

Parent JOIN THE FIGHT.
END DUCHENNE.
Project
Muscular
Dystrophy

PMPD

Annual Conference 2018
Scottsdale, Arizona

SEPTEMBER IS DUCHENNE ACTION MONTH

SAVE THE DATE! DUCHENNE ACTION MONTH RETURNS THIS SEPTEMBER 2018.

This is the perfect opportunity for you, your family, your colleagues, and your community to join the fight to end Duchenne! The month will be packed full of simple and powerful actions you can take to raise awareness and funds to support PPMD's vital work in the fight to end Duchenne.

- Host a **Race to End Duchenne .1K** to raise funds for PPMD sometime during the month, either in your town, at a local school, or even in your office!
- Have your child's school or your co-workers wear red to **raise awareness** on September 7 (World Duchenne Awareness Day)!
- Host a tailgating or watch party while your favorite **Coach To Cure MD** team plays on September 29 and get sponsors or sell tickets as you join the fight to end Duchenne on the field!
- Create your own **'do it yourself' event** to get your local community involved!
- Ask your state governor or town mayor to recognize **World Duchenne Awareness Day** with a proclamation or have a landmark building "GO RED" with lighting!
- Restock your family's supply of great **PPMD merchandise** so that you can raise awareness all month long!

These are just a few of the ideas we have to help our community raise awareness and funds to support PPMD's mission to end Duchenne this September, but they may require you speaking to your child's school, your employer, or your town to start mapping out the logistics ahead of time — and we are here to help.

Visit parentprojectmd.org/actionmonth to get started!



THIS SEPTEMBER LET'S
ALL DO SOMETHING...

TO HELP RAISE AWARENESS.
TO JOIN THE FIGHT.
TO END DUCHENNE.

WELCOME TO PPMD'S 2018 ANNUAL CONFERENCE

Dear Friends,

Welcome to PPMD's Annual Conference! We are so thrilled to be with all of you at this beautiful resort against the stunning backdrop of the Arizona landscape. Could it be a little cooler? Sure, I wouldn't mind that. But as your East coast and Midwestern PPMD family keeps saying – at least it's a dry heat!

For 24 years, PPMD has gathered this community together to learn the latest updates in research and care, to put our advocacy priorities into action, and to reconnect with friends...friends that have become family. What overwhelms me though, year after year, is the way our Annual Conference continues to grow and expand.

When we first gathered in 1994, the Annual Conference had a handful of researchers and scientists talking to a room of parents and grandparents. It wasn't much more than a day-long meeting because truthfully there wasn't that much to discuss. But there was hope.

Today, we have over 500 attendees! We have dozens of researchers, scientists, clinicians, and physicians. Parents and grandparents aren't just coming – entire families are joining us. Representatives from pharmaceutical companies and small biotechs with Duchenne therapies in the packed therapeutic pipeline are here, as are vendors for tools specifically geared towards people with Duchenne. Our agenda is now three very full days, and it's only that short because we know no one could spend the week to ten days it would take to cover everything happening in our community!

Hope has never been so abundant. Gene therapy isn't the future – it is here and we look forward to what's happening next. A variety of therapeutic approaches are attempting to attack Duchenne from every angle,



“
HOPE HAS NEVER BEEN
SO ABUNDANT”

knowing that it will likely take a combination of therapies to stop the progression of this disease. The way we care for our loved ones living with Duchenne, continues to improve as our understanding of this journey increases. Our voices are being heard as we advocate for our community, not only in the halls of Congress at the federal level, but locally as well.

To paraphrase one of my favorite quotes from Gandhi, we are making the change we want to see in the world. It may not be as fast as we would all like – nothing ever is in our Duchenne world – but change is happening.

So thank you for joining us these next few days for what promises to be a meaningful experience for all of us. You being here makes our Annual Conference stronger and we appreciate your ongoing commitment in the fight to end Duchenne.

Stay cool and we hope you'll take advantage of all the wonderful moments this Conference has to offer!

Pat Furlong
Founding President & CEO,
Parent Project Muscular Dystrophy



SPECIAL EVENTS & MEET UPS



DUCHENNE TEENS & ADULTS SOCIAL

Wednesday, June 27, 7:00–9:30pm

PPMD and PPMD's Adult Advisory Committee (PAAC) will host a social for all teens and adults living with Duchenne. Join us for a casual get together, light appetizers, and drinks.



NEWLY DIAGNOSED MEET AND GREET

Wednesday, June 27, 7:00–9:30pm

This small gathering is for our newly diagnosed families only and will be held on Wednesday evening.



WELCOME RECEPTION

Thursday, June 28, 6:30–8:30pm in Camelback Foyer

Join us on Thursday evening for our Welcome Reception and Resource Fair and Poster Session. Meet and mingle with other conference attendees while visiting our exhibitor booths and posters. Appetizers and drinks will be provided at this reception.



ANNUAL CONFERENCE CELEBRATION DINNER

Saturday, June 30, 7:45–10:30pm

Join us on Saturday evening for our Annual Conference Celebration Dinner. This year, PPMD will host a lively, interactive dance party celebrating the Duchenne community. This is a celebration you will not want to miss and the perfect opportunity to unwind with friends new & old.

SIGN UP FOR...



KIDS TRACK

Friday, June 29, 9:30am–12:30pm in Ballroom G
Saturday, 8:45–11:00am and 1:30–3:30pm in Camelback H

Kids Track kicks off on Friday with an amazing planetarium show! Come meet some new friends and interact with old friends. Then join us for an amazing show under the stars in our very own planetarium.

The fun continues on Saturday, June 30 with age appropriate breakout groups and activities for each level. This year's Kids Track theme is outer space. Later in the day on Saturday, June 30, there will be a movie and other age specific games and activities.

Registration Details: Registration is \$65 per child. Minimum age is 5 or kindergarten. If you did not register your child ahead of time, please speak to someone at PPMD Conference Registration. Kids Track registration also includes a free t-shirt, lunch on Friday, and dinner Saturday night.



RACE TO END DUCHENNE .1K

Friday, June 29 at 6:00pm
Outside the Camelback Ballroom

PPMD will host the Race to End Duchenne .1K on Friday evening! Join us after a long day of presentations for this burst of fun and activity. No one hosts a zany race quite like PPMD. Watch as parents, families, kids of all ages, scientists and researchers, industry sponsors, and ANYONE and EVERYONE gather outside the Camelback Ballroom for this momentous race. Together we will tackle .1K (about the length of a football field!). Yep – that is right. Don't worry, we know this might be a lot, so we will arrange for water stations and even refreshments at the end of the race!

Your registration includes a race shirt, bib, swag bag, and all the fun of racing. You won't want to miss this exciting event. It's a great way to start the day, by raising awareness and having fun doing it!

Registration Details: Registration is \$20 per adult and \$10 per child (kids under 5 are free). Visit PPMD Conference Registration by the end of lunch on Friday to secure your spot!



THANK YOU TO PPMD'S RESOURCE FAIR EXHIBITORS!

The PPMD Resource Fair is an unique opportunity for resource providers and the community to connect about practical services, equipment, and more.

The Resource Fair is located in the East Foyer and will be open Thursday, June 28, from 6:30pm – 9.30pm, and displays may remain open through Friday, June 29 at 2:00pm.

PREMIERE EXHIBITORS



STANDARD EXHIBITORS



THANK YOU TO PERKY JERKY FOR SPONSORING THE RACE TO END DUCHENNE .1K AT PPMD'S 2018 ANNUAL CONFERENCE!



In addition to sponsoring our Race to End Duchenne program in multiple cities, we're working on some exciting new things with Perky Jerky for 2018 and beyond!

FOR FAMILIES



A Duchenne diagnosis can leave families feeling isolated, overwhelmed, and confused. The most important thing for you to remember is that you are not alone.

PPMD is here to connect you to news, resources, advancements in research, and—most importantly—each other. That's one of the reasons we are so glad you are at our Annual Conference this year.

Even if you have been on this journey for years, it may be worth visiting PPMD's new website and exploring all of the resources we provide that could help assist you and your family at various points along the way. Now, more than ever, there is reason to hope – hope for therapies, hope for research, hope for care, hope for a cure. We hope that the resources and guidance that we offer here might be helpful for each family member along this journey.

View these resources at parentprojectmd.org/families

FOR NEWLY DIAGNOSED

If you are the parent of a very young child or a new diagnosis, please spend time in the Newly Diagnosed section of our site. And let the PPMD family know who you are – we are here for you and want to make sure you have the tools you need to begin this journey. There is a lot of information to absorb and the last thing we want to do is overwhelm you. So check out this section, explore, and when you are ready, do not hesitate to reach out. You are never alone.

parentprojectmd.org/newlydiagnosed

ASSEMBLING A CARE TEAM

It is extremely important that you work with a comprehensive, multidisciplinary neuromuscular team that has experience and expertise managing all aspects of patients and families living with Duchenne. This comprehensive team will allow each specialist to give input into the best and most appropriate care for you and your child.

parentprojectmd.org/careteam

CERTIFIED DUCHENNE CARE CENTERS

PPMD is dedicated to ensuring that all families have access to comprehensive, optimal Duchenne care. For this reason, we have started the Certified Duchenne Care Center Program, creating a network of sites capable of providing the highest level of comprehensive Duchenne care. Find out if there is a site near you and learn more about what goes into our certification process.

parentprojectmd.org/carecenters

GENETIC TESTING & INTERPRETATION

Decode Duchenne provides free genetic testing, interpretation, and counseling to people with Duchenne or Becker muscular dystrophy who have been unable to access genetic testing due to financial barriers. The program is administered by Parent Project Muscular Dystrophy through The Duchenne Registry, and is supported by Sarepta Therapeutics and PTC Therapeutics.

parentprojectmd.org/decode



FOR FAMILIES

Certified Duchenne Care Centers: PPMD's Certified Duchenne Care Center network has now grown to 18 incredible centers across the United States, serving more than 2,550 patients and their families. parentprojectmd.org/carecenters



COMMUNITY RESOURCE CENTER

The PPMD Community Resource Center is meant to be a one-stop online resource for every child, adult, and family living with Duchenne. Here you can find some of the favorite resources, products, and organizations of our community members.

As a community resource and online tool, we need you to contribute your experiences and tell your stories to this secure social network of families around the world fighting to end Duchenne.

parentprojectmd.org/resources

INSURANCE INFORMATION

We have all had frustrations getting healthcare paid for — whether it is getting access and coverage for appointments, procedures, equipment, and/or medications. Coverage is especially difficult when new medicines or procedures are recommended. To make this process easier, PPMD has assembled resources that will help families and medical providers at each stage of the healthcare access process.

parentprojectmd.org/accessresources

CLASSROOM RESOURCES

PPMD wants to make sure that families within our Duchenne community are equipped with as many tools as possible to ensure a smooth start to the school year. Because it's hard enough just to figure out the secret code to get through the front entrance, let alone to have to navigate considerations around IEPs (individualized education programs), accessibility, and what information to provide to whom about your child's medical care. So, consider this your Back-To-School Survival Kit (at least related to Duchenne).

parentprojectmd.org/classroom

END DUCHENNE TOUR

Combining each of the pillars that make up PPMD's mission, the End Duchenne Tour brings updates on research, advocacy, and care to families across the country, featuring a roster of leading experts in the Duchenne space. This is your opportunity to interact with vetted leaders in Duchenne, connect with local families on the same journey, and, when possible, explore your area Certified Duchenne Care Center. All meetings are free and kids are welcome to attend and participate in PPMD's Kids Track.

parentprojectmd.org/tour

FOR FAMILIES



PPMD's Advocacy Conference: Never advocated before? No problem! 25 percent of our attendees every year are brand new advocates. PPMD will get you well prepared for your meetings and you will be with other experienced family advocates while on the hill. parentprojectmd.org/advocacy

PPMD'S CONNECT

Online interactions are great, but nothing can replace in-person connections with other families living with Duchenne. PPMD has local Connect groups all over the country for this very reason.

Previously known as FACES, these groups serve many functions, but primarily exist to help you along this journey. Reaching out to your local group will let them know that you are there. Maybe you don't want anything, or maybe you just want to "listen" for awhile — that's ok.

No one needs to go through this experience alone. Finding one or two families to connect with will help you to know that there is hope and that you have support whenever you need it.

parentprojectmd.org/connect

EXPLORE RESEARCH & CLINICAL TRIALS

Duchenne research continues to progress, with multiple therapies in clinical trial. Stay up-to-date on the latest research and learn about actively recruiting clinical trials and studies.

parentprojectmd.org/exploretrials

JOIN THE DUCHENNE REGISTRY

If you have Duchenne or care for someone living with Duchenne, join The Duchenne Registry to share your data. The information you give advances research and treatments for Duchenne, and helps you learn about and enroll in actively recruiting clinical trials and research studies.

duchenneregistry.org

BECOME AN ADVOCATE

Join the fight to end Duchenne by raising your voice in Washington at PPMD's Advocacy Conference. We also encourage you to sign up to receive Action Alerts, so that you can stay up-to-date with the latest advocacy news, as well as reminders to contact legislators on the most pressing issues.

parentprojectmd.org/advocacy



FOR FAMILIES

Race to End Duchenne:

Whether you are a seasoned athlete or a first time runner/walker, we have an event where you can make your miles count even more as you pound the pavement to raise funds for PPMD's mission! racetoendduchenne.org



FUNDRAISING EVENTS

We are only as strong as our community. Join the fight, and together, we will end Duchenne.

Race To End Duchenne

Participate in a Race to End Duchenne near you to join hundreds of individuals and families around the country raising money for Duchenne. You can also show your commitment by supporting or sponsoring a participant. Host a Race to End Duchenne .1K—a run that most anyone can do, in any space. It's a great opportunity to educate a new community about Duchenne (and raise funds for PPMD's essential work along the way).

racetoendduchenne.org

DIY Fundraising to End Duchenne

Do it your way! PPMD's mission is to end Duchenne but we need YOU! Have a great idea for a fundraiser? Looking for ways to raise money to support research? You can do just about anything to raise funds to end Duchenne. And PPMD is here to help you every step of the way.

parentprojectmd.org/diy

Coach To Cure MD

For over a decade, PPMD and college football teams have partnered to raise money and awareness through Coach to Cure MD. This September tradition has multiple ways you and your family can participate on game day — and have a great time doing it. Find out how you can get in the game and join the PPMD team!

coachtocuremd.org

EXPLORE CLINICAL TRIALS



The cornerstone of Parent Project Muscular Dystrophy's mission is to identify and support promising Duchenne muscular dystrophy research that can impact all those living with Duchenne now, during their lifetime. No one in this community will deny the importance of a robust research program, and for 24 years, PPMD has supported innovation we believe has the possibility to treat every single person living with Duchenne.

The following clinical trial and research study FAQ sheets are family-friendly summaries of clinical trials and research studies for people with Duchenne and Becker, as well as female carriers, that will be presented at this year's conference. There are also FAQ sheets for current pre-clinical research that is soon to be in clinical trial. These FAQ sheets have been written for this year's program book in consultation directly with the Researchers.

NOTE: The following FAQ sheets are intended to cover only the research studies that will be presented at this year's conference.

You can find a comprehensive list including additional studies online at parentprojectmd.org/exploretrials.



EXPLORE CLINICAL TRIALS



OBSERVATIONAL



RESTORING OR REPLACING DYSTROPHIN



INFLAMMATION



IMPROVING MUSCLE MECHANICS

ACTIVELY RECRUITING



Therapeutic Approach	Study	Industry/ Institution	Age (years)	Ambulation Status	Mutation Specific	Steroid Use	Location	Page
	Cardiac MRI Biomarkers and Genotype-Phenotype Correlations	NIH	7-21	Either	No	No specific requirement	United States	12
	Genetic Modifiers	UCLA	Any	Either	No	No specific requirement	United States	13
	Ataluren	PTC Therapeutics	5+	Ambulatory	Yes - Nonsense mutation	Currently using	United States + Other	14
	Eteplirsen - Exon 51 Skipping (Study 4658-102)	Sarepta Therapeutics	6 mon - 24 mon	Either	Yes - Deletion amenable to exon skipping	No specific requirement	Europe	15
	Nationwide GALGT2 Gene Therapy	Nationwide Children's Hospital	8+	Ambulatory	No	Currently using	United States	16
	Nationwide Micro-Dystrophin Gene Delivery	Nationwide Children's Hospital	3 mon - 7 yrs	Ambulatory	Yes - Other	No specific requirement	United States	17
	PF-06939926 (Formerly BMB-D001) Mini-Dystrophin Gene Therapy	Pfizer	5-12	Ambulatory	No	Currently using	United States	18
	SGT-001 Micro-Dystrophin Gene Transfer (IGNITE DMD trial)	Solid	4 - 17	Either	No	Currently using	United States	20
	SRP-4045 and SRP-4053 - Exons 45 and 53 Skipping (ESSENCE study)	Sarepta Therapeutics	7-13	Ambulatory	Yes - Deletion amenable to exon skipping	Currently using	Other	21
	SRP-5051 - Exon 51 Skipping (PPMO Study)	Sarepta Therapeutics	12+	Either	Yes - Deletion amenable to exon skipping	No specific requirement	United States	22
	WVE-210201 - Exon 51 Skipping	WAVE Life Sciences	5-18	Either	Yes - Deletion amenable to exon skipping	No specific requirement	United States + Other	23
	CAP-1002 (HOPE-2 trial)	Capricor	10+	Either	No	Currently using	United States	24
	Givinostat	Italfarmaco	6-17	Ambulatory	No	Currently using	United States + Other	25
	MNK-1411 (BRAVE study)	Mallinckrodt	4-8	Ambulatory	No	Never used	United States	27
	Vamorolone	ReveraGen	4-7	Ambulatory	No	Never used	United States + Other	28
	Idebenone (SIDEROS trial)	Santhera	10+	Either	No	Currently using	United States + Other	30
	RG6206 Anti-Myostatin Adnectin	Roche	6-11	Ambulatory	No	Currently using	United States + Other	31

EXPLORE CLINICAL TRIALS



NOT YET RECRUITING



Therapeutic Approach	Study	Industry/ Institution	Page
	Edasalonexent (Formerly CAT-1004)	Catabasis	32

PRECLINICAL



Therapeutic Approach	Study	Industry/ Institution	Page
	Nationwide Exon 2 Skipping for Duplication 2	Nationwide Children's Hospital	37
	Wave Exon 53 Skipping	Wave Life Sciences	38
	Carmeseal-MD™	Phrixus	39

FEMALE CARRIERS



Therapeutic Approach	Study	Industry/ Institution	Age (years)	Ambulation Status	Mutation Specific	Steroid Use	Location	Page
	Nationwide Carrier Study	Nationwide Children's Hospital	18+	Either	No	No specific requirement	United States	40

ONGOING



Therapeutic Approach	Study	Industry/ Institution	Page
	Ezutromid (PhaseOut DMD study)	Summit	33
	NS-065/NCNP-01 - Exon 53 Skipping	NS Pharma	34
	Domagrozumab (Formerly PF-06252616)	Pfizer	35
	Pamrevlumab (Formerly FG-3019)	Fibrogen	36

JOIN THE DUCHENNE REGISTRY TO ADVANCE RESEARCH & SPEED DEVELOPMENT OF NEW TREATMENTS

Join The Duchenne Registry to stay up-to date on ALL therapies that are currently in development. A main goal of The Duchenne Registry is to connect registrants with actively recruiting clinical trials and research studies.

Just by joining **DuchenneRegistry.org** and completing our online surveys, you are actually participating in research studies and providing data that is crucial in the fight to end Duchenne. Join today or update your account at **DuchenneRegistry.org**.



CARDIAC MRI BIOMARKERS AND GENOTYPE-PHENOTYPE CORRELATIONS

VALIDATING CARDIAC MRI BIOMARKERS AND GENOTYPE-PHENOTYPE CORRELATIONS FOR DUCHENNE MUSCULAR DYSTROPHY



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

This trial is actively recruiting participants.

What is the goal or purpose of this study?

The goal of this study is to help researchers identify and validate cardiac MRI biomarkers to better understand the health of the heart and changes in heart health over time in boys with Duchenne muscular dystrophy. The study aims: i) to identify and characterize several cardiac MRI biomarkers for boys with Duchenne; ii) to define the sensitivity of the well-characterized cardiac MRI biomarkers for detecting early cardiac involvement; and iii) to use these validated cardiac MRI biomarkers to better understand the genotype-phenotype correlation in boys with Duchenne.

Who is funding this study?

This study is funded by the National Institutes of Health.

Who is eligible to participate in this study?

To participate in this study you must be:

- Healthy boy or pediatric patient with Duchenne age 7 to 21
- Able and willing to complete an approximately 75 minute or less MRI exam without sedation or mechanical ventilation
- On a stable drug regimen (if taking prescription drugs) for at least 3 months prior to participation

To participate in this study you may not have:

- Renal insufficiency
- Non-MRI compatible implants (e.g., neurostimulator, pacemaker, implanted cardioverter defibrillator)
- Claustrophobia that prevents an MRI exam
- Known allergy to MRI contrast agents
- Serum potassium level of more than 5.0 mmol/L
- Signs and symptoms of heart failure

What do I have to do if I decide to participate in this study?

This study involves three groups.

- Group 1a boys are healthy volunteers who will complete a cardiac MRI without contrast.
- Group 1b boys are boys with Duchenne who will complete a cardiac MRI with contrast, blood tests, 24 hour heart monitoring, and pulmonary function tests.
- Group 2 boys are boys with Duchenne who will complete a cardiac MRI with contrast, blood tests, 24 hour heart monitoring, and pulmonary function test, then have a second MRI scan with contrast 6 months later.
- Group 3 boys are boys with Duchenne who will complete a cardiac MRI with contrast, blood tests, 24 hour heart monitoring, pulmonary function tests, and genetic tests.

Special considerations:

Some boys in Group 1a and 1b will be asked to have a repeat MRI scan at UCLA or at Children's Hospital of Orange County (CHOC).

How long will this study last?

Depending upon your participation in the study, you will either have one visit, or you will return to the clinic for a second cardiac MRI in 6 months. You will not receive follow up information once your participation in the study has ended.

Where does this study take place?

This study takes place at Ronald Reagan UCLA Medical Center and Children's Hospital of Orange County (CHOC).

How many visits to the study site are necessary?

- Boys in Group 2 will come to the clinic for two visits. The first visit will be at the start of the study and the second visit will take place six months later.

- All other boys (Group 1b and 3) will come to the clinic for a single visit, except for ten boys (Group 1b) who will have cardiac MRI scans repeated at CHOC or UCLA, depending upon where the initial MRI scan was obtained.

Can any visits be done locally?

No, the MRI equipment requires that the cardiac MRI exams take place at UCLA or CHOC hospitals.

Is there any funding to help pay for travel?

Yes, if you are a boy in Group 3 and you live a significant distance from the study site, you may be eligible for reimbursement for travel expenses related to accommodation, transportation, lodging and meals.

Will I get paid for participating in this study?

Yes, please contact the study group for specific information.

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 the latest cardiac MRI methods before they are widely available, and having access to medical specialists that may not be 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 (NCT02834650).

GENETIC MODIFIERS

GENETIC MODIFIERS OF DUCHENNE AND BECKER MUSCULAR DYSTROPHY



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

This study is actively recruiting participants.

What is the goal or purpose of this study?

The Genetic Modifier Study is trying to figure out what genetic changes are causing some people with Duchenne or Becker to be more mildly affected and others to be more severely affected. In other words, what genes are modifying the person's disease. Researchers will try to identify genes and gene variants that may modify the disease process and that will move the community closer to find effective treatments for Duchenne and Becker.

Who is funding this study?

This study is funded by the NIH and PPMD.

Who is eligible to participate in this study?

All males (any age) with Duchenne or Becker are eligible to participate.

What do I have to do if I decide to participate in this study?

Participation in this study requires:

- A brief one page questionnaire that is emailed to you, and returned by email.
- Blood draw or saliva collected near your home, at UCLA or at your local doctors office.
- A brief annual health survey (optional but helpful) by email or phone.

Where does this study take place?

This study takes place at the Center for Duchenne Muscular Dystrophy, David Geffen School of Medicine at UCLA in Los Angeles, CA.

Can any visits be done locally?

Yes. Study participants do not travel to UCLA. Participants complete all study procedures where they live.

Can I request a genetic report or my sequencing data to be sent to me?

- No. This is a research study, no report will be generated and our protocol does not allow us to return their data to the participants.
- On the other hand, if we find something clinically relevant we will discuss the findings with the participants.

Will I get paid for participating in this study?

No.

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), 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 http://cdmd.ucla.edu/pages/genetic_modifiers



ATALUREN

LONG-TERM OUTCOMES OF ATALUREN IN DUCHENNE MUSCULAR DYSTROPHY (041)



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

Recruitment is underway around the world.

What is the goal or purpose of this study?

The goal is to study the long-term safety and efficacy effects of ataluren in patients with Duchenne muscular dystrophy caused by a nonsense mutation (nmDMD).

What is ataluren?

Ataluren is an oral protein restoration therapy designed to enable the formation of a functioning protein in patients with genetic disorders caused by a nonsense mutation. A nonsense mutation is a premature stop signal in the genetic code that interrupts the production of an essential protein.

What is the status of Translarna?

- In August 2014 Translarna received marketing authorization in the European Union for the treatment of nonsense mutation Duchenne in ambulatory patients aged five years and older, representing the first-ever treatment approved for the underlying cause of the disease.
- PTC is expanding commercial access to Translarna across Europe, the Middle East, Latin America, and Asia Pacific.
- PTC has engaged in dialogue with the US Food and Drug Administration regarding a path forward to bring Translarna to patients in the US.

Who is funding this study?

This study is funded by PTC Therapeutics

Who is eligible to participate in this study?

To participate in this study you must be:

- Male with Duchenne, age 5 years
- Positive for a nonsense point mutation in the dystrophin gene
- Using systemic corticosteroids (prednisone/prednisolone or deflazacort) for a minimum of 12 months immediately prior to start of study treatment, with no significant change in dosage or dosing regimen for a minimum of 3 months immediately prior to start of study treatment
- Able to perform 6MWD 150 meters
- Able to perform timed function tests within 30 seconds
- Willing and able to comply with scheduled visits, drug administration plan, study procedures, laboratory tests, and study restrictions.

What do I have to do if I decide to participate in this study?

This study involves clinic visits every 12 weeks during the double-blind period and every 24 weeks during the open-label period.

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 72-week of double-blind, placebo controlled and 72-week open-label period.

Where does this study take place?

This study will be conducted internationally with sites around the world.

How many visits to the study site are necessary?

Approximately 11 visits over the 144 week duration of the study.

Can any visits be done locally?

No, visits to the study site will be required to ensure consistency.

Is there any funding to help pay for travel?

Reimbursement will be determined on a case-by-case basis.

Will I get paid for participating in this study?

No, there is no payment for study participants.

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.

Where can I learn more about this study?

You can learn more about this study at www.ClinicalTrials.gov (NCT#03179631).

ETEPLIRSEN - EXON 51 SKIPPING (STUDY 4658-102)

AN OPEN-LABEL STUDY TO EVALUATE SAFETY, TOLERABILITY, PHARMACOKINETICS, AND EFFICACY OF ETEPLIRSEN IN YOUNG PATIENTS WITH DUCHENNE AMENABLE TO EXON 51 SKIPPING



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research study?

Study 4658-102 is a Phase 2 study that is currently recruiting participants for Cohort 2 (6 - 24 months) in Europe. Cohort 1 (24 - 48 months) is fully enrolled.

What is the purpose of this research?

This study is being conducted to assess the safety, tolerability, pharmacokinetics, and efficacy of eteplirsen, an investigational exon 51 skipping therapy, in young patients with Duchenne who are amenable to exon 51 skipping.

Who is sponsoring this study?

The study is sponsored by Sarepta Therapeutics, Inc.

What are the eligibility requirements of this study?

Eligibility criteria include but are not limited to:

- Male between 6 months to 24 months of age (inclusive)
- Diagnosis of Duchenne with a deletion mutation amenable to exon 51 skipping
- Parent(s) or legal guardian(s) who is willing to provide written informed consent

Key exclusion criteria include:

- Received treatment that might have an effect on muscle strength or function within 12 weeks prior to dosing
- Received previous or current treatment with any experimental treatment
- Clinically significant illness other than Duchenne
- Clinically significant laboratory abnormality
- Any other condition that could interfere with the patient's participation

The Principal Investigator (study doctor) determines whether or not a patient meets the requirements and is eligible for participation in the study.

Where will this study take place?

This study is currently being conducted at five clinical sites in Europe. To view participating study sites, visit www.clinicaltrials.gov (Identifier NCT03218995).

What can be expected by participating in this study?

All patients enrolled in the study receive weekly treatment with the investigational therapy eteplirsen, for at least 48 weeks and up to 96 weeks. Safety is regularly assessed throughout the study via the collection of side effect information, laboratory tests, electrocardiograms (ECGs), echocardiograms (ECHOs), vital signs, and physical examinations. Exploratory efficacy assessments including age- appropriate functional measurements are also being collected.

Will I be paid for allowing my child to participate in this study?

Reasonable costs associated with participation in the study (e.g., travel, parking, meals) are reimbursed. Additional information regarding reimbursement can be provided by a participating study site.

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 NCT03218995). You may also email medinfo@sarepta.com or call +1-617-274-4000.



NATIONWIDE GALGT2 GENE THERAPY

VIRAL GENE TRANSFER FOR GALGT2 (RAAVRH74.MCK.GALGT2) AS A SURROGATE GENE THERAPY



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

This Phase 1/2 trial is enrolling participants by invitation only.

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 GalNAc 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, GalNAc 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 GalNAc 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 GalNAc 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?

Participants must be ambulant boys with Duchenne, age 4 years or older.

What do I have to do if I decide to participate in this study?

This is a dose escalation trial where participants will receive a modified intravascular limb infusion (ILI) procedure that will be used to sequentially deliver viral vector to each lower limb via a major lower limb artery. Safety endpoints will be assessed by changes in hematology, serum chemistry, urinalysis, immunologic response to rAAVrh74 and GALGT2. Efficacy measures will be used as secondary outcomes including a combination of functional 6 minute walk test (6MWT) and direct muscle testing for strength (MVICT) of lower limb muscles.

Where does this study take place?

The clinical trial will take place at Nationwide Children's Hospital (NCH) in Columbus, Ohio, and will include multiple visits to this site.

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 this 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?

Although there is no guarantee of personal benefit by participating in this clinical trial, 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.

Where can I learn more about this study?

You will be able to learn more about this study at ClinicalTrials.gov (NCT03333590).

NATIONWIDE MICRO-DYSTROPHIN GENE DELIVERY

SYSTEMIC GENE DELIVERY CLINICAL TRIAL FOR DUCHENNE



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

This Phase 1/2a clinical trial is actively recruiting participants.

What is the goal or purpose of this study?

The goal of this study is evaluate safety and efficacy in 12 boys with Duchenne following a single infusion of rAAVrh74.MHCK7.micro-dystrophin.

Who is funding this study?

This study is sponsored by Nationwide Children's Hospital and was funded by Parent Project Muscular Dystrophy.

Who is eligible to participate in this study?

To participate in this study you must be a boy with Duchenne, between 3 months and 7 years of age, ambulatory, and have a Duchenne gene mutation between exons 18-58. For a complete list of all the inclusion and exclusion criteria, please visit ClinicalTrials.gov.

What do I have to do if I decide to participate in this study?

This study involves a single infusion in the arm of the rAAVrh74.MHCK7.micro-dystrophin. This will take place at Nationwide Children's Hospital. There will be close follow-up for 3 years, including several muscle biopsies, laboratory tests, physical therapy assessments and physical exams.

Where does this study take place?

This study takes place at Nationwide Children's Hospital in Columbus, Ohio, and Washington University School of Medicine in St. Louis, MO.

Where can I learn more about this study?

You can learn more about this study at www.ClinicalTrials.org (NCT03375164). You can also contact the study coordinator Alana Mahley at Alana.Mahley@nationwidechildrens.org or 614-355-2606.



PF-06939926 (FORMERLY BMB-D001) MINI-DYSTROPHIN GENE THERAPY

A STUDY TO EVALUATE THE SAFETY AND TOLERABILITY OF PF-06939926 GENE THERAPY IN DUCHENNE



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

- This is a Phase 1b, first-in-human/first-in-patient, multi-center, open-label, non-randomized, ascending dose, safety and tolerability study of a single intravenous (IV) infusion of PF-06939926 in ambulatory subjects with Duchenne muscular dystrophy.
- Additional details about this study can be found at www.ClinicalTrials.gov (Identifier: NCT#03362502).

What is PF-06939926 Gene Therapy?

- Duchenne is caused by an absence of dystrophin, a protein that helps keep muscle cells intact. In the absence of dystrophin, muscle cells deteriorate.
- PF-06939926 is an investigational, recombinant adeno-associated virus, serotype 9 (AAV9) carrying a shortened version of the dystrophin gene (mini-dystrophin).
 - An AAV9 capsid was chosen for the delivery of this mini-dystrophin, because of its mild immune responses (safety) and its ability to enter muscle tissue when tested in pre-clinical studies.
- Because the human dystrophin gene is too large to fit in the AAV9 capsid, a mini-dystrophin was developed that may help retain muscle function similar to that of a patient with a more mild disease, like Becker muscular dystrophy.

What is the goal or purpose of this study?

- The goal of this study is to test how safe and tolerable a single infusion of PF-06939926 is in ambulatory boys with Duchenne.
- Other objectives include measurement of dystrophin expression and distribution, and assessments of muscle strength, quality, and function.

Who is funding this study?

Pfizer Inc.

What are the requirements to be considered eligible to participate in this study?

- Diagnosis of Duchenne confirmed by medical history and genetic testing;
- Body weight between 15 and 50 kg;
- Receipt of glucocorticoids for 6 months and a stable daily dose for at least 3 months prior to study entry;
- Ability to walk independently and to rise from floor within seven (7) seconds;
- Ability to tolerate magnetic resonance imaging (MRI) without sedation and with no contraindications to these procedures;
- Ability to tolerate muscle biopsies under anesthesia with no contraindications to these procedures;
- Negative test results at the start of this study for antibodies against AAV9 and for T-cell (immune) response to dystrophin.

What do I have to do if I decide to participate in this study?

- This study is partitioned into 5 basic periods: Screening, Baseline evaluation, Treatment, First year Follow-up, and then Long-term Follow-up with annual visits for 4 years.
- Participants will receive a single IV infusion of PF-06939926 over approximately a 2-hour period.
- Participants will be required to remain in-patient at the clinical research site for at least 24 hours post-infusion.
- Participants will be required to undergo safety tests, including MRI imaging, as well as assessments to measure muscle strength, quality and function. Muscle biopsies will also be collected to assess the presence of mini-dystrophin in the tissue.
 - Some of the assessments required at a study visit may need to be completed across multiple days.

How long will this study last, and will I have access to the drug/treatment once the study has ended?

- The anticipated length of participation in the study is approximately 5 years.
- This study will seek to enroll 12 boys, with two cohorts of 6 boys each. Within each of the cohorts, the dosing interval between the first and second participant will be at least 6 weeks. If no safety concerns are identified 3 weeks after the second participant is infused, then dosing may proceed at 3-week intervals.
- Participants in the study will receive a single IV infusion of PF-06939926 during the study.
- No re-administration of the PF-06939926 mini-dystrophin gene therapy is currently being considered at any time before, during, or after the study.

Where does this study take place?

- This study will be completed across approximately four clinical research sites in the United States only; Duke University Medical Center and University of Utah Hospital are currently active clinical research sites.
- Please check www.ClinicalTrials.gov, Identifier: NCT#03362502, for details regarding the status of these sites.

Will I get paid for participating in this study?

- Participants will not be paid for taking part in this study.
- Reasonable travel and accommodations will be provided or reimbursed.

Why should I consider participating in this study?

- This is the first study of PF-06939926 in humans.
- While no personal benefit can ever be guaranteed by participation in a clinical study, there are other benefits, including allowing participants to play an active role in their own health care (or that of their child), gaining access to new research treatments before they are widely available, having access to medical specialists that are normally not as accessible, and helping others by contributing to a better understanding of Duchenne.

Where can I learn more about this study?

You can learn more about this study at www.ClinicalTrials.gov (NCT#03362502)



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS



SGT-001 MICRO-DYSTROPHIN GENE TRANSFER (IGNITE DMD TRIAL)

IGNITE DMD – MICRODYSTROPHIN GENE TRANSFER STUDY IN ADOLESCENTS AND CHILDREN WITH DUCHENNE



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

In November 2017, Solid Biosciences initiated a Phase 1/2 clinical trial for its lead gene transfer candidate, SGT-001. The clinical trial, called IGNITE DMD, is a randomized, controlled, open-labeled, single-ascending dose study to investigate the safety, tolerability and efficacy of SGT-001 in both ambulatory and non-ambulatory male children and adolescents with Duchenne. At time this overview was printed, IGNITE DMD was on clinical hold by the U.S. Food and Drug Administration (FDA). For more background and information on the status of this clinical trial, please go to www.solidbio.com/media or www.clinicaltrials.gov.

Where is this research being done and who is funding this research?

The first active site for this clinical trial is at the University of Florida. The program is funded by Solid Biosciences.

What is the goal or purpose of this research?

The goal of this research is to evaluate the safety and efficacy of SGT-001, an adeno-associated virus (AAV) mediated gene transfer that contains an engineered version of the dystrophin gene (microdystrophin). The microdystrophin gene in SGT-001 has been designed to produce a functional form of dystrophin protein in skeletal and cardiac muscles. This therapeutic approach is being studied for its potential to treat Duchenne patients, regardless of specific dystrophin mutation.

Who is eligible to participate in this trial?

The IGNITE DMD study is designed to enroll approximately 16 to 32 patients with Duchenne who will be randomly assigned to either an active treatment group or a delayed treatment group. It is planned that non-ambulatory adolescents aged 12 to 17 years will receive SGT-001 initially, then ambulatory children aged four to 11 years will be dosed at a later stage of the clinical trial. More information about eligibility for the clinical trial can be found at www.clinicaltrials.gov, NCT03368742.

Where is this clinical trial taking place?

The University of Florida in Gainesville, FL.

Where can I learn more about this research?

www.solidbio.com
www.ClinicalTrials.gov (NCT03368742)

SRP-4045 AND SRP-4053 - EXONS 45 AND 53 SKIPPING (ESSENCE STUDY)

A 96 WEEK TRIAL FOR INDIVIDUALS WITH DUCHENNE MUSCULAR DYSTROPHY AMENABLE TO EITHER EXON 45 OR EXON 53 SKIPPING



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research study?

This trial is a Phase 3 study.

What is the goal or purpose of this study?

The purpose of this research study is to evaluate the safety and efficacy of SRP-4045 and SRP-4053 in individuals with Duchenne who have a deletion that is amenable to exon 45 or exon 53 skipping.

Who is sponsoring this study?

This study is sponsored by Sarepta Therapeutics.

Who might be eligible to participate in this study?

As of April 2018, ESSENCE has reached targeted enrollment in the US and is no longer enrolling patients in the US. Enrollment is continuing in other countries. The study is enrolling individuals with Duchenne with deletions amenable to exon 45 or exon 53 skipping. Twice as many patients will receive active treatment as will receive placebo (2:1 randomization).

Eligibility criteria include but are not limited to:

- Genotypically confirmed diagnosis of Duchenne, with genetic deletion amenable to exon 45 or exon 53 skipping
- Male, 7 – 13 years of age
- Stable dose of oral corticosteroids for at least 24 weeks before entry
- Stable pulmonary and cardiac function
- Average 6MWT (6-Minute Walk Test) between 300 - 450 meters

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 criteria for the study, and therefore whether or not the patient is eligible to participate.

Where will this study take place?

~60 global sites are planned to participate in ESSENCE. Please see clinicaltrials.gov for updates (Identifier NCT02500381).

Will all individuals enrolled in this trial receive treatment with the investigational therapy?

No, some patients will receive active treatment and some patients will receive placebo (2:1 randomization).

Why is Sarepta including a placebo group of patients in this study?

Comparing patients receiving active drug against patients receiving placebo helps to understand how a drug affects patients and to determine whether the investigational agent is effective and safe.

Why should I consider participation in this study?

While no benefit can be guaranteed from participation in any clinical study, enrolled participants may:

- Have access to clinicians with expertise in the treatment of Duchenne
- Contribute to medical research and what is currently known about the progression of Duchenne
- Contribute to information that may accelerate the development of Duchenne therapies.

Choosing to participate in a clinical study is an important and personal decision. It is recommended that you speak with your doctor, family members, and/or friends about participating in this study.

Will I be paid for participating in this study?

Generally, travel costs associated with participation in the study will be paid up-front, and incidental costs (parking, meals, etc.) are reimbursed. We do not offer a stipend for participation in the ESSENCE trial. Additional information regarding reimbursement and compensation can be provided by a participating study site.

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.

All patients will receive weekly infusions over 96 weeks. Patients in the treated group will receive an infusion of investigational therapy, and patients in the untreated group will receive a placebo infusion. Neither the doctor nor the patient will know whether or not the infusion is placebo or investigational therapy during the 96 weeks. After 96 weeks, an open-label extension will continue for up to 96 weeks. During the open-label extension, all patients will receive investigational agent, and no patients will receive placebo.

Clinical efficacy will be assessed at regularly scheduled study visits, including functional tests such as the six minute walk test. All patients will undergo a muscle biopsy at baseline and a second muscle biopsy at either Week 48 or Week 96.

Safety will be assessed through the collection of side effect information, laboratory tests, electrocardiograms (ECGs), echocardiograms (ECHOs), vital signs, and physical examinations throughout the study.

Blood samples will be taken periodically throughout the study to assess levels of drug in the bloodstream (pharmacokinetics) and to monitor patient safety.

Where can I learn more about this clinical study?

ESSENCE has reached targeted enrollment in the US and is no longer enrolling patients in the US. Enrollment is continuing in other countries. To learn more information about the ESSENCE study, please visit www.clinicaltrials.gov (Identifier NCT02500381) or essencetrial.com. You may also email medinfo@sarepta.com or call +1-888-727-3782.



SRP-5051 - EXON 51 SKIPPING (PPMO STUDY)

A FIRST-IN-HUMAN, OPEN-LABEL, MULTI-CENTER STUDY TO EVALUATE THE SAFETY, TOLERABILITY AND PHARMACOKINETICS OF SINGLE ESCALATING DOSES OF SRP-5051 IN INDIVIDUALS WITH DUCHENNE AMENABLE TO EXON 51 SKIPPING.



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research study?

Study 5051-101 is an open-label Phase 1 study that is currently recruiting participants in the US. Participants who complete the study may be eligible to enroll in an extension study.

What is the goal or purpose of this research?

This Phase 1 study is being conducted to assess the safety, tolerability, and pharmacokinetics of SRP-5051, in participants with Duchenne who are amenable to exon 51 skipping.

Who is sponsoring this study?

The study is sponsored by Sarepta Therapeutics, Inc.

What are the eligibility requirements of this study?

Eligibility criteria include but are not limited to:

- Diagnosis of Duchenne muscular dystrophy
- Deletion mutation in the dystrophin gene amenable to exon 51 skipping
- Male, age 12 years and older
- Stable dose of oral corticosteroids for at least 12 weeks or has not received corticosteroids for at least 12 weeks prior to screening
- Has not had treatment with Exondys 51® or drisapersen within 6 months prior to screening
- No participation in experimental gene therapy studies or any experimental gene therapy for Duchenne at any time
- Stable cardiac and pulmonary function

Note: There are no requirements for ambulation status

Additional criteria apply. Ultimately, the Principal Investigator (study doctor) determines whether or not a participant meets the requirements and is eligible for participation in the study.

Where will this study take place?

This study is currently being conducted at approximately 10 clinical sites in the United States. To view participating study sites, visit www.clinicaltrials.gov (Identifier NCT03375255).

What can be expected by participating in this study?

Participants will receive one dose of SRP-5051 via IV infusion, followed by approximately 12 weeks of safety observation. Participants will stay in the clinic for 2 nights after receiving study drug. During this time, study doctors will collect blood and urine for pharmacokinetic (PK) tests, and perform other safety assessments such as physical exams and vital sign collection. Participants will return to the clinic for 3 additional visits at Week 2, Week 4, and Week 12. These visits will be one-day visits lasting approximately 3-4 hours during the 12 weeks following dosing. Safety, including side effects and laboratory tests, will be monitored on an ongoing basis for all participants. Additionally at the Week 4 visit, a needle muscle biopsy will be collected from a muscle in the leg.

After completion of the study, participants may be provided the option to participate in an open-label extension study with monthly dosing of SRP-5051.

The Principal Investigator (study doctor) and/or the study site contact will review all study requirements with potential participants during the informed consent process both for the single dose study, and for the open-label extension study.

Why should I consider participation in this study?

- While no benefit can be guaranteed from participation in any clinical study, enrolled participants may:
- Have access to clinicians with expertise in the treatment of Duchenne

- Become more familiar with what participation in a clinical study entails
- Contribute to medical research and what is currently known about the progression of Duchenne
- Contribute to information that may accelerate the development of Duchenne therapies.

Choosing to participate in a clinical study is an important and personal decision. It is recommended that you speak with your doctor, family members, and/or friends about participating in this study.

What is SRP-5051?

SRP-5051 is an investigational drug, meaning it has not been approved by the FDA or any other governmental agency. SRP-5051, is a PPMO compound designed to enable skipping of exon 51. PPMO has a cell-penetrating peptide (CPP) attached to a PMO backbone. The mechanism of action is sequence-specific binding to RNA targets. Pre-clinical (animal) studies have demonstrated targeted delivery to skeletal, cardiac, and smooth muscle animal tissues.

Will I or my child be paid for participating in this study?

Generally, costs associated with participation in the study (e.g., travel, parking, meals) are reimbursed within fair market value. If there isn't a site geographically close to you, travel will be provided and arranged by a third party vendor. Additional information regarding reimbursement and travel can be provided by the participating study site.

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 NCT03375255). You may also email medinfo@sarepta.com or call +1-888-727-3782.

WVE-210201 - EXON 51 SKIPPING

A MULTICENTER, DOUBLE-BLIND, PLACEBO-CONTROLLED, PHASE 1 STUDY OF WVE-210201 ADMINISTERED INTRAVENOUSLY TO PATIENTS WITH DUCHENNE (WVE-DMDX51-001)



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

This is a Phase 1 clinical trial that is actively recruiting.

What is the goal or purpose of this study?

The primary objective is to evaluate the safety and tolerability of a single dose of the investigational drug WVE-210201 compared to placebo. WVE-210201 is for patients with Duchenne who are amenable to exon 51 skipping.

Who is funding this study?

This study is funded by Wave Life Sciences.

Who is eligible to participate in this study?

Patients and/or caregivers should discuss the eligibility requirements of the clinical trial with their doctor.

Key eligibility criteria include:

- Genetically-confirmed diagnosis of Duchenne that is amenable to an exon 51 skipping therapy
- Male, aged 5 to 18 years old
- Ambulatory or non-ambulatory

What do I have to do if I decide to participate in this study?

Patients and/or caregivers should discuss potential participation in the study with their doctor.

How long will this study last, and will I have access to the drug/treatment once the study has ended?

- Patients will receive a single dose of the study drug (WVE-210201 or placebo).
- Wave intends to initiate an open-label extension (OLE) study, and patients who complete participation in the Phase 1 clinical trial may be eligible for participation in the OLE.

Where does this study take place?

This study will be conducted at multiple sites in the US, Europe, and Canada. Since this is the first clinical study of WVE-210201, it is important that patients live within close proximity to one of the sites for participation.

Will I get paid for participating in this study?

- There is no compensation for participating in the trial, but reasonable travel expenses incurred due to the study requirements will be reimbursed.
- Patients and/or caregivers should discuss specific questions regarding travel reimbursement in the clinical trial with the study investigator.

Where can I learn more about this study?

You can learn more about this study at www.wavelifesciences.com and www.ClinicalTrials.gov (NCT# NCT03508947). Email clinicaltrials@wavelifesci.com for additional information.



CAP-1002 (HOPE-2 TRIAL)

CAPRICOR'S PHASE 2 TRIAL OF CAP-1002, A CELL-BASED THERAPY FOR DUCHENNE



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What is the HOPE-2 trial?

HOPE-2 is a Phase 2 clinical trial that will evaluate the safety and efficacy of repeat doses of CAP-1002 in boys and young men with Duchenne muscular dystrophy and reduced upper limb function.

What is CAP-1002?

CAP-1002 is a biologic product consisting of cardiosphere-derived cells (CDCs) derived from donated heart muscle. These cells have been shown to be potentially immune-modulatory, anti-inflammatory, anti-fibrotic, and regenerative. In a previous clinical trial, CAP-1002 delivered directly into the arteries of the heart of boys and young men with Duchenne-related heart disease was shown to be generally safe and well tolerated and demonstrated signals of significant and sustained improvement in cardiac and skeletal muscle function compared to participants who received usual care only.

At what stage is this research?

HOPE-2 is actively enrolling participants. Approximately 84 participants will be randomized at 10-15 medical centers in the United States. Visit the HOPE-2 page at [ClinicalTrials.gov](https://clinicaltrials.gov) for a current listing of trial sites.

Who is funding this trial?

This trial is sponsored and funded by Capricor, Inc.

Who is eligible for this trial?

HOPE-2 is one of the very few trials to focus on both individuals with Duchenne who are ambulatory and non-ambulatory and at least 10 years of age or older. Eligible participants must be taking steroids regularly for at least 12 months prior to screening and have reduced upper limb strength.

What happens during this trial?

- Participants in HOPE-2 must first read and sign an informed consent form before completing any trial assessments.
- Participants who qualify will be randomly assigned to receive intravenous (IV) infusions of CAP-1002 or placebo (an inactive substance). The chance of being assigned to the CAP-1002 group is 50/50, like flipping a coin. The treatment assignment, which remains blinded to the participant, participant's family, and study doctor, does not change throughout the course of the trial. Participants will receive an IV infusion of CAP-1002 or placebo every three months for a total of 4 doses.
- HOPE-2 participants will complete medical check-ups, blood tests, quality of life questionnaires, cardiac MRI, the Performance of Upper Limb module, strength testing, pulmonary function tests, and North Star Ambulatory Assessment.

How long will I be in this trial?

- Participation in HOPE-2 will last for about 13 months, including the screening period, requiring about 6-7 visits to the study center.
- If the results from the HOPE-2 trial suggest an appropriate risk/benefit profile of CAP-1002, Capricor, upon the recommendation of the Data Safety Monitoring Board (DSMB), plans to introduce an open-label extension study to offer CAP-1002 to participants who were randomized to placebo and completed the original trial.

Will I get paid to participate in this trial?

- Participants that complete the HOPE-2 trial and complete all required visits will receive \$425. Participants will be paid for only those visits and procedures that were completed.
- Additionally, Capricor will pay for trial-related travel expenses (for example, reasonable airfare, hotel, meals, ground transportation, etc.) for a HOPE-2 participant and one travel companion.

Where can I learn more?

You can find more information about Capricor, CAP-1002, and clinical research in Duchenne at the following sites:

- www.capricor.com
- www.hope2trial.com
- <https://clinicaltrials.gov/ct2/show/NCT03406780>
- www.ParentProjectMD.org

GIVINOSTAT

RANDOMISED, DOUBLE BLIND, PLACEBO CONTROLLED, MULTICENTRE STUDY TO EVALUATE THE EFFICACY AND SAFETY OF GIVINOSTAT IN AMBULANT PATIENTS WITH DUCHENNE



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

This Phase 3 trial is actively recruiting participants in US, Canada, and Europe.

What is the goal or purpose of this study?

The main goal of this phase 3 study is to evaluate the efficacy of Givinostat compared to placebo to slow the disease progression in Duchenne boys. The study will also assess safety and tolerability of the drug.

Has Givinostat been tested before?

- Givinostat has been tested in a number of clinical studies in adults and pediatric populations. Concerning the clinical experience in dystrophinopathies, a Phase 2 clinical trial (DSC/11/2357/43) has been conducted to evaluate the safety and the potential Givinostat as a treatment for Duchenne.
- DSC/11/2357/43 was an open label 2-part, Phase 2 study in ambulatory children aged 7 to less than 11. Clinical results showed that Givinostat significantly counteracted histological disease progression after 12 months of treatment, in particular:
 - It significantly increased muscle fibers size, and it significantly decreased total fibrosis, fatty replacement, necrosis, the mean number of hypercontracted fibers;
 - It significantly increased regenerative fibers with no depletion of the pool of satellite cells.

- The functional tests showed an overall stability after 12 months of treatment in this population, with small changes in some parameters. Overall the drug was safe and well tolerated. The study was completed in December 2017, 18 boys are still on treatment in the long-term safety study (DSC/14/2357/51). The results of this Phase 2 study, 12 months of treatment were published in *Neuromuscular Disorders*, in October 2016 (Bettica et al, 26(10):643-649).

Who is funding this study?

This study is sponsored by Italfarmaco SpA.

Who is eligible to participate in this study?

- Clinicians are recruiting approximately 192 ambulant Duchenne boys. Of these, 128 subjects will be assigned to Givinostat and 64 will be assigned to placebo. To participate in this study you must be:
- Male with Duchenne diagnosis confirmed by genetic testing
 - Age 6 years
 - On stable corticosteroid for at least 6 months
 - Able to perform the 4 stairs climb in not more than 8 seconds
 - Able to rise from floor in less than 10 seconds
 - Additional details regarding inclusion and exclusion criteria will be available on www.ClinicalTrials.gov (NCT#02851797).

What do I have to do if I decide to participate in this study?

This study involves clinic visit every 12 weeks for 18 months, with more frequent visits during the first 3 months. After the screening period, boys will be randomized to receive Givinostat oral suspension or placebo oral suspension to be taken twice a day with food (e.g. during breakfast and during dinner).

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 18 months of double-blind, placebo controlled period. When a subject has concluded 18 months of treatment (Givinostat or Placebo), regardless his walking ability, he will be asked to participate in an open-label long-term safety study with Givinostat, until the drug will be available on the market. All subjects in this long-term safety study (DSC/14/2357/51, NCT#03373968) will take Givinostat.

Where does this study take place?

This study takes place at many sites in US, Canada, and Europe.



GIVINOSTAT (CONTINUED)



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

How many visits to the study site are necessary?

Approximately 15 visits over the 18 months duration of the study. During the screening period, the patient will have 2 visits, and he will be asked to come back to the site for an additional visit to have an MRI scan which could be done in a different hospital. Then, he will come back within 1 month for the first administration of the study treatment. During the first month of treatment the patients will come back to the site weekly, then every 2 weeks for the second months, then at the third months and then every 3 months.

Can any visits be done locally?

Yes, a study nurse is appointed to collect blood sample at your home (if you agree) in some visits.

Is there any funding to help pay for travel?

Yes, there is a reimbursement for the travel.

Will I get paid for participating in this study?

No, you won't. Costs associated with participation in the study (e.g. travel, parking, hotel, meals) are reimbursed. The reimbursement process is different from countries, in some countries Italfarmaco through the hospital will reimburse your travel expenses incurred for the participation of the study, in other countries (e.g. US and Canada) a travel agency can be involved to plan your travel and reimburse the travel expenses.

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.

Where can I learn more about this study?

- You can learn more about givinostat studies at www.italfarmaco.com and www.ClinicalTrials.gov (Epidys trial NCT#02851797, long-term trial NCT#03373968 and Becker trial NCT#03238235).
- You may contact clinical study staff to inquire about clinical study.

MNK-1411 (BRAVE STUDY)

A STUDY TO ASSESS THE EFFICACY AND SAFETY OF MNK-1411 IN BOYS WITH DUCHENNE

What stage is this research?

This trial is actively recruiting participants.

What is the goal or purpose of this study?

The goal of this research is to demonstrate that MNK1411 is safe and effective as a treatment for Duchenne.

Who is funding this study?

This study is funded by Mallinckrodt ARD Inc.

Who is eligible to participate in this study?

To participate in this study you must:

- Be male between 4 and 8 years of age with a documented diagnosis of Duchenne
- Have had no previous systemic treatment with corticosteroids
- Be able to tolerate subcutaneous injections twice weekly
- Have serum potassium within the reference range at screening or upon retesting within the screening period
- Have no symptomatic cardiomyopathy (in the opinion of the investigator)

What do I have to do if I decide to participate in this study?

This study involves 11 clinic visits over a period of up to 56 weeks. Every participant will receive two subcutaneous injections weekly. This trial does have a placebo arm. The primary outcome measure is the 10 meter walk/run test.

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 about 56 weeks, including 4 weeks screening, 24 weeks of double blind treatment and an optional 24 weeks of open label treatment. Long term access to the study drug after study closure is not available at this stage of the study.

Where does this study take place?

This study takes place at up to 55 study sites in 15 countries including the USA, EU, and Middle East.

How many visits to the study site are necessary?

11 clinic visits

Can any visits be done locally?

No, visits must be done at a participating investigator site.

Is there any funding to help pay for travel?

Yes, all study visit related travel expenses will be provided to study participants and their caregivers.

Will I get paid for participating in this study?

No, but all travel expenses are paid by the 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 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.
- Participation this study will also help to determine whether MNK1411 is a safe and effective therapy for boys with Duchenne.

Where can I learn more about this study?

You can learn more about this study at www.ClinicalTrials.gov (NCT#03400852).



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS



VAMOROLONE (VBP15)

A POTENTIAL STEROID ALTERNATIVE CUSTOMIZED FOR DUCHENNE



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

[There are multiple trials going on in parallel:](#)

- A Phase 2a clinical trial (VBP15-002) of 48 Duchenne boys (4 to <7 years, steroid naïve) is completed. This is a 2 week treatment, with a focus on safety and biomarkers. Data is currently under interpretation.
- A Phase 2a extension clinical trial (VBP15-003) of the same 48 boys is fully enrolled and ongoing. This is 24 weeks of treatment. Last patient last visit is Q2 2018.
- A Long Term Extension study (VBP15-LTE) is open for the same 48 boys, enabling dose escalation. This is a 2 year treatment period, and is ongoing.
- A Phase 2b clinical trial (VBP15-004) for boys with Duchenne muscular dystrophy is currently recruiting. This is a 48 week study of about 120 boys with Duchenne (4 to <7 years, steroid naïve). This study plans to recruit at about 30 sites in the following countries (US, CANADA, UNITED KINGDOM, ISRAEL, AUSTRALIA, SWEDEN, GERMANY, ITALY, NETHERLANDS, CZECH REPUBLIC, BELGIUM). The first 24 weeks (~6 months) will include four arms (low dose vamorolone, high dose vamorolone, prednisone, and placebo; 1:1:1:1), and for the second 24 weeks all Duchenne boys will transition to vamorolone (high dose or low dose).

What is the goal or purpose of this research study?

- The goal of this research is to see if an investigational drug called vamorolone is effective (improves or stabilizes muscle strength and function) and has fewer side effects than steroids in boys with Duchenne.
- Vamorolone is hoped to retain the beneficial anti-inflammatory and muscle strengthening aspects of corticosteroids (prednisone, deflazacort), while decreasing some of the undesirable side effects (bone fragility, stunted growth, insulin resistance, mood changes, delay of puberty and others).
- Vamorolone has additional activities such as a mineralocorticoid receptor antagonism and membrane stabilization that may increase benefit to boys with Duchenne.

Who is funding this research study?

- Many non-profit foundations and governments have funded the vamorolone program. A complete description of funders can be found at <http://www.reveragen.com/about-us/partnerships/>
- The National Institute of Neurological Disorders and Stroke (1R44NS095423-01) and the National Institute of Arthritis and Musculoskeletal and Skin Diseases (1U34AR068616-01) have provided funding for VBP15-002 and VBP15-003 trials.

- The European Union's Horizon 2020 research and innovation program under grant agreement No 667078 has provided funding for VBP15-004 (via Newcastle University, UK).
- To date, ReveraGen has worked through public-private partnerships to develop other aspects of the vamorolone development program:
- PPMD (\$750,000) and Foundation to Eradicate Duchenne (\$250,000) are co-funding the chronic toxicology studies
- The \$2.1M Phase 1 trial was 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 research study?

- Boys with Duchenne who have never taken steroids and who are ages 4, 5, or 6 at study entry.
- Additional eligibility criteria may apply and can be discussed with a study team member.

How long will this research study last, and will I have access to the investigational drug once the study has ended?

- The newly recruiting VBP15-004 study will last approximately one year.
- After completion of the study, there may be an extension study to allow continued access to vamorolone (long term extension).

Where does this research study take place?

- The newly recruiting VBP15-004 study will take place at sites located in the USA, Canada, Europe, Israel, and Australia.
- Detailed site information can be found on clinicaltrials.gov (NCT03439670).

How many visits to the study site are necessary?

Approximately 15 visits over about 12 months.

Can any visits be done locally?

Visits must be done at one of the participating sites.

Will I get paid for participating in this research study?

No, but travel, stay, and meal expenses are paid for by the study.

Why should I consider participating in this research study?

- Participation will help determine whether vamorolone is an effective therapy for Duchenne and how its effectiveness and side effects compare to prednisone.
- Showing effectiveness for Duchenne could have implications for many other disorders where steroids are used.
- The vamorolone program includes many innovations in clinical trial design that, if successful, will speed other drug development programs, including blood biomarkers and mobile health outcomes.

Where can I learn more about this research study?

- Information will be posted on the ReveraGen website, as well as www.clinicaltrials.gov.
- Contact Andrea Smith at asmith@trinds.com



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS



IDEBENONE (SIDEROS TRIAL)

PHASE 3 STUDY ASSESSING THE EFFICACY, SAFETY AND TOLERABILITY OF THE INVESTIGATIONAL DRUG IDEBENONE IN PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY RECEIVING GLUCOCORTICOID STEROIDS (SIDEROS)



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

The SIDEROS clinical trial is a Phase 3 clinical trial which began recruiting participants in September 2016.

What is the goal or purpose of this study?

The primary objective of this study is to assess the efficacy and safety of the investigational drug idebenone in slowing the loss of pulmonary function in boys with Duchenne receiving glucocorticoid steroids.

What is idebenone and how does it work?

Idebenone is thought to work in Duchenne by increasing the energy output of the cells' mitochondria – the parts ("factories") of the cell that generate all of a cells' energy. Mitochondria produce the energy necessary for the cell functioning through a process called "cellular respiration" which requires oxygen and produces energy. During cellular respiration, some toxic forms of oxygen (called oxygen free radicals) can be produced. These free radicals must be neutralized by other substances to avoid cellular damage. Idebenone is expected to act as a neutralizer of these toxic forms of oxygen. Thus, idebenone is expected to have an antioxidant effect, and consequently prevent cellular damage. Idebenone is optimized to dissolve in water and lipids and able to cross the mitochondrial membrane.

Who is funding this study?

This study is funded by Santhera Pharmaceuticals.

Who may be eligible to participate in this study?

- To participate in this study, you must be male with Duchenne, at least 10 years old and have stable use of glucocorticoid steroids continuously for at least 12 months with no dose adjustments for the past six months, with the exception of weight changes. This includes prednisone or deflazacort, and any dosing regimens. Changes between prednisone and deflazacort are allowed if dose is comparable.
- Participants must have a baseline Forced Vital Capacity between 35%-80% and be able to provide reliable and reproducible pulmonary function testing.

What do I have to do if I decide to participate in this study?

- This study involves a screening assessment, and boys will be randomized to receive either 900 mg/ daily of idebenone (2 tablets, 3 times daily with food) or placebo for 18 months. Parents will be asked to keep a daily diary of medication intake.
- Participants will complete safety assessments, pulse-oximetry, and pulmonary function tests at study site visits every three months, as well as be given a hand-held spirometry device to complete weekly pulmonary function assessments at home.
- Funding will be provided to assist families with costs to participate in the study.

How long will this study last, and will I have access to the drug/treatment once the study has ended?

- Patients' active participation is up to 21-22 months (88-92 weeks) including screening and follow-up visits, and 18 months receiving study medication or placebo.
- Patients completing the study through week 78 will be eligible to participate in an open-label extension study and will continue to receive medication until the trial is terminated or idebenone is approved.

Where can I learn more about this study?

Please visit www.siderosdmd.com for updates on this trial. Information about the trial is also available at www.DuchenneConnect.org and www.clinicaltrials.gov.

RG6206 ANTI-MYOSTATIN ADNECTIN

ROCHE INVESTIGATIONAL MOLECULE RG6206 AN ANTI-MYOSTATIN ADNECTIN



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

RG6206 is an investigational molecule in a Phase 3 clinical study (NCT03039686) in ambulatory boys with Duchenne muscular dystrophy that is currently actively recruiting.

What is the purpose of this research?

The main goal of the Phase 3 study (NCT03039686) is to determine whether this investigational molecule RG6206 may be a safe and effective treatment for ambulatory boys with Duchenne.

What is RG6206?

- RG6206 is an investigational molecule that is designed to bind to a protein called myostatin and limit its function.
- Myostatin is a naturally occurring protein that is made mostly in skeletal muscle cells. These are muscles like the ones found in the arms and legs. Everyone has some myostatin and its natural function usually is to stop muscles growing too much.
- Limiting myostatin has been shown in some studies in people to increase muscle size.

Where is this research being carried out?

This is a global study. Sites are already open in the United States and Japan. Additional sites are open or planned to be opened in Canada, Argentina, UK, Belgium, France, Spain, Sweden, Netherlands, Germany, Italy, and Australia in 2018. In the United States there are multiple study sites open and/or planned in the following states: CA, FL, GA, IL, KS, MD, MO, NY, OH, PA, Washington DC, NV, AZ, and Canada.

Who would be eligible to participate in this clinical study?

- Your son may be eligible to participate if he has Duchenne confirmed by medical history with genetic testing and is:
- 6 to 11 years of age (inclusive) and weight at least 15kg (33 lbs)
- Able to walk without assistance and climb stairs on his own, 4 stairs in 8 seconds or less
- Receiving corticosteroids

Note that all Duchenne mutations are eligible for this study and full eligibility requirements will be assessed at a participating study site.

How many boys will be enrolled in the study?

Approximately 159 boys with Duchenne will be enrolled in this study.

How long will the study be?

The study has two parts:

- PART 1: consists of an initial 48 week randomized placebo-controlled phase during which all boys will receive either RG6206 or placebo (2:1 ratio, meaning twice as many participants will receive RG6206 versus placebo)
- PART 2: consists of a 48-week open-label phase during which all boys will receive RG6206

How is the study medication given?

RG6206 or placebo is administered once weekly by an injection under the skin (subcutaneous injection) in to the tummy (abdomen). The three first doses must be done at the study site, while the remaining administrations may be done at home.

Will I have access to the study drug once the study has ended?

After completion of the 48 week open-label phase, depending on the results of the study, your child may be given the option to enter a separate extension study to continue to receive RG6206.

Is there any funding to help pay for travel?

Eligible participants may receive, at no cost, transportation services with a travel concierge service to cover the costs of study-required travel, according to local guideline.

Where can I learn more about this research?

- Visit ClinicalTrials.gov (search for NCT03039686)
- Visit www.roche-duchenne-clinicaltrials.com
- Please contact your treating physician to discuss the clinical study



EDALONEXENT (FORMERLY CAT-1004)

CATABASIS' PHASE 3 TRIAL WITH EDALONEXENT



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

This will be a global Phase 3 trial in Duchenne muscular dystrophy to evaluate the efficacy and safety of edalonexent. We are not yet recruiting for this Phase 3 trial.

Who is eligible to participate in the trial?

The trial is anticipated to enroll approximately 125 patients with the following criteria:

- Boys with a diagnosis of Duchenne (any confirmed mutation) between the ages 4 and 7 years
- Ability to complete the timed function tests
- No corticosteroid use within the past 6 months
- The key inclusion and exclusion criteria for this clinical trial will be available on www.clinicaltrials.gov.

What do I have to do if I decide to participate in the trial?

- The trial is planned to be a randomized, double-blind and placebo-controlled study with 2 boys receiving edalonexent for every 1 boy receiving placebo. Edalonexent capsules will be taken orally, three times per day.
- Patients will have clinic visits every 3 months to assess North Star Ambulatory Assessment, time to stand, 4-stair climb, 10-meter walk/run, muscle strength, safety measures, assessments of growth, cardiac and bone health. Neither muscle biopsies nor MRIs will be required for this study.
- After 12 months in the study, all boys are expected to receive edalonexent in an open-label extension.

Is there any funding to help pay for travel?

Yes, Catabasis will fund travel-related expenses. Information on travel support will be available through the clinical study coordinator at the clinic site and through our patient travel coordinating agency, Colpitts.

What are the results from the Phase 2 MoveDMD® trial?

- MoveDMD is a Phase 2 clinical trial in boys with Duchenne Muscular Dystrophy with an ongoing open-label extension.
- In the Phase 2 MoveDMD trial and open-label extension, edalonexent substantially slowed Duchenne disease progression through more than one year of treatment compared to an off-treatment control period. Consistent improvements in assessments of muscle function and MRI were observed through 48 weeks of oral 100 mg/kg/day edalonexent treatment compared to the rates of change in the pre-specified control period for boys prior to receiving edalonexent treatment. Additionally, supportive changes in non-effort based measures of muscle health were seen, with significant longer-term reductions in muscle enzymes and C-reactive protein (CRP), supporting the durability of edalonexent treatment effects.
- Edalonexent has been well-tolerated with no safety signals observed throughout the trial.

Where is this research being done and who is funding this research?

- This research is being done by Catabasis, a clinical-stage biopharmaceutical company with a mission to bring hope and life-changing therapies to patients and their families. Catabasis is focused on the treatment of rare diseases, including Duchenne.

What is the goal or purpose of this research?

Edalonexent is an oral small molecule investigational drug candidate that inhibits NF-kB. Dystrophin is a protein that keeps muscles healthy by maintaining the structure of muscle cells. In boys with Duchenne, the absence of dystrophin combined with mechanical stress in muscle leads to activation of NF-kB, which in turn drives muscle damage and prevents muscle regeneration. By inhibiting NF-kB, we believe that edalonexent may reduce muscle damage and increase muscle regeneration in boys with Duchenne.

Where does the clinical trial take place?

We expect to have Phase 3 clinical trial sites in North America, Europe, Asia, and Australia. The list of recruiting sites will be shared closer to the start of the study.

Where can I learn more about this research?

- You can learn more at www.catabasis.com.
- You can contact Catabasis directly with any questions at DMDTrials@catabasis.com.
- Please check www.ParentProjectMD.org for updates to this FAQ sheet.

EZUTROMID (PHASEOUT DMD STUDY)

A 48 WEEK PHASE 2 CLINICAL STUDY TO ASSESS THE ACTIVITY AND SAFETY OF UTROPHIN MODULATION WITH EZUTROMID IN AMBULATORY BOYS WITH DUCHENNE MUSCULAR DYSTROPHY



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

Ezutromid is in a Phase 2 proof-of-concept clinical trial, called PhaseOut DMD. Enrollment is now complete and top-line results from the full trial are expected in the third quarter of 2018.

What is utrophin and utrophin modulation?

- Utrophin is a naturally occurring protein that is functionally and structurally similar to dystrophin. Utrophin is produced naturally during the early stages of muscle fiber development but is turned off in maturing muscle fibers, as dystrophin steadily increases to perform the same functional role. When a muscle fiber is damaged, utrophin is produced during the repair process. Summit's utrophin modulation approach aims to use small molecule drugs to maintain the production of utrophin to compensate for the absence of dystrophin in patients with Duchenne. This approach has the potential to treat all patients with Duchenne.
- Ezutromid is an orally administered utrophin modulator that has the potential to benefit all patients regardless of their dystrophin mutation. This is Summit's most advanced utrophin modulator, and the Company is also advancing an earlier-stage pipeline of future generation utrophin modulators. Summit believes that the approach of utrophin modulation could be complementary to other therapeutic approaches currently in development for Duchenne or that are approved.

What is the goal or purpose of this study?

- PhaseOut DMD is a 48-week open label trial that aims to provide proof of concept for ezutromid and utrophin modulation through measurements of magnetic resonance parameters of the leg and through measuring utrophin protein and muscle fiber regeneration by analysis of muscle biopsies. Exploratory endpoints include the 6-minute walk distance, the North Star Ambulatory Assessment and patient reported outcomes.
- At the end of the 48-week period, patients participating in the trial have the opportunity to be transitioned into an open-label extension phase that lasts until either ezutromid is approved or its development is discontinued.

What were the inclusion criteria for the PhaseOut DMD trial?

Some of the key inclusion criteria for being able to participate in this trial included:

- age ranging from 5th to 10th birthday with confirmation of Duchenne diagnosis through genetic testing or muscle biopsy,
- ability to walk a prespecified distance that is at least 300 meters unassisted in a 6-minute walk test, and
- being on a stable regimen of corticosteroids for at least 6 months prior to enrollment.

When will data from PhaseOut be reported?

- Summit reported interim 24-week data from the trial in the first quarter of 2018. After 24 weeks of treatment, ezutromid maintained utrophin expression, significantly and meaningfully reduced muscle damage and significantly reduced muscle inflammation.
- Summit expects to report top-line data from the full trial in the third quarter of 2018.

What are the next steps for the development of ezutromid?

Summit is advancing preparatory activities for a placebo-controlled clinical trial of ezutromid and a potential regulatory filing based on the full trial results, should they be positive.

How long until ezutromid could be available on the market?

- Ezutromid will not be available on the market in the United States until Summit files an application for marketing approval of ezutromid for Duchenne with the U.S. Food and Drug Administration (FDA) and the FDA approves the application. There are two potential paths to seeking regulatory approval in the United States for ezutromid if the 48-week results from PhaseOut DMD are positive. One option is to run a placebo-controlled clinical trial and apply for FDA approval based on the results from that trial. Another option is to file for accelerated approval with the FDA based on the full 48-week results from PhaseOut DMD, which could potentially allow for earlier market access to ezutromid. A placebo-controlled clinical trial could then serve as a confirmatory trial for full approval. We plan to update the community on our plans after the results from the full PhaseOut DMD trial.

Where can I learn more about PhaseOut DMD and other future clinical trials?

- Sign up to receive news on our clinical trials and utrophin program at www.utrophintrials.com
- More information on PhaseOut DMD is available at: <https://clinicaltrials.gov/ct2/show/NCT02858362>



NS-065/NCNP-01 - EXON 53 SKIPPING

SAFETY, DOSE FINDING, AND EFFICACY STUDIES OF NS-065/NCNP-01 IN BOYS WITH DUCHENNE



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

- Study NS-065/NCNP-01-201 which is a Phase 2 study completed enrollment and dosing at clinical sites in North America (United States and Canada).
- All participants elected to enroll into Study NS-065/NCNP-01-202 which is an extension study of NS-065/NCNP-01-201. All participants continue to be dosed.

What is NS-065/NCNP-01

NS-065/NCNP-01 is a new drug designed to skip exon 53 in the dystrophin gene.

What is the goal or purpose of this study?

- Study NS-065/NCNP-01-201 was conducted to evaluate the safety and tolerability of a low dose and high dose of NS-065/NCNP-01 injection delivered as a weekly intravenous infusion in patients with Duchenne who are amenable to exon 53 skipping. Additional objectives of the study assess its effect on induction of dystrophin, muscle function and strength, pharmacokinetics and pharmacodynamics.
- Study NS-065/NCNP-01-202 is being conducted to continue evaluating the safety and tolerability of low and high doses of NS-065/NCNP-01 Injection over an additional 72-week period (total 96 weeks of treatment in Study NS-065/NCNP-01-201 and 202). Additional objectives of the study will assess its effect on muscle function and strength.

Who is funding this study?

The study is sponsored by NS Pharma, Inc.

Who was eligible to participate in this study?

To be in Study 201 boys must have a confirmation of Duchenne by genetic test with a mutation amenable to skipping exon 53 and be between the ages of 4 and less than 10 years old. They must be able to walk independently and able to complete standard timed tests. Boys must have been on a stable dose of steroid for at least 3 months.

Did all boys enrolled in this trial receive treatment with the investigational therapy?

- Study NS-065/NCNP-01-201 was a 2-period, randomized, placebo-controlled, dose finding study for 24 weeks.
- Period 1 was a 4-week, blinded, placebo-controlled phase and Period 2 was a 20-week, open-label phase where all participants receive the study medication.
- Study NS-065/NCNP-01-202 is an open-label, extension study of NS-065/NCNP-01-201 for an additional 72 weeks.
- All participants who completed Study 201 have enrolled into Study 202 and all participants continue to receive the investigational therapy, NS-065/NCNP-01.

How long will this study last, and will I have access to the drug/treatment once the study has ended?

- Study NS-065/NCNP-01-201 lasted 24 weeks.
- Study NS-065-NCNP-01-202 will last an additional 72-weeks.

Why did Study NS-065/NCNP-01-201 include a placebo group of participants?

In Study 201 the initial 4-week placebo arm served as a safety control arm. The safety information collected during the 4-week placebo period will be compared to those participants who received NS-065/NCNP-01.

Where do the studies take place?

This study is conducted at selected participating centers of the Cooperative International Neuromuscular Research Group (CINRG) in the North America. For more information on site locations, please visit www.clinicaltrials.gov. Identifier NCT02740972 and NCT03167255.

Where can I learn more about this study?

You can learn more about this study at www.ClinicalTrials.gov (NCT# NCT02740972 and NCT03167255).

DOMAGROZUMAB (FORMERLY PF-06252616)

DEVELOPMENT OF A MYOSTATIN INHIBITOR (DOMAGROZUMAB) AS A POTENTIAL THERAPY FOR DUCHENNE



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

- This is a Phase 2 randomized, double-blind, placebo-controlled, multiple ascending dose study to evaluate the safety, efficacy, pharmacokinetics and pharmacodynamics of Domagrozumab (PF-06252616) administered to ambulatory boys diagnosed with Duchenne muscular dystrophy. Enrollment of participants completed in May 2017.
- Additional details about this study can be found at www.ClinicalTrials.gov (NCT#02310763) or www.dmdmyostatintrial.com.

What is Domagrozumab?

Domagrozumab is an investigational drug (not approved for commercial use) designed to block the activity of myostatin. Myostatin is a protein that acts in the body to prevent muscle from growing too large. This drug is being tested to see if blocking myostatin will help to increase muscle strength and function in boys with Duchenne.

What were the inclusion (enrollment) criteria for this trial?

The main inclusion criteria were that participants must be ambulatory boys with Duchenne who are 6 to <16 years of age.

What happens during the course of the study?

- Participants receive monthly IV infused doses for up to 2 years at their study site.
- Based on the design of the study, all participants receive the investigational drug during half or all of the duration of the study.
- Participants undergo safety evaluations at regular intervals (including monthly clinical exams and blood tests).
- Participants undergo functional evaluations (including the 4 stair climb and 6 minute walk test every 2 months and MRI scans to evaluate the leg muscles).

Where is the trial taking place?

Study sites are in the United States, Canada, Italy, United Kingdom, Poland, Bulgaria, Australia, and Japan.

What happens next?

- Participants completing the Phase 2 study of PF-06252616 may be eligible to enroll in the open-label extension study of Domagrozumab (PF-06252616). Information about the open-label extension study may be found at www.ClinicalTrials.gov (NCT#02907619).
- The 1 year primary analysis will be conducted in the summer of 2018.

- The Primary Analysis is a pre-specified analysis that will evaluate the differences in the treatment groups (domagrozumab compared to placebo) to complete a 4 stair climb test following 1 year on the study. This analysis is able to be conducted because all enrolled patients will have completed at least 1 year on the study (period 1). At the time of the primary analysis, patients will still be ongoing in the second year of the study (period 2). The second year provides additional important information about long-term efficacy and safety. The complete study data analysis is on-schedule for the summer of 2019.

Where can I learn more about this study?

The current study is posted on www.ClinicalTrials.gov (NCT#02310763)

Is Pfizer pursuing other possible treatments for Duchenne?

- Pfizer is conducting a phase 1b, first-in-human/first-in-patient, multi-center, open-label, non-randomized, ascending dose, safety and tolerability study of a single intravenous infusion of PF-06939926 in ambulatory subjects with Duchenne muscular dystrophy (Duchenne).
- Pfizer is an active participant in advocacy/academic/industry consortia.



PAMREVLUMAB (FORMERLY FG-3019)

AN INVESTIGATIONAL THERAPEUTIC MONOCLONAL ANTIBODY TO INHIBIT THE ACTIVITY OF CONNECTIVE TISSUE GROWTH FACTOR (CTGF)



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

This trial is active, but no longer recruiting.

What is the goal or purpose of this study?

– This is a Phase 2, open-label, single arm trial of pamrevlumab (FG-3019) to estimate its efficacy in non-ambulatory patients with Duchenne muscular dystrophy. The rationale for using pamrevlumab in patients with Duchenne is based on data that show that CTGF promotes muscle fibrosis and reduces the ability of damaged muscle cells to repair. Pamrevlumab binds to CTGF and may prevent this cascade. In a preclinical study using an mdx mouse model, pamrevlumab reduced muscle fibrosis and improved muscle function. These data suggest that treatment with pamrevlumab may slow the loss of muscle function. The use of pamrevlumab is investigational in this study. Pamrevlumab is not an FDA-approved drug for any indication and its efficacy and safety have not been demonstrated yet.

Who is sponsoring this study?

This study is sponsored by FibroGen, Inc., a clinical stage biopharmaceutical company (www.fibrogen.com).

Who is eligible to participate in this study?

To participate in this study you must be age 12 years or older with Duchenne, and non-ambulatory (wheelchair dependent). Please see ClinicalTrials.gov (NCT 02606136) for additional inclusion and exclusion criteria.

What do I have to do if I decide to participate in this study?

Each eligible participant will receive pamrevlumab every two weeks by intravenous infusion for up to 3 years. All participants will be closely monitored for safety. Efficacy assessments will be performed routinely; pulmonary and muscle function tests approximately every 3 months, MRIs approximately once a year. This study also includes collection of quality of life data in a questionnaire.

How long will this study last?

The anticipated length of the study is approximately 3 years.

Where does this study take place?

This study is ongoing at several sites across the United States: Cincinnati Children's in Cincinnati, OH; Washington University in St. Louis, MO; UCSF Benioff Children's Hospital in San Francisco, CA; Children's Hospital Colorado in Aurora, CO; Rare Disease Research in Atlanta, GA; Children's Hospital of Philadelphia in Philadelphia, PA; Children's Hospital Boston in Boston, MA; Shriners' Hospital for Children in Portland, OR; University of California, Los Angeles in Los Angeles, CA; Children's Medical Center Dallas in Dallas, TX.

Where can I learn more about this study?

- You can learn more about this study at www.fibrogen.com and www.clinicaltrials.gov (NCT02606136).
- Please check www.DuchenneConnect.org for updates to this FAQ sheet.

NATIONWIDE EXON 2 SKIPPING FOR DUPLICATION 2

EXON 2 SKIPPING THERAPY TO INDUCE INTERNAL RIBOSOMAL ENTRY SITE (IRES) ACTIVATION IN DUCHENNE PATIENTS WITH EXON 2 DUPLICATIONS



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

This research is pre-clinical, meaning it has not advanced to clinical trials involving people yet. The first-in-human trial is anticipated to begin in the third quarter of 2018.

What steps need to be completed before moving into a clinical trial?

Virus manufacturing for a first-in-human trial is complete. Pre-clinical, IND-enabling toxicity studies are essentially complete. IND submission is anticipated in summer 2018.

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

4 months.

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, which was discovered in patients with very mild Becker who were ambulant into their seventh decade.

Who is funding this study?

The preclinical development of this study has been funded by CureDuchenne and the Beauhawks Foundation. Manufacturing of the clinical viral vector lot has been funded by the Beauhawks Foundation. Discussions for funding for the clinical trial are underway.

Who will be eligible to participate in this study?

The final enrollment criteria are not set, but we anticipate enrolling pre-ambulant (> age 6 months) and ambulant 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. A pre-enrollment check for antibodies to the virus will be performed.
- Delivery of the vector will likely require sedation and delivery to both legs via a catheter.
- Muscle biopsies will be required at several time points: before and 12 weeks after vector delivery (open biopsies), and at 6 and 12 months after vector delivery (needle biopsies).

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, treatments such as immunosuppression or 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.ClinicalTrials.gov and www.nationwidechildrens.org/center-for-gene-therapy.



WAVE EXON 53 SKIPPING

WAVE'S STEREOPURE ANTISENSE OLIGONUCLEOTIDE (ASO) FOR EXON 53 SKIPPING IN DUCHENNE



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

This research is in the preclinical phase, meaning it has not advanced to clinical trials involving people yet.

Where is this research being done and who is funding this research?

Wave Life Sciences is conducting discovery research in Cambridge, MA, and in collaboration with Dr. Matthew Wood at the University of Oxford in the UK. This research is being funded by Wave Life Sciences.

What is the goal or purpose of this research?

The goal of this early research is to work toward developing a medicine for people with Duchenne muscular dystrophy amenable to exon 53 skipping.

What steps need to be completed before moving into a clinical trial?

Required research studies must be completed and regulatory applications filed and approved prior to initiating a clinical trial.

Where would a clinical trial take place?

The company expects to run a global clinical trial and hopes to have clinical trial sites in the United States. Sites have not been selected at this time.

Where can I learn more about this research?

Please visit www.wavelifesciences.com or email medicalinformation@wavelifesci.com for additional information.

CARMESEAL-MD™

POLOXAMER 188 NF



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

- In the US: Our research is at the clinical stage, our product has an open IND and approval for an initial clinical trial.
- In Europe: While we are developing Carmeseal-MD in the US under FDA supervision, it is already available to patients in Europe, Argentina, and New Zealand as an unlicensed medicinal product ("Special") when prescribed by a specialist.

Where is this research being done and who is funding this research?

Phrixus studies were supported financially by the SMARTT (Science Moving towards Research Translation and Therapy) program at NHLBI. 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 as well as an improvement in upper body strength. 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 and we are starting to collect data from our first patients. Specifically, we have not observed any adverse events, however, data from biomarkers is favorable (such as a 60% reduction in CPK).

What steps need to be completed before moving into a clinical trial?

We have raised approximately \$1 million to conduct P-004, a 10 patient open label trial at Cincinnati Children's Hospital with Dr. John Lynn Jefferies as Principal Investigator. All regulatory activities have been completed.

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

We expect the trial at CCHMC to begin in May or June 2018.

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 Children's Hospital (John Jefferies, supported our pre-IND meeting), and the University of Tennessee Health Science Center (Jeff Towbin, also supported our pre-IND meeting). We are also in discussions with Cedar-Sinai in Los Angeles.

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. 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 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.



NATIONWIDE CARRIER STUDY

CHARACTERIZATION OF CLINICAL SKELETAL AND CARDIAC IMPAIRMENT IN CARRIERS OF DUCHENNE AND BECKER



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



FEMALE CARRIERS

What stage is this research?

This is an actively recruiting longitudinal observational clinical study.

What is the goal or purpose of this study?

To study the neuromuscular, cardiovascular and psychological/cognitive impact of being a genetic carrier of Duchenne or Becker muscular dystrophy.

Who is the sponsor of this study?

This study is sponsored by Parent Project Muscular Dystrophy.

What are the inclusion (enrollment) criteria for this study?

- 18 years of age and older
- Able to complete testing in English
- A genetically confirmed mutation in the Duchenne gene with an affected child OR
- Duchenne and Becker mothers with NO somatic mutation in the Duchenne gene (females who are not carriers of Duchenne/Becker but have an affected child)

What do I have to do if I decide to participate in this study?

The study consists of three visits which take place on an annual basis. Each visit spans the course of one to two days and includes a number of procedures that address the neuromuscular, cardiovascular, psychological, and cognitive impact that you may experience. If you are interested in taking part in this study, please contact the study coordinator at Nationwide Children's Hospital to find out more about what's involved and decide if you would like to schedule a study visit.

Where does the study take place?

This study takes place at Nationwide Children's Hospital in Columbus, Ohio.

Are travel expenses to the study site reimbursed?

The study is currently able to provide up to \$500 in reimbursement for travel and lodging expenses. Please contact the study coordinator for additional information about reimbursement.

Why should I consider participating in this study?

While a primary benefit of the study is in understanding the impact of being a genetic carrier of Duchenne or Becker, the study also provides and pays for several tests such as an MRI and Cardiovascular Stress test which may be of value to you or your physician.

Where can I learn more about this study?

Contact study coordinator:
Eric Camino
614-722-2715
Eric.camino@nationwidechildrens.org
www.clinicaltrials.gov (NCT02972580)

CLINICAL TRIAL GLOSSARY



BELOW IS A LIST OF SOME HELPFUL TERMS YOU MAY HEAR AS YOU BEGIN TO EXPLORE THE CLINICAL TRIAL PROCESS

ACRONYMS

Here are a few acronyms you may hear a lot:

NIH

National Institutes of Health

CDC

Centers for Disease Control

FDA

Food and Drug Administration

HHS

Department of Health and Human Services

NINDS

National Institute of Neurological Disorders and Stroke

NIAMS

National Institute of Arthritis and Musculoskeletal and Skin Diseases

NCATS

National Center for Advancing Translational Sciences

BIOMARKER

Typically some type of lab test or value that can be objectively measured and evaluated. The biomarker can serve as an indicator of the normal biological process, disease process, or pharmacologic responses to a therapy/treatment. A biomarker in Duchenne could be dystrophin in a muscle biopsy or an MRI of a muscle. Right now there are no approved or "validated" biomarkers for Duchenne, but companies are using them in trials to add to the clinical evidence and hopefully have them validated over time.

BLINDING OF STUDY

At least one or more parties involved in the trial, such as the researcher or the people in the study, do not know which people have been assigned which treatment group.

CARRIER

In Duchenne, women who carry one copy of the genetic change that causes Duchenne or Becker. The woman also has another copy of the Dystrophin gene that does not contain the genetic change. Carriers have a 50% chance of passing the mutation on to each pregnancy.

CLINICAL ENDPOINT

Measure of events or outcomes such as symptoms, functional abilities, laboratory tests, etc., that help objectively determine if the therapy or medication is working.

CLINICAL RESEARCH

The study of health and illness in people. It can include things like the cause, prevention, diagnosis, treatment, or progression of a disease.

ADVERSE EVENT (AE)

An event that happens in a study to the patient while receiving the treatment/therapy. It may or may not be caused by the treatment or therapy.

ASSENT

Children under the age of 18 are not legally able to provide informed consent. Instead, they are asked for their assent (meaning they agree to take part in the study). Assent is obtained in addition to parental/guardian informed consent. Assent is usually obtained from the ages of 7-17, but this does vary depending on the local IRB.



CLINICAL TRIAL RANDOMIZATION

In a clinical trial, people are assigned by chance to separate groups that compare different treatments. A participant cannot choose which group to be in and neither can the researcher

COMPASSIONATE USE

See "Expanded Access"

DATA SAFETY MONITORING BOARD (DSMB)

Group of experts that advises the study investigators about the study safety. They can make recommendations throughout the study that could affect whether the study continues or is modified or is stopped because of safety concerns.

DNA

Deoxyribonucleic acid (DNA) is the chemical inside the nucleus of all cells that carries the genetic instructions for making living organisms.

DNA SEQUENCING

A method of testing that is like proofreading a sentence. With DNA sequencing you determine the exact genetic code of the area you are studying. Different people can have slightly different sequences (variants) with little or no impact on how well the gene works. This is normal variation and is what makes us each unique. However, some errors in a gene's sequence can have serious consequences and cause diseases like Duchenne.

DOUBLE BLIND STUDY

Two or more parties involved (such as the researcher and the person in the study) do not know which people have been assigned which treatment groups.

EXCLUSION CRITERIA

A list of things that exclude a patient from being in a study.

EXPANDED ACCESS

A means by which companies can make investigational new drugs available, under certain circumstances, to treat a patient(s) with a serious disease or condition who cannot participate in a controlled clinical trial. FDA must approve the use, the company must be willing to supply the drug, and a treating physician must be willing to monitor the patient.

GENE

A gene is, in essence, a part or portion of DNA that gives the body instructions on how to make specific proteins such as dystrophin.

HETEROZYGOTE

A person who has one copy of a gene with a genetic change. Humans typically have two copies of most genes. When you are a heterozygote for a particular gene change, it means one copy of your gene contains the gene change, while the other copy does not.

INCLUSION CRITERIA

A list of features such as age, mobility status, disease state, etc., that must be met to be in a study.

INFORMED CONSENT

The voluntary permission from a person stating they are willing to be in a clinical trial. Before a person gives their consent, the study coordinator will review with them information about the trial, including what the study is trying to show (potential benefits, risks and inconveniences, alternative therapies available) and of the person's rights and responsibilities.

INSTITUTIONAL REVIEW BOARD (IRB)

An independent group of professionals designated to review and approve the study which includes the protocol, informed consent forms, study advertisements, and patient brochures. The IRB's job is to ensure that the study is safe and effective for anyone who will be in the study. It is also the IRB's responsibility to ensure that the study adheres to the FDA's regulations.

The IRB is responsible for:

- making sure the risks are as low as possible and that the risks are worth the benefits, and
- making sure all federal, institutional, and ethical guidelines are followed.

The IRB must review and approve components of a study including:

- the protocol
- informed consent forms
- study advertisements/recruitment materials
- informative handouts (i.e. fact sheets or brochures).

INTERMEDIATE CLINICAL ENDPOINT (ICE)

A measure of a therapeutic effect that is considered reasonably likely to predict the clinical benefit of a drug.

INVESTIGATIONAL NEW DRUG (IND)

A drug that has not been approved for general use by the FDA, but is being studied in clinical trials to study its safety and effectiveness. The IND program at FDA is how a pharmaceutical company obtains permission to ship an experimental drug across state lines (usually to clinical investigators) before the drug has been approved. The FDA reviews the IND application for safety to assure that research subjects will not be subjected to unreasonable risk. If the application is cleared (accepted), the candidate drug usually enters a Phase 1 clinical trial.

MUTATION

A change in genetic material. Mutations can be passed down through families or can occur by accident in a person for the first time. A mutation changes how that gene is supposed to work.

NATURAL HISTORY STUDY

A study that follows a group of patients over time who have, or are at risk of developing, a specific medical condition or disease. A natural history study collects health information in order to understand how the medical condition or disease naturally develops and progresses.

NEW DRUG APPLICATION (NDA)

The compiling of all non-clinical, clinical, pharmacological, pharmacokinetic, and stability information required about a drug by the FDA in order to approve the drug for marketing in the U.S.

OPEN LABEL STUDY

Everyone involved (i.e. people, doctors, pharmacists) in the trial know which people have been assigned which treatment group.

OUTCOME MEASURE

A test that is used to objectively determine the function of a patient. Common outcome measures in Duchenne are the 6-minute walk test (6MWT), North Star Ambulatory Assessment (NSAA), 10-meter walk/run. Non-ambulatory measures include reachable workspace and performance of upper limb (PUL).

PATIENT REPORTED OUTCOME (PRO)

Any report about a person and their health that comes directly from the person, without interpretation of that information by a clinician or anyone else. Examples include the surveys on DuchenneConnect that ask about symptoms, mobility, and quality of life.





CLINICAL TRIAL GLOSSARY

PHARMACOKINETICS (PK)

The study of how a drug is processed, metabolized, and gotten rid of from the body.

PHENOTYPE

Traits or characteristics that are observational such as brown hair or eye color.

The phenotype is determined by a person's genetic makeup, or genotype. In Duchenne a phenotype trait could be scoliosis as it is an observable trait.

PRE-IND MEETING

Sponsors looking for pre-IND guidance can request a "Type B" meeting with the FDA. This is an opportunity for a company to gain valuable feedback from the FDA on any questions regarding drug development. Although the FDA does not require these meetings, they are recommended because such meetings can confirm the requirements of the development process.

PRIMARY ENDPOINT

The main event or outcome that is being used to determine if the therapy or treatment actually works.

PROTOCOL AMENDMENT

A written description of a change(s) to a protocol.

SECONDARY ENDPOINTS

Secondary endpoints are only used to help interpret the primary endpoint, but they cannot be used by themselves to prove a therapy or treatment is effective. Secondary endpoints may also help provide information about future research.

SERIOUS ADVERSE EVENT (SAE)

An event in a study that includes any of the following: (a) inpatient hospitalization or prolonging of a hospital stay, (b) significant disability, (c) death or life threatening event, (d) requires treatment to prevent permanent damage, or (e) results in a birth defect.

SINGLE BLIND STUDY

Either the researcher or the person in the study do not know which treatment group the person has been given.

SPONSOR

The group, company, or individual who is paying for the clinical research. Sponsors include individual researchers, foundations, voluntary groups, health care institutions, government agencies, and pharmaceutical, biotechnology, and medical devices companies.

STUDY PROTOCOL

The written description of a clinical study. It includes the study's objectives, design, endpoints (or what is being measured), methods, inclusion, and exclusion criteria (who can participate).

SURROGATE ENDPOINT

In clinical trials, a surrogate endpoint is an indicator or sign used in place of another to tell if a treatment works. Surrogate endpoints do not guarantee that a treatment works, but they can give an earlier indication that the therapy is reasonably likely to have benefit. Biomarkers can be used as surrogate endpoints (see biomarkers). In cancer, surrogate endpoints include a shrinking tumor. In Duchenne, though not yet validated, dystrophin levels or MRI imaging of muscle are exploratory surrogate endpoints.

X-LINKED DISORDER

The gene for the disease is located on the X chromosome. Males have one X chromosome and one Y chromosome. Females have two X chromosomes.

NOTES

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JOIN THE RACE TO END DUCHENNE

We are thrilled to announce the launch of the new name and look for our endurance program, Race to End Duchenne. What began as one race with 80 runners has grown into a movement across multiple types of endurance events, with more than 600 participants annually in races around the world. Whether you are a seasoned athlete or a first time runner/walker, we have an event where you can make your miles count even more as you pound the pavement to raise funds for PPMD's mission!

Visit racetoendduchenne.org and join the Race to End Duchenne!

CREATE YOUR OWN EVENT

PPMD's mission is to end Duchenne but we need YOU! Have a great idea for a fundraiser? Looking for ways to raise money to support research? You can do just about anything to raise funds to end Duchenne. And PPMD is here to help you every step of the way. Visit our newly launched 'do-it-yourself' website for all the tools you need to create a custom event that raises money and awareness in your community.

Visit parentprojectmd.org/diy and start a DIY event!

LET'S TACKLE DUCHENNE

For 11 years, college and high school coaches across the country have been helping PPMD raise awareness by wearing Coach To Cure MD patches on the last Saturday in September. This year, during the weekend of September 29, we need your help to make this 11th season of Coach To Cure MD our best season yet! Join our team and take part in one of the easy – and fun! – activities we have put together to help you help us end Duchenne.

Visit coachtocuremd.org and tackle Duchenne!



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