FANCONI ANEMIA RESEARCH FUND, INC.

FAMILY NEWSLETTER

#24 A Semi-annual Newsletter on Fanconi Anemia for Families, Physicians and Research Scientists Fall, 1998

Amanda (Pennsylvania), Janelle (Canada) and Amy (Oregon)

Families Learn, Share and Relax at Annual Family Meeting

Thirty families from seven countries met at Aurora University at Lake Geneva, Wisconsin, for our 8th annual FA Family Meeting, August 25-30, 1998. This new location had ample meeting rooms, gorgeous views of Lake Geneva, wonderful food and a wide variety of outdoor activities. It provided a relaxing setting for families to share experiences and learn. Because we arrived at the end of summer, the activities staff had already departed. Local families recruited volunteers, and children enjoyed a wide range of day and evening activities.

Presenters updated families on research developments and described future treatment approaches. Readers should review the *Science Letter* for detailed information. A few highlights:

- Hans Joenje announced that his laboratory has isolated the FA-G gene. Scientists have now identified three of at least eight Fanconi anemia genes. As with FA-A and FA-C, researchers have no clear idea as to the function of the protein encoded by FA-G.
- John Wagner presented data concerning the high success rate of matched sibling donor transplants (from 75.6% to 85%). However, most patients lack matched sibling

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Joenje Laboratory Isolates New FA Gene

Hans Joenje, Ph.D. (The Netherlands) announced at the Lake Geneva FA Family Meeting that his laboratory recently has isolated the gene for complementation group G (FA-G). Joenje and associate, Johan deWinter, assisted by collaborators in the laboratories of Dr. Martin Digweed (Berlin), Dr. Holger Hoehn (Wurtzberg), and other major FA scientists will publish their FA-G gene findings in the November 1, 1998 issue of the leading scientific journal, *Nature Genetics*.

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HIGHLIGHTS

MEDICAL NEWS

Standards of Clinical Care Conference to Assist Practitioners, Patients

The Fanconi Anemia Research Fund convened a conference entitled "Toward Standards of Clinical Care for FA Patients" in Portland, Oregon from May 28-30, 1998. Inspiration and direction for this conference came from clinicians, concerned that there are no generally accepted guidelines to assist practicing physicians as they move from diagnosis to treatment of Fanconi anemia.

Nine professionals representing a wide range of specialities set forth draft guidelines which were then debated by over thirty physicians. Each major topic was discussed at length: The Diagnostic Workup; Management of Hematological Failure; Evaluation and Management of Growth Failure; Bone Marrow Transplantation–Sibling Donors; Bone Marrow Transplantation–Unrelated Donors; Longitudinal Studies; Social Work Issues; Future Therapies. An effort was made to achieve consensus. Our discussion leader, Eva Guinan, MD, kept everyone on task and skillfully guided the group toward consensus covering a wide range of topics.

This conference should generate two major publications in the near future. The first will be an FA "Clinical Standards Guide," published by the FA Research Fund. We will distribute this guide by mail and through our website. We will ask for physician feedback and update the guide on a regular basis. The second publication will be a major article co-authored by caucus section leaders and submitted to a leading clinical journal for publication.

Many pediatricians, family doctors and even hematologists have little or no experience in treating this disorder. This guide should be of great assistance to physicians as they work with FA families to provide the best therapeutic intervention currently available. ◆

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.

Work Closely with Your Dentist: The Need for Aggressive Diagnosis with FA Patients

During the last few years, several older patients in our support group have developed cancer of the mouth. Some are regular attendees at Family Meetings and personally well known to many of us. We hope and pray for good outcomes as they undergo therapy to address this worrisome complication.

Elise Bolski, DDS and spouse of Dr. Wayne Rackoff, prepared an article for our *Science Letter*. (Rackoff is known to many of us for his insightful presentations at scientific meetings, knowledge of Fanconi anemia and instigation of clinical trials for FA patients). Bolski states that dentists who treat FA patients should be encouraged to become very knowledgeable about this illness and aware of their patients' increased risk of cancers of the gastrointestinal tract, including oral mucosal and tongue carcinoma. She states "**Suspicious lesions, including ulcerations, persistently swollen tissue and leukoplakia should be subjected to biopsy.**" Bolski recommends that dental check-ups should begin at the age of 18 months **and continue semi-annually.**

We strongly recommend that all families read this article. Families should give a copy of this article to any dentist who treats their FA patient. Cancer of the oral cavity, detected at a very early stage, may stand a greater chance of being cured. ◆

Annual Family Meeting

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donors, and success rates for alternative transplants remain discouragingly low (31% to 34%). Several new approaches are now being explored to improve these outcomes, including non-myeloablative therapy, megadose stem cell transplant, transplanting lymphocytes with a "suicide" gene (so that T-cells could assist engraftment but be eliminated if severe GVHD occurs), and genetic correction of stem cells.

• Christopher Walsh will begin a gene therapy trial for ten FA-A patients this fall (see *Science Letter* for details). He expressed both caution and optimism. Dr. Walsh believes his laboratory can isolate special cells (SP cells), or pluripotent stem cells. The vector used by his laboratory is 10 to 100 times more potent than the previous vector used in a gene therapy trial, and it penetrates these very early stem cells. His laboratory has succeeded in putting corrected cells into FA mice and then eliminating the remaining FA stem cells. This has resulted in curing the FA defect in mouse bone marrow. This upcoming trial will not include eliminating remaining FA bone marrow. Much work must be done before this approach can be proved efficient in human beings, but this trial is an important beginning.

• Blanche Alter presented an overview of the May conference on "Standards of Clinical Care for FA Patients" held in Portland, Oregon May 28-30 (see article, page 2). She also gave very helpful guidelines to assist patients in monitoring routinely for possible malignancies. Readers are strongly urged to read her recommendations in the *Science Letter*.

• Several presenters gave helpful information on subjects of general interest to all FA families. Susan Olson discussed diagnosis and prenatal diagnosis of FA. She described a new method which utilizes in vitro fertilization and sampling of a single cell from a 6-8 day embryo to determine the health and HLA status of the embryo. Preimplantation diagnosis is feasible for families in which a disease mutation is known. Frank Smith gave a lucid overview of stem cell therapy and gene therapy. Martin Johnston gave an outstanding overview of the process of cell division and the events which could lead to the development of myelodysplastic syndrome (MDS) or leukemia in FA patients.

The schedule for each day allowed time for family activities or small group discussions on a wide variety of topics. Nancy Cincotta led many sessions dealing with issues such as day to day coping with Fanconi anemia and bereavement issues. Nancy also led a group for FA patients and another for siblings of patients. As always, those attending her groups expressed high praise and enthusiasm for these valuable sessions.

Many physicians and researchers stayed to meet with small groups of parents concerning their special needs and questions. Nasrollah Shahidi, believed by many to have the most experience in treating this disease, met with small groups of parents. Shahidi's practical advice, extensive knowledge, warm concern and encouragement have been highly valued by all parents fortunate enough to have worked with him. ◆

Controlling Nose and Mouth Bleeding with Amicar

by Wayne Rackoff, MD, Rich Harris, MD, Jeff Lipton, MD, Blanche Alter, MD

Amicar (amino caproic acid) is a drug used to help control bleeding. It is most effective in bleeding from the mucosa of the nose and mouth. It blocks the breakdown of clots that are formed naturally in the body. It is not effective for bleeding from all sites in the body, because it works by being secreted into the lining of body cavities (e.g., it is secreted into the saliva).

This drug should only be used after consulting your hematologist. There are situations, such as bleeding in the urinary tract (kidneys/bladder), when Amicar should *not* be used. If bleeding is related to low platelets, Amicar may be helpful, but a platelet transfusion may also be needed. Amicar may be useful to *prevent* bleeding after dental procedures, but always consult your hematologist before using this drug.

Amicar may cause nausea and vomiting. It is expensive but may be stored at home for a fairly long time. You should speak to your hematologist about whether he or she recommends keeping Amicar at home. Never take more than the prescribed dose, because an excessive amount may cause harmful blood clots. ◆

FUNDRAISING

Summerville Fund Awards New FA Family Challenge Grant

The Summerville Fund of the Oregon Community Foundation (OCF) has granted a $60,000 challenge grant for FA research, but FA families must meet that challenge by December 31, 1998! Be sure to ask your donors to write "Summerville" on the memo line of their check, and thank them for helping us double their gift!

We are deeply grateful to William Swindells and the Summerville Trustees for their generous support. Their gifts, totaling $120,000, have helped to fund FA research in the laboratory of Dr. Maureen Hoatlin at Oregon Health Sciences University. The challenge grant matching funds will be available for FA research outside of Oregon. ◆

Lisa Nash Reports on Highly Successful Fundraiser

We decided to do a big fundraiser for FA this fall, and chose a silent auction. We combined our fundraiser with other events put on by the Jewish Community Festival, held at our Jewish Community Center. The Festival included a bone marrow drive to find a donor for our daughter, Molly, and anyone else needing a bone marrow transplant. It was the perfect location and timing for our fundraising effort!

Prior to the auction we contacted restaurants, shops, beauty salons, resorts, hotels and toy stores (the beanie babies were one of the most successful items). We tried to think of anyone who might be willing to donate to the silent auction. Contacting a large number of people and businesses attracted a lot of attention. We tried to stay with moderately priced items as people were more likely to bid on them. Items ranged in value from a $6.00 bottle of wine to a $500.00 gift certificate from a jewelry store.

The silent auction turned into two events with over 150 items auctioned. People really were into bidding! The hardest part came afterwards. That's when we contacted the winners, collected money and delivered items. Our auctions were on September 13 and 28, and we are still trying to get in touch with people and collect money! It was a great time and we feel that it was really profitable.

In soliciting items for the auctions, we contacted Waldenbooks, part of a nationwide chain of book stores. They do not contribute items, but they hold in-store book fairs. We worked with the manager of the store to pick a good day. He then contacted other Waldenbooks in our area and six wanted to participate in our effort. The stores then decided how long the fair would go (this can be from two hours to all day). Customers were asked if they wanted part of the cost of nonsale items to go to Fanconi anemia. Most people were willing, and 20% of those sales went to FA. Waldenbooks will let you advertise, but this must be planned in advance and approved by the company. We just made fliers and posted them at the entrance to the stores and at the cash registers.

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Meyer Memorial Trust Awards Funds for FA Regional Network Project

The FA Research Fund will be able to expand its services to patients, families, physicians and researchers thanks to a newly received grant from the Meyer Memorial Trust of Portland, Oregon. The Trust has awarded $150,000 over a three-year period to support a number of improvements in our FA Family Education and Support Network.

Our goal is to create a regional network that will allow the Fund to:

- conduct regional family meetings for family support and education;
- assist families in raising funds for FA;
- encourage sharing of information about medical resources within each region.

This grant enables our Fund to hire a Family Services Assistant to provide fundraising assistance to families and maintain a new patient family database. In order to succeed, we will need the help of families to bring staff in contact with new funding sources in each region. ◆

Family Fundraising Efforts

From **January 1, 1998** through **June 30, 1998**, seventeen families conducted special fundraisers including letter campaigns, yard sales, United Way promotions, long-distance telephone card sales and children's hop-a-thons, raising a total of $141,378. An additional forty-six families either made personal donations or had donations made in honor of their families or in memory of an FA patient, totaling $35,134. During this six-month period United Way and Combined Federal Campaign funds more than doubled, yielding $13,510 in donations for family support.

We appreciate all those who made special efforts to raise needed funds for research and family support. Raising funds may not be easy, but nothing about FA is easy. Unfortunately, the responsibility for attracting and funding prominent researchers falls ultimately on families struggling with FA. Remember that researchers are seeking a cure for you, your child or your relative. Without research there will never be a cure!

We urge all families to take part in helping us meet our future financial goals toward funding research.

We are deeply grateful to the following families who were involved in fundraising events during the first six months of 1998. Remember that funds raised since June 30 will be included in our next newsletter.

$75,000+

Dave & Lynn Frohnmayer

$10,000+

Andrew & Vicki Athens
Pat & Maria Gleason
Allen Goldberg & Laurie Strongin

$5,000+

Mark & Susan Trager

$1,000+

Peg LeRoux
Eric & Beth Losekamp
Chris Scaff
Bill & Connie Schenone
Jeff & Debby Slater
Steve & Melissa Turner

Up to $1,000

Boyd & Lisa Bourgeois
Gary & Melody Ganz
Rene LeRoux
Steve & Alison McClay
Marlene Stone & Robin Paulson
Rick & Lynn Sablosky

The following people made personal donations or had gifts donated in their honor or in memory of a family member:

Byron & Denise Adamson
Al & Janeth Acosta
Ken & Jeanne Atkinson
Cheri Bank
Mark & Linda Baumiller
Gilbert Bodier
Michael & Diane Bradley
Chris & Susan Collins
Bill & Pat Danks
Darla Patrick & Dottie Day
Donna DellaRatta
Joseph & Tracey DeMarco
Pat & Mary DiMarino
Ed & Janice Duffy
Nathan & Ann Eckstadt
Michael Greenberg
Chris Hull
Susan (DaRosa) Jackson
Jeff & Beth Janock
Leardon Keleher
John & Karilyn Kelson
Robert & Jennifer Kiesel
Jee-Ai Kim and SeJin Kwon
Barb Lawrence
Dennis & Sharon Lower
Lynette Lowrimore
Bill & Jackie Lucarell
Stuart Cohen & Deane Marchbein
James Mathieson
Jack & Pam McCarty
Cecilia Meloling
Mike & Myra Lewis
Griff & Cecilia Morgan
Sheila Muhlen
Jack & Lisa Nash
Ron & Fredi Norris
Kevin & Lorraine O'Connor
Kashar Porter
Shirley Ricker
Nancy Rising
Terry & Therese Robertson
Glen Russo
Bob & Andrea Sacks
Eric & Lori Salo
Severt & Beatric Score
Bryan & Karen Siebenthal
Dave & Mitzi Speelman
Mike & Beth Vangel
Marc & Sandi Weiner
Reese & Nancy Williams

"If FA families don't step forward to raise funds for research, who in the world will?"

~ FA mother

FAMILY NEWS

Ruthie Saunders: A Special Gift

by Sharon Saunders

There is so much to tell, nuances of good and bad, the difficulties in making decisions, the unbelievable stupidity and intelligence of people, that it is difficult just to begin. And so, I'll begin in the present, because the present is good.

My daughter Ruthie is now 17 years old and in her second year of high school in Jerusalem. She loves dogs, collecting stamps, drooling over the Back Street Boys, watching TV, creating art, playing drums and hanging out with her friends. She's pretty terrific. She has an amazing sense of humor and a very strong sense of self. Ruthie knows what's good and can easily separate the wheat from the chaff. She has what is now popularly called "emotional intelligence."

Ruthie was born in a small town in upstate New York. She was very small, with a small head, the absence of thumbs, tiny eyes and a few coffee-colored spots on her back and tummy. Because of the absence of thumbs and the attendant radials she was tested by Dr. Arleen Auerbach and the diagnosis was Fanconi anemia. Her biological parents went home and decided that for everyone concerned a new home should be found for her. Was this bad—were they wrong? I have a great daughter because of it and Ruthie is alive and well with a full life.

It's hard to make these determinations, to understand that something bad on a larger scale can also be good. Because Ruthie had the abnormalities of the hands she was diagnosed almost immediately. Because it was known that she had FA from the beginning she wasn't given many blood transfusions or androgens. Her bone marrow transplant at the age of nine went without a hitch. She had no problems with the radiation, no GVHD, no fungal problems and within 9 days her blood tests showed the signs of success.

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Celine Van Heerden

A Tough Diagnosis

by Marius and Samantha Van Heerden, South Africa

Our daughter was born on November 25, 1996. She was born with both thumbs absent. Her birth weight was 1.9 kg.

When Celine was six weeks old her doctor ordered a syndrome test (she had features of a child with a particular syndrome, e.g., almond-shaped eyes and short stature). After six weeks the test came back negative, which was a huge relief. At five months the pediatrician said that we had to do a chromosome test on Celine. The results came back and the doctor said that there was an increase in the breaking of the chromosomes. He suspected that it was Fanconi anemia. This test was repeated when she was a year old. We received the same results. Celine was diagnosed with Fanconi anemia.

We took Celine for a second opinion to a children's disease doctor in Johannesburg. His opinion was the same. We were told to have her followed by the hematology and oncology unit at the Johannesburg hospital.

Celine's blood count is still normal, although her white cells and platelet count have dropped a bit. We go every three months to have her blood checked.

Celine started walking on the 19th of February at 15 months. We are so proud of her. ◆

The Dietrich Family: A Portrait of Caring, Determination and Courage

by Ralf and Cornelia Dietrich

Hello. We are Cornelia and Ralf Dietrich from Germany. We have three daughters: Sarah Ninja (18), Elisa (16), and Valeska (13). At age five, Sarah Ninja was diagnosed with severe aplastic anemia. Her counts were: Hb 8; platelets 12,000; neutrophils between 500 and 1,000. The consequences were severe infections, nosebleeds and general weakness. At age six she required blood transfusions every 4 to 5 weeks. Cortisone was given for 3 months, but did not result in any improvement. At age seven Sarah Ninja was tested for FA, and so were her sisters. The result revealed a disaster. Not only did our oldest daughter have FA, but so also did our youngest. Only Elisa is healthy, but her bone marrow is not a match for Valeska or Sarah Ninja. Fortunately, Valeska was doing fine.

Early on, Lynn and Dave Frohnmayer provided us with important information. Since we had grave concerns regarding unrelated donor BMTs, the only option left for Sarah Ninja was oxymetholone. After 3 months of oxymetholone, Sarah Ninja's neutrophil and platelet counts improved, but the red blood count did not show any improvement. However, after 12 months of oxymetholone, we were able to suspend blood transfusions for more than a year. A chicken pox infection, however, wiped out the progress we had made. We raised her dose temporarily to 3 mg per kilo of body weight. The blood counts increased, but she developed liver adenomas (tumors). Over a period of many months we tapered the dose down to .5 mg per kilo body weight, and the liver adenomas shrank by fifty percent. Unfortunately, with the lower oxymetholone dose the blood counts decreased again.

Sarah Ninja has been receiving blood transfusions with red blood cells for 12 years, with an interruption only of 1 year. During the last three years she has had transfusions every two weeks. All together, she has probably had more than 170 red blood cell transfusions. She is taking Desferal regularly to remove excess iron from her body. In addition she

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In Loving Memory

Jamie Alexander
6/26/85 – 5/18/98

Linda Burton
7/24/65 – 9/9/98

Jennifer Hyland
10/27/83 – 9/22/98

Derek (DaRosa) Jackson
8/2/85 – 8/26/98

Dirk Juergens
11/2/65 – 8/6/98

Hiroshi Kawasaki
7/7/94 – 4/6/98

Mary Katherine Keleher
3/27/69 – 6/18/98

David Kwon
3/4/90 – 3/23/98

Aaron Moskowitz
12/7/88 – 3/11/98

Jacob Wedlake
7/21/95 – 6/26/98

We Welcome New Families Who Have Joined Our Support Group

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Orleans, Ontario
Canada K1C 3E9
(613) 841-8092
Christina ~ DOB 6/30/98

Ezat Faizyar
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Pleasanton, CA 94566
(925) 461-8491
Yasameen ~ DOB 3/11/94

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DOB 12/11/69

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Michelle Lee ~ DOB 1/2/96

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011 54 64 215124 Fax
Constanza ~ DOB 1/23/89
Gustavo ~ DOB 8/13/85

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Oakville, WA 98568
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Dacoda (Cody) ~ DOB 5/10/96

Joan & Ken Peters
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Meghan ~ DOB 7/30/93

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Celine ~ DOB 11/25/96

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(706) 317-5730

Ruthie Saunders

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Because her biological parents were first cousins, Ruthie had the autosomal recessive genes that caused her to have FA, but she also had two siblings who were HLA matches for her. Disaster on one hand, and unbelievable luck on the other.

Ruthie was born with just about everything in the way of abnormalities an FA kid can have: microcephaly, strabismis, the missing thumbs and radials, short stature, and tetralogy of the fallop (a serious heart problem). When she was little she couldn't have a heart operation because she was underweight. Then the blood problems reared their ugly head after a bout with both chicken pox and scarlet fever (at the same time). The operation was set to happen when she was 10, after her bone marrow transplant, but I hastily cancelled it because the doctors couldn't guarantee me any large measure of success. We decided to wait.

Intuition is a great thing when dealing with all the baffling components of FA decision making. When Ruth was 14, a very prominent Israeli doctor returned from the States to work at the children's hospital in Petach Tikvah and he did a beautiful job of correcting her heart.

If there is any real advice I can give parents of FA children it is to trust yourself and never be ambivalent. Listen to those warning bells inside you. You know how to differentiate a caring doctor with your best interests at heart from a doctor whose main concern is using your child as a statistic for publication.

One quick story illustrates this point. I once took Ruthie to the clinic for a blood workup. The pediatric hematologist leaned over and wanted to know if he could ask me a personal question. "Why," he asked, "are you trying so hard to save the life of a child with multiple handicaps?" He pointed out that, after all, I had no insurance to do the transplant overseas and that Dr. Shimon Slavin of Hadassah Hospital advised me not to do the transplant in his unit but to go to Hôpital St. Louis in Paris and have it done with Prof. Eliane Gluckman. I had to raise $120,000 for this to happen. "Why don't you just let nature take its course?" he asked. "She won't thank you for being alive when she's 18. I know what I'm talking about. I have an 18-year-old daughter who is legally blind and she is miserable." I exploded and called him a Nazi, among other things, and I often wonder what would have happened if Ruthie's biological parents had kept her and had gone to this doctor?

Ruthie is a success story. At 17 she is alive, vital, and considering that she is a teenager, pretty darn happy. Attitude is crucial. There have been tremendous obstacles along the way, lots of difficulties, but throughout it all there has been the sense that somehow there is a reason for all this, that there is a greater plan at work. I don't purport to understand it, I only know that the world is an imperfect place, and that as human beings our greatest challenge is to perfect it.

Hadasseh Hospital in Jerusalem has a list of potential bone marrow donors. If a family will send or fax me the HLA typing of their child, I will put it through the computer. My fax is: 011 972 2 675 7217.

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.

Joenje Isolates New FA Gene

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This is the second FA gene discovery attributed to the outstanding researchers associated with the Joenje laboratory. In 1996, Hans, together with European and North American collaborators, cloned the gene for FA-A.

The gene now known as FA-G was identified previously as a gene that corrects the mutant form of an experimental Chinese hamster cell, when in the presence of certain cross-linking chemicals. The function of the FA-G protein is still unknown. But this discovery provides an additional piece of the puzzle that needs to be solved in order to understand what, precisely, goes wrong in FA cells.

So far, pathogenic (disease-causing) mutations in the FA-G gene have been found in four FA families. Patients previously excluded by genetic testing from FA-A and FA-C may now potentially be tested for group FA-G, on the basis of mutation screening for this newly identified group. ◆

Dietrich Family

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has received platelet transfusions for 3 years, and now has had a total of over 250. At first, she had the transfusions every 9 days, but now the platelets last only 3 or 4 days, in spite of the fact that she is receiving platelets only from the same HLA-matched donors. She has taken G-CSF for three years since her white blood cell count dropped to dangerously low levels.

As a result of this treatment regimen she is relatively stable. We haven't had a life-threatening crisis in the last 3 years. No severe nose and gum bleeding, no internal bleeding, and no abscesses occur any more. Although Sarah Ninja's bone marrow is no longer capable of producing measurable quantities of red blood cells, platelets or white blood cells, our life has become a bit more relaxed as a result of the transfusion regimen and the G-CSF.

Of course, many fears and profound questions remain: How long can her body tolerate the high iron count (in spite of the Desferal)? Will she require red blood cell and platelet transfusions even more frequently? Will the blood bank be able to find suitable HLA-matched donors of platelets? Will she be able to cope psychologically if later on she needs transfusions every two days, or every day? In what direction are the abnormal clones in her bone marrow developing? Could thrombopoietin help improve her platelet count, once it becomes available? Can the G-CSF protect her against cancer or leukemia because of her elevated white blood cell counts? Or will G-CSF promote the proliferation of leukemia cells? How long can her liver tolerate the many transfusions and the G-CSF? She has had liver adenomas for 8 years. How will they affect her in the future? Is there a growing risk that they will become malignant? We have been able to keep Sarah Ninja alive in spite of the poor prognosis given by her physicians. But we also ask, will our treatment regimen reduce her chances to benefit from future progress in the area of bone marrow transplants or gene therapy?

At the moment, we are less worried about Valeska's condition. She has been stable taking low doses of oxymetholone for 5 years. She is now 13, and hasn't required one single transfusion.

We learned how to live our life without making plans for the future. We treasure every day we are allowed to spend together. We give a lot of time and energy to our German version of the FA Research Fund.

We'd be delighted to respond to families who call or write us with questions.

Sarah Ninja Dietrich

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Editors' note: *The Dietrichs' personal struggle tells only one part of their story with Fanconi anemia. The rest requires a chapter, but a short summary will have to do. Ralf and Cornelia have organized the German support group. They have raised funds, tirelessly, for FA research. Ralf has worked very closely with Hans Joenje from The Netherlands to coordinate the collection of blood samples for complementation and mosaicism studies. He provides support to countless families. By recording endless cassettes of video tapes, he has chronicled the histories of FA families, in order to help others learn. Ralf and Cornelia have devoted their lives to a search for better therapies and eventually a cure for FA.*

Ralf acknowledges that the daily demands of this illness with its many crises have absorbed much of his family's energies. He wants readers to know that in spite of the limitations FA has placed on the life of Sarah Ninja, she has maintained a quality of life which she values immensely. She attends school when possible. She is extremely artistic; she makes and sells a wide range of beautiful articles including scarves and jewelry which bring joy to many. The Dietrichs truly treasure each day, in spite of the worries and fears FA has brought to their family. Beyond that, they have contributed beyond measure to the well-being of all families afflicted with FA. ◆

Keeping Count

by Carol Siniawski

Tracking Blood Counts

We have been keeping track of Jake's CBCs (complete blood counts) since he was diagnosed over four years ago. In hopes of helping other families through this journey, I would like to share details of our chart, so that readers can decide if it would help to track these data. The chart may need to be modified, depending upon each patient's situation.

Along the top of the chart I note the date of the blood draw and Jake's general health. Head colds, stomach viruses, time needed to stop a nose bleed are all noted. The latter was helpful when we started seeing an Ear, Nose and Throat doctor about the nose bleeds. We could see the lessened severity and frequency of his nose bleeds from one winter to the next.

The next columns are marked as follows: platelets (needed to stop bleeding); hemoglobin (which affects energy level); and reticulocytes (percentage of immature red blood cells). When Jake's hemoglobin drops he gets tired. An elevated reticulocyte count might indicate that his body is trying to produce more red blood cells. In the next column I note his white blood count, neutrophils (the combination of segs and bands which is expressed as a percentage) and his absolute neutrophil count (ANC). The latter is obtained by taking the percentage of cells that are neutrophils and multiplying this number times the total number of white cells. The ANC can be very useful in determining if he has a sufficient number of neutrophils to fight off most infections. The next column is the MCV, which is the size of the red blood cells. An increase in this number can indicate that the bone marrow is under increased pressure to produce cells. This number was useful to us when Jake was receiving iron supplements. We would watch that count, as well as his hemoglobin and reticulocyte counts, to help us decide when or if we could lower his iron dosage.

The next column is for red blood cell transfusions, followed by a column for platelet transfusions. By scanning the sheet quickly, I can tell how many transfusions he has had and when he had them. The last column was used for making notes on when he started medications, when he stopped, or when the dosage changed.

Those are column titles that you see along the top of the page. Below each column title, I took a few rows and made some detailed notes. I wrote in each column what each identified cell did for the body, like fight infection or stop bleeding. Then I talked with Jake's doctor and got a feel for what kind of counts were acceptable for FA kids. I wrote down what the doctor believed would be a "watchout" level: the lowest possible count before we intervened medically by starting a new medication or getting a transfusion.

I called the last row "test accuracy." I want to know when to react to a small change and when not to. For instance, a change in platelets of 1000 can mean very little. The test for platelet counts is not always precise, and can vary within ranges. I need to know how sensitive the testing is. I need to know when to let the change in numbers concern me and when not. That discussion with the doctor was very helpful. I recommend this type of discussion for anyone keeping charts.

If you'd be interested in seeing our chart, I'd be happy to share it. Should you decide to keep a chart, please contact your doctor and review it with him/her, because the situation may vary for each patient. A review with your doctor would insure you have the same interpretation of the numbers and their meanings. Also, consider having the doctor provide you with all past CBCs, so you can see the long-term trends. Our doctor pulled all of Jake's counts, even those done before his FA diagnosis. Watching trends is very useful. For instance, it was helpful for me to see that Jake's counts dropped due to some kind of stomach virus, but would bounce back up when he got better. I learned not to get upset by one CBC. I look for trends, and become concerned when the counts drop and stay low. Now I wait for three CBCs before I admit that his counts are low and are probably staying down. The data are helpful in discussions about Jake's response to his medication.

Maintaining a Medical Binder

When you are seeing multiple doctors, it is helpful to keep a binder. In the front of my binder I keep a log of dates, which includes every doctor visit (include name and specialty), dates of tests performed, when medications (including dosage) were started, changed or terminated. This is basically a log of dates indicating when all major events occurred.

I also keep a mini glossary here including an enlargement of the diagram of hematopoiesis found on page 111 of our *Fanconi Anemia Handbook*. I keep the blood count chart (above) in this front section.

Next, I include as many sections as the number of doctors Jake is see-

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FAMILY MEETING AT LAKE GENEVA

The following are comments from families who participated in the family meeting held for the first time this year at Lake Geneva, Wisconsin.

Leslie Roy, Family Support Coordinator with South Africans Colin, Charles, Dawn and Monique

"Enriching, enlightening, critically significant."

Concerning the support groups: "Good bonding, sharing, informational and directional."

Emily (Colorado) and A.J. (New York) share their talents.

"We enjoyed the family free time (especially renting boats)…. The adult dinner and karaoke was great. Thank you for arranging separate group talk time for the FA kids and their siblings. This is the only place in the world they could do this! Thanks!!!"

"The ability to attract the top professionals in their various disciplines and present them in both a seminar and informal Q/A session is both essential and impressive. We really appreciate this opportunity and are continually thankful for it. Of course, an added benefit is sharing with the other families. For a first attempt—the Children's Program was done well, but can't compare to the structure of Camp Sunshine."

"Thanks for a wonderful conference and quality family time as well as allowing FA friends sharing time! Very Helpful. Very educational. Thanks again."

"I like the fact that we had family time this year (more time). We had a wonderful time here. A great learning experience. Linda, good luck in your endeavors!! Leslie, thank-you so much for all your support!!"

Canadians Pierre, mother Michelle and sister Sylvie

Keeping Count

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ing. These can be color-coded (red for hematologist, yellow for urologist, white for orthopedist, etc.). Behind each tab I have all the details related to that specific doctor. I include a list of my questions and the answer given, detailed notes of consultations, test results with dates. This single binder contains all of the medical information on your child. As I consult with one doctor, I can quickly reference another doctor's recommendation, and be prepared to answer questions regarding any aspect of Jake's treatment. I found this very helpful once I decided to be a very involved parent and I started to take responsibility for medical decisions being made. I felt empowered to ask more questions, and to seek clarity on those things that were fuzzy to me.

I hope some of this has been helpful to you. Please let me know if you have discovered other ideas that were helpful to you in tracking your child's medical situation.

Editors' Note: *Some of our children take many different medications and the dosages and specific drugs can change very often. One doctor will not always know what another specialist has prescribed. At the beginning of almost every doctor visit, the nurse or physician will ask "What medication is he/she on?" Going by memory is sometimes impossible! We got in the habit of regularly updating medications on the computer, and handing medical personnel this detailed list at each visit. In addition, we prepared and regularly updated a detailed summary of our child's medical history, which was useful every time we saw a new physician, physical therapist, etc.* ◆

Big Boys Don't Cry

On the Outside That Is

By Robert Naseff, Ph.D

Ted had been talking freely about himself and his child, and then he stopped and looked up at the ceiling. The ten other men in the room, seated in a circle along with me as the leader, all waited patiently and curiously for him to continue. We had gathered to discuss the challenges for fathers of children with disabilities. Before long, the waiting became uneasy, so I asked if there was anything else he wanted to share. Still looking at the ceiling, he answered hesitantly, "There's so much I want to say, but if I say any more, I'll cry… and I don't think I'll be able to stop."

It became obvious that he was looking up in order to keep the tears in his eyes from overflowing. As he slowly lowered his head and faced the other men, a tear rolled slowly down his left cheek. What an awkward but tender expression of male emotion. The man who was sitting on Ted's right reached over and put his arm around his comrade. This incident was the catalyst for the other men to open up, and many did so with tears in their eyes and deep feeling in their voices.

One man's reluctant openness released the other men from the taboo against expressing their depth of feeling. Is it because we have held it in so long that men believe that if we cry the tears won't stop? For as we approached school age, most of us were taught that "big boys don't cry." To enforce that social command, those who couldn't hold back the tears had to endure the humiliation of being called a "girl" or a "sissy" or a "fairy." But where do the tears go? Perhaps it is as my own father once observed to me that we men just cry on the inside.

My own story is similar to those of the men in the room, for I too have a child with a disability. Nineteen years ago, swept away by the electricity of the moment, my heart pounded with excitement as I held my newborn son's soft delicate body next to my heart. He was all I had dreamed he would be as our eyes met and locked onto each other for the first time. Visions of playing baseball and building model airplanes together and having a warm, close relationship danced in my mind's eye.

Tariq's life flowed through the first eighteen months of his life as he rolled over, raised his head, began creeping, then crawling, cruising, triumphantly walking, and then talking. Then he got an ear infection, and the train went off the track. That exciting time when every day seemed to bring a new accomplishment was gone. He stopped talking, stopped playing normally, and began flapping his arms in a strange repetitive manner. His life and mine have never been the same.

Eventually, after years of early intervention, my boy was diagnosed with autism and mental retardation. He never spoke again and never learned to read or write. Now eighteen, he's still extremely active and doesn't understand danger. It was confusing and bewildering not knowing which end was up—feeling so badly and yet having an adorable child with a serious "problem." A grief beyond words, but there was no death and a totally normal looking child whom I have loved as much as life itself.

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Big Boys Don't Cry

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I thought I would change him, and make him the boy I wanted him to be. But he has changed me, and helped me to become the man I needed to be. He taught me the meaning of unconditional love—to honor his sacred right to be loved for who he is, not what he has achieved lately, how he looks, or how much money he will earn. What a priceless lesson that he has taught me in his silence, without words—like a Buddha.

The "New Man"

Until the 1970's the role of the father in child development was largely ignored. While regarded as providers and protectors, fathers were not expected to be involved in day-to-day parenting activities, with the notable exception of discipline. Who, for example, doesn't remember hearing "Wait until your father gets home?" In emphasizing the undeniable importance of mothers, social scientists lost sight of the father and the larger family context in which children grow and develop. The word "parent" became synonymous with "mother". This same trend applied to fathers of children with disabilities.

By the time fathers were "rediscovered," many men were frustrated with their traditional roles. Many had found that the "duty" to be a successful breadwinner had sometimes choked the natural instinct to nurture, and that they could instead be tender and nurturing with their children and provide discipline too. And, as more and more women worked outside the home, fathers became of necessity more involved in the day-to-day care of their children. Michael Lamb, Ph.D., a leading scholar on fathers, revealed some significant differences between mothers' and fathers' behaviors with their newborn infants. Mothers spend more time attending to the infants' basic needs while fathers tend to play more. Fathers are also observed to be more vigorous and rougher in their play than mothers.

Mothers and fathers initially respond differently to a child with a disability. Fathers seem less emotional and focus traditionally more on long-term problems such as the financial burden. Mothers respond more openly with their emotions and are more concerned with the burdens of the daily care of the child. Fathers who are less involved in daily interaction with their children tend to have a prolonged period of denial about the disability and its implications. The growing literature about men tells us that men express their feelings differently and tend to avoid the direct expression of feelings other than anger or frustration.

We contemporary fathers don't fit the image of the perfect dad as Robert Young portrayed in "Father Knows Best." On the other hand, we are far from the idiocy that is depicted in the popular current image of Homer Simpson. Our struggle is to continue the traditional role while taking on new responsibilities that we have not been prepared for. *In fact, we are creating new images of fatherhood for our children to improve upon.*

Connecting Through Sorrow

What are men expected to do in the face of loss? Keep the lid on emotions, take charge of practical details, support others, and take on the loss as a challenge of traditional masculinity are all part of the script. On the other hand, men are *not* expected to lose control over their emotions, to openly cry, to worry, or to express overwhelming sadness. In caring for one's special child, a father can find himself more cheerful when actively involved with his wife in helping to meet their child's needs and wants. Standing by and watching a child's struggle and a wife's pain is more heartbreaking than pitching in and helping with the work. This sort of involvement can break the irresistible pull of grief. Many men find it hard to talk openly about their feelings with their wives because they think they are expected "to be strong." Whether this is true or not, men tend to believe so.

Each must find his own way through the grief. For men, perhaps the greatest frustration is that they cannot fix everything and make it better for their wives or their children. In this respect, too, love is not enough. The pain that accompanies this realization makes you go places you never planned to go. There is an immediate need to connect despite our clumsy attempts at sharing and intimacy. It takes time to acknowledge feelings and let them be heard. Even though fathers initially see themselves primarily as support people to their wives and children, they are able to acknowledge their own needs once they get together with each other. Breaking through this wall of their own emotions helps fathers to work through and complete their own grief. Here are a few helpful hints for men in this process:

1. Find other men to share thoughts and feelings with. The National Fathers' Network at www.fathersnetwork.org is a great place to start. There are many essays by

fathers there who are going through what you are experiencing.

2. Keep a journal of your thoughts, feelings, and experiences. Sometimes our innermost feelings come out when we write.
3. Think about what you would feel if you weren't angry, grumpy, or irritable. It could be something more tender.
4. Realize that when a woman wants to talk about a problem, she doesn't always want you to fix it.

Sometimes it seems that men grieve on the inside, so that women can grieve on the outside. Here are some hints gleaned from my years of experience working with fathers about what men want from their partners:

1. Remind your partner that you don't want or need him to fix everything, merely to listen and show his caring is often enough.
2. Tell your partner what he is doing right which helps him feel valued and secure in the marriage.
3. When possible, ask for help in finding solutions—even if you think you already have one. When this is the goal, it is easier for a man to listen and not be overwhelmed by a woman's emotions.
4. Plan time together as a couple.

Conclusion

It's obvious that men have a different tone of voice than women. What's not so obvious, but equally true, is that men have a different tone of grieving and feeling. We are not defective, but we are different. Rich and Debbie Auerbach of Philadelphia have a charming four-year-old son, Adam, who has autism. Accepting and coping with their very active child has been trying for this couple, particularly because Rich himself has cerebral palsy. At a recent support group for parents, Rich shared his insight that Adam and other children with disabilities, and he himself "are not the children of a lesser God."

Indeed with their gift for teaching their families and the world about unconditional love, they may be more like guardian angels.

Robert Naseef's story of his journey with Tariq and his work with families of children with special needs is told in his book, *Special Children, Challenged Parents: The Struggles and Rewards of Raising a Child With a Disability*. This book is available through bookstores, at www.amazon.com and through Carol Publishing at 1-800-447-BOOK (2665). ◆

Pat Gleason (PA), Vicki Athens (MI), Lorraine O'Connor (NY), Mary DiMarino (NY) doing Karaoke on Parents Night Out at Lake Geneva.

Nash Fundraiser

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After the book fair is over, it takes a couple of weeks to tally everything. They will then send a check directly to the FA fund. We are waiting to learn the amount we raised through the book fair. **This was probably the easiest way to do a fundraiser. We let all of our family and friends know and they all went shopping on that day. Waldenbooks will do the book fair once a year for each charity if you keep in contact with them.**

If anyone has any questions, please contact us and we would be more than willing to help you in any way.

Lisa & Jack Nash
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Our heartiest congratulations to the Nash family for their very creative, successful efforts!! ◆

New Director Joins FA Research Fund Staff

Months ago, Linda DeSpain informed the Frohnmayers that she planned to resign as Executive Director of FARF before the end of the year. After five years of guiding our organization, she wanted to return to university, obtain an advanced degree, and re-enter the teaching profession.

We always knew that Linda would not be with us forever. When first hired, she promised to give us two years of her time. The added three were a wonderful bonus. Linda has excelled in so many aspects of this very demanding job. She has developed strong relationships with the scientific community, has steadily increased our funding base and has earned the affection and trust of our FA families. She has managed a complex, multi-faceted office, learned the very cutting edge of FA science, brought us into the world of cyberspace, and arranged outstanding family and scientific conferences. During her tenure we have witnessed the rapid expansion of scientific knowledge concerning our rare disorder. We will miss Linda's warmth, her common sense, her dedication to our mission and her cheerful "can-do" attitude. With much sadness at our loss, we wish her well as she pursues her goals.

Our search for Linda's replacement led us to Joachim Schulz. Joachim is from Germany, yet speaks and writes fluent English. He has a quick sense of humor, a self-deprecating wit, and a warm, caring personality. Joachim was attracted to this position because he truly sees the chance to make a tremendous difference in the lives of children. In his words this is not just "a job—it's a calling." In response to incidents of child abuse in his local community,

Joachim Schulz,
New executive director of FA Research Fund

Joachim led a community-wide crusade to establish a program for the prevention of child abuse. He was the leading force in creating an entire organization, from building a dynamic board of directors to hiring competent staff. He obtained federal and state grants as well as funds through private donations. And he did all of this on a volunteer basis.

Joachim brings to this position a diverse set of skills and experience, most notably in the areas of fundraising, business management, publishing and community organization. He impressed our search committee as extremely intelligent, inquisitive, creative, and energetic. We have every confidence that our organization will flourish under his management.

Linda and Joachim are job-sharing until Linda's departure on November 1. The transition period has been a wonderful masterpiece of cooperation. In your own ways, please convey your heartfelt thanks to Linda and your enthusiastic welcome to Joachim. ◆

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