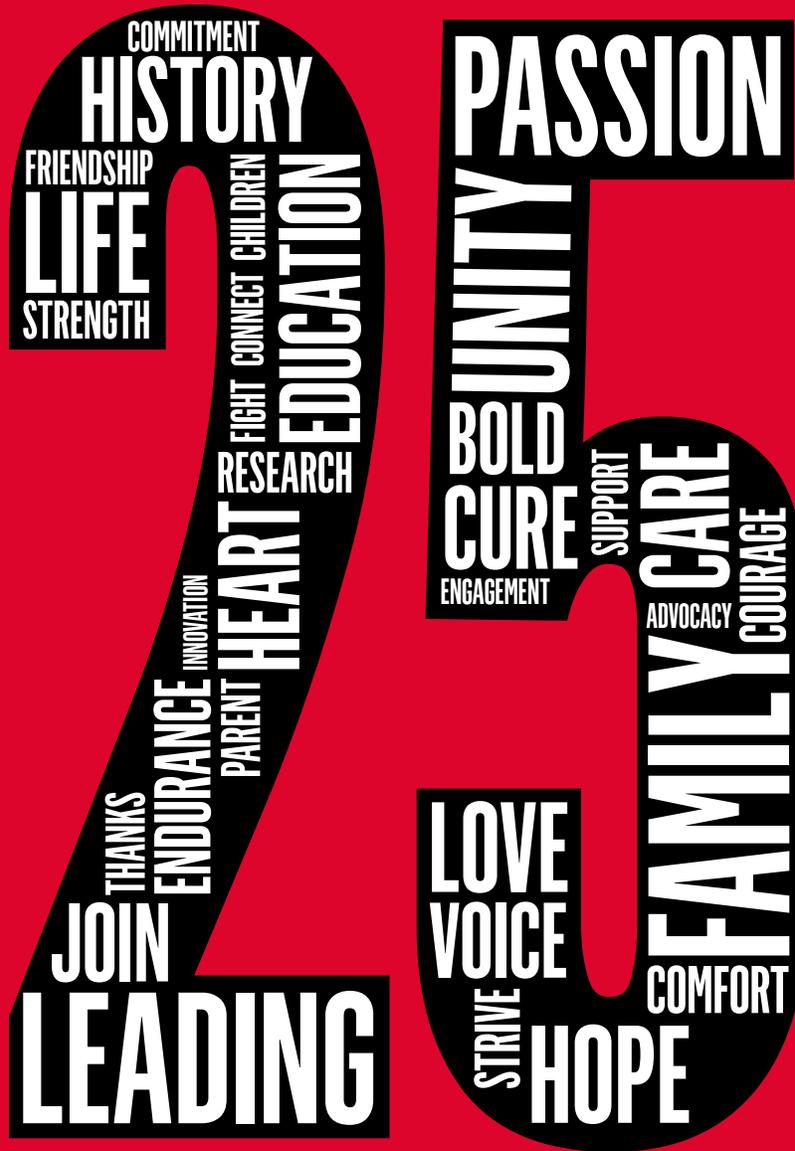


Parent Project Muscular Dystrophy

JOIN THE FIGHT.
END DUCHENNE.

June 26 – 30, 2019
Orlando, FL



JOIN THE FIGHT



SAVE THE DATE: DUCHENNE ACTION MONTH RETURNS SEPTEMBER 2019

September will be packed full of simple and powerful actions you & your community can take to raise awareness and funds to support PPMD's vital work in the fight to end Duchenne.

- The easiest way to take action this **September is with a Facebook fundraiser on September 7 (World Duchenne Awareness Day)**. Go to PPMD's Facebook page, and in just a couple of clicks, you can raise awareness & help fund our fight!
- Create your own **'do it yourself'** event to get your local community involved!
- Stock up on great new **PPMD merchandise** so that you can raise awareness all month long!

Visit endduchenne.org/actionmonth to get started!

DIY FUNDRAISING TO END DUCHENNE

Join PPMD and fund the fight to end Duchenne *your way*! From bake sales and game nights, to full-scale golf tournaments and live auctions, the PPMD team will help make your ideal fundraising event a reality. You know your community best and we are here to help you host something that is both impactful and fun. PPMD will help guide you and give you all the tools that you will need to be successful in the fight to end Duchenne!

Get started today. Visit endduchenne.org/diy.

RACE TO END DUCHENNE

Have you always wanted to run a race? Is being active your thing? Whether you're a seasoned athlete or a first-time race participant, joining the Race to End Duchenne team is a great way to take action. The Race to End Duchenne program raises nearly \$1 million a year thanks to our dedicated team members, who do everything from run marathons to bike in road races to conquer triathlons, all while raising funds and awareness about Duchenne.

Visit racetoendduchenne.org and join today!

LET'S TACKLE DUCHENNE

The last Saturday in September will mark the 12th Annual Coach To Cure MD. College and high school football coaches across the country will once again help PPMD raise awareness by wearing Coach To Cure MD patches to #TackleDuchenne. This September 28, we need YOU to join our team.

Taking part is fun and easy – visit coachtocuremd.org to help your team #TackleDuchenne!

WELCOME HOME



I'm a big fan of serendipity, of full circle moments. It's hard to believe that PPMD's first Annual Conference was 25 years ago AND that very first meeting was also in Orlando!

So much has changed since that first meeting. There were maybe 40 to 50 people in attendance. I'm not sure if we had a printed agenda, but if we did it probably didn't take up a full page of paper. There just wasn't that much to discuss.

But it was the beginning. Our beginning. A moment that led us to today – 600 families & experts and an agenda that packs a week's worth of content into three days.

There has never been so much promise in Duchenne research. We have never had a therapeutic pipeline so packed full of potential treatments. With 25 years of experience in this community, we understand how complex Duchenne is and how this multisystem disease will require a multisystem solution.

When we started 25 years ago, we were parents and grandparents meeting with a few experts to learn more about Duchenne – the bleak truth that there was little research being done and little federal or regulatory support.

Not only are parents and grandparents joining us this week, but siblings, friends, co-workers, and most importantly, people living with Duchenne – who aren't just part of the conversation, they are leading the conversation.

Today, we have dozens of pharmaceutical companies and biotechs in attendance, we have the respect and attention of federal agencies, and we have 25 Certified Duchenne Care Centers, all represented and here in Orlando with us to hear our stories and better understand our Duchenne journeys.

“

**You are my people.
This is where life is lived.
There are the moments, the people,
That make life worth living.
These are the people who make life beautiful.
These are my people.
This is my tribe.**

”

This week, amidst all of the incredible presentations, breakout groups, and our first town hall discussion, we hope you will find moments to reconnect with old friends and make new ones. I've always believed that the greatest benefit of attending our Annual Conference is the opportunity to find other families like your own that get you, that understand what you are living through.

I say it every year – none of us wanted to be part of the Duchenne community, none of us wanted to live life affected by a rare disease. But 25 years in, I am in constant awe of each and every one of you here today.

You are my people. You are my tribe.

Thank you and have a wonderful Conference!

Warmly,

Pat Furlong
Founding President & CEO
Parent Project Muscular Dystrophy



SPECIAL EVENTS & MEET UPS



DUCHENNE TEENS & ADULTS SOCIAL

Wednesday, June 26, 7:00–10:00pm

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 26, 7:00–10:00pm

This small gathering is for our newly diagnosed families only and will be held on Wednesday evening. If you are a newly diagnosed family and need child care, PPMD staff members will be happy to provide that care in the adjoining meeting room.



WELCOME RECEPTION

Thursday, June 27, 6:15–8:45pm

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 29, 7:30–10:30pm

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 and our 25th anniversary. This is a celebration you will not want to miss and the perfect opportunity to unwind with friends new & old.

DON'T MISS....



INTERACTION ALLEY

Friday, June 28,
11am–2:30pm
& 4:30–6:30pm
Saturday, June 29,
11am–4pm

A new experience at this year's PPMD Annual Conference is Interaction Alley. Here you will be able to make one on one appointments with experts who can help you navigate physical therapy challenges, challenges with school, learning & behaviors, or even take some time to meet with our Duchenne Registry Genetic Counselors. Kids will also have fun at the Microsoft Xbox exhibit (on Friday, June 28 only) where the new adaptive controller and other adaptive devices will be set up to try.



TOWN HALL

Saturday, June 29,
5:15–6:15pm

This year PPMD will end the 25th Annual Conference by hosting an interactive, one-of-a-kind Town Hall. This session will be a conversation. All presenters, attendees, PPMD staff, and literally everyone at the PPMD Annual Conference will be invited to discuss what matters to them as the Duchenne community looks to the future. We will ask questions, listen to the answers, share our concerns, and dig deep into the strategies, goals, and hurdles the future presents. You will not want to miss being part of this Town Hall and having your views, your voice heard.



KIDS TRACK

Thursday, June 27,
3:15–5:15pm
Friday, June 28,
9:30am–12:30pm
Saturday, June 29,
2:30–5:00pm

Kids Track kicks off on Thursday with an Amazing Animals show! Come meet an alligator, skunk, parrot, and some other animals at this hands-on experience. Reunite with old friends and make some new friends too! The fun continues on Friday and Saturday with age appropriate breakout groups and activities for each level.

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 and all served meals at the conference.



SIB SHOP

Saturday, June 29,
2:30–5:00pm

As a program of the Kids Track, PPMD will offer a Sib Shop for those children who are siblings to people with Duchenne. This breakout for the siblings has become a successful element of PPMD's educational events and is a safe place for siblings to be together, talk, and really open up on topics that they face.

Registration Details: Registration is \$65 per child. If you did not register your child ahead of time, please speak to someone at PPMD Conference Registration.

Sib Shop registration also includes a free t-shirt and all served meals at the conference.



THANK YOU TO PPMD'S RESOURCE FAIR EXHIBITORS!

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

The Resource Fair will be open Thursday, June 27, 6:15 – 8:45pm, and displays may remain open through Friday, June 28 at 2:30pm.

PREMIERE EXHIBITORS

Cincinnati Children's Hospital
changing the outcome together

KINOVA

 **Microsoft**


A DIVISION OF
TRAVISMEDICAL

 **NATIONWIDE CHILDREN'S**
When your child needs a hospital, everything matters.™

STANDARD EXHIBITORS

 **The Assistance Fund**

 **CUMBERLAND®**
PHARMACEUTICALS

 **DUCHENNE**
Family Assistance Program


EVEREST
TO END DUCHENNE


IMAGING DMD
ON TOP OF THE FIGHT FOR A CURE


Jett
FOUNDATION


millennium
respiratory services
Respiratory Homecare Specialists

PERFECT
LIFT
robot


ReveraGen
BioPharma



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
endduchenne.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.

endduchenne.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.

endduchenne.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.

endduchenne.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.

endduchenne.org/decode



FOR FAMILIES

Certified Duchenne Care Centers: PPMD's Certified Duchenne Care Center network has now grown to 25 incredible centers across the United States, serving more than 3,300 patients and their families. endduchenne.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.

endduchenne.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.

endduchenne.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).

endduchenne.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.

endduchenne.org/tour



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. endduchenne.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.

endduchenne.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.

endduchenne.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.

duchenregistry.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.

endduchenne.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.

endduchenne.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 25 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 preclinical research that is soon to be in clinical trial. These FAQ sheets have been written for this year's program book in direct consultation 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 endduchenne.org/exploretrials.



EXPLORE CLINICAL TRIALS



ACTIVELY RECRUITING



Therapeutic Approach	Study	Industry/ Institution	Age (years)	Ambulation Status	Mutation Specific	Steroid Use	Location	Page
	Ataluren - Long-Term Outcomes (041)	PTC Therapeutics	5+	Ambulatory	Yes - Nonsense mutation	Currently using	United States + Other	12
	Ataluren - The Dystrophin Study (045)	PTC Therapeutics	2-7	Either	Yes - Nonsense mutation	No specific requirement	United States	13
	Ataluren - The Dystrophin Study (046) in participants currently taking ataluren	PTC Therapeutics	Any	Ambulatory	Yes - Nonsense mutation	No specific requirement	United States	14
	Cardiac MRI Biomarkers and Genotype-Phenotype Correlations	NIH	7-21	Either	No	No specific requirement	United States	15
	Edasalonexent (PolarisDMD trial)	Catabasis Pharmaceuticals	4-7	Ambulatory	No	None in past 6 months	United States + Other	16
	Genetic Modifiers	UCLA	Any	Either	No	No specific requirement	United States	17
	Givinostat	Italfarmaco SpA	6-17	Ambulatory	No	Currently using	United States + Other	18
	Idebenone (SIDEROS trial)	Santhera Pharmaceuticals	10+	Either	No	Currently using	United States + Other	20
	MNK-1411 (BRAVE study)	Mallinckrodt Pharmaceuticals	4-8	Ambulatory	No	Never used	United States	21
	PF-06939926 Mini-Dystrophin Gene Therapy	Pfizer Inc	5-12	Ambulatory	No	Currently using	United States	22
	RG6206 (SPITFIRE study)	Roche/Genentech	6-11	Ambulatory	No	Currently using	United States + Other	24
	SGT-001 Micro-Dystrophin Gene Transfer (IGNITE DMD trial)	Solid Biosciences	4-11	Ambulatory	No	Currently using	United States	25
	SRP-4045 and SRP-4053 - Exons 45 and 53 Skipping (ESSENCE study)	Sarepta Therapeutics	7-13	Ambulatory	Yes - Deletion amenable to exon 45 & 53 skipping	Currently using	United States + Other	26
	SRP-5051 - Exon 51 Skipping (PPMO study)	Sarepta Therapeutics	12+	Either	Yes - Deletion amenable to exon 51 skipping	No specific requirement	United States + Other	27
	SRP-9001 Micro-Dystrophin Gene Transfer	Sarepta Therapeutics	4-7	Ambulatory	No	Currently using	United States	28
	Vamorolone	ReveraGen Biopharma	4-7	Ambulatory	No	Never used	United States	29

EXPLORE CLINICAL TRIALS



NOT YET RECRUITING



OBSERVATIONAL

Therapeutic Approach	Study	Industry/ Institution	Age (years)	Ambulation Status	Mutation Specific	Steroid Use	Location	Page
	Ifetroban	Cumberland Pharmaceuticals	7+	Either	No	Stable	United States	31
	Suvodirsen - Exon 51 Skipping (DYSTANCE 51 study)	Wave Life Sciences	5-12	Ambulatory	Yes - Deletion amenable to exon 51 skipping	Currently using	United States + Other	32



RESTORING OR REPLACING DYSTROPHIN

ONGOING



Therapeutic Approach	Study	Industry/ Institution	Page
	CAP-1002 (HOPE-2 trial)	Capricor Therapeutics	33
	Carrier Study	Nationwide Children's Hospital	34
	Pamrevlumab	FibroGen	35
	Viltolarsen (Formerly NS-065/NCNP-01) - Exon 53 Skipping	NS Pharma	36

PRECLINICAL



Therapeutic Approach	Study	Industry/ Institution	Page
	Wave Preclinical Exon Skipping	Wave Life Sciences	37



COMBATING FIBROSIS



REDUCING INFLAMMATION



IMPROVING MUSCLE GROWTH AND PROTECTION



RESTORING THE CELLS ENERGY



IMPROVING HEART FUNCTION

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. 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.



ATALUREN - LONG-TERM OUTCOMES (041)

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



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL

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.
- In 2018 the indication for Translarna was expanded to include patients as young as 2 years old.
- PTC is expanding commercial access to Translarna across Europe, the Middle East, Latin America, and Asia Pacific.
- In the US, PTC is conducting a study to assess dystrophin production in patients taking Translarna. The study was initiated towards the end of 2018 and is currently enrolling. You can find more information at www.clinicaltrials.gov. PTC plans for a potential U.S. regulatory re-submission in 2020.

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?

Travel expenses for participants in the trial will be reimbursed

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 may not be normally be 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).

ATALUREN – THE DYSTROPHIN STUDY (045)

A STUDY TO ASSESS DYSTROPHIN LEVELS IN PARTICIPANTS WITH NONSENSE MUTATION DUCHENNE MUSCULAR DYSTROPHY (NMDMD)



What stage is this research?

This trial is actively recruiting participants within the United States.

What is the goal or purpose of this study?

The goal of this study is to assess the production of dystrophin in participants with nonsense mutation Duchenne muscular dystrophy.

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.
- Translarna (ataluren) has received marketing authorization in the European Union for the treatment of nonsense mutation Duchenne in ambulatory patients aged two years and older.

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 2 to 7 years
- Positive for a nonsense point mutation in the dystrophin gene
- Willingness to undergo 2 needle muscle biopsies, 1 at the study start and 1 after 40 weeks
- No previous exposure to ataluren treatment
- Willing and able to comply with scheduled visits, drug administration plan, study procedures, muscle function tests, laboratory tests, and study restrictions

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

- The study will evaluate the levels of dystrophin before and after 40 weeks of ataluren therapy using needle muscle biopsies using two validated assay methods, electrochemiluminescence (ECL) and immunohistochemistry. There will be periodic visits in between for evaluation.
- Participants will be required to travel to UCLA (Los Angeles, CA) for the biopsy visits.

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 40 weeks during which all participants will receive ataluren.
- At the end of the study, participants may have the opportunity to continue ataluren through an on-going extension study.

Where does this study take place?

This study takes place at multiple sites within the United States; Biopsies are performed at UCLA (Los Angeles, CA).

How many visits to the study site are necessary?

In addition to the screening visit there will be a total of 3 visits – 2 at UCLA for biopsies and 1 at study site for evaluation and dosing adjustment.

Can any visits be done locally?

Visits are only conducted at the study sites and UCLA.

Is there any funding to help pay for travel?

Travel expenses for participants in the trial will be reimbursed.

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 may not normally be 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 (NCT03648827).
- Please check parentprojectmd.org for updates to this FAQ sheet.



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



ATALUREN – THE DYSTROPHIN STUDY (046) IN PARTICIPANTS CURRENTLY TAKING ATALUREN

A STUDY TO ASSESS DYSTROPHIN LEVELS IN PARTICIPANTS WITH NONSENSE MUTATION DUCHENNE MUSCULAR DYSTROPHY (NMDMD) WHO HAVE BEEN TREATED WITH ATALUREN



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL

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 assess the production of dystrophin in participants with nonsense mutation Duchenne muscular dystrophy who are being treated with ataluren.

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.
- Translarna (ataluren) has received marketing authorization in the European Union for the treatment of nonsense mutation Duchenne in ambulatory patients aged two years and older.

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
- Positive for a nonsense point mutation in the dystrophin gene
- Willingness to undergo a needle muscle biopsy
- Currently being treated with ataluren 10, 10, 20 mg/kg for ≥ 9 months, with no gap in treatment of greater than (>) 1 month, in an ongoing PTC-sponsored nmDMD clinical trial prior to study entry
- Willing and able to comply with scheduled visits, drug administration plan, study procedures, muscle function tests, laboratory tests, and study restrictions

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

- The study will evaluate the levels of dystrophin after ataluren therapy using muscle biopsies using two validated assay methods, electrochemiluminescence (ECL) and immunohistochemistry.
- Participants will be required to travel to UCLA (Los Angeles, CA) for the biopsy visits.

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

The participant will remain in their current ataluren clinical trial until the end of that study.

Where does this study take place?

This study has one visit that takes place at UCLA (Los Angeles, CA).

Can any visits be done locally?

No, visits are only conducted at UCLA.

Is there any funding to help pay for travel?

Travel expenses for participants in the trial will be reimbursed.

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 may not normally be 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 (NCT03796637).
- Please check parentprojectmd.org for updates to this FAQ sheet.

CARDIAC MRI BIOMARKERS AND GENOTYPE-PHENOTYPE CORRELATIONS



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

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 males with Duchenne muscular dystrophy. The study aims:

- to identify and characterize several cardiac MRI biomarkers for patients with Duchenne;
- to define the sensitivity of the well-characterized cardiac MRI biomarkers for detecting early cardiac involvement; and
- to use these validated cardiac MRI biomarkers to better understand the genotype-phenotype correlation in males 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 males 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 males are healthy volunteers who will complete a cardiac MRI without contrast.
- Group 1b males are patients with Duchenne who will complete a cardiac MRI with contrast, blood tests, 24 hour heart monitoring, and pulmonary function tests.
- Group 2 males are patients 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 males are patients 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 patients 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 six 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, Stanford University, and Children's Hospital of Orange County (CHOC).

How many visits to the study site are necessary?

- Patients 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 patients (Group 1b and 3) will come to the clinic for a single visit, except for ten patients (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, Stanford or CHOC hospitals.

Is there any funding to help pay for travel?

Yes, if you are a patient 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).



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ONGOING



PRECLINICAL



EDASALONEXENT (POLARISDMD TRIAL)

CATABASIS' PHASE 3 POLARISDMD TRIAL OF EDASALONEXENT IN DUCHENNE MUSCULAR DYSTROPHY



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL

What stage is this research?

PolarisDMD is enrolling males in a global Phase 3 trial in Duchenne muscular dystrophy to evaluate the efficacy and safety of edasalonexent.

What is the goal or purpose of this research?

Edasalonexent inhibits NF- κ B, a protein which plays a fundamental role in skeletal and muscle disease in Duchenne. By inhibiting NF- κ B, edasalonexent has the potential to decrease inflammation and fibrosis, promote muscle regeneration, and slow disease progression. Our goal is for edasalonexent to become an oral new foundational therapy to slow disease progression for all people affected by Duchenne at all ages as a single agent and in combination with other therapies.

Who is eligible to participate in the trial?

The trial is enrolling approximately 125 patients with the following criteria:

- Patients ages 4 to 7 (up to 8th birthday) with a diagnosis of Duchenne (any confirmed mutation)
- Ability to complete the timed function tests, including stand from supine without assistance in ≤ 10 seconds, 10MWT and 4-stair climb
- Not on corticosteroids for at least 6 months
- The key inclusion and exclusion criteria for this clinical trial is available on www.clinicaltrials.gov (NCT# 03703882)

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

- The trial is a randomized, double-blind and placebo-controlled study with two patients receiving edasalonexent for each patient receiving placebo. All patients may have the opportunity to receive edasalonexent after 12 months through our open-label extension, GalaxyDMD.

- Edasalonexent capsules are taken orally, three times per day with food. The soft-gel capsules are available in two sizes, one similar to the size of a Tic-Tac and another similar to the size of a jelly bean. A special straw is provided to help facilitate capsule swallowing.
- Clinical trial site visits are every three months to measure key assessments and safety, including labs. Neither muscle biopsies nor MRIs are required for this study.

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

After the 12-month PolarisDMD trial, all patients may have the opportunity to receive edasalonexent in our open-label extension, GalaxyDMD.

Is there any funding to help pay for site travel?

Yes, Catabasis will fund travel-related expenses as described by the clinical study coordinator at the clinic site and through our patient travel coordinating agency.

What are the results from the Phase 2 MoveDMD® trial and open-label extension?

- Preserved muscle function and slowed disease progression were observed in all assessments of muscle function (North Star Ambulatory Assessment, time to stand, 4-stair climb and 10-meter walk/run) through 72 weeks of edasalonexent treatment compared to the off-treatment control period.
- Supportive changes in measures of muscle health were seen, with significant reductions in muscle enzymes, C-reactive protein (CRP), and MRI T2 rate of change compared to the off-treatment control period, supporting the durability of edasalonexent treatment effects.
- Significantly decreased heart rate towards age-normative values, supporting potentially beneficial cardiac effects of edasalonexent.

- Patients grew similarly to unaffected males. Height increased by an average of 2.1 inches/year, while weight increased by an average of 2.9 lbs/year, both in line with typical height and weight increases of unaffected patients.

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.

Where does the clinical trial take place?

PolarisDMD has clinical trial sites in North America, Europe, Israel, and Australia to improve patient access. For more information on expected and active clinical sites, please visit www.catabasis.com/patients-families/polaris-dmd-clinical-trial.php.

Where can I learn more about this research?

- You can learn more at www.catabasis.com/patients-families/polaris-dmd-clinical-trial.php
- You can contact Catabasis directly with any questions at DMDTrials@catabasis.com.
- Please check parentprojectmd.org for updates to this FAQ sheet.

GENETIC MODIFIERS

GENETIC MODIFIERS OF DUCHENNE AND BECKER MUSCULAR DYSTROPHY



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 PPMDC.

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



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ONGOING



PRECLINICAL



GIVINOSTAT

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



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NOT YET RECRUITING



ONGOING



PRECLINICAL

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 trial is to evaluate the efficacy of givinostat compared to placebo to slow the disease progression in males with Duchenne muscular dystrophy. 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 adult and pediatric populations with cancer and arthritis. We completed a Phase 2 clinical trial (DSC/11/2357/43) to evaluate the safety and the potential efficacy of Givinostat among males with Duchenne.
- DSC/11/2357/43 was an open label, 2-part, Phase 2 trial in ambulatory males with Duchenne who were 7 years up to 11 years old. Trial results showed that Givinostat significantly reduced histological disease progression after 12 months of treatment, in particular:
 - o It significantly increased the size of muscle fibers, and it significantly decreased total fibrosis, fatty replacement, necrosis, the mean number of hypercontracted fibers;
 - o It significantly increased regenerative fibers with no depletion of the pool of satellite cells.

Analysis of functional testing found that participants' muscle strength and function were stable after 12 months of treatment, with small changes in some parameters. Overall, the drug was safe and well tolerated. The study was completed in December 2017. 18 patients are still on treatment in a long-term safety study (DSC/14/2357/51). The results of the 12-month Phase 2 trial 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 224 ambulatory males with Duchenne. Of these, 160 participants will be randomly assigned to Givinostat and 80 will be randomly assigned to placebo. To participate in this study you must be:
- Male with a Duchenne diagnosis confirmed by genetic testing
 - Age ≥ 6 years
 - On a stable corticosteroid for at least 6 months
 - Able to perform the 4 stair climb in not more than 8 seconds
 - Able to rise from the floor in 3 up to 10 seconds
 - There are other inclusion and exclusion criteria for this study. You can find all of the details on www.ClinicalTrials.gov (NCT#02851797).

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

- This study involves 15 clinic visits during an 18-month period. Visits are more frequent during the first 3 months. After the screening period, patients will be randomly assigned to receive givinostat oral suspension or placebo oral suspension taken twice a day with food (e.g. during breakfast and dinner).
- Trial visits include physical function testing, blood tests, urine tests, and physical exams.
- Participants also complete three MRI tests to measure changes inside the muscle.

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 trial is 18 months. Participants who finish the double - blind placebo - controlled study can enroll in an open-label long-term safety study to receive givinostat oral liquid suspension until the drug becomes available on the market. Participants who finish the 18-month study are eligible for the open label study regardless of their walking or stair climbing ability. All participants in this long-term safety study (DSC/14/2357/51, NCT#03373968) will take givinostat oral liquid suspension.

Where does this study take place?

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

How many visits to the trial site are necessary?

Trial participants complete approximately 15 visits over 18 months. During the screening period, patients attend two visits. Once those are finished, patients have an MRI test, which might be at another site. Then patients return for one more visit to receive the first dose of the study drug. During the first month of treatment, patients attend weekly visits, then every two weeks in the second month, then once in the third month, and then every three months.

Can any visits be done locally?

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

Is there any funding to help pay for travel?

Yes. In the US and Canada, a travel agency will arrange your travel for you. In other countries, you will be reimbursed for your travel costs. Ask your site for more details.



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ONGOING



PRECLINICAL

Will I get paid for participating in this study?

No, you won't. Costs associated with participation in the trial (e.g. travel, parking, hotel, meals) are reimbursed. The reimbursement process is different in every country. In some countries Italfarmaco will arrange for your expenses to be reimbursed by your site. In other countries (US and Canada), we offer central travel support through a travel agency.

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 can email us at patientadvocacy@italfarmaco.com to ask more questions.



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)



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ONGOING



PRECLINICAL

What stage is this research?

The SIDEROS clinical trial is a Phase 3 clinical trial.

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 males with Duchenne receiving glucocorticoid steroids.

What is idebenone and how does it work?

Idebenone is thought to have a dual effect in Duchenne – mitochondrial activation and protection. Idebenone is thought to increase 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 participants 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 and travel to and from the study site.

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.clinicaltrials.gov (NCT02814019).

MNK-1411 (BRAVE STUDY)

A STUDY TO ASSESS THE EFFICACY AND SAFETY OF MNK-1411 IN MALES 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
- 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. An open Label Extension is offered to all participants until either the compound is marketed or in case MNK would stop development of MNK-1411. 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 study includes 4 weeks of screening, 24 weeks of double blind treatment and an optional open label treatment. An open Label Extension is offered to all participants until either the compound is marketed or in case MNK would stop development of MNK-1411.

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 (after the first 56 weeks: visits during the extended Open Label period will take place every three months).

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 patients with Duchenne.

Where can I learn more about this study?

You can learn more about this study at www.ClinicalTrials.gov (NCT#03400852) or <https://www.bravetrialdmd.com/>



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ONGOING



PRECLINICAL



PF-06939926 MINI-DYSTROPHIN GENE THERAPY

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



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NOT YET RECRUITING



ONGOING



PRECLINICAL

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).
- Preliminary planning is underway in anticipation of a global, pivotal study of PF-06939926.

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 ability to enter muscle tissue efficiently when tested in pre-clinical studies, and an acceptable safety profile in prior human 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 males 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 imaging assessments, such as with 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 five basic periods: screening, baseline evaluation, treatment, first-year follow ups, and then long-term follow up with annual visits for four years.
- Participants will receive a single IV infusion of PF-06939926 over approximately a two-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 five years.
- This study will seek to enroll up to 15 patients across multiple cohorts. Within each of the cohorts, the dosing interval between the first and second participant will be at least six weeks. If no safety concerns are identified three weeks after the second participant is infused, then dosing may proceed at shorter intervals. Recruitment and identification of the patients for participation is complete.
- 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 during, or after the study.

Where does this study take place?

- This study will be completed across three clinical research sites in the United States only; Duke University Medical Center, University of Utah Hospital, and UCLA Medical Center 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)



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ONGOING



PRECLINICAL



RG6206 (SPITFIRE STUDY)

ROCHE/GENENTECH CLINICAL TRIAL TO EVALUATE THE EFFICACY, SAFETY, AND TOLERABILITY OF RO7239361 IN AMBULATORY MALES WITH DUCHENNE MUSCULAR DYSTROPHY



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL

What stage is this study?

RG6206 (or RO7239361) is an investigational molecule in a SPITFIRE Phase 2/3 clinical study (NCT03039686) in ambulatory males with Duchenne muscular dystrophy.

What is the purpose of this study?

The main goal of the SPITFIRE Phase 2/3 study (NCT03039686) is to determine whether this investigational molecule RG6206 is effective, safe, and tolerated for ambulatory males 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 from growing too much.

Has RG6206 been studied before?

- Yes. An initial clinical trial (Phase 1) with RG6206 was completed in healthy adults. Further information can be found here: NCT03100630 on ClinicalTrials.Gov.
- RG6206 is currently being studied in a Phase 1/2 study to investigate its safety and tolerability in males with Duchenne. The trial is ongoing, but is no longer open to new patients. You can find more information here: NCT02515669 on ClinicalTrials.Gov.

Where is this study being carried out?

- The Phase 2/3 clinical study is global. Sites are located in the United States, Canada, Japan, Argentina, UK, Belgium, France, Spain, Sweden, Netherlands, Germany, Italy, and Australia.
- In the United States there are multiple study sites. Further information on sites and their status can be found here: NCT03039686 on ClinicalTrials.Gov.

Who are the participants in this clinical study?

The participants in this trial are males with Duchenne confirmed by medical history with genetic testing and are:

- 6 to 11 years of age (inclusive) and weighs 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
- Has a North Star Ambulatory Assessment (NSAA) total score ≥ 15 points at the Screening Visit
- Participants in this study can have any Duchenne mutations. Full screening assessments for participation in the study will be conducted at the study sites.

How many boys will participate in the study?

Approximately 159 males with Duchenne will participate in this study.

How long will the study be?

The study has two sequential parts:

- PART 1: consists of an initial 48-week randomized placebo-controlled phase during which all patients will receive either RG6206 or placebo (2:1 ratio, meaning twice as many participants will receive RG6206 versus placebo)
- PART 2: consists of a 192-week open-label phase during which all patients will receive RG6206

How is RG6206 given in the study?

RG6206 or placebo is administered once weekly by an injection under the skin (subcutaneous injection) in to the tummy (abdomen), thigh, or back of upper arm after initial training at the site. The three first doses must be done at the study site, while the remaining administrations are done at home.

Where can I learn more about this study?

- Please contact your treating physician to discuss the clinical study
- Visit ClinicalTrials.gov (search for NCT03039686)
- Call the Roche/Genentech Clinical Trial Information Support Line
Phone: 1-888-662-6728
Hours: Monday-Friday, 5am-5pm Pacific Time

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



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

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. 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 only, current 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. Currently, the study is within the ambulant, children's cohort ages 4-11, once the Data and Safety Monitoring Board (DSMB) review the data, they may permit non-ambulant adolescents, ages 12-17, to participate again. 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)



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PRECLINICAL



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



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PRECLINICAL

What stage is this research study?

This trial is a Phase 3a 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?

~80 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). However, all patients will receive active treatment in the Open Label Period after completing the 96-Week Double Blind Period.

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

SRP-5051 - EXON 51 SKIPPING (PPMO STUDY)

A PHASE 1 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



What stage is this research study?

Study 5051-101 is an open-label Phase 1 study that is currently recruiting participants in the US and Canada. Participants who complete the study may be eligible to enroll into 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 eteplirsen or drisapersen within six months prior to screening
- No participation in experimental gene therapy for treatment of 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 based on the protocol.

Where will this study take place?

This study is currently being conducted at over ten clinical sites in the United States and Canada. 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 two 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 three 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.
- 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.

*Passini et al. Presented at the 13th Annual Meeting of the Oligonucleotide Therapeutics Society (OTS) | 24-27 September 2017 | Bordeaux, France





SRP-9001 MICRO-DYSTROPHIN GENE TRANSFER

A 48-WEEK, RANDOMIZED, DOUBLE-BLIND, PLACEBO-CONTROLLED, SYSTEMIC, GENE-DELIVERY CLINICAL TRIAL FOR DUCHENNE MUSCULAR DYSTROPHY USING SRP-9001 WITH A 96-WEEK EXTENSION



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ONGOING



PRECLINICAL

What stage is this research?

This trial is a Phase I/2 study that is currently recruiting participants in the United States.

What is the goal or purpose of this study?

The purpose of this study is to evaluate the safety and efficacy of exogenous gene transfer in Duchenne patients by measuring biological and clinical endpoints in two parts: a 48-week randomized, double-blinded, placebo-controlled period (Part 1), and a 96-week, double-blinded extension period (Part 2).

Who is funding this study?

The study is sponsored by Sarepta Therapeutics, Inc.

Who is eligible to participate in this study?

Key inclusion criteria include but are not limited to:

- Established clinical diagnosis of Duchenne and documented dystrophin gene mutation of Duchenne phenotype.
- Indication of symptomatic muscular dystrophy by protocol-specified criteria.
- Ability to cooperate with motor assessment testing.
- Stable dose equivalent of oral corticosteroids for at least 12 weeks.
- Male, 4-7 years of age, inclusive.

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

After providing informed consent, patients will be evaluated for eligibility. Enrolled patients will perform functional tests, undergo laboratory work, and muscle biopsies throughout the course of the study.

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 144 weeks.

Where does this study take place?

This study currently takes place at one clinical site in the United States. To view the participating study site, visit www.clinicaltrials.gov (Identifier NCT03769116).

How many visits to the study site are necessary?

25

Can any visits be done locally?

No.

Is there any funding to help pay for travel?

Yes, costs associated with travel and parking are reimbursed.

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 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# 03769116).
- Please check parentprojectmd.org for updates to this FAQ sheet.



What stage is this research?

- Vamorolone is a multi-functional drug that shows anti-inflammatory, membrane stabilization, and mineralocorticoid receptor antagonist activities. While it is a steroidal drug, it is not a corticosteroid, and current data suggest that it does not lead to some of the side effects associated with corticosteroids in Duchenne (deflazacort, prednisone), such as growth stunting. Open label Phase 2 studies (VBP15-002; VBP15-003) have been completed in 46 Duchenne patients, and the data obtained suggested improvements in strength and mobility over a 6-month treatment period. 43 of these patients enrolled in a 2-year long-term extension study (VBP15-LTE); some have completed the LTE and enrolled in Expanded Access for continued vamorolone treatment. Currently, patients with Duchenne are being enrolled in the pivotal clinical trial (VBP15-004; Phase 2b/3), the trial requested by the FDA that was designed to get critical information about how vamorolone is working (efficacy) in order to file for drug approval. During the first 6 months of this trial, one quarter of the patients will receive a placebo, one quarter will receive prednisone, and half will receive vamorolone (either 2 mg/kg/day or 6 mg/kg/day). After 6 months, ALL of the patients will transition to vamorolone. Upon finishing the trial, there is an Expanded Access Program available so participants can continue to receive vamorolone. We are enrolling 120 males with Duchenne (4 to 6 years, steroid naïve); we plan to complete enrollment by the end of 2019. This study is recruiting at about 30 sites in the following countries: US, Canada, United Kingdom, Israel, Australia, Sweden, Netherlands, Czech Republic, Belgium, Spain, and Greece.

Other trials that are finished or ongoing:

- A Phase 2a clinical trial (VBP15-002) of 48 Duchenne males (4 to 6 years old, steroid naïve) is complete. This was a 2- week treatment trial, with a focus on safety and biomarkers. This study has been published: <https://www.sciencedirect.com/science/article/pii/S1043661818309757/pdf?md5=2ce96ac43a71cbf25159a7dfed62a384&pid=1-s2.0-S1043661818309757-main.pdf>
- A Phase 2a extension clinical trial (VBP15-003) in Duchenne males is also complete. Patients received 6 months of treatment with vamorolone; 46 of the 48 patients who enrolled in VBP15-002 entered and finished this study. The study showed that vamorolone was safe up to the highest dose (6 mg/kg) and showed dose-responsive improvements in strength and mobility measures.
- The Long Term Extension study (VBP15-LTE) is a 2-year continuation study of the same patients who were enrolled in VBP15-003, with all patients increasing vamorolone dose to 2.0-6.0 mg/kg/day. At the time of writing, 43 patients continue in or have finished this study; no one has withdrawn from the trial due to a safety concern. In this trial, we are evaluating the long-term safety of vamorolone, and comparing safety to patients treated with prednisone in a natural history study.
- Expanded access is available for participants who have finished vamorolone trials and wish to continue vamorolone treatment. We are continuing to monitor long-term safety of vamorolone during expanded access.

What is the goal or purpose of this research?

- The goal of this research is to see if vamorolone (an investigational drug) is effective (improves or stabilizes muscle strength and function) and safe in patients with Duchenne.
- Vamorolone is being developed as a potential replacement for corticosteroids (deflazacort, prednisone); it is not used in combination with corticosteroids.
- If trials show that vamorolone is safe and effective, it could potentially be approved for 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, VBP15-003, and VBP15-004 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:
 - o PPMD (\$750,000) and Foundation to Eradicate Duchenne (\$250,000) have co-funded the chronic toxicology studies
 - o MDA USA, and three UK foundations (Joining Jack, Duchenne Research Fund, Duchenne Children's Trust) provided \$2M for the Phase 1 trial.



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ONGOING



PRECLINICAL



VAMOROLONE (CONTINUED)



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ONGOING



PRECLINICAL

- o 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).

- o Grants for design of a clinical trial in younger (2-4 year) and older (8-18 year) Duchenne males has been provided by DuchenneUK and Save Our Sons.

- ReveraGen has received funding via option agreements with Actelion, Idorsia and Santhera.

Who is eligible to participate in this research study?

- Males 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 currently recruiting VBP15-004 study is a 1-year study for each patient enrolled.
- After completion of the study, participants may continue to receive vamorolone via ReveraGen's Expanded Access Program, under IND (see clinicaltrials.gov, NCT03863119) in the US, or possibly via compassionate use in other countries.

Where does this research study take place?

- The actively 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. There are no mandated site visits for the Expanded Access Program, but your child must continue to receive regular clinical care with visits to the physician prescribing vamorolone.
- All drug dosing is taken by mouth and is taken each morning at home. Vamorolone is a flavored liquid suspension.

Can any visits be done locally?

Visits for the VBP15-004 trial 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 a safe and effective therapy for Duchenne.
- If vamorolone is shown to be safe and effective for Duchenne, vamorolone may be available to other children with Duchenne instead of corticosteroids. If vamorolone is safe and effective in Duchenne, this could lead to other studies of vamorolone in disorders where corticosteroids are used.
- The vamorolone program includes many innovations in clinical trial design that, if successful, may speed other drug development programs, including blood biomarkers and mobile health outcomes.
- ReveraGen is focused on keeping patients at the center, providing continued access to vamorolone for trial participants.

Where can I learn more about this research study?

- You can learn more about this study at <https://vision-dmd.info/> or www.reveragen.com, or www.clinicaltrials.gov.
- For parent queries, contact Suzanne Gaglianone at Suzanne.gaglianone@reveragen.com; for physician queries contact Laurie Conklin, MD at laurie.conklin@reveragen.com



A PHASE 2 STUDY OF DAILY, ORAL ANTI-FIBROTIC THERAPY TO PREVENT HEART MUSCLE DISEASE AND IMPROVE HEART MUSCLE FUNCTION IN AMBULATORY AND NON-AMBULATORY DUCHENNE PATIENTS

What stage is this research?

Ifetroban research is at the clinical stage, meaning it has advanced to clinical trials involving people. Ifetroban has been studied in over 26 clinical trials and dosed in over 1,300 people including healthy volunteers. There are three active clinical trials evaluating ifetroban in other rare diseases. Animal studies mimicking Duchenne demonstrated ifetroban prevents heart muscle disease and prolongs survival. Recruitment is anticipated to begin in June 2019.

What is the goal or purpose of this study?

The goal of this clinical trial is to see if ifetroban, a novel targeted drug product, can be safely and effectively used for the treatment of Duchenne heart muscle disease.

Who is funding this study?

This study is funded by Cumberland Pharmaceuticals.

Who is eligible to participate in this study?

To participate in this study you must be male, 7 years of age or older, diagnosed with Duchenne and with stable cardiac function.

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

This study involves taking an oral medication once a day for 12 months, having your finger pricked on the first day of treatment and the 7th day of treatment to measure the oral medication levels in two drops of your blood. We will follow the health of your heart using an MRI before you start treatment and after receiving oral medication for 6 months and 12 months. As you are having your MRI,

blood will be drawn to make sure your blood cells and chemistry levels are normal. Your lung function, daily activity, quality-of-life and muscle strength will be checked before you start treatment and after receiving oral medication for 6 months and 12 months. We will call you periodically to see how you are feeling between visits.

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

The anticipated length of the study is 2 years. Treatment duration is 12 months. There is an optional open-label extension to all participants that complete 12 months of treatment.

Where does this study take place?

This multicenter Phase 2 study takes place at several U.S. sites including Riley Children's Hospital (IN), Vanderbilt's Children's Hospital (TN), Children's National Medical Center (DC), University of California Los Angeles (CA), Emory's Children's Healthcare of Atlanta (GA), and Nationwide Children's Hospital (OH).

How many visits to the study site are necessary?

Three visits over 12 months.

Can any visits be done locally?

All three visits must be done at one of the participating centers. Some procedures can be completed at home.

Is there any funding to help pay for travel?

Yes. Cumberland will provide patients a stipend for their participation, time and effort in the study. If an overnight stay is needed, Cumberland will provide a travel stipend to cover meals, hotel, and mileage.

Why should I consider participating in this study?

Participation will help determine whether ifetroban is an effective therapy for Duchenne heart muscle disease and whether quality-of-life, daily activity and muscle strength improve. 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 may not normally be available to you or your child, and helping others by contributing to the better understanding of Duchenne heart disease.

Where can I learn more about this study?

You can learn more about this study at www.ClinicalTrials.gov (NCT# NCT03340675) or by contacting Cumberland Pharmaceuticals at 615-564-2188 or imaciasperez@cumberlandpharma.com.





SUVODIRSEN - EXON 51 SKIPPING (DYSTANCE 51 STUDY)

A RANDOMIZED, DOUBLE-BLIND, PLACEBO-CONTROLLED, EFFICACY AND SAFETY STUDY OF SUVODIRSEN IN AMBULATORY PATIENTS WITH DUCHENNE MUSCULAR DYSTROPHY (DYSTANCE 51)



What stage is this research?

This is a Phase 2/3 clinical trial

What is the goal or purpose of this study?

The primary objective is to evaluate the efficacy and safety of the investigational drug suvodirsen compared to placebo in ambulatory males with Duchenne 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 DMD that is amenable to an exon 51 skipping therapy
- Male, aged 5 to 12 years old
- Ambulatory, able to walk independently for at least 10 meters in 20 seconds or less

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?

- The duration of treatment during the placebo-controlled portion of the trial is 48 weeks.
- Patients may be eligible to enter into an open-label phase of the trial to receive ongoing treatment with suvodirsen after they complete the placebo-controlled portion of the phase 2/3 clinical trial.

Where does this study take place?

We currently expect that this study will be conducted in the U.S., Europe, Canada, Japan, and Australia.

Will I get paid for participating in this study?

- There is no compensation for participating in the trial, but reasonable 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.ClinicalTrials.gov (NCT03907072).
- Email clinicaltrials@wavelifesci.com for additional information.
- Please check www.parentprojectmd.org for updates to this FAQ sheet.

CAP-1002 (HOPE-2 TRIAL)

HOPE-2 DUCHENNE CLINICAL TRIAL

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 males 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 patients 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?

Enrollment for HOPE-2 is currently on hold. An interim analysis is being performed by the Sponsor of the clinical trial and will be completed by the third quarter of 2019. The outcome of the analysis will determine whether to continue enrollment. Information on trial status and sites is kept current on the HOPE-2 page at ClinicalTrials.gov.

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. Pursuant to the protocol, participants are to receive an IV infusion of CAP-1002 or placebo every three months for a total of four 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) and availability of resources, Capricor 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





CARRIER STUDY

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



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL

What stage is this research?

Closed enrollment

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?

We are not looking for new participants.

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:
Thelma Asare
614-355-2606
Thelma.asare@nationwidechildrens.org
- www.clinicaltrials.gov (NCT02972580)

PAMREVLUMAB

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



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 4 years. All participants will be closely monitored for safety. Efficacy assessments will be performed routinely; pulmonary and muscle function tests approximately every three 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 four 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 parentprojectmd.org for updates to this FAQ sheet.



ACTIVELY RECRUITING



NOT YET RECRUITING



ONGOING



PRECLINICAL



VILTOLARSEN (FORMERLY NS-065/NCNP-01) - EXON 53 SKIPPING

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



What stage is this research?

- Study NS-065/NCNP-01-201 is a Phase 2 study which has completed enrollment (16 patients) 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.
- Submission of a rolling New Drug Application (NDA) for viltolarsen (NS-065/NCNP-01) was initiated for the treatment of Duchenne in patients amenable to exon 53 skipping.

What is viltolarsen (NS-065/NCNP-01)?

Viltolarsen (NS-065/NCNP-01) is an investigational 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 viltolarsen (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 were to assess the medication's 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 viltolarsen (NS-065/NCNP-01) injection up to an additional 144 week period or until the product is commercially available (total 168 weeks of treatment in Study NS-065/NCNP-01-201 and 202). Additional objectives of the study are to assess the medication's 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 males must have had 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 have been able to walk independently and to complete standard timed tests. Patients must have been on a stable dose of steroid medication for at least 3 months.

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

- Study NS-065/NCNP-01-201 was a 2-period, randomized, 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 received the study medication.
- Study NS-065/NCNP-01-202 is an open-label, extension study of NS-065/NCNP-01-201 for an additional 144 weeks.
- All participants who completed Study 201 have enrolled into Study 202 and all participants continue to receive the investigational therapy, viltolarsen (NS-065/NCNP-01).

How long will this study last?

- Study NS-065/NCNP-01-201 lasted 24 weeks.
- Study NS-065-NCNP-01-202 will last for up to an additional 144 weeks (total 168-week treatment) or until the product is commercially available.

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 viltolarsen (NS-065/NCNP-01).

Where do the studies take place?

This study was and continues to be 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. Identifiers 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).

WAVE PRECLINICAL EXON SKIPPING

WAVE LIFE SCIENCES – EXON SKIPPING RESEARCH

What stage is this research?

Wave is applying learnings from our exon 51 clinical program to further our exon skipping research in Duchenne to advance potential therapies targeting other exons. We have multiple research programs in the preclinical phase, meaning they have not advanced to clinical trials involving people yet.

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

This exon skipping research is being conducted and funded by Wave Life Sciences.

What is the goal or purpose of this research?

The goal of our research is to work toward developing medicines for people with Duchenne muscular dystrophy amenable to exon skipping. We have selected and are advancing a development candidate for exon 53 skipping. In addition, we are pursuing preclinical research toward investigational exon skipping therapies for people amenable to exon 44, 45, 52, 54, and 55 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 for any of our preclinical exon skipping programs.

Where would a clinical trial take place?

If our preclinical programs are ready to advance into the clinic, we expect to run global clinical trials and hope to have clinical trial sites in the United States. Due to the preclinical stage of our research, sites have not been selected at this time.

Where can I learn more about this research?

- Please email medicalinformation@wavelifesci.com for additional information.
- Please check <https://www.parentprojectmd.org/research/clinical-trials/explore-clinical-trials/> for updates to this FAQ sheet.



ACTIVELY RECRUITING



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ONGOING



PRECLINICAL



CLINICAL TRIAL GLOSSARY

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

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.

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.

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.

CLINICAL TRIAL GLOSSARY



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

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).



CLINICAL TRIAL GLOSSARY

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.

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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NOTES

SHOP TO END DUCHENNE



ONE TOUGH MOTHER TANK

Make sure you visit the PPMD Store while you're at the Conference and stock up on our new End Duchenne swag!

Celebrate the moms and dads in your life with our great new t-shirts and tank tops. Everyone has **One Tough Mother** or a **Super Dad** in their life that needs a t-shirt that recognizes just how awesome they are. Or maybe you just want to celebrate you!

You'll score a hole-in-one (see what we did there?) with PPMD's brand new golf shirt! This exclusive **Adidas Climacool Golf Shirt** is embroidered with our PPMD logo and features both moisture-wicking fabric and UPF 50+ UV protection. Climacool offers strategically placed mesh ventilation for cooling. Even if you don't play golf, you can raise awareness and look sharp in this great new shirt!

And don't forget to stock up on PPMD's stylish and practical **Race to End Duchenne** long-sleeve running shirts, windbreakers, and hats! We have great running gear for athletes of all experience and for every season of the year.

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