, Lord, you gave Luke a mountain. A mountain so high. But we will climb and it isn't a hill any longer. You gave Luke a mountain this time. ♦

Berlin Transplant Renews Hope

by Susan Collins, Delray Beach, FL

Today, September 11, we are 53 days post-transplant and in University Children's Hospital, Charité, Berlin, Germany. I never believed that either of those things was possible. Our son, Christian, who just turned 7, had been transfusion dependent for about three years. In fact, three years ago when his counts really bottomed out and his first clonal abnormality showed up, we were planning to take him to transplant right away. However, after much prayerful consideration, it became painfully obvious to us that Christian could not possibly survive the protocols that were then available.

Christian has multiple birth defects including heart problems, significant endocrine problems, seizure history, chronic respiratory illness, and more. With so many strikes against him already, we simply knew that he could not survive the toxicity of total body irradiation (TBI). We therefore accepted that we would probably lose him either to leukemia or to an infection. But we also kept an open mind, and promised ourselves that we would re-evaluate transplant options if changes were made in the protocols.

Thankfully, we began hearing about the use of fludarabine and then about lower toxicity protocols. After much investigation and again much prayerful consideration, it was very clear that bringing Christian here to Berlin was our only option. We once again felt hope. In addition to the attractiveness of the lower toxicity busulfan and fludarabine protocol, we felt strongly that we did not want a T-cell depleted graft, for fear of lack of engraftment or a late rejection. We also were extremely concerned about germ control. We knew that everything would have to click perfectly for

Christian to have any real chance. The combination of extremely tight germ control and longer inpatient stay added to the appeal of coming here. Finally, we also came to believe that the use of busulfan instead of TBI might help reduce the risk of secondary post-transplant cancers that have plagued other FA patients.

When we arrived here, Christian had approximately 35% blasts and a highly proliferic monosomy 7 clonal abnormality. He had suffered repeated viral infections and was in the hospital at home and here for a total of almost two months before his transplant on July 21. Christian had a 6/6 unrelated bone marrow donor.

And now for a truly miraculous closing. Christian seems to have achieved a stable engraftment with no GVHD. His last platelet transfusion was on Day 21, and this week his platelets have gone over 200K. He has not received red cells since Day 27 and his current hemoglobin is 10.4. His absolute neutrophil count (ANC) has not dipped below 1000 since Day 24. These counts have all occurred without the use of any growth factors. We remain painfully aware of the long and dangerous road ahead. However, whatever the future may hold cannot diminish the magnitude of this miracle. We are most excited to be bringing Christian out of the hospital to our Berlin apartment this week. We will not be returning to the States until after Day 100, near the end of October. ♦



Ailton

Ailton was, and always will be, very special, because he taught me a lot about his faith and this helped me in my search to understand life. For me to understand life is to know that “death” relieves one from a world that is so unjust and disloyal into an eternal life in a calm place where there are no longer fears or threats.

I feel Ailton deserves to be in “Heaven” because he was exceptionally dignified for a person of only twelve years. He faced his reality with the hope of making the best out of life. With the help of God, he achieved his aim because he was sure that God would receive him.

His last words on this earth were, “Mom, don’t worry, everything is going to be all right. I am with Jesus.” These words keep me going, and I hope one day I’ll be with him and God.

Antionietta Medeiros, Mother
Brazil ♦

Danielle DiMarino - A Loving Remembrance

A year has gone by since Danielle passed away, and it is still hard to believe she is gone. Danielle DiMarino was born on May 7, 1987 and passed away on August 24, 1999. She was a sixth grade student at H.C. Crittenden Middle School. Danny, as we called her, was twelve years old. Only twelve years to live an extraordinary life, but live it she did. She was a dark haired, dark eyed baby, attached to her parents and her little white star pillow. From the moment she was born we knew she was special. A curious infant and a talkative toddler, she was always full of questions and wonder.

From the time she was born, she faced numerous adversities, painful medical procedures, and depressing hospital stays. However, had you not known our family you would never have known any of this. She never let on about her illness. That’s the type of person she was. Danny was an amazingly happy person despite the many obstacles she faced each day. Her daily goal was to be just like everybody else, a carefree pre-teen (looking forward to being “13”), worried only about her clothes, boys, and her friends. She loved dancing, music,



singing, and knew all the lyrics to every song. With her striking voice, we often joked she would be a D.J. some day. She truly loved life and lived for each moment. She laughed loudly and loved fiercely.

Danielle faced every situation with a determination of spirit and a smiling countenance. She never gave up and definitely never gave in. Her friendships were built on loyalty and unconditional acceptance. Only now do we realize why Danny was so special. She was placed here to teach us a great lesson...how precious and fragile each and every life is. She was, and always will be, our shining Star.

Never forgotten – always in our hearts,
Mom, Dad and Jen ♦

Paula Ceresa

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p53 gene is abnormal in about half of precancerous mouth growths, and also in about half of all cancers.

For the next 12 weeks, Paula traveled to Chicago to take ONYX-015, which was administered as a mouth-wash. She reported that her mouth felt better than before. In July, 2000, she wrote “the dysplasia on my tongue seems gone and another area on the

roof of my mouth and the gum around one tooth are much better—almost disappeared. It’s only slightly red and hardly tender at all.” A biopsy was performed at 12 weeks, and confirmed improvement in Paula’s precancerous condition. She was therefore admitted to the second 12 week phase of this trial. The trial is now complete, and Paula is awaiting results of a biopsy.

We are all hoping for the best, Paula! ♦



In Loving Memory

Jorge Cabrera

1/27/90 - 9/14/00

Ailton Medeiros Augusto

5/8/87 - 1/19/00

Lynn Welfare Mendenhall

12/23/53 - 7/8/00

Skyleigh Moureau

4/4/80 - 7/11/00

Logan Waldrip

3/23/90 - 4/22/00

Michael Ungerer

8/20/91 - 3/99

Excerpts from Lynn Welfare Mendenhall's Eulogy

by Fori McLean

When I think of Evelyn Welfare Mendenhall, I think of a young lady with an indomitable spirit who never met a stranger. If you know Lynn like I know Lynn, you know what I am talking about.

She was an open book and she expected you to be the same. Each person she met instantly became an extended family member. I think Lynn possessed more tenacity in her pinkie finger than most people have in their entire bodies. She had to!

When Lynn was a toddler, the doctors told her parents that she probably wouldn't live beyond the age of 2. But she surprised everyone. Everyone, except God!! He had marvelous plans for Lynn. Lynn, you brought us joy for over 46 years.

Lynn was small, but only in stature. Her heart was as large as the Grand Canyon and she was truly one of the most altruistic people I have ever known. She was always "loving up" on someone and making him or her feel special. When you were with Lynn, she was extremely kind and caring about your needs. One of Lynn's assets was her ability to draw other people out of their comfort zone and communicate with them from the heart.

Lynn loved people and she loved music. The combination was always a winner. When Lynn worked with elderly folks at retirement homes, she never failed to light up many faces. She would roller skate into the room, come dressed as a clown, or in some other whimsical way she would entertain those whom society had cut off from the mainstream. She would play the piano or guitar and before long, feet were tapping or mouths were lip-



Lynn with Chris Hull

synching the words Lynn was singing.

Lynn knew life is not fair. Yet, she seldom complained. She was too blessed to be depressed for very long. She accepted that life is often bad, but she acknowledged that God's grace is always good.

When you think of Lynn, what comes to mind? Her cancer? Her Fanconi anemia? Yes, those contributed to the demise of her earthly existence, but by no means did they define Lynn. She did not wallow in self-pity. Lynn embraced life every moment!!

Today, I stand before you and ask you to join with me in praising our Heavenly Father for allowing Lynn to be included in His plan. ♦

Fanconi Canada: Energetic and Productive!!

*by Lorne Shelson & Annette Waxberg,
Toronto, Ontario*

In the first nine months of 2000, Fanconi Canada has raised almost \$22,000 Canadian dollars for research, and we're still going strong. The Cronins have been particularly successful in their efforts, having raised almost \$18,000. These funds will allow us to meet the first year of our commitment to co-fund a 3-year post-doctoral fellowship in FA research with the Canadian Institutes of Health Research. The application deadline is November 1st.

We have expanded distribution of our bilingual newsletter beyond our forty families to include supporters and interested doctors across Canada. Fanconi Canada is also sponsoring Dr. Martin Champagne, a transplanter at Hôpital Ste-Justine in Quebec, to attend the FA Scientific Symposium in Amsterdam. Our website is in draft form and should be up by the end of the year.

The 2nd Annual Ontario Region FA Family Picnic was held in July near Toronto. We had a lovely day talking, swimming, picnicking, doing arts & crafts, flying kites and playing Frisbee. The next family get together will be our "Holiday Happening" in December. This gathering will also serve as our Annual Meeting during which we will complete our reorganization by appointing permanent directors and officers. ♦

We Welcome New Families Who Have Joined Our Support Group

John and Audrey Barrows
1608 Victoria St.
Baldwin, NY 11510
516-867-2491
Kelsey ~ DOB: 11/6/91

Catherine Bergin
62 Whipple Street #1
Worcester, MA 01607
508-792-9335
Ronald ~ DOB: 1/15/89

Karen Bowser
3106 253rd Street Ct. E
Spanaway, WA 98387
253-847-8942
Mallory

Tanya Goodrich
10513 8th St. E.
Edgewood, WA 98372
253-942-8786
Cameron ~ DOB: 10/3/95

Randy and Christy Kelley
4365 Milner Road, West
Birmingham, AL 35242
205-980-2535
Hunter ~ DOB: 1/7/95

Bruce and Marilyn Masters
91 Michener Ave.
Mt. Pearl, NF, Canada A1N 4G2
709-368-7607
Jason ~ DOB: 5/5/95

Tony and Lina Nahas
28723 Magnolia Way
Saugus, CA 91350
661-296-3056
Peter ~ DOB: 9/11/90

Lorraine Nazer
14 Linton Close Welling
Kent, England DA16 3EL
011-208-316-6423
Karen Marie ~ DOB: 2/14/85

P. Michael and Kay Proctor
61238 Hwy. 207
Hepner, OR 97836
541-676-9827
Roy Michael Hale ~ DOB:
12/25/87

Ron and Alice Schaefer
7940 Summerplace Dr.
Citrus Heights, CA 95621
916-729-4160
Nikelle ~ DOB: 1/10/71

Maria van der Laan-Goos
Boomberglaan 28
1217 RS Hilversum
The Netherlands
011-31-35-624-6130
Barbara ~ DOB: 3/11/87

Use of Logo

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

Is There a Cancer Fighting Diet?

by Carol Ceresa, Nutritionist

Wouldn't it be wonderful if we could prevent or delay the onset of cancer, especially since children and adults with Fanconi anemia are at increased risk? I believe in the value of overall good nutrition and in carefully evaluating and trying some products and supplements that may be of benefit. To have the best strategies for fighting cancer, we need to consider the basics.

Cancer starts with a single cell that gets out of control. The causes of cancer are not entirely clear. Cancer promoters, called carcinogens, include viruses and chemicals, as well as lifestyle and environmental factors (air pollution, smoking, lack of exercise and poor diet). No single food or nutrient causes or prevents cancer. However, there are steps that will help reduce your risk for cancer.

Eat a plant-based diet.

Vegetables and fruits have a complex composition with more than 100 vitamin, mineral, fiber and other beneficial substances. Phytochemicals, meaning plant chemicals, may offer protection from cancer. Phytochemicals are substances that plants naturally produce to protect themselves against viruses, bacteria, and fungi. Some of these naturally occurring substances are carotenoids, flavonoids, indoles, isoflavones and protease inhibitors. As with vitamins and minerals, different plant foods supply different kinds of protection. How much is enough? At least 5 servings/day of fruits and vegetables. Which ones are best for fighting cancer?

Include antioxidants to “round-up” and destroy the cell-damaging free radicals that are by-products of cells burning oxygen.

Free radicals can damage body cells and tissues, as well as DNA, which is your body's master plan for reproducing cells. The following three antioxidant vitamins and selenium, a mineral, play key roles in neutralizing free radicals:

- Beta-carotene (which turns into Vitamin A in the body). This is supplied by dark green and deep orange fruits and vegetables: carrots, sweet potatoes, pumpkin, winter squash, bok choy, broccoli, spinach, and V-8 or other vegetable juices.
- Vitamin C, contained in oranges, cantaloupe, strawberries, papaya, kiwi, red bell pepper, broccoli, brussels sprouts.
- Vitamin E, found in wheat germ, almonds, walnuts, sunflower seeds and vegetable oils.
- Selenium, present in seafood, lean meats and whole grains.

Eat foods rich in phytochemicals:

- Allyl sulfides: onions, garlic.
- Ellagic acid: strawberries, raspberries, blackberries, cranberries, walnuts, pecans.
- Flavonoids: black or green tea, citrus, red grapes.
- Lycopene: tomatoes and tomato products, red grapefruit.
- Phytoestrogens: soybeans, grains.
- Resveratrol: peanuts.

Be sure that you are building on the foundation of a diet that is as healthful as possible.

Eating a nutritious diet each day can help to maintain or improve your feeling of well being, your strength and energy level as well as shoring up your

body's nutrient stores and defenses. You must have an adequate level of protein (meat, poultry, fish, dried beans, eggs & nuts); foods rich in calcium (milk, yogurt, cheese or calcium enriched soy, wholesome grains and starches [6-11 servings/day]); as well as the fruits & vegetables rich in cancer fighting substances. Although there are very specific recommendations for number of portions depending on age and activity level, each of us should have at least:

- 4 daily servings of whole grain/high fiber bread or cereal.
- 3 daily servings of low-fat milk or other high-calcium foods.
- 2 small daily servings of lean meat, poultry, fish or vegetable protein.
- 1 serving of beans or lentils every other day.
- 30 minutes every day of moderate physical activity.

Consider vitamin and mineral supplements and other supplements.

Talk with your health care professional before taking vitamin and mineral supplements or any other alternative therapy. This will help to ensure that you take safe and appropriate amounts of vitamins and minerals. A good rule is: food first, vitamins and minerals in appropriate amounts (100% of the recommended daily allowances) plus recommended levels of antioxidants for FA; and cautious use/trial of other supplements that have been discussed and approved by your physician/health care provider.

Your diet is an important part of your defense against cancer. Eating the right kinds of food, every day, can help you feel better and stay stronger. ♦

Family Meeting at Lake Geneva



Thanks for the great, profoundly life-changing weekend. I'll need a long time to process all this intellectually and emotionally, but I know now how grateful I am to FARE.

The volunteers were great. It was especially wonderful not having the anxiety that if an adult session went over time, we had to miss something to get the kids.

Great job on the conference! The speakers were wonderful! I know this was a small family turnout, but maybe if people can't make it because of financial reasons, there could be some assistance. Maybe we could get miles donated. I would hate to have families miss out on this wonderful experience.

Although we were unable to stay for more than the weekend, we thought the scientific content was one of the best ever.



The family camp already has given direct positive effects for my son, Johan, which we would not have achieved without your help and initiatives. We will always remember how the family camp this year really helped to give our family new hope. I am also now in close contact with Ralf Dietrich in Germany and I have established e-mail contacts with several European families.

It will always be an expense, but the idea of meeting and talking with the doctors who are trying to save or someday transplant your child is priceless!



This has really been an eye-opener for me, and has given me energy so that I can develop a good treatment strategy for my son.

Thanks so much for organizing this great weekend and for being there for us and other FA families!

FUNDRAISING

Call to Fundraise for FA

by Lorraine McQueen, Richmond, VA

I am writing to urge each and every FA family to raise funds for the FA Research Fund. Our son, Sean, was diagnosed with FA on October 18, 1999 (you never do forget the “diagnose day,” do you?). Like most families, our first reaction to the news was disbelief. As much as my husband and I tried to convince ourselves that it just couldn't be, we both knew it was true. Finally, all of the pieces to Sean's unique medical puzzle fit.

At first we just cried a lot. We were completely at a loss as to how we should live our lives and felt immobilized by the diagnosis. We were forever coming up with lists of questions that no one could answer. Our doctors told us that because Sean is mosaic, he “should be fine” so we should “just wait and see how he does.”

I remember hearing the doctor telling us to “wait and see” and thinking, “If you knew a train was going to come speeding along and hit you, would you just “wait and see” what happened when it did? We knew it was time for action!

There are so many aspects of this disease that are beyond our control. However, we, as parents of these fantastic FA kids, are not powerless! One important way that each and every one of us can engage in battle against this disease is to raise funds for the FA Research Fund. FAREF is our best hope of finding a cure.

Our first fundraising effort was a letter writing campaign. At first we felt a bit uncomfortable about the idea of writing a letter to all of our family and friends to ask them for money. We knew, though, that letting our pride

get in the way of making every effort for our son would be a mistake.

The process was really quite easy. We simply drafted a letter describing Sean's diagnosis and asking people to make a donation to the Fund. We then e-mailed the letter to Leslie Roy with a list of addresses, and the Fund handled the rest. The response that we got was fantastic! Our friends were so grateful to have some information about this mysterious disease and to have a concrete way to help. It was amazing to us how much people really do want to help if you just show them how.

We then decided to move on to our next project, which is a Casino Night and Silent Auction to be held in Richmond, Virginia on September 30, 2000. We started by sending out letters to local companies and stores, soliciting for financial sponsorship of the

Lynn Mendenhall's CD A Lasting Gift to our Fund

Before her death, Lynn Welfare Mendenhall was determined to complete work on her CD, one of her final gifts to our research fund. The CD contains sixteen pieces, thirteen vocals and three instrumentals. Lynn plays the piano or guitar for each vocal selection. Knowing Lynn's immense musical talents, this CD will be a treasured, lasting gift to all of us.

Each CD costs \$16.00 if mailed inside the USA or \$17.80 if mailed outside the USA. Net proceeds will be donated to FAREF. Checks should be payable to SpachWorks and mailed to



Lorraine & Kevin McQueen

“Play for FA” event and for items for the silent auction. We were again amazed to see how eager so many were to help. We were smart enough to accept the offer of friends to help us in our efforts and formed a “Friends for Sean” committee. Thus far, we have gathered over 200 auction items and 15 corporate sponsors. This project has certainly kept us busy these past few months, but it is going to be great!

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Lynn's brother or to her mother at:

John S. Welfare
7201 Folger Dr.
Charlotte, NC 28270

Susan Welfare
4214 Cheltenham Rd
Charlotte, NC 28211

Inquiries can be sent by e-mail to John at aguabonita@aol.com or to Susan at SWell04725@aol.com. The family plans to fill all orders before Christmas. This would be a wonderful holiday gift to friends and loved ones. ♦

Family Fundraising Efforts

by Leslie Roy

This year we sent out a questionnaire to families asking for their input regarding several items affecting FARF. One of the questions we asked was, "Have you fundraised for research in the past or are you willing to fundraise now?" The responses were varied and insightful. Here are a few of the responses:

"I am a single parent of two children with little time for anything else. I am, however, trying to make time to fundraise as I believe it is an excellent cause."

Every FA family faces difficult challenges each and every day. Nonetheless, we hope you all agree that FARF is an excellent cause for our limited energies. Research supported by FARF is our best hope for finding a cure for this illness.

"I haven't had the time nor do I know what to do. I'm overwhelmed with the effort it would take."

Most families today are stretched far beyond their capacity. If a family is willing to send a fundraising letter, our office will help you write the letter, address envelopes with your donor addresses, stamp and mail them. We also have an excellent fundraising book written by an FA parent that we would gladly mail out to anyone considering fundraising.

"Yes, we send a letter to church family and friends and our own family every year."

"Yes, we have done fundraisers and will continue to do so until we find a cure."

"Yes, but I haven't done it more often because of time restraints and finding an "audience" outside of those who've contributed in the past."

We appreciate each and every effort that families make toward raising funds. Remember that your best

audience is, in fact, those who have contributed in the past. They want to know what progress has resulted from their contributions, and what is now needed to cure you or your child. Past donors become invested in our cause. We need to ask for their help on a yearly basis, thank them for past support, and tell them why we continue to need their help.

"Research is making painfully slow progress, but must continue. The more research, the better the chance it will help our children now and in the future."

Research does seem painfully slow in providing answers, but FA is a rare and complicated disease. Many rare diseases have no researchers working on their behalf. We are fortunate to have a growing group of dedicated scientists working diligently year after year for answers that will save FA children and adults.

Researchers continue to submit grant proposals for funding, and our family support group continues to grow. We would never want to turn down a worthy grant proposal due to lack of funds, nor would we want to limit the number of families that we can serve. To this end I encourage everyone to consider what part you can play in helping FARF reach our goals.

Thanks to all who made a special effort to raise funds during the first six months of 2000. We report below on funds received from January 1 through June 30. We raised \$558,718, which includes \$63,035 in memorial contributions and \$9,636 from United Way/Combined Federal Campaign contributions.

We are not reporting on amounts raised in excess of \$20,000. Large sums raised by a few families can make some feel that their own efforts are

meaningless. **Yet every single effort, large or small, will help us reach our goal of curing this disorder.** Do whatever you can, and we will be empowered to do more.

Funds raised during the first six months were attributed to the following families who either did a fundraiser, directed memorial funds to FARF or gave a personal donation.

\$20,000 and up

Dave & Lynn Frohnmayer
Deane Marchbein & Stuart Cohen

\$10,000 - 19,999

Chuck & Katy Hull
Mike & Beth Vangel

\$5,000 - \$9,999

Susan & Chris Collins
Laurie Strongin & Allen Goldberg
Bill & Connie Schenone
Lorraine & Kevin McQueen

\$1,000 - \$4999

Vicki & Andrew Athens
Mark & Linda Baumiller
Darryl Blecher & Diana Fitch
Randy & Nancy Bloxom
Ray & Diane Cronin
Joseph & Tracy DeMarco
Antonino & Marie DiMercurio
Ed & Janice Duffy
Pat & Maria Gleason
Susan Jackson
Beth & Jeff Janock
Jack & Lisa Nash
Jan & Leonard Riley
Matt & Diane Senatore
Steve & Melissa Turner

Up to \$1000

Eddie & Sandy Allen
Serge & Brenda Arsenault
Ken & Jeanne Atkinson
Lynn Baervoets

Keith & Lily Baggett
 Roger & Sarah Baker
 Donna Barnes
 Gilbert Bodier
 David & Carole Boudreau
 Richard Briga
 Tad & DeeDee Burzynski
 Lynnette Chandler
 Brad & LeaAnn Curry
 Bill & Pat Danks
 Day Family
 James & Carol Dillon
 Pat & Mary DiMarino
 Paige Ellis Family
 Fred Engel
 Ezat & Laila Faizyar
 Nancy Fena
 Susan Gannon
 Dave & Paula Guidara
 Mitchell & Tirzah Haik
 Frank & Kelly Hamilton
 Jeff & Judy Hoffman
 Brian Horrigan & Amy Levine
 Leardon Keleher
 John & Karilyn Kelson
 Alfredo Leontic
 Peg LeRoux
 Eric & Beth Losekamp
 Lynnette Lowrimore
 Steve & Allison McClay
 Cecelia Meloling
 Griff & Cecelia Morgan
 Shiela Muhlen
 Lisa & Kenny Myhan
 Bob & Alice Nicholson
 Ron & Fredi Norris
 George & Kathryn Reardon
 Lynn & Rick Sablosky
 Bob & Andrea Sacks
 Erik & Lori Salo
 Martin & Linda Sankey
 Ron & Alice Schaefer
 Bryan & Karen Siebenthal
 Mark & Susan Trager
 Marc & Sandy Weiner
 Susan Welfare
 Nancy & Reese Williams
 Elizabeth & Gerald Wisz
 Marge Zaborney ◆

A Novel Fundraiser

Mike Vangel's father was pulled over for speeding the other day. The police officer said he wouldn't write him up a ticket (which would have been for over \$100) if he promised to make a donation to his favorite charity. So he sent our research fund \$150.00!! As Beth Vangel commented, "Hey, we'll take the \$\$\$ anyway we can get it!" ◆

Family Meeting

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want to learn and share should be able to attend this camp!

Through the advocacy of Vicki Athens, a group of nineteen young volunteers, associated with the Detroit Rotary Club, served as camp volunteers, along with returning volunteers. These energetic youngsters, led by the empathetic and hilarious Rob Sawyer, greatly enriched the program for all of our children. A dance performance by young teens was a highlight of the meeting. One youngster even ventured to Six Flags, a nearby entertainment park. Taking your evaluations into account, plans are already underway to make next year's camp even more entertaining and meaningful for all youngsters in attendance.

Evening bonfires and volleyball matches, Karaoke night and dancing, and parasailing over Lake Geneva showed the capacity of many to have fun in spite of sadness and stress. Medical and scientific presentations occurred over the weekend, for families unable to extend their stay into a work week. Readers are strongly encouraged to read the Medical News section of this newsletter, plus the *Science Letter*, for detailed information on all medical and scientific presentations. ◆

A Time of Heartache and Hope

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days, but enjoy the good ones.

Murad will need a kidney transplant and a bone marrow transplant. I pray that all FA patients have the courage to live each day to the fullest. We love Murad so much.

Annie is 13 this year and is okay. She is doing well in school. She loves her little brother a lot, and helps with his bathing and changing. We miss our other babies a lot. We have lost a son, Zain, and a daughter, Mariam, one two-days old, the other at two months. I have also had four miscarriages. But some things were not meant to be.

I now have a hernia and gallstones. A week after moving into our new home, we were robbed. A lot of bad things have happened all at once. I hope good times are coming. We live in hope and have to take each day as it comes. Today the sun is shining and we are going to have fun in the park.

God bless you for helping us.

The Shaikh Family ◆

Call to Fundraise for FA

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After we sleep a lot in October, we will gear up for a "Play for FA" golf tournament this spring. The opportunities really are endless!

Kevin and I are certainly not fundraising experts. There is no magic in what we are doing. We are just parents of a very special little boy whom we love very much. We know that it is up to us to save him and we have no time to waste. The clock is ticking!

I know life is hectic and we all have much too much to do every day. Please consider putting fundraising for FARF at the top of your "To Do" list. These children are too precious! ◆

FA Malignant Tumor Cells and Leukemia Cells Needed for Research

Dr. Bagby's laboratory in Portland is beginning studies on cancer and leukemia cells from children and young adults with Fanconi anemia. His team believes that the recent identification of new functions of the Fanconi anemia proteins may lead to a new molecular understanding of the processes that cause cancer and leukemia. Fresh, frozen, or paraffin embedded samples of tumors and/or bone marrows may be sent directly to Dr. Bagby at the address below. Please have your physician or the pathology department notify Dr. Bagby at least one day in advance. Material should be sent by overnight mail to:

Grover C. Bagby, MD
Director, Oregon Cancer Center
OHSU CR145
Attention: Winifred Keeble
3181 SW Sam Jackson Park Road
Portland, Oregon 97201
e-mail: grover@ohsu.edu

Editors' Note: If you are planning surgery for the removal of potentially cancerous or cancerous cells, please don't let the laboratory discard tissue after their analysis. These samples can be of enormous value to scientists who wish to understand and eventually treat these complications of FA. In advance, we thank you for helping to push scientific discovery ahead!



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Conference Fee and Transportation Costs Tax Deductible as Medical Expense

by Darryl Blecher, Pittsburgh, PA

A parent attended a medical conference in another city concerning a chronic disease suffered by his child, in an effort to learn more about the disease and methods to treat it. The child's doctor had recommended the conference, which was sponsored by an organization that supports research and education about the disease.

The IRS, under Section 213, ruled that the trip was primarily for, and essential to, the child's medical care. Therefore, the parents could deduct transportation costs and the conference registration fee. Parents could not

deduct meals and lodging costs because neither the parent nor the child was receiving medical care from a physician at a hospital away from home.

Given the above example, transportation expenses to the Family Meeting or to a Scientific Symposium should be considered legitimate deductible medical expenses. Check with the IRS or your own tax preparer as well. Such a deduction is subject to the limitation that only those medical expenses in excess of 7% of one's adjusted gross income are deductible. ♦