

Heart to Heart

Volume 8, Number 2 • Fall 2011 • Winter 2012

Highlights...

RESEARCH BRIEFINGS

- Update on 2009-2010 Research Grant Recipientspg. 1
- Cardiomyopathy Specialist: John Jefferies, M.D.pg. 4

CCF UPDATES

- Ninth Annual Golf Classic Triumphspg. 5
- Highlights from CCF Family Fundraiserspg. 6
- Taking a Stand Against Sudden Cardiac Arrestpg. 8
- New Guidelines on HCM Diagnosis and Treatmentpg. 8

FOR FAMILIES

- Patient Perspective: From Athlete to Sports Reporterpg. 9
- Expert Q&A: New Guidelines on Genetic Testingpg. 10
- Teen & Young Adult Ambassador: Matt Protas.....pg. 10

CCF-Funded Researchers MAKING PROGRESS

CCF's funded research continues to make a significant impact in the field of pediatric cardiology. In 2009 and 2010, CCF awarded seed funding to five investigators studying pediatric cardiomyopathy. To date, their findings have been published in peer-reviewed journals, presented at scientific conferences and, in some cases, expanded with additional funding from the **National Institutes of Health**.

Identifying and Detecting Genetic Mutation-Causing Pediatric Cardiomyopathy

Stephanie Ware, M.D., Ph.D.

associate professor of pediatrics and co-director of cardiovascular genetics at Cincinnati Children's Hospital

Dr. Ware received CCF funding in 2008 and a 2-year grant extension in 2010 to identify the most common genetic mutations for pediatric cardiomyopathy and to develop clinical genetic tests to detect these mutations.

Dr. Ware has found a cause for more than 60 percent of the approximately 100 studied cases and discovered that in more than 15 percent of the cases, the disease is caused by more than one mutation. As two thirds of cases are idiopathic, Dr. Ware's study is a significant development in the genetics of pediatric cardiomyopathy. "Many times, the causes in children may be the same as adults, but we are finding differences as well. It is important to study this disease specifically in children," says Dr. Ware. Her study also is examining how some of the genetic changes determine the severity of the disease.

During the course of the grant, there has been tremendous progress in genetic technology, and Dr. Ware has modified her study to make use of this new technology. Dr. Ware states, "We still need to determine the best way to interpret the results, and this interpretation needs to be revisited as our knowledge continues to increase."

While continuing her study to identify new genes, Dr. Ware is developing a clinical genetic test that is specific to pediatric cardiomyopathy but will complement existing cardiomyopathy testing panels. "This would mean that physicians would have a consistent way to screen for the most common causes of the disease, which will help develop more targeted therapies for children," adds Dr. Ware.

This year Dr. Ware presented her findings at the **Weinstein Cardiovascular Development Conference** in May 2011 and at the **American Society of Human Genetics Meeting** in November 2010 and October 2011. Her paper entitled "Genetic diagnosis in pediatric cardiomyopathy: clinical application and research perspectives" was presented at **CCF's Second International Scientific Conference** and published in the April issue of *Progress in Pediatric Cardiology*.

Generating Heart Muscle and Vascular Cells from Stem Cells to Study Causes of Cardiomyopathies

Charles Murry, M.D., Ph.D.

professor of pathology and bioengineering and director of the Center for Cardiovascular Biology at University of Washington

As one of CCF's 2009 grant recipients, Dr. Murry's research focused on the growing field of stem cell therapy. His study involved developing an efficient

continued, page 3



A Cause for Today... A Cure for Tomorrow

**Children's
Cardiomyopathy
Foundation**

The *Heart to Heart* newsletter is published biannually by the Children's Cardiomyopathy Foundation (CCF), a national organization dedicated to saving lives and improving the quality of life for children with cardiomyopathy. CCF's mission is to accelerate the search for a cure by supporting research, educating physicians and patients, and increasing awareness and advocacy related to the needs of children with cardiomyopathy and their families. CCF is a publicly supported tax-exempt organization as described under section 501(c)(3) of the Internal Revenue Service.

Board of Directors

Lisa Yue, *President & Founder*
Brian Nold, *Treasurer*
Raymond Yue, *Secretary*
Carney Hawks
Carolyn Kong
Ian Sandler
Eddie Yu

Medical Advisory Board

Wendy Chung, MD, PhD
Columbia University Medical Center
Steve Colan, MD
Children's Hospital Boston
Daphne Hsu, MD
Children's Hospital at Montefiore
Steve Lipshultz, MD
University of Miami Medical Center
Jeff Towbin, MD
Cincinnati Children's Hospital

Staff

Lisa Yue, *Executive Director*
Sheila Gibbons, *Development & Communications Senior Manager*
Renee Thekkekara, *Development & Communications Coordinator*
Christine Colón, *Patient Outreach & Support Manager*
Harriet Salk, *Patient Support Coordinator*
Becky Delgado, *Administrative Assistant*

All submissions and correspondence regarding the newsletter should be directed to:

Children's Cardiomyopathy Foundation
P.O. Box 547
Tenafly, NJ 07670
Tel: 866.808.CURE
Fax: 201.227.7016
info@childrenscardiomyopathy.org
www.childrenscardiomyopathy.org

The Children's Cardiomyopathy Foundation, including all parties to or associated with *Heart to Heart* will not be held responsible for any actions readers take based on their interpretation of articles from this newsletter. As always, readers are encouraged to discuss medical evaluations and treatments with their own physicians.

© Copyright 2011-2012 Children's Cardiomyopathy Foundation. All rights reserved.

From Lisa Yue, CCF Founder & President

As a steering committee member on the **Sudden Cardiac Arrest Coalition (SCAC)**, CCF has had a busy year supporting many legislative issues. Recently, the Senate agreed to set aside \$2.5 million for the **Rural and Community Access to Emergency Devices Program**. This competitively awarded program provides funding to states to buy automatic external defibrillators (AEDs) for locations where sudden cardiac arrest is likely to occur. We achieved this victory in spite of the federal deficit thanks to the collective efforts of the Coalition, Senator Tom Harkin and your grassroots support.

There is much more to do in the advocacy area, which is why CCF has decided to invest more time and resources into developing a federal government relations and advocacy program. We need to have a stronger voice at the federal level and ensure that policymakers address issues of importance specific to the cardiomyopathy community. We have enlisted the help of **Drinker Biddle & Reath**, a national law and consulting firm with expertise in health government relations, to represent CCF in Washington D.C.

This is a new arena for us but a very important one, and we are excited about what we can achieve in the months ahead. We are committed to improving the lives of children with cardiomyopathy and will keep you informed of our new public policy initiatives.

Lisa Yue

A Holiday Wish



Some kids wish for new toys.
Our kids wish to get better.

Watch your mail for our **HOLIDAY APPEAL**, which shares with you CCF's wishlist.

We hope your family and friends will join us this holiday season in making these wishes come true.

Research Briefings *continued from page 1*

system for generating heart muscle and vascular cells from stem cells derived from human fibroblasts, which are readily accessible connective tissue cells found in the skin and most other tissues. Dr. Murry's research team is the first group to use induced pluripotent stem cells (iPSC) to create human heart tissue in a dish and show that it can contract and survive to form new heart muscle after transplantation. This was tested by implanting the engineered heart tissue constructs into the hearts of rats with impaired immune systems.

Through his study, Dr. Murry was able to optimize the conditions of differentiation, tissue engineering and tissue conditioning, revealing new research possibilities. Because obtaining cardiac tissue of children with cardiomyopathy in large quantities is difficult, the ability to generate pediatric cardiac tissue from a more accessible source like skin tissue is a significant development. "By making cardiac muscle tissue that is genetically identical to the patient, we can better understand the basis for cardiac dysfunction and establish novel treatments at an accelerated rate," states Dr. Murry.

His next step will be to take skin cells from cardiomyopathy patients, convert them into pluripotent stem cells, and then direct the cells into heart muscle that carries the genetic mutation that causes the disease. The contraction and relaxation of the generated heart muscle tissue, as well as its susceptibility to rhythm disturbances, can then be studied. "This will set up the basis for understanding what causes the contractile and electrical problems in cardiomyopathic hearts as well as for drug screens to identify new treatments," adds Dr. Murry.

A manuscript detailing Dr. Murry's study has been published in the June 24, 2011 issue of *Circulation Research* titled "Growth of Engineered Human Myocardium with Mechanical Loading and Vascular Coculture." The article was the most widely read article published in *Circulation Research* this year, and the authors were recognized at the **American Heart Association**

journal's annual dinner in November.

Determining the Role of T-cells in the Development of Pediatric Dilated Cardiomyopathy

Jay Reddy, Ph.D.

associate professor of veterinary and biomedical sciences at University of Nebraska-Lincoln

Dr. Reddy received a four-year joint research grant from CCF and the **American Heart Association** in 2009 for his study on how Coxsackievirus B3 (CVB3) in some individuals leads to chronic inflammation of the heart muscle (myocarditis) and progresses to dilated cardiomyopathy (DCM). His study focuses on how the CVB3 virus causes certain T-cells — blood cells that fight infections — to become self-reactive and attack the heart's muscle cells. He believes that certain T-cells latch onto and attack cardiac myosin or heart muscle proteins causing DCM to develop.

To determine how great a role these cardiac myosin-specific T-cells have in contributing to DCM, Dr. Reddy generated mice models with myocarditis caused by the CVB3 virus and developed a genetically engineered chemical reagent (major histocompatibility complex class II tetramer). In the next phase of his research, he will use this chemical reagent on the heart and spleen cells of the infected mice to identify and isolate the cardiac myosin-specific T-cells from other types of non-disease inducing T-cells. Then he will transfer the cardiac myosin-specific T-cells into healthy mice and observe if DCM develops as predicted.

Dr. Reddy's study ultimately could lead to treatment specifically designed for viral-induced DCM. "Nearly 50 percent of the cases of myocarditis or DCM can have some evidence of exposure to CVB3," notes Dr. Reddy. "If we prove that this virus triggers an autoimmune response and thereby provides a mechanism for DCM to develop, we can come up with extremely targeted therapies.

His work to date has been accepted for publication in three major journals this

year. "Detection of cardiac myosin heavy chain-specific CD4 cells by using MHC class II/IAk tetramers in A/J mice" was published in the September issue of *Journal of Immunology Methods* and "Detection of autoreactive CD4T cells using major histocompatibility complex class II dextramers" was published in the July issue of *BMC Immunology*. Later this year, his article, "Identification of novel mimicry epitopes for cardiac myosin heavy chain— that induce autoimmune myocarditis in A/J mice" will be published in the journal *Cellular Immunology*.

Characterizing Molecular Mechanisms of Heart Muscle Cell Regeneration

Bernhard Kuhn, MD

assistant professor of pediatrics at Harvard Medical School and cardiologist at Children's Hospital Boston

Dr. Bernard Kuhn's 2010 research grant is on cardiac regenerative medicine. His study focused on the molecular mechanisms related to heart muscle regeneration and the identification of those genes responsible for controlling the production of heart muscle cells. Dr. Kuhn's research is based on the premise that cardiomyopathy is associated with loss of heart cells or cardiomyocytes, which are irreplaceable and may be associated with heart failure. Dr. Kuhn's study explores the ability to create new functional cardiomyocytes to replace the heart's deficient cells and restore normal pumping function.

For the past two years, Dr. Kuhn has been developing a novel technology known as the Fucci live-cell-cycle reporter system to visually monitor the heart muscle cell cycle in real time using fluorescent dyes and tags to create a 3-dimensional model of the heart. This technology showed that mouse hearts do create new muscle cells contrary to belief, and it highlighted which cardiomyocytes regenerate. "Demonstrating that there is cardiomyocyte proliferation in mice is a huge turnaround in how cardiomyopathy is studied and understood," says Dr. Kuhn. "This is a very important step forward, one that looks at and validates a neglected mechanism of heart growth and regeneration."

continued, page 4

Research Briefings *continued from page 3*

After months of study, Dr. Kuhn found that the technology also can identify gene products that regulate the growth of heart muscle cells.

Dr. Kuhn's next step is to use microarray technology to understand on a molecular level how the heart makes new cardiomyocytes. By identifying the genes that are responsible for cardiomyocyte proliferation, he can determine which genes have the potential to repair the damaged heart. Dr. Kuhn's findings could transform therapeutic options for heart failure. "We intend to apply for National Institutes of Health Funding to aid in finding those invaluable genes that lead to the expansion of the heart muscle population," states Dr. Kuhn.

3D-Engineered Tool to Investigate Childhood Hypertrophic Cardiomyopathy

J. Carter Ralphe, M.D.

assistant professor of pediatrics at University of Wisconsin-Madison and chief of cardiology at the American Family Children's Hospital

Dr. J. Carter Ralphe received a 2010 award to study 3D-engineered cardiac tissue associated with childhood hypertrophic cardiomyopathy (HCM). This is the first study showing that engineered cardiac tissue can be generated from cardiac cells of a newborn mouse. This offers a more effective way of studying mutations in cardiac myosin binding protein C (cMyBP-C), a muscle protein that regulates the heart's contraction and is linked to the cause of HCM.

Traditionally animal models are used to uncover specific mutation physiology and to study why different mutations in a gene vary in severity and age of onset, but it is expensive and demands an enormous time commitment. Dr. Ralphe's 3D model is more cost-effective and more efficient. "We found that it takes roughly three months with the 3D construct versus two years and \$20,000 required to generate animal models containing HCM-causing mutations," says Dr. Ralphe. "We now have a rapid screening tool that might provide insight into how families' specific mutation

affects the function of the muscle cells."

Dr. Ralphe's 3D technology will help researchers more easily genetically manipulate the contractile properties of the heart and test how alterations in cardiac genes contribute to abnormal heart function such as HCM. Furthermore, examining how specific mutations respond to physiologic stress will help to explain why different mutations in the same protein lead to such a range in disease severity.

Dr. Ralphe received a 5-year \$1.9 million grant from the **National Institutes of Health** (NIH) to expand the study to include a panel of 12 human mutations and to evaluate environmental stressors on HCM development in each of the human mutations. Dr. Ralphe says CCF's grant played a critical role in securing the NIH grant. "I am immensely grateful for CCF's support, which gave me the resources to collect critical data and jump start my

research," said Dr. Ralphe.

There have been several publications and presentations related to Dr. Ralphe's research. "Neonatal mouse-derived engineered cardiac tissue: a novel model system for studying genetic heart disease," was published in **Circulation Research** and "HCM-causing mutation in myosin binding protein-C accelerates contractile kinetics in non-hypertrophied engineered cardiac tissue" and "Engineered cardiac tissue as a non-hypertrophied model system," were both published in **Pediatric Research** in 2010. "Functional insights from a 3D engineered tissue construct of unremodeled murine MYBPC3 myocardium," was presented at the **American Heart Association Basic Cardiovascular Science** meeting in July 2010 and the **American Heart Association Scientific Sessions** meeting in November 2010.



Spotlight on Cardiomyopathy Specialist: JOHN JEFFERIES, M.D., M.P.H.

John Jefferies, M.D., M.P.H., is associate professor of pediatric cardiology and the director of advanced heart failure and cardiomyopathy at the **Cincinnati Children's Hospital**. Dr. Jefferies is a clinical cardiologist who has translational and clinical research programs. He works closely with **Jeff Towbin, M.D.**,

chief of pediatric cardiology. Dr. Jefferies credits working with Dr. Towbin for more than 10 years for his interest in pediatric cardiomyopathy (PCM).

Dr. Jefferies' research involves cardiovascular genetics with specific interest in a newly recognized form of cardiomyopathy — left ventricular non-compaction cardiomyopathy (LVNC). "LVNC is the third most common cardiomyopathy behind dilated cardiomyopathy and hypertrophic cardiomyopathy," notes Dr. Jefferies. "A LVNC diagnosis is becoming more frequent in infants and adolescents as awareness of the disease increases; however, there is only limited published data and much of this is in adult populations."

To expand on the understanding of LVNC, Dr. Jefferies and his team have built the largest international cohort of patients with LVNC. Currently, their areas of study include finding and evaluating treatments, learning more about the variability of the disease, and using gene identification to yield more screening options for families with LVNC. "It is a huge opportunity to make lasting impacts on the care of these patients," says Dr. Jefferies.

His work with families affected by the disease also has been a great inspiration. "Nothing is more motivating than a parent asking you to help their child," says Dr. Jefferies. "Being a father, I understand that your child's well-being can be the driving force for almost everything you do."

NINTH ANNUAL Golf Classic Triumphs



Eddie Yu, Edgar Sabounghi, Tim Malley and Kevin Kavanagh

CCF hosted its **Ninth Annual Golf Classic** at the exclusive Montclair Golf Club on July 18 to a sold-out crowd of top finance and legal professionals. Players shared in a day of good weather and challenging golf to help raise \$411,000. Afterwards, CCF supporters enjoyed cocktails, a silent auction and dinner.

One of the highlights of the dinner was guest speaker **Matt Protas** whose high school athletic career ended when he was diagnosed with hypertrophic cardiomyopathy. He spoke about his difficult journey from diagnosis to acceptance and how the disease has given him a new sense of purpose. Now a senior at the University of Connecticut, Matt has agreed to be

CCF's first teen and young adult ambassador and will work with CCF to develop more support services for teens and young adults with cardiomyopathy. (See story on page 10.)

Net proceeds from the event will go towards supporting research and education initiatives on pediatric cardiomyopathy. "The golf classic continues to be a great success," says CCF Board Member **Ian Sandler**. "We have our generous donors to thank. Their incredible support year after year helps us advance our mission of one day finding a cause and a cure for pediatric cardiomyopathy."

The winning foursomes were: **Ivan Friedman, David Lerner, Matt Maggio, Andy Stock** (first prize); **Richard Brennan, Ryan Brennan, Matthew Glass, Edward Keating** (second prize); **Dennis Lu, Jon Eckert, Kevin Baer, Jim Higgins** (third prize); and **Mark Lawrence, Paul Arrouet, Dave Austin, Terry Yun**



Top: Tim Milton
Bottom: Brad Roberts, Eric Nagel, Kerri Cohen, and Joe Salerno

(fourth prize). The prize for closest to the pin went to **Scott Mallek** and **Matthew Glass**. **Tim Sargen** and **Seth Bernstein** won the prize for longest drive, and **Gene Pagnozzi** took home the raffle prize.

2011 Event Sponsors: Sunil Aggarwal; Anchorage Advisors, LLC; Bank of America; Barclays Capital; Richard Barrera & Purnima Puri; BGC Partners; Steven Bleier; BNP Paribas Bank; Chris Berry & Matt Lambert; The Boyce Family; Andrew Brenner; John O'Meara & Chaney Sheffield; Brigade Capital Management LLC; Euro Brokers High Yield/Robert Burke; Cantor Fitzgerald; Dustin Cappelletto; Sheree Chiou & Jason Kirschner; Citigroup; Colbeck Capital; Jason Colodne; Emil Costa; Credit Suisse; DDJ Capital; Lucas Detor; Deutsche Bank Securities; Robert & Karen Dishner; Daniel Elkaim; Fried, Frank, Harris, Shriver & Jacobson LLP; Eric & Monica Friel; Rob Gallivan & Family; Jim Geregthy; Gleacher & Company/Mark Colm; Goldman Sachs; The Henderlong Family; Hunton & Williams LLP; ICAP High Yield; Jefferies & Company/Scott Haberman; Jefferies & Company/Brian Hewitt; David & Sarah Johnson; JP Morgan; Katten Muchin Rosenman LLP; George Khouri; Kolatch Family Foundation; Clint Kollar; Kramer Levin Naftalis & Frankel LLP; Daniel & Lauren Krueger; The Lehner Family; The Lund Family; Mackay Shields; Macquarie Capital (USA) Inc.; The Maldutis Family; James Martin; Drew McKnight; Mike & Deb McLaughlin; Gil Nathan; The Ornstein Family; Gene Pagnozzi; Regiment Capital; Richards Kibbe & Orbe LLP; Riva Ridge Capital Management; Edgar Sabounghi; The Sandler Family; Adam Savarese & Family; Senator Investment Group; Simpson Thacher & Bartlett; Stone Lion Capital Partners LP; Taconic Capital Advisors LP; Kevin Tan; Vanderbilt Appraisals; Eddie Yu & Lisa Yue

Save the Date!

4th Annual

All-In For A Cure No-Limit Texas Hold 'Em

**Wednesday, February 8, 2012
Crimson, New York City**

Join CCF for an exciting evening of charity poker!

Try your luck at winning the grand prize: entry to the **World Series of Poker Main Event.**

Visit our event web page under "News & Events" for more information.

More event photos are online under "News & Events/Golf Classic."

FAMILY FUNDRAISERS

Timothy Grant Jewelry CHARITY GOLF OUTING FOR PEDIATRIC HEART DISEASE



Tim, Darby and Lisa Siefert

Lisa and Tim Siefert, and Lorrie Ruud held a golf fundraiser in Lake Zurich, Ill. to honor the Siefert's daughter, **Darby**, age 5, who has hypertrophic cardiomyopathy and to pay tribute to their son **Shane** who died in an accident in March. The community-supported event raised an impressive \$15,500 for CCF.

Ticketed admission included a barbecue lunch, a full round of golf and dinner. The evening activities

included raffles and a silent auction featuring a variety of donated items, including a dress designed by Project Runway star **Peach Carr**, tickets to **Chicago Bears** and **Chicago Blackhawks** games, and a basketball autographed by **Ron Artest** of the **LA Lakers**. The event sponsorships and auction items were contributed by the Siefert's friends and family, local businesses, and patrons of the two Siefert jewelry stores.

CCF Parent Ambassador **Jennifer Kirkham** was the evening guest speaker. Jennifer lost her son Judah, age 1, to dilated cardiomyopathy last year. "The event was terrific," according to Jen. "I was so impressed by their supporters, who didn't know anything about cardiomyopathy but who were willing to show their support to this family who has been through a lot."

The Siefert's have requested that the proceeds from the golf outing go towards **CCF's Family Assistance Program**, which will launch later this year. "Tim and I want to support those families and children who might not be getting the medical care that they need because they don't have enough money or insurance," Lisa says.

CASEN RILEY 3-on-3 Basketball Tournament for Pediatric Cardiomyopathy

For a second year in a row, **Heather** and **Casey Riley** held an all-day basketball tournament in Abilene, Texas to honor their son **Casen**, who passed away unexpectedly to hypertrophic cardiomyopathy at six months old. Heather also organized raffles and sold t-shirts she designed with Casen's picture. \$5,000 was raised for CCF, topping last year's total of \$3,000.

"The Foundation has been very helpful in providing me with information and support," says Heather. She plans on making the tournament a yearly event in memory of Casen's birthday. In June, she and her husband welcomed their second child, Carter. They look forward to the day when Carter can shoot hoops in his big brother's honor.



Casey, Carter and Heather Riley

MORE FAMILY FUNDRAISERS...

Wedding Dance for a Cause

Leah Maloney and **David Tully** paid tribute at their wedding to David's brother **Bradley** who passed away from hypertrophic cardiomyopathy in 2008. Nearly 100 guests made donations for the chance to dance with the wedding couple. To date, the Tully family has raised more than \$10,000 for CCF in Bradley's memory.



Birthday Party in Honor of CCF

Cary and **Hannah Fliegler** organized a party at Stonybrook Day Camp in Randolph, N.J. to celebrate the birthdays of their three children, **Sophia**, **Sam** and **Aria**, and to mark the sixth anniversary of Sophia's heart transplant from cardiomyopathy. She and her siblings asked for donations to be made to CCF in lieu of birthday presents. More than \$3,100 was raised for research.



Run for Brooke

Aimee Gillum participated in the Nike Women's Half Marathon in San Francisco, Calif., and raised more than \$1,600 in sponsorships. She ran in honor of her friend's daughter, **Brooke Balck**, who was diagnosed with dilated cardiomyopathy and recently received a heart transplant.

Movie Madness at CCF Golf Classic

In lieu of birthday gifts, CCF staff member **Becky Delgado** asked friends and family to donate western, mobster and Disney themed DVDs, which she assembled into beautiful silent auction gift baskets for CCF's Annual Golf Classic fundraiser.



Hockey Night for Judah

Jen Kirkham honored her son **Judah**, who passed away from dilated cardiomyopathy in 2010 at the age of 1, with an American Hockey League game fundraiser in Peoria, Ill. The local hockey team, the **Peoria Rivermen**, donated 30 percent of ticket sales to CCF. Friends, family and fans who attended the game enjoyed lunch before the game and participated in a raffle to help raise more than \$800.

Brat Fry and Bake Sale Tribute

Sara Huss' daughter **Peyton Kay Huss** would have turned five this June. To mark the occasion and to pay tribute to Peyton's life, which was cut short by dilated cardiomyopathy, Sara held a brat fry and bake sale at her local grocery store in Wrightsman, Wis. She also sold CCF curebands and raised nearly \$700.

North Carolina Group Chili's Fundraiser

North Carolina Support Group leaders **Angela Henderson** and **Melissa Perna** arranged a CCF night at Chili's in Durham, N.C. CCF received a portion of the evening sales along with donations from guests. The event brought together CCF families as well as **Michael Carboni, M.D.**, of **Duke University Medical Center** and his team of nurses. Angela lost her son **Jonathan** to restrictive cardiomyopathy in 2005 and Melissa's son **Troy**, age 11, has left ventricular non-compaction cardiomyopathy.

Healthy Heart Awareness at School

Students and staff at **Balmville Elementary School** in Newburgh, N.Y. celebrated **Healthy Heart Month** with a fundraiser and assembly. Arranged by **Sharon Tramm**, whose daughter **Kristen** passed away from dilated cardiomyopathy, students participated in a basketball free throw and art poster contest. At the school assembly, pediatric cardiologist **Eric Fethke, M.D.** gave a presentation on cardiomyopathy and staying heart healthy.

Laughter is Good for the Heart

Jeanne Curtin hosted a comedy night at The Gold Room in Portland, Maine in honor of her son **Austin**, age 15, who has hypertrophic cardiomyopathy. Guests were entertained all night by comedians and special guest musician **Dave Andrew**. Proceeds from the ticket sales and 50/50 raffle went to CCF.

Dress Down for a Good Cause

St. Anne's School in Garden City, N.Y., chose CCF for the school's monthly dress down day in memory of **Eddie Casey**, who passed away from dilated cardiomyopathy at age 3 while waiting for a heart transplant. His older sister **Elizabeth** and fellow students donated their dollars to dress without uniforms for a day.

Lillybelle's Motorcycle Ride

Jon Ruggeri and his fellow motorcycle enthusiasts organized a 100-mile ride between Downingtown, Pa., and Elkton, Md., to raise more than \$1,200 in honor of granddaughter **Lillybelle Chatman-Royce**, age 3, who has dilated cardiomyopathy. At the end of the ride, riders, friends and family celebrated with burgers and beer, a 50/50 raffle, and live music on the grounds of St. Anthony's Church in Downingtown.



Second Chance at Life Luncheon

CCF member **Michelle Perona** threw a fundraiser in Ansonia, Conn., to mark the first anniversary of her son's heart transplant. **Jesse, Jr.**, age 1, was diagnosed with dilated cardiomyopathy soon after birth. Relatives and friends came out for an Italian feast, raffles, and arts and crafts activities.

Treasure Hunt for CCF

Jeff Jensen designed a CCF Geocoin for **Geotagging**, a GPS outdoor treasure hunt game, in honor of his 12-year-old daughter **Shawna** who lives with hypertrophic cardiomyopathy. The CCF pathtag is sold on eBay (geocoin #18952) and 50 percent of the proceeds will be donated to CCF.



Taking a Stand Against SUDDEN CARDIAC ARREST

The Children's Cardiomyopathy Foundation (CCF), as part of the Sudden Cardiac Arrest Coalition (SCAC) and in cooperation with the Congressional Heart and Stroke Caucus, hosted the "Take a Stand Against Sudden Cardiac Arrest" event on Wednesday, October 26, 2011 in recognition of National Sudden Cardiac Awareness Month.



Rep. Roe demonstrating CPR

children is crucial because it allows for early treatment, which may include an implantable defibrillator, to protect against sudden cardiac death," states Lisa Yue, CCF president and founder. "Also, knowing cardiopulmonary resuscitation (CPR) and how to use an automatic external defibrillator (AED) can be a life saving measure in the case that an at-risk child has a sudden cardiac arrest."

The event featured four concurrent training sessions on CPR and the use of AED in the Rayburn House Office Building in Washington, D.C.

Representative Lois Capps (D-CA) was honored for her work in introducing the "Teaching Children to Save Lives Act," legislation which would provide grants to assist schools with teaching student CPR and how to use an AED. "By training the

next generation of students in CPR and AED response, they may one day save the life of a classmate, friend, family member or complete stranger," said Capps.

Representative Phil Roe (R-TN) was recognized for his heroic actions in September in helping to save the life of a 52-year-old father of three. Dr. Roe spoke on the importance of having access to an AED in an emergency and their ease of use. He thanked the coalition for reminding the public that immediate bystander intervention with CPR and AEDs can more than double survival rates.

Benjamin Abella, M.D., of the center for resuscitation science in the department of emergency medicine at the University of Pennsylvania emphasized that in SCA cases bystander action can mean the difference between life and death. "As compelling as the statistics are, however, it is the stories of survival that really bring the message home," said Dr. Abella. The day culminated with the personal stories of three SCA survivors who were all saved by bystanders using CPR and an AED.

NEW GUIDELINES on the DIAGNOSIS AND TREATMENT of Hypertrophic Cardiomyopathy

New guidelines jointly published by the **American Heart Association** and the **American College of Cardiology Foundation** on hypertrophic cardiomyopathy (HCM) offers pragmatic steps on the diagnosis and treatment of this frequently undiagnosed heart disease. The practice guidelines represent a consensus of expert opinion on the disease and were developed to assist healthcare providers in the clinical management of their HCM patients.

According to this new science-based guideline, many people properly diagnosed with HCM can have a normal life span with appropriate treatments. "This disease, once regarded as mysterious and almost universally associated with poor outcomes, is now a highly treatable genetic heart disease," says Barry Maron, M.D., director of the **Hypertrophic Cardiomyopathy Center** at the **Minneapolis Heart Institute Foundation** and co-chair of the committee that developed the recommendations.

The guidelines were published in the November 8, 2011 edition of *Circulation*. Key recommendations include:

- HCM patients should not participate in intense or competitive sports; however low-impact exercise as part of a healthy lifestyle is acceptable.
- Because HCM is often inherited, all first-degree relatives of someone known to have the disorder should be screened.
- Children of parents with HCM should be tested every 12 to 18 months since the disease may present later in life.
- Cardiac MRIs should be used to diagnose HCM when electrical tests and ultrasounds are inconclusive.
- Patients should be offered genetic testing and counseling, and be evaluated for risk of sudden death.
- Patients judged to be high risk based on symptoms or family history should be offered implantable defibrillators to prevent sudden death.
- Drug therapy can help control symptoms, but for patients with severe symptoms who do not respond to medication surgical intervention may be necessary.
- Surgeries should be performed by doctors who have performed at least 20 similar procedures or at a center dedicated to the treatment of HCM.

PATIENT PERSPECTIVE

From Rising Star on the Field, To Rising Reporter on the Sidelines: AMANDA HOUSENICK'S STORY



Amanda Housenick

Before **Amanda Housenick** began a career as a sports writer for *The Morning Call* in Allentown, Penn., she was a rising high school field hockey star in her hometown of Mountaintop, Penn. That was until her world came to a second-splitting stop when she was diagnosed with hypertrophic cardiomyopathy (HCM).

Amanda was at the top of her game with big east colleges scouting her; but practices were a struggle with shortness of breath and wheezing. At the age of 15, she was diagnosed with exercise-induced asthma and given an inhaler; but the wheezing and burning lungs continued.

A year later, Amanda was officially diagnosed with HCM. The devastating diagnosis ended her playing career. Amanda was heart broken and depressed. "Sports was my life. I played year-round— field hockey, soccer, basketball— and I played everyday," recalls Amanda. "I felt that I had lost my identity when I was told I couldn't play anymore."

It wasn't just the game she loved; it was being part of a team and the camaraderie of fellow teammates. "I loved the spaghetti dinners before a game, making team spirit signs and the pep rallies," says Amanda. After her diagnosis, it was hard for Amanda to even see her former teammates wearing their uniforms on game day.

Six weeks after her diagnosis, Amanda was presented an opportunity to answer phones at *The Times Leader* in Wilkes-Barre, Penn. She thought it might help her feel better to have a new interest, and indeed it did.

After only six weeks at the job, she wrote her first byline story, and she is now making her mark in the field of sports writing. She won the acclaimed **Associated Press Sports Writer Award** in 2003, but her biggest accomplishment was being named Phillies beat writer in 2008 at the age of 29.

As the only female sports reporter at *The Morning Call*, the 3rd largest paper in the state, her stories have been published in associated papers including, the *Los Angeles Times* and the

Chicago Tribune. She has interviewed multiple Hall of Famers including **Mike Schmidt**, **Jim Bunning**, and **Robin Roberts**. Her most memorable assignment, she says, was a three-piece story about a Bloomsburg University basketball player, **Adam Bohman**, whose brother, a college swimmer, collapsed and died of dilated cardiomyopathy during his swim practice.

She also has written about her own experience with HCM and now feels she is in a unique situation to raise awareness of this misunderstood disease. "I never actually had exercise-induced asthma or the microscopic hole in my heart as my cardiologist had diagnosed me with when I was 12," says Mandy.



Amanda interviews Ryan Howard, Philadelphia Phillies first baseman

If it were not for the exercise-induced symptoms, she says she would have never known that she had the disease and could have eventually collapsed from heart failure. "This disease can be caught early with the right treatment and a good cardiologist. It just needs more awareness; I want to help bring that chance to people." Amanda adds, "I don't want to see parents burying their children like **Hank Gathers'** mom or the countless others who have endured this deadly, yet often quiet killer."

Now Amanda wears a heart monitor and exercises sensibly. She still reminisces about having to leave the game she loved. "I sometimes have that dream that I am playing one more time," says Amanda. "But now I have a job I absolutely love, and one of the reasons I found my career and this job, is because of what I went through when I was 15 years old." She also met the love of her life and future husband on the job.

Amanda is positive about the future and about what message she wants to send about living with HCM, "I want people to understand this disease. I want kids and teenagers to see a healthy, 32-year-old woman who is confronting her disease, taking the necessary steps to find the cause, and is still living life to the fullest."

Introducing CCF Teen and Young Adult Ambassador: **MATT PROTAS**



Matt Protas has been appointed as CCF's first **Teen and Young Adult Ambassador**. Matt will use his first-hand knowledge of

living with cardiomyopathy to coach and mentor other teens and young adults struggling to find their way after being diagnosed.

Matt Protas was at the height of his high school wrestling career when he was diagnosed with hypertrophic cardio-myopathy (HCM) and told that he would have to give up competitive sports. At the time, he was 15 and completely devastated. "I was angry and sad because I was an accomplished athlete who could never play sports again because of HCM," remembers Matt. With the support of his family, friends and physician, Matt learned to accept his diagnosis and use it as motivation in his daily life.

Matt is now 21, a physiology and neurobiology major at the University of Connecticut and a licensed emergency medical technician in New Jersey and Connecticut. He hopes to eventually pursue a career in medicine. This summer Matt worked for **Carolyn Ho, M.D.**, medical director at the cardiovascular genetics center at **Brigham & Women's Hospital** in Boston, on two studies focused on preventing or slowing the progression of HCM in asymptomatic individuals that have a genetic mutation for HCM.

"As CCF's Teen and Young Adult Ambassador, I am committed to helping CCF's members, both children and their parents, get through the fear and isolation connected with being diagnosed with cardiomyopathy," says Matt. "The wonderful staff at CCF are doing great work, and I am honored to be involved with such a great organization."

If you or your child would like to speak with Matt, please call Chris Colón at 866.808.2873, ext. 905 and she will put you in touch with Matt.



Q & A

Q: What do patients and families need to know about the new issued guidelines?

A: These guidelines now provide first-time guidance for the medical community—specifically cardiologists, geneticists, and genetic counselors—who are involved in the care of families with inherited heart rhythm (cardiac channelopathies) and heart muscle (cardiomyopathies) diseases. It is important to remember that the diagnostic, prognostic and therapeutic role of the genetic test is very much disease-specific.

Among the cardiomyopathies, the genetic test will have far greater impact for families with hypertrophic cardiomyopathy than families with dilated cardiomyopathy, at least right now. Also, as with the availability of any "new" test in medicine, it will take time for insurance companies to recognize the clinical utility of the genetic test and implement favorable insurance positions towards genetic testing. Therefore, the medical community and the families with these diseases need to continue to lobby that genetic testing is as helpful as almost any of the cardiac tests that we routinely order and which are routinely reimbursed.

Spread Holiday Cheer While **SUPPORTING CCF!**

- ✓ **Buy your gifts online using goodsearch.com or igive.com** and select the "Children's Cardiomyopathy Foundation" as your charity to support. A portion of your purchase will go directly to CCF at no extra charge.
- ✓ **Use CCF's red curebands and other CCF merchandise as fun stocking stuffers.** Download a cureband order form (childrenscardiomyopathy.org/site/merchandise.php) and check out CCF's online merchandise store (cafepress.com/cardiomyopathy).
- ✓ **Ask friends and family to make a donation to CCF in lieu of holiday gifts** or give someone an honorary tribute gift. CCF can customize your holiday message on our "Hope" tribute card.

with

Michael J. Ackerman, M.D., Ph.D.

New Guidelines for Genetic Testing

An international panel of experts from the **Heart Rhythm Society (HRS)** and the **European Heart Rhythm Association (EHRA)** presented new guidelines on cardiovascular genetic testing at the **HRS 32nd Annual Scientific Sessions** in May 2011.

The guidelines, "Expert Consensus Statement on the State of Genetic Testing for the Channelopathies and Cardiomyopathies," review the importance of genetic testing and provide the latest knowledge on screening patients with cardiac conditions at risk for sudden cardiac death. The recommendations, which were published in the August 2011 issue of *HeartRhythm Journal* and *Europace*, focus on genetic testing for 13 inherited cardiac conditions including six channelopathies, five cardiomyopathies, out-of hospital cardiac arrest survivors and post-mortem testing in sudden death cases.

Dr. Michael Ackerman, director of the sudden death genomics laboratory at the **Mayo Clinic** in Rochester, Minn., was co-lead author of the new guidelines. In this issue, he shares with CCF his long-term outlook on genetic testing recommendations and best practices.

Q: When is the ideal time to begin the process of genetic testing for someone with a diagnosis of cardiomyopathy?

A: In general, when a cardiologist has strong clinical suspicion for the presence of an inheritable cardiomyopathy, like hypertrophic cardiomyopathy, then genetic testing for that disease is recommended.

Q: What impact will the new recommendations have on the number of people seeking genetic testing for heart conditions?

A: Hopefully, it will help physicians realize that disease-specific genetic testing for several heart conditions is no longer a research-based test but have matured into clinically available tests with recognized clinical utility.

Q: Should a family with a cardiomyopathy diagnosis ask for genetic testing as part of the affected person's care plan?

A: If they have a cardiomyopathy like hypertrophic cardiomyopathy where the genetic test will be informative to them and to their relatives, families need to be proactive and ask about the genetic test. However, there are other cardiomyopathies, like dilated cardiomyopathy, where the need for genetic testing is not yet clear or convincing.

Q: Should those who received a genetic test in the past be retested again as the knowledge of genetics advances?

A: Yes, if a patient has an inherited cardiomyopathy and underwent research based genetic testing and was given negative results, they should explore repeat testing with a clinically available test. The research-based assays previously performed had about an 85 to 95 percent sensitivity-to-detect rate, meaning that if there truly were a mutation present in one of the known disease genes, those research assays would have missed 5 to 15 percent of those mutations. The current generation genetic tests have about a 99 percent sensitivity-to-detect rate. Importantly, this does not

mean that the current tests detect all mutations. That yield is disease-dependent. For example, with the current generation test, about 50 percent among all patients with hypertrophic cardiomyopathy will be mutation-positive.

Q: The new guidelines suggest it may be helpful to seek out a center experienced in genetic evaluation and family-based management of the specific cardiac conditions to which the guidelines apply. What are some tips for making sure a center is experienced in those areas?

A: Most centers of excellence for inherited cardiomyopathies will be readily apparent by the "sniff" test. In other words, do they smell like a center of excellence for that specific disease? How many patients have they evaluated? Do they offer all the therapeutic interventions for that disease? Does the center perform research studies on pediatric cardiomyopathy and do they publish the results in scientific research articles? Are the doctors known in the field for that disease?

HearttoHeart

Volume 8, Number 2 • Fall 2011 • Winter 2012

Children's Cardiomyopathy Foundation

P.O. Box 547, Tenafly, NJ 07670

Non-Profit Org.

U.S. Postage

PAID

Permit #129

Paramus, NJ



MEMBER SUPPORT SERVICES

CCF offers a variety of ways for members to share information and provide support to one another.

CCF offers a member forum, local support group meetings and toll-free phone sessions.

For more information about these services or the below scheduled events, please contact Chris Colón at ccolon@childrenscardiomyopathy.org.

Local Support Groups

- **North Carolina**

Duke University Medical Center
Durham, N.C.

January 26, 2012 at 6:30 p.m.
February 23, 2012 at 6:30 p.m.
March 22, 2012 at 6:30 p.m.
April 26, 2012 at 6:30 p.m.
May 24, 2012 at 6:30 p.m.

Room 4902, McGovern-Davidson
Children's Health Center

CCF Forum Guest Q&A Sessions

- **Promoting Well-Being in Pediatric Cardiomyopathy**

Victoria Norton, Ph.D.
Rocky River Behavioral Pediatrics
January 16 - 23, 2012

CCF is TOP-RATED HEALTH ORGANIZATION



CCF was recently named as a top-rated health non-profit by America's leading charity evaluators **GreatNonprofits**, **CharityNavigator**, **GuideStar** and **GlobalGiving**.

We thank everyone for their support and wonderful reviews. If you have not already done so, please share your story about CCF on greatnonprofits.org. Every review increases our visibility and helps further our mission.