

# Heart to Heart

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## From the PRESIDENT

This issue focuses on our CCF families — their amazing stories with new methods of treatment and the wonderful work they are doing to raise awareness and funding for research on cardiomyopathy. As I read about their grassroots efforts, I am reminded of an article about the Komen Breast Cancer Foundation. While they are now a multi-million dollar foundation well respected in the medical research community, this was not always the case. They started out with the lone voice of their founder but over time more families joined in their cause and with each new voice added to their chorus, their message became clearer and stronger. As their voices became louder, people of influence began to take notice of what was then an ignored disease. It started with one voice but with many voices, it brought about a symphony of change.

Similarly, cardiomyopathy in children is still considered a “silent killer”, not well understood, and invisible to the eyes of major funders and the media. However we have the ability to change this — just like Susan Komen did with breast cancer. We can make a difference in our own individual way, whether it's contributing to CCF's annual appeal, planning a small fundraiser, volunteering professional skills or speaking to the media and the community. Each action brings our voices together in the search for a cure. I hope that you will be inspired, as I was, by the words of the Komen Foundation's “One Voice” campaign (featured to the right).

May the holidays be a special time for you and your family!

Lisa Yue  
President & Founder

## New Medical Advancements for Pediatric Cardiomyopathy SAVES Lives

**Breakthroughs** have been made in the effort to find alternative treatments to sustain and save the lives of infants and young children battling cardiomyopathy. Treatments that were initially created to support the hearts of teens and adults have now proven to be effective on much younger, smaller hearts. Two medical advances have met with great success with two CCF families. One involves **biventricular pacing** to treat dilated cardiomyopathy and the other involves a ventricular assist device called the **Berlin Heart** for children with severe heart failure and in need of a transplant.

### Biventricular Pacing

Biventricular pacing is a procedure in which a small pacemaker is placed under the skin of the upper chest or abdomen. The device is connected to leads (soft insulated wires) that are either inserted inside the heart



Jacob Urowsky, 18 months after surgery

(endocardially) or attached to the outside of the heart (epicardially). The device is battery-powered and sends tiny electrical impulses to the two lower heart chambers, the right and left ventricle, to beat in a more synchronized pattern. As a result, the heart is able to pump more effectively, thus improving the heart's ability to supply blood and oxygen to the body. This type of resynchronization therapy has been performed on adults for years, but this procedure has only been tested on young children a few years ago. Implanting a pacemaker in a child, especially an infant, was almost unheard of in 2003 until it was successfully utilized for **Jacob Urowsky**.

In 2002, at the age of four months, Jacob was diagnosed with dilated cardiomyopathy after

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### WORDS OF INSPIRATION

*I am one voice,  
one voice singing in the dark,  
just one lonely voice.  
My song can be a spark;  
come and join me.  
We can light a fire so bright  
the world will be dancing in our light  
and we will not be silent anymore.  
We are one voice; we will not be silent.  
We will keep on singing 'til we're heard.  
We are one voice; we will not be silent  
'til our song is sung around the world.*

- Komen Foundation's  
“One Voice” campaign

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Children's



Cardiomyopathy  
Foundation

*“A Cause For Today... A Cure For Tomorrow”*

The *Heart to Heart* newsletter is published biannually by the Children's Cardiomyopathy Foundation (CCF), a national health organization dedicated to saving lives and improving the quality of life for those affected by pediatric cardiomyopathy. Pediatric cardiomyopathy is a chronic and life-threatening heart disease that affects roughly 30,000 children in the U.S. CCF's mission is to accelerate the search for a cure by stimulating research, educating physicians, and increasing awareness and advocacy related to the needs of affected children and their families. CCF is a publicly supported tax-exempt organization as described under section 501(c)(3) of the Internal Revenue Service.

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# Genetic Discrimination:

## A Consideration for

**Genetic testing** is defined in the *New England Journal of Medicine* as "the analysis of human DNA, RNA, chromosomes, proteins and certain metabolites to serve a number of medical purposes." It can be used to diagnose a genetic disease and to identify who may be at a heightened risk of developing a particular disease. With pediatric cardiomyopathy, where 68%-80% of cases are idiopathic (no known cause), genetic testing can help to pinpoint the exact cause of the disease, leading to proper diagnosis and more effective therapy. Genetic testing can also help to identify other "at risk" family members, such as parents, siblings, children or other relatives. The results can provide life-saving information on who should be monitored more carefully and who does not need to be followed long term.

Currently there are a handful of clinical labs that do specialized genetic testing on cardiomyopathy. Scientists continue to research genetic mutations responsible for pediatric cardiomyopathy with the goal of developing diagnostic chips that could screen children and their families for a variety of genetic causes for cardiomyopathy. Although this is an exciting possibility, this option is complicated by the fact that legal protections are not in place to protect families from genetic discrimination related to genetic testing and research. Genetic discrimination occurs when a person is treated differently based on their genetic information (family history of a disease or a genetic test result of an individual or of a family member) because he or she has an inherited disorder or is at risk of developing an inherited disorder.

According to studies published in the *American Journal of Medical Genetics* and *Genetics in Medicine*, the majority of people with inherited disorders or inherited risk are very concerned about genetic discrimination. The concern is that their genetic information may be used to deny, limit or cancel their health and life insurance and to discriminate against them in

the workplace. However, the reality is that documented cases of discrimination are rare. The only known publicized case taken to court occurred in 2001. Terri Seargent was diagnosed with Alpha-1 Antitrypsin Deficiency after losing her brother to the disease. Her self-insured employer dismissed her after receiving a bill for preventative treatment. Mrs. Seargent filed a complaint with the **Equal Employment Opportunities Commission** and was vindicated.

The possibility of genetic discrimination can negatively affect a family's decision to screen members, especially minors under 18 years of age. As a result, some people are not taking advantage of genetic tests that could make a significant difference in their healthcare decisions and outcomes. This fear also prevents many people from becoming involved in medical research that could provide better information and choices for those affected by hereditary disease in the future.

The current situation is that there is essentially no risk for people who are covered by government programs (Medicare, Medicaid, Military) or medium-to-large employers (20 or more employees) with group health insurance plans. This accounts for the vast majority (roughly 80%) of people in the U.S. with public or private health insurance. The 1996 enacted **Health Insurance Portability and Accountability Act (HIPAA)** does protect people with group health insurance against being denied insurance, having their insurance canceled or having their rates individually increased due to any pre-existing condition. Federal employees have additional protection. An executive order enacted in 2000 prohibits discrimination in federal employment based on genetic information.

Risks are greater for people who purchase health insurance on their own rather than through an employer because there will be greater scrutiny of the individual's health status and medical records.

# Genetic Testing

In terms of life or disability insurance, discrimination may be a more serious issue because plans are usually purchased by individuals, not by groups, and requires review of an individual's risk status. While it may still be possible to obtain life insurance, increased premium charges may apply.

While most states have enacted legislation on discrimination in health insurance and in the workplace, not every state offers the same protection and some states have not passed anti-discrimination laws. Advocacy groups such as the **Genetic Alliance**, the **Coalition for Genetic Fairness**, and the **Council for Responsible Genetics** believe that the current state adopted legislation is mostly a patchwork of clauses that leave gaps through which discrimination can still occur. Thus, these groups are pushing for more comprehensive and far-reaching federal protection.

On February 17, 2005, The United States Senate took the first step in prohibiting discrimination on the basis of predictive

genetic information with respect to health insurance and employment. This piece of legislation known as **The Genetic Information Nondiscrimination Act** recognizes that "all individuals, whether they are healthy or sick, and all medical information, genetic or otherwise, should be afforded the same protections under the law." The legislation specifically:

- 1) Prevents health insurers from denying coverage in group or individual markets and in adjusting premiums based on an individual's predisposition to a genetic condition.
- 2) Establishes basic legal protections that will enable and encourage individuals to take advantage of genetic screening, counseling, testing and new genetic therapies.
- 3) Impedes both employers and insurers from requiring applicants to submit to genetic tests.
- 4) Maintains strict use and disclosure requirements of genetic test information.
- 5) Imposes penalties against employers and insurers who violate these provisions.

Despite President Bush's full support and unanimous approval in the Senate, the Genetic Information Nondiscrimination Act still awaits action in the House of Representatives. Once this passes, it will be brought to the President to be signed into law.

If this legislation passes, it only protects "at risk" individuals who are carriers or have a dominant mutation but no signs or symptoms of the disease. It does not cover people with pre-existing genetic diseases who have manifested symptoms. In the meantime, there are other ways to reduce the risk of discrimination if a family chooses to do genetic testing or participate in genetic research:

- 1) Inquire about whether genetic test results can be kept separate from a patient's normal medical records.
- 2) Inquire about whether a patient can be tested anonymously under a pseudonym without recording any information about their actual identity.
- 3) Decline to seek reimbursement from the patient's health insurer for testing or seek reimbursement only if the test results are favorable.

Slowly, legislative and grass-root forces are coming together to ensure that the hopeful promises of science and medicine can become a reality for all of society. All those involved agree that individuals need to overcome genetic discrimination concerns and move forward with genetic testing and research for the benefit of a healthier medical future.

## Georgia Patient Support Group Established

Fellow CCF family member, **Audrey Callahan** has taken the initial steps to reach out to other parents and children living with pediatric cardiomyopathy. With CCF's guidance, she has established a CCF support group with the **Sibley Heart Center**, a division of the **Children's Healthcare of Atlanta**, Georgia.

This new local-level support group offers families in the Atlanta metro area an opportunity to gather every other month to talk,

inform, comfort and sustain one another in their common struggle with pediatric cardiomyopathy. Currently, ten families, nine of whom have registered with CCF, participate in the group.

Several key hospital staff members serve as medical advisors to the support group. They include cardiologist **Dr. William Mahle**, **Megan Ramsey**, the Program Specialist for the **Kids at Heart** program, and **Alex Berg**, a nurse and Heart Transplant Coordinator at Sibley.

Audrey's hope is to expand the group to include other families in the Atlanta area, to one day have annual fundraisers, and to help support state-wide initiatives such as those that screen teen athletes for cardiomyopathy. She would also like to organize family picnics so that parents and, especially, children can meet others who are battling this disease. "When we were first diagnosed, we thought it would be so nice to call someone who was going through the same things to talk to. That's what we're trying to offer other families now," adds Audrey.



Dr. William Mahle & Megan Ramsey of Sibley Heart Center



CCF family member Amy Urowsky & Megan Ramsey

## New Medical Advancements for Pediatric Cardiomyopathy Saves Lives *continued from page 1*

initial feeding problems and then an irregular heartbeat was detected. Jacob was also found to suffer from a condition known as dysynchrony or “left bundle branch block” in which the lower chambers of the heart do not pump blood in unison with one another or with the upper chambers. Because there is a delay between the contraction of the right side and the left side of the heart, the heart has less time to fill with blood and cannot pump enough blood to the rest of the body. Heart failure symptoms are worsened as a result. It is a condition that can occur independently of cardiomyopathy and physicians do not know whether one condition causes the other.

Upon diagnosis, Jacob was immediately admitted to the cardiac intensive care unit (CICU) at the **Children’s Healthcare of Atlanta** in Georgia. His ejection fraction (measurement of pumping efficiency) was only 12% at the time and his heart was very enlarged. The team of specialists prescribed medications at first, but when those failed to improve Jacob’s heart function, doctors concluded that a heart transplant would be Jacob’s best chance for survival although his prognosis would still be questionable. After painstaking research and consultations with families of transplanted children, Jacob’s parents, Amy and Todd Urowsky made the difficult decision to not go forward with a transplant. Jacob was then moved to a hospice center for end-stage care. However, miraculously, Jacob began to resume his eating, something he had not done since his initial diagnosis. He was readmitted to the CICU and administered **Coreg**, a drug typically prescribed to adults with heart failure but more recently being tested on children with dilated cardiomyopathy. Jacob’s condition improved dramatically. Four months later when Jacob was almost one year old, the biventricular pacing procedure was recommended based on his two heart disorders. Nearly three years after the pacing system was implanted, Jacob’s heart is pumping efficiently. He no longer needs medication and his cardiomyopathy seems to have disappeared.

According to Jacob’s cardiologist at Children’s Healthcare, **Dr. Peggy Strieper**,

the biventricular pacing procedure may be a better alternative than heart transplant for some children. “In children, heart transplants are only palliative, they’re not curative,” says Dr. Strieper. “In addition it can be very difficult for patients and families to deal with the complications that can result after transplant.”

There are strict guidelines for a child to be a candidate for biventricular pacing:

- The child must have primary cardiomyopathy (e.g. disease only affects the heart and is not related to another disease)
- The child must have dysynchrony
- The child’s ejection fraction must be less than 35%
- Medications do not adequately treat heart failure symptoms, affecting quality of life
- Symptoms must manifest themselves and include:
  - **Babies** - not growing well, breathing too fast, exhibiting heart failure
  - **Toddlers** - not keeping up with others their age, not performing activities they could do before, exhibiting shortness of breath

Risks associated with the procedure include internal bleeding and perforation, but the risks are far less than those associated with a heart transplant. The one caveat is that with any experimental procedure there is no data to show how long the pacing system will work in a child’s body.

To date, Children’s Healthcare has performed 15 such procedures on children, with the youngest child younger than eight months. “Three of the children have not done well; the rest have done beautifully and some have even been removed from the heart transplant list. They have not been hospitalized afterwards, and their symptoms have lessened, though only Jacob’s have gone away completely,” says Dr. Strieper. The procedure has so far met with a 70% success rate, and it is now performed in most major pediatric wards throughout the country.

Dr. Strieper adds, “Biventricular pacing is a very valuable tool. It’s out there; it’s available and parents should ask the cardiologist for more information about the procedure and if their children are candidates.” Jacob’s mother Amy adds, “Never give up hope! Find out everything

you can. Don’t be afraid to ask anything... As a parent, you are your child’s advocate.”

### Berlin Heart

Another procedure that has made more recent headlines is a new **ventricular assist device (VAD)** known as the Berlin Heart. A VAD is a mechanical pump that takes over the heart function of the damaged ventricle and restores normal blood flow to the lungs and rest of the body. The Berlin Heart, also known as **Excor**, does much of the work of the patient’s natural heart without replacing it. The device is implanted surgically under the breastbone (sternum) with 2-4 tubes (cannula). Each of the tubes is tunneled through the heart muscle and chest wall and then attached to the Berlin Heart pump, which is a pneumatic pump. This includes a reservoir for blood collection and a flexible membrane which allows for the pumping action. The bulk of the device rests outside of the body with only the tubes connecting it to the heart. The German-made pump comes in various sizes for a range of patients, including newborns.

Currently, the Berlin Heart is the only artificial heart pump being used for longer term bridge-to-recovery or bridge-to-transplant of infants and young children who continue to deteriorate on standard medical therapy. Children can survive on the Berlin Heart for up to a year. In the past, the usual method of circulatory support in children was **ECMO (Extra-Corporeal Membrane Oxygenation)** set up in the intensive care unit. It requires the child to be on a breathing machine and leaves them immobilized. VAD devices, however, allow children to leave their hospital beds while still recovering.

For years, artificial pumps have been used successfully on adults waiting for transplants. But only recently have US researchers started to focus on bridge-to-transplant or ventricular-assist devices for the tiniest heart patients. Because there are so few children (30-40 cases/year in the U.S.) who would benefit from such a device, there is no commercial interest in producing child size pumps nor the incentive to seek **U.S. Food and Drug Administration (FDA)** approval. The Berlin Heart device has been used 20 times in the U.S. and Canada and in each case it

was used with emergency government approval. The German manufacturer, also known as Berlin Heart, is currently trying to expedite the FDA approval process on humanitarian grounds. Nevertheless, this procedure is relegated to the most desperate cases when conventional medical therapy is not enough.



Serafina Akard, 3 weeks post transplant

In May 2005 at the age of 2 years, **Serafina Akard** became one of the few U.S. patients to benefit from this new medical technology. Serafina Akard was first diagnosed with dilated cardiomyopathy at 18 months after an initial misdiagnosis of pneumonia. Upon proper diagnosis, Serafina was managed for several months with oral medications but when her heart began to fail five months later she was admitted to the intensive care unit at **Lucile Packard Children's Hospital (LPCH)** at **Stanford University Medical Center** in California. Her parents, Michael and Suzanne Akard were told that Serafina would need a heart transplant and that the only way to sustain such a young and fragile heart during the wait was with **ECMO**, the heart-lung bypass machine. Unfortunately, ECMO only supports the heart for about 2-4 weeks and the wait for a donor heart for a child of Serafina's age would be far longer than that.

With few medical options, the team of pediatric cardiologists at LPCH suggested using the Berlin Heart. This radical treatment had been used in 2004 at LPCH on

a 5 month-old baby boy whose heart was affected by a viral infection and getting weaker on ECMO. The Berlin Heart device sustained the baby boy for seven weeks until he received a donor heart. Although still considered an investigative

device and not approved by the FDA, Serafina's physicians were able to get special approval for "compassionate care". A German doctor arrived within a week with the artificial heart pump and monitored its use on Serafina. She was on the bridge-to-transplant device for 8 weeks, until a healthy heart became available on June 26th.

Serafina is doing extremely well following her transplant with zero rejection. Serafina's cardiologist, **Dr. David Rosenthal** of Lucile Packard Children's Hospital points out, "Before the Berlin Heart, Serafina was very fragile and her organs were shutting down." While on the Berlin Heart she was able to make good progress and be in perfect condition to receive a donor heart. Michael Akard adds, "The Berlin Heart provided a successful bridge-to-transplant, to the extent that Sera was able to laugh, play and even walk a little bit prior to transplant. Recovery has been remarkably swift thanks to her strong physical condition while on the Berlin Heart".

Aside from the standard risks of heart

surgery, device infection or malfunction, the Berlin Heart can cause blood clot formation. Blood thinners are usually administered to prevent the possibility of clotting and stroke but this may lead to another problem of excessive bleeding. Dr. Rosenthal adds, "This is a useful but very high-risk device for children with advanced heart failure who are deteriorating on maximum medical therapy. There is a significant complication rate from both the device and the underlying disease. On the other hand, in carefully selected patients, it can be life-saving." Studies have shown a 60-80% success rate in the use of the device, which has primarily been used in Europe to treat viral infections of the heart muscle in children. To date, there have been 240 VAD's implanted worldwide with approximately 127 children implanted with the Berlin Heart. Roughly 12-15 hospitals in the U.S. (e.g. **Children's Hospital of Pittsburgh, Saint Louis Children's Hospital, Lucile Packard Children's Hospital**) and Canada have successfully implanted the device in children.

Similar to the Urowskys and their experience with biventricular pacing, the Akards are trying to spread the word about the Berlin Heart in the hopes that it might save the life of another child. Suzanne Akard advises, "If you have a young child and talk of a transplant comes up, do talk to the doctors about the Berlin Heart. It may not be appropriate for every cardio-myopathy child, but it is absolutely worth asking about."

## CCF Attends MEDTRONIC's Annual Patient Link Conference

CCF was invited to the **Medtronic Foundation's Patient Link Conference** held June 15th in Minneapolis, Minnesota. The purpose of the meeting was to expand the capacity of Patient Link organizations to better serve their constituents. In 2004, CCF was awarded a Medtronic Patient Link grant for its proposed **Parent Empowerment Program**. The conference included speakers from various Medtronic business units, industry leaders, non-profit consultants and 49 other lay advocacy groups. The conference focused on understanding the skills, attributes and resources necessary for sustaining organizational growth and provided opportunities for groups to network and share best practices.



CCF's Lisa Yue & Linda Mears at the Medtronic Headquarters

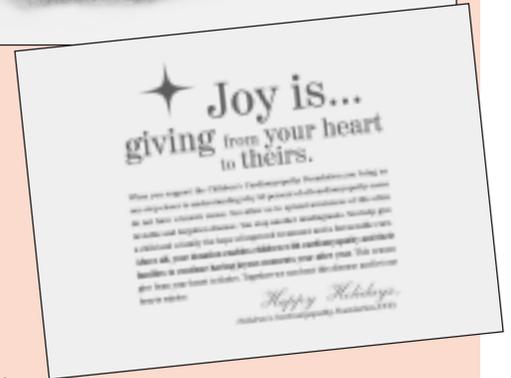
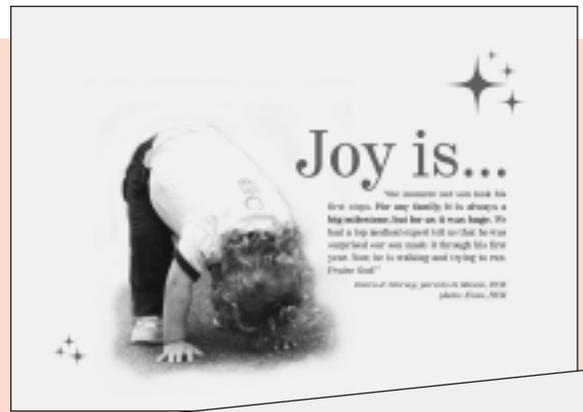
# “JOY” Holiday Appeal Campaign kicks off in November

This holiday season CCF is taking a new approach to our annual appeal campaign with the launch of our **Joy Campaign** and **Annual Friendship Program**. These two initiatives are part of a national fundraising and awareness-building effort to increase CCF's donor network and to strengthen the connection between donors and the families they assist. The theme of this year's annual appeal campaign is “**Joy is...**” which will focus on the experiences of families with affected children and emphasize that joy can still be found in unexpected moments in their lives.

In September, CCF asked its network of families to share those moments of pure joy with us. From the many responses received, CCF was inspired to create holiday appeal cards highlighting these messages of joy. These heart warming cards will be sent to all CCF supporters and potential donors starting mid November.

In order to broaden CCF's donor network and encourage families affected by cardiomyopathy to join in this year's fundraising and awareness efforts, the Annual Friendship Program was developed. The program invites CCF family members and relatives to mail 20 or more holiday appeal cards with a short personal note to their own network of family, friends and colleagues. CCF also offers the option of sending the cards on their behalf with a printed message composed by the family.

For more information on how to order these joyful cards, please contact CCF at 201-227-8852, ext. 901



## Joy is...

“Joy is... the moment our son took his first steps. For any family, it is always a big milestone but for us it was huge. We had a top medical expert tell us that he was surprised our son made it through his first year. Now he is walking and trying to run. Praise God!”

*Daren & Stormy, parents to Mason, DCM*

“Joy is... at the end of each day when I read the girls a story, tuck them into bed and thank God for that wonderful day. Having two children with cardiomyopathy makes me ever more aware of just how precious life is.”

*Audrey, mom to Grace & Megan, DCM*

“Joy is... a happy toddler who is walking, talking and playing when at 4 months the doctors were saying they didn't know if she would make it. It is so wonderful that she is alive!”

*Rebecca, mom to Julia, DCM*

“Joy is... having our son break 100 in golf when a few months ago his participation in sports abruptly ended with his diagnosis. No more baseball, football, or basketball, but there is still golf. What a joyous moment to see him recapture his passion for sports!”

*Victor & Patti, parents to Joey, DCM*

“Joy is... seeing your child do a happy dance in front of two elderly women who never dreamed the child would live to be ten years old!”

*Suzie, mom to Autumn, HCM*

“Joy is... watching him run, kick, throw a ball and try to ride a bike just like any other toddler. Joy is knowing he is a tough kid and will be able to handle anything that comes his way. Joy is knowing that we are not alone in all of this!”

*Jennifer, mom to Andrew, HCM*

To read additional “Joy Stories” sent in by parents of children with cardiomyopathy, please visit our website at [www.childrenscardiomyopathy.org](http://www.childrenscardiomyopathy.org)

## Consider WORKPLACE & COMMUNITY Giving

When making a donation to CCF, don't forget about employer matching gifts and workplace giving programs (United Way or your company annual appeals). In 2004, CCF received \$22,000 from matching gifts alone – enough to fund one pilot research study. Larger corporations customarily give a portion of their

# Third Annual **GOLF CLASSIC** Raises over \$220,000



Mark DeSalvo & Betsy Liozzi with CCF board members Tami Horan & Brian Nold

On Thursday September 15, CCF hosted its **Third Annual Golf Classic** at the **New York Country Club** in New Hempstead, New York. The sold out event helped to raise \$220,765 gross, surpassing last year's total. Heavily supported by the financial service industry, this year marked a tremendous increase in sponsorships from well-established firms.

The Annual Golf Classic is CCF's main fundraising effort, and net proceeds from this year's event will be earmarked for CCF's annual research grant program. Through a competitive grant review

income to charity for tax purposes and your employer may have a charitable or community giving program that CCF can also apply to for funding. In general, corporate foundations are more prone to give to a cause that is recommended by an employee. In addition, if you are member of a community club, social organization or men's/women's association that holds yearly fundraising events, please consider nominating CCF as a beneficiary organization. These are all easy but effective means for raising additional funds for research and education on pediatric cardiomyopathy.

process, each year CCF awards seed funding to investigators interested in furthering medical understanding and treatment advancements on pediatric cardiomyopathy.

Generous people from the tri-state area came out for the all-day event and played golf, enjoyed a delicious barbecue lunch, and ended the evening

with a cocktail reception and three-course dinner. They also took part in a silent auction of over 50 donated items as well as sports memorabilia provided by **Grandstand Sports**.

Donated **Titleist, Ping, Cutter & Buck**, and **Nike** golf items were awarded to the first, second, and third prize foursome winners. First prize went to **Mike Kirsh, Ira Cohen, Stuart Goldstein, and Brian Hewitt**. Second prize winners included **Matthew Glass, Ted Burdick, Dan Allen, and Kevin Ulrich**. **Sam Kim, Jinho Jan, Jim Om, and Chung Lee** were the third prize winners. Other contest winners included **David Pucciarello** (Closest to the Pin), **Clint Kollar** (Men's Longest Drive), **Ann Bell** (Women's Longest Drive) and **Paul Haskel** (Closest to the Line).



Lincoln Isetta & Jason Kirschner



Steve Levitan, Ian Sandler, Tom Janover & James Gereghy



Kevin Ulrich, Mathew Glass, Ted Burdick & Dan Allen

## *Many thanks to our event sponsors -*

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## JOURNAL SUMMARY: Management of Pediatric

**HYPERTROPHIC CARDIOMYOPATHY (HCM)** is a genetic disorder characterized by myocardial hypertrophy (thickening of the heart muscle) without an identifiable cause. HCM is thought to occur 1 in 500 in the general population and remains the most common cause of sudden death in children and adults under 35 years old.

### Diagnosis

Since 1989, at least 10 genes and 150 mutations have been identified to cause HCM. Three genetic defects account for the majority of cases and include the beta MHC, myosin-binding protein C, and troponin T genes. 90% of patients with a beta MHC gene defect will present with HCM in childhood whereas with the other genetic mutations, HCM does not normally present until adulthood. While genetic testing exists, it remains expensive on a diagnostic level and is confined to only a few research labs, thus limiting its availability to most cardiologists treating children. In addition, published data indicate that clinical genetic testing for sarcomeric mutations only account for 60% of HCM cases. Prenatal molecular diagnosis of HCM has recently been proven feasible, but because of the genetic and clinical variability of the disease, it may not become routine.

Two-dimensional echocardiography remains the primary diagnostic method for identifying children with HCM, with an increased focus on diastolic performance (the "resting" rate). Diastolic abnormalities have been shown to be frequent in children with HCM and to be the main cause for exercise intolerance. Tissue Doppler imaging is also a useful diagnostic tool for

predicting adverse clinical outcomes such as death, cardiac arrest and ventricular tachycardia (life-threatening arrhythmias).

Sudden death can occur in children, although the risk factors are likely different than in adults. Clinical evaluations should therefore include annual 24 or 48 hour Holter monitoring for identifying abnormal heart rhythms (arrhythmia) and assessing the risk of sudden death in young patients with HCM. Additionally, cardiopulmonary stress testing (exercise stress test) performed in a controlled hospital setting has recently been proven safe for HCM patients and is useful for identifying young patients at risk for sudden cardiac death. Increasingly, magnetic resonance imaging (MRI) is more commonly being used to determine the severity of heart muscle damage. These evaluative tests aid the physician in choosing a treatment regimen for the patient.

Current screening recommendations for first-degree relatives of affected family members include annual evaluation by echocardiogram in children ages 12-18 years and ongoing evaluation at five-year intervals. Evaluation of children younger than 12 should be dictated by family history and clinical assessment. Affected children and young adults should be observed annually.

### Management

Primary goals in the treatment of pediatric HCM are alleviation of symptoms and prevention of sudden death. Determining the risk category of a patient is an important part in determining their treatment. The highest risk for sudden cardiac death has been associated with the following observations: previous cardiac arrest, sustained arrhythmia, family history of premature HCM related cardiac death particularly if sudden in a close relative or multiple relatives, identification of a high-risk genetic mutation, abnormal blood pressure response to exercise and/or non-sustained arrhythmia from a Holter test.

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**“While genetic testing exists, it remains expensive on a diagnostic level and is confined to only a few research labs, thus limiting its availability to most cardiologists treating children.”**

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In regards to medical management, physical activity restrictions remain important. The latest **American Heart Association** statement on exercise guidelines advises patients with genetic cardiovascular disease to refrain from competitive athletics and isometric activities.

For patients who have symptomatic HCM, beta-blockers continue to be the frontline therapy. Disopyramide, either

# Hypertrophic Cardiomyopathy

Full length article available on [www.pubmed.gov](http://www.pubmed.gov)

Current Opinion in Cardiology 2005, 20:80-83

Anji T. Yetman, University of Colorado Health Sciences Center

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alone or in conjunction with a beta-blocker, has been shown to be useful in improving heart function. Verapamil is best reserved for patients who do not have significant symptoms related to obstruction because death has been reported to occur in patients receiving a calcium channel blocker.

Surgical myectomy continues to be the standard therapy for children with severe symptoms from left ventricular outflow obstruction (constricted blood flow due to narrowing of the pumping chamber) and who do not respond to drug therapy. This procedure provides long-term relief of symptoms, and if performed at an experienced medical center is a relatively low risk surgical procedure.

On the other hand, alcohol septal ablation, which is sometimes performed in adults with HCM, is a high-risk procedure for left ventricular outflow tract obstruction. The procedure has not been subjected to randomized, controlled studies or long-term follow-up and is therefore not recommended for children with HCM.

While a diagnosis of HCM usually leads to an evaluation for an implantable cardiac defibrillator (ICD), careful patient selection is advised. Because there is still the risk of complications during implantation, this therapy should be considered for secondary prevention after an episode of resuscitated sudden death or for primary prevention of patients with multiple risk factors. Advances in

implantable defibrillators now make this therapy feasible in younger children.

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**“Earlier diagnosis leads to earlier surveillance and management, which must continue over a child’s lifetime and can greatly affect long-term quality of life.”**

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## Conclusions

There are important differences from adults in the approach to the diagnosis and management of HCM in children and adolescents. Care regarding evaluation and therapy must be taken given the potential life-long implications. Earlier diagnosis leads to earlier surveillance and management, which must continue over a child’s lifetime and can greatly affect long-term quality of life. Advances in the precision and use of noninvasive evaluation are important, as well as the development and evaluation of safe and effective therapies aimed at slowing the progression of the disease, alleviating symptoms and lowering the risk of sudden life-threatening events.

## Mutated Gene Causes Serious Heart Disease in Newborns

An international research group led by Professor **Manfred Kilimann** of **Uppsala University** in Sweden has identified the genetic cause of a severe heart disease in newborn children. The findings were published in the June issue of the **American Journal of Human Genetics**.

Cardiomyopathies are diseases of the heart muscle tissue that can lead to heart failure and sudden death. Childhood cardiomyopathy affects one in 100,000 children nationally. This is a group of complex diseases resulting from more than 200 known causes.

Professor Kilimann’s team studied a particular cardiomyopathy known as “fatal congenital nonlysosomal heart glycosinosis” (FCNHG). Children with this disease have an enlarged heart and abnormal heart rhythms, and often die in the first few weeks of life. It is a glycogen storage disease, with some similarity to Pompe disease.

The researchers, including partners in Germany, Britain, and the U.S., determined that the problem was in a gene (PRKAG2) involved in energy metabolism and known to cause a milder form of cardiomyopathy in teens and young adults. All three affected infants studied had the same mutation.

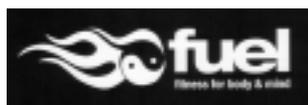
Heart transplantation is currently the only cure for FCNHG, and the study doesn’t change that. However, it does provide hope for more reliable diagnosis and genetic counseling. Also, the study showed that the mutation was not passed from parent to child, giving parents of an affected child reassurance that the risk of having another affected child is low.

*Reprinted with permission from the Summer/Fall 2005 edition of the “Orphan Disease Update” from the National Organization for Rare Disorders ([www.rarediseases.org](http://www.rarediseases.org)).*

## CCF Participates in San Francisco Charity Run

On July 31, CCF was one of 28 **Cause to Run** charities participating in the annual **San Francisco Marathon**. 12,000 runners registered for this year's marathon, half marathon, and 5K runs. The event also encouraged participants to pledge support and raise funds for their charity of choice. CCF's team had 16 runners for our cause this year, double the number of last year. The captain that led our team's finish was **Chris Achuck**, brother-in-law of CCF board member **Raymond Yue**. More than \$1,000 was raised for CCF this year.

## Spinning for Research



On  
September  
17, **Fuel  
Fitness for**

**Body and Mind** in Cresskill, New Jersey hosted a morning spin class marathon to benefit CCF. Fuel Fitness donated 100% of its registration fees and marathon pledges to CCF. **Gerard Bochese**, owner of the gym, felt the event was a great way to get in shape and support a worthy cause. Fuel Fitness has been an ongoing supporter of CCF's work, acting as a sponsor and auction donor at our annual golf event, promoting our "cureband" awareness wristbands and helping to broaden our community supporters.

## Shop for a Cause

On October 27, CCF participated in **Bloomindale's, The Shopping Benefit**. The special shopping day was held at Bloomindale's four New Jersey stores and entitled ticket holders to 15-20% savings on merchandise, fashion presentations, book signings, sweepstakes, children's activities and culinary tastings. CCF received 100% of the proceeds from every ticket that CCF sold (\$10 each) and an additional \$5 for every ticket holder that attended the event.



# SPOTLIGHT on CCF FAMILIES

## A Mother's Tribute to her Daughter

By Janie Dale, mother to Vanessa,  
12/1/04-4/7/05 DCM



Vanessa Dale

Our beautiful baby girl, Vanessa Katherine, was born Dec 1, 2004. Christmas and New Years was simply the best as we traveled between families introducing our healthy family... so we thought. A follow up visit to her pediatrician at 2 months was the entrance to the most gut-wrenching roller-coaster ride imaginable when Vanessa was diagnosed with dilated cardiomyopathy (DCM). There were a few bad weeks at the hospital, then a few good weeks at home, and then back and forth for two months. On her last checkup we were told she was in severe heart failure. Less than 18 hours later, she died in my arms on April 7, 2005. While she was only four months old it seems as though she had shared a lifetime with us.

While we miss our little angel more than we could possibly express, my husband and I realize that we must choose to either envelop ourselves in self-pity and withdraw from the world, or latch on to the deeper meaning and a better understanding of the frailty of life and to embrace the remaining moments we have. With the support of friends and family, we have chosen the latter and are looking for ways to add meaning and purpose not only to Vanessa's life, but to ours as well. Throughout our journey, the **Children's**

**Cardiomyopathy Foundation (CCF)** and its forum of families with affected children provided support at a desperate time for us. Following Vanessa's passing we have been inspired by the CCF and the **Chad Foundation**, two organizations founded by parents who lost children to the disease. This January we are teaming up with both CCF and the Chad Foundation to conduct a free echo-screening event at a nearby high school for 100-200 kids, during which we will host some fundraising activities to support CCF's research efforts. We hope that this event will help identify any at risk children and increase awareness of pediatric cardiomyopathy.

Additionally, we have discussed with cardiologists at **Texas Children's Hospital** in Houston our desire to help fund the establishment of a database in their cardiomyopathy lab that would be dedicated to sorting through the genetic links of various heart diseases.

While we will never be able to hold our precious Vanessa again, through these efforts, her life can continue to touch and affect others, perhaps long after we are gone.

## A MOM'S RUN for her Son

On October 30, **Jennifer Dickson** participated in a 5-person relay team in the **Cape Cod Marathon**. She ran in honor of her 18-month-old son Drew and all children living with cardiomyopathy.

At the age of six months, Drew was diagnosed with Hypertrophic Cardiomyopathy (HCM) as well as Wolf-Parkinson-White syndrome. One year later, his heart shows no signs of cardiomyopathy, though the other heart disease remains. The date of the run ironically coincided with the date of Drew's diagnosis and hospitalization last year, and Jennifer was determined to mark this anniversary with a positive,

life-affirming activity.

In preparing for her run, Jennifer wanted to design a t-shirt that would bear the names of all the children in CCF's family support network. When she shared this idea with other CCF parents, the idea was met with such enthusiasm that now Jennifer is producing additional t-shirts for purchase through CCF. With each



Jennifer & Drew Dickson

purchase, a portion of the t-shirt price will be donated to CCF's patient support fund.

Although Jennifer shuns publicity for herself, she says "I am glad that the attention is creating awareness of the disease. I would do anything for the kids." While her own son is fortunate and will be able to participate in sports, Jennifer knows that this is not the case for most of the other affected children.

## HEARTS OF HOPE Benefit in New Jersey raises nearly \$20,000

**Peter and Danielle Torok**, CCF family members from New Jersey hosted the first annual **Hearts of Hope Benefit** raising close to \$20,000 for CCF. The Toroks became involved with CCF after their daughter Anna was diagnosed with dilated cardiomyopathy at the age of 6 months. Now 2 1/2 years old, Anna has made a remarkable recovery without the need for a heart transplant, and continues to thrive on a regimen of heart medication. "From where we stood two years ago, upon Anna's initial diagnosis, to where we stand today we feel very blessed!" says Danielle.

Hoping to create awareness and fund more research on pediatric cardiomyopathy, the Toroks planned a cocktail reception and raffle for October 16. Held at the Women's Club of Caldwell, New Jersey, 81 people from the surrounding area attended the weekend affair. The event was supported by in-kind donations from local businesses, family members and friends. In true volunteer spirit, family and friends also served as bartenders, waitresses, and entertainers that night.

"In addition to raising money, I think we have definitely made strides in increasing awareness about pediatric cardiomyopathy. We look forward to having the Second Annual Hearts of Hope Benefit next year," added Danielle.

## A MASQUERADE PARTY in CCF's Honor

When Audrey Callahan's high school friends, Stacy Galarza and Whitney Bryant, decided to throw a 30th birthday party, CCF family member Audrey Callahan suggested CCF as the beneficiary organization.

**Doug and Audrey Callahan** and their two children, Grace, 2, and Megan, 1 both have dilated cardiomyopathy. According to Audrey, who has been a CCF member since 2003, CCF has provided not only information about the disease but also assurance that she and her husband are not alone in their struggle. She thought a themed dinner party would be a fun way to help support CCF's work.

The masquerade themed fundraiser was held on June 4 at TJ's Landing in Buckhead, Georgia. Nearly 40 people attended with everyone having a wonderful time celebrating with the Callahans and supporting CCF.

*CCF offers fundraising packets and guidelines for other families who are interested in hosting similar fundraising events.*



Stacy Galarza, Audrey Callahan & Whitney Bryant

Address Service Requested



Tel: 866.808.CURE  
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www.childrenscardiomyopathy.org

The Children's Cardiomyopathy Foundation (CCF) is a national non-profit health organization focused on pediatric cardiomyopathy - a chronic and life-threatening heart disease that affects children. CCF is dedicated to improving treatments and finding cures through the support of medical research, education, and increased awareness and advocacy.

P.O. Box 547, Tenafly, NJ 07670

The Newsletter of the Children's  
Cardiomyopathy Foundation

Volume 2, Number 2 • Fall 2005 • Winter 2006

# Heart to Heart

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# Welcome to Heart to Heart

The Newsletter of the  
Children's Cardiomyopathy Foundation  
Volume 2, Number 2 • Fall 2005 • Winter 2006

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### • Foundation News

**Cover:** CCF families benefit from new medical advances – biventricular pacing & the Berlin Heart ventricular assist device.

### • Family Information

**Page 2:** Genetic discrimination is a key concern in genetic testing.

**Page 3:** First local support group launched in Georgia.

### • Medical News

**Pages 8 & 9:** Review recent medical journal summaries on pediatric cardiomyopathy.

### • Fundraising Update

**Page 7:** Third Annual Golf Classic raises over \$220,000.

**Page 10 & 11:** Learn about the fundraising and awareness efforts of CCF family members.

FALL 2005 • WINTER 2006

## CCF Research Goals

The principal mission of CCF is to support biomedical and clinical research towards the early and precise diagnosis, the effective and appropriate treatment and the ultimate prevention and cure of pediatric cardiomyopathy.

*From a research and treatment perspective, the priorities are:*

- 1) to support studies to better understand the disease
- 2) to set up research resources to encourage promising research
- 3) to consolidate treatment expertise into regional centers of excellence

## Making a Difference... CCF 2005-2006 ANNUAL APPEAL Campaign Underway

As the year draws to a close, it is a special time for reflection. CCF is celebrating the holiday period with the launch of our **Joy Campaign and Friendship Program**. See page 6 for more information about this year's fundraising and awareness initiative.

**In this season of giving, please remember CCF with a gift.**

To make a donation, please contact CCF at 866-808-CURE  
or [info@childrenscardiomyopathy.org](mailto:info@childrenscardiomyopathy.org)