



FAMILY NEWSLETTER

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



Researchers, treating physicians, and FA parents attended the FA Scientific Symposium in October.

HIGHLIGHTS

| | |
|--|----|
| Discoveries Reported | 2 |
| Gene Therapy Trial to Begin | 2 |
| Bone Marrow Transplant Conference Planned | 2 |
| Comparison Between Complementation Group and Mutations, and Clinical Outcomes | 3 |
| Fludarabine-Based Regimen for Alternate Donor Hematopoietic Cell Transplantation.... | 4 |
| New Promising Vectors for Gene Therapy | 4 |
| Preimplantation Genetic Diagnosis Articles | 5 |
| Medical Advice From our E-Mail Group..... | 8 |
| Family News..... | 9 |
| Fundraising | 16 |

Fanconi Anemia Scientific Symposium

One hundred fifty-six researchers, treating physicians, and fifteen FA parents from fourteen countries met in Amsterdam for the Twelfth Annual International FA Scientific Symposium, October 26-29, 2000. Countries represented were Tunisia, France, England, Canada, Spain, Italy, Germany, Argentina, Israel, Japan, South Africa, Russia, The Netherlands and the United States. Fifty-one scientists and treating physicians gave formal presentations. Evaluations from attendees confirmed, once again, that our annual scientific meeting is an outstanding investment of our precious research dollars.

Research topics covered six areas: Gene Discovery and Regulation; Cancer and Leukemia; FA Protein Function and Hematopoiesis; Mosaicism,

continued on page 15

Plan to Attend August Family Meeting!

Our 10th annual FA Family Meeting is seven months away, but now is the time to start planning. This year, we will offer a limited number of scholarships to help families defray travel and lodging expenses (*see article, p. 11*).

From August 10-14, 2001, FA families, treating physicians, and researchers will meet at the picturesque lakefront setting of Aurora University's George Williams Lake Geneva campus in Williams Bay, Wisconsin. We will learn from our experts, meet and share experiences with other FA families, and relax. Lake Geneva is an easy, two-hour drive from Chicago's O'Hare airport, or a forty-five minute drive from the airports in Milwaukee or Madison, Wisconsin.

We will have two days of science and medical presentations on the

continued on page 14

Discoveries Reported

Two articles concerning FA-related discoveries were published on February 23 in the journal, *Molecular Cell*. "Positional Cloning of a Novel Fanconi Anemia Gene, *FANCD2*" by Markus Grompe, Robb Moses, Alan D'Andrea and collaborators describes the identification of an FA gene which plays a crucial role in the FA gene pathway. *FANCD2* is also the first FA gene found to be conserved in lower animal species and plants. The discovery of *FANCD2* now means that there are at least eight complementation groups.

"Interaction of the Fanconi Anemia Proteins and BRCA1 in a Common Pathway" by Alan D'Andrea, Markus Grompe and collaborators, states that the FANCA, FANCC, FANCE, and FANCG proteins form a complex in the nucleus of the cell. This complex is required for the activation of the FANCD2 protein.

In normal, non-FA cells, the FANCD2 protein plays an important role in DNA repair. In normal cells damaged by X-rays or other agents, the FANCD2 protein is found in distinct spots (or foci) in the nucleus. These foci are not found in cells from FA patients regardless of the complementation group. Interestingly, the breast cancer protein 1 (BRCA1) is located with the FANCD2 protein in these same foci after the same kinds of DNA damage. The FANCD2 protein therefore provides the missing link between the FA protein complex and the BRCA1 repair machinery. Disruption of this pathway results in the cellular and clinical phenotype common to all FA complementation groups. ♦

Gene Therapy Trial to Begin

A clinical gene therapy trial for Fanconi anemia will open shortly at Indiana University in conjunction with the recently formed International Collaborative Fanconi Anemia Group. The institutions currently involved include Indiana University, the Fred Hutchinson Cancer Research Center, and St. Jude Research Hospital, as well as representatives from Brazil and Germany. The FDA has approved the protocol, and final modifications are now being submitted.

This is a pilot study to evaluate the safety and feasibility of performing gene therapy on patients with Fanconi anemia. A retroviral vector for clinical use with the FA complementation group C gene has already been produced at the National Gene Vector Laboratory located at Indiana University. A retroviral vector for the FA complementation

group A is currently being evaluated for clinical use.

The protocol will use retroviral gene transfer to place a normal FA gene into blood-producing cells obtained from a patient's bone marrow. Peripheral blood and stored umbilical cord blood collections may also be used as the source of blood-producing cells. A retroviral marker vector will be transferred to a small portion of the blood-producing cells. Participants in the clinical trial will be monitored closely for the transfer of the FA gene into their blood cells. The fate of the cells corrected with the normal FA gene and the marker vector will be compared to determine the efficacy of gene correction. The researchers hope that this approach may eventually correct or prevent the bone marrow failure of Fanconi anemia. ♦

Bone Marrow Transplant Conference Planned

FA bone marrow transplant experts from around the world will gather in Chicago on April 28, 2001, for a one-day conference. Grover Bagby, Oregon Health Sciences University and John Wagner, University of Minnesota, have graciously donated their time to organize this meeting. The conference will include approximately twenty-four treating physicians who specialize in transplantation and its complications. The small number of attendees and targeted focus of the meeting should assure that this crucial topic is addressed comprehensively, with ample time for discussion. To encourage open dialogue and a thorough exchange of ideas, this meeting will

not be open to observers.

Short presentations will be followed by panel discussions. Participants will cover the following topics: matched sibling donor transplants; unrelated and related mismatched transplants; sources of stem cells (bone marrow, cord blood and peripheral blood stem cells); and pre- and post-transplant complications. The FA Research Fund is sponsoring this conference. We trust that the sharing of information and opportunity for informal discussion and debate will ultimately produce better transplant outcomes. Our sincere thanks to donor John Holmes of Ice Bear, Inc., for making this workshop possible. ♦

Comparison Between Complementation Group and Mutations, and Clinical Outcomes

Christopher Mathew, PhD, Guy's Hospital, London, and collaborators published an article in *Blood*, December 15, 2000, on the association between complementation group and mutation type and the clinical outcome in Fanconi anemia. The authors studied 245 patients from all known complementation groups. Disease mutations were identified in 169 patients. The authors noted that FA-G patients had severe bone marrow failure and a higher incidence of leukemia. Birth defects or anomalies

were more common in the rare groups FA-D, FA-E, and FA-F. In FA-A, patients who inherited mutations from both parents which knocked out protein production from the *FANCA* gene had an earlier onset of anemia and a higher incidence of leukemia than those with mutations producing an altered protein. In FA-C, there was a later age of onset of aplastic anemia and fewer birth defects in patients with the 322delG mutation; there were more anomalies in patients with the IVS-4 mutation. ♦

Four Patients Transplanted with Related, Mismatched Donors Do Well

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

All four patients engrafted early (between days nine and eleven post-transplant). Immune reconstitution occurred at 8 months for the first two and 12 months for the third. It is too soon to evaluate the fourth patient. All four are alive and well; the first three patients are alive almost three years, two years and one year post-transplant.

No patient has yet experienced acute or chronic graft-versus-host disease. The third patient had post-transplant complications (CMV infection and EBV lymphoma) which have been resolved. The number transplanted is very small and the time, post-transplant, for the fourth patient is inadequate for a full evaluation. Nevertheless, Boulad is optimistic about these very promising results. ♦

Chapel Hill, NC Will Perform Complementation Group Testing on FA Families

by *Chris Walsh, MD*

We will perform complementation group testing on peripheral blood and bone marrow aspirate samples. Patients, through their own physicians, can send bone marrow aspirate and blood samples to my lab (see address below). Samples can be sent at room temperature and should contain anticoagulant such as EDTA for overnight shipment. Notification of the lab 1-2 days before shipment would be appreciated. There is no cost to families.

Helmut Hanenberg, MD, Will Do Complementation Analysis for FA Families

Helmut Hanenberg will do complementation analysis for all known complementation groups. Peripheral blood is the easiest and quickest material to analyze, although he can use any cell type that can be grown in culture. Hanenberg recommends sending 8 - 12 mls of blood because of the long distance from the United States to Germany. There is a likelihood of greater than 95 percent of finding the defective gene using his method. At the present time, there is no cost for this service. For information on shipping and handling, contact:

Dr. Helmut Hanenberg
Department of Pediatric
Hematology & Oncology,
Children's Hospital,
Heinrich Heine University
Moorenstr. 5
40225 Duesseldorf, Germany
e-mail: Helmut.Hanenberg@uni-
duesseldorf.de
phone: 011 49 211 811-6103
fax: 011 49 211 811-6436

In addition, if patients are undergoing tumor biopsy or surgical excision, those samples can also be sent to the following address:

Dr. Chris Walsh
Rm. 7101 Thurston Bldg.
CB#7352
Chapel Hill, NC 27599
phone: 919-966-9116
fax: 919-966-0907
e-mail: cwals@med.unc.edu

Fludarabine-Based Preparative Regimen for Fanconi Patients Undergoing Alternate Donor Hematopoietic Cell Transplantation

Margaret L. MacMillan, MD and John E. Wagner, MD, University of Minnesota

Until recently, graft failure was the major obstacle to successful bone marrow transplantation in patients with Fanconi anemia using an alternate donor (i.e., a donor other than a matched sibling). The risk of graft failure was particularly high in patients with somatic lymphocytic mosaicism (the presence of >10% lymphocytes insensitive to DEB). We thought that the high rate of graft failure might be partly due to insufficient suppression of the immune system. Therefore, we developed a new preparative regimen by adding fludarabine to the commonly used preparative regimen of cyclophosphamide, total body irradi-

ation and anti-thymocyte globulin.

To date, fourteen patients enrolled on this new protocol at the University of Minnesota can be evaluated. All 13 who survived at least one month post-transplant successfully engrafted. Seven of the fourteen patients are alive and free of disease. Opportunistic infections remain a major complication of bone marrow transplantation. We routinely screen all FA patients before transplantation to identify and treat any hidden infections. In addition to a high-resolution chest CT and sinus x-ray, all patients are seen by our Infectious Disease service before transplantation. Despite aggressive man-

agement of any suspected or proven infection, patients with infections prior to transplantation do not do well after transplantation. Therefore, it is imperative that patients be considered for alternate donor bone marrow transplantation as soon as they have met one of the following three criteria:

1. Aplastic anemia as defined as having at least one of the following: platelet count < 20 x 10⁹/L; ANC < 5 x 10⁸/L; hemoglobin < 8 g/dL.
2. Myelodysplastic syndrome (MDS) with or without chromosomal anomalies.
3. Hematologic malignancy such as acute myeloid leukemia (AML).

It takes an average of 3 months to identify a suitable unrelated bone marrow donor for a patient. Therefore, it is best if Fanconi anemia patients are seen in consultation early on, preferably just as the blood counts begin to decline. This timely consult and initiation of a donor search will enable us to arrange for a transplant as soon as it is necessary, with the goal of decreasing the risk of life-threatening opportunistic infections.

At the University of Minnesota we are also very interested in identifying the long-term post-transplant issues of FA patients. Two years ago we conducted a pilot study of late effects after transplantation and quality of life of patients with Fanconi anemia. With the help of the Fanconi Anemia Research Fund, we will be conducting a second survey this spring. We hope that many patients and families will participate so that we can learn how we may optimize the health and quality of life of our patients and their families. ♦

New Promising Vectors for Gene Therapy

As the functions of the proteins encoded by the Fanconi genes are being identified, scientists remain increasingly optimistic that gene therapy of bone marrow stem cells might be an effective treatment for bone marrow failure in children and adults with Fanconi anemia. While this kind of therapy will not place the normal gene in all cells of the body, Fanconi researchers have high hopes that the bone marrow abnormalities might resolve.

One of the technical problems with gene therapy for bone marrow stem cells is that the true stem cells are rare and most of them are not actively dividing. Unfortunately, many of the first generation retroviral vectors (the packages that carry genes into the cells) aren't able to get the gene into any cell unless it undergoes cell division. Now, new vectors are being tested that are capable of putting the gene into cells that are not dividing. Investigators

working with these vectors are seeking to develop evidence that stem cells from patients with Fanconi anemia, particularly those with the A and C complementation groups, can be successfully treated.

The researchers developing these vectors are seeking to obtain samples of bone marrow cells from children and adults with Fanconi anemia type A and type C. Patients and parents who would like additional specific information on these studies and information on how samples should be prepared for shipping to these laboratories should call or write to:

Dr. Grover Bagby
Director OHSU Oregon Cancer
Center
CR145
3181 SW Sam Jackson Park Road
Portland, OR 97201
phone: 503-494-0524
FAX: 503-494-7086

Preimplantation Genetic Diagnosis

by John Wagner, MD, Pediatric Bone Marrow Transplant Program, University of Minnesota

Preimplantation Genetic Diagnosis (PGD) allows carriers of a genetic disease to know the health and HLA status of an embryo prior to achieving a pregnancy. Couples who desire additional children and can afford the time and expense of such a procedure might consider PGD.

A family must first be tested for complementation group assignment and specific disease mutations. The next step is to obtain HLA typing on the mother, father, and child affected with Fanconi anemia.

The most important step is identifying a good *in vitro* fertilization (IVF) team. IVF does not have to be performed at the location of the PGD team, although that is possible. IVF

first requires daily injections to hyperstimulate the ovaries. The eggs are harvested approximately 14 days after initiating the injections. Each of the mother's eggs is fertilized with the father's sperm. After two to three days, the cells will have divided about three times. At the 8 cell stage, a single cell is removed and tested by the PGD team. The cell from a well-growing embryo is then tested for Fanconi anemia and HLA identity. Embryos that are HLA matched to the FA patient and free of disease may be used for implantation. Embryos that are free of disease but not HLA matched can also be implanted or frozen for later implantation, depending upon the desire of the family. Various options

are possible and should be discussed.

Within four weeks after implantation, a pregnancy test will be performed. If the mother is pregnant, chorionic villus sampling (CVS) or amniocentesis will be scheduled to confirm that the fetus is healthy. In addition, HLA typing will be performed. If both characteristics are confirmed, an arrangement will be made for the collection and shipment of the umbilical cord blood to the transplant center at the time of birth. A collection kit and instruction manual will be shipped to the family and/or obstetrician directly. On arrival, the cord blood will be tested for stem cell number, infectious disease contamination (from the delivery process) and HLA (a third time). Prior to transplantation the newborn donor will be evaluated for FA once again (a third time). These tests are repeated because of their critical importance.

Procedural Steps

- 1) Conference with transplant physician and genetic counselor
- 2) HLA type mother, father and child affected with Fanconi anemia; confirm absence of healthy HLA-matched sibling donor
- 3) Obtain complementation group assignment and mutation analysis
- 4) Referral to PGD center
- 5) Referral to IVF center
- 6) CVS/amniocentesis to confirm health and HLA status of fetus
- 7) Cord blood collection and harvesting
- 8) Confirmatory testing on cord blood/newborn baby
- 9) Transplantation of HLA-matched umbilical cord blood ♦

Molecular Testing for Pre-implantation Genetic Diagnosis at The Rockefeller University

by Arleen Auerbach, PhD

For pre-implantation diagnosis with *in vitro* fertilization (PGD/IVF) for Fanconi anemia, it is necessary to use molecular testing for FA detection. Whereas prenatal DEB testing is sensitive for detection of any FA complementation group, to do PGD it is necessary to know which of the FA genes is defective in the family. This is because it is necessary to compare the defective gene in the affected child to the same gene in the embryo, to determine whether the embryo is also affected with FA. Once the complementation group is determined, detection of at least one of the mutations in the gene that is defective in the patient is helpful for PGD. Polymorphic markers in the defective gene can also be used to aid in diagnosis by PGD.

Our Laboratory of Human Genetics and Hematology at The Rockefeller University is able to help FA families with complementation testing and mutation analysis who are registered in the International Fanconi Anemia Registry (IFAR) and are participating in our research program. The informed consent form approved by our Institutional Review Board for the IFAR study allows us to give the results of complementation studies and mutation testing to families who want the information, if they have indicated this on the signed form. Our lab has federal and state approval for clinical diagnosis and DNA-based diagnosis for Fanconi anemia. We charge \$650 for DEB testing to confirm the diagnosis of Fanconi anemia, if it hasn't already been done by our laboratory. There are no other charges to FA families for complementation or mutation analysis. E-mail contact: auerbac@mail.rockefeller.edu ♦

New Molecular Diagnostic Services Offered to FA-A Families Considering Prenatal Genetic Diagnosis (PGD)

To date, families pursuing prenatal genetic diagnosis (PGD) have been in complementation group C, which accounts for 15% of all FA families. This technology has not yet been an option for patients in FA-A, which accounts for 60-65% of FA patients.

For PGD to work, scientists must examine one cell in each pre-embryo to see if it is a normal or FA affected cell. This is relatively easy to do with FA-C. The *FANCC* gene is of average size. In addition, there are six common mutations in the *FANCC* gene which cause FA. Scientists can screen rapidly for the presence of these six mutations. Almost all FA-C families have these common mutations.

In contrast, the *FANCA* gene is very large. Most of the disease mutations in this gene are private, or specific to only one family. There is no rapid, accurate test to locate a family's specific mutations. Therefore, the mutations in *FANCA* are not amenable to the same type of analysis as are the mutations in *FANCC*.

Sherri J. Bale, PhD, Clinical Director of Gene Dx, Inc., states that it is possible to use genetic markers to determine if one cell is healthy or not. *FANCA* is on chromosome 16. Each FA carrier has two copies of chromosome 16, one normal and the other carrying a disease mutation. Using genetic markers, it is possible to determine which chromosome 16 from each parent was inherited by the FA patient. This approach is often called "linkage analysis" because scientists are determining which genetic marker is "linked" to the mutation in the gene, without actually identifying the mutation itself.

The procedure is very simple. DNA from both parents, the FA patient, and any healthy siblings is "typed" at several

genetic markers. This information is then used by the *in vitro* laboratory to test fertilized embryos prior to implantation. In many cases it will be possible to determine with a very high degree of accuracy whether or not an embryo has inherited the chromosomes carrying the disease mutations from each parent.

GeneDx, Inc. of Rockville, MD, is now providing linkage analysis to FA-A families in which there is at least one living patient with the disease, and both parents are available for genetic studies. It is also helpful if there are unaffected siblings of the FA patient who can be studied, as this can increase

the accuracy of the analysis. The analysis is done in the laboratory on DNA obtained from a simple cheek swab, which can be collected at home using materials provided by GeneDx, Inc. At this time, the cost to perform this analysis on parents, the FA patient, and any unaffected siblings is \$1,500.

GeneDx, Inc. is a full-service genetic testing and diagnosis company dedicated to serving the diagnostic and genetic counseling needs of individuals and families with rare hereditary disorders. For further information contact Sherri Bale, PhD, FACMG, Clinical Director, GeneDx, Inc. at 240-453-6285. ♦

Preimplantation Genetic Diagnosis for Fanconi Anemia at the Reproductive Genetics Institute

The Reproductive Genetics Institute (RGI) in Chicago, Illinois has had extensive experience in preimplantation genetic diagnosis (PGD). They have worked with over 1,000 couples. Over 2,000 *in vitro* fertilization (IVF) cycles have resulted in more than 200 healthy children. PGD with IVF and embryo transfer is done as part of an experimental study.

Over the past two years, the RGI has included HLA diagnosis of embryos for the purpose of cord blood stem cell or bone marrow transplantation. The Nash family was their first successful attempt using PGD which resulted in a transplant. RGI will continue to offer its services to other FA families.

In the Nash family, the specific mutation in *FANCC* was known. The same techniques can be used with embryos to detect a specific known mutation in *FANCA*. When the mutation is not known, it is often possible

to use linked genetic markers and family studies (linkage analysis) to predict if an embryo has inherited both the mother's and the father's mutations.

RGI is willing to work with families in complementation groups A and C. Another laboratory must first assign a family to a complementation group. Parents, the FA patient, and unaffected siblings need to be HLA-tissue typed. It is very useful to the Reproductive Genetics Institute if another laboratory has determined a patient's specific FA mutations.

In some instances when the specific mutations cannot be determined, the RGI may develop linkage analysis for a family. This analysis would take 8 to 10 weeks. The cost of developing a system to diagnose both FA and HLA in embryos is \$4,000.

In addition, the cost is \$5000 per *in vitro* fertilization cycle (\$2,500 to determine if the embryos are FA affected

continued on page 19

Present Odds for PGD Not Encouraging

We are now aware of four FA-C families who have attempted PGD a total of seventeen times. One family experienced nine failed attempts. The Nash family has had the only success thus far. As laboratories gain experience, we trust that these numbers will improve. ♦

Molly Nash Update

Molly Nash, the first FA patient to receive a cord blood transplant following successful preimplantation genetic diagnosis (PGD), continues to do well, apart from digestive tract complications. Molly's transplant occurred on October 26, 2000. Her blood counts are now completely normal.

Molly has had a feeding tube since she was six months old. In addition, she has complications resulting from her transplant. Chemotherapy and radiation were very hard on her digestive tract, and she also developed a post-transplant adenoviral infection. She has had diarrhea, vomiting and pain on a fairly regular basis since her transplant. Her infection has cleared, but the digestive tract problems remain. Biopsies do not show graft-versus-host disease. Tube feedings and TPN (IV feedings) provide her nutritional needs, but they also suppress her appetite. The Nashes have learned patience and perseverance from this whole ordeal and know that Molly's digestive problems will take time to resolve.

The Nashes plan to do bone marrow drives once Molly is out of protective isolation. They hope to raise public awareness for FA. Their story appeared in *McCalls Magazine*, *People Magazine*, and in the *Ladies' Home Journal* in February. They will be featured in *Parenting Magazine* in June or July. They were on the television

Laurie Strongin Writes of Unsuccessful PGD Attempts

Laurie Strongin has written a very moving article about her family's attempts to achieve a pregnancy through prenatal genetic diagnosis (PGD). Laurie and her husband, Allan Goldberg, attempted this procedure nine times, but were not successful. With considerable detail, Laurie poignantly chronicles the emotional and physical toll of their many attempts. Because of the length of the article, it will not be published here. However, anyone wishing to read this account can obtain a copy of Laurie's article through the FA Research Fund office. ♦

D'Andrea Wins Pediatrics Honor

Alan D'Andrea, MD, Harvard Medical School scientist and FA researcher, has been awarded the 2001 E. Mead Johnson Award. This coveted honor, given over the past 60 years, constitutes the highest research prize in pediatrics. We congratulate D'Andrea on this magnificent achievement! Alan writes: "One very positive side of this award—I will be able to present a high profile lecture on Fanconi anemia at the annual Pediatrics Meeting in Baltimore in April."

We are deeply grateful for the efforts of D'Andrea and all FA researchers for their labors to raise awareness of FA and to advance FA science. ♦



Adam and Molly Nash

program *20/20*.

Molly's mother, Lisa, writes "We know how lucky and blessed we are with two wonderful kids. Now we need to help others who are where we were. We owe that to Molly. We hope to help FA for many, many years until there is a cure for this terrible disease, and no

more suffering. That is what we are living for."

We thank the Nash family for their courageous willingness to share their experiences and to endure all of the risks of widespread publicity. Awareness of FA has been increased dramatically as a result. ♦

Medical Advice From our E-Mail Group

Blanche Alter, MD, on FA post-transplant surveillance:

I am a pediatric hematologist/oncologist at the National Cancer Institute (NCI), with a long-standing interest in FA. I have recently joined this e-mail list, and would like to respond to the question raised by Mr. Jackson with regard to "what is next" for FA patients who have had a successful bone marrow transplant (BMT). It is clearly very exciting and gratifying when BMT succeeds, provides a cure for aplastic anemia or leukemia, and eliminates the need for transfusions and/or androgens. However, it must be recognized that BMT (or cord stem cell transplant, or even gene therapy) is designed to replace or fix only the bone marrow. The genetic defect in other body organs is not repaired by transplant.

Unfortunately, as many of the FA families know, one of the long term concerns after BMT is the possibility of an increased risk of cancer. In particular, cancers of the head and neck have been reported following BMT in FA patients, especially cancers of the mouth and tongue. The size of the risk and whether there are excess occurrences of other specific types of cancers, over and above what is seen in the untransplanted FA patient, remain unclear. These important questions will be the focus of studies that are now in the planning stage here at the NCI.

For now, the consensus recommendation of the group of FA experts convened by the FARF in 1998, with regard to cancer surveillance, is for close monitoring of the head and neck with at least annual dental evaluations looking for white patches or sores in the mouth. New symptoms, such as persistently swollen glands in the neck, persistent pain in the mouth, tongue or

throat, chronic sores inside the mouth, or new white patches on the tongue, gums or cheeks, should be brought to your caregiver's attention without waiting for the next scheduled check-up. Since little is known about the very long term outcomes of FA patients who have had a BMT, each patient should remain under close medical observation. Specific concerns of patients and families should be brought to the attention of their physicians.

John Wagner, MD, on trying to prevent post-transplant fungal infections:

In general, we recommend discontinuing oxymetholone and starting itraconazole one month prior to BMT. Although we have not yet proven its benefit, it is hoped that itraconazole will reduce the risk of fungal infection after transplant. This presumes that the liver function tests are near normal.

While itraconazole may cause liver problems, this side effect is reversible by simply stopping the drug. The alternative is amphotericin, which has a far greater likelihood of toxicity. Amphotericin can cause kidney problems, which would prevent us from using a full dosage of other crucial drugs. Therefore, itraconazole is recommended as a first choice. Amphotericin would be used if a fungal infection were suspected or the patient were considered to be at excessive risk, i.e., colonized with fungus or a history of fungal infection.

John Wagner, MD, on clonal abnormalities:

Monosomy 7 is worrisome. But, in the absence of any longitudinal study, its real significance for a given patient is hard to predict. In contrast to non-FA patients with monosomy 7 where a bad course is the rule, this may not

be true for FA patients. Such cytogenetic clones may come and go. The bottom line is that we don't know the true significance of monosomy 7.

Nonetheless, closer surveillance is recommended if a cytogenetic study reveals a monosomy 7. This means bone marrow examinations every 4 months for a while to see if the clone goes away spontaneously, progresses (i.e., both in terms of proportions of cells involved and the addition of other cytogenetic abnormalities), and alters the way the cells look under the microscope (i.e., development of myelodysplasia or leukemia). In my own experience, I am suspicious that abnormalities of chromosomes 1 and 3 may be as frequent and as ominous, but this, too, is not proven. Any chromosome abnormality should be viewed as a call for closer surveillance just as it is for chromosome 7. ♦

David Williams, MD, will Join Staff at Cincinnati Children's Hospital

Dr. David Williams, a pioneer in gene therapy research and well known to many FA families, has announced he will be moving to the Children's Hospital Medical Center in Cincinnati in the upcoming year. He will be teaming up with Dr. Richard Harris of the Blood and Marrow Transplant program at Cincinnati Children's Hospital. Dr. Harris has performed more than 60 related and unrelated donor transplants in children with FA. ♦



Nastya Samosyuk

Family from Belarus Shares Experiences

by Alexander and Valentina Samosyuk, Belarus

We are the Samosyuk family from Belarus, one of the newly independent states of the former Soviet Union. We write to share our experiences in the hope of helping others, to thank those who have helped us, and to establish contacts with FA families in other countries.

Our eight-year-old daughter, Nastya, has Fanconi anemia. She was born with an extra thumb on her right hand. Later we learned that she had very low blood counts, and at the age of four, she was diagnosed with Fanconi anemia.

The correct diagnosis was the only help we got from her doctors. Different medical specialists told us openly that they didn't know how to treat this disease. We were desperate. Yet we refused

to simply watch our daughter die. We tried different things, including alternative medicine. The results were far from good. We kept looking for people who knew about this disease.

Three years ago, we found the FA Research Fund on the internet. We got all the available information, including newsletters and the *FA Handbook*, a book that changed our lives completely. All of a sudden we realized that people can cope with this tragic phenomenon in their lives. That was a ray of hope. The book was provided by people who didn't even know us and had probably never even heard of Belarus. We will always be thankful to the Frohnmayers and others who helped write the *Handbook*.

continued on page 13

A Successful Unrelated Transplant for Emily

By Terry & David Estes

Emily was diagnosed with FA at age 4. She was treated with oxymetholone and then transfusions, as the counts worsened and the medication failed to work. In January, 2000, we left for Minnesota to go to transplant. The hardest part of the whole process was leaving our son behind with relatives and being so far from home. Also, knowing what was to come and what Emily would experience was very tough. The day they gave Emily total body irradiation was one of the most emotional for us as parents. I can't describe the feeling you get when you see your child strapped into position and have to walk out, leaving her behind, knowing that you are allowing someone to

wipe out her immune system. This is very hard on the parents.

She received her new bone marrow on February 11, 2000. On day +8 (Feb.18) her new counts started to show. This was a day of so much joy and happiness for us as parents. Emily could not have cared less since she was experiencing all of the side effects of the chemo and radiation. She had sores in her mouth and through her GI-tract, and couldn't talk or eat. Day by day her counts continued to climb. On March 5 she was discharged from the hospital. We were very careful, almost paranoid, about what she did, and what or whom she was around. Once

continued on page 11



Emily Estes

Living in Hope

by Janet Graham

Greetings to all FA families! I am 31 years old, I live in Australia, and I have been dealing with FA since the age of six.

When I was ten, I had a bone marrow transplant at the Prince of Wales Children's Hospital, in Sydney, Australia. My donor was my perfectly matched brother. At the time of my transplant, I was given only a 10% chance of survival. This was because I was in extremely poor health at the time, and the fact that this center had never done an FA transplant before.

Many complications arose after my transplant. I suffered severe GVHD for six months. My body peeled as though I had third degree burns all over. I lost all my fingernails. I had severe thrush which affected the mouth, nose and eyes, and I still struggle with the side effects of this. I was fed intravenously through a central line. However, the line became infected and during surgical removal, it snapped in half and traveled through the aorta to the heart and lungs. Emergency surgery was performed to remove the central line. Nine months after transplant, I was discharged from the hospital. It took almost two years before I recovered from the transplant and its complications.

In February 2000, I was diagnosed with cancer of the esophagus. I was not given chemotherapy or radiation because of the side-effects, and the great uncertainty that these would be helpful. Instead, physicians opted for surgery. My tumor was 10 cm down in the esophagus, was 5 cm long and covered the full circumference of the esophagus. During a 6 1/2 hour procedure, physicians removed 3/4 of the esophagus and half of the stomach. They reconnected the remaining stomach to the top of the esophagus, using part of my small intestine. Doctors

were unable to create a stomach valve and consequently I suffer from stomach upsets and reflux. The good news is that I have been free of cancer since my surgery.

I keep very busy with my small business—a computer training service and web page design business. I teach in the vocational adult education sector which can prove very challenging at times. I teach people who usually have no computer experience. I run my business primarily from my home. My main hobby is water color painting, which I find relaxing and energizing. I also like to cook and, of course, eat. My husband does not currently work at a job, but is happy helping with the home duties and looking



Janet Graham

after our son, Joshua. Joshua will be four in May. He goes to pre-school, is very active, and loves the outdoors. That I have a son is a miracle in itself!

Living in Hope, Janet ♦

Fanconi Canada Supports Science, Helps Families

Lorne Shelson, President of Fanconi Canada, reports on the accomplishments of this organization over the past year. Fanconi Canada helped fund the research project, *Understanding Fanconi anemia through functional analysis of the Fanconi anemia (FA) group C protein*, undertaken by Dr. Madeleine Carreau of the Centre Hospitalier Universitaire de Quebec. Fanconi Canada sent a leading hematologist/oncologist to the FA Scientific Symposium in Amsterdam, and has joined with the Canadian government to fund a Postdoctoral Fellowship dedicated to FA research. The website (www.fanconicanada.org) provides information to FA families and researchers, and accepts on-line credit card donations from Fanconi Canada supporters. The second annual Ontario Region FA Picnic brought together many FA families. The organization

distributes a newsletter and brochure.

Shelson writes: "We have been running a race to help science find a cure. We have worked hard to raise funds and we thank all of you who have helped with that effort. The progress that has been made in understanding FA and developing treatments since families began fundraising for scientific research has been truly phenomenal. We desperately need to push scientific discovery ahead vigorously. For all FA patients, continued research is our best hope."

Those of you who participate in our e-group know that Annette and Lorne Shelson regularly post articles of great interest and relevance to FA families. Our congratulations and thanks to Fanconi Canada families for their hard work, impressive results, and dissemination of important information to all of us! ♦

Family Meeting Scholarship Project to Defray Costs of Attending our Annual Meeting

In August, 2000, 30 families attended the Family Meeting. With an active group of over 300 FA families, the FA Research Fund would like to double the number of families attending. However, many FA families are overwhelmed financially because of the medical costs associated with this disease and cannot afford to attend.

Fortunately, a recent large donation was made to the Fund by FA parents Bill and Jackie Lucarell, specifically to provide assistance to families to attend the Family Meeting. In addition, the Fund has received a two-year grant from Unimed Pharmaceuticals for this purpose. Because of these generous donations, the FA Research Fund has established a scholarship fund for a two-year project to defray the attendance and travel costs of families who need assistance. Currently, \$35,000 is available for this project. We will continue to raise funds for this purpose.

Participants first will be encouraged to seek other support, often available through local service organizations. Those still unable to attend without help from the Fund will be eligible for assistance. Priority will be given to the following: newly diagnosed families; those who have never attended a Family Meeting; those facing an imminent major treatment decision for which the meeting's educational program could be of immediate value; those unable to find an equivalent education and support service in their country; or those providing special assistance for language-translation or other program support at the request of the Family Support Coordinator.

The Fund wants to make this program as accessible to families as possible. If you aren't certain you meet the above criteria, we encourage you to contact the Fund. There could be

special circumstances in your situation that could enable you to qualify for the program.

Families will complete a written application available through the FARF office. The Family Support Coordinator will convene a small committee

to select scholarship recipients. Family financial information and the names of those selected will be confidential. We hope that at least thirty FA families will receive scholarship assistance during the next two years. ♦

A Successful Unrelated Transplant

continued from page 9

she came out of the hospital she never had to have a transfusion, has had no more nosebleeds and no real complications. She did experience a lot of stomach pain, diarrhea, and a bout with shingles just before going back home to Georgia. The few problems she experienced were minor for an FA patient. She was and continues to be a real trooper. She never once complained or wanted to stop the whole process.

For an 8-year-old she is a very tough little girl. She was on IV fluids and an NG-tube (a tube which allows feeding through a nasal passage) until we came home to Georgia in order to keep her kidneys functioning and to keep her weight up. After coming home she was on fluids at night for about 3-1/2 months. Once they started decreasing her CSA (cyclosporin) at 9 month the fluids were gradually decreased until she was taken off the CSA completely.

At no point through all of this has Emily shown any signs of GVHD. She is now almost one year post-transplant and is on only zantac for reflux (she has been on this since birth) and bactrim to protect against infection (this will end at one year post-transplant). She has normal blood counts. She has more energy than she's had in

years. We will continue to home school Emily for some time, but for the most part she is leading a very normal, happy life with no effects from the transplant ordeal.

When Emily was diagnosed 5 years ago we were told that our daughter would die. We were then told she had a 25% chance if she had an unrelated transplant. We were even told by a couple of FA families that if we were wise, we'd not go through with an unrelated transplant due to the bad odds. We let God direct us and it has been nothing less than a miracle for Emily.

We did our homework concerning which transplant center and which transplanters to choose. We never gave up hope, and there is hope!! Dr. Wagner and Dr. MacMillan are working tirelessly to achieve better transplant results and Emily is proof of that. Emily was primarily under Dr. MacMillan's care the whole time we were in Minnesota. We couldn't ask for a sweeter, more knowledgeable person to deal with. Emily received the best of care from the hospital nursing staff, the BMT clinic personnel, and the doctors. We can never say how thankful we are to all of them for the care she received and the ongoing work they do. Let me end by saying to all of you who are in question of what to do, or feel the devastation we felt: there is hope, there is hope, there is hope. Never give up! ♦

Life with Fanconi Anemia

by Janelle Redekop, Calgary, Canada

I had a very bad viral infection when I was 5 years old and in kindergarten. I was staying at my Auntie Cathie's at the time, while my dad was working and my mom had gone away on a little vacation. Auntie Cathie had noticed that I wasn't myself and had taken me to the doctor to get my blood tested. I was usually the healthiest in the family. They took my blood and realized that I had something very serious and unusual, so they rushed me to the Alberta Children's Hospital.

It was the day my mom got back from her trip. As soon as she got home, there was a message on the answering machine saying that I was at the hospital and that she had better come quickly! My dad was already there. I remember the doctor saying, "We're checking it out and you might have to stay a night or two." A night or two ended up being a whole month!

I missed over a year of school while I was at home with a low immune system, and I couldn't go back until my parents took me to the U.S. to get a chicken pox vaccination. I returned to school in grade 1, when I should have been in grade 2. Ever since then, I have always been a year older than my classmates. I am now in grade 4, I'm 10 years old, and living with FA.

I have low hemoglobin, platelets, and white cell counts, just as a usual FA patient would. My platelets are usually between 10 and 15 thousand, so I have to be careful not to get injured. Because of low hemoglobin I have had MANY transfusions! This raises my iron to a dangerous level that could damage my heart and other organs, so it is necessary to use a nightly IV needle of Desferal. I really dislike it! I don't mind the needle, I just don't like the reason we're doing it. I also can't have as many sleepovers with friends. It takes

a long time to stop the bleeding in the morning, and I always have bruises on my tummy.

I also get teased all the time because of my deep voice from the androgens. Kids don't realize how much it hurts and affects a person. This really seems like the worst nightmare, and A LOT of the time it is, but what keeps me going, besides family, friends and God, are the many great things that have happened in my life!

When I was seven years old, I was invited by an actress to stay at her home in Toronto. I watched her do a TV episode, and tapings of the cartoon Sailor Moon, which was my favorite show at that time. I was then recognized by the Royal Canadian Mounted Police (RCMP), as one of their honored patients in a fundraiser for bone marrow research. Thankfully, since then the RCMP have helped us raise money for FA on a regular basis!

The hospital included me on their special trip to Disneyland for the day. It was an exciting adventure for the 96 children who went along. I was thrilled to have my neighbor who works for Air Canada as my group leader! Her daughter, Candice, was kind enough to give me private swimming lessons in their back yard pool, which keeps me safe from the germs and injuries that might happen in a public swimming pool. I was given an unexpected gift card from McDonalds, which allows me to have free French fries anytime. This helps cheer me up on the way to the hospital.



Janelle and Jack Redekop

Over the past five years many newspaper and television articles have covered my condition. Personally, I feel comfortable in front of the cameras since my dream is to be an actress, and the younger I start the better. I'm also hoping this will make others more aware of FA. One of the things I really enjoyed was being a TV Co-host for the Children's Hospital Telethon. I have just been given the privilege of being chosen the Alberta Children's Hospital Champion Child for 2001. This means that I will be the children's spokesperson at the hospital's public events and presentations. I then meet up with the other Champion Children from Canada and the United States at Disneyworld, Florida in April!!

All these things I have mentioned are not as important to me as the wonderful friends I have made during the long journey of FA. I look forward to seeing them all every summer at the FA camp. This is one place I can truly have fun, and be myself! I feel we all relate to one another in a supportive and understanding way. This gives me the confidence to go on with my everyday life. ♦

Family from Belarus

continued from page 9

Ralf Dietrich, the Executive Director of the FA Fund in Germany, has played an extremely important role in Nastya's life. Three years ago he flew to Warsaw to meet Nastya and our family. Since then he has been like a guardian angel for Nastya. We appreciate his help in arranging tests for our daughter. His recommendations do not replace the advice of doctors, but have been very helpful and effective. Thanks to medicine provided by Schearing Plough, Nastya had very stable blood counts last year. Unfortunately, her counts dropped dramatically after we tried to reduce the dose of androgens because of serious side effects. It took us months to improve her condition again.

Any serious bleeding can change the whole situation for Nastya, because it is extremely difficult to arrange platelet transfusions. Each time we need one, we have to go to Minsk, the capital city, which is more than 350 kilometers from the city where we live. The situation is aggravated by economic hardship in the country. Clinics often don't have modern equipment, let alone androgens and supportive medicines. None of the drugs that Nastya needs is available in Belarus. Besides, we are the only FA family in the country and we cannot expect too much attention from doctors who are already overwhelmed with many other children's diseases caused by the Chernobyl accident.

In such a situation, any family would probably feel very alone to fight its own problem. We are happy to say that we don't have this feeling. Our family is regularly invited to attend FA support group meetings in Germany, and learn about the most recent research in the field. We feel we are part of the FA community.

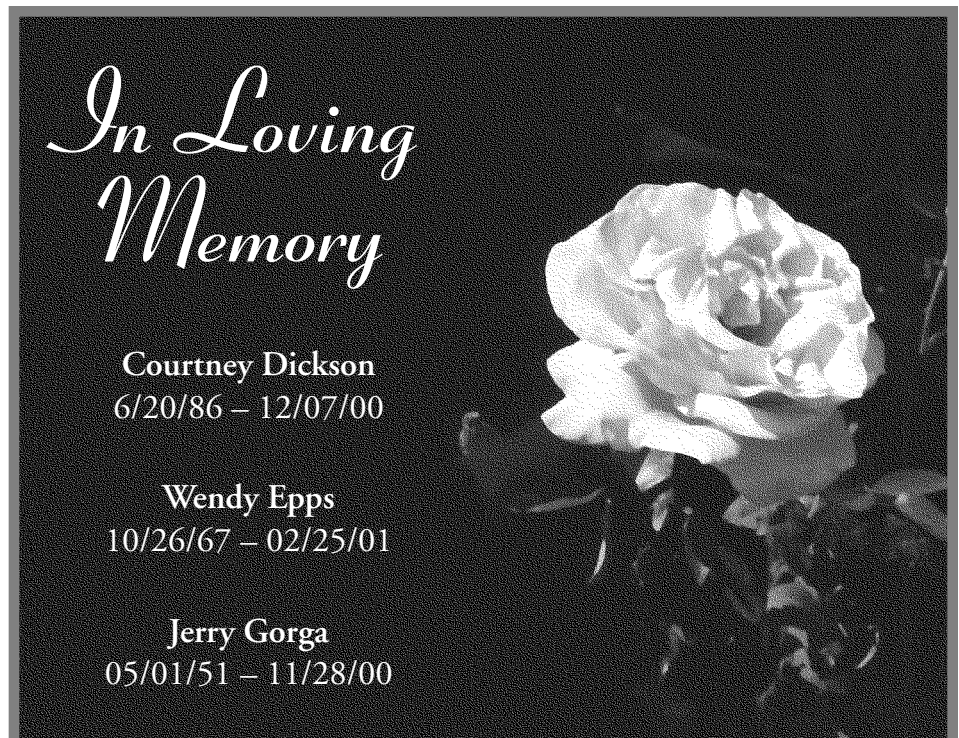


From left to right: Pawel Stankiewicz, MD, Warsaw, Poland; Ralf Dietrich, FARF Germany; Alexander Samosyuk and his wife Valentina with their two children Nastya and Irina.

About two years ago, we decided to have another baby. This decision would not have been possible without hope for a prenatal diagnosis. Ralf Dietrich arranged for us to have this test in Germany. Thanks to all those who helped us, we have a child who brings us much joy and happiness. If our experience would be of use to any other family, we invite them to con-

tact us. We would also be glad to hear from any family who would like to share their experience or simply establish contact with us.

Alexander and Valentina
Samosyuk
108/1 Kievskaya Street
224020 Brest
Belarus



We Welcome New Families Who Have Joined Our Support Group

Rosaleen Moran

Abbeystown
Caherlistrance, Co. Galway
Ireland
011 93 31242
Bernadette ~ DOB: 12/10/73
Frances ~ DOB: 3/23/82,
deceased 1/90

Roberto and Karen Enrieu

5 Dalton Gardens
Belgrave Road
Wyken, Coventry
England CV2 5BY
roberto.enrieu@bigfoot.com
Michelle

Elizabeth and Graham Walker

55 Dunlop Crescent
Dreghorn, Irvine, Ayrshire
Scotland KA11 4HN
011 12 94 217696
DOB – 8/29/75, transplanted
8/14/97 matched sibling

Andrew and Jennifer Gough

6323 E. Lafayette
Scottsdale, AZ 85251
(480) 663-1749
amgough@home.com
Shannon ~ DOB: 6/27/00

Kees and Roos van Straten

Grote Barteldweg 17
7391 CK Twello
The Netherlands
011 31 571 276924
k.vs@planet.nl
Jordy ~ DOB: 4/13/89
Chiara ~ DOB: 7/07/94, sibling
BMT April 1999
Selina ~ DOB: 7/12/86

Janet Graham

PO Box 49
Fairy Meadow
NSW
2519 Australia

Address Corrections to the Directory

Johnnie and Debra Byrd

2218 14th St. #A
Two Rivers, WI 54241-2851

Kim Frock

317 Luther Drive
Westminster, MD 21158
Listed under Missouri in the
directory

Greg Gill

31514 Rolling Meadow Ct.
Coarsegold, CA 93614-8713

Joseph and Patricia Grieco

61 Dari Road
Middle Island, NY 11953-2658

Dr. D.M. Halepoto

B-No. A8
New Wahdat Colony
Nyderabad Sindh
Pakistan

Robin Paulson

3128 Island Drive
Redding, CA 96001
Same phone

E-mail Corrections:

Fabio Frontani
fabio.frontani@mailcity.com

Charles and Dawn Church

chardawn@xsinet.co.za

Plan to Attend Family Meeting!

continued from page 1

weekend, followed by one day of psycho/social sessions on Monday. We have shortened the entire meeting by one day. Families can choose to attend part or all of the program.

Presenters will address topics such as FA 101 (a session for newly diagnosed families); decision-making guidelines for timing of treatment decisions; long-term clinical management; advancements in bone marrow transplantation; prenatal genetic diagnosis; cancer prevention and treatment; gene therapy; and understanding genes and disease mutations. On our third day, Nancy Cincotta will lead discussions on coping and living with FA. We are also inviting a nutritionist to discuss the effects of a healthful diet on the immune system.

Our wonderful volunteers from the Michigan Rotary Youth Group, plus volunteers who have helped us for years, will be back this summer. They make it possible to offer a comprehensive, age-appropriate children's program during medical presentations and group meetings. Evening activities will include bonfires, a magic show, and our now-famous karaoke night.

Families have received information related to cost and a pre-registration form. Contact the FA office for additional information or with questions.

Anyone who has attended a Family Meeting will agree that this is one of the best ways to learn about FA and gain up-to-date medical information and needed support. We hope to see you there! ♦

FA Scientific Symposium

continued from page 1

Transplantation, and Novel Therapies; FA Protein Complexes and DNA Repair; and Diagnosis and Murine (Mouse) Models.

Participants generally agreed that the most significant findings presented at this meeting were the cloning of the *FANCD2* gene by Markus Grompe, Oregon Health Sciences University, and the work presented by Alan D'Andrea, Dana Farber Cancer Institute, Boston, and others, concerning the essential role of FA proteins in DNA repair. There is now considerable evidence that FA proteins form a complex in the nucleus of the cell, that this complex "turns on" the *FANCD2* gene, and that this gene plays an

important role in DNA repair. D'Andrea also observed that the *FANCD2* gene interacts with the breast cancer gene, *BRCA1*. Researchers noted the need for additional confirmation and analysis of the FA gene pathway (see page 2).

Of therapeutic relevance were findings concerning bone marrow transplant protocols and outcomes for those with unrelated or mismatched related donors. Wolfram Ebell, MD, Charity Hospital, Berlin, presented promising early results using a protocol which eliminates irradiation in FA transplant patients. Margy MacMillan, MD, University of Minnesota, described post-transplant infection problems in FA patients and the possibility that coming to transplant before the white count is exceedingly low might improve

transplant outcomes. Presentations described efforts to improve gene therapy. Participants concurred that much more work needs to be done in this promising area.

Researchers in the laboratory of Hans Joenje, The Netherlands, have cloned and characterized the gene for complementation group E. This is the sixth FA gene to be isolated to date.

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

On behalf of all FA families, the editors give heartfelt thanks to Grover Bagby, Oregon Health Sciences University and Chair of our Scientific Advisory Board, for generously giving his time to organize and plan the Scientific Symposium, and to Hans Joenje and his colleagues at the Free University of Amsterdam, for graciously hosting this event. ♦

German Support Group Celebrates 10th Anniversary

The German Support Group, founded in 1990 by Ralf and Cornelia Dietrich, held its 14th family meeting from October 30 to November 5, 2000 in Gersfeld, Germany. Over 100 participants, including 25 FA experts and 24 FA families from Germany, Bulgaria, Belarus, Russia, Turkey, the US, Italy, The Netherlands and Hungary met to learn and celebrate ten years of progress and friendship. Seventeen presentations included lectures from Chris Walsh, gene therapy; Markus Grompe, the *FANCD2* gene; Blanche Alter, FA and cancer; Wolfram Ebell, bone marrow transplantation with fludarabine; and Hans Joenje, the *FANCE* gene. Traute Schroeder-Kurth, MD, University of Würzburg, gave an overview of 35 years of FA research, and Holger Hoehn, MD, University of Würzburg, presented the German FA awards for 2000 to Grompe, Joenje, Johan de

Winter, PhD, Free University of Amsterdam, and Schroeder-Kurth.

Families also discussed recent fundraising results in Germany and the support of research projects by the German FA Fund. There was much music, dancing, and many enjoyable activities for children and adults. A video tape from leaders of FA support groups in Argentina, Canada, France, Italy, The Netherlands and the United States congratulated the German Support Group on ten years of wonderful accomplishments. The program ended with a special workshop on cancer and leukemia in FA.

Our heartiest congratulations to the German Support Group on its 10th anniversary. You have helped enormously to push scientific discovery ahead, and have been a source of knowledge and support to all the rest of us! ♦

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.

FUNDRAISING

An Inspirational Gift to our Fund

by Vicki Athens

February 5 marked the 100th day of school, and this meant a celebration for the students of Owen Elementary School in Trenton, MI. The children celebrated the day by bringing in a collection of 100 of something. Hundreds of jellybeans and marbles, paper clips and noodles, earrings and Pokemon cards filled the school.

The collection of one child, Nathan Tracy, can inspire us all. Nathan, a second grader, is a friend of the Athens family, and has participated in various fundraising efforts. Nathan wanted to find a way to help his friend Andrew and others with FA. So, Nathan spoke to family, friends, neighbors, and people at church. He explained his 100's day assignment and Fanconi anemia. He asked them to sign his book, and if they would give him \$1.00, he would donate it to FA research. Because of the generosity of everyone, Nathan collected 100 signatures to take to school,



Nathan Tracy

and he collected \$100 for FA.

We believe the answers to FA are within our reach. It is our obligation to fund research. When the task of raising funds seems insurmountable, Nathan's example of raising \$100 in a weekend can inspire us all. Thanks Nate! ♦

On-Line Credit Card Donations

On-line credit card donations are now possible on the FA Research Fund Home Page (www.fanconi.org).

The Fanconi Anemia Research Fund has recently installed an on-line payment system with *PayPal*. Credit card donations can be made through *PayPal* without leaving the FA Research Fund web site. Look for the *PayPal* button below the *Donations* line on the FARF home page. ♦

FARF Eligible for Combined Federal Campaign

The FA Research Fund has just been notified that we are eligible to participate in the 2001 Combined Federal Campaign (CFC). Our organization will appear in the listing of "National/International Organizations" which is published in each local campaign brochure. The CFC identification number donors will use to design-

Event Insurance

More and more families have been holding wonderful events in their communities to raise funds for Fanconi anemia research. We very much support these efforts. However, we have been advised by our attorney and our insurance carrier that "event insurance" is necessary to protect the sponsoring family and the FA Research Fund from liability in the event that a participant (at the dance, the auction, golf tournament, etc.) incurs an injury. Many hotels will not allow an event to occur on their premises without written documentation of such insurance.

We have talked to our insurance carrier and have found that the best way to handle this is for the sponsoring family to determine if a "rider" can be purchased on their homeowner's insurance for such a one-time event. These "riders" are usually quite inexpensive. The Fund would be willing to pay all or part of this expense. In addition, if the sponsoring family is not able to purchase event coverage through their insurance carrier, the Fund will attempt to make other insurance arrangements to cover the potential liability. ♦

nate their contribution to FARF is 1183.

Please make this information known to friends and neighbors who are employees of the U.S. government. This is a painless way to allow them to earmark their annual giving and to extend our FARF donor base. ♦

Coping with Fanconi Anemia Through Knowledge and Fundraising: One Family's Efforts

By Christie Kelley

As many of you know, fundraising is a crucial part of funding research in hopes of finding a cure for Fanconi anemia. Our six-year-old son, Hunter, was diagnosed with FA in March of 2000. While the diagnosis gave us new insight into previous medical problems, the news has been devastating.

At first, all we could do was cry. We really did not know where to turn. We had so many questions for our doctors and so many of them went unanswered. We knew that we had to get as educated as possible on FA and the treatments for it. After joining the support group in May, we decided to attend the family meeting in Wisconsin. That was the best thing we could possibly have done! We gathered so

much information and for the first time since Hunter's diagnosis, we felt like we might be able to move forward with our lives. It was then that we decided that feeling sorry for ourselves would not get us the results that we wanted for Hunter and others with FA. We decided that we wanted to make a difference.

Our first fundraising effort grew from a neighborhood letter campaign into a costume ball/silent auction. As some friends were going door to door in our neighborhood of 300 homes handing out a letter describing Hunter's diagnosis and asking for a donation to the Fund, a different group of friends was organizing a costume ball and silent auction to be held the end



Hunter Kelley

of October. It was really amazing how many people were willing to help. Something small quickly grew into a huge event! With local radio and TV coverage and fliers going home in backpacks from many schools, total strangers were calling wanting to know how they could help. With the help of many friends, we put together a very successful event in about nine weeks. It was a lot of work but it was also a lot of fun! After the event, we felt we had not only raised a great deal of money for research but we had also helped educate the city about FA. Plans are already in the works for The Second Annual Costume Ball and Silent Auction benefiting Hunter Kelley and the Fanconi Anemia Research Fund.

We would like to urge everyone to try some form of fundraising. You will be amazed at how easy it is and how many people are willing to help. Even if you start out with something small like we did, you may see that it could quickly blossom into something much more than you ever imagined!! ♦



Thanks to our Volunteers!

For years FARF has benefited from a dedicated group of volunteers led by coordinator Arthur Golden. In this picture, Dave Frohmayer stops by to thank some of the volunteers as they work hard to send out a fundraising letter. From left to right: Bill Wiley, Roberta Phillips, Arthur Golden, Dave Frohmayer, Mary Ellen Eiler, and Vi Johnson.

Family Fundraising Efforts for the Past Six Months

From July 1 to December 31, families raised \$587,242. Our Fund also received \$11,620 from the Combined Federal Campaign and United Way. Our special thanks to all of you who have worked so hard to raise needed research dollars.

An additional \$36,530 was donated in loving memory of children and young adults we have lost all too soon to this devastating disease.

\$40,000 & up

Dave & Lynn Frohnmayer
Randy & Christie Kelley
Kevin & Lorraine McQueen

\$20,000 - \$39,999

Andrew & Vicki Athens
Bill & Jackie Lucarell

\$10,000 - \$19,999

Michael & Beth Vangel

\$5,000 - \$9,999

Joseph Chou
Allen Goldberg & Laurie Strongin
Jeff & Judy Hoffman
Charles & Katy Hull
Robert & Mary Nori

\$1,000 - \$4,999

Chris & Susan Collins
Ray & Diane Cronin
Ed & Janice Duffy
Gary & Melody Ganz
Susan Jackson
Jeff & Beth Janock
Robert & Jennifer Kiesel
Eric Kjos-Hanssen & Turid Frislad
Peg LeRoux
Deane Marchbein & Stuart Cohen
Gil & Peggy McDaniel
Sheila Muhlen
Jack & Lisa Nash
Robert & Andrea Sacks
Bill & Connie Schenone
Jim & Carol Siniawski

Mark & Susan Trager
Mark & Sandy Weiner
The Welfare Family

Up to \$1,000

Ken & Jeanne Atkinson
Tracy & Melody Austin
John & Audrey Barrow
Mark & Linda Baumiller
Randy & Nancy Bloxom
Paul Brodie
Tad & DeeDee Burzynski
Brian & Margaret Curtis
Bill & Pat Danks
The Day Family
James & Carol Dillon
Pat & Mary DiMarino
Antonino & Marie DiMercurio
Paige Ellis
Fabio Frontani
Pat & Maria Gleason
The Gorga Family
Dave & Paula Guidara
Mitchell & Tirzah Haik
Roger & Eleanor Herman
Eugene & Renee Lemmon
Rene LeRoux

Tucker Lovejoy
Dennis & Sharon Lower
Greg & Lynnette Lowrimore
Susie Mandel
Tom & Marilyn Massino
Jack & Pam McCarty
Steve & Allison McClay
Cecelia Meloling
Lynda Moureau
Kenny & Lisa Myhan
Louis & Virginia Napoles
Bob & Alice Nicholson
Ron & Freddi Norris
Lynn & Shirley Quilici
The Russo Family
The Scaff Family
Erik & Lori Salo
Robert & Linda Scullin
Tommy & Brenda Seiford
Matt & Diane Senatore
Calvin & JoAnn Shields
Bryan & Karen Siebenthal
Jeff & Debby Slater
Anne Marie Thorstenson &
Martin Persson
Jennifer White

A Special Word of Thanks from our Executive Director

by Mary Ellen Eiler

I want to thank all of you for your exceptional fundraising efforts this past year. Thanks to you, we far exceeded our fundraising goal for research and family support. In fact, FA families raised 22% more money in 2000 than in 1999. Not bad! Ten families raised over \$10,000 each this last year, and several of those ten raised far more than that. But, more importantly, more families than ever took part in fundraising this year—23% more than 1999—

from writing letters to their friends and families to holding major fundraising events. Whether you raised thousands or \$100 this year, you get lots of credit and thanks from me. I can only imagine how hard it is to deal with Fanconi anemia. Despite that, you found the strength to rise to the occasion to let your friends know about the need for funds. Thank you very much. ♦

New Staff

Since the last newsletter, we have experienced some turnover among our staff. Susan Castillo resigned as Director of Development at the end of December to fulfill her duties as a senator in the Oregon State Legislature. Joachim Schulz, our Executive Director, resigned in July to open a consulting business. He has since become a Director of Development with Holt International Adoptions, which is based in Eugene. We miss them both and wish them well in their new endeavors!



Jill Emerson

Jill Emerson joined us in early December as an administrative and family support assistant. She recently retired from the North Slope Borough School District in Barrow, Alaska, where she had a 25-year career as a schoolteacher and technology coordinator. She also worked for the Alaska Department of Education as a co-leader of the Alaska Math Consortium. Jill received her Bachelors and Masters degrees at the University of Oregon. She is delighted to return to the relative warmth of Eugene after the many years in Barrow!



Mary Ellen Eiler

Mary Ellen Eiler was selected by the Board of Directors for the Executive Director position and began her duties at the end of July, 2000. She served on FARF's Board of Directors from 1997-2000, and was Board President for two years.

Mary Ellen retired in 1997 from a 31-year career with the state of Oregon, first as a regional administrator for Children's Services Division and, later, as the superintendent for the Oregon Youth Authority.

Mary Ellen is an exceptionally hard worker, who devotes countless weekends and evenings to our Fund, in addition to her regular work schedule. She is well-organized, thorough, and tenacious. She has a keen intellect, and has quickly gained an understanding of the complexities of Fanconi anemia. We are deeply grateful for her dedication to this effort, and her willingness to devote her precious time to our cause.

PGD for Fanconi Anemia at the Reproductive Genetics Institute

continued from page 6

or not, and \$2,500 to determine HLA compatibility with the FA patient). There are additional costs for medication and the IVF procedures. The RGI can carry out the *in vitro* fertilization and embryo transfer at the Chicago facility. The RGI has relationships with IVF centers around the world, so that the IVF process can sometimes be done locally, while the genetic analysis is done at the Chicago facility.

For further information and contacts:

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Christina Masciangelo, M.S.,
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Allen Horwitz, M.D., Ph.D.,
Director of Medical Genetics;
Yury Verlinsky, Ph.D.,
Director of RGI

Use of Logo

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



The Thirteenth Annual International FA Scientific Symposium

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Hilton Hotel Portland, Oregon



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We'll Miss You, Leslie

Leslie Roy resigned from her position as Family Support Coordinator on March 9. She moved to California to help her aunt, who has early-stage Alzheimer's. She began work at Orange Coast Community College on March 15; she also hopes to complete her goal to finish her Bachelor's degree.

Leslie worked for three years as our office manager; for the past six years, she has provided support to FA families around the world. Leslie became very knowledgeable about Fanconi anemia, and tirelessly assisted families in their efforts to become better informed. She created e-groups so that families could share knowledge and give support to one another. Families describe Leslie as compassionate, deeply caring, and extremely helpful. Families could always count on Leslie to help them through times of crisis and loss. Leslie devoted much of her life to our families and to this cause, and many of us became her close friends.

We will miss you, Leslie.