FA family newsletter

A Semi-Annual Newsletter On Fanconi Anemia For Affected Families, Caring Physicians & Research Scientists.

Newsletter #14 Summer/Fall 1993

Family Meeting Brings Hope

Forty families from 21 states and three countries attended the third annual Family Symposium, held in Bloomington, Minnesota on July 16, 17 and 18. This family meeting wasn't our biggest, but t valuations told us that the lectures, the small groups and the informal encounters were the most fulfilling and hopeful yet. For those who could not attend, we missed you. Lay summaries (see supplement) capture most of the scientific presentations. Here are some highlights:

- **Dr. Arthur Nienhuis** is one of the world's pioneering gene therapy experts. During a fascinating lecture on gene therapy for Fanconi anemia, he informed us that human clinical trials for those FA patients in group C (the newly discovered gene) could begin in early 1994. Laboratory studies have already demonstrated that some of the defective FA cells from patients in this group can be corrected using a retrovirus carrying the healthy gene. This is stunning progress!
- **Dr. Markus Grompe** has been collaborating closely with the National Institutes of Health to identify FA group C patients who could be included in the forthcoming NIH group C gene therapy trial. Grompe pioneered the discovery that FA individuals of Ashkenazi Jewish descent have a high likelihood of being in complementation group C. His lecture explained a variety of ways in which mutations of the FA(C) gene can be detected in different patients, including those of non-Jewish descent.
- **Dr. Richard Harris** revealed heartening progress in successful treatment and survival of FA patients undergoing marrow transplantation. Using an HLA matched sibling donor and before leukemic transformation, all 21 patients, treated with a preparatory regimen developed by Dr. Harris, are alive and well. These patients have survived from two months to 70 months post transplant.

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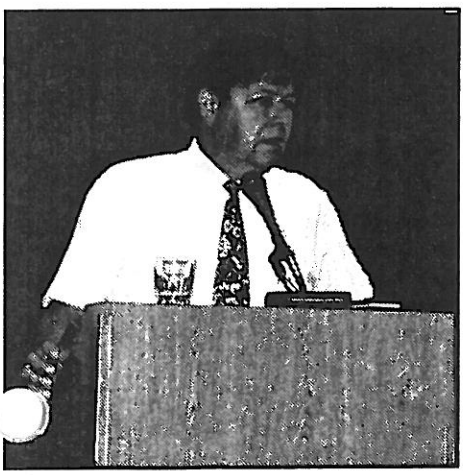
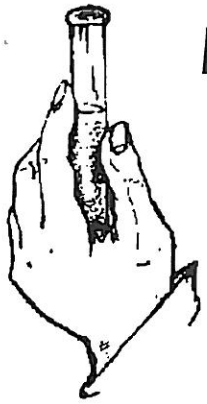
Informal discussions enrich meeting.



Art Nienhuis, MD Addresses Family Meeting.

HIGHLIGHTS



Medical News

Researcher Finds Gene in FA Families

The important discovery of one of the Fanconi anemia genes (FACC) has enabled research to proceed rapidly in a number of different directions. Of tremendous interest to FA families is that it is now possible to determine which individuals are defective in the one gene discovered to date.

Researchers at the Oregon Health Sciences University, led by **Dr. Markus Grompe**, have been working hard to determine which FA patients belong to complementation group C. They first analyzed 17 FA patients, and found that three (or 18%) had mutations in the FACC gene.

Interestingly, two of the three patients had identical mutations. Upon further questioning, both families were of Eastern European (Ashkenazi) Jewish descent. Researchers suspected that this particular mutation might be common among Jewish FA patients. A total of twelve Jewish FA families were examined, and ten of these or 83% had the same splice mutation. This indicates that this mutation causes the majority of Fanconi anemia in Ashkenazi Jews and that most Jewish FA patients belong to complementation group C.

Dr. Grompe and his colleagues next analyzed 314 Jewish individuals from the general population (not affected with FA) to detect the frequency rate of FA carriers. They discovered that .6% were carriers. This means that if a known FACC carrier marries a spouse of Jewish descent, their chances of having a baby with FA would be 1/600.

Dr. Grompe has now completed the analysis of 43 families in the OHSU cell repository. He has found 12 FACC families, of which 10 are Jewish. Only 2/31 (6%) of the non-Jewish families analyzed to date belong to complementation group C.

The National Institutes of Health is now developing a protocol for a gene therapy trial. Patients in FACC are potential candidates for this trial. In addition, accurate carrier detection and prenatal diagnosis is now possible in families belonging to complementation group C.

Oregon Health Sciences University will continue to analyze FA cell lines for mutations in FACC, and will keep families informed of their results. This process will be repeated as each additional gene is discovered.

Mutation Analysis of the Fanconi Anemia Gene FACC

Arleen D. Auerbach and Peter C. Verlander

Fanconi anemia (FA) is a genetically diverse disorder defined by hypersensitivity of cells to specific DNA damaging agents. There are at least four different genes which may result in this disease; the gene for one of these (FACC) has been cloned in Dr. Buchwald's laboratory in Toronto. Our laboratory at The Rockefeller University has screened DNA from approximately 175 racially and ethnically diverse FA families for mutations in this gene. These families are entered into the international Fanconi Anemia Registry (IFAR) at The Rockefeller University, and have had diepoxybutane (DEB) testing for FA in our laboratory. We believe that our methods have sufficient sensitivity to detect 95% of all mutations in FACC. Some of our findings are summarized below.

Mutations in the FACC gene were found in approximately 15% of all FA patients. We have identified 6 different mutations in FACC that appear to be related to the disease. Approximately 50% of the patients with FACC mutations are Jewish, of Ashkenazic and Sephardic descent, and all have two copies of the same mutation. This specific mutation was not found in any non-Jewish patient in our population sample. A single, different mutation was found in 40% of the patients with FACC abnormalities, while the other 4 FACC mutations occurred less frequently, and together accounted for 10% of cases. Non-Jewish patients with FACC mutations were all Caucasian, of Northern European or Italian ancestry.

We have found that the severity of FA is related to the specific mutations present in the DNA of the patient, and to whether both parents carry the same mutation. Jewish patients who have a mutation in FACC have a severe clinical picture, with multiple congenital abnormalities, and relatively early onset of hematologic disease. The majority of the other patients with mutations in FACC have a milder form of the disease.

We recommend that you share the information in this report with your physicians. Anyone wishing information on the results of our testing in their families, or regarding carrier screening in their extended family may contact us at:

The Rockefeller University
1230 York Avenue
New York, New York 10021
Telephone: (212) 327-7533
FAX: (212) 327-8232

Editors' note: For more information about the International Fanconi Anemia Registry, see our booklet, *Fanconi Anemia: A Handbook for Families & Their Physicians*, pp 25-26.

It's not where we are, it's the direction we are going that is important.

– Oliver Wendell Holmes

Linda and Dennis Solin Honored

At the opening dinner of the Family Meeting, **Linda** and **Dennis Solin** were honored for their years of tireless effort on behalf of FA families throughout the world. In 1989, Dennis Solin generously volunteered his office and staff as headquarters for our early fundraising efforts. His professional skills were invaluable in creating our formal organization. He secured our tax exempt non-profit status. As President of our Board, Dennis has provided the leadership and guidance to help create what a noted researcher recently called the "prototype organization for an orphan disease". Your editors conservatively estimate the value of Dennis Solin's direct and in-kind contributions to our Fund over the years to exceed $60,000.

Before becoming our coordinator, Linda Solin worked as a full time volunteer for our Fund for a year and a half. As a student intern, she donated her entire stipend to our research Fund. For the last three years, she has been our overall coordinator. She has helped organize four international scientific meetings, three family meetings, and a pilot project FA family week at Lake Sebago, Maine. She has been successful in obtaining numerous grants for scientific research and family support. Families have deeply appreciated the quality of help and emotional support routinely received from Linda.

Editors Postscript:

With deep regret, we announce Linda's decision to resign her position at the FA Research Fund. She will join the District Attorney's office as Director of the newly created Child Advocacy Program. Linda will be missed greatly by all of the FA families who have come to know her and appreciate her special contributions to our effort. We wish Linda well in her new position. She has our heartfelt thanks for her pivotal role in making our organization what it is today.

Scientific Advisors and FA Research Fund Board Meet

Thanks to a generous grant from special friends, we held the first ever joint meeting between our Board of Directors and our Board of Scientific Advisors in Chicago, Illinois on June 26, 1993. This unusually productive meeting reviewed research in progress and charted new avenues for targeted research funding in the future.

Our reviewers were unanimous in their strong recommendation that we provide continued funding support to discover the remaining FA genes and to hasten studies into the mechanisms of the gene defects. They identified strategies to involve DNA repair researchers who work with yeast and animal models. A particularly valuable outcome was the collective effort to identify new laboratories and new scientists with expertise related to FA. Attendees helped develop an agenda for scientific presentations for our 5th international scientific symposium. That meeting will be held in early December in St. Louis, in conjunction with the American Society of Hematology meeting.

We deeply appreciate the uncompensated and selfless service of these distinguished scientific advisors.

European Scientists Collaborate

Hans Joenje, PhD reports that his research team at the Free University, Amsterdam is collaborating with researchers at the University of Leiden,The Netherlands, France, Germany, Italy, and **Manuel Buchwald's** laboratory in Toronto, Canada, to form an international network for FA research ("EUFAR"). EUFAR's objectives include: to determine the number of genes causing FA and to clone those genes; to determine how these genes control chromosomal stability and prevent cancer; and to determine the FA gene mutations found in European FA patients.

The success of EUFAR greatly depends on the collaboration with FA patient support groups which now exist in Germany, Italy, France, TheUnited Kingdom and The Netherlands. The German support group has already played an essential role in collecting blood samples from many FA patients and their family members.

Family Meeting

(Continued from page 1)

- Encouraging statistics show the feasibility of matched unrelated donor transplantation as a potential therapy for FA patients. Given the great increase in the number of marrow donors in the National Marrow Donor Program, **Dr. Norma Ramsay's** presentation suggested that this resource will be of increasing importance for FA families who lack a matched sibling donor.

Three eminent physicians lucidly explored treatment suggestions.

- **Dr. Nasrollah Shahidi** presented valuable insights into the effects of viral and bacterial infections on the bone marrow, the effects of transfusions on marrow function, and the use and misuse of androgen therapy. He explored the potential beneficial effects of vitamin therapy and good hygiene in possibly preventing malignancies.
- **Dr. Blanche Alter** discussed the implications of clonal abnormalities in FA patients. She gave an overview of laboratory experiments using various colony stimulating factors with FA bone marrow cells. In the laboratory, FA cells were most responsive to stem cell factor (SCF). Unfortunately, because of a variety of safety concerns, SCF is not expected to be available for clinical trials for some time.
- **Dr. Joseph Gertner** explored the advantages and disadvantages of the use of human growth hormone with FA children. He outlined possible risks, and cautioned that significant results might be expected primarily in FA patients deficient in human growth hormone.

Numerous small group discussions were received with great enthusiasm by FA children, their siblings, and parents. These sessions provided a much needed outlet for the sharing of feelings, fears, problems, coping strategies and hopes.

FA Families Attend Camp Sunshine

In the spring of 1993, the FA Support Group was invited to attend Camp Sunshine at Lake Sebago, Maine. This is a unique camp for families who have a child with a life-threatening illness. Because of the short notice (and long distances!) only eleven FA families were able to attend this five-day camp. However, all who attended were extremely impressed by the Camp Sunshine program and felt they benefitted enormously from this experience.

Children spent their days in well-organized, varied activities. Parents could participate in morning discussion groups and explore their feelings about living and coping with their child's medical problems. They took long walks, read, played tennis and enjoyed the beauty of Camp Sunshine. Organized evening programs provided entertainment for all ages. Apart from the often considerable expense of getting to Camp Sunshine, all other expenses (food, lodging) were free to the families.

Our eleven FA families reflected on their five days at Camp Sunshine. Here is what some of them wrote:

"I liked the feeling of belonging. I finally found people who feel the same way I do - and it's not wrong! It's a place where you are comfortable, your children are comfortable and you are understood."

"This was a very positive experience. I was concerned prior to arriving that it might be very depressing but it was a lot of fun! The care and love expressed by the volunteers is incredible."

"It's a safe haven where the family can heal the family fabric."

"The support groups were so great! They helped us become sane!"

"It's a place you can go and leave many of the daily burdens of caring for an ill child behind. It's a happy and caring environment where you can grow and learn through the workshops and the experiences of other families facing a similar situation. The staff members at Camp Sunshine have thought of everything to make your stay fun and pleasant. I [illegible] refreshed and stronger."

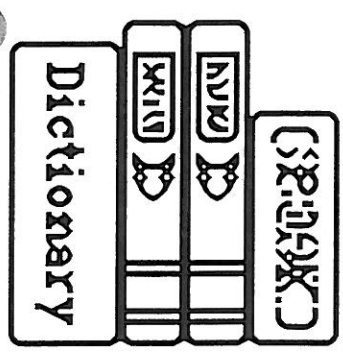
Families enjoy the beach at Camp Sunshine. From left to right: Christen Athens, Vicki Athens, Adam Day, Andrew Athens, Rich Day and Dotty Day.

Andrew Athens enjoys his time at the beach.

At our July Family Symposium, we asked everyone to evaluate the possibility of using Camp Sunshine for our next family meeting. We would try to combine the camp program with the formal scientific and medical presentations which so enrich our family meetings. We are now in the process of evaluating responses.

Nancy Cincotta led stimulating discussion groups at Camp Sunshine.

Resources & Tips for Families



Bone Marrow Transplant Handbook

Susan K. Stewart, editor of the BMT Newsletter, has just published a most welcome new 157 page book entitled *Bone Marrow Transplants - A Book of Basics for Patients*. The book is written in lay language by and for marrow transplant patients. The authors were assisted by medical specialists from numerous transplant centers.

This new book covers a wide variety of important topics, from coping with stress to issues concerning related, unrelated and mismatched transplants, to post transplant issues and complications. The book contains a helpful glossary of medical terms.

The Director of Nursing of the transplant program at the Fred Hutchinson Cancer Research Center said of this book: *"Our families value the resource because it is technically accurate, easy to read, comprehensive and universally appropriate regardless of where they are treated".*

This new book can be ordered from the BMT Newsletter, 1985 Spruce Avenue, Highland Park, Il 60035. Enclose a five dollar donation payable to "BMT Newsletter". Reduced prices can be obtained for multiple copies. Phone (708) 831-1913 or FAX (708) 831-1943.

A Resource About Siblings

Barbara Azrialy wrote to us, asking that we inform you of her recent publication. She is the author of *The Sibling*, a handbook for understanding the brother or sister of a child with a handicap.

The author includes excerpts from relevant articles and books, information from interviews with scores of siblings, and insight from personal and professional experiences.

The book can be purchased for $11.00 (including postage and handling) at:

Azrialy
5414 Newcastle #61
Encino, CA 91316

Questions should be directed to the author at (818) 344-8011.

Transportation Assistance Available

The Aplastic Anemia Foundation of America, New England Region, has initiated a patient assistance program to ease transportation costs for aplastic anemia patients. The program provides reimbursement for mileage and parking up to $200 per 12 month period for the patient. Any individual who has been diagnosed and resides in the New England states (Maine, Vermont, New Hampshire, Massachusetts, Connecticut or Rhode Island) is eligible for the assistance program. Please contact:

Aplastic Anemia Foundation
of America
New England Region
P.O. Box 1645
Brookline, MA 02146

Massachusetts Offers Unique Programs

Two unique programs offer medicaid coverage to Massachusetts residents independent of their financial status. The Kaileigh Mulligan program is available for families whose child or children meet very strict medical guidelines. Among other criteria, the child must be at risk for hospitalization and have a daily need for two distinct, skilled nursing services. For more information, contact Louise Bannister at 617-348-5527.

Common Health is for Massachusetts children who qualify medically for SSI but whose families are not financially eligible. Families pay monthly premiums which entitle them to Medicaid. For additional information, call 617-348-5059.

Does your state offer similar programs? Let us know, so that we can spread the word!

Do you need help with insurance reimbursement?

BMT Newsletter maintains a list of attorneys in several states who are experienced in litigating and/or settling bone marrow transplant insurance reimbursement problems. To obtain the list, phone 708-831-1913 or write

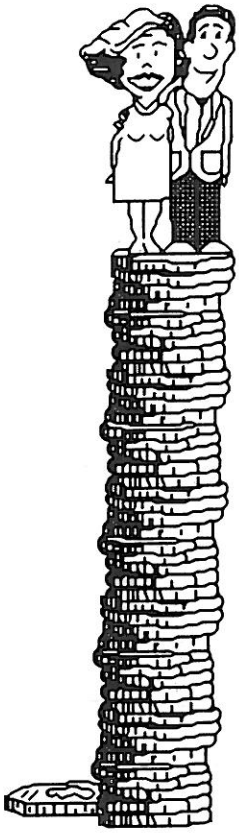
BMT Newsletter,
1985 Spruce Ave.,
Highland Park, Il 60035.

We reprint two helpful articles from the *Aplastic Anemia Foundation of America Newsletter*, Summer, 1993:

EMLA CREAM, developed by Astra USA, is the first needle-free anesthetic that can penetrate skin to stop the pain of injections and other invasive procedures. Recently approved by the FDA, this product can relieve both the physical and therefore emotional pain patients experience when faced with daily and weekly needle use. The cost is estimated to be between $1 and $3 an application.

DENTAL CARE can be tricky for aplastic anemia patients because of the risk of bleeding gums. Dentists recommend purchasing a brush with the smallest head possible and running the bristles under very hot water for 1 to 2 minutes before brushing. This makes the bristles more pliable and softer on the gums. Use only a toothbrush and toothpaste recommended by the American Dental Association. For further dental care, ask your dentist to prescribe an anti-microbial mouthwash to aid in plaque reduction and gingivitis. It is important that patients get regular dental checkups because of drug side effects.

Families Share Fundraising Ideas



Seven FA families shared their insights and techniques on fundraising at the Family Symposium. All of us were inspired by their ideas and impressive efforts.

Fredi Norris presented an articulate, impassioned plea for each family to do whatever it can to raise research dollars. She was concerned to note that only 45 out of 256 families raised or contributed money last year. She emphasized that the amount raised is less important than the fact that everyone does his or her best to help.

Marlene Stone, grandmother of FA patient **Jessica Paulson**, has done an annual fundraiser each year for the past four years. She had to swallow her pride to ask friends for help, but the importance of the goal enabled her to proceed. Her daughter, **Robin**, is now able to approach her friends and ask for their help. Marlene read Robin's poignant, personal fundraising letter. It touched us all deeply. Feel free to write our Fund for a copy of this effective, moving letter.

Pat and **Bill Danks** spoke with their usual humor and effectiveness about mobilizing a community to plan and execute a highly successful fundraising event. The Danks' Beef and Beer parties are tons of fun, bring in loads of money and - to hear the Danks tell it - require no work because your friends do it all!

Linda Sankey outlined her plans for a craft fair this fall, and asked families to support the first ever FA Family Cookbook. **Martin Sankey**, who kindly donates his time and energy to produce this newsletter, spoke of working within one's corporation to obtain in-kind donations.

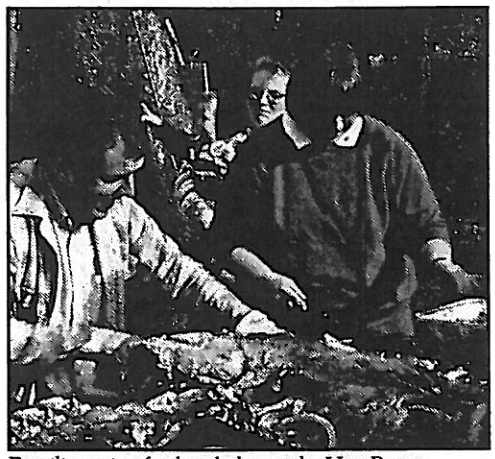
Ed Brookover gave many useful tips on how to write a good fundraising letter. His own letters are outstanding, and he kindly offered to help any FA family with the writing, editing or execution of a fundraising appeal. Fundraising is actually a favor to others because it gives them a chance to help you!

Darryl Blecher and his wife, **Diana Fitch**, formed a committee of their friends and let this committee plan and executive their fundraising drive. Darryl spoke with enthusiasm about how they mobilized their entire community to support their efforts.

Leonard Riley conducted a successful fundraising effort following his daughter's successful bone marrow transplant. He spoke eloquently of the need to help not only our own children, but the many FA children yet to be born who might be spared what our children have endured.

In April, 1993, **Gary Gangwer** and the Emanuel United Church of Christ hosted a tremendously successful Hog Roast. Proceeds to date total $10,800, which will be shared by the FA Research Fund and Riley Children's Hospital.

Gary and his friends roasted twelve hogs (1800 pounds!!) and served pork to 873 people. A pie and cake auction and raffle tickets for items such as airline tickets and cellular phones brought in additional donations. Clowns painted faces on children which netted an additional $800. Food and raffle items were all donated by the community. Over 120 volunteers worked to make this event a huge success.

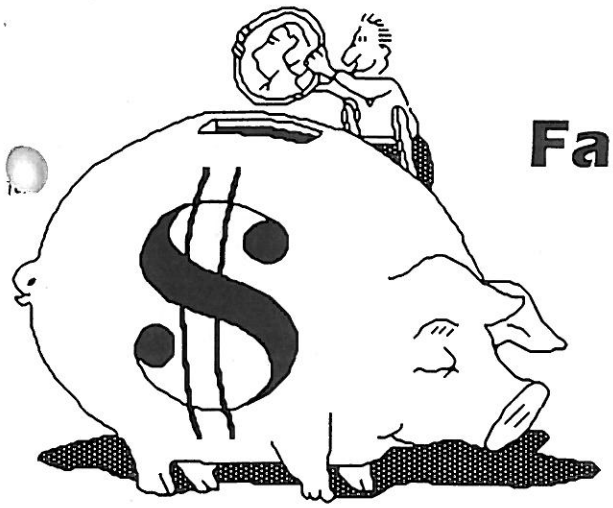


Families enjoy food and play at the Hog Roast.

Gary believes that this event brought people in his church and community together as never before. A strong feeling of camaraderie and the conviction that people can pull together to create something important came from this effort. On the Sunday following the Hog Roast, Gary sang "You'll Never Walk Alone" to the church congregation. There was not a dry eye in the church.

Mary and **Pat DiMarino** astonish and impress us with their many creative ways to bring in research dollars. Over the holidays, Mary mailed a fundraising letter, which netted $3,400. A delicatessen and bagel shop displayed a tin can with Danielle's picture and an explanation of FA, and this raised almost $900. A relative organized "jeans" day to raise research dollars, and also sold red ribbons for $3 each to benefit FA research. In the spring the DiMarino's community sponsored a baseball game to raise funds, and Pat DiMarino organized a golf tournament. Whew!! Mary writes:

"To be honest with you, when I first started this fundraising endeavor, I was very uncomfortable and apprehensive about asking people for money. Then I thought about my daughter's precious life and as you can see, I haven't stopped yet!! Pat and I are overwhelmed at the generosity of people. We are fortunate and blessed to have such good people behind us. It makes us feel less alone, knowing people care.



Family Fundraising Efforts

During the past six months, thirty FA families conducted fundraisers, and another eighteen families made generous contributions to our effort. Tons of thanks for your wonderful help! We still have a long ways to go before meeting our Tiger challenge, so your continued assistance is deeply appreciated. Our efforts support scientific research, and have helped produce stunning results in just a few years. We must continue our hard work! If each family could raise $1,000 per year, we could maintain our present level of research support. For many, this goal is impossible, but others can do more. Every single contribution, large or small, is vital to our success.

The following families have assisted our efforts during the past six months. Thanks to each and every one of you!!

$15,000 - 20,000:
Ron & Fredi Norris

$5,000 - 10,000:
James & Donna DellaRatta
Pat & Mary DiMarino
Debbie & Jeffrey Slater

$1,000 - 5,000:
Vicki Athens
Al & Laura Berman
Phyllis Cafaro
June & Hugh Delvalle
Dr. Sidney & Ethel Farkas
Dave & Lynn Frohnmayer
Deanne Marchbein & Stuart Cohen
Linda & Bob Scullin
Sandy & Marc Weiner

$500 - 1,000:
Dotty Day
Janice & Ed Duffy
Gary Gangwer
Leardon Keleher
Jennifer & Robert Kiesel
Matt & Diane Senatore
Mark & Susan Trager

Up to $500:
Pamela Baxter
Diane & Michael Bradley
Delores, Carol Ceresa, Paula Guidara
Pat & Bill Danks
Maria Duran
Jim Galvin
Greg Gill
Mr & Mrs. Kwang S. Ha
Ousama & Souha Halteh
Irene & John Kalman
Leslie & Barbara Lawrence
Eugene & Renee Lemmon
Bill & Jackie Lucarell
Marilyn & Tom Massino
Jack & Pamela McCarty
Alison & Steve McClay
Lorraine & Kevin O'Connor
Ron & Cindy Poe
Pat & Ken Rau
George & Kathy Reardon
Leonard & Jan Riley
Terry & Therese Robertson
Martin & Linda Sankey
Dick & Judi Selke
Mitzi & Dave Speelman
Marlene Stone
Michael Tauber
Alex & Marlene Violassi

We also report, with appreciation yet great sadness, that contributions were received in loving memory of **Bryan (BJ) Gill.**

Jackie Lucarell pays special tribute to her aunt and uncle for their sacrifice and unwavering efforts to support FA research:

*"My aunt and uncle, **Frank** and **Marilou Linsley**, have been contributing to Fanconi anemia research since your family was on television in January of 1989. They have been making monetary donations of $50-$100 regularly ever since and continue to do so, even though my uncle's job is on shaky grounds.*

They are not wealthy people but they both have hearts of gold. They are very dedicated to finding a cure for FA. They are an inspiration to all of us and we can't be grateful enough for all they have done for Jimmy and all FA victims.

I am very proud of my aunt and uncle. Along with their children they have donated over $3500 to FA research."

From all of the FA families, a heartfelt thanks to Frank and Marilou Linsley!

Pam Baxter sends this touching message:

*"The week of May 3rd our New Bloomfield schools took part once again in raising funds for **Bradley** and **George**.*

Seeing these wonderful children bring in coins, dollars and checks was the medicine my heart needed. Love comes in many forms and in this case the love is the children. The boys are learning it is as important to be related by love as to be related by blood.

Use this money to find a CURE, not only for Bradley, George and my sake, but for the sake of our extended family, our Community."

From Our Families

We provide excerpts from two beautifully written and informative letters by **Neil Frank**. Neil and **Iris** are the loving parents of 20 month old **Rebecca**.

Neil and Iris knew from ultrasounds before Rebecca's birth that Rebecca had "duodenal atresia" (stomach not connected to the rest of her digestive tract) and a likely missing kidney. Neil describes Rebecca's birth and subsequent surgeries:

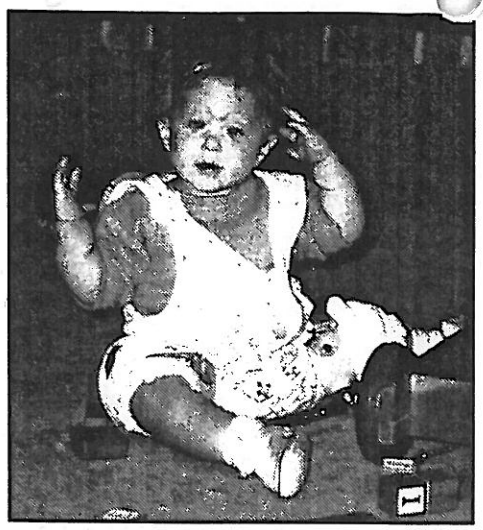
"Rebecca's birth was carefully monitored and managed, and she emerged appearing healthy and whole to this untrained eye. But an experienced hand noticed immediately that Rebecca's deformed thumbs indicated any number of serious problems.

Our family pediatrician appeared within thirty minutes of Rebecca's birth, accompanied by one of the foremost general pediatric surgeons in this part of the country. We were so grateful and relieved by this quick response.

The prenatal diagnosis was confirmed. Otherwise all of her organs and vital signs were normal. Rebecca's APGAR scores were very good. However, x-rays showed that many of her organs were displaced from their expected location. It was, as one doctor put it, "as if they had made a milk shake of her insides". With no completed digestive tract, she could not be nursed or fed orally. She was maintained in a neonatal isolette connected to various IVs and catheters throughout the night before her transfer to another hospital for surgery.

It is bitter beyond imagination to watch other newborns held and cuddled by happy parents while you can only stroke and talk to yours in her isolette. It is nearly devastating to pack up your little overnight bag and walk out of a maternity ward the following day with only the cards and gifts of well-wishers, but no baby to carry home.

This sorrow is relieved only by a happy ending. We had that, but it was a long time coming. Rebecca's digestive tract surgery was successful. However, a day or so after surgery, her kidney function began its decline. This was the beginning of the blackest period in our lives. We were told



Rebecca Frank

there was only one kidney located in the pelvic region, that it was small and might not be adequate to maintain our daughter. The doctors could not predict whether the kidney would grow with Rebecca or ever recover its function. Transplant was not an alternative in a newborn, less than seven pounds in weight, even if a donor were available. Dialysis was not a promising option for such a small, fragile child.

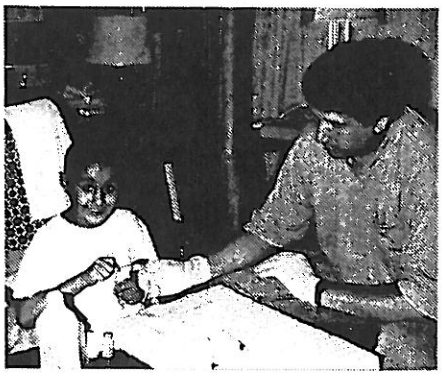
This was the period wherein we thought our child would die. We were told to prepare for that event. She didn't. We were so infinitely grateful for a second chance. It

Family Rejoices Following Successful Transplant

In 1992, **Hernaldo Hernandez**, age 8, had a successful bone marrow transplant at Children's Hospital, Cincinnati. His father, **Hernan**, writes of the family's gratitude and joy:

"After a long search in our community, the Cathedral of Saint Thomas More, Arlington, VA decided to back up our effort to raise money for the transplant. In a couple of weeks, both the American and the Hispanic communities shared a common goal for the first time: 'save the life of an FA kid'. The commitment was successful. A year later we feel we are a blessed family. Our Hernaldo is doing reasonably well after the BMT, and all those memories will be unforgettable. They have meant a deep change in our lives as a family, as individuals and as good people."

Hernan wanted to thank the administrators, doctors, nurses and other workers at Children's Hospital for making his family's dream come true. He gives special credit to **Dr. Richard Harris**, *"a good person and bright researcher"*.



Hernaldo and Hernan Hernandez

forged our resolve to deal with any event, any outcome, any hardship, so long as we could have our baby.

I try to hide my rueful amusement at the compliments we receive from friends and family for our "courage", our "strength" in "what (we) are going through" with Rebecca. Surely they are well intentioned. But we have no sense of the slightest hardship or deprivation, or any consciousness of special strength or courage. We're so happy we still have her and that she's doing well that nothing required of us seems like a sacrifice, only a pleasure, in view of the goal.

Rebecca's kidney problems were successfully treated surgically. The defect was diagnosed as caused by a deformed connection of the ureter to the kidney, causing a "water pump" effect, a backup that stressed the kidney and impaired its function. The angle at which the ureter was implanted impeded drainage. This is apparently not an uncommon FA associated birth defect, as are the defective kidney placement and function.

Rebecca has the one kidney, the deformed, now corrected, ureter implant, and a second blind ureter, dead-ended for failure of the second kidney to form and provide it with a destination. Her kidney function will never be normal in medical terms, but her function is more than adequate, for now, for her to lead a normal life. She has taken no special medication since her fifth month and is currently under no diet restrictions. As precaution, however, we maintain her on a commercially available special formula recommended for infants with kidney problems. Check-ups with the nephrologist are less frequent, an indication of her stability.

I am interested in hearing about other experiences regarding kidney problems in FA patients, particularly children. I am moderately concerned about the pelvic position of one kidney. Ordinarily positioned in the back, is Rebecca more prone to have her kidney injured in the ordinary rough and tumble of child play and adolescent athletics? Should she have special protection or restrictions? Taking a long range optimistic view, will this impact on her child bearing due to the position, and complicate the risks thereof beyond the sufficiently serious risk of pre-natal stress and kidney dysfunction?"

Neil notes that Rebecca's hip dislocations were not detected until the age of six months:

"Rebecca spent the first five weeks of her life in the neonatal care unit at Presbyterian Babies Hospital in New York. Although she was x-rayed, CAT scanned, probed and tested in every imaginable fashion, hip dislocation was never diagnosed or even mentioned during this period.

The issue was first raised by our family pediatrician, who heard faint "clicks" on manipulating Rebecca's legs, at about six months of age. This discovery was sufficient to refer us to a pediatric orthopedist immediately.

For three months thereafter, Rebecca wore a Pavlik harness, a common treatment for congenitally displaced hips in infants. This treatment was successful in correcting the dislocation of the right hip, but failed to correct the left one. The congenital deformity of the socket, directly related to FA, required surgical correction.

Several physicians and friends speculated that if a diagnosis had been made at the earliest stage, i.e., as a neonate, and the Pavlik harness applied at that time, both hips might have been corrected without surgery.

No conclusive evidence supports this hypothesis. However, the lesson for other parents of FA children where diagnosis is made soon after birth, is to be very sensitive to this deformity. It tends to be relegated to a subordinate status inasmuch as it is not life-threatening. However, diagnosis and treatment of the hip deformity at the earliest possible stage would more likely result in successful non-surgical correction.

Our pediatric orthopedic surgeon had previous experience performing hip surgery on Fanconi children. One important lesson I have learned as the parent of a child with a rare disease, is that the most impressive medical credentials are irrelevant compared to actual experience in treating patients who suffer from the disease. Our surgeon had both, and is a warm, compassionate, good-humored person to boot.

I emphasize the importance of a good family general practice pediatrician. Specialists specialize, and take a narrow view from the point of their assigned task. But FA children have all of the ordinary medical problems of newborns and infants that other children have, in addition to their special needs. At least one knowledgeable, concerned professional must be responsible for looking at the big picture in planning for any child's medical needs, albeit in consultation with the specialists.

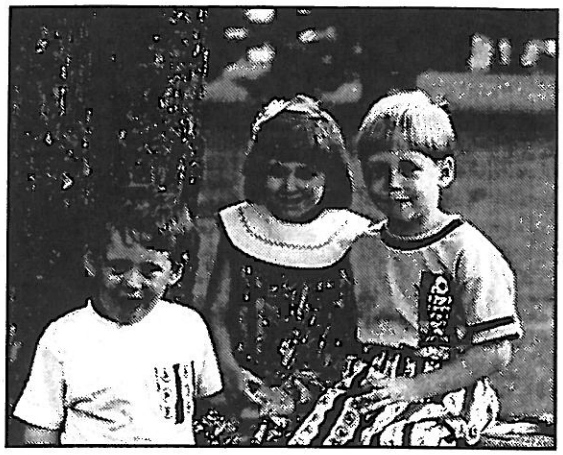
Our family pediatricians had never encountered an FA child. But both took up the challenge of caring for Rebecca with dedication and zeal. They gave us the confidence of knowing that there are professional people who consider Rebecca's welfare on a broad personal plan, rather than an impersonal challenge to their medical skills.

Rebecca now has licked her digestive-tract deformities, her hip deformities and her urologic kidney problems. Intellectually and neurologically she appears unaffected. She presently has no hematologic symptoms. She is so far unimpeded in her play by her floating thumbs. We count our blessings. If we had to endure a million more times the anxiety, depression, disruption, effort and expense, we should have

Continued on Page 10 (See Rebecca)

Sandy Weiner wrote a beautiful, poignant article about their son, **Avi**, which appeared in the July, 1993 edition of *Parent* Magazine. Avi's unique sense of humor, love of music, and engaging personality were the gifts he shared with those fortunate enough to have known him. Readers will be deeply moved by this lovely article.

Catelyn Duffy is a typical five year old girl through enjoying all that life has to offer her. Although she has lower than normal blood counts, this fact hasn't stopped her from being an extremely energetic young girl. Catelyn has been enjoying her summer vacation between nursery school and kindergarten with her two younger brothers, **Matthew** and **Michael**. She loves to be in and around the water and is particularly proud now that she has mastered an ability to swim underwater; she had great fun in the hotel pool at the recent family symposium, demonstrating all of her swimming techniques. The "babysitting club" in Minneapolis also gave her the opportunity to meet her old friends and establish many new friendships.

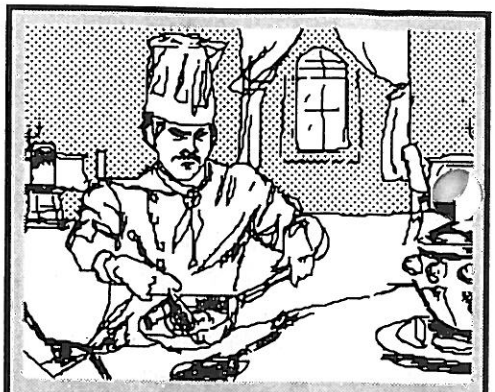


Catelyn has great empathy when she senses that other children are struggling. Her compassion for others showed itself last school year when a young Chinese girl entered class, speaking no English and clearly having difficulty integrating into a new environment. Catelyn quickly took the child under her wings, making her feel as comfortable as possible; she is always concerned about everyone being happy.

In our family Catelyn takes on the role of social director. When visitors arrive, she'll quickly organize a party or dance show. You'll always find her tapes and tape recorder close by to provide the background theme music. It's rare to hear Catelyn talk negatively about anything - she's even managed to convince herself that her blood tests don't hurt.

Four successful hand surgeries give Catelyn ample opportunities to exhibit Dr. Joe Upton's great work and explain to all that "God has made me special and loves me very much". Catelyn has great fun with her two brothers even though she does get frustrated when her overprotective parents restrict her from getting involved in the daily tumbling, tackling, and bed jumping Olympic events. However, she's always taking the time to show her less worldly brothers the finer points involved in catching frogs and fishing at the lake.

This September, our little girl will be entering kindergarten. Six years ago, we didn't even know if we'd ever arrive to this point in time. We are extremely thankful for all the joy that Catelyn has brought into our lives. We are also very grateful for the many medical advances over that time and hopeful for many more leading to an eventual cure for FA. Catelyn's insatiable lust for life demands nothing less.



Martin and Linda Sankey are busy preparing the first ever Fanconi Anemia Family Cookbook. This cookbook will be completed by October, and could be an ideal holiday gift for family and friends. Send your favorite recipes to:

Martin and Linda Sankey
1239 Woodledge Drive
Mineral Ridge, OH 44440

If you wish, you can include a picture and biographical information about your child/children. A recipe could also be dedicated to a lost loved one. Let's all support this wonderful idea!

Rebecca (*Continued from page 9*)

done so cheerfully to have come this far. That all seems so unimportant when you love a child and have hope for a happy ending."

Neil expressed his highest regard for Presbyterian Babies Hospital in New York, and for the many highly skilled physicians who gave Rebecca expert care. The Franks would be pleased to discuss their family's medical experiences with persons seeking expert medical care in the New York region.

Neil and Iris Frank can be reached at:
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Your News Needed

The single biggest criticism of this Newsletter is that it does not contain enough news from our families. Older patients would like to hear from their contemporaries, and parents want to hear how other parents are coping. Everyone likes to read about the accomplishments, successes and challenges of our children. **But to print those stories, we have to receive them from you!** So send us your pictures and your news!

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Your Help Needed

We know that many FA families are not yet aware of our support group. They could benefit from receiving our newsletter, attending our Family Meetings and meeting those of us who share the same concerns.

Could you please contact the primary medical center in your region or state? Tell hematologists, social workers and nurses about our support group. Please urge them to refer families to us. Contact Lynn Frohnmayer for a packet containing materials to share with medical professionals.

Your efforts will help break the isolation felt by other families. Please help!

FA family newsletter

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