

CHARGE Accounts



Spring 2005

A Quarterly Newsletter for Families and Friends

Vol. 15 No. 1

CHARGE Syndrome:
CHARGE has four
major features -
Coloboma, Choanal atresia,
Cranial nerve abnormalities, and
Characteristic ears
More information on website

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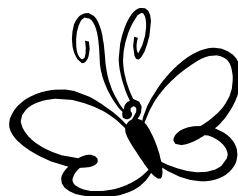
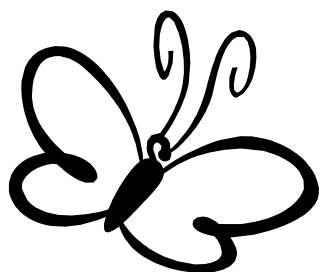
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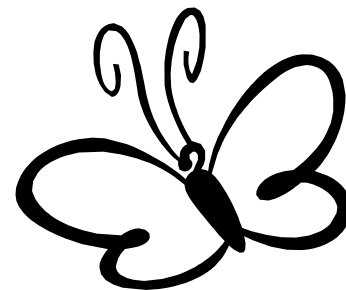
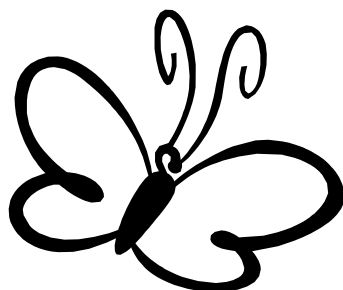
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2005 Calendar

- June 15** Conference Registration Deadline
June 27 Hotel Registration Deadline
July 22-24 7th International CHARGE Syndrome
Conference, Miami Beach, Florida



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YOUR HELP IS NEEDED!

Please share your stories, parent tips, questions, book reviews and suggestions for other features you would like to see included.
Remember this is your newsletter!

Our Experience with DNA Testing

by Michael Schwartz in Ventura, CA, father of 7-year-old, Danny (CHARGE)

PERSONAL ACCOUNTS

I recently received the DNA results from the Netherlands for my son Daniel. The Clinical Genetics Center Nijmegen (KGCN) found that he has a mutation on the CHD7 "CHARGE gene." This is the lab that discovered the gene, as reported in the September 2004 issue of Nature Genetics (vol. 36, no. 9, pp. 955-957). The full text of the article is available at www.nature.com/naturegenetics for US \$30 .

I found it interesting and almost fun to figure out the logistics to have Danny's blood shipped from the United States to the Netherlands to be tested. In case any of you plan to do it, here is how I did it. *Editor's note: Genetics clinics frequently do DNA testing at outside and out of the country labs. Your geneticist or genetic counselor can usually arrange all of the logistics, fill out all the forms, and communicate the results to you in a timely, understandable manner.*

1. If you have health insurance, call the company and see what you need to do to get them to pay. The cost at KCGN is 650 euro (approximately US \$840 at current exchange rates). My insurance company has not paid and probably won't.

2. Get a copy of the Request Form for DNA Analysis CHARGE Syndrome from KGCN. E-mail dnadiagn@antrg.umcn.nl and they can e-mail you the form. The form has instructions about what sort of tubes to put the blood in, where to send it, etc.

3. Get a prescription or order from the doctor to have the blood drawn and sent to KGCN to be tested for DNA analysis for CHARGE. I brought Danny's doctor the Nature Genetics article so he would know what I was talking about.

4. Get on the phone and figure out who can draw the blood, who can ship it to the Netherlands, how the shipping bill can be paid, etc. Since KGCN wants the blood within 4 days of being drawn, you want to get the logistics figured out in advance. One lab I called did not have an account with a shipping company but also would not release the samples to me to ship "because I might drop them." The County Hospital finally agreed to draw the blood for free and gave the vials to me (with some packaging material they had) to ship.

5. Fill out the KGCN request form and shipping forms. I used DHL/Airbourne Express, which sent me in advance their paperwork to fill out and their special "Lab Pack" envelopes. They also gave me a sample of the "commercial invoice" I had to prepare. I described the "goods" as one lab pack containing 4 vials of blood for DNA testing, not hazardous, not infectious, with an arbitrary total value I assigned of \$10. I also collected a couple of small boxes to use in packaging the vials.

6. Get the blood drawn and shipped to KGCN. I did it on a Monday morning to coincide with the shipping company's schedule.

7. Make sure the blood gets there. I kept tracking my package on the shipping company's web site. Why is it still in Oxnard some 31 hours after I delivered it there? Eventually, it's in Los Angeles. Now it's in New York! Then, magically, in Brussels faster than the laws of physics could get it there. You know the poem, "Welcome to Holland"? I kept hoping the blood samples wouldn't end up in Italy! Finally, somebody in Nijmegen signed for it. I keep bugging the lab with e-mails. The University wasn't closed for some Dutch holiday, was it? No, the return message assured me in English, the package was received and was in good condition.

8. Wait for the results. The KGCN form says 3-6 months. My results were done in two months but were sent to my doctor, who didn't tell me about them until I asked him months later. I was surprised that I felt a little sad when I got the results. Seeing scientific "proof" that Danny has CHARGE Syndrome somehow seemed to make his problems more concrete. But if no mutation had been found, I probably would have been more upset, starting from scratch to see if he should have some other diagnosis.

9. Pay the bill. The bill was in Dutch with a cover letter in English, sent to me through my doctor's office. KGCN does not accept credit cards. I had my bank convert euros to dollars and wire the money to KGCN's bank. My bank charged me a \$40 fee, which I think is typical.

10. I sent a claim to my insurance company. The claim is still pending although the indications I have so far are that they probably will not pay because it is a foreign lab. I figured going into it that I probably would not be reimbursed. Maybe someone more clever or tenacious can figure out a way to get their insurance to pay.

Although the results do not have a lot of practical use to us at this point, I feel it filled in a piece of the puzzle and was worthwhile. KGCN advised me to send in samples for myself and my wife (at 650 euro each) to establish that it was a de novo mutation rather than inherited from either of us. We don't plan to spend the money at this point. We will wait until Danny's unaffected twin brother is old enough to have children and then see what tests are available. *Editor's Note: Identification of a mutation in an individual with CHARGE allows prenatal diagnosis in future pregnancies.*

Dr. Conny van Ravenswaaij and the staff at KGCN were most courteous and helpful during this process. I visited the Netherlands years ago but I still got a kick out of tracking little vials of Danny's blood across the world and trying to translate bills in Dutch.

DNA Testing for CHARGE Syndrome

by Kyna Byerly, Genetic Counselor

As most of you know, last fall Dr. Conny van Ravenswaaij's group in the Netherlands announced they had identified a gene for CHARGE. The gene is called CHD7 (which stands for chromodomain 7) and is on chromosome 8. Some background information may be helpful in discussing DNA testing. Genes are made up of a sequence of DNA code. The code is a sequence of bases, represented by four letters: GACT. So a gene is a long sequence of bases: AUG GGC GUA GCG.....etc. Genes code for proteins. Each three bases are a "codon" and represent a specific thing: 'start' 'stop' or a specific building block of the protein (an amino acid). The cell 'reads' the DNA code and makes a protein to match. Proteins need to be made in the right amount, at the right time, and have the right structure and shape to do their job.

DNA is organized into genes; genes are organized on chromosomes. Everyone has two copies of each chromosome, thus two copies of CHD7. If one gene is missing (deleted) less protein is made. As you can imagine, a big deletion involving many genes may have more of an effect than a small deletion involving only one gene. A deletion of a whole gene, or just a small part of a gene, can cause problems. Chromosomes are big enough they can be seen with a microscope and analyzed (the study of chromosomes is called cytogenetics). A chromosome deletion found through standard cytogenetic analysis is a deletion of a large amount of DNA involving many genes. Smaller deletions (micro deletions) are harder to detect. Even smaller changes (mutations) in the DNA sequence can still have big effects. Changes in a single base can cause a non-functional protein. Single base mutations can be detected by sequencing - a longer, harder process that involves determining the exact sequence of base pairs for the entire gene, thousands of bases long! In other words, the smaller the change in the DNA, the harder it is to detect. DNA testing can identify changes that can lead to genetic syndromes, like CHARGE.

A small percentage of people with CHARGE tested have deletions involving the CHD7 gene. Large deletions can be detected by a type of testing called *genomic microassay analysis*. A much larger percentage have small mutations in CHD7. These mutations can be detected by *gene sequencing*. Some people with CHARGE clinically will not have a small mutation or deletion in CHD7. This does not mean they do not have CHARGE; it means there is another cause; probably another gene which can cause CHARGE which has not been identified yet. Gene sequencing is the best type of testing for CHARGE, as most people have small mutations, not deletions. So far, it appears that around half of all people with CHARGE have mutations in the CHD7 gene identifiable by gene sequencing.

Where is DNA testing being done? There are currently three labs that do clinical, fee-for-service DNA testing for CHARGE. One is Dr. van Ravenswaaij's group in the Netherlands, Clinical Genetics Center Nijmegen. They offer sequencing of the CHD7 gene, prenatal diagnosis, and clinical confirmation of mutations detected in a research lab. They charge about \$840 at current exchange rates. See Michael Schwartz's article for his experience with this lab. A second, newly available option is GeneDx, Inc. in Gaithersburg, MD. They also offer sequencing of the CHD7 gene, prenatal diagnosis, and clinical confirmation of mutations detected in a research lab. They charge \$3900. A third option is Signature Genomic Laboratories, in Spokane WA. They offer genomic microassay analysis, which does not detect as many mutations as sequencing. Some labs may be able to do testing on cells from inside the cheek, as opposed to a blood draw.

A fourth center is Baylor College of Medicine, in TX. This testing is done on a research basis only. Some of you may have participated in this research. According to Seema Lalani, they have studied 111 patients. While they started with a different type of testing, they are now sequencing the CHD7 gene. The results are still being analyzed so they do not yet know the exact number of cases with a mutation identified. They will notify all research participants directly of the results. This work has been on research basis with no cost to the families. They would recommend testing by GeneDx or the Netherlands lab to get timely results. However, they are accepting new samples for research. If a mutation in CHD7 is not found, the samples will be saved for future analysis if another gene is identified. Families interested in participating in this research may contact Seema Lalani, MD or Susan Fernbach, RN directly at 832-826-5713 or visit www.cardiogene.org/contacts.htm.

How can you get DNA testing, and should you? You will need a doctor to order the testing; results will be sent to the doctor requesting the test. Hospitals and larger labs are usually able to draw and send out samples to outside labs. Genetics clinics have a lot of experience in doing DNA testing through outside, even out of country labs. Consultation with your geneticist or genetic counselor can help you determine whether or not to pursue DNA testing, and by which method. They can also facilitate the whole process, i.e. get the right forms, arrange for the sample to be shipped, and call you with the results. DNA testing can be a complicated, long and expensive process; however there are resources available to help you determine if you want to pursue it, get it accomplished and understand the results once they are in.



PRESIDENT'S NOTE

Bruce Appell

Hello Everyone,

Another school year is winding down, the air is beginning to warm, and summer is peering around the corner. Before you know it, we will be visiting and sharing life experiences with one another on the beaches of Miami.

By now, you should have received your registration information for the 7th International CHARGE Syndrome Conference. Marion, Susy, Neal, Dennis, and all of our Board Members are working very hard to make this a memorable experience. I know just last night Uncle Kenny was sitting at our dining room table coming up with ideas for Uncle Kenny's craft corner. It is unbelievable how much time, effort, and hard work goes into planning and preparing for a conference, especially one of this magnitude. We hope that you will be able to attend and take advantage of the wonderful opportunities and learning experiences that will be available for you. Please encourage your child's therapists, educators and medical advisors to take advantage of the conference and attend as well.

We are entering a busy time of year. For those of you who have children graduating from primary, high school and college programs we want to say congratulations and wish them well. As you leave on your vacations we hope you have a safe journey. For those who have loved ones serving in the military services we say a pray for their safe return home. And as always, we wish our children with CHARGE, their siblings, parents, and extended families the strength they need to face all challenges ahead.

The CHARGE Accounts newsletter is intended for general information only. Medical or treatment information and/or opinions are not necessarily endorsed nor recommended by CHARGE Syndrome Foundation, Inc. or its officers. Readers are reminded that the best source of medical advice is always their child's physician.

FOUNDATION ACCOUNT

EXECUTIVE DIRECTOR'S NOTE

Marion Norbury

The 7th International CHARGE Syndrome Conference is less than four months away. We hope you are making plans to attend. Remember you can help us in our planning if you register early. However, you have until June 15 to pay for your registration. If you have questions about the conference or need a registration packet, please contact the Foundation office at 1-800-442-7604.



SILENT AUCTION NEEDS YOUR HELP

Please contact Dennis O'Toole if you can donate items for the Silent Auction. You can reach Dennis at 440-331-4869 or email at dennis@chargesyndrome.org. With everybody's help, we can support the Foundation while having fun at the conference.

Committee Chairs

Conference – Neal Stanger

Research – Meg Hefner

Fundraising – Brownie Shott

Education/Outreach – Jim Thelin

**All board members have a
special email address:**

[\(firstname\)@chargesyndrome.org](mailto:(firstname)@chargesyndrome.org)

CHARGE Syndrome in the American Journal of Medical Genetics

Meg Hefner, M.S.

The last two years have been very exciting for CHARGE. In 2003, the first Behavior Symposium was held at the Cleveland CHARGE Syndrome Conference. In 2004, the first major gene for CHARGE syndrome was reported. And in 2005, a special issue of the American Journal of Medical Genetics devoted entirely to CHARGE Syndrome was published. It is available from the Foundation for purchase. It may be found also at a medical library.

For many years, the medical community and the CHARGE Foundation conferences focused primarily on the medical and developmental aspects of CHARGE. Thanks to the efforts of Tim Hartshorne (psychologist and father of a young man with CHARGE), behavior in CHARGE has become a major field of study. Tim organized the Behavior Symposium and asked each presenter to send us a written summary of their research and presentation. We received about a dozen articles. Tim, Sandra Davenport and I edited those articles and submitted them as a group to the American Journal of Medical Genetics. The editors of the journal were delighted to have such a comprehensive look at behavior in a genetic syndrome. They accepted all of the articles, including one written by three parents. In addition, the Journal had received a number of other submissions about CHARGE. They decided to dedicate an entire issue of the Journal to CHARGE Syndrome. The March 15, 2005 issue of the journal is a Special Issue on CHARGE syndrome. These articles are also available to you and the professionals who work with your children. Please keep in mind that they are written for medical and educational professionals – some translation may be needed!

Purchase entire journal: The Foundation has a limited number of the entire issue of the journal available for purchase at \$20 each (postage included). To purchase, contact the CHARGE Syndrome Foundation, Inc., 409 Vandiver Dr Ste 5-104, Columbia MO 65202-1563. Orders will be limited to one copy.

Editor's Note: In the Winter 2004 Issue of Accounts, we asked for interested members to apply for a position on the Foundation's Board of Directors. We explained that there were six positions available for persons to serve on the Board. When the deadline for applications arrived, we had received five applications. Since the bylaws state that "The six (6) persons receiving the highest number of votes will be elected," it was decided that we would not hold a formal election but would declare these five duly elected. The vacant position will be filled by the new board after the 2003 Conference. Again, if anyone is interested in applying for this position, please contact the Foundation office for an application form.

We have printed the applications from the new Directors below as a way of introducing them to you.

CHARGE Syndrome Foundation, Inc. Board Candidates' Statements 2005

Randall Thomas Goodwin Family member of individual with CHARGE

Hours available per week: as many as it takes to support my duties

Committee interests: Biennial Conference, Fund Raising, Lee E. Norbury Memorial Scholarship, Membership

Qualifications, Goals and Interests:

My name is Randall T. Goodwin and I am a fabrication supervisor with Lockheed Martin Aeronautics, located in Fort Worth, Texas. I have been with Lockheed Martin for the past two years. Prior to this I was on active duty with the United States Marine Corps, retiring after 20 years of service. I am a veteran of the Gulf war, and have held Detachment Commander billets at 2 American Embassies (Panama City, Republic of Panama and Port-Au-Prince, Republic of Haiti). I have managed large budgets and have lead many people. I also have an uncanny ability to get things accomplished in a timely manner.

I, like many of you, was thrown into the world of CHARGE not knowing the first thing about it. I will be the first to say that I don't know everything about CHARGE, but I know there are paths that will take me to people who might. One thing that I am sure of is that there are a lot of parents out there that know very little about CHARGE. One of my goals, whether elected to the board or not, is to help provide educational material/direction for those who elicit information and help. Having a son born in a hospital that has one geneticist, who three years later still did not recognize CHARGE, has made me dig in and educate myself. It shouldn't have to be that way. CHARGE is recognizable by its many different features. Educating professionals that care for our kids is another goal of mine. One of my best assets is my wife, who is currently training in the Texas Sparkle program to become an advocate for parents and kids that need help.

The application asks for my qualifications. I have searched for a reply to that. All that I can come up with is that I am ready, willing and eager to help the CHARGE Foundation in any way that I can. If given the opportunity to serve on the Board of the CHARGE Syndrome Foundation, I will serve the members to the best of my ability.

Bonnie Haggerty Family member of individual with CHARGE

Hours available per week: at least 10 hours, more if needed.

Committee interests: Public Relations: communication/networking (newsletter, web page, e-mail, support groups), Professional Services (e.g. medical/Educational issues)

Qualifications, Goals and Interests:

Parent of a daughter, Patty (age 20), with CHARGE; paraprofessional within Special Education 1993-present; currently have 105 credits toward Special Education degree, graduate of Partners in Policy Training; organized a parent support group in hometown; currently working with DBCC to develop task force to improve transportation for individuals with deaf/blindness

Although we are members of the Federation of the Blind and the National Family Association, the CHARGE Syndrome Foundation has been vital to our lives. I would like to focus on educational interventions for all our children, including older individuals. I would also like to focus on behavior, and the changes an individual with CHARGE goes through into adulthood. Having met numerous experts within the educational avenue, including those at Perkins and at Helen Keller National Center, I would hope to improve an already fabulous avenue for individuals with CHARGE. I would also hope to focus on public relations throughout America. It is my hope to give back to the Foundation the immense help, joy and enlightenment we have received.

Marilyn Ogan Family member of individual with CHARGE

Hours available per week: 12-15 hours per week, with more available during Summer months.

Committee interests: Biennial Conference, Membership, Public Relations: communication/networking (newsletter, web page, e-mail, support groups).

Qualifications, Goals and Interests:

Previous Board Member, including Vice-President; 2001 Conference Chair; Parent of CHARGER; Vice-President of PTCO (parent organization) at Indiana School for the Deaf; Graduate of Partners-in-Policymaking (Indiana)

I have a high interest in educational and community advocacy for individuals with disabilities. I would like to see the CHARGE Syndrome Foundation, Inc. become a resource for advocacy, in the sense that we can disseminate information about what has worked for other families in community settings. I would also like to see the Foundation continue as a resource for individuals and families diagnosed with CHARGE on issues like the numerous Deafblind problems encountered once transitioning to adult services in the community, from the educational setting. As our "children" grow and reach maturity, there are issues regarding guardianship, services, transportation, etc., and we have many families who have "been there; done that." Yet the Foundation has little packaged information available to the families with older children. I would like to see the Foundation work in collaboration with groups like Perkins and NFADB to develop information specific to CHARGE/Deafblind issues for the older individual.

I feel this has the potential to expand our membership numbers, as well. While we see a large number of families with newly diagnosed children turn to the Foundation for information, we seem to lose the older families.

I am pleased the Foundation has expanded to a new office outside of Marion Norbury's home. I feel this only emphasizes the fact of our growth as a clearinghouse for information about CHARGE Syndrome. I believe we just need to expand our information/resources to include the families that are facing futures beyond childhood.

Bonnie Shott Family member of individual with CHARGE (current Board member)

Hours available per week: 3-5 or more as the need arises

Committee interests: Biennial Conference, Fund Raising, Lee E. Norbury Memorial Scholarship, Membership

Qualifications, Goals and Interests:

As a parent of a child with CHARGE Syndrome and a current Foundation Board member, I would like to continue working to create a strong Foundation that is able to help meet the needs of many of those affected by CHARGE Syndrome, whether they are individuals with CHARGE Syndrome, their families and friends or professionals. Although we have made progress as a Foundation, we have much more to do in the coming years in the areas of awareness, education, research and more. I would like to continue working as a board member to assist with the necessary work. My past experience as the Executive Director of an agency that delivered services to people with disabilities in their homes, my current experience as a volunteer in our school district and my current experience as a business owner have all provided me with tools to bring to the Foundation. Couple that experience with my passion for all our children and adults who have CHARGE Syndrome, and I believe I can continue to be a good board member of the Foundation.

Neal Stanger Family member of individual with CHARGE (Current Board Secretary)

Hours available per week: 30-40

Committee interests: Biennial Conference, Fund Raising, Lee E. Norbury Memorial Scholarship, Membership, Public Relations: communication/networking (newsletter, web page, e-mail, support groups), Professional services (e.g. Medical/Educational liaisons), Research

Qualifications, Goals and Interests:

In August of 1993 our daughter Megan was born. She is a blond haired, blue-eyed girl with more energy than I have ever seen in another human being. In 1994 she was diagnosed with CHARGE Syndrome. By the end of 1994 I had decided that there needs to be more known about CHARGE Syndrome by myself, and more importantly the medical community and the general population.

After attending the Portland conference in 1995 I realized that I wanted to get more involved in the CHARGE Syndrome Foundation. I was elected onto the board in 1997 and I served as the Communications chair for the first two years. I set up our website (www.chargesyndrome.org), got all the board members email addresses (neal@chargesyndrome.org) and set up numerous list serves for the board of directors and committees so that we could easily communicate with each other. In 1998 I suggested to the board that we have a silent auction at the Houston Conference. The auction was a huge success and we have since had three total auctions raising well over \$30,000 for

the Foundation. In 1999 I was elected to the Executive Committee and have since held the positions of Secretary, Treasurer and Vice President. Besides the Executive Committee positions that I have held, I also handled the program and agenda for the 2003 conference in Cleveland, and I am currently the Conference Committee chairperson for the upcoming conference in Miami this summer. I have sat on most committees and task forces over the years and most recently I was very involved in finding a new office for the Foundation in Columbia, Missouri. On the fundraising aspect, I have been responsible for raising funds and donations in excess of \$75,000. This also includes certificates that have been donated for airline tickets when our board and Executive board get together for face-to-face meetings.

My goals for the foundation are simple. I would like the foundation to continue being a family run, family driven support system. I am also hoping that we could eventually have enough funding to do everything we want, and never have to look at the financial side of it, just the benefits for our children. I would also still like to get more awareness for CHARGE Syndrome in the general population. By your supporting me again and voting for me to stay on the board I would be one step closer to fulfilling these goals.

RESEARCH ACCOUNT

University of Tennessee Survey on Factors Related to Communication Development

A special mailing was recently sent from the Foundation Office to the members about the *UT Survey on Communication Development*. It was also distributed on the listserv. It is an online survey. You go to <http://surveys.utk.edu/charge/index.htm>. The survey is anonymous and confidential because you give no identifying information.

The survey will take about an hour to complete and you may need to have some of your child's records to help you with developmental milestones.

What is the purpose of all this? We found out that the acquisition of symbolic language is related to (1) solving hearing problems, (2) the ability to walk independently, and (3) communication training early in life. We are asking you to share what you know about these topics. We are going to use what is known from the professional literature and the information that parents tell us about problems and solving problems to create a booklet for parents on communication development. It will be introduced at the Conference in Miami Beach.

We really would appreciate your participation. And we also urge you to participate in the Balance Survey that is being conducted by a group from SUNY.

Jim Thelin and the University of Tennessee Research Team
865-974-1796 or jthelin@utk.edu.

Looking for information about your child with CHARGE

Gina Pucci is a Master student at SUNY Brockport in Rochester, NY. She is completing her thesis on the *Effect of CHARGE Syndrome on Balance in Children during Physical Activity*. Her focus is looking at all variables that may affect a child with CHARGE balance related problems. If interested in filling out a questionnaire and taking part in this research, please contact Gina via mail, phone or email of your interest. A questionnaire will then be sent out to you to complete!

Gina Pucci
3901 Spanish Bay Court, Elkton, MD 21921
(716) 984-4519 gpucci@ccps.org



Memorial Gifts Create Lasting Tribute to Central Michigan University Student

When Central Michigan University (CMU) sophomore **Joanna M. Russ** was tragically killed in an automobile accident on March 11, 2004, her family and friends knew they couldn't let her efforts in conducting research to help children with disabilities fade away. In a lasting tribute to Joanna, they established the **Joanna M. Russ Memorial Undergraduate Research and Presentation Grant** through memorial gifts designated to CMU, including substantial gifts from psychology department faculty members and Joanna's parents, Walter and Annie Russ. The annual grant will help offset students' costs of conducting research or presenting the results of their research.

Joanna, a psychology major, wanted to become a psychologist and work with special needs children and their families. She was especially interested in helping children with autism. "When Joanna was in high school, she worked with a set of autistic toddlers to fulfill her National Honor Society community service hours," said Annie Russ. "She did very well with them, and it just came naturally to her. She would think about things such as what color shirt to wear to get the kids to respond, and she had good luck with it."

Joanna started taking classes at CMU in the fall of 2002, but was unhappy with the lack of activities to keep her engaged. "She was a very driven, straight-A student, so she needed a lot of activity and stimulation," said Annie. Looking for a way to get Joanna more involved in university programs, Mike Owens, CMU's associate dean of students, introduced her to psychology professor Tim Hartshorne. "Joanna was very interested in autism and severe disabilities, so I asked her to join my research team to study CHARGE Syndrome," said Hartshorne, who was in the midst of conducting two simultaneous studies funded by the CHARGE Syndrome Foundation.

"CHARGE Syndrome is a relatively rare condition (1 in 12,000 births is an estimate) that was first identified in 1979. Some of the children with CHARGE display behavior that can be classified as autistic, obsessive compulsive, attention deficit hyperactivity, and tic disorder," said Hartshorne. "The major challenges with CHARGE are vision and hearing problems resulting in many children being classified as deafblind; swallowing problems leading to the need for feeding tubes; balance problems causing significant delays in learning to walk; and communication difficulties related to the deafblindness."

"Joanna made a big contribution to our study by getting us to include questions on communication. She wondered whether communication skills development and/or the method of communication used by the child influenced behavior or sensory processing. We have found that communication skill problems are related to more severe behavior difficulties," said Hartshorne.

The Joanna Russ research and presentation grant is open to CMU students of all academic majors. Research related to CHARGE Syndrome will be given priority, but projects can fall under any of the following categories:

- The cause or nature of behavior difficulties in children with CHARGE Syndrome;
- Educational, family, medical, social, or other difficulties experienced by children with CHARGE Syndrome or other genetic syndromes;
- Studies related to autism and autism spectrum disorder;
- Studies related to other low incidence disabilities such as deafblindness.

Applicants must have a minimum 3.0 GPA and present evidence of faculty sponsorship. The psychology department will select the number of recipients and amount awarded each year.

Although the loss is unbelievably hard, Annie said the family is comforted by the many letters they've received from Joanna's friends, teachers, and the special needs children and families that she worked with. "We had no idea that she had touched so many lives, that her influence on others was so encompassing," she said. The family also finds comfort in knowing that Joanna's compassion for helping special needs children will be carried on by other students at CMU. "It started out as an 'in lieu of flowers.' We had no idea it would develop into something this big," said Annie. "My husband's co-workers held fund-raisers and raffles to raise more than \$3,500. Joanna's high school and college friends and teachers, her roommate's family, and the family of the children Joanna babysat all emptied their pockets to help out."

"Joanna's love for others will live on forever in so many hearts and minds, and it's her love that created all of this. Her spirit, her life, everything that she wanted to do will live on in her scholarship."



We thank everyone for their generous support . . .

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Matching Gift Program and
Eric Stovicek NJ
Bob & Kim Anderson IL
Corrine Doogan IL
Ronald Podoll KY
Frank & Joan Overton MI
Morgan Gibson TX
George & Phyliss Saul NY
Audrey Dwyer NY
Anne Winter FL
Bonnie Haggerty MA
William & Sandy Muir PA
Karen & Ken Wojcik MN
Ann Garner AL
Cathleen Josephson NY
Michelle Partridge AL
Michael & Laurie Schwartz CA
Julie & Zane Shook KS
Don & Cathy Kauffold IL
Timothy & Nancy Hartshorne MI
Jacqueline D'Urso CA
Ronald & Laverne Krause OH
Patricia Mosher Nova CANADA
Cyndie Pfohler NY
Florence Finnerty PA
Dennis & Colleen O'Toole OH
Maria Corsini & Brian Still WA
Tracey & Philip Cutcliffe CANADA
Emily & James Murray NJ
Steven & Susan Hiscutt TN
United Way of Kentucky KY
United Way of Kitsap WA
Allstate Giving Campaign VA
Community Foundation for SE
Michigan - Roberts Family Fund
CHARGE Across America
Michael Brennan OH

Spanfelner Memorial

*In memory of Will Spanfelner, son of
Michael & Amy Spanfelner of Chico, CA*
Paul Garcia & Catherine Steinbach
CA
Peter & Darlene Duryea PA
Richard & Marta Holm CA
Andrew & Nancy Garcia CA
George & Carol Vranesh CA
Keith & Edna Waller CA
Mr. & Mrs. William B. Rometo OH
Dale Jones CA
TM Duche Nut Co., Inc. CA
W. James Edwards CA
The Staff of Hooker Oak School CA
Peter & Kellie Paredero CA
John & Dorothy Rath CA
Scott & Cynthia Chalmers CA
Fred & Carol Richelieu CA
Joseph & Barbara Osness CA
Dennis & Sandra Cole OH
Tyson & Laurie Henry CA
Patricia Calhoon CA
Blanche Rodgers & Mary Jo Corsetti
PA
Paul & Linda Kondrick CA
Glenn Johnston, OD CA
Melissa & Matthew Davis MA
Michael & Kathy Lyon CA
Jerry & Carla Dunham CA
Walter & Sharon Dahlin CA
Michael & Diedra Cross CA
James & Jean Jorgenson CA
Leonard & Alsia Costa CA
Lee & Sierra Grider CA
Jack & Joyce Winter CA
Eugenie Rovai CA
Douglas & Nancy Meyers & Cynthia
King CA
Patricia Davies CA
John Lucchesi CA
Rebecca & Kevin Gallagher CA
Kevin & Janie Murray CA
James & Mary Clancy CA

Research Fund

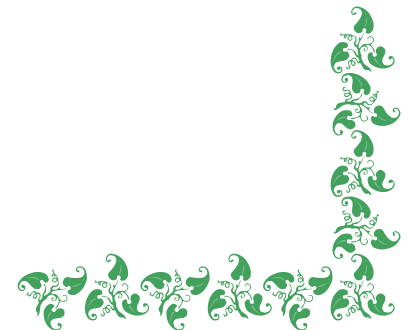
David & Barbara Cash MI
John & Cheryl Kruger MI
Maureen Cash & Brian Bartkowiak
MI
George & Linda Nahitchevansky NY
Kelly Drew NC
Angela Mugeridge CA
Keith & Jennifer McDaniel OK
In honor of Christian Roberts
Don & Julie Graham MD
In memory of Meghan Graham

Conference Fund

Marilyn & Rick Ogan IN
Linda Sontag PA

Special Requests

Treva Schaeffer OH
on behalf of Joseph Lavelle
Rocky River Junior Women's Club
OH *In honor of Clare O'Toole*
Helen Dillinder TN
In honor of Hunter Doran
Irene & Jim Lucas CA
*In honor of great-granddaughter
Cheyenne Brittell*
Ruth Downey MN
In honor of Michael Kruger
Betty McKnight OH
In honor of Makenzie Dougherty
Harold Reich & Sandra Swartz CA
Jo Ann & Forrest Stichman CA
Edna & Bernard Carlin CA
*In honor of Danny Schwartz and
honoring Ruth & Jerry Schwartz*
Carolyn Fleet TX
In honor of Garland Goodwin



CHARGE SYNDROME
AWARENESS WEEK
JULY 18-22, 2005

This is the time to spread the word about CHARGE Syndrome.

Contact the media, public officials, schools and other
organizations in your area
and
tell them about CHARGE.

Distribute brochures to your library, clinics, newspapers,
tv and radio stations.

Write a short description of CHARGE Syndrome and promote
the Foundation and the Conference in July.