WE SEE STRENGTH...
WE SEE STRENGTH IN...
CONNECTING.

“I define connection as the energy that exists between people when they feel seen, heard, and valued; when they can give and receive without judgment; and when they derive sustenance and strength from the relationship.”

- Brené Brown

Welcome to our nation’s capital and to my favorite time of year – PPMD’s 21st Annual Connect Conference!

I feel like every year we tell you it’s the biggest conference we’ve had, but that’s because every year it grows more and more comprehensive as the Duchenne landscape grows more and more vast.

This year we will hear from over twenty industry partners, dozens of researchers and scientists, and an array of care providers covering the topics that concern you most. We welcome our pre-conference Transitions Meeting and invite you and your family to attend the open sessions. We are excited to host the east coast premiere of the wonderful documentary Anthony’s Adventure about Jill Castle’s son Anthony and their family’s trek to the Grand Canyon. Kids Track returns with our craziest, most action-packed program ever, including mad scientists. And, we’ll have robotics Sunday morning! That’s right, our agenda is so big this year that we have a packed day on Sunday, including an up-close look at robotic technology being developed to assist people with Duchenne.

But more important than any of this is how PPMD’s Annual Connect Conference connects us all. No one understands what you and your family are dealing with better than other people on the same journey as you. This community, over the last 20+ years, has been my sustenance, my courage, and my strength.

You’ve been there to celebrate with me, you’ve been there to wipe away my tears.

And you’ve supported PPMD by attending our Annual Connect Conference, by making donations, by running in races, and hosting events. You’ve supported PPMD every time you’ve reached out to another family dealing with Duchenne and offered them advice, kindness, friendship.

This is an incredible time in Duchenne research. We can see the finish line approaching on several therapies—and yet the closer we get, the higher the highs and lower the lows seem to be. It’s during these extremes when our support for each other, our empathy is perhaps the most critical.

We are a community of fiercely passionate and brave families fighting to end Duchenne. We see strength in each other. We see strength in YOU.

Thank you for attending this year’s conference and I can’t wait spend the next several days with you and your family!

Warmly,

Pat Furlong, PPMD President
NEW AT THIS YEAR’S CONNECT CONFERENCE

Every year PPMD strives to bring you the latest research, advocacy, and care updates. This year, we have expanded our already packed agenda with new events that will educate and inspire you and your family.

Duchenne/Becker Transitions Meeting

Wednesday, 2:00pm–7:30pm • Lincoln 3/4
Thursday, 9:00am–12:00pm • Thurgood Marshall Ballroom

This year, PPMD’s Annual Connect Conference includes a meeting track dedicated exclusively to adults with Duchenne and Becker. Presenters will specifically address topics of interest to teens and adults: driving, independent living, home modifications, life after college, dating and family life, managing personal care attendants, robotics as mobility options, navigating ‘the system’, and more. This meeting will also have a strong tone of self-advocacy and empowerment as we work to ensure that we transform this ‘moment into a movement.’ Wherever you are on your Duchenne journey, transition issues are important and early planning helps.

NOTE: Portions of the Transitions meeting will be open and welcome to all Connect Conference attendees ages 14 and older (parents included). Other segments of the Transitions meeting will be open only to adults and teens living with Duchenne/Becker ages 14 and older (closed to parents). Specific agenda details can be picked up at Connect Conference Registration.

Welcome Reception
Poster Session & Vendor Fair

Thursday, 6:30pm • Expo Hall C

Please join us for appetizers and information! This year’s welcome reception will feature dozens of posters from researchers around the world working in the Duchenne field, in addition to numerous vendors offering products and services for the Duchenne community.

Additional Vendor Fair hours:
- Thursday, 11:00am–1:00pm and 5:00pm–9:00pm
- Friday, 7:00am–2:00pm

NOTE: For a full list and map of vendors and poster presentations, please pick up a handout at Connect Conference Registration.
End Duchenne Rally & Hill Day

Thursday, 5:40pm–6:30pm • Thurgood Marshall Ballroom
Friday, 9:00am–12:30pm • Off-site (Capitol Building Steps)

With this year’s Annual Connect Conference in Washington, DC, PPMD wanted to take advantage of being in our Nation’s Capital—and ensure that our Duchenne community leaves an indelible imprint on federal policymakers. Friday morning of the conference will kick off with an End Duchenne Rally at the foot of the Capitol, followed by an opportunity for all attendees to have one meeting with the office of a Member of Congress. For those attendees who signed up in advance, you will be trained and provided all the tools needed Thursday afternoon for a fun and successful Friday morning.

Let’s ensure that anyone within eyesight of the U.S. Capitol on June 19th knows that it is time to End Duchenne—join us for our End Duchenne Rally & Hill Day!

Schedule:
• Advocate Training for Hill Visits
  Thursday, 5:40pm–6:30pm (Thurgood Marshall Ballroom)
• End Duchenne Rally & Hill Day
  Friday, 9:00am–12:30pm (Off-site: Capitol Building Steps)

NOTE: If you would like to attend and did not register in advance, please speak to someone at Connect Conference Registration. Directions to Capitol Hill from the hotel will be given out during training. Please note the additional registration fee for those attendees planning on joining this important effort and being part of the Hill visits helps to offset the cost of scheduling and will also provide you with an End Duchenne t-shirt to be worn by all advocates.

Kids Track: Mad Science & Sib Shop Program

Friday, 3:30pm–5:00pm • Lincoln 3/4
Saturday, 9:30am–12:30pm • Lincoln 3/4
Saturday, 2:30pm–5:00pm • Hoover (Kids), Lincoln 3/4 (Sib Shop)

One of our most exciting and fast-growing programs at the Connect Conference is our Kids Track! This year, we have expanded our Kids Track to give those attending more creative outlets to express themselves.

This year, the program has a “Weird Science” theme and includes:
• One and a half hour session the afternoon of Friday, June 19 (Lincoln 3/4)
• Three hour session and lunch the morning of Saturday, June 20 (Lincoln 3/4)
• A special “Sib Shop” session the afternoon of Saturday, June 20 (Lincoln 3/4), or
• The afternoon session of Kids Track on Saturday, June 20 (Hoover)

During these sessions there will be a wacky & weird science entertainer, special group games, crafts and activities (broken into 3 age groups; minimum age is 5 or kindergarten), and a very special team of teen robot inventors who will inspire the kids with their cool technology and robot building & battling skills.

Kids Track registration also includes a free t-shirt, lunch on Saturday, and dinner Saturday night.

NOTE: Minimum age is 5 or kindergarten. If you did not register your child ahead of time, please speak to someone at Connect Conference Registration.
East Coast Premiere of “Anthony’s Adventure”  
Friday, 8:00pm • Thurgood Marshall Ballroom  
Last May, a group of 30+ volunteers carried Anthony Castle to the bottom of the Grand Canyon. A 14-year-old with an infectious smile and Duchenne muscular dystrophy, Anthony warmed hearts around the world with his story of courage and determination. While many people thought Anthony’s bucket list item to put his feet in the Colorado River was too ambitious, the nonprofit Daring Adventures, friends, and family gathered support to make his dream come true. Anthony and his family demonstrate incredible strength and determination—focusing on what CAN be done instead of what can’t.

“Anthony’s Adventure,” is an uplifting story with an empowering message of the importance of living in the present and appreciating life. Prepare to be inspired!

NOTE: This film is appropriate for all ages!

New Robotics Session  
Sunday, 9:15am–11:45am • Thurgood Marshall Ballroom

PPMD is excited to announce an entire session at this year’s Connect Conference dedicated to our Robotics Initiative. Traditionally PPMD’s Connect Conference has wrapped up on Sunday morning. But this year we are taking advantage of the time we have on Sunday to bring you a look at some of the exciting robotic technology PPMD is funding, as well as other technologies we are considering at various stages of development. Not only will you hear from the engineers behind these devices, you will get an up close look at what they are and how they work.
PANELS AT-A-GLANCE

Keeping Your Family Intact: Surviving the Diagnosis
Thursday, 2:20pm–3:00pm
During this panel discussion moderated by Mary-Lou Weisman, author of Intensive Care: A Family Love Story, hear first-hand accounts & perspectives from different family members when it comes to “making it work.”

Combination Therapies: Ultimately Treating Duchenne
Saturday, 8:00am–9:15am
This panel discussion will take a deeper look at combination therapies: the opportunities, hurdles, and goals for ultimately treating Duchenne.

Different Together: Explaining Phenotype Variability in Duchenne and its Implications in Clinical Trials
Saturday, 5:00pm–6:00pm
This panel session will examine the differences in Duchenne; why cases progress differently.

PPMD’s Robotics Program
Sunday, 9:15am–11:45am
Meet innovators looking at robotics technology for the Duchenne community. Each panelist will present on their technology and then will participate in an open discussion. After a robust Q&A session, we will ask each of the panelists to break apart in the room so that attendees can visit each panelist and examine their technology first hand.
IMPACTING THE DUCHENNE DRUG DEVELOPMENT PIPELINE

PPMD has learned that successful therapy development requires a commitment, not just to funding individual drug projects, but also to improving the entire ecosystem surrounding trials—from decreasing the variability of clinical care, to drafting regulatory guidelines with the FDA, to developing tools that decrease the time and cost of conducting trials.

You can help us help accelerate this process. Through donations, advocacy, trial participation, survey participation, and staying active in this community, you give PPMD strength and support our comprehensive approach to end Duchenne.

It’s easy to see from this chart—Duchenne is complex. But it is no match to the passion, determination, and strength of this community.
# CLINICAL TRIAL FAQs

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# PPMDCONNECT
Becker Natural History Study
Becker Muscular Dystrophy: A Natural History Study to Predict Efficacy of Exon Skipping

- **What stage is this research?**
  This study is actively recruiting participants.

- **What is the goal or purpose of this study?**
  This is a natural history study to characterize the Becker muscular dystrophy clinical presentation and also to provide information regarding the possible effectiveness of a therapy currently in development for Duchenne muscular dystrophy (exon skipping). Researchers will correlate specific genetic changes with a range of clinical outcomes including physical development, mental development, and quality of life in patients with Becker. The observed variability between individuals with Becker will be studied to deepen our understanding of molecular mechanisms relevant to the optimization of exon skipping therapeutic approaches, as well as to optimize study designs and outcome measures for future clinical trials in Becker.

- **Who is funding this study?**
  This study is funded by the National Institutes of Health (NIH).

- **Who is eligible to participate in this study?**
  To participate in this study you must be a male with Becker, age 4 and above, and have an in-frame dystrophin gene deletion where the boundaries of the mutations are confirmed.

- **What do I have to do if I decide to participate in this study?**
  At each study visit you will have a physical and neurological exam by a study physician, and review your health medication, and cardiac history. You will have strength, function, and breathing testing performed by a physical therapist, and you will also complete quality of life questionnaires. At your first study visit you will also have a blood draw and a skin biopsy, and adults will have the option to have a muscle biopsy.

- **Where does this study take place?**
  This study will be run at select Cooperative International Neuromuscular Research Group (CINRG) network sites and affiliates. The participating centers include:
  - University of Florida in Gainesville, FL
  - Newcastle University in Newcastle upon Tyne, United Kingdom
  - Children’s Healthcare of Atlanta/Emory University in Atlanta, GA
  - Penn State Hershey Medical Center in Hershey, PA
  - Duke University in Durham, NC
  - Centro Clinico Nemo in Milan, Italy
  More sites may be added in the future so be sure to check www.clinicaltrials.gov or the website for the CINRG group: www.cinrgresearch.org for updated site lists, as well as specific contact information at each participating site.

- **How many visits to the study site are necessary?**
  There will be a total of 4 visits — a baseline evaluation and 3 annual follow-up visits over a 3-year period.

- **Can any visits be done locally?**
  No, they must be done at a participating CINRG or affiliate center.

- **Is there any funding to help pay for travel?**
  The study does not provide any funds for travel; however please inquire at each individual site as some CINRG sites may have alternative resources for travel reimbursement or assistance.

- **Will I get paid for participating in this study?**
  No, you will not be paid for your participation in this study. You will not be charged for additional tests and procedures that are performed only because you are participating in this research. Your responsibility to pay for other medical treatment will not be changed by your participation in this study.

- **Why should I consider participating in this study?**
  While no personal benefit can be guaranteed by participating in this study, there are other benefits, including allowing you to play an active role in your own health care (or that of your child), gaining access to medical specialists that are normally not available to you or your child, and helping others by contributing to the better understanding of Becker.

- **Where can I learn more about this study?**
  You can learn more about this study at www.clinicaltrials.gov (NCT01539772) and www.cinrgresearch.org.
  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Cough in Duchenne and Becker
Peak Cough Flow and Cough Clearance in Duchenne or Becker Muscular Dystrophy

What stage is this research?
This study is actively recruiting boys and young men with Duchenne or Becker muscular dystrophy.

What is the goal or purpose of this study?
The goal is to determine whether physiologic measures (peak cough flow, measures of respiratory muscle strength including MIP, MEP, SNIP, and spirometry) can predict the ability to clear the airways of secretions with cough (spontaneous cough clearance, as measured by a nuclear medicine study) in children with neuromuscular disease. It will also determine whether airway clearance is augmented by high frequency chest wall oscillation.

Who is sponsoring this study?
This study is sponsored by the University of Pittsburgh and Respirtech, Inc.

Who is eligible to participate in this study?
Researchers are recruiting boys and young men with Duchenne or Becker who are between the ages of 6 years to 21 years old. Participants must not need mechanical ventilation during the day.

What do I have to do if I decide to participate in this study?
If you and your child decide to take part in this study, your child would be asked to first perform several pulmonary function tests (spirometry and measurement of respiratory muscle strength and peak cough flow). Next, subjects would inhale a radioactive “tracer” that can be seen on a special imaging camera. Your child would then be asked to cough several times over the next several minutes. Following this, they would wear a special oscillating chest vest for 30 minutes, during which they would be asked to periodically cough. The study typically lasts about 3 hours in total.

Where does this study take place?
This study is taking place at the Children’s Hospital of Pittsburgh in Pittsburgh, PA.

Will I get paid for participating in this study?
Yes, families will be compensated to thank them for their time. Families will also be reimbursed for mileage costs.

Why should I consider participating in this study?
While no personal benefit can ever be guaranteed by participation in a study, there are other benefits, including allowing you to play an active role in your own health care (or that of your child), gaining access to medical specialists that are normally not available to you or your child, and helping others by contributing to the better understanding of Duchenne.

Where can I learn more about this study?
For more information, please contact Paul Rebovich, MS at 412-692-5873, or paul.rebovich@chp.edu.
You can learn more about this study at www.clinicaltrials.gov (NCT02034305).
Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Deflazacort
A Pharmacokinetic Study of Oral Deflazacort in Children and Adolescents with Duchenne

- **What stage is this research?**
  This Phase 1 clinical trial is actively enrolling participants.

- **What is the goal or purpose of this study?**
  The main purpose of this study is to characterize the single-state and steady-state dosing of oral deflazacort in pediatric and adolescents patients with Duchenne.

- **Who is the sponsor of this study?**
  Marathon Pharmaceuticals (www.marathonpharma.com) is the sponsor of this study.

- **What are the inclusion (enrollment) criteria for this trial?**
  Participants need to be boys with Duchenne who are between the ages of 4–16 years old. Participants must not have received deflazacort within 30 days, or previously discontinued deflazacort due to an intolerable reaction. Additional details regarding the inclusion and exclusion criteria are posted on www.clinicaltrials.gov.

- **What do I have to do if I decide to participate in this study?**
  Participants will receive oral deflazacort administered once daily at 0.9 mg/kg for eight days. Participants may then enter a long-term extension study, at which point treatment will be continued at the same dose and frequency until such time that deflazacort becomes commercially available or the study is terminated.

- **Where does the trial take place?**
  There are currently 4 sites in the US: Los Angeles, CA; Chicago, IL; Rochester, NY; and Salt Lake City, UT. Please check www.clinicaltrials.gov for details regarding these sites and how to contact the study coordinator.

- **Why should I consider participating in this study?**
  While no personal benefit can ever be guaranteed by participation in a clinical trial, there are other benefits, including allowing you to play an active role in the health care of your child, gaining access to medical specialists that are normally not available to your child, and helping others by contributing to the better understanding of Duchenne.

- **Where can I learn more about this study?**
  www.clinicaltrials.gov (code NCT02251600)
  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
DP ARF Ultrasound
Double Push Acoustic Radiation Force (DP ARF) Ultrasound for Monitoring Degeneration in Duchenne Muscular Dystrophy

• What stage is this research?
  This study is actively recruiting participants.

• What is the goal or purpose of this study?
  This is a pilot clinical trial to assess the ability of a new ultrasound-based imaging method, Double-Push Acoustic Radiation Force (DP ARF) ultrasound, to monitor the progression of Duchenne muscular dystrophy.

• Who is sponsoring this study?
  This study is sponsored by the University of North Carolina, Chapel Hill, in collaboration with the National Institute of Neurological Disorders and Stroke (NINDS).

• Who is eligible to participate in this study?
  The study population will include 30 boys with Duchenne, enrolling at ages 5 to 10 years. To be considered for enrollment, boys must have a clinical onset of Duchenne by age 5, and must have the ability to stand, either alone or with assistance. Male siblings with no known neuromuscular disorders, ages 5 to 10 years, are also eligible for enrollment as controls.

• What do I have to do if I decide to participate in this study?
  All boys will be imaged 3 times annually for 4 years. In addition to DP ARF imaging every 4 months, the boys will undergo standard quantitative muscle testing (QMT) and timed function tests (TFT) of time to standing, 6-minute walk, and 30-feet walk. Age at loss of ambulation will also be recorded for each boy.

• Where does this study take place?
  This study takes place at the University of North Carolina, Chapel Hill (UNC-CH).

  • How many visits to the study site are necessary, and can any visits be done locally?
    » A total of 12 visits – 3 per year for 4 years. All visits must be at UNC-CH.

  • Is there any funding to help pay for travel?
    » Yes, this study provides reimbursement for air travel (for the patient and an accompanying adult) and car travel (by the mile). Hotel accommodations in Chapel Hill are also provided.

• Will I get paid for participating in this study?
  No, there is no payment for participating, other than the funding to help with travel costs.

• Why should I consider participating in this study?
  While no personal benefit can ever be guaranteed by participation in a study, a potential indirect benefit may be the satisfaction of being a part of the advancement of medical science to improve monitoring and treatment of Duchenne and the other 30+ types of muscular dystrophies in children and adults.

• Where can I learn more about this study?
  You can learn more about this study at ClinicalTrials.gov (NCT01506518) and http://dmdultrasound.bme.unc.edu. Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Duchenne Natural History Study
Longitudinal Study of the Natural History of Duchenne Muscular Dystrophy

• What stage is this research?
The study is actively recruiting Duchenne muscular dystrophy and healthy control participants.

• What is the goal or purpose of this study?
The purpose of this study is to establish the largest long-term assessment of people with Duchenne muscular dystrophy. In this study, the investigators associated with the Cooperative International Neuromuscular Research Group (CINRG) will take a detailed look at Duchenne participant’s physical abilities, the medical problems they experience, and how they use health care services. Physical abilities will be compared to a group of healthy controls.

The second purpose of this study is to find out whether small, normal differences in the genetic makeup of people with Duchenne (called “single nucleotide polymorphisms” or “SNPs”) affect how their disease progresses and relates to muscle strength/size and steroid response.

The third purpose of this study is to study genetic variations associated with Duchenne.

The final purpose of this study is to determine whether certain biomarkers are present in people with Duchenne and not in healthy controls.

• Who is sponsoring this study?
The study is sponsored by the U.S. Department of Education, National Institutes of Health (NIH), and Department of Defense. Parent Project Muscular Dystrophy is funding the study of the New Young Duchenne Cohort.

• Who is eligible to participate in this study?
Researchers are now recruiting for the New Young Duchenne Muscular Dystrophy Cohort. 100 male Duchenne participants, age 4 – 7 years old, are needed and we have 17 slots currently available. The diagnosis of Duchenne must be confirmed by genetic test and/or muscle biopsy.

Researchers are now recruiting for the Healthy Control Cohort. 120 male healthy controls, ages 6–17 years old, are needed and we have 25 slots currently available.

• What do I have to do if I decide to participate in this study?
Duchenne Muscular Dystrophy Cohort: At each study visit you will have a physical and neurological exam by a study physician, and your health medication will be reviewed. You will have strength, function, and breathing testing performed by a physical therapist, and you and/or your parent/legal guardian will also complete quality of life questionnaires. Optional saliva and/or blood samples will be collected for genetic and/or biomarker (markers that may help us understand more about Duchenne) analysis.

Healthy Control Cohort: At each study visit you will have strength and function testing completed. Optional blood samples will be collected for biomarker (markers that may help us understand more about Duchenne) analysis.

• Where does this study take place?
The study is running at 20 participating centers of the Cooperative International Neuromuscular Research Group (CINRG) network. The participating CINRG centers include 11 sites in the US and additional sites in Canada, Argentina, Italy, Sweden, India, Israel, and Australia. More site and study updates are available so be sure to check www.clinicaltrials.gov or the website for the CINRG group: www.cinrgresearch.org for additional information.

• How many visits to the study site are necessary?
  » There will be up to 12 visits for Duchenne muscular dystrophy participants.
  » There will be 2 visits for typically developing controls.

• Can any visits be done locally?
  » No, they must be done at a participating CINRG center.

• Is there any funding to help pay for travel?
  » The study does not provide any funds for travel; however please inquire at each individual site as some CINRG sites may have alternative resources for travel reimbursement or assistance.

• Will I get paid for participating in this study?
No, you will not be paid for your participation in this study. You will not be charged for additional tests and procedures that are performed only because you are participating in this research. Your responsibility to pay for other medical treatment will not be changed by your participation in this study.

• Why should I consider participating in this study?
While no personal benefit can ever be guaranteed by participation in a study, there are other benefits, including allowing you to play an active role in your own health care (or that of your child), gaining access to medical specialists that are normally not available to you or your child, and helping others by contributing to the better understanding of Duchenne.

• Where can I learn more about this study?
You can learn more about this study at www.cinrgresearch.org and www.clinicaltrials.gov (NCT00468832).
Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Eteplirsen (PROMOVI/Study 4658-301)
An open-label, multi-center, 48-week study with a concurrent untreated control arm to evaluate the efficacy and safety of eteplirsen in duchenne muscular dystrophy

• What stage is this research study?
  PROMOVI (study 4658-301) is a Phase 3 study, currently recruiting participants at clinical sites in the United States.
  
• What is the goal or purpose of this research?
  This Phase 3 study is being conducted to evaluate the safety and efficacy of eteplirsen, Sarepta’s investigational exon 51 skipping therapy. The data collected in this study will build upon over three years of data from the company’s ongoing Phase 2b clinical study.
  
• Who is sponsoring this study?
  The study is sponsored by Sarepta Therapeutics.
  
• Who is eligible to participate in this study?
  Researchers are recruiting approximately 80 males with Duchenne amenable to exon 51 skipping for the treated group and approximately 80 males with Duchenne not amenable to exon 51 skipping for the untreated group.
  Key inclusion criteria include but are not limited to:
  • Ambulatory male with Duchenne, aged 7 – 16 years old
  • TREATED GROUP: Deletion mutation amenable to exon 51 skipping
    * Deletions amenable to exon 51 skipping include, but are not limited to, deletions of exons 45-50, 47-50, 48-50, 49-50, 50, 52, and 52-63.
  • UNTREATED GROUP: Deletion mutation not amenable to exon 51 skipping**
    ** A patient with a deletion amenable to skipping a single exon (i.e., 53, 45, 50, 6, 8, 23) or to skipping double exons (2 exons) may be eligible. Participation is not limited to specific deletions. Patients who do not have out-of-frame mutations and are not amenable to any potential exon-skipping therapy are not eligible to participate.
  • Stable dose of corticosteroids for at least six months
  • Stable pulmonary and cardiac function
  Additional criteria apply and will be reviewed with patients during the screening process. The Principal Investigator (study doctor) determines whether a patient meets the inclusion and exclusion criteria for the study, and therefore whether or not the patient is eligible to participate.
  
• Where does this clinical study take place?
  This study is being conducted at clinical sites in the United States. To view participating study sites, including those that are actively recruiting patients, visit www.clinicaltrials.gov (Identifier NCT02255552).
  
• Will all boys enrolled in this trial receive treatment with the investigational therapy?
  This study will include a group of patients amenable to exon 51 skipping who will receive treatment with the investigational exon 51 skipping therapy eteplirsen (the treated group). The study will also include a group of patients not amenable to exon 51 skipping who will not receive treatment with the investigational exon 51 skipping therapy (the untreated group).
  
• Why is Sarepta including an untreated group of patients in this study?
  Patients in the untreated arm will serve as a control arm to patients in the treated arm. Patients enrolled in the untreated and treated arms of this clinical study will perform similar physical assessments and procedures throughout the study (e.g., 6MWT). The information collected from these assessments and procedures will then be compared between the treated and untreated groups to help evaluate the safety and efficacy of eteplirsen.
  
• Why should I consider participating in this study?
  While no benefit can be guaranteed from participation in any clinical study, we believe you may:
  • Have access to highly experienced clinicians with strong expertise in treating Duchenne, who might not normally be accessible to you and your family
  • Receive notification and knowledge of future clinical studies for which you or your son may be eligible
  • Gain better understanding of how the patient’s Duchenne is progressing
  • Have the opportunity to become more familiar with what participation in a clinical study entails
  • Contribute to what is now known about Duchenne progression
  • Have the opportunity to help others by contributing to medical research that may accelerate the development of Duchenne therapies
  Choosing to participate in a study is an important personal decision. Talk with your doctor and family members or friends about deciding to join a study.
  
• Will I be compensated for participating in this study?
  Generally, reasonable costs associated with participation in the study will be pre-paid and/or reimbursed in accordance with the approved travel policy for the study. Additional information will be provided by individual study sites.

(continued on p. 14)
If enrolled, what can I expect during the study?
The principal investigator (study doctor) and/or the study site contact will review study requirements with patients during the screening process for both arms of the trial.

Patients enrolled in the treated group will visit study sites for dosing, functional assessments, medical testing, and two biopsies over the course of the year-long study. Dosing visits will occur weekly and functional assessments will be conducted quarterly.

Patients enrolled in the untreated/control group will not receive the investigational therapy eteplirsen, and will visit study sites up to 12 times over the course of the year-long study for functional assessments and other medical tests. Biopsies will not be required from this group, though minimally invasive procedures (i.e., blood draws) will occur.

Safety, including adverse event monitoring and routine laboratory assessments, will be continuously monitored for all patients.

Where can I learn more about this clinical study?
To learn more about this study, you or your doctor may contact the study research staff using the contacts posted on www.clinicaltrials.gov (Identifier NCT02255552). You may also email trialinfo@sarepta.com or register on www.skipahead.com to receive information from Sarepta, including updates on our clinical studies.
Eteplirsen (Study 4658-204)
An open-label, multi-center study to evaluate the safety and tolerability of eteplirsen in patients with advanced stage Duchenne muscular dystrophy

- **What stage is this study?**
  Study 4658-204 is a Phase II study that is ongoing, but no longer recruiting participants.

- **What is the goal or purpose of this research?**
  This Phase II open-label study is being conducted to explore safety and tolerability of Sarepta’s potential exon 51 skipping therapy eteplirsen in patients with advanced stage Duchenne muscular dystrophy (Duchenne) who are amenable to exon 51 skipping. Exploratory objectives include the effect of eteplirsen on pulmonary function tests (PFTs) and other functional clinical measures.

- **Who is sponsoring the study?**
  The study is sponsored by Sarepta Therapeutics.

- **What were the eligibility criteria to participate in this study?**
  Researchers recruited males with Duchenne amenable to exon 51 skipping.
  Key inclusion criteria included but were not limited to:
  - Male 7 - 21 years of age
  - Diagnosis of Duchenne with a mutation that is amenable to exon 51 skipping, confirmed by a genetic report
  - Stable dose of oral corticosteroids for at least 24 weeks or has not received corticosteroids for at least 24 weeks
  - Non-ambulatory, or incapable of walking ≥300 meters on the 6-Minute Walk Test (6MWT)
  - Score of ≤4 on the Brooke Score for Arms and Shoulders
  - Stable cardiac and pulmonary function
  - Use of contraceptives for sexually active males throughout the study
  - Willing to provide consent and comply with the study
  Additional criteria applied and were reviewed with patients during the screening process. The Principal Investigator (study doctor) determined whether a patient met the inclusion and exclusion criteria for the study, and therefore whether or not the patient was eligible to participate.

- **Where is the study taking place?**
  This study is being conducted at clinical sites in the following US states: California, Iowa, Maryland, Massachusetts, Missouri, New York, Ohio, and Washington. To view sites conducting the study, visit [www.clinicaltrials.gov](http://www.clinicaltrials.gov) (Identifier NCT02286947).

- **Are all boys enrolled in this trial receiving treatment with the investigational therapy eteplirsen?**
  All enrolled patients are receiving weekly infusions with the investigational exon 51 skipping therapy eteplirsen.

- **What are the participants doing in the study?**
  Patients are receiving once weekly intravenous (IV) infusions of 30 mg/kg eteplirsen for up to 96 weeks. An extension to the dosing period may be considered prior to the end of the 96-week planned dosing period. Safety is regularly assessed throughout the study via the collection of adverse events (AEs), laboratory tests, electrocardiograms (ECGs), echocardiograms (ECHOs), vital signs, and physical examinations. Exploratory efficacy assessments including PFTs, upper extremity testing, and other functional measurements occur every 12 weeks over the first year of treatment and approximately every 24 weeks over the second year of treatment.

- **Are there any preliminary results available from this study?**
  The study is ongoing and no results are available at this time.

- **Where can I learn more about this clinical study?**
  To view additional information about this study, you may visit [www.clinicaltrials.gov](http://www.clinicaltrials.gov) (Identifier NCT02286947). You may also email trialinfo@sarepta.com for more information.
Follistatin Gene Transfer – Becker
Follistatin Gene Transfer to Patients With Becker Muscular Dystrophy and Sporadic Inclusion Body Myositis

• What stage is this research?
This Phase 1 trial is enrolling participants by invitation only.

• Where is this research being performed and who is funding this research?
This research is being led by Dr. Jerry Mendell at Nationwide Children’s Hospital Research Center in Columbus, Ohio. It is being funded by PPMD’s GIFTED Program.

• What is the goal or purpose of this research?
Follistatin is a muscle growth-stimulating protein. This research is intended to build upon preliminary studies in mice with muscular dystrophy and in non-human primates which demonstrated that the follistatin gene, when injected into muscles, can cause significant increases in the size of injected muscles and improvements in the strength of injected muscles. If successful, the investigators can potentially prolong a patient’s ability to walk.

The gene will be carried into the muscle by a virus called adeno-associated virus (AAV). This virus occurs naturally in muscle and does not cause any human disease.

• Who is eligible to participate in this study?
Again, this study is recruiting patients by invitation only. Adult male Becker patients (>18yo) with a proven mutation of the dystrophin gene and continued ambulation after age 15 years old are being recruited. Participants must have identifiable atrophy of the quadriceps muscle with muscle weakness ≥2 standard deviations below predicted using quantitative muscle testing. This study is also recruiting patients with sporadic inclusion body myositis.

• What do participants have to do in this study?
Participants with either of these diseases will have shots of the follistatin gene injected directly into their thigh muscle on one or both legs (one time only). One hundred and eighty days following the gene delivery, participants will undergo testing to see if their muscle strength has improved and muscle biopsy to look closely at the muscle to see if the muscle fibers are bigger. Between the time of the gene transfer and the muscle biopsy, participants will be carefully monitored for any side effects of the treatment. This will include an MRI of the thigh muscle before treatment and at day 180 following treatment. Blood and urine tests, as well as physical examination will be done on the participants during the screening visit and several times throughout the study to make sure that there are no side effects from the gene injections.

• What are the future plans for this research?
If this study is successful, the investigators will expand the research to a Phase 2 study and will also make plans to test it in patients with Duchenne muscular dystrophy.

• Where can I learn more about this research?
You can learn more about this research at www.nationwidechildrens.org/center-for-gene-therapy and www.clinicaltrials.gov (NCT01519349).

Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Follistatin Gene Transfer – Duchenne
Clinical Intramuscular Gene Transfer of rAAV1.CMV.huFollistatin344 Trial to Patients With Duchenne

• What stage is this research?
  This Phase 1/2 trial is enrolling participants by invitation only.

• Where is this research being performed and who is funding this research?
  This research is being led by Dr. Jerry Mendell at Nationwide Children’s Hospital Research Center in Columbus, Ohio.

• What is the goal or purpose of this research?
  The proposed clinical trial is an outgrowth of the safety record and functional improvement seen in the Becker follistatin gene therapy trial. Follistatin is a muscle growth-stimulating protein. In this study, the investigators propose to inject the follistatin gene into muscle using a virus called adeno-associated virus (AAV1.CMV.huFS344). This virus occurs naturally in muscle and does not cause any human disease. If successful, the investigators can potentially prolong a patient’s ability to walk.

• Who is eligible to participate in this study?
  This study is recruiting patients by invitation only. Six male Duchenne patients, age 7 years or older, with a proven mutation of the dystrophin gene are being recruited. Participants must have impaired muscle function based on clinical evidence, and they must be on stable dose of prednisone for three months at time of enrollment or be started on oral dose of daily prednisone regimen for 30 days prior to gene transfer.

• What do participants have to do in this study?
  The viral vector will be delivered to the legs of 6 Duchenne patients via multiple, direct intramuscular injections of rAAV1.CMV.huFollistatin344. The number of injections per muscle will depend on the size of the patient. A total dose of 2.4E12 vg/kg (1.2E12 vg/kg/limb) will be delivered. This dose will be divided between gluteal muscles, quadriceps and tibialis anterior. This is a wider distribution of vector than given to Becker patients, who overall improved the distance walked on the 6MWT without adverse events related to viral transduction into a single muscle.

  The primary objective of this study is safety. Endpoints will include hematology, serum chemistry, urinalysis, immunologic response to rAAV1 and follistatin, and reported history and observations of symptoms. Efficacy measures will be used as secondary outcomes and include the distance walked on the 6MWT, functional tests by PT, life quality questionnaire, MRI, EIM, and muscle biopsy. Participants will have follow up visits on days 7, 14, 30, 45, 60, 90, 180 and 9, 12, 18 and 24 months post-gene transfer.

• Why should I consider participating in this study?
  While no personal benefit can be guaranteed by participating in this trial, there are other benefits, including allowing you to play an active role in your own health care (or that of your child), gaining access to new research treatments before they are widely available and having access to medical specialists that are normally not available to you or your child, and helping others by contributing to the better understanding of Duchenne and Becker.

• Where can I learn more about this research?
  You can learn more about this research at www.nationwidechildrens.org/center-for-gene-therapy and www.clinicaltrials.gov (NCT02354781).

  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
FOR-DMD
Finding the optimal steroid treatment for Duchenne muscular dystrophy

• **What stage is this research?**
  This study is actively recruiting participants.

• **What is the goal or purpose of the FOR-DMD study?**
  This study will look at the benefits and side effects of the three most widely prescribed steroid treatments for Duchenne muscular dystrophy (DMD). The type of steroids commonly prescribed for DMD are called corticosteroids. Corticosteroids are a type of drug similar to natural hormones produced by the adrenal glands that reduce inflammation and suppress the immune response. They are often prescribed to boys with DMD. These steroids may have an effect on stabilizing or even improving muscle strength for a period of time but not all boys respond to treatment. The main steroid that is used is called prednisone. Deflazacort is also used in some countries. These are not “anabolic steroids” which is what athletes use illegally to build up muscle — these do not have an effect in DMD. Sometimes they are also referred to as ‘glucocorticoids’.

  - We will compare three different treatment groups:
    - Daily prednisone
    - Daily deflazacort
    - Intermittent prednisone (10 days on / 10 days off).

  The study is randomised (your child’s treatment group will be decided randomly, as in drawing names from a hat or tossing a coin) and double-blind which means that neither participants nor their doctors will know which group the boy is in (until the study is completed).

  All three steroid treatments are commonly used in boys with DMD and have been shown to be beneficial. Benefits include an increase in the length of time that the boys can continue to walk, reduction in the development of curvature of the spine, a longer time of adequate breathing, and possibly protection against the development of heart problems.

  However, we do not yet know which steroid treatment has the most benefit and most tolerable side effects. Therefore, this is a trial of present day steroid use which is needed because the practice of prescribing steroids varies a lot across doctors. This means that patients may not be getting the best possible treatment and management of side effects. All boys in this study will be receiving treatment with steroids and will be managed as per the recognized standards of care.

• **Who is eligible to be in this study?**
  To be in this study your child must have a confirmed diagnosis of DMD by genetic test. He must be between 4 and 7 years old and NOT previously treated with steroids except by inhaler or as an ointment. 300 participants are needed, and 145 have enrolled to date.

• **Where will this study take place?**
  This study will take place in at least 43 muscle clinics in the US, Canada, UK, Germany, and Italy and other countries may also be included later. Please visit www.clinicaltrials.gov for a complete list of all study sites. The principal investigators are Dr. R Griggs at the University of Rochester, NY and Prof. K Bushby at Newcastle University, UK.

• **What will happen during the study?**
  If you are interested in the study and your child appears to be eligible, he will be invited to visit the study site for a screening visit. At this appointment the study will be explained to you and your child in detail and some tests will be performed to allow the study doctors to ensure your child meets all the necessary requirements to participate in this study. After the screening period, you and your child will visit the study site at 3 months and then every 6 months after that. There will be a total of around 8–13 visits depending on when your child is enrolled. At each visit your child will be assessed to monitor benefits and side effects of corticosteroids. We hope that many children will be able to find a muscle clinic that is participating in the study reasonably close to where they live and that the study visits will take the place of their routine follow up. We expect your child will be in the study for 3–5 years.

• **Is there any funding to help pay for travel?**
  For participants in the United States and Canada, we are able to reimburse reasonable travel expenses (airfare, mileage, hotel, meals, etc.) incurred to reach the study center closest to the participant’s home. We are able to offer this assistance through a grant provided by the Muscular Dystrophy Association (MDA). After the screening visits, the frequency of clinic visits should not be any more than is usual for follow up in DMD.

• **Will I or my child get paid for participating in this study?**
  No.

• **Will I have access to the drug once the study has ended?**
  Yes, your doctor will discuss treatment options at the end of the study to decide the best steroid treatment plan for your child. The only issue is that deflazacort is not currently available in the US and we don’t know if it would be available in the US if the study showed it to be better than the other treatments. We are discussing this with the FDA. Deflazacort is already available in many countries and, in the US, it can be ordered from other countries by physician prescription.
**Will participating in this study prevent my child from taking part in other clinical trials?**

We are aware that trials of other potential new therapies may start during the course of the study. As steroid treatment is part of the normal standard of care in DMD we do not believe that being in this study would prevent your boy from being in another study later if there were one that he were eligible for. For example, if your child is/will be participating in Sarepta Therapeutics’ exon-skipping trials (i.e., PROMOVI), he may also participate in FOR-DMD at the same time. Moreover, although we hope that the majority of boys will finish the whole trial, as with any clinical study, you are entitled to withdraw at any time if you no longer wish to participate.

**Why should I consider participating in this study?**

While no personal benefit can ever be guaranteed from being in a clinical trial, there are other benefits, including:

- Allowing you to play an active role in DMD research
- Access to medical specialists that might not normally be available to your child
- Access to high standard medical care and management in DMD
- Contributing to the better understanding of DMD and what the best steroid treatment is

All participants will be getting active drug (there is no placebo group) and will be followed up and managed during the trial according to current standards of care.

**What should I do if I decide I want to take part in this study?**

Please visit www.clinicaltrials.gov for a complete list of study sites and the study coordinator at each site. Please call or email the study coordinator at the site nearest your home if you would like to participate. You can also contact the FOR-DMD US Project Manager, Kimberly Hart, at telephone 585-275-3767 or email Kim_Hart@urmc.rochester.edu.

**Who is funding this study?**

This study is funded by the US National Institutes of Health (NINDS). Parent Project Muscular Dystrophy and the MDA have also provided funding for this study.

**Where can I learn more about this study?**

You can learn more about this study at www.for-dmd.org and www.clinicaltrials.gov (NCT01603407). You can view the recent PPMD webinar on the FOR-DMD study: https://www.youtube.com/watch?v=W7B24zpBrDw

Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Gene Transfer of Micro-Dystrophin
Clinical Intramuscular Gene Transfer Trial of rAAVrh74.MCK.Micro-Dystrophin to Patients with Duchenne Muscular Dystrophy

• What stage is this research?
  This Phase 1 clinical trial is enrolling participants by invitation only.

• What is the goal or purpose of this study?
  The goal of this study is to evaluate safety and biological activity of the micro-Dystrophin vector as part of the process in determining if it can act as a potential dystrophin replacement for Duchenne muscular dystrophy. There will be two groups: one high dose and one low dose group. Both groups receive escalating doses injected into the Extensor Digitorum Brevis (EDB) muscle to determine the maximum tolerated dose (MTD). The EDB muscle is a muscle on the upper part of the foot. Each group will have three participants.

• Who is funding this study?
  This study is funded by the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD).

• Who is eligible to participate in this study?
  To participate in this study you must be a boy at least 7 years old or older, wheelchair-dependent, confirmed dystrophin mutations based on mutation compatibility with microdys cDNA, able to cooperate with muscle testing, and if sexually active, must be willing to practice reliable method of contraception. For a complete list of all the inclusion and exclusion criteria, please see the listing on clinicaltrials.gov.

• What do I have to do if I decide to participate in this study?
  This study involves injection of the micro-dystrophin therapy into the EDB muscle (on the upper part of the foot). It will also involve some lab studies (blood and urine) and return visits over the course of 2 years. There will also be a muscle biopsy on the EDB muscle on day 180 in one foot compared to the placebo treated muscle biopsy in the opposite foot.

• How long will this study last, and will I have access to the drug/treatment once the study has ended?
  The anticipated length of the study is 2 years.

• Where does this study take place?
  This study takes place at Nationwide Children’s Hospital, Columbus, Ohio.

• How many visits to the study site are necessary?
  >> At least 13 visits over the course of 2 years (baseline, injection visit (days 0-2), follow up visits on days 7, 14, 30, 60, 90, and 180 day, and at the end of the 1st and 2nd year).

• Can any visits be done locally?
  >> No, all visits must be done at Nationwide Children’s Hospital.

• Is there any funding to help pay for travel?
  >> Please check with the study coordinator at the trial site.

• Why should I consider participating in this study?
  This study will help determine if this particular micro-dystrophin is safe for people with Duchenne which will help determine if further larger studies should be done.

While no personal benefit can ever be guaranteed by participation in a clinical trial, there are other benefits, including allowing you to play an active role in your own health care (or that of your child), gaining access to new research treatments before they are widely available and having access to medical specialists that are normally not available to you or your child, and helping others by contributing to the better understanding of Duchenne and Becker.

• Where can I learn more about this study?
  You can learn more about this study at www.DuchenneConnect.org and www.ClinicalTrials.org (NCT02376816) as well as http://www.nationwidechildrens.org/gene-therapy-clinical-studies--1.
HT-100
Akashi’s Phase 1/2 Clinical Program in DMD

- **What stage is this research?**
  This clinical trial program is actively recruiting participants.

- **What is the goal or purpose of this trial?**
  The main purpose of this study is to test the safety and tolerability of different, increasing doses of an experimental medication called HT-100 in boys and young men with Duchenne muscular dystrophy. The study medication, HT-100, is a medicine that may help promote healthy muscle regeneration, diminish inflammation and the resulting damage to muscle, and decrease the scar tissue that forms in the muscles of children with Duchenne. HT-100 does not appear to be mutation specific, meaning it is potentially applicable to all boys and young men with Duchenne.

  The clinical trial is also evaluating exploratory measures of efficacy of HT-100.

  Akashi Therapeutics (formerly Halo Therapeutics) has an approved IND from the FDA allowing the company to conduct this research and has received Orphan Drug status for HT-100 for Duchenne.

- **Who is funding this research?**
  The drug is being developed for Duchenne by Akashi Therapeutics, Inc. (formerly Halo Therapeutics). This research is funded by the Nash Avery Foundation, Charley’s Fund, Parent Project Muscular Dystrophy, and more than 20 other Duchenne patient foundations.

- **Who is eligible to participate in this trial?**
  This trial is open to males with Duchenne, ages 6–20 years old. Ambulatory or nonambulatory boys can enroll, and participants may be either corticosteroidnaive or on corticosteroid therapy for at least 12 months (stable dose and regimen).

  Recent, substantial change in use of cardiac medications or medications affecting muscle function and/or significantly compromised cardiorespiratory function would exclude you from this trial.

- **What do I have to do if I decide to participate in this trial?**
  Single and multiple ascending doses of HT-100 will be given to participants. Safety and tolerability will be assessed. Pharmacokinetic sampling, or measurements of the amount of HT-100 in the bloodstream, will also be taken.

  This initial study of safety and tolerability will be followed immediately by a 12 month, open label extension study. All boys and young men who complete the initial study will be eligible to participate in the extension study.

  All participants who complete the open label extension study will be eligible to participate in an open label study that will provide ongoing access to medication until HT-100 is available as a marketed product.

- **Where does this clinical trial take place?**
  There are 5 sites in the US: UC Davis in Sacramento, CA; Kennedy Krieger Institute in Baltimore, MD; Washington University School of Medicine in St. Louis, MO; Nationwide Children’s Hospital in Columbus, OH; and Cincinnati Children’s Hospital in Cincinnati, OH.

- **How many visits to the study site are necessary?**
  There are 3 HT-100 clinical protocols that allow for continuous dosing over an extended period of time. For the initial study, there are 9 separate visits to the study site, some of which require an overnight stay and some of which occur over multiple days. For the extension study, the participant has the option to remain in the study for up to 12 months with 7 separate single day visits to the study site, 1 every other month, with phone conversations planned between the onsite visits. For the longterm extension study, the participant has the possibility of continuing for a period of years and there are 2-single-day visits to the study site each year, 1 every 6 months, for as long as the participant remains in the study. At the time of the participant’s last dose in either the extension or longterm extension study, the visit to the study site can occur over multiple days if it is more convenient for the participant and family.

- **Is there any funding to help pay for travel?**
  Yes, there is some funding available to help pay for travel. In addition, Akashi Therapeutics (the sponsor) is making available a travel coordinator to assist families with travel planning.

- **Will I get paid for participating in this study?**
  No, there is no stipend for participating in this study.

- **Why should I consider participating in this study?**
  While no personal benefit can ever be guaranteed by participation in a clinical trial, there are other benefits, including allowing you to play an active role in the health care of your child, gaining access to medical specialists that are normally not available to your child, and helping others by contributing to the better understanding of Duchenne.

- **Where can I learn more about this research?**
  You can learn more at www.akashirx.com (http://akashirx.com/) and www.ClinicalTrials.gov (ID NCT01847573 and NCT01978366). Patients interested in participating in this trial can email Akashi using this address: trialinfo@akashirx.com (mailto:trialinfo@akashirx.com), or email the study contact at the site nearest your home:
  » California: University of California, Davis Medical Center, Sacramento; Erica Goude (email Erica.goude@ucdmc.ucdavis.edu)
  » Maryland: Kennedy Krieger Institute, Baltimore; Dr. Genila Bibat (email: bibat@kennedykrieger.org))
  » Missouri: Washington University School of Medicine, St. Louis; Linda Schimmoeller (email: schimmoellerl@neuro.wustl.edu)
  » Ohio: Cincinnati Children’s Hospital Medical Center; Ma Leilani (Lani) Relucio (email: lani.relucio@cchmc.org) AND Nationwide Children’s Hospital, Columbus; Mallory Rowell (email: mallory.rowell@nationwidechildrens.org)

  Please check www.DuchenneConnect.org (http://www.duchenneconnect.org/) for updates to this FAQ sheet.
Microsoft Band
Microsoft Band as an Outcome Measure for Boys with Duchenne Muscular Dystrophy

• **What stage is this research?**
  This study is actively recruiting boys with Duchenne muscular dystrophy.

• **What is the goal or purpose of this study?**
  Researchers want to learn what functional activities can be measured while boys with Duchenne wear the Microsoft (MS) Band.

• **Who is sponsoring this study?**
  This study is sponsored by the Cooperative International Neuromuscular Research Group (CINRG).

• **Who is eligible to participate in this study?**
  Researchers are recruiting boys with Duchenne, ages 4 years – 9 years old. Participants must have reliable access to wireless internet.

• **What do I have to do if I decide to participate in this study?**
  If you and your child decide to take part in this study, your child would be asked to wear the MS Band for seven straight days (10 hours per day and while sleeping). You would then take a break for a week, and following this break, your child would wear the band for another seven straight days. During this time, you would be asked to complete daily logs about your child’s activity and ensure the MS Band is plugged in and the battery fully charged each night. You will be asked questions about your child’s daily routines and family experiences with the MS Band. Six months after your initial wearing period, you will be asked to complete these activities for a third period of seven straight days.

• **Where does this study take place?**
  This study will be run at two CINRG network sites: the University of Pittsburgh in Pittsburgh, PA, and the University of California, Davis, in Sacramento, CA.

• **Will I get paid for participating in this study?**
  Yes, families who take part will be compensated to thank them for their time.

• **Why should I consider participating in this study?**
  While no personal benefit can ever be guaranteed by participation in a study, there are other benefits, including allowing you to play an active role in your own health care (or that of your child), gaining access to medical specialists that are normally not available to you or your child, and helping others by contributing to the better understanding of Duchenne.

• **Where can I learn more about this study?**
  For more information, please contact Gabriela Niizawa at 412-383-9775.
  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
MRI and Cardiopulmonary Function
Assessment of Cardiopulmonary Function in Duchenne Muscular Dystrophy

• What stage is this research?
  This is an actively recruiting clinical trial.

• What is the goal or purpose of this study?
  The goals of this study are to monitor long term changes in heart and breathing muscles in children with Duchenne and to develop better imaging approaches to evaluate these muscles.

• Who is the sponsor of this study?
  This study is sponsored by CureDuchenne and the University of Florida.

• What are the inclusion (enrollment) criteria for this trial?
  Participants need to be boys with Duchenne who are between the ages of 5–13 years old.

• What do I have to do if I decide to participate in this study?
  Participation includes Magnetic Resonance Imaging (MRI) of the heart and breathing muscles, breathing tests (called pulmonary function tests), exercise tests (using a stationary bike) and possibly an echocardiogram (ultrasound of the heart).
  These tests will be performed from one to four times per year for up to four years.

• Where does the trial take place?
  This study takes place at the University of Florida in Gainesville, FL.

• Are travel expenses to the study site reimbursed?
  Travel reimbursement may be available based on the amount of charitable funding and the needs of the participant.

• Why should I consider participating in this study?
  Although the results from this study will not directly benefit you, the goal of this study is to learn more about the heart and breathing muscles. This information may help to develop better ways to evaluate heart and lung function.

• Where can I learn more about this study?
  You can learn about this study at www.clinicaltrials.gov (code NCT02195999)
  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Myoblast Transplantation
Myoblasts Transplantation in Duchenne Muscular Dystrophy Patients

• What stage is this research?
  This Phase 1/2 clinical trial is actively enrolling participants.

• What is the goal or purpose of this study?
  The purpose of this study is to investigate whether the transplantation of normal donor myoblasts throughout one muscle, the extensor carpi radialis (forearm), of the patients will maintain or improve the strength of that muscle, slowing disease progression.

• Who is the sponsor of this study?
  Dr. Jacques Trembaly at the Centre de recherche du CHU de Québec is the sponsor of this study.

• What are the inclusion (enrollment) criteria for this trial?
  Ten participants will be recruited for this trial. Participants must be males with Duchenne muscular dystrophy who are at least 16 years old. Additional details regarding the inclusion and exclusion criteria are posted on www.clinicaltrials.gov.

• What do I have to do if I decide to participate in this study?
  Participants will be transplanted with myoblasts grown from the muscle biopsy of a healthy donor. Thirty million myoblasts will be injected per cm cube. The contralateral muscle will be injected with saline to serve as a control. The strength of both muscles will be measured at three months post transplantation to verify whether the myoblast transplantation showed a significant improvement of the injected muscle. If there is no significant improvement the protocol will be terminated immediately for that patient. If there is a significant improvement, the patient will be maintained under immunosuppression until six months post transplant and his strength will be re-evaluated.

• Where does the trial take place?
  This trial takes place at two Canadian sites — the Children’s Hospital — London Health Sciences Centre in London, Ontario, and Centre Hospitalier Universitaire de Québec in Québec, Québec.

• Will I be reimbursed for travel expenses?
  Yes, both the participant as well as the donor participant will be reimbursed for study related travel expenses.

• Will I get paid for participating in this trial?
  No, you will not be paid for your participation in this trial.

• Why should I consider participating in this study?
  While no personal benefit can ever be guaranteed by participation in a clinical trial, there are other benefits, including allowing you to play an active role in your health care, gaining access to medical specialists that may not normally be available to you, and helping others by contributing to the better understanding of Duchenne.

• Where can I learn more about this study?
  Please visit www.clinicaltrials.gov (code NCT02196467) for further details including contact information. Please check www.DuchenneConnect.org for updates to this FAQ sheet.
PF-06252616
Development of a myostatin inhibitor as a potential anabolic therapy for the muscular dystrophies.

• What stage is this research?
This Phase 2 study is actively enrolling participants — please see www.ClinicalTrials.gov for details (NCT#02310763). The study is designed to evaluate the safety, tolerability, efficacy, pharmacokinetics and pharmacodynamics of PF-06252616.

• What is PF-06252616?
PF-06252616 is an experimental anti-myostatin monoclonal antibody. Myostatin acts in the body to help regulate muscle growth by inhibiting (blocking) muscle differentiation and growth. Blocking the activity of myostatin may have therapeutic application in treating muscle wasting diseases, such as Duchenne muscular dystrophy. Based on the proposed mechanism of action of PF-06252616, there is the potential to increase muscle mass and function in boys with Duchenne who have evidence of reduced muscle mass.
Preclinical evidence of increased muscle size and function has been demonstrated in mice and nonhuman primates.
The Phase 1 study of PF-06252616 in healthy adults provides evidence of increased muscle measured by MRI scanning after 1 month of dosing.

• Who is funding this research?
At this time, Pfizer is fully supporting the clinical development of PF-06252616.

• What are the inclusion (enrollment) criteria for this trial?
Participants must be ambulatory boys with Duchenne ages 6 to 9 years old. Eligible participants must also able to perform the 4 stair climb in > 0.33 stairs/second ≤1.6 stairs/second (with or without the use of handrails).
The diagnosis of Duchenne must be confirmed in the subject’s medical history and by genetic testing obtained during routine medical care. Please note: The investigational therapy is not mutation-specific, so boys with any DMD gene mutation may participate.
Eligible participants must be on glucocorticosteroids for a minimum of 6 months prior to signing informed consent to join the study. Additional screening evaluations will be conducted at the study site to confirm eligibility. Full details are available on ClinicalTrials.gov (NCT#02310763).

• What do I have to do if my child meets the inclusion criteria and I decide that I may want my child to participate in this study?
Participants will receive monthly IV infused doses of either PF-06252616 or placebo for up to 2 years at their study site. Participants will undergo safety evaluations at regular intervals (including monthly clinical exams and blood tests; scans at 1–3 times per year). Participants will undergo functional evaluations (including the 4 stair climb and 6-minute-walk test every 2 months and scans to measure muscle volume and quality 3 times per year). Based on the design of the study, all participants will receive the investigational drug during all or some of the study.

• Where does the trial take place?
There are currently sites open for enrollment in the United States (Los Angeles, CA; Cincinnati, OH; Baltimore, MD; and Iowa City, IA); Canada (London, Ontario); Japan (Tokyo); and the United Kingdom (Newcastle upon Tyne). Please check www.clinicaltrials.gov for details regarding the trial sites. Additional sites are under evaluation and will be posted on clinicaltrials.gov if and when active.

• What, among other things, should I consider when deciding whether my child should participate in this study?
While no personal benefit can ever be guaranteed by participation in any clinical trial, participation in such a clinical study may:
» allow you to play an active role in the health care of your child;
» permit you to gain access to medical specialists that may not normally be available to your child;
» enable you to potentially help others by contributing to a better understanding of Duchenne.
A discussion with the study investigator will also help identify any potential risks to participation.

• Is Pfizer committed to pursuing possible treatments for Duchenne?
Our investment in Duchenne currently extends beyond PF-06252616. Although this is our most advanced compound we are actively studying other compounds with diverse mechanisms of action and modalities to potentially help Duchenne patients and their families.
We have been active in building our internal Duchenne efforts including: recruitment of additional scientists and clinicians with experience in Duchenne, evaluating additional programs for Duchenne, designing clinical trials, and exploring research collaborations.

• Where can I learn more about PF-06252616?
The current study is posted on www.ClinicalTrials.gov (NCT#02310763). Visit Pfizer.com/pipeline, Pfizer’s online database where you learn more about our portfolio of new medicines and find more about our Research and Development efforts around the world.
Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Spironolactone and Eplerenone
Therapeutic Potential for Aldosterone Inhibition in Duchenne Muscular Dystrophy

• What stage is this research?
This Phase 3 trial is actively recruiting participants.

• What is the goal or purpose of this study?
The purpose of the study is to see if both spironolactone and eplerenone (Inspra), generic drugs used in other types of heart conditions, help maintain heart and lung function in boys with Duchenne.

• Who is sponsoring this study?
This study is sponsored by The Ohio State University.

• Who is eligible to participate in this study?
Participants must be males with Duchenne, at least 10 years old, non-ambulatory, not taking a corticosteroid (prednisone or deflazacort), and not taking eplerenone or spironolactone. Please see www.ClinicalTrials.gov for additional inclusion and exclusion criteria.

• What will participants do in this study?
Participants will be randomly assigned to receive either eplerenone or spironolactone, and they will take one tablet of the assigned drug once daily for 12 months.

All participants will have blood drawn at the baseline visit as well as months 1, 2, 3, 6, 9 and 12.

All participants will have a cardiac MRI scan and pulmonary function tests at baseline and 12 months.

• Where does this study take place?
The baseline visit will take place at one of the following:
  » The Ohio State University (OSU) in Columbus, OH
  » Mattel Children’s Hospital in Los Angeles, CA
  » University of Colorado Hospital in Aurora, CO
  » University of Utah Hospital in Salt Lake City, UT

Blood draw at 1, 2, 3, 6, and 9 months can be done at a local laboratory where you reside.

The 12 month visit will be done at the same site where you had your baseline visit.

• Will I be paid for participating in this study?
There is no payment for participating in the study but you will be reimbursed for travel, food and lodging expenses for the baseline and 12-month visit.

The blood draws, cardiac MRI scans, and pulmonary function tests will be paid for by the study if it is not part of your regular clinical visit. The study drug will also be provided free of charge.

• Why should I consider participating in this study?
While no personal benefit can ever be guaranteed by participation in a clinical trial, there are other benefits, including allowing you to play an active role in your or your child’s healthcare, gaining access to medical specialists that are normally not available to you or your child, and helping others by contributing to the better understanding of Duchenne.

• Where can I learn more about this study?
You can learn more about this study at www.ClinicalTrials.gov (NCT02354352)
You may also contact:
  » Dr. Subha Raman at The Ohio State University, (614) 688-8020 or raman.1@osu.edu
  » Dr. Nancy Halnon at Mattel Children’s Hospital, (310) 267-7618 or nhalnon@mednet.ucla.edu
  » Dr. Scott Auerbach at University of Colorado, (720) 777-8723 or scott.auerbach@childrenscolorado.org
  » Dr. Michael Puchalski at University of Utah, (801)587-9104 or Michael.puchalski@hsc.utah.edu

Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Steroids in Young Boys
Historically Controlled Trial of Corticosteroids in Young Boys with Duchenne Muscular Dystrophy

• What stage is this research?
This Phase 2 clinical trial is actively enrolling participants.

• What is the goal or purpose of this study?
The purpose of this study is to investigate the efficacy (effectiveness) of oral weekend corticosteroid use in infants and young boys with Duchenne who are under age 30 months. While it has been known for many years that corticosteroid use benefits boys with Duchenne, most clinicians do not consider treating until after age 3 or 4 years of age. The primary reason for the delay is that daily corticosteroid use has many side effects including short stature, obesity, and osteoporosis. A recent randomized blinded study of weekend oral corticosteroid use over one year showed equal improvement in strength with fewer side effects, particularly as related to growth and cushingoid changes.

• Who is the sponsor of this study?
Washington University School of Medicine is the sponsor of this study.

• What are the inclusion (enrollment) criteria for this trial?
24 participants will be recruited for this trial. Participants must be boys with Duchenne who are between 1 month and 30 months of age, with no prior use of corticosteroids. Additional details regarding the inclusion and exclusion criteria are posted on www.clinicaltrials.gov.

• What do I have to do if I decide to participate in this study?
Participants will take oral prednisone (5mg/kg/day) on Friday and Saturday mornings with breakfast. Assessments will be performed periodically to determine if oral weekend prednisone use improves gross motor development in infants and young boys with Duchenne. Assessments will include the Bayley-III Scales of Infant Development and the North Star Ambulatory Assessment (NSAA). Ultrasound of muscles will be performed at the primary site (Washington University). Each boy will be followed for one year.

• Where does the trial take place?
This trial takes place at 6 sites in the US: University of California, Davis in Sacramento, CA; Nemours Hospital in Orlando, FL; Laurie Children’s Hospital of Chicago, IL; Washington University in St Louis, MO; Nationwide Children’s Hospital in Columbus, OH; and University of Texas South Western Medical Center of Dallas, TX.

• Why should I consider participating in this study?
While no personal benefit can ever be guaranteed by participation in a clinical trial, there are other benefits, including allowing you to play an active role in the health care of your child, gaining access to medical specialists that are normally not available to your child, and helping others by contributing to the better understanding of Duchenne.

• Where can I learn more about this study?
Please visit www.clinicaltrials.gov (code NCT02167217) for further details including contact information for each trial site.
Please check www.DuchenneConnect.org for updates to this FAQ sheet.
BMS-986089
Bristol-Myers Squibb’s Candidate for Myostatin Inhibition

• **What stage is this research?**
  BMS-986089 is in a Phase 1 clinical trial in healthy volunteers that began in 2014.

• **What is BMS-986089?**
  BMS-986089 is an investigational protein that binds to myostatin. Myostatin is a protein produced primarily in skeletal muscle cells that prevents muscle cell growth and differentiation. Animals lacking myostatin or animals treated with substances that block the activity of myostatin have significantly larger muscles.

• **Where is this research being done?**
  This research is being done by Bristol-Myers Squibb (BMS). Information regarding ongoing clinical trials can be found on www.ClinicalTrials.gov and on the BMS clinical trial patient and caregiver website, www.bmsstudyconnect.com.

• **What is the goal or purpose of this research?**
  The primary goal of the Phase 1 study is to assess the safety, tolerability, immunogenicity, drug levels, and drug effects of single and multiple doses BMS-986089 in healthy adult subjects.

• **When can we expect to see clinical trials for patients with muscular dystrophy?**
  A Phase 1b/2 study in Duchenne patients is expected to start in the fall 2015. Up-to-date information regarding ongoing clinical trials can be found at www.ClinicalTrials.gov and at the BMS clinical trial patient and caregiver website, www.bmsstudyconnect.com.

• **Where would a clinical trial take place?**

• **Who would be eligible to participate in a clinical trial?**

• **Where can I learn more about this research?**
  http://www.bmsstudyconnect.com will post the clinical trial once it is actively recruiting patients. www.ClinicalTrials.gov will post the clinical trial once it is actively recruiting patients.

  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
**CAT-1004**
Catabasis’ Upcoming MoveDMD Trial

- **What stage is this research?**
  Phase 1 trials in adults with CAT-1004 have been completed. A Phase 1 / 2 clinical trial in boys with Duchenne, called MoveDMD, is expected to start in the second quarter of 2015.

- **Where is this research being done and who is funding this research?**
  This research is being done by Catabasis, a clinical-stage biopharmaceutical company focused on the discovery, development and commercialization of novel therapeutics based on its SMART linker technology platform. Catabasis is initially focused on the treatment of rare diseases, including Duchenne muscular dystrophy (DMD). Parent Project Muscular Dystrophy is supporting patient travel for MoveDMD.

- **What is the goal or purpose of this research?**
  CAT-1004 is an oral small molecule investigational drug candidate that has been observed to inhibit activated NF-kB. Dystrophin is a protein that keeps muscles healthy by maintaining the structure of muscle cells. In boys with DMD, the absence of dystrophin combined with mechanical stress in muscle leads to activation of NF-kB. Activated NF-kB drives muscle damage and prevents muscle regeneration.

- **What is the current state of this research and what steps need to be completed before moving into a clinical trial?**
  Catabasis has completed a series of studies in animal models of Duchenne in which CAT-1004 and similar compounds reduced muscle inflammation and degeneration, reduced fibrosis and improved muscle function.
  The Phase 1 / 2 clinical trial in boys with Duchenne is expected to start in the second quarter of 2015.

- **What is your best estimate for the length of time it will take to move this research into clinical trials?**
  We expect for MoveDMD to start enrolling patients in the second quarter of 2015.

- **Where would a clinical trial take place?**
  We plan to have three centers participate in the MoveDMD trial: University of Florida, Gainesville, Florida; Shriners Hospitals for Children, Portland, Oregon; and The Children’s Hospital of Philadelphia, Philadelphia, Pennsylvania.

- **Who would be eligible to participate in a clinical trial?**
  Requirements for participating in this clinical trial include:
  - Boys with a diagnosis of DMD (any confirmed mutation) between the ages 4 and 7 years
  - Ability to walk independently
  - No corticosteroid use within the past 6 months and no plans to start corticosteroids in the next 6 months
  The key inclusion and exclusion criteria and additional details about this clinical trial are available at www.clinicaltrials.gov.

- **Where can I learn more about this research?**
  You can contact Catabasis directly with any questions at Joanne.Donovan@catabasis.com.
  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
**GALGT2 Gene Therapy for DMD**

Viral gene transfer for GALGT2 (rAAVrh74.MCK.GALGT2) as a surrogate gene therapy for DMD

- **What stage is this research?**
  This research is clinical, meaning it has advanced to clinical trials involving people. An IND application has been approved by the FDA. Funding for an initial trial of intramuscular injection of the virus (rAAVrh74.MCK.GALGT2) has been obtained. IRB approval is pending. We anticipate enrolling the first patients in the intramuscular injection study in August, 2015.

- **What is the goal or purpose of this study?**
  The goal of this study is to introduce the GALGT2 gene into the body by using a viral vector (an adeno-associated virus, or AAV). Because the virus carries GALGT2 rather than a version of the dystrophin gene, it is a “surrogate” gene therapy.

GALGT2 encodes the protein GaINAc transferase (beta 1,4-N-acetylgalactosamine galactosyltransferase). This is an enzyme that transfers a complex sugar molecule onto a few specific proteins, including dystroglycan.

Usually, GaINAc transferase is found only at the neuromuscular junction (NMJ), where some components of the dystroglycan-associated protein complex are different than elsewhere in muscle. Importantly, at the NMJ, utrophin is present instead of dystrophin.

In the mdx mouse, viral gene transfer of GALGT2 results in expression of GaINAc transferase across the entire muscle membrane (instead of just at the NMJ), as well as upregulation of utrophin across the entire muscle fiber.

In the mdx mouse, this expression can correct muscle functional deficits to the same degree as does microdystrophin gene expression. Furthermore, overexpression of GALGT2 corrects muscle pathology in mouse models of other muscular dystrophies, including LGMD2A and congenital muscular dystrophy (MDC1A).

This AAV viral vector is known not to cause disease. The vector includes a gene promoter that is specifically activated in muscle tissue, so the gene should not be significantly activated in other tissues. The AAV-delivered gene is not integrated into chromosomal DNA.

Because the GaINAc transferase is already expressed in patients, there should not be any immune response generated to the transferred gene’s protein product.

- **Who is funding this study?**
  Development, preclinical testing, and the clinical trial have been funded by the NIH.

- **Who is eligible to participate in this study?**
  The first-in-human gene transfer trial will consist of intramuscular injections into the extensor digitorum brevis (EDB) muscle on the side of the foot in recently non-ambulant patients over the age of 9 years. A subsequent phase of the trial will consist of intravascular delivery in ambulant boys.

- **What do I have to do if I decide to participate in this study?**
  Each EDB muscle (foot) will receive an injection; one will be saline, and the other will contain the viral vector. Muscle biopsy will be performed on each EDB muscle at either 6 or 12 weeks after injection.

- **Where does this study take place?**
  The clinical trial will take place at Nationwide Children’s Hospital (NCH) in Columbus, Ohio.

- **How many visits to the study site are necessary?**
  We anticipate that the study will require 13 visits to NCH over two years.

- **Can any visits be done locally?**
  No

- **Is there any funding to help pay for travel?**
  We will be seeking funding to help pay for travel.

- **Will I get paid for participating in this study?**
  No

- **How long will this study last, and will I have access to the drug/treatment once the study has ended?**
  The study will require follow up for two years after injection. Patients who participate in the initial intramuscular injection study are unlikely to be eligible for follow up vascular delivery studies, due to the expected development of antibodies to the viral capsid. If the treatment were to be approved by the FDA, plasmapheresis to clear anti-AAV antibodies may allow future treatment, although this cannot be guaranteed at present.

- **Why should I consider participating in this study?**
  The initial first-in-human intramuscular injection study is necessary to confirm the expression of the GALGT2 gene in humans, and to assess the safety of this expression. This information is needed to proceed to a future efficacy trial.

There is no likelihood of personal benefit by participation in this initial clinical trial, but there may be other benefits. These include allowing you or your child to participate in the advancement of a new and promising therapy, and helping others by contributing to the better understanding of Duchenne and Becker.

- **Where can I learn more about this study?**
  You will be able to learn more about this study at www.DuchenneConnect.org and www.ClinicalTrials.org, and www.nationwidechildrens.org/center-for-gene-therapy.
**Isofen3**
A combination drug for the treatment of Duchenne

- **What stage is this research?**
  This upcoming study will be a Phase 2 clinical trial. Isofen3 is a combination of Ibuprofen (a nonsteroidal anti-inflammatory agent-NSAID) and isosorbide dinitrate (a drug that releases nitric oxide). Each of these is licensed worldwide for individual use.

- **What is the goal or purpose of this study?**
The goal of this study is to determine if the combination of two drugs, Ibuprofen and isosorbide dinitrate, is capable of slowing the progression of Duchenne. In animal studies the drug combination has been shown to help with muscle repair and myogenesis, with reducing inflammation, and with enhancing muscle function. A pilot study in nonambulant patients with either Becker or Duchenne or Limb Girdle muscular dystrophy plus two Phase 1 studies in healthy volunteers showed the drug to be safe and tolerable with improvement in some of the measures studied.

- **Who is funding this study?**
The study is not funded and is on hold.

- **Who is eligible to participate in this study?**
  To participate in this study you must be above 6 years with a confirmed diagnosis of Duchenne, nonambulatory, and be able to meet certain heart and respiratory function criteria, with no recurrent headaches. You will need to pass an initial screening visit and meet all inclusion criteria and sign a consent form.

- **How long will this study last, and will I have access to the drug/treatment once the study has ended?**
The anticipated length of the study is a total of 42 months with enrollment being 6 months and the study lasting for 36 months. Depending on the results, you may have access to the drug while data is being analyzed and before FDA approval.

- **Where does this study take place?**
The study site locations are still being determined.

- **How many visits to the study site are necessary?**
  There are 14 visits in which you will receive non invasive assessments (except blood draw) of your health status. All visits must be done at a study site, as specific assessment of your muscle function has to be completed by a physiatrist trained in the study procedure.

- **Will I get paid for participating in this study? Is there any funding to help with travel?**
  No, this study is on a voluntary basis. Presently there is no funding for travel.

- **Why should I consider participating in this study?**
  While no personal benefit can ever be guaranteed by participation in a clinical trial, there are other benefits, including allowing you to play an active role in your own health care (or that of your child), gaining access to new research treatments before they are widely available and having access to medical specialists that are normally not available to you or your child, and helping others by contributing to the better understanding of Duchenne and Becker.

- **Where can I learn more about this study?**
  This trial will be posted on www.ClinicalTrials.gov once it is actively recruiting.

  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Strength Training
Development of a Strength Training Protocol in Duchenne Muscular Dystrophy

- What stage is this research?
This study is not yet recruiting participants. Recruiting is expected to begin by July 2015.

- What is the goal or purpose of this study?
Parents of boys with Duchenne have received little guidance on the potential of exercise to maintain muscle function. This pilot study focuses on assessing whether a mild to moderate strengthening exercise program can be safely implemented in boys with Duchenne.

- Who is funding this study?
This study is funded by the NIH – NIAMS (National Institute of Arthritis and Musculoskeletal and Skin Diseases).

- Who is eligible to participate in this study?
To participate in this study you must be between the ages of 7–9 years with a confirmed diagnosis of Duchenne, able to walk independently for at least 100 meters (~length of a football field) without an assistive device, able to climb 4 stairs, and currently using corticosteroids (prednisone or deflazacort).

- What are participants doing in this study?
Muscle strength tests and exercises will be performed for the thigh muscles. Safety is a focus for this study, and safety measures include MRI and Magnetic Resonance Spectroscopy (MRS) measurements and a small blood draw to determine creatine kinase levels. Some participants will perform an in-home exercise program for 12 weeks.

- Where does this study take place?
The first part of this study (Aim 1) will take place at the University of Florida (UF) in Gainesville, Florida. The second part (Aim 2) will take place at UF and will also take place at the location of the participant’s residence.

- How many visits to the study site are necessary?
For Aim 1, participants will spend 1–2 weeks at UF.
For Aim 2, participants spend 1 week at UF and then return to UF for single visits after 6 weeks and 12 weeks. During weeks 2–12, participants perform an exercise program three days/week at their home.

- Will I get paid for participating in this study? Is there any funding to help with travel?
Yes, gift cards are provided after completion of visits at UF. The cost of airfare and hotel for the subject and one parent/legal guardian are provided as part of this study.

- Where can I learn more about this study?
You can learn more about this study at www.ClinicalTrials.gov (NCT02421523).
Please check www.DuchenneConnect.org for updates to this FAQ sheet.

(continued on p. 22)
### VBP15
A Novel Steroid Customized for Duchenne

- **What stage is this research?**
  This program is clinical, meaning it has advanced to clinical trials involving people. Phase 1 clinical trials in healthy adult volunteers are underway, and Phase 2a Duchenne studies are expected to begin later in 2015.

- **What is the goal or purpose of this study?**
  The goal of this research is to develop a drug (VBP15) that has optimized four subactivities of traditional steroids for Duchenne: increase the anti-inflammatory effect, decrease side effects, increase mineralocorticoid antagonism, and stabilize myofiber membrane walls against dystrophin deficiency.

- **Who is funding this study?**
  To date, ReveraGen has worked through public-private partnerships to develop VBP15:
  - PPMD ($750,000) and Foundation to Eradicate Duchenne ($250,000) are co-funding the chronic toxicology studies required by regulatory agencies
  - The $2.1M Phase 1 trial is funded by MDA (50%), and three UK foundations (Joining Jack, Duchenne Research Fund, Duchenne Children’s Trust)
  - Additional funding has provided by the Save Our Sons, NIH TRND, CDMRP Department of Defense, CureDuchenne, and the Duchenne Alliance Research Foundation (Save Our Sons, Michael’s Cause, Pietro’s Fight, Alex’s Wish, and Ryan’s Quest).

- **Who is eligible to participate in this study?**
  The Phase 1 clinical trials are enrolling healthy adult volunteers between the ages of 18 Years to 65 Years.
  The Phase 2a clinical trial (to begin in late 2015) will enroll ~25 steroid naïve Duchenne boys ages 4–7 yrs of age.
  The Phase 2b clinical trial (to begin in 2016) will enroll ~100 steroid naïve Duchenne boys ages 4–7 yrs of age.

- **What do I have to do if I decide to participate in this study?**
  The Phase 2a clinical trial will be carried out by the Cooperative International Neuromuscular Research Group (www.cinrgresearch.org). Contact Lauren Morgenroth (lhache@childrensnational.org) or info@reveragen.com.

- **How long will this study last, and will I have access to the drug/treatment once the study has ended?**
  The Phase 2a study will include an extension study. Those who opt to enter the Phase 2a and Phase 2a extension study will have a 2 week drug treatment, 4 week drug holiday, then 6 month extension study on drug.

- **Where does this study take place?**
  The Phase 1 clinical trials in normal volunteers are taking place at PRA in Lexington, Kansas.
  The Phase 2a clinical trials in Duchenne boys will take place in CINRG sites in the USA.
  - How many visits to the study site are necessary?
    - Phase 2a = ~5 visits (once every two weeks);
    - Phase 2a extension = ~7 visits (once a month)
  - Can any visits be done locally?
    - There will be sites throughout USA, but visits must be done at one of the CINRG sites.
  The Phase 2b clinical trials will be throughout Europe.

- **Will I get paid for participating in this study?**
  All expenses are paid for by the study.

- **Why should I consider participating in this study?**
  Participation will help determine whether VBP15 is an effective therapy for Duchenne. Showing effectiveness for Duchenne could have implications for many other disorders where glucocorticoids are used. The VBP15 program includes many innovations in clinical trial design that, if successful, will speed other drug development programs, including blood biomarkers and mobile health outcomes (Microsoft Band).

- **Where can I learn more about this study?**
  Information will be posted on the CINRG and ReveraGen websites, as well as www.clinicaltrials.gov.
  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
ARM210
Using ARM210 to Improve Muscle Strength and Function in Duchenne Muscular Dystrophy

• **What stage is this research?**
  This research in Duchenne muscular dystrophy is late-stage pre-clinical, meaning it has not advanced to clinical trials involving people with Duchenne yet. A first time in human Phase 1 clinical safety study in adult healthy volunteers is planned to start in 3Q 2015. In addition, a related compound in development by ARMGO has completed several clinical trials for heart failure and cardiac arrhythmias. Positive activity in patients was demonstrated, validating ARMGO’s novel therapeutic approach.

• **Where is this research being done and who is funding this research?**
  This research is being done by ARMGO Pharma, Inc. The Muscular Dystrophy Association (MDA) awarded the company $1 million to develop the compound, ARM210, for treatment of Duchenne.

• **What is the goal or purpose of this research?**
  ARMGO has identified a new class of small molecule therapeutics (called Rycals®) that restore normal balance of calcium within muscle cells by correcting the activity of a type of channel called the “ryanodine receptor calcium channel complex” (RyR). In mice that lack dystrophin, Rycal ARM210 corrected a calcium leak occurring through the RyR and improved daily activity, strength, muscle force, and muscle damage. These studies help establish the rationale for conducting a clinical trial with this compound in Duchenne.

• **What is the current state of this research and what steps need to be completed before moving into a clinical trial?**
  Formal non-clinical toxicology studies required by the FDA have been completed. ARM210 was found to be safe and well tolerated. A first time in human Phase 1 clinical safety study in adult healthy volunteers is planned to start in 3Q 2015.

• **What is your best estimate for the length of time it will take to move this research into clinical trials in Duchenne patients?**
  Following successful completion of a Phase 1 clinical safety study in adult healthy volunteers and discussions with clinical experts and regulators, an initial clinical study in Duchenne patients is planned to start in late 2016 or early 2017.

• **Where would a clinical trial take place?**
  It is too early to know where a clinical trial for this research would be located. Many complex factors go into determining the right location(s) for a clinical trial.

• **Who would be eligible to participate in a clinical trial?**
  Again, it is too early to know what the inclusion criteria would be for a future clinical trial.

• **Where can I learn more about this research?**
  You can learn more about ARM210 at ARMGO’s website (http://www.armgo.com/).
  www.ClinicalTrials.gov will post all clinical trials once they are actively recruiting patients.
  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
• What stage is this research?
  This compound is in preclinical research, meaning it has not yet advanced to clinical trials.

• What is the goal or purpose of this research?
  The purpose of this preclinical research is to identify the optimal dosing level and regimen for use in a future clinical study. The research compound, AT-300, is a novel modulator of stretch-activated calcium channels. It is intended to help restore normal levels of calcium in Duchenne skeletal and cardiac muscle. Abnormally high levels of calcium in Duchenne muscle contribute to loss of function and eventually to muscle cell death. AT-300 would be relevant for all boys and young men with Duchenne, regardless of mutation. Akashi Therapeutics is conducting this research and has received Orphan Drug status for AT-300 for Duchenne.

• Who is funding this research?
  The drug is being developed for Duchenne by Akashi Therapeutics, Inc.

• What steps need to be completed before moving into a clinical trial?
  We need to complete the determination of the optimal dose level and regimen, and then conduct a series of preclinical safety studies to establish preliminary safety before filing for an IND and conducting a study in humans, most likely in healthy volunteers.

• What is your best estimate of when AT-300 could enter a clinical trial?
  The initial clinical study, a safety study that would most likely be conducted in healthy volunteers, could start as soon as the second half of 2016.

• Where can I learn more about this research?
  You can learn more at www.akashirx.com (http://akashirx.com/)
Biglycan
A Unique Utrophin Upregulator

• What stage is this research?
This research is pre-clinical, meaning it has not advanced to clinical trials involving people yet.

• Where is this research being done and who is funding this research?
This research is taking place at Tivorsan and in the laboratory of Dr. Justin Fallon at Brown University. This work was funded by PPMD’s End Duchenne GAP program. Current funding is coming from private investors in Tivorsan, recently awarded PPMD and MDA foundation grants to Tivorsan, and an NIH grant (“U01 mechanism”) to Dr. Fallon.

• What is the goal or purpose of this research?
The goal of this research is to use a protein called recombinant human biglycan (rh-biglycan) to increase utrophin and neuronal NOS (nNOS) at the muscle cell membrane, resulting in reduced muscle damage and improved muscle function. Utrophin is a molecule that is related to dystrophin in structure and form and can "stand in" for dystrophin when present in larger than normal quantities.

• What is the current state of this research?
Independent laboratories have reproduced the beneficial effect of rh-biglycan (rhBGN) in mice that lack dystrophin. A reliable method to manufacture the protein has been established and a scalable production process is being optimized. Additional pre-clinical studies are currently in process.

An optimized version of rhBGN has been developed and this molecule, called TVN-102, has been designated the lead clinical candidate.

Tivorsan has initiated discussions with FDA to define its IND-enabling preclinical studies and early clinical development plan.

• What steps need to be completed before moving into a clinical trial?
Manufacturing of TVN-102 needs to be scaled-up to produce quantities and purity necessary for use in humans. Safety testing must be completed and the pharmacology properties of TVN-102 must be determined.

• What is your best estimate for the length of time it will take to move this research into clinical trials?
12–18 months

• Where would a clinical trial take place?
It is too early to know where a clinical trial for this research would be located. Many complex factors go into determining the right location(s) for a clinical trial.

• Who would be eligible to participate in a clinical trial?
Again, it is too early to know what the inclusion criteria would be for a future clinical trial. However, TVN-102 therapy is applicable to all forms of Duchenne, regardless of the underlying mutation.

• Where can I learn more about this research?
You can learn more about this research at www.Tivorsan.com. www.ClinicalTrials.gov will post all clinical trials once they are actively recruiting patients.
Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Carmeseal-MD™
Poloxamer 188 NF

• What stage is this research?
  In the US: Our research is at the pre-clinical stage, our product has not advanced to clinical trials involving people yet. We have completed a pre-IND (Investigational New Drug) meeting with FDA and plan to start a clinical trial next.
  In Europe: While we are developing Carmeseal-MD in the US under FDA supervision, it is already available to patients in Europe as an unlicensed special.

• Where is this research being done and who is funding this research?
  Phrixus studies are currently supported by the SMARTT (Science Moving towards Research Translation and Therapy) program at NHLBI which is providing preclinical, manufacturing, formulation and regulatory support. Technical work is being conducted at SRI International. Our work to date has been funded by the National Institutes of Health (NIH) through SBIR grants, the Biosciences Research and Commercialization Center as well as Coalition Duchenne and Duchenne Alliance.

• What is the goal or purpose of this research?
  Our goal is to demonstrate that Carmeseal-MD has beneficial effects in patients with both Duchenne and Becker muscular dystrophy: An improvement in cardiac and respiratory function via protection of heart muscle and diaphragm. Carmeseal-MD acts as a molecular band-aid by binding to and then sealing microscopic tears in muscle cells caused by the lack of functional dystrophin. This prevents the uncontrolled leakage of calcium which in turn increases the performance of heart muscle and diaphragm and prevents their degeneration.

• What is the current state of this research?
  Carmeseal-MD has been shown to be effective in three dystrophic animal models (mdx and mdx/utr double-knock out mice, GRMD dogs) and two models of heart failure (rats with surgically induced heart failure, micro-embolism induced dog heart failure model). All pre-clinical studies are complete, including three-month GLP toxicology studies in rats and dogs, except for actual manufacturing of clinical supplies and stability studies.

• What steps need to be completed before moving into a clinical trial?
  We have written agreement with FDA that all that is needed is the manufacturing of the clinical supplies and one month stability studies.

• What is your best estimate for the length of time it will take to move this research into clinical trials?
  3–6 months after further funding.

• Where would a clinical trial take place?
  We are already collaborating with leading research clinicians and have verbal commitments from several to conduct the first trials. High likelihood centers include Cincinnati Childrens Hospital (John Jefferies, supported our pre-IND meeting); University of Tennessee Health Science Center (Jeff Towbin, supported our pre-IND meeting); University of Chicago (Beth McNally, on our SMAB); Nationwide Children's Hospital, Columbus, OH (Linda Cripe, on our SMAB); UPMC, Pittsburgh, PA (Jonathan Finder, supported our pre-IND meeting); and University of Michigan, Ann Arbor, MI (Mark Russell).

• Who would be eligible to participate in a clinical trial?
  Carmeseal-MD is expected to be useful for all patients with Duchenne or Becker, regardless of genetic mutation. While inclusion and exclusion criteria still need to be finalized, we expect enrollment of non-ambulatory patients with early cardiac and respiratory dysfunction. This approach maximizes our chances of seeing a positive effect and provides a unique clinical trial opportunity for patients who cannot perform the six-minute-walk test.

• Where can I learn more about this research?
  Please visit www.phrixuspharmaceuticals.com/index.htm. Updates to this FAQ sheet will be posted on www.DuchenneConnect.org.
DMD: iPS Cell Therapy
Duchenne Muscular Dystrophy: iPS Cells and Therapeutic Applications

- **What stage is this research?**
  This research is pre-clinical, meaning it has not advanced to clinical trials involving people yet.

- **Where is this research being done and who is funding this research?**
  This research is being done in the lab of Dr. Rita Perlingeiro at the University of Minnesota and is funded by grants from PPMD and MDA.

- **What is the goal or purpose of this research?**
  Induced pluripotent stem cells (iPS) are adult cells that have been reprogrammed to an embryonic stem cell-like state.
  There has been tremendous excitement for the therapeutic potential of iPS cells in treating genetic diseases. Our current research builds on our successful proof-of-principle studies for Duchenne performed with mouse wild-type and dystrophic iPS cells as well as control (healthy) human iPS cells. These studies demonstrate equivalent functional myogenic engraftment to that observed with their embryonic counterparts following their transplantation into dystrophic mice. Our goal now is to apply this technology to iPS cells obtained from patients with Duchenne by establishing methods to genetically correct the disease, and to evaluate the regenerative potential of resulting genetically corrected iPS cells in dystrophic mice. We are also developing a protocol for making iPS cells and their muscle derivative using integration-free methods.

- **What steps need to be completed before moving into a clinical trial?**
  Safety and efficacy must be established in our pre-clinical work before moving into a clinical trial.

- **What is your best estimate for the length of time it will take to move this research into clinical trials?**
  3+ years

- **Where would a clinical trial take place?**
  It is too early to know where a clinical trial for this research would be located. Many complex factors go into determining the right location(s) for a clinical trial.

- **Who would be eligible to participate in a clinical trial?**
  Again, it is too early to know what the inclusion criteria would be for a future clinical trial.

- **Where can I learn more about this research?**
  You can learn more about this research at the website for Dr. Perlingeiro’s lab:
  http://www.med.umn.edu/lhi/research/PerlingeiroLab/index.htm
  www.ClinicalTrials.gov will post all clinical trials once they are actively recruiting patients.
  Please check www.duchenneconnect.org for updates to this FAQ sheet.
DT-200
A Selective Androgen Receptor Modulator to Improve Muscle Strength & Function in Duchenne

• What stage is this research?
The DT-200 development program has completed three initial Phase 1 trials in 78 healthy adult volunteers showing the SARM is safe and well tolerated for up to 14 days treatment. Testing in Duchenne has not yet begun.

• Where is this research being done and who is funding this research?
DT-200 is wholly owned by Akashi Therapeutics, who is leading the clinical development program. Akashi is currently seeking funding for the next development step, a four week Proof of Concept (POC) clinical trial.

• What is the goal or purpose of this research?
DT-200 is a selective androgen receptor modulator (SARM). Selective androgen receptor modulators (SARMs) have been developed to mimic the muscle building effects of androgens (testosterone), without their undesirable side effects. Akashi hopes that the more precise action of this drug will confer better long term safety and tolerability in both adult and pediatric muscle diseases compared to androgens. DT-200 is effective in multiple animal models, including mice that lack dystrophin. Importantly, compared to other SARMs in clinical development, DT-200 shows significantly greater preference for skeletal muscle. Accordingly, Akashi plans to develop DT-200 with the objective of improving muscle strength and function in both adult muscle diseases, such as Charcot Marie Tooth disease (CMT) and Facioscapulohumeral Muscular Dystrophy (FSHD), and in pediatric myopathies, such as Duchenne and Spinal Muscle Atrophy (SMA).

• What is the current state of this research and what steps need to be completed before moving into a clinical trial?
Three Phase 1 studies with DT-200 were successfully completed in Belgium and Germany, having confirmed the product is safe and well tolerated at the once daily oral dose of 0.5 mg. Akashi met with the UK Regulatory Agency (MHRA) for scientific advice and input into the design of a Proof of Concept clinical trial. Akashi plans to initiate this four week POC trial in the UK in healthy adult volunteers. The main objective of the POC trial is first to evaluate DT-200’s ability to increase mass, strength and function of healthy muscle. It is anticipated the POC trial will take approximately nine months to complete.

• What is your best estimate for the length of time it will take to move this research into clinical trials?
If the POC trial in healthy adult subjects is positive, Akashi anticipates initiation of clinical trials in adult and pediatric muscle diseases within six months from the conclusion of the POC trial.

• Where would a clinical trial take place?
Phase 2 trials would be initiated in both the US and Europe.

• Who would be eligible to participate in a clinical trial?
It is too early to know what the inclusion criteria would be for a future clinical trial.

• Where can I learn more about this research?
You can learn more about DT-200 (and other Akashi initiatives) by consulting the Akashi website at: http://akashirx.com www.ClinicalTrials.gov will post all DT-200 clinical trials once they are actively recruiting subjects.
Please check www.DuchenneConnect.org (http://www.duchenneconnect.org) for updates to this FAQ sheet.
Exon 2 Skipping for IRES Activation
Exon 2 skipping therapy to induce Internal Ribosomal Entry Site (IRES) activation in Duchenne patients with exon 2 duplications

- **What stage is this research?**
  This research is pre-clinical, meaning it has not advanced to clinical trials involving people yet.

- **What steps need to be completed before moving into a clinical trial?**
  Completion of pre-clinical efficacy studies to determine the minimal efficacious dose. Completion of IND-enabling in-life toxicity studies in mice.

- **What is your best estimate for the length of time it will take to move this research into clinical trials?**
  One year.

- **What is the goal or purpose of this study?**
  The goal of this study is to induce skipping of either one of both copies of exon 2 in patients with exon 2 duplications. We intended to induce exon skipping by the use of a virus carrying several copies of a modified small nuclear RNA (U7snRNA) targeted to exon 2, where it interferes with splicing. Skipping of one copy of exon 2 will be expected to result in a wild-type Duchenne transcript and expression of a full-length dystrophin protein. Skipping of both copies of exon 2 will be expected to result in the activation of an IRES in exon 5 that results in the production of a highly functional version of the dystrophin protein.

- **Who is funding this study?**
  The preclinical development of this study has been funded by Cure-Duchenne. Funding for the clinical trial has not yet been obtained.

- **Who is eligible to participate in this study?**
  The final enrollment criteria are not set, but we anticipate enrolling ambulate patients who carry a duplication of exon 2.

- **What do I have to do if I decide to participate in this study?**
  This study will involve frequent visits to Nationwide Children’s Hospital. Delivery of the vector will likely require sedation and delivery to both legs via a catheter. Muscle biopsies before and 12 weeks after vector delivery will be required.

- **How long will this study last, and will I have access to the drug/treatment once the study has ended?**
  The study will require follow up for two years after injection. Patients who participate in the trial are unlikely to be eligible for later gene delivery with the same vector, due to the expected development of antibodies to the viral capsid. However, animal studies suggest that genes delivered by AAV viruses will last for years. If the treatment were to be approved by the FDA, plasmapheresis to clear anti-AAV antibodies may allow future treatment, although this cannot be guaranteed at present.

- **Why should I consider participating in this study?**
  Patients who naturally express the IRES-driven isoform of the dystrophin protein walk in to their seventh decade. Studies in an animal model of exon 2 duplication Duchenne show that skipping can be done very efficiently with the virus.

- **Where can I learn more about this study?**
  You will be able to learn more about this study at www.DuchenneConnect.org and www.ClinicalTrials.gov, and www.nationwidechildrens.org/center-for-gene-therapy.
Laminin-111
Laminin-111, Integrin and Utrophin as a Potential Therapy for Duchenne Muscular Dystrophy

• What stage is this research?
This research is pre-clinical, meaning it has not advanced to clinical trials involving people yet.

• Where is this research being done and who is funding this research?
LAM-111 research for the treatment of merosin-deficient congenital muscular dystrophy (MDC1A) is currently being performed at the University of Nevada, Reno; and Alexion Pharmaceuticals in Connecticut.

Prothelia’s funding has been primarily through US Government grants. Prothelia has received funding from several advocacy groups including PPMD’s End Duchenne GAP program, Struggle Against Muscular Dystrophy (SAM), and Hope for Gus. The current effort has shifted focus towards the use of LAM-111 for treatment of MDC1A and is being funded by Alexion.

• What is the goal or purpose of this research?
Animal data shows that human laminin-111 upregulates (increases) the molecules integrin and utrophin. Both integrin and utrophin work together to restore lost muscle cell adhesion when dystrophin is missing at the muscle membrane. This approach should help all patients regardless of their dystrophin mutation.

• What is the current state of this research?
This project is in preclinical development with demonstrated effectiveness in the mdx and dyW mouse models of Duchenne and MDC1A, respectively. Current efforts are focused on MDC1A, though positive clinical data for MDC1A may accelerate clinical development for Duchenne. Further studies in a more severely affected animal model of Duchenne may be required to justify further development of LAM-111 for treatment of Duchenne.

• What steps need to be completed before moving into a clinical trial?
A scalable process must be constructed that produces sterile recombinant human laminin-111 (rhLAM-111). Prior to any testing of rhLAM-111 in clinical (human) studies, we must ensure it is safe and effective in the mouse and large animal models of muscular dystrophy.

• What is your best estimate for the length of time it will take to move this research into clinical trials?
Uncertain. Current efforts are focused on MDC1A.

• Where would a clinical trial take place?
There are several candidate locations, including hospitals in Cincinnati (OH), Boston (MA), and Rochester (NY). However, there are many complex factors that go into determining the right location/countries for a clinical trial and further assessments will be made once we approach clinical development.

• Where can I learn more about this research?
www.Prothelia.com
Please check www.DuchenneConnect.org for updates to this FAQ sheet.
NBD Peptide
Using NF-κB blockers to Decrease Inflammation and Improve Muscle Function in Duchenne

• What stage is this research?
  This research in Duchenne is pre-clinical, meaning it has not advanced to clinical trials involving people with Duchenne yet.

• Where is this research being done and who is funding this research?
  This research is being done in Dr. Denis Guttridge’s laboratory at The Ohio State University in collaboration with colleagues at The Ohio State University and Nationwide Children’s Hospital. The work has been funded by the NINDS branch of the NIH.

• What is the goal or purpose of this research?
  The NF-κB pathway has been shown to be involved in promoting inflammation and compromising muscle function in response to the loss of dystrophin in Duchenne. Dr. Guttridge’s group has used a small molecule called “NBD” to specifically block this pathway. In mice that lack dystrophin, NBD significantly improves the function of breathing muscles and allowed the mice to maintain whole body function. Also, in mice that lack dystrophin and utrophin, the drug significantly improved cardiac function. Skeletal muscle improvements were also observed when NBD was administered to dogs lacking dystrophin.

• What is the current state of this research and what steps need to be completed before moving into a clinical trial?
  Discussions have taken place with the FDA in order to submit a pre-Investigational New Drug application.
  Studies in a large animal model have been completed and the findings have now been published.
  NBD is being produced on a large scale at a commercial vendor.
  Non-GLP pharmacology and toxicology testing have been completed. Formal GLP toxicology studies have been planned.

• What is your best estimate for the length of time it will take to move this research into clinical trials?
  At this time, it is too early to estimate when a clinical trial in Duchenne would start. However, preparations are in place to complete the final studies leading to an IND submission.

• Where would a clinical trial take place?
  The Phase 1 safety trial is planned for Nationwide Children’s Hospital, Columbus, Ohio, directed by Dr. Jerry Mendell. For Phase 2, it is too early to know where a clinical trial will take place and much will depend on the safety profile determined in Phase 1.

• Who would be eligible to participate in a clinical trial?
  Again, it is too early to know what the inclusion criteria would be for a future clinical trial.

• Where can I learn more about this research?
  You can learn more about Dr. Guttridge’s research at The Ohio State University website (http://biomed.osu.edu).
  www.ClinicalTrials.gov will post all clinical trials once they are actively recruiting patients.
  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
RTC13
RTC13 Read-Through Compound-Development of a drug that corrects nonsense mutations in patients with Duchenne

• What stage is this research?
This research is preclinical, meaning it has not advanced to clinical trials involving people yet.

• Where is this research being done and who is funding this research?
This research is being done at the University of California Los Angeles (UCLA) and is or has been funded in the past by the Muscular Dystrophy Association (MDA), the National Institutes of Health (NIH), and the Department of Defense (DoD).

• What is the goal or purpose of this research?
We have identified a molecule called RTC13 that can restore a full length dystrophin protein in skeletal muscles of Duchenne patients affected by nonsense mutations. They are generally caused by single point mutations in the dystrophin gene that lead to the inappropriate presence of specific sequences (UAA, UAG, or UGA) called stop codons. These stop codons cause a premature arrest in the synthesis of the dystrophin protein. As a result, no dystrophin is produced in skeletal muscles and heart. We have recently shown that this drug can restore dystrophin expression in muscles of mdx mice, a widely used animal model for Duchenne. Our goal is to optimize the dose necessary to achieve therapeutic effects in Duchenne patients, and to conduct the safety and toxicology studies required to file an Investigational New Drug (IND) application to the Food and Drug Administration (FDA). It has been estimated that approximately 13% of Duchenne patients could benefit from readthrough of nonsense mutations. Importantly, because the drug restores full length dystrophin, the protein being produced is expected to be fully functional and should be able to halt or at least counteract disease progression.

• What is the current state of this research?
Our data demonstrate that RTC13 can efficiently restore dystrophin into muscle of Duchenne models with substantial beneficial effects achieved on muscle function. We are currently optimizing an orally viable formulation of the drug that can be administered to patients as this represents the best option to treat the disorder.

• What steps need to be completed before moving into a clinical trial?
We have completed proof of concept studies in the mdx mouse model for Duchenne. We are now focusing on conducting the toxicology studies needed to demonstrate that the compound is safe to use in children and young adults. The steps necessary to conduct toxicology and safety studies will require extensive economical resources. As such, effort will be placed on identifying potential funding sources through US government grants and other advocacy groups.

• What is your best estimate for the length of time it will take to move this research into clinical trials?
2–3 years

• Where would a clinical trial take place?
It is too early to know where a clinical trial for this research would be located.

• Who would be eligible to participate in a clinical trial?
Again, it is too early to know what the inclusion criteria would be for a future clinical trial.

• Where can I learn more about this research?
You can learn more about this research at http://bertonilab.neurology.ucla.edu/index.html.
www.ClinicalTrials.gov will post all clinical trials once they are actively recruiting patients.
Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Tamoxifen
Using tamoxifen to improve muscle strength in Duchenne and Becker

• What stage is this research?
This research in Duchenne is currently at the transition from pre-clinical to clinical, meaning that pre-clinical data and previous experience with this drug are sufficient to start clinical trials involving people. Tamoxifen is the generic name for an approved drug (Nolvadex) that is used to treat estrogen-dependent breast cancer.

• Where is this research being done and who is funding this research?
This research is being done at Dr. Urs Ruegg’s laboratory by Dr. Olivier Dorchies at the University of Geneva using mdx mice. They have shown that tamoxifen triggers substantial improvements in muscle quality and strength in mdx mice. Parent Project Muscular Dystrophy has been helping to fund this research.

• What is the goal or purpose of this research?
The main goal is to collect pre-clinical data that are convincing to take tamoxifen into a clinical trial in Duchenne and perhaps Becker. Low doses (threshold 0.3 mg/kg body weight per day) were effective to improve muscle function. This very low dose suggests that the drug acts on a high-affinity target, probably one of the estrogen receptors, ERα and ERβ. Studies are ongoing to investigate the exact mechanism of action using mice deleted of either ERα or ERβ and antagonists of these receptors.

Ultimately, tamoxifen may be part of a “cocktail” along with other treatments that slow or stop the loss of strength in Duchenne or Becker. It is also possible that tamoxifen could take the place of prednisone as an alternative with fewer side effects or that lower prednisolone doses could be used in combination with tamoxifen.

• What is the current state of this research?
Drs. Ruegg and Dorchies have tested various doses of tamoxifen in both young and old mice. Benefits of treatment included lower creatine kinase levels, 40% less fibrosis in diaphragm and heart than in untreated mice, and near normal improvements in muscle strength. Although mice are not humans and it is never entirely certain how results will translate, a robust treatment response in mice that lack dystrophin is the gold standard for moving drugs into clinical trials for Duchenne.

The laboratory of Dr. Dominic Wells at the University of London has shown that there is good reproducibility and that the combination of tamoxifen plus prednisolone gives a slightly stronger effect than any of the two alone.

• What steps need to be completed before moving into a clinical trial?
While current data are encouraging, we don’t yet know if tamoxifen will have the same positive effects in Duchenne and Becker patients. We need to understand more about its mechanism of action and need to investigate if tamoxifen prevents muscle necrosis and/or enhances muscle regeneration. This would be an additional benefit. These experiments can go on in parallel with a clinical trial.

Noteworthy, the pharmacological profile of tamoxifen is well known; it has also been given as an anti-tumor agent to children aged 5 to 12, and no undesired effects have been noted.

• What is your best estimate for the length of time it will take to move this research into clinical trials?
There are two points of view: Some think that it should be tested in dystrophic dogs before a clinical trial, whereas others don’t see the benefit of a canine study, given that tamoxifen has been used for more than 30 years to treat breast cancer. Without a study in dogs, a clinical trial with Duchenne patients may start around the end of 2015. If a study in dogs is required, a clinical trial would start 2-3 years later.

• Where would a clinical trial take place?
Many factors go into determining the right location(s) for a clinical trial. It is likely that Dr. Lee Sweeney will be the coordinator of a first pilot trial and that PPMD will provide funding.

• Who would be eligible to participate in a clinical trial?
It is too early to know what the inclusion criteria will. The improvements seen in the mice with tamoxifen are very likely not dependent on a particular type of mutation in the dystrophin gene, so tamoxifen is potentially applicable to all boys and young men with Duchenne and Becker. This drug is relatively inexpensive, about USD 2000-4000/year.

• Where can I learn more about this research?
www.ClinicalTrials.gov will post this clinical trial once it is actively recruiting patients.

Please check www.DuchenneConnect.org for updates to this FAQ sheet.
Coenzyme Q10 and Lisinopril
Clinical Trial of Coenzyme Q10 and Lisinopril in Muscular Dystrophies

• **What stage is this research?**
  This clinical trial is closed to new enrollment, but actively following previously enrolled participants.

• **What is the goal or purpose of this study?**
  This is a clinical trial to test a medication used for the heart, called lisinopril (an angiotensin converting enzyme (ACE) inhibitor) and supplement called Coenzyme Q10, to ameliorate the decline in cardiac muscle functions that occurs in muscular dystrophies. The goal of this study is to determine if Coenzyme Q10 alone, lisinopril alone, or a combination of Coenzyme Q10 and lisinopril is more effective at delaying the onset of cardiac symptoms in patients with Duchenne, Becker, or Limb Girdle muscular dystrophy.

• **Who is funding this study?**
  This study is funded by the Department of Defense (DOD).

• **Who was eligible to participate in this study?**
  Participants must have a confirmed genetic diagnosis of Duchenne, Becker, or Limb Girdle muscular dystrophy (certain type 2 only), be 8 years of age or older, and have no clinical cardiac symptoms with a normal left ventricular fractional shortening (>28%) on echocardiogram. They must not have used Coenzyme Q10, lisinopril, or beta blockers in the past for longer than six months.

• **Where does this study take place?**
  This study enrolled participants at select Cooperative International Neuromuscular Research Group (CINRG) network sites. These sites include: Children’s National Medical Center, Washington, DC; Ann & Robert H. Lurie Children’s Hospital of Chicago, Chicago, IL; Carolinas Medical Center, Charlotte, NC; University of Pittsburgh, Pittsburgh, PA; University of Tennessee, Memphis, TN; National Center of Neurology and Psychiatry, Tokyo, Japan; and Alberta Children’s Hospital, Calgary, Alberta, Canada.

• **When is the study anticipated to be completed and results available?**
  The study is anticipated to be completed in December 2016 and results available in 2017.

• **Where can I learn more about this study?**
  You can learn more about this study at www.cinrgresearch.org and www.clinicaltrials.gov (NCT01126697).

Please check www.DuchenneConnect.org for updates to this FAQ sheet.
**Eteplirsen (Study 4658-203)**

An open-label, multi-center study to evaluate the safety, efficacy and tolerability of eteplirsen in early stage Duchenne muscular dystrophy

- **What stage is this research study?**
  Study 4658-203 is a Phase 2 study that is currently recruiting participants at clinical sites in the United States.

- **What is the goal or purpose of this research?**
  This Phase II open-label study is being conducted to assess the safety, tolerability, efficacy and pharmacokinetics of the potential exon 51 skipping therapy eteplirsen in patients with early stage Duchenne muscular dystrophy (Duchenne) who are amenable to exon 51 skipping.

- **Who is sponsoring this study?**
  The study is sponsored by Sarepta Therapeutics.

- **Who might be eligible to participate in this study?**
  Researchers are recruiting approximately 20 males for the treated group and approximately 20 males for the untreated group.
  Key inclusion criteria include but are not limited to:
  - Male 4-6 years of age.
  - Diagnosis of Duchenne, genotypically confirmed.
  - Stable dose of oral corticosteroids for at least 12 weeks or has not received corticosteroids for at least 12 weeks.
  - Intact right and left biceps muscles or two alternative upper arm muscle groups.
  - Parent that is willing to provide consent and comply with study procedures.

  Additional criteria apply and will be reviewed with patients during the screening process. The Principal Investigator (study doctor) determines whether a patient meets the inclusion and exclusion criteria for the study, and therefore whether or not the patient is eligible to participate.

- **Where will this study take place?**
  This study will be conducted at clinical sites in the United States. To view participating study sites, including those that are actively recruiting patients, visit www.clinicaltrials.gov (Identifier NCT02420379).

- **Why is Sarepta including an untreated group of patients in this study?**
  Patients in the untreated arm will serve as a control arm to patients in the treated arm. Patients enrolled in the untreated and treated arms of this clinical study will perform similar physical assessments and procedures throughout the study (e.g., 6MWT). The information collected from these assessments and procedures will then be compared between the treated and untreated groups to help evaluate the safety and efficacy of eteplirsen.

- **Why should I consider participating in this study?**
  While no benefit can be guaranteed from participation in any clinical study, we believe you may:
  - Have access to highly experienced clinicians with strong expertise in treating Duchenne, who might not normally be accessible to you and your family
  - Receive notification and knowledge of future clinical studies for which you or your son may be eligible
  - Gain better understanding of how the patient’s Duchenne is progressing
  - Have the opportunity to become more familiar with what participation in a clinical study entails
  - Contribute to what is now known about Duchenne progression
  - Have the opportunity to help others by contributing to medical research that may accelerate the development of Duchenne therapies

Choosing to participate in a study is an important personal decision. Talk with your doctor and family members or friends about deciding to join a study.

- **Will I be compensated for participating in this study?**
  Generally, reasonable costs associated with participation in the study will be reimbursed in accordance with the approved travel policy for the study. Additional information will be provided by individual study sites.

- **If enrolled, what can I expect during the study?**
  The Principal Investigator (study doctor) and/or the study site contact will review study requirements with all patients during the screening process.
  Patients enrolled in the treated group will receive weekly infusions of the investigational therapy eteplirsen. Patients will visit sites for functional testing and laboratory work, biopsies will be conducted, and MRI assessments will be collected from all patients during the course of the study.
Patients enrolled in the untreated group will not receive the investigational therapy eteplirsen or undergo muscle biopsy. Functional tests, laboratory work, minimally invasive procedures (i.e., blood draws) and MRI assessments will be collected from untreated patients during the course of the study.

Safety, including adverse event monitoring and routine laboratory assessments, will be continuously monitored for all patients in the study.

**Where can I learn more about this clinical study?**
To learn more about this study, you or your doctor may contact the study research staff using the contacts posted on www.clinicaltrials.gov (Identifier NCT02255552). You may also email trialinfo@sarepta.com or register on www.skipahead.com to receive information from Sarepta, including updates on our clinical studies.
• What stage is this research?
  This is an active trial, but enrollment for Phase 1 has been completed. We are anticipating that this study will be extended into a Phase 2, with enrollment starting in September 2015.

• What is the goal or purpose of this study?
  This study focuses on developing Magnetic Resonance Imaging (MRI) as a tool to monitor disease progression in Duchenne and to serve as an outcome measure for clinical trials. The aim of the study is to determine whether noninvasive MRI outcome measures can replace muscle biopsies in evaluating the effectiveness of new treatments in future clinical trials.

  In Phase 1 of this study we demonstrated that MRI studies of the legs in ambulatory boys are very sensitive to disease progression, can be reliably implemented across sites, are predictive of loss of ambulation, and correlate with function. This is extremely important as it provides strong evidence that MRI can be valuable as a biomarker in clinical trials.

  In Phase 2 of this study we will perform MRI studies of the arms and study the effect of loss of ambulation on the MRI biomarkers. The reason for studying the arms and MRI biomarkers in boys that lose the ability to walk is to set the stage for the inclusion of non-ambulatory boys in future clinical trials.

• Who is funding this study?
  This study is funded by the NIH – NIAMS/NINDS.

• Who is eligible to participate in this study?
  Phase 1: Enrollment is complete.
  Phase 2: Participants in this study will be males with Duchenne, ages 5–18 years ambulatory and non-ambulatory upon entering the study.

• What are participants doing in this study?
  Phase 1: MRI and Magnetic Resonance Spectroscopy (MRS) measurements are performed on the participants’ leg muscles, and muscle strength and functional tests such as walking and climbing four steps are also performed. Additionally, a small sample of skin cells will be taken from the participants and stored in established tissue banks.

  Phase 2: MRI and Magnetic Resonance Spectroscopy (MRS) measurements are performed on the participants’ arm as well as leg muscles, and muscle strength and functional tests such as walking and climbing four steps are also performed. Additionally, a blood and urine sample will be taken from the participants and stored in established tissue banks.

• Where does this study take place?
  This study is taking place in 3 different cities in the US: Gainesville, FL at the University of Florida; Philadelphia, PA at Children’s Hospital of Philadelphia (CHOP); and Portland, OR at the Oregon Health and Science University (OHSU) and Shiners Hospital for Children-Portland.

• When will the study be completed?
  The estimated study completion dates is May 2020.

• Are there any preliminary results available?

• Where can I learn more about these studies?
  You can learn more about this study at www.ImagingDMD.org and www.ClinicalTrials.gov (NCT01484678).

  Please check www.DuchenneConnect.org for updates to this FAQ sheet.
• What stage is this research?
A Phase 1b clinical trial in patients with Duchenne is ongoing in the United Kingdom but is no longer recruiting. Preliminary results are expected to be reported in the third quarter of 2015. This current trial is to confirm whether a balanced diet will allow for better absorption of SMT C1100. A Phase 2 open label trial in patients is planned to start in the second half of 2015, and a larger international Phase 2 in early 2016.

• What is the goal or purpose of this research?
Utrophin is a naturally occurring protein that is similar to dystrophin and scientists have shown that modulating its production can compensate for the missing dystrophin and help to restore healthy muscle function.

SMT C1100 is a small molecule utrophin modulator that has the potential to benefit all Duchenne patients regardless of their dystrophin mutation. This is Summit's most advanced utrophin modulator with second and future generation molecules also in development. The approach of utrophin modulation is anticipated to be complementary to other therapeutic approaches currently in development.

• Who is funding this research?
The research is being supported by Summit Therapeutics and the UK government.

• What is the current state of this research?
Preliminary results from a Phase 1b clinical trial in boys aged between 5 and 11 years old were reported in 2014. The study showed SMT C1100 was well tolerated at all doses tested. All the boys had variable blood plasma level of SMT C1100 with only two of the boys achieving levels similar to those of the adult volunteers in the 2012 Phase 1 study. Initial evidence suggests the variability in drug uptake may be due to differences in diet and to other disease-related factors. In addition, in the majority of patients creatine kinase levels were reduced during dosing. Further evaluation of the data from this trial is on-going and it is expected further results will be reported at future scientific meetings.

Preclinical studies established that SMT C1100:
» Increases utrophin protein in dystrophin deficient muscle cells from Duchenne patients to levels expected to have significant therapeutic benefit
» Significantly increases the amount of utrophin in the mdx mouse model of Duchenne
» Improves whole muscle function in a study with an endpoint similar to the 6-minute-walk test
» Reduces muscle degeneration, fibrosis and chronic inflammation

• Where will future trials take place? Will there be study sites in the US?
Clinical trials could take place in either Europe or the US or both, and the harmonization program between the FDA and EMA allows for trial data from one region to be used in clinical trial applications in the other.

• Why should I consider participating in future studies?
While no personal benefit can ever be guaranteed by participation in a clinical trial, there are other benefits, including allowing you to play an active role in the health care of your child, gaining access to medical specialists that are normally not available to your child, and helping others by contributing to the better understanding of Duchenne.

• Where can I learn more about SMT C1100?
You can learn more at www.summitplc.com.
Follow Summit on Twitter: @summitplc
www.ClinicalTrials.gov will post the trials as soon as they are recruiting.
Please check www.DuchenneConnect.org for updates to this FAQ sheet.

SMT C1100
A Small Molecule Utrophin Modulator for Duchenne

#PPMDCONNECT
Tadalafil
A Randomized, Double-Blind, Placebo-Controlled, Phase 3 Trial of Tadalafil for Duchenne Muscular Dystrophy

• What stage is this research?
This Phase 3 clinical trial is active, but no longer enrolling participants.

• What is the goal or purpose of this study?
The main purpose of this study is to determine if tadalafil can slow the decline in walking ability of boys who have Duchenne muscular dystrophy. The study will also assess the safety of tadalafil and any side effects that might be associated with it in boys who have Duchenne.

• Who is the sponsor of this study?
Eli Lilly and Company (www.lilly.com) is the sponsor of this study.

• What were the inclusion (enrollment) criteria for this trial?
Participants are boys with Duchenne who are between the ages of 7–14 years old and ambulatory. Participants must have been on a corticosteroid therapy for at least 6 months prior to screening, and must have had a left ventricular ejection fraction (LVEF) ≥50% as determined by echocardiogram. Additional details regarding the inclusion and exclusion criteria are posted on www.clinicaltrials.gov.

• What are participants doing in this study?
Participants receive study treatment (tadalafil or placebo) for the first 48 weeks of the study, and can then continue into an extension period of at least 96 weeks during which all participants will receive tadalafil. The primary outcome measure that is measured at study visits is the 6-minute-walk test.

• Where does the trial take place?
There are currently sites in 16 states in the US, as well as multiple sites in 14 other countries (Asia, Canada, Europe, and South America). Please check www.clinicaltrials.gov for a complete list of study sites.

• When will the study be completed?
The estimated study completion date for the placebo-controlled period is December of 2015, with initial results expected in the first half of 2016.

• Are there any preliminary results available?
This trial is ongoing and no preliminary results are available.

• Where can I learn more about this study?
Translarna™ (ataluren)
A New Drug for Nonsense Mutations by PTC Therapeutics

• What is the current status of Translarna™ (ataluren)?
PTC Therapeutics is conducting the Ataluren Confirmatory Trial in Duchenne muscular dystrophy (ACT DMD), a Phase 3 trial of the drug in patients with nonsense mutation Duchenne. This randomized, double-blind, placebo-controlled trial is designed to evaluate the efficacy and safety of Translarna™ (ataluren). The study enrolled approximately 230 participants at 54 sites in North America, South America, Europe, Israel, Asia, and Australia. Top-line data from this trial is expected by the end of 2015.

Translarna™ (ataluren), received marketing authorization in the European Union in August 2014 for the treatment of nonsense mutation Duchenne muscular dystrophy in ambulatory patients aged five years and older, representing the first-ever treatment approved for the underlying cause of the disease. The approval in the EU is subject to PTC’s obligation to provide data from the ongoing Phase 3 ACT DMD trial by the end of 2015.

In the US, PTC began a rolling New Drug Application submission to the FDA in December 2014. The data from the ACT DMD trial should form the basis for finalizing the NDA submission. PTC has begun building its US infrastructure in preparation for a potential US launch in the first half of 2016.

Open label studies are still ongoing with those patients who participated in previous Translarna trials.

Translarna is available for siblings of patients in certain open label PTC clinical trials for nonsense mutation Duchenne muscular dystrophy. Translarna will be made available to these siblings in advance of commercial availability in their regions, after a treating physician and the patient or guardian make a clinical decision to use Translarna, and consistent with any other applicable regulatory requirements.

Translarna is being studied in patients with cystic fibrosis (CF) in the Ataluren Confirmatory Trial in Cystic Fibrosis (ACT CF), an ongoing Phase 3 trial that is expected to be fully enrolled in the second half of 2015.

A Phase 2 clinical trial of Translarna in patients with mucopolysaccharidosis type 1 (MPS 1) enrollment is initiating and some data is expected by the end of 2015.

A Phase 2 clinical trial of Translarna in aniridia, a disorder of the eye, is expected to initiate in 2015.

• What is a nonsense mutation?
A nonsense mutation is a premature stop signal in the genetic code that interrupts the production of a protein. Proteins are essential to the proper working of every cell in the body. Nonsense mutations result in incomplete proteins that do not function properly and in turn cause a genetic disorder. For example, in nonsense mutation Duchenne, dystrophin, which is an essential protein in the muscle, is incomplete and non-functional, leading to muscle wasting and progressive loss of muscle strength.

• What is the goal or purpose of this research?
Translarna is a new drug designed to enable the formation of a functioning protein in a patient with a genetic disorder due to a nonsense mutation. Translarna is taken orally and has the potential to treat the root cause of the disorder by overriding the premature stop signal so that a functional protein can be made. It does not alter a patient’s genetic code or introduce genetic materials into the body.

• Who is funding this research?
Translarna was discovered and developed by PTC Therapeutics. Its development has been supported by the FDA Office of Orphan Products Development, Parent Project Muscular Dystrophy, the Muscular Dystrophy Association, Cystic Fibrosis Foundation Therapeutics Inc., and the National Center for Research Resources.

• When will Translarna be commercially available?
Translarna is approved in in the European Union. PTC Therapeutics is undertaking country by country reimbursement negotiations to make commercial access possible. Due to the urgent need for access, PTC Therapeutics has been establishing reimbursed early access programs, or EAP programs, in selected countries to provide reimbursed access to Translarna to nonsense mutation Duchenne patients who meet applicable criteria. The EAP programs intend to make the product available to patients before commercial product becomes available in selected territories. Access is currently provided in Spain, France, Italy, Greece, Israel, Turkey, and Colombia. PTC is also working with the authorities in Brazil and other countries to make Translarna available under EAP programs there. As of April 30th, PTC has 82 Duchenne muscular dystrophy patients on commercial therapy through either reimbursed early access programs or commercial sales.

• How can I find out if Translarna would benefit my child?
Approximately 13% of boys with Duchenne have a nonsense mutation. Patients should discuss the appropriateness of genetic testing with their child’s physician or a genetic counselor. Usually, only a small amount of blood is required to perform the test. The blood sample must be sent to a specialized laboratory that has expertise in Duchenne. Translarna is specifically for patients with a nonsense mutation. It will not benefit patients whose Duchenne is caused by a different mutation, such as a deletion or duplication.

• Where can I learn more about Translarna?
You can learn more about Translarna at www.ptcbio.com.
Please check www.DuchenneConnect.org for updates to this FAQ sheet.
What stage is this research?
This Phase 3 trial was completed with 64 randomized and treated patients ages 10–18 years old that were not using concomitant glucocorticoids, irrespective of mutation status and ability to ambulate.

What was the goal or purpose of this study?
The primary objective of this study was to assess the efficacy of idebenone (Catena®/Raxone®) in improving or delaying the loss of respiratory function.

Who was funding this study?
This study was funded by Santhera Pharmaceuticals.

What is Catena® and how does it work?
Catena® is the U.S. brand name for Santhera’s 150 mg film-coated idebenone tablets. In Europe, the drug brand name is Raxone®. Idebenone works in Duchenne muscular dystrophy to increase the energy output of the cell’s mitochondria—the parts (“factories”) of the cell that generate all of a cell’s activities. Specifically, the drug acts as an electron carrier to provide additional electrons to the mitochondria, which use them to generate energy. Idebenone can carry and drop off electrons within the mitochondria numerous times. In addition to helping cells make extra energy, idebenone is a powerful antioxidant and can neutralize destructive free radicals in cells. All of these activities help dystrophic muscle cells to maintain their cellular energy supply, which is reduced as a result of the lack of dystrophin and protect cells from oxidative stress.

What were patients doing in this study?
Participants were randomized to receive either pharmaceutical-grade idebenone 900 mg/day (300 mg 3 times a day with meals) or matching placebo for 52 weeks. Patients underwent hospital and home-based respiratory assessments.

When was this study completed?
The last patient visit occurred in January 2014 and the study ended in April 2014.

What are the results of the study?
The results of the study were published in the medical journal The Lancet (Buyse et al., 2015; 385: 1748-57). The results of the DELOS trial demonstrated that Catena®/Raxone® significantly reduced the annual decline in Peak Expiratory Flow (PEF as percent predicted, PEF%) by 66% compared to patients taking placebo. Other respiratory function endpoints such as Forced Vital Capacity (FVC) and Peak Expiratory Flow (PEF) by 35% compared to patients taking placebo. The annual decline in FVC in the placebo group was 10.7% compared to 2.4% in the Catena®/Raxone® group (p=0.03). The annual decline in FVC in the placebo group was 9.0% decline in FVC in the placebo group versus a 5.7% decline in the Catena®/Raxone® group (p=0.08).

Will there be additional clinical trials in the future, and if so, when?
It is too early to know if additional trials will be required.

How long will it take for Catena®/Raxone® to be approved and available for use in the United States?
Santhera has initiated discussions with U.S. and European regulators and is preparing for filing of marketing authorization in North America and in Europe. Santhera will provide periodic updates about the regulatory process towards approval.

Where can I learn more about Catena®/Raxone®?
You can learn more at www.santhera.com. The results of the study were published in The Lancet in April 2015 (Buyse et al., 2015; 385: 1748-57).

Please check www.DuchenneConnect.org for updates to this FAQ sheet.
GET INVOLVED

Coach To Cure MD

Save the Date! Coach To Cure MD returns for its 7th year on September 26, 2015.

Looking to get in the game this year?
Help us #TackleDuchenne with 3 easy plays!
1) Talk. Contact your local football team/league to see if they are involved.
2) Tailgate. Have your own event to spread awareness and raise funds for Duchenne.
3) Touchdown. Are you near a school that is looking to have a Duchenne family join them on game day? Find out on CoachToCureMD.org. Contact Danielle Garrigan at Danielle@parentprojectmd.org for more details.

Keep up with everything Coach To Cure MD related:
- Like us on Facebook: Facebook.com/coachtocuremd
- Follow us on Twitter: Twitter.com/coachtocuremd

Run For Our Sons

Join the hundreds of friends and families that pound the pavement every year to help us end Duchenne. An amazing race may be in your own backyard!

Now recruiting for the following worldclass races:
- Disneyland Half Marathon Weekend (Sept. 4–6)
- GoodLife Fitness Victoria Marathon (Oct. 11)
- Under Armour Baltimore Running Festival (Oct. 17)
- TCS New York City Marathon (Nov. 1)
- Avengers Half Marathon Weekend (Nov. 13–15)
- Walt Disney World Marathon Weekend (Jan. 7–10, 2016)
- Star Wars Half Marathon Weekend (Jan. 14–17, 2016)
- Chevron Houston Marathon (Jan. 17, 2016)

Run to make a difference. Run to raise awareness.
Run for those who can’t.

Run For Our Sons.

Visit RunForOurSons.org to learn more.
World Duchenne Awareness Day

People around the world will help participate in the second annual World Duchenne Awareness Day on September 7. Over the coming months, we will tell you about exciting ways you can mark this international event.

Every day, each of us works to fight Duchenne in our families and in our communities, and we’re making an impact. Imagine the impact we can have if we work together as a global community?

Help us make September 7 incredible and join us in World Duchenne Awareness Day!

www.WorldDuchenneAwarenessDay.org

DuchenneConnect

DuchenneConnect is an international registry and resource serving the needs of the Duchenne and Becker community.

Register on DuchenneConnect.org and you will:

• **Advance research** by providing data that is used by researchers and doctors to improve the care and treatment of those with Duchenne and Becker.

• **Learn about actively recruiting clinical trials** and research studies, and how you can participate.

• **Participate in PCORnet**, a national effort to speed up research and answer questions important to many different patient communities.

• **Gain access to valuable educational materials** including a webinar series & bimonthly newsletter.

Already registered?
Please remember to login to your account and update your medical history at least once per year.

Are you a Professional?
Join our email list to receive our newsletter and other important updates, including clinical trial news.

Questions?
Contact the DuchenneConnect Coordinator at 201.937.1408 or coordinator@duchenneconnect.org. The DuchenneConnect Coordinator is a certified genetic counselor who is available to answer your questions.

About DuchenneConnect

DuchenneConnect is a program of Parent Project Muscular Dystrophy (PPMD), the largest, most comprehensive nonprofit organization in the United States focused on finding a cure for Duchenne. PPMD is the sole guardian of DuchenneConnect and its material.

Learn more about how your information is used at DuchenneConnect.org, or contact us: coordinator@duchenneconnect.org.
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