All of us who face cardiomyopathy struggle for answers. What can we do to accelerate research and improve treatment? What can we do to raise awareness? Often there are more hurdles than answers: funding, enrolling study participants, sharing research findings and engaging researchers. CCF is working to address these needs by providing research grants, establishing a tissue and blood repository, planning scientific workshops and reaching out to researchers to encourage their participation in our work.

This season’s newsletter is devoted to genetic testing and research participation. Many times participating in a study or genetic test is easy and only involves sending in a blood sample during your child’s routine visit. Nonetheless, genetic testing and research participation are new topics to most families and this newsletter attempts to demystify some key features - what is it, where is it done and the benefits of getting it done.

You can also support research by asking your doctor what studies your family should enroll in. Because your doctor cares about your child’s health, just by asking, you will raise awareness and engage doctors. Everyone helps when they contribute to this cause - every question puts focus on the cause, every dollar helps us get closer to a cure. Thank you for your past contributions and for working with us to find more answers to pediatric cardiomyopathy.

Respectfully,

Lisa Yue
President & Founder
The Children’s Cardiomyopathy Foundation (CCF) has teamed up with Healthcare Branding Group, Inc. (HBG), a leading brand development and creative communications practice located in Chicago. The partnership will provide CCF with a combination of strategy development and creative communication services devoted to enhancing the overall awareness and recognition of the Foundation.

This summer, CCF was awarded a generous Parent Empowerment Program grant from the Medtronic Foundation’s Patient Link division. The goal of the Parent Empowerment Program is to build a support network for children with cardiomyopathy and their families while providing access to patient friendly information. The program will include a combination of print and electronic educational materials with medical information and guidelines for living with the disease.

HBG and CCF will work together to develop accessible, up-to-date, and easy to understand information in order for families to be informed and motivated to better manage their child’s care. To date, there have not been any patient materials or support services developed for families and children affected by this chronic disease. A national research study will begin in early 2005 with the program being rolled out later that year.

HBG will also be working with CCF on a number of initiatives to advance the Children’s Cardiomyopathy Foundation’s overall structure, resources, process and brand. The firm has extensive experience and knowledge of healthcare medicine and science combined with business, marketing and philanthropy. For over twenty years, HBG has been working with a variety of healthcare clients to identify emerging opportunities to enhance their brand.

For more information on HBG please visit www.healthcarebranding.com

IMPORTANT Parent Survey This Month!

CCF families, we need your help! Your opinion is important to us so please visit our web site, www.childrenscardiomyopathy.org, in early December to participate in our Parent Survey. As we continue to develop materials to support and educate parents of children with cardiomyopathy, we want to incorporate your thoughts and experiences into our work. Thank you in advance for your time and consideration.
CFC RESEARCH GRANT PROGRAM Launched

This summer CFC launched a national research grant program with a formal request for proposals for innovative basic, translational and clinical research relevant to the cause or treatment of cardiomyopathy in children. CCF will award each selected investigator a maximum of $50,000 in total direct costs over a one or two year study period.

The grant award program is designed to provide seed funding to investigators for the testing of initial hypotheses and collecting of preliminary data to help secure long-term funding by the National Institute of Health and other major granting institutions. This year’s grant deadline was October 29 and applications are currently being reviewed. CCF’s Medical Advisors and Board of Directors will make award decisions based on a proposed study’s scientific excellence, relevance to pediatric cardiomyopathy and impact to health outcomes in children. Final decisions will be made in December with final disbursement of funds by year-end. CCF plans to distribute at least $125,000 in this grant award period. More information about CCF’s grant guidelines and application for the 2005 review period are online at www.childrenscardiomyopathy.org/main/grants.htm

CCF Funded Research, continued from page 1

RAAS Gene Polymorphisms Influence Cardiac Remodeling in Children with Hypertrophic Cardiomyopathy, Seema Mital, MD; Etiology-Specific Outcomes in Pediatric Hypertrophic Cardiomyopathy, Steve Colan MD; and Impaired Functional Status in Children with Cardiomyopathy: A Report From the Pediatric Cardiomyopathy Registry, Lynn Sleeper, ScD.

In addition to the AHASS presentations, eight manuscripts are being submitted to top rated scientific journals for publication in 2005. Most of these publications evolved from CCF’s partnership with the National Heart, Lung and Blood Institute funded Pediatric Cardiomyopathy Registry to promote further analysis and dissemination of findings on the disease. The resulting manuscripts and lead authors that will acknowledge CCF include:

Molecular Genetic Stratification of Familial Hypertrophic Cardiomyopathy
- Wendy Chung, MD, PhD
This paper introduces a new clinical genetic screening test that was developed to evaluate families affected with hypertrophic cardiomyopathy with a high incidence of sudden death in young adults. The abstract was presented at the American College of Cardiology and Eastern Society for Pediatric Research 2004 sessions.

Incidence & Etiology of Dilated Cardiomyopathy in Children
- Jeffrey A. Towbin, MD
This study sheds light on the incidence, etiology and survival rates associated with dilated cardiomyopathy in childhood as it relates to gender, age, ethnic background and disease characteristics.

Predictors of Etiology in Pediatric Cardiomyopathy
- Gerald F. Cox, MD, PhD
This paper examines the percentage of families with diagnosed children that have a known etiology to the disease and the factors that determine this.

Etiology-Specific Outcomes in Pediatric Hypertrophic Cardiomyopathy
- Steven D. Colan, MD
This research looks at the frequency of various causes of hypertrophic cardiomyopathy in children and the corresponding etiology-specific survival rates.

Outcome Predictors in Pediatric Hypertrophic Cardiomyopathy
- Steven E. Lipshultz, MD
This outlines patient factors such etiology, age, and clinical symptoms, at the time of diagnosis that may affect the subsequent risk of death or heart transplant in children with hypertrophic cardiomyopathy.

Noonan Syndrome & Cardiomyopathy: Outcome Predictors
- Bonnie A. Salbert, MD
This longitudinal study examines the natural history, risk factors and survival rates associated with children with cardiomyopathy related to Noonan’s Syndrome.

Characteristics of Duchenne & Becker Muscular Dystrophy in the Pediatric Cardiomyopathy Registry
- David Connuck, MD
This study attempts to describe the clinical characteristics and determine the relevant outcome factors for Duchene and Becker Muscular Dystrophy, which are x-linked forms of cardiomyopathy.

“Exciting inroads in the research and education area have been made with CCF’s support.”

An Analysis of Medical Therapy Offered to Children with Dilated Cardiomyopathy
- William Harmon, MD
This analysis reviews the current state of therapy for children with dilated cardiomyopathy and supports the need for specific management guidelines for pediatric heart failure patients.
**The Basics of GENETIC TESTING**

Genetic testing is taking on a more important role in the overall management of cardiomyopathy. A genetic test is defined as an analysis of human DNA, RNA, chromosomes, proteins, and certain metabolites that allows for earlier identification and diagnosis of individuals at risk of developing a disease. Genetic testing is usually preformed for a variety of reasons: to confirm a clinical diagnosis or prognosis, to establish the cause and inheritance pattern of the disease, to identify at risk relatives in need of regular follow-up and to identify relatives who did not inherit the causative mutation, freeing them of ongoing monitoring. If a genetic mutation can be identified in an affected individual, all other family members can be screened via a blood test to see if they have the same mutation and are more susceptible to developing symptoms later in life. Families can also make informed family planning decisions with the possibility of prenatal testing or pre-implantation (in vitro fertilization) screening to ensure that future children would not be affected.

**What is the Genetic Testing Process?**

Before any genetic testing is done, participants need to familiarize themselves with all the steps. The first step is to discuss the appropriateness of genetic testing with a cardiologist and/or geneticist and to determine which types of tests are applicable. At this point, the role of a genetic counselor is important in ensuring that participants have a full understanding of the benefits, risks and implications of the test results for both themselves and for other family members. During the process, participants will need to determine whether and how they will disclose the results to other family members and what follow-up medical care will be necessary.

**Where is the Testing Done?**

Once it has been determined that genetic testing is appropriate, a decision will need to be made about where it is performed. Due to the complexity of most diseases, no laboratory will offer comprehensive testing for all genetic mutations causing a disease. A doctor, geneticist or genetic counselor should be able to recommend various testing facilities based on the type of testing required. Depending on the patient's objectives and timeframe for answers, genetic testing can be performed in either a commercial or a research laboratory.

Commercial diagnostic laboratories are clinically approved for genetic tests and generally need to meet review standards under the Clinical Laboratory Improvement Amendments (CLIA) to ensure the quality of their testing procedures. These labs can determine, on a fee per test basis, whether specific genetic mutations are present in an individual. Although the turnaround time is much quicker than a research lab, the costs of these tests can be high and insurance may not cover it. The limitation with commercial labs is that they only offer testing for the most common or known mutations for a genetic disorder. Therefore, if a family has a rare genetic disorder with an unclassified cause or without a family history (i.e., sporadic case), genetic testing at a diagnostic lab may not provide the answers that are needed.

An alternative to diagnostic testing is to enroll in a research study at a larger teaching hospital or medical center. As part of the research, the laboratory will offer free genetic testing as an incentive to enroll more patients into their study. The advantage of this arrangement is that these laboratories will screen patients for many genes, including rare types, until a potential mutation is identified. The disadvantage is that it may take years to complete without any guarantee of benefits from the study. Even if a genetic variant is identified, it may take several more years of researching other families before it can be confirmed as a disease-causing mutation. Additionally, due to the new patient privacy laws and the implications of genetic mutation identification, many research laboratories will not report the discovery of a mutation to the patient or their doctor unless a CLIA-certified laboratory confirms the results. This would then entail some costs to the enrolled patient.

**Testing Costs and Insurance**

As mentioned, genetic testing done at a commercial laboratory can be very expensive because it is a rare test that is labor-intensive and involves multiple levels of review. Participants will need to evaluate the pros and cons of paying for the tests out of pocket or using medical insurance. Even if an individual decides to have their insurance cover the fees, not all genetic tests are covered. While federal and state laws prohibit discrimination based on genetic information, some individuals still choose not to bill their insurance to avoid any potential discrimination and privacy issues that may arise for other family members (sisters, brothers, children). On the other hand, disclosing positive test results can help certain individuals get coverage on preventative or other medical interventions specific to the diagnosed disorder.

More information on genetic testing can be found at the Genetic Health website (www.genetichealth.com).
Clinical Genetic Testing for Pediatric Cardiomyopathy

Cardiomyopathies are the most common disorders resulting in heart failure, with dilated cardiomyopathy (DCM) responsible for the majority of cases.

Other forms of cardiomyopathy, such as hypertrophic cardiomyopathy (HCM), are also important causes of heart failure. Over the past few years, breakthroughs in understanding the basic mechanisms have occurred and it is now becoming increasingly important that the genetic basis for these disorders be clearly recognized by caregivers and scientists. It has also become clear that these diseases are genetically highly heterogeneous, meaning that multiple genes have been identified for each of the major forms of cardiomyopathy and that most patients have unique mutations in these genes. These data suggest that a simple genetic diagnosis of most patients with cardiomyopathy is not practical with current technologies.

At least 30-40% of DCM cases and more than 50% of HCM cases are inherited, but currently there are few options for genetic testing for patients with inherited cardiomyopathies and even less options for those with sporadic (i.e., no obvious family history) forms of the disease. This is a result of the large number of genes associated with most cardiomyopathies, as well as the large sizes of many of these genes. For example, one study involving the screening of a large cohort of HCM patients for mutations in sarcomere proteins (the proteins that make the heart muscle contract) lead to a genetic diagnosis in only about 30% of unrelated index patients, although when only cases with a family history of HCM were considered this increased to 57%. This study demonstrates that even in HCM, which involves considerably fewer genes than DCM, the rate of mutation detection is too low to be acceptable for most clinical genetic tests. For autosomal dominant DCM, where the patient has one copy of the defective gene while the other is normal, it appears that for each of the genes identified to date, mutations are identified in only 1-2% of patients screened.

Patients with X-linked cardiomyopathies or patients with cardiomyopathy associated with conduction disease do however have some clinical testing options. These forms of cardiomyopathy appear to be associated with mutations in a small number of genes, allowing certified diagnostic (CLIA) laboratories to offer genetic testing. CLIA stands for Clinical Laboratory Improvement Amendments and provides a set of rules by which diagnostic laboratories are regulated. To receive CLIA certification a diagnostic laboratory must participate in an accreditation program, such as that run by the College of American Pathologists, to ensure that the laboratory is operating at high standards. Patients identified with X-linked DCM (XLCM or Barth syndrome) can be screened for mutations in dystrophin and G4.5 (TAZ), the latter being strongly advised if there is evidence of concurrent neutropenia or 3-methylglutaconic aciduria. In addition, mutations in G4.5 have been detected in patients with pure DCM (without other clinical manifestations of Barth syndrome), endocardial fibroelastosis and left ventricular noncompaction. There are several CLIA-certified laboratories that offer testing for DNA variants in these genes.

In a patient presenting with DCM associated with conduction disease or with left ventricular noncompaction, there is a strong likelihood that a mutation in lamin A/C will be detected. Currently there are several CLIA approved laboratories that offer lamin A/C testing in the United States. A number of laboratories also offer testing for mitochondrial mutations that are associated with cardiomyopathies. The John Welsh Cardiovascular Diagnostic Laboratory at the Baylor College of Medicine has recently released tests for a number of genes linked to cardiomyopathies, including G4.5 (TAZ), SCO2, Lamin A/C, and SURF1, and will continue to add new tests periodically.

“Clinical evaluation of these individuals is important regardless of their perceived health status, since many cardiomyopathies have been shown to demonstrate incomplete penetrance, meaning that some individuals carrying a genetic mutation may be healthy, and express the disease differently, ranging from sub clinical to severe heart problems.”
Clinical Genetic Testing, continued from page 5

diagnostic service by the Laboratory for Molecular Medicine at Harvard Medical School in the United States, as well as by other laboratories around the world.

Although the probability of identifying a disease-associated mutation in an individual affected with some forms of cardiomyopathy, such as DCM, is currently small, important steps can be taken to improve the likelihood of success. All patients presenting with a cardiomyopathy that is known to be of familial origin or is thought to be sporadic in nature should be given a thorough genetic evaluation, which should include pedigree analysis. The patient should be questioned in detail regarding his family medical history, in order to identify other family members who may also be affected or at risk. At a minimum, all first-degree relatives of the patient (i.e., parents, siblings, and children), as well as all family members suspected of being affected or being at risk should be clinically evaluated. Clinical evaluation of these individuals is important regardless of their perceived health status, since many cardiomyopathies have been shown to demonstrate incomplete penetrance, meaning that some individuals carrying a genetic mutation may be healthy, and express the disease differently, ranging from sub clinical to severe heart problems. Once a complete family medical history is taken and clinical evaluations of family members performed, analysis based on the resulting data may indicate a possible mode of inheritance, as well as define the spectrum of diseases that must be considered. This information can then be used to narrow the number of possible candidate genes for mutational analysis. In larger families, the generation of a complete and accurate pedigree may allow linkage analysis to be performed, further narrowing the possible candidate genes, assuming that sufficient family members are available and willing to participate in such a study. By narrowing the list of candidate genes to be screened, the probability of successfully identifying the disease-associated mutation can be greatly improved. Due to the complexity of genetic analysis, its interpretation, and the psychosocial issues surrounding genetic analysis, we strongly recommend that all patients and their families receive genetic counseling. A true partnership between the patient, clinician, and genetic counselor will provide the highest standard of patient care.

The John Welsh Cardiovascular Diagnostic Laboratory at Baylor College of Medicine in Houston, Texas provides diagnostic testing for patients with acquired (viral) or inherited forms of heart muscle disease. It was established in 2001 by Jeffrey A. Towbin, MD, who is the laboratory’s medical director, Karla R. Bowles, PhD, FAMG, laboratory director, and Neil E. Bowles, PhD, assistant director. The genetic tests currently offered focus on genes associated with pediatric cardiomyopathies, such as those in patients with Barth, Leigh syndromes or Emery-Dreifuss muscular dystrophy. In addition the lab offers testing for dilated cardiomyopathy, left ventricular non-compaction, and endocardial fibroelastosis. For more information, please contact Dr. Karla Bowles at 832-824-4155 or visit their website at www.bcm.edu/pedi/cardio/research/welsh.html

New Partnerships, continued from page 1

American Heart Association (AHA): Working with the Cardiovascular Disease in the Young Council (CDYC), one of 13 AHA councils, CCF and top specialists in the field will contribute their knowledge and expertise to developing an online and printed pamphlet on pediatric cardiomyopathy for the public. Spearheaded by CCF, the project will be managed by Dr. Catherine Webb, Vice Chair of CDYC, and Dr. Kathryn Taubert, Vice President of Science & Medicine at the AHA. The resource, to be available early 2005 on the AHA website, will cover: types of cardiomyopathy, causes, symptoms, diagnosis, treatment, research, prognosis, living with the disease and offer resource listings and a glossary. The AHA is the largest and most influential national health agency disseminating information about cardiovascular diseases and stroke. AHA’s national call center receives more than 53,000 calls per month and their website attracts close to 5 million unique visitors.

National Society of Genetic Counselors (NSGC): Together with a team of genetic counselors from John Hopkins, University of Colorado, University of Chicago and the National Society of Genetic Counselors, CCF will support the development of a booklet targeted to newly-diagnosed or at risk children with cardiomyopathy. Due to the complexity of the disease, many affected children do not have a clear understanding about what it means to live with a chronic heart disease or may have difficulties articulating their questions and concerns. This booklet, the first of its kind, will provide parents as well as health care professionals with an age appropriate resource to explain the disease in an easy to understand manner. Topics will include: the disease, the heart, common symptoms, causes, genetics, evaluation, treatment and lifestyle as it relates to cardiomyopathy in children. More information about ordering the book will be available on CCF’s website in the Spring of 2005.
NEW Research Studies

The below research abstracts highlight two different types of studies currently recruiting patients. One is a National Institute of Health sponsored study at their clinical center in Maryland and the other is an institution specific, privately funded study in Texas.

Isoproterenol Challenge to Detect Arrhythmogenic Right Ventricular Cardiomyopathy

Location: National Institute of Health Warren G. Magnuson Clinical Center - Bethesda, MD

Study Summary: Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a familial heart muscle disease that can cause sudden death because of abnormal heart rhythm (ventricular tachycardia) or heart failure due to weakened heart muscle. It is important to diagnose ARVC in affected families because treatment such as implantable defibrillators may prevent sudden death. However, ARVC is hard to diagnose with current imaging and diagnostic techniques. This proposed study will examine the accuracy of a new test using the drug isoproterenol to identify heart rhythm problems, and therefore confirm diagnosis of patients with known ARVD and screen family members who may have the disease but do not have clear-cut evidence of it. Study participants will have blood tests, cardiac tests, including imaging studies of the heart, in addition to the drug test.

Eligibility: ARVC patients and their family members (age greater than 5 years).

Study Period: Started May 20, 2004

Subject Compensation: NIH is responsible for all testing expenses. Patients are responsible for travel expenses.

Investigator: Thea McAreavey, MD, FACC
Tel: 301-496-9320
Email: dmcareavey@cc.nih.gov

Accuracy of a Screening Echocardiogram in Detecting Idiopathic Hypertrophic Cardiomyopathy During the Athletic Pre-Participation Physical

Location: Baylor University Medical Center - Dallas, TX

Study Summary: Many athletes have died from idiopathic hypertrophic cardiomyopathy (HCM) while participating in sports without any preceding symptoms and a normal pre-participation physical examination. Recently, portable echocardiogram machines have become available to detect HCM and prevent sudden death, but the accuracy of such machines and the use of them in the pre-participation physical have not been studied. This proposed study will evaluate the accuracy of the screening echocardiogram in patients with known HCM among other athletes being screened. The test will determine how many of the HCM known patients were detected as “not normal” and how many had false negatives. Of those athletes that are screened, it will also determine how many had false positive screening echocardiograms by correlating their results with a follow up full echocardiogram.

Eligibility: HCM patients (age 10-25 years) with no surgical procedures related to HCM (no chest scars), be physically able to walk and run, and with a confirmed HCM diagnosis by echocardiogram within the last 2 years.

Study Period: May 2005 (exact date TBA)

Subject Compensation: $200 honorarium for those that qualify and complete the study. All travel expenses for the subject and one parent will be covered.

Investigator: Fabian E. Pollo, PhD
Tel: 214-820-6300
Email: fabianp@baylorhealth.edu

Before deciding to participate in research, important articles to read include:

- Medical Decision Making: Informed Consent in Pediatric Research
  -Congenital Heart Information Network (www.tchin.org)

- Informed Consent: Participating in Genetic Research Studies
  -Genetic Alliance (www.geneticalliance.org)

- Genetic Information, Privacy and Discrimination
  -National Society of Genetic Counselors (www.nsgc.org)

To subscribe to Heart to Heart, the newsletter of the Children’s Cardiomyopathy Foundation, please email name and mailing address to newsletter@childrenscardiomyopathy.org with the title “Subscribe” in the header.
Hypertrophic cardiomyopathy (HCM) is the most common single-gene cardiac disorder and it is predominately inherited in an autosomal dominant manner. Each child of an individual diagnosed with HCM has a 50% chance of inheriting the condition. Because of the extreme variability in how HCM presents, it has been difficult to determine diagnosis for pre-symptomatic individuals. This has been especially problematic for young children of a diagnosed individual. Although the genetic mutation may have been inherited at the time of conception, it may take years before it fully expresses as HCM. Therefore, a child that is evaluated clinically and found to have normal heart function could still develop HCM later in life. For years, researchers have been trying to develop a reliable genetic screening test to confirm whether suspected individuals have an HCM-causing genetic mutation and be predisposed to complications from the disease.

Common Mutations
Based on numerous genetic studies of HCM among young adults and adults, it has been found in both familial and sporadic (i.e., no family history) cases that HCM is predominately a disease of the cardiac sarcomere proteins. HCM can be caused by mutations in any one of 11 genes (MYH7, MYBPC3, TNNI3, TPM1, ACTC, MLC2, MLC3, RyR, Titin and MYH6), and in rare instances two mutations have been detected in a single family. More than 200 individual mutations have been identified in relation to these 11 genes. Detection in one of the above more common genes can confirm the diagnosis of HCM and subsequently lead to: 1) further screening for HCM and arrhythmias, 2) assessment of risk for sudden cardiac death, 3) modifications in vigorous athletics or activity and 4) guidelines on obtaining antibiotic prophylaxis prior to dental and surgical procedures.

New Clinical Test
In the spring of 2004, a new clinical panel test was introduced to the benefit of the patient and medical community. The Laboratory for Molecular Medicine (LMM), a certified clinical diagnostic lab operating within Harvard Medical School-Partners Healthcare Center for Genetics & Genomics, now offers three panel tests of direct DNA sequencing of ten cardiomyopathy associated genes. The tests have greater than 99.9% accuracy to detect mutations in the sequence analyzed. Panel A screens for the five most common HCM genes: MYH7, MYBPC3, TNNI2, TNNI3, and TPM1. If a mutation is not detected in panel A, the individual may test for panel B which includes 3 less common HCM genes: ACTC, MLC2 and MLC3. Panel C screens for mutations for two glycogen storage disease genes mimicking HCM caused by sarcomere mutations: LAMP2 and PRKAG2. In adults, the detection rate of mutations for panel A is 50-60% and for panel B 5-10%. Panels A and B together can detect from 55-70% of all adults with clinical symptoms of HCM. The Lab also offers “known mutation testing”, a sequence analysis that determines the presence or absence of a known genetic mutation in suspected family members.

It is important to note that the current screening panel does not include all genes that may cause HCM and up to 30% of patients tested do not have a mutation detected. Therefore, the failure to detect a mutation does not completely rule out the diagnosis of HCM (except when screening for a known familial mutation). The mutation detection rate has been higher for patients with a family history of HCM while the rate among children without a family history of HCM has not been systemically studied. Currently research is being conducted to determine the detection rate in children.

The genetic test, which has a turnaround time of 3 weeks, requires submitting blood, saliva, biopsy or autopsy tissue sample of a HCM affected patient. Participation can be done through a physician by completed a requisition form (available on the website) and a patient consent form. The fees for panel A and B screening are $3,000 and $1,150 respectively for one individual and $500 for screening of each additional family member once a mutation is identified in a patient. This fee may be covered by insurance if submitted with a physician letter.

Whether to Test
Compared to adult testing, determining the cause of HCM in children is a more complicated process. Certain steps need to be taken before a child is considered for sarcomere mutation testing. A cardiologist and geneticist from a cardiovascular center who has experience in diagnosing and treating HCM patients should be consulted. First, it will need to be determined whether the disease is restricted to the heart or is a systemic disorder affecting other organs. HCM in children can be associated with malformation syndromes (e.g., Noonan syndrome), neuromuscular disorders (e.g., Friedreich ataxia) or metabolic conditions (e.g., Pompe, fatty acid oxidation defects, and mitochondrial diseases). There are more than 20 of these rare systemic causes of HCM in children. If a child does not have one of these known causes, the next step would be to screen the parents for evidence of heart disease and work with a genetic counselor or geneticist to review the family medical history. Depending on whether there is a family history or not, an informed decision can be made about whether the mutation detection rate is worth participating in this new HCM panel testing.

For more information on HCM panel genetic testing, please contact the Laboratory for Molecular Medicine at Harvard University (617) 768-8500 or visit their website at www.hpcgg.org/lmm.
CCF Founder Honored as a HERO for HEALTHCARE

President and Founder, Lisa Yue was recently selected by Good Housekeeping from hundreds of nominations to be one of five “Heros for Health” award recipients. Last year’s winners included Goldie Hawn and Jessica Lynch. The award luncheon will be held in New York City on November 17 and will be hosted by Campbell Brown, co-anchor of NBC’s “Today” Weekend Edition. The $10,000 award provided by Good Housekeeping’s sponsor, General Electric, will go towards funding more patient education and support services related to pediatric cardiomyopathy.

Do You Know Someone Famous with CARDIOMYOPATHY?

What do Ashton Kutcher and Denzel Washington’s character in Nick Cassavettes movie John Q have in common? The unfortunate answer is cardiomyopathy. Ashton Kutcher’s twin brother contracted cardiomyopathy when he was 13 and required a heart transplant. In the movie John Q Denzel Washington plays a desperate father determined to save his son who has cardiomyopathy and is in need of a heart transplant. This little known heart disease has had a terrible impact as well on a number of high profile athletes. The world of professional and amateur athletics appears to have been disproportionately affected by cardiomyopathy. Its victims include: NBA player Reggie Lewis of the Boston Celtics who died of a cardiomyopathy related cardiac arrest during a pick-up basketball game and Olympic figure skater Sergei Grinkov who collapsed and later died during a routine practice for Stars on Ice. Others who have died include college basketball star Hank Gathers of Loyola Marymount and college football star Ricky Bell of USC.

CCF in the NEWS

CCF is working hard to increase public awareness of the disease and the Foundation’s resources. View the latest news stories about CCF and our family members:

• **When Tiny Hearts Give Out**
  - The Bergen Record, July 13, 2004
  Article searchable online at www.northjersey.com

• **Woman Helps Her Kids, Others With Cardiomyopathy**
  - Sterling Heights Mirror Newspapers, September 30, 2004
  Article searchable online at www.hometownlife.com

• **Father Copes by Creating**
  Article searchable online at www.hometownlife.com

• **Teeing Up for a Kids’ Charity**
  Article searchable online at www.investmentnews.com

• **From the Heart**
  - The Peddie Chronicle, Fall 2004 edition

• **2004 Heros for Health Awards**
  - Good Housekeeping, December 2004
  Article searchable online at magazines.ivillage.com/goodhousekeeping

In order to increase awareness of the disease and advocate for more research funding, we need to inform the public about the realities of this devastating disease. While having a celebrity spokesperson would help to garner more attention, each of us can help to increase community awareness by talking to the local media. If you are interested in sharing your story or know of someone famous with cardiomyopathy, give CCF a call and we can help to get the story out to the local and national media outlets.
**Have a HEART**

Are you interested in helping to raise funds but don’t have time to organize a big event? The Hopeful Hearts paper heart sales program has been a popular way to raise funds, and we hope to expand this simple concept with more volunteer participation. Preprinted hearts, supplied by CCF, can be sold at school events, fundraisers, retail stores (supermarkets, banks) and clubs (book, women’s, golf clubs, gyms) for $10, $5 or any dollar amount. These decorative hearts with personal messages written by purchasers can either be posted at business establishments or presented to a hospital’s child life center for children recovering in the cardiology unit. If you would like more information about this fundraising program, please contact CCF at 201-227-8852.

---

**CCF Part of Bloomingdale’s Shopping Benefit**

On October 26, CCF participated in The Shopping Benefit, Bloomingdale’s annual charity shopping event. The special shopping day was held at Bloomingdale’s New Jersey stores in Riverside Square, Willowbrook, Short Hills and Bridgewater Commons, and featured fashion presentations, book signings, sweepstakes, children’s activities and culinary tastings. CCF ticket holders were entitled to 15% - 20% savings off Bloomingdale’s merchandise on the day of the event with 100% of ticket proceeds going to CCF.

---

**Second Annual GOLF CLASSIC Raises $178,000**

The Second Annual CCF Golf Classic held on Tuesday, September 14, 2004 at the New York Country Club in New Hempstead, NY was a tremendous success drawing 160 people from the tri-state area and raising close to $178,000 gross, surpassing last year’s total. The annual Golf Classic is the main source of CCF revenue for research and education initiatives on pediatric cardiomyopathy. The event is important in raising awareness of the disease and broadening CCF’s donor base. The net proceeds from this year’s event will be earmarked for CCF’s annual research grant program. Every year, CCF awards research funds of varying levels to investigators interested in furthering medical understanding and treatment advancements on pediatric cardiomyopathy.

The all-day event included a BBQ lunch, full round of golf, cocktails and dinner. During the cocktail reception and dinner there was a silent and live auction of 60 donated items as well as sports memorabilia provided by Steiner Sports. The highlight of the evening was when New York Giants legend Joe Morris made his appearance to emcee the evening dinner and live auction. During the dinner, Doctor Wendy Chung spoke about the impact of the Pediatric Cardiomyopathy Comprehensive Care Program that CCF helped to establish at the Children’s Hospital of New York. Joe Morris followed with an inspirational speech on teamwork and the need to work together to bring about positive change for underserved causes.

Callaway golf clubs were awarded to all first, second and third prize foursome winners. First prize went to the foursome headed by long time CCF supporter, Robert Voreyer and included other Morgan Stanley players Austin Zeigler, Aaron Hood and James Rhodes. The MacKay Shields team took second with Carney Hawks, Brian Nold, Won Choi and Mark Spellman. Third place went to the foursome with Bobby Frahm, Matt Glass, Mike Donoghue and Ted Burdick. Other contest winners included: Kevin Greaney (closest to the line), Jason Kirschner (closest to the pin and Jet Blue Challenge Champion) and J.J. Olszowy (longest drive).
CCF's second charity fashion show, "Fall into Fashion", with Neiman Marcus was held in Troy, Michigan on September 12. Doctor Richard Humes from the Children's Hospital of Michigan was the guest speaker at the sold out Sunday brunch. Based on the success of both the Spring event in New Jersey and Fall event in Michigan, CCF teamed up with Neiman Marcus again for a follow up "Holiday Style" benefit in Troy, Michigan. The elegant cocktail reception was held on November 18 and featured a strolling fashion show, silent auction and guest speakers.

MANY THANKS to our Event Sponsors

Abbott & Abbott Construction • Abrams Capital • Angelo Gordon & Co. • Avenue Capital Management • Bond Street Capital • Brencourt Advisors • Chapdelaine Corporate Securities & Co. • Chatham Asset Management • Chesapeake Partners • CIBC World Markets Real Estate Finance Group • Citigroup Global Markets • Credit Suisse First Boston • James Cullianane, Stark Investments • Deutsche Bank Securities • FIMAT USA • Forex Capital Markets • Fuel Fitness for Body & Mind • Howard & Sharon Golden • GoldenTree Asset Management • JDV Equipment Corporation • JMG Capital Management • KBC Financial Products • Kolatch Family Foundation • Michele Kolsky-Assatly, Coldwell Banker • MacKay Shields • Edward Mule • Dan Ornstein • Singer Family Foundation

Many thanks to all our anonymous sponsors and individual donors as well!

TEAM EFFORT to Raise Funds & Awareness

Scott Newport has an inspirational story to tell about his life with his son. Scott's son, Evan, has hypertrophic cardiomyopathy associated with Noonan's Syndrome and was not expected to live past his second birthday when he was first diagnosed. In spite of the doctor's predictions, Evan is now 2 1/2 years old. As part therapy and part hobby, Scott began making furniture from discarded, damaged wood that he painstakingly refinished himself. Scott, a builder and carpenter by trade, eventually turned his hobby into a fundraising vehicle for organizations that help his son. Scott is now selling his handcrafted furniture at MT Hunter, a Birmingham, MI furniture store, with the full proceeds benefitting CCF. CCF family member Denise Hoglund pitched in to design Scott's display materials and CCF helped to secure press coverage. Scott and his family have already raised $750 for CCF through a hometown garage sale that attracted the attention of the local media and community organizations.

We Want to Hear from You!

Families affected by cardiomyopathy as well as physicians and nurses are encouraged to submit articles, news updates and/or personal stories for the newsletter. Articles can be up to two pages single spaced in length and sent as a MS Word file to newsletter@childrenscardiomyopathy.org. Photographs and artwork can be submitted as standard image files (.tif, .gif, .jpeg). The submission deadline for our next issue is February 18, 2005.
Welcome to Heart to Heart
The Newsletter of the Children's Cardiomyopathy Foundation
Volume 1, Number 2 • Fall 2004 • Winter 2004

INSIDE THIS ISSUE:

• Foundation News
  Cover News: CCF funded research recognized at the American Heart Association and American College of Cardiology 2004 Scientific Sessions.

• Family Information
  Page 2: CCF partners with more organizations to develop new patient materials. See parent survey inside.
  Page 7: New research studies recruiting patients.

• Medical News
  Page 4 & 5: The genetic testing process and options.
  Page 8: HCM clinical testing now available.

• Fundraising Update
  Page 10: Second Annual Golf Classic is a great success.

MAKING A DIFFERENCE...
CCF 2004-2005
ANNUAL FUND DRIVE UNDERWAY

As the year draws to a close, it is time to reflect upon the achievements of the past and to look ahead to the challenges of the future.

In this season of giving, please remember CCF with a gift. Our goal for this annual appeal is to raise $75,000 to sponsor an International Scientific Workshop.

To gather the best scientific minds together with the aim of improving treatment and understanding the genetic causes of cardiomyopathy in children.

• To facilitate more dialogue
• To present research findings
• To develop a research agenda
• To initiate collaborative research

For more information about making an annual gift, please contact CCF at 201-227-8852 or info@childrenscardiomyopathy.org

The Children’s Cardiomyopathy Foundation is a IRS 501(c)(3) public charity so all donations are tax-deductible to the extent permitted by law.