

# Rare Disease Research

*Providing Exceptional Solutions for Clinical Trials in Rare Diseases*

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# Who are we?

- *Rare Disease Research* (RDR) is an independent research center that specializes in clinical trials for patients with neurogenetic disorders
  - **Duchenne Muscular Dystrophy**
  - Spinal Muscular Atrophy
  - Limb-Girdle Muscular Dystrophy
  - Myasthenia Gravis
  - Niemann Pick Type C
  - Epilepsy
  - Cerebral Palsy
  - Metachromatic Leukodystrophy, and more
- As an independent research center, we are able to work with any sponsor, indication, or study
  - Even studies for a single patient!!

# Who are we?

## OUR MISSION

To **provide access to investigational treatments** for pediatric and adult patients with rare conditions that otherwise would not have the opportunity to participate in cutting-edge clinical research efforts

## OUR VISION

Cultivating **trustworthy relationships** with our collaborators in order to facilitate access to investigational therapies **for ALL our patients** with Rare Diseases

# Our DMD Trials

*Actively recruiting or upcoming*

# PTC 124-GD-048-DMD



<b>Phase</b>	Phase 1
<b>Sponsor</b>	PTC Therapeutics
<b>Investigational medication</b>	Ataluren (PTC124), orally administered powder 3x daily
<b>Trial Schedule</b>	24 weeks of open label treatment, optional 52-week extension  Visits: Screening, baseline, and Weeks 4, 12, and 24
<b>Key Inclusion</b>	<ul style="list-style-type: none"><li>• ≥6 Months to &lt;2 Years</li><li>• Nonsense mutation of dystrophin gene</li></ul>



# 4658-402 “MIS51ON”



<b>Phase</b>	Phase 3b
<b>Sponsor</b>	Sarepta Therapeutics
<b>Investigational medication</b>	Eterlipsen, IV infusion once weekly
<b>Trial Schedule</b>	Part 1: 4 - 144 weeks of open label treatment Part 2: up to 144 weeks of double-blind treatment
<b>Key Inclusion</b>	<ul style="list-style-type: none"><li>● 4 - 13 years old, inclusive</li><li>● Out-of-frame deletion mutation of the DMD gene amenable to exon 51 skipping</li><li>● Able to walk independently without assistive devices</li></ul>



# CAP-1002-DMD-04 “HOPE-3”

<b>Phase</b>	Phase 3
<b>Sponsor</b>	Capricor Therapeutics
<b>Investigational medication</b>	CAP-1002 Human Allogeneic Cardiosphere-Derived Cells, IV infusion OR placebo
<b>Trial Schedule</b>	4 infusions of CAP 1002 or placebo over 3 months, optional 12 month extension
<b>Key Inclusion</b>	<ul style="list-style-type: none"><li>• At least 10 years of age</li><li>• Ambulatory OR non-ambulatory</li><li>• Treatment with glucocorticoids for at least 12 months</li></ul>



# Dyne 251-DMD-202 “DELIVER”

<b>Phase</b>	Phase 1/2
<b>Sponsor</b>	Dyne Therapeutics
<b>Investigational medication</b>	DYNE 251 OR placebo, IV infusion
<b>Trial Schedule</b>	24-Week placebo-controlled period 24-week open-label period 96-week long term extension
<b>Key Inclusion</b>	<ul style="list-style-type: none"><li>● 4-16 years of age, inclusive</li><li>● Ambulatory or non-ambulatory</li><li>● Receiving stable dose of glucocorticoids</li><li>● Mutation amenable to Exon 51 skipping</li></ul>



# C3391003



<b>Phase</b>	Phase 3
<b>Sponsor</b>	Pfizer
<b>Investigational medication</b>	PF-06939926 OR placebo
<b>Trial Schedule</b>	1 IV infusion on Days 1 and 390
<b>Key Inclusion</b>	<ul style="list-style-type: none"><li>● <math>\geq 4</math> to <math>&lt; 8</math> years of age</li><li>● Receiving a stable daily dose of prednisone or deflazacort</li><li>● Ambulatory</li></ul>



# SRP 5051-201- “Momentum”



<b>Phase</b>	Phase 2
<b>Sponsor</b>	Sarepta Therapeutics
<b>Investigational medication</b>	SRP 5051, IV infusion
<b>Trial Schedule</b>	Part B: infusion every 4 weeks for up to 2 years
<b>Key Inclusion</b>	<ul style="list-style-type: none"><li>● 7 to 21 years of age</li><li>● amenable to exon 51-skipping</li><li>● Stable dose of corticosteroids for at least 12 weeks</li></ul>



# Our DMD Trials

*No longer enrolling*

# EDG-5506-002 “ARCH”

<b>Phase</b>	Phase 1b
<b>Sponsor</b>	Edgewise Therapeutics
<b>Investigational medication</b>	EDG 5506, daily oral dose
<b>Trial Schedule</b>	12 month open label treatment period
<b>Key Inclusion</b>	<ul style="list-style-type: none"><li>● **Becker muscular dystrophy**</li><li>● Ambulatory at screening</li></ul>



# “Epidys”



<b>Phase</b>	Phase 3
<b>Sponsor</b>	Italfarmaco
<b>Investigational medication</b>	Givinostat OR placebo
<b>Trial Schedule</b>	18 months of treatment
<b>Key Inclusion</b>	<ul style="list-style-type: none"><li>• At least 6 years of age</li><li>• Ambulatory</li><li>• Systemic corticoid use for at least 6 months</li></ul>

# Fibrogen 093, 094, and 079

<b>Phase</b>	Phase 3
<b>Sponsor</b>	Fibrogen
<b>Investigational medication</b>	Pamrevlumab or placebo
<b>Trial Schedule</b>	52 weeks of pamrevlumab or placebo administered every 2 weeks

# Reach out to us!

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