H.O.P.E. for DUCHENNE MD
Helping Our Parents Explain Duchenne MD

Parent Project Muscular Dystrophy
Leading the Duchenne muscular dystrophy community

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Therapeutics Initiates Clinical Trials of PTC124

PTC124 is a first-in-class, orally delivered, investigational new drug for the treatment of genetic defects resulting from nonsense mutations. Nonsense mutations are single-point alterations "stop signs" in the DNA that halt the translation process, producing a short, nonfunctional protein. PTC124 is a unique drug that allows the cellular machinery to bypass the nonsense mutation and continue the translation process, restoring the full-length functional protein.

It is estimated that 15-20% of boys diagnosed with DMD have this type of mutation. It is easy to dismiss this information if your son has a deletion or duplication in his dystrophin gene, because PTC124 will not be useful in that instance. It is easy to understand how one could feel left out and sad. But, wait – there is a reason we should all be interested, enthusiastic, and hopeful.

Boys with this type of mutation (premature stop codon) who participate in this clinical trial will potentially produce full-length dystrophin in muscle for the first time EVER. For many years, researchers and physicians have been speculating that the production of full-length, functional dystrophin may cause an immune response because the body will look at this protein as foreign; while others have suggested that dystrophin is not foreign, because all boys produce what are referred to as "revertent fibers" (muscle cells with dystrophin, but not in sufficient quantity to be useful or functional.) The reality is that the answer is unknown.

This trial is a critical step for our sons — all of them. If PTC124 is successful, the boys will produce dystrophin. If there is no immune response, we will have answers to this very critical question of whether dystrophin is a foreign protein. If dystrophin production is tolerated and strength is preserved or regained, the doors will be wide open for all strategies that restore dystrophin: exon skipping, viral gene therapy, direct DNA injections, and stem cells. And those strategies will affect every boy.

For more details, please visit our website home page (www.parentprojectmd.org) and look for the RED flag under Research & Treatment News.

More than 280 people make their way to Cincinnati, Ohio to participate in PPMD's 10th Annual Conference

On Thursday, July 8, several parallel meetings were held, all directed toward improving care for our sons. Dr. Doug Biggar convened experts to discuss osteoporosis in DMD. Clearly bone fragility is a concern in DMD, based on diminished mobility as well as the use of steroids. While this was quite apparent, we have more questions: What is the importance of the bone density score in DMD? How important is it to give calcium supplements? When is the appropriate time to give bisphosphonates (Fosamax, Didrocal, and Actonel), and what is the most effective intervention for our sons? You will be hearing more on this issue in the coming months.

Because our sons need and deserve excellent care, because we wish all clinics were "experts" in DMD, and because we wish all clinics were capable of providing support and hope, Dr. Norb Weidner from Children's Hospital Cincinnati conducted a focus group with 15 parents in an effort to understand what types of support services would be most useful and helpful. The real questions are: What types of services are needed? How can we improve care? How can clinics provide more support for families?

We kicked off this year's Annual Conference by providing those new to our conference (or those interested in a review) with broadbased information to prepare them for the intense days to follow. Conference topics covered all aspects of care as well as research, discussing what we can now describe as optimal care. In addition, we held breakout sessions on emotional issues in DMD, learning problems, physical therapy, advocacy, and networking.

Presentations on care were extensive. Physicians agree that boys with DMD need physical evaluation and baseline studies, including an echocardiogram. We need to understand the specific mutation or genetic error in each child. There is general agreement by experts in DMD, that steroids are considered the "gold standard." While there are some questions remaining (e.g., optimal age for intervention and regimen), steroids should be recommended early to boys with DMD. Dr. Jonathan Finder discussed Respiratory Consensus (now in publication) and the benefit of the Cough Assist. All families need access to optimal care.

We all noticed a change in the atmosphere at this conference. The research presentations were intense, overwhelming, and wonderful. Researchers discussed translation and upcoming clinical trials not in a context of "someday soon," but in the context of 2005 and 2006. Dominick Wells, P.h.D. (UK) was asked to summarize the meeting on Saturday afternoon. After several very long, intense days, a certain hush came over the room. Dr. Wells concluded his summary by listing possible clinical trials on the horizon: therapeutic interventions such as BBIC (protease inhibitor), Myodur (calpain inhibitor), Myostatin inhibition (Wyeth), and Pentoxifillin (CINRG); gene repair mechanisms, oligonucleoties (exon skipping), PTC124 (premature stop codon drug) and gene therapy approaches, adeno-associated virus (AAV6 or AAV8) containing a micro dystrophin, and direct DNA injections (Wolff). The possibilities are real and near-term. We are seeing progress and we can feel a change. We are hopeful that these strategies will make a difference for this generation of boys.

On Saturday evening, we celebrated 10 years of PPMD and marked 10 years of working together as a community, 10 years of networking, and 10 years of hope.

On Sunday, we said our goodbyes and left Cincinnati with more hope than ever, whispering a prayer of godspeed for our researchers, our doctors, our representatives, ourselves and our sons.

We are already planning for next year. Mark your calendars: July 7-10, 2005. Westin Hotel — Cincinnati, Ohio.

YOU'RE RUNNING A WHAT?

For the first time ever, Parent Project Muscular Dystrophy (and specifically Duchenne muscular dystrophy) will be represented at the Walt Disney World® Marathon. This exciting endeavor is set to take place January 9th, 2005. PPMD will be one of many charities that are represented, and our runners will be part of thousands striving to reach the finish line. The difference is that we don't run this race for ourselves…we **Run for Our Sons**.

If you haven't heard about the **Run for Our Sons** team that PPMD is organizing for the Walt Disney World® Marathon and Half Marathon, please visit our website at www.parentprojectmd.org for more information.

All details (from race requirements to course descriptions, dates, times, and registration forms) are available for review. There are many ways to participate in the **Run for Our Sons** team, even if you can't run the marathon itself.

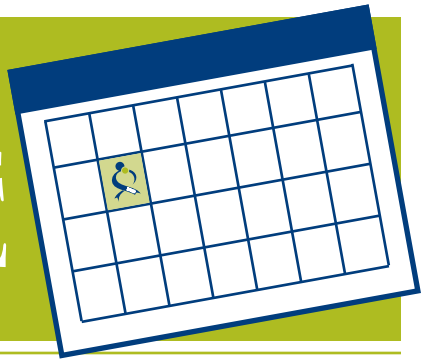
PPMD has a wonderful new and easy way to secure sponsorship through a company called Just Giving (www.justgiving.com/parentprojectmd). You can use this site to create a personal fundraising page to send to friends and family. Check out Kimberly's page (www.justgiving.com/pfp/kimberly) for ideas and to see how effective it has been to date.

There are many rules and regulations for the Walt Disney World® Marathon and Half Marathon, some of which have strict deadlines. If you are interested in being part of the team, please send your registration form to Kimberly (kimberly@parentprojectmd.org) as soon as possible.

Be part of history as PPMD proudly sponsors the RUN FOR OUR SONS.

For more information on participating in the marathon, please contact Kimberly@parentprojectmd.org.

SAVE THE DATE



Parent Project Muscular Dystrophy announces the start of our 2005 calendar. There are some exciting events coming up that we're sure you won't want to miss. Mark your calendars today!

JANUARY 9, 2005 • Run for Our Sons at the Walt Disney World® Marathon in Orlando, Florida. For information on the marathon and ways you can participate, visit our website (www.parentprojectmd.org) or refer to the column in this newsletter.

February 13-19, 2005 • 4th Annual DMD Awareness Week — Your Heart Is A Muscle, Too! Join families from around the world in giving voice to DMD. Look for more details regarding DMD Awareness Week 2005 on our website later this fall.

MARCH 6-8, 2005 • PPMD's Annual Advocacy Conference in Washington, D.C. Participation at PPMD's Advocacy Conference is a great way to help create change for DMD. This event is designed to convince Congress of the need for a much greater investment in muscular dystrophy research. Given the federal budget challenges, it is more critical now than ever that Congress hears from you!

JULY 7-10, 2005 • PPMD's 2005 Annual Conference, Westin Hotel, Cincinnati, Ohio The 2005 Annual Conference will once again be held in Cincinnati ,OH. Join us as top DMD experts from around the world present the latest information on DMD research, treatments, and care options. Look for more details and registration information in early 2005.

FOCUS ON FUNDRAISING

Each year, families from around the country host events to help raise money for the fight against DMD. From golf tournaments to dinner dances, bake sales to bowl-a-thons, fundraising events are a wonderful way to generate funds and create awareness for DMD.

One popular fundraising event is the raffle. There are two raffles currently underway to benefit PPMD that need your support. We know that for many, hosting a fundraiser isn't possible, but supporting the efforts of families like yours is an important way to participate and strengthen the DMD community.

HOUSE OF HOPE RAFFLE

On January 13, 2005, one lucky person will become the proud new owner of a house in Port St. Lucie, Florida. Yes, that's right: a brand new house in Florida for the price of a $20 raffle ticket! The Poole Family of Long Island, New York is thrilled that this idea has taken off and that people are getting involved. Below is a link to the House of Hope website. On the site, you can view a the diagram of the house, read about the raffle, and even purchase tickets. Please help the Poole family and all those affected by DMD by purchasing your House of Hope raffle tickets. www.houseofhopepsl.org

BOMBARDIER QUEST 500 4x4 ATV RAFFLE

On September 8, 2004, someone will ride away on their very own new Bombardier ATV! This raffle is organized by the Herrera family in Montana. Tickets for this raffle are still available for only $10 by calling Ron Herrera at (406) 443-5218. Ron and his wife Nina are long-time supporters of PPMD, and they hope this raffle will help raise funds for DMD research. Supporting their efforts is easy — just call to enter for your chance to win and help make the Herrera's fundraising efforts a success.

PPMD Office News

To better serve the DMD community and increase productivity, PPMD will be consolidating our offices and transferring all data management to the New Jersey office.

Please direct all correspondence (general information, literature requests, conference registrations, fundraising information, and all donations) to the NJ office:

PARENT PROJECT MUSCULAR DYSTROPHY
158 Linwood Plaza, Suite 220 • Fort Lee, NJ 0024
tel: (201) 944-9985 • fax: (201) 944-9987

This restructuring should help PPMD work more efficiently and enable our Executive Director, Pat Furlong, to devote less time to day-to-day management and more time to research and advocacy efforts.

SO WHO IS THE PPMD OFFICE STAFF, AND HOW CAN I REACH THEM?

Pat Furlong is the Founder/President and Executive Director. Pat can still be reached in the Ohio office at (513) 424-0696 or pat@parentprojectmd.org

Kimberly Galberaith is the Associate Executive Director; she can be reached in the NJ office at (201) 944-9985 or kimberly@parentprojectmd.org.

Ryan Fisher will be working full-time in the NJ office as our Office and Database Manager (ryan@parentprojectmd.org). He will work alongside Cecilia Peterson (cecilia@parentprojectmd.org), who will be working part-time with us to help process and track all grass-roots fundraising events.

INTERESTING OFFICE STATISTICS:

DID YOU KNOW THAT OVER THE COURSE OF A WEEK, PPMD…

Receives approximately 350 phone calls • Receives over 300 e-mails

Has between 400 and 500 hits on our website • Sends out an average of 10 information packages

Never forgets how precious time is for our boys.

Nothing is more important to us than the young men who inspire us to fight Duchenne muscular dystrophy (DMD) every day. This year, Parent Project Muscular Dystrophy (PPMD) will be marking 10 years as the leading resource for the DMD community by organizing the creation of a commemorative quilt to celebrate these remarkable young men.

"Celebrating Our Sons" Quilt

THE DEADLINE FOR TURNING IN YOUR QUILT SQUARE IS RAPIDLY APPROACHING: SEPTEMBER 13TH 2004

We are inviting all parents, grandparents, family members, friends, and those diagnosed with DMD around the world to design using the theme, "Celebrating Our Sons." To do so, simply cut out a 5" square of white cotton fabric and let your creative side take over. Designs may be in the form of embroidery, fabric paint, fabric transfers (iron-ons), or anything other method that inspires you.

All finished squares must be delivered to PPMD's Development Office no later than September 13th in order to be included in the final quilt.

Our quilt will be beautifully put together and then displayed on our website for all to see the tapestry of our community. Then in the late fall, we will put the "Celebrating Our Sons" quilt up for auction. All proceeds from the quilt raffle will go toward DMD research.

FINAL SQUARES SHOULD BE MAILED TO:
Cecilia Peterson
c/o Parent Project Muscular Dystrophy
158 Linwood Plaza, suite 220
Fort Lee, NJ 07024

Help us celebrate the diversity, the inspiration and the hope in our DMD community.

HELP US CELEBRATE OUR SONS.

UPDATE ON THE GREAT S.T.I.R. CHALLENGE

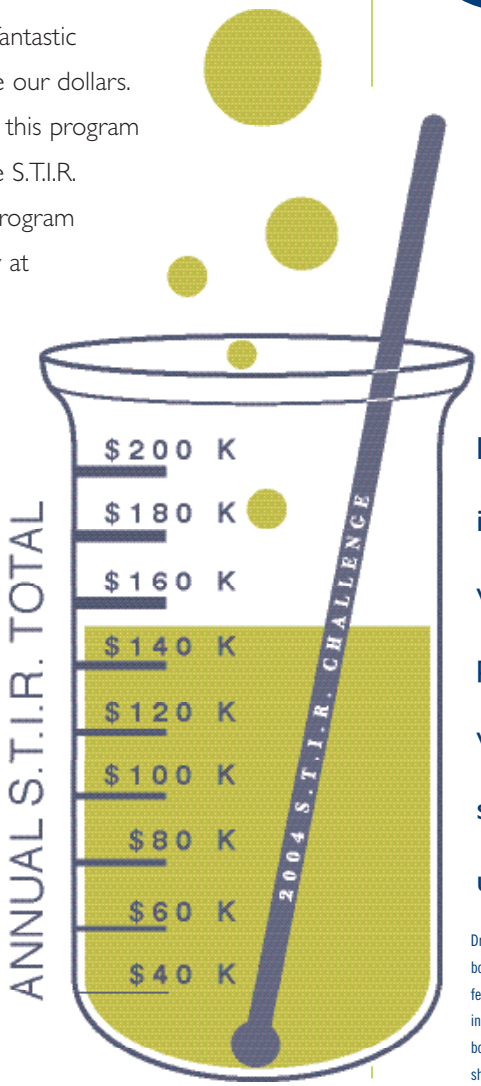
Many of you received a letter encouraging you to participate in PPMD's monthly giving program, S.T.I.R.

You may remember reading about the $50,000 matching gift opportunity and our goal to increase donations and raise $200,000 by the end of 2004 (details online at www.parentprojectmd.org).

Here's an update on where the Great S.T.I.R. Challenge stands:
Since our letter campaign began in late June, enrollment in S.T.I.R. is up almost 30%! Individuals giving between $10 and $1000 a month have helped our annual S.T.I.R. number jump to $151,797! Please help us by joining S.T.I.R. and asking others to join.

This generous matching gift is a fantastic opportunity for PPMD to double our dollars. If you have not yet signed up for this program — of if you would like to receive S.T.I.R. brochures so you can help the program grow — please contact Kimberly at kimberly@parentprojectmd.org

Remember: it takes all of us together, doing whatever we can and participating in so many ways, as we continue Striving To Impact Research!



$151,797 as of 8/15/04. For updates on our progress, please check online at www.parentprojectmd.org.

Q: Why is nutrition so important for boys with DMD?

A:

Craig McDonald and colleagues studied mobility in boys with and without DMD. The study involved wearing pedometers. It was found that on average, boys with DMD moved 40% less than boys without DMD. If we extract this information and try to imagine a diet for boys with DMD, we might think about ways to increase protein and decrease carbohydrates. We are working with nutritionists, and will be making suggestions about diet and nutrition in upcoming months.

Dr. Craig McDonald and colleagues used the SAM (Step Activity Monitor) to quantitatively measure physical activity in boys with Duchenne muscular dystrophy (DMD). Analysis of data reveals that each day, boys with DMD spent 40% fewer minutes than did controls at a higher activity level. The DMD boys spent a greater proportion of time each day in the inactive or sedentary range and the low-activity range as compared with age-matched boys (controls). The boys with DMD also took significantly fewer steps per day at a high activity level than did controls. These findings show that boys with DMD had a marked decrease in high levels of physical activity at a young age.