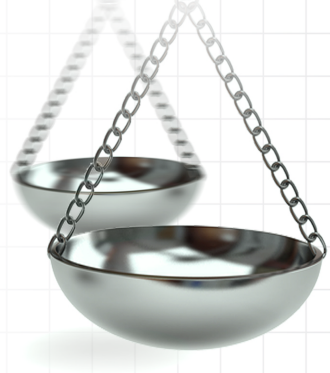
Plain Language Summary of Publication

Ataluren for the treatment of people living with nonsense mutation Duchenne muscular dystrophy: a plain language summary of Study 041



Journal of Comparative Effectiveness Research

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Summary

What is this summary about?
This article describes results from Study 041. Study 041 was a clinical study of ataluren, a treatment for people living with nonsense mutation Duchenne muscular dystrophy (nmDMD for short). Over time, people living with nmDMD experience muscle loss and reduced muscle strength and function (decline in muscle function). The researchers wanted to know whether 72 weeks of treatment with ataluren slowed the decline in muscle function in a large group of people living with nmDMD. To determine this, they compared the effects of ataluren with the effects of a placebo, which is a treatment that looks the same as the study treatment but has no active ingredients. The study also looked at the safety of ataluren over the 72 weeks of treatment. Ataluren has previously been compared with placebo in 48-week long clinical trials.

How to say (double click on the sound icon to play the sound)
Ataluren: a-ta-LEWR-ren
Corticosteroid: KOR-tih-ko-STEH-roid
Dystrophin: DIS-tro-fin
Duchenne muscular dystrophy: doo-SHEN MUH-skyoo-lur DIH-struh-fee

What were the results?
In this study, 359 people took at least one dose of ataluren or placebo, even if they later switched or didn't stick to the treatment plan. Over 72 weeks, ability to walk and physical function declined less in people receiving ataluren than in people receiving placebo. This means that their physical abilities were maintained for longer if they took ataluren. The number of side effects were generally similar between people who took ataluren and those who took placebo.

What do the results of Study 041 mean?
These results help to confirm that 72 weeks of treatment with ataluren (compared with 72 weeks of placebo treatment) slows down the decline in muscle function experienced by people living with nmDMD. Results also confirmed that side effects reported were similar with ataluren and placebo. These results likely show that people who receive ataluren can maintain their independence for a longer time. This study is the largest phase 3 clinical study of people living with nmDMD that has been done so far.

Who sponsored this study and this summary?

Study 041 and this plain language summary were **sponsored** by PTC Therapeutics.

Sponsor: a company or organization that oversees and pays for a clinical research study.

Becaris PUBLISHING

Where can readers find more information?

The original article discussed in this summary is titled 'Confirmatory long-term efficacy and safety results of ataluren in patients with nmDMD from Study 041, an international, randomized, double-blind, placebo-controlled, phase III trial'. The original article is free to read at: https://becarispublishing.com/doi/full/10.57264/cer-2024-0238

Who is this article for?

This plain language summary is intended for people living with DMD, their families and caregivers, and healthcare professionals who care for people living with DMD.

Living with Duchenne muscular dystrophy?

What is Duchenne muscular dystrophy?

Duchenne muscular dystrophy is also known as DMD. DMD is a **genetic condition**, meaning that it is caused by changes in a **gene**. A change in a gene is called a **mutation**. People living with DMD have changes in the *DMD* gene. These changes may be passed down from the mother to their sons (inherited), but may sometimes happen randomly.

These changes in the *DMD* gene stop the body from making a **protein** called dystrophin properly. Dystrophin is needed to keep muscles healthy and allows them to work properly. DMD causes muscle weakness and reduced muscle strength and function over time. It mainly affects boys. People living with DMD require specialist support at all stages as their condition progresses.

Several different changes in the *DMD* gene can occur; some cause the dystrophin protein to be too small and some mean it cannot be made at all. Some patients with DMD have a change called a **nonsense mutation**. This change signals the body to stop making the dystrophin protein before it is finished. This results in the formation of dystrophin that does not work properly. This form of DMD is called nonsense mutation DMD (nmDMD).

There is no cure for DMD; however, there are treatments available to help to treat symptoms and help to maintain muscle function. Some treatments are targeted for specific types of *DMD* gene changes. Improved medical care means that quality of life and life expectancy is improving for people living with DMD.

Duchenne muscular dystrophy (DMD for short): a rare, genetic muscle disorder that most commonly affects boys. Several genetic changes cause DMD, one of which is a nonsense mutation.

Rare condition (or disease): a health problem affecting a small percentage of the population.

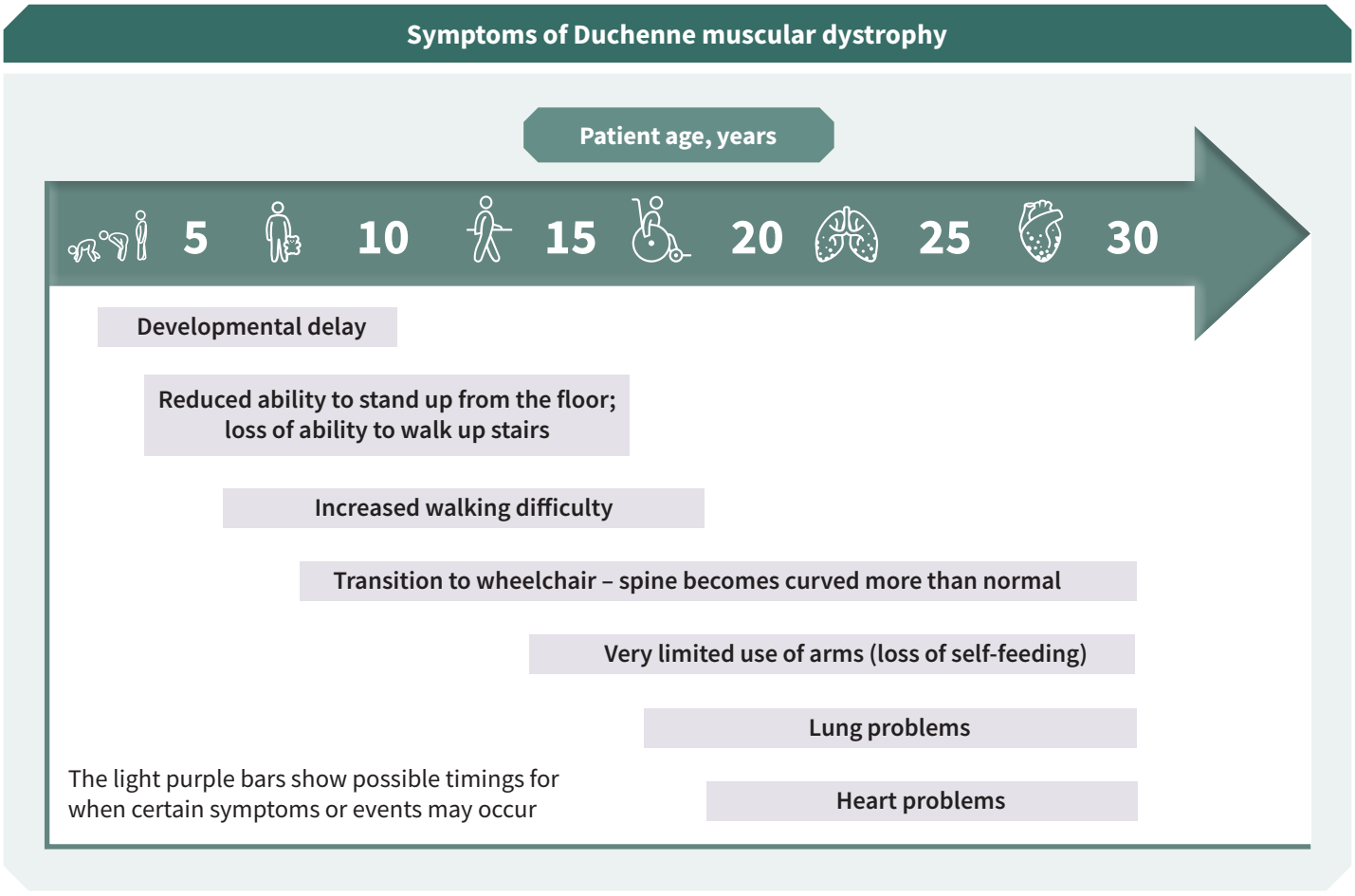
Genetic condition (or disease): a health problem caused by changes (mutations) in DNA that affect genes and alter the instructions for the body's growth, function or development.

Gene: a piece of DNA that tells the body how to make proteins. It can be passed down from biological parents to children.

Mutation: a change in a gene.

Proteins: building blocks of the body, made up of smaller pieces called amino acids. The sequence of amino acids is determined by DNA.

Nonsense mutation: a specific type of change in a gene that alters its instructions for making a protein. This change means that the cell stops making a protein before it is complete, resulting in a version of the protein that does not work properly.



Ataluren: a treatment for people living with nmDMD

What is ataluren and how does it work?

Ataluren is a treatment for people who have nmDMD and is approved in several countries. Only people with this type of DMD can receive ataluren.

Under normal conditions (without nmDMD), a 'go sign' in the *DMD* gene signals **mRNA** to be made. This mRNA translates instructions from the *DMD* gene so that the cell can make dystrophin protein. In people with nmDMD, a nonsense mutation creates a 'stop sign' earlier than normal in the gene. This stop sign is like having a full stop in the middle of a sentence: if the sentence ends too early then it does not make sense, and so likewise, the cell cannot properly understand the gene's instructions for making dystrophin. The early 'stop sign' signals the cell to stop making dystrophin protein before it is finished. This results in the formation of dystrophin that does not work properly. Ataluren works by enabling dystrophin to be made properly. Ataluren comes in a sachet and is taken by mouth three times a day.

Ataluren: a treatment for people living with nonsense mutation Duchenne muscular dystrophy that is taken by mouth three times a day.

mRNA: a molecule made from a DNA template. It translates instructions from genes during the process that cells use to make proteins

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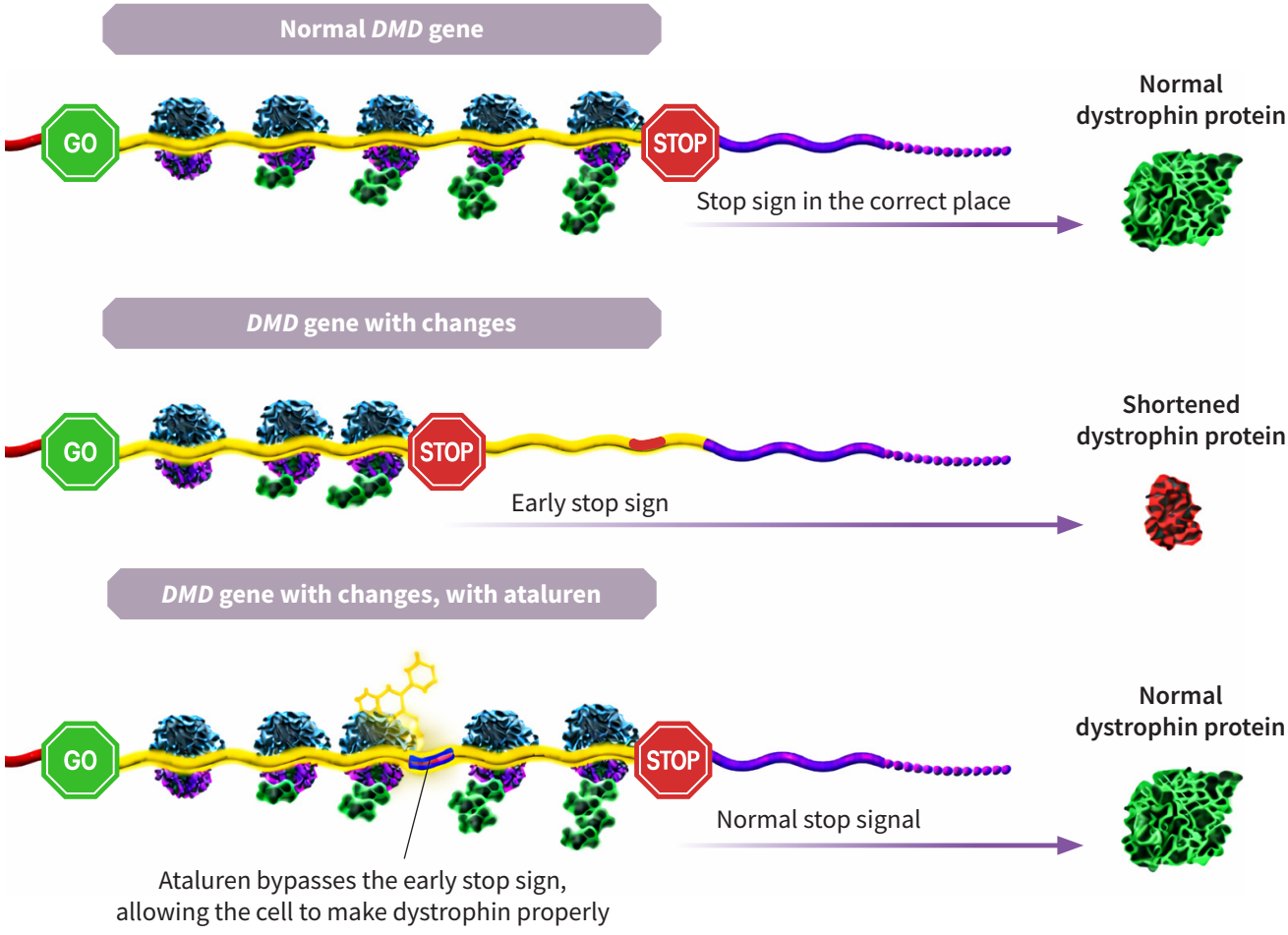
How does ataluren work?



About Study 041: a study of ataluren in people living with nmDMD

Why was this study done?

Study 041 was a **phase 3 clinical trial** to assess the effects of ataluren in people living with nmDMD. The researchers wanted to confirm that ataluren slows muscle function decline (its **efficacy**), and to look at the side effects of ataluren (its **safety**) over 72 weeks of treatment. The researchers also wanted to assess ataluren over a longer time period and in a larger number of people than in previous studies, which were 48 weeks long.

Phase 3 clinical trial: a large study that compares an active study treatment to a placebo or standard treatment. These studies look at whether the study treatment is safe and if it works effectively.

Efficacy: the ability of the active study treatment to improve the condition being studied.

Safety: monitoring of any expected and unexpected health issues that may be linked to the study treatment.

What happened in this study?

To make sure this assessment was reliable, the study was **placebo-controlled**, **randomized** and **double-blind**.

- **Placebo-controlled** means that some of the people in the study received **placebo** instead of active study treatment. The sachet of placebo looked exactly like the sachet of ataluren but did not have any ataluren in it. Researchers used placebo to help to make sure that any of the effects they saw in the people who took ataluren were caused by the active study treatment.

Placebo: a treatment that looks the same as the study treatment but has no active ingredients. It is sometimes called a dummy treatment.

- **Randomized** means that the people in the study were allocated at random to their treatment (either ataluren or placebo three times a day for 72 weeks). This made sure that people's characteristics could not impact which treatment they received.
- **Double-blind** means that none of the people in the study, researchers, study doctors or other study staff knew which treatment each person was taking. Some studies are done this way to reduce **bias** because knowing what treatment each person is taking can affect the results of the study.

Who took part in this study?

A total of 360 males with nmDMD took part in Study 041. They were from 18 different countries or regions, and their average age was around 8 years. Around half of the people could walk between 300 and 400 metres in 6 minutes at the beginning of Study 041. Everyone included in the study was using **corticosteroids**. Standing up from the floor took 5 or more seconds for around two-thirds of the people in the study.

Bias: a predisposition that leads to an altered view of the true effects of the study treatment.

Corticosteroids (also called steroids): medicines that are used to reduce inflammation in the body. They mimic the effect of a hormone called cortisol that the body naturally produces. Current treatment guidelines recommended that people living with Duchenne muscular dystrophy take corticosteroids with ataluren to manage their symptoms.

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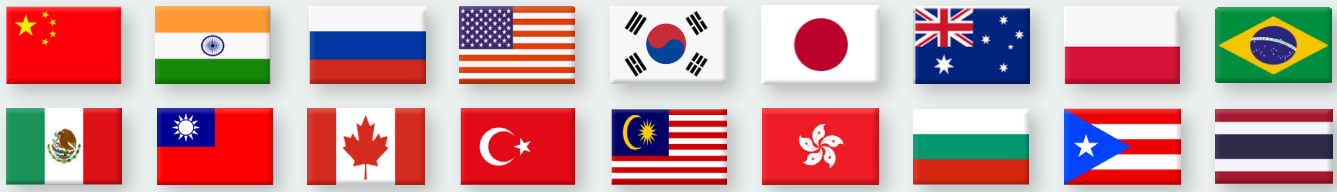
Study 041 design



To be included, people had to:

- Be **male** and aged **5 years or older**
- Have visible **signs of DMD**
- Have a **nonsense mutation** in the ***DMD* gene**

360 people from 18 countries received either ataluren or placebo, taken three times a day by mouth

One person in this group withdrew from the study before the first assessment after receiving one dose of ataluren

Ataluren
184 people

Placebo
176 people

360 people received at least one dose of ataluren or placebo. The researchers looked at the safety of ataluren in this group (**as-treated population**)

Ataluren
183 people

Placebo
176 people

359 people were assigned to take either ataluren treatment or placebo and had walking assessments at the start of the study and at least one visit after the study began (**intention-to-treat group**)

The intention-to-treat group included two smaller groups of people called **subgroups**

These were used to assess the effect of ataluren in people at particular stages of DMD

Subgroup 1
Aged 5 years or older

- People who could walk a distance of 300–400 metres in 6 minutes at the start of the study

Ataluren
86 people

Placebo
83 people

Subgroup 2
Aged 7–16 years

- People who could walk a distance of at least 300 metres in 6 minutes at the start of the study
- People who could not stand up from the floor in less than 5 seconds at the start of the study

Ataluren
92 people

Placebo
93 people

Of the 360 people who were randomized, just over half (51%) received ataluren and just under half (49%) received placebo. One person received a single dose of ataluren but did not complete any study assessments so was not included in the **intention-to-treat (ITT)** group. This means that 359 participants were included in the ITT group. This group included everyone who was originally assigned to take either ataluren treatment or placebo, even if they later switched or didn't stick to the plan. People in this group had walking assessments at the start of the study and at least one visit after the study began.

The ITT group included two smaller groups called **subgroups**.

Intention-to-treat (ITT) group: a group consisting of everyone who was originally assigned to take either the treatment or placebo, even if they later switched or didn't stick to the plan. People in this group had walking assessments at the start of the study and at least one visit after the study began.

Subgroup: a group of people within the overall population of a clinical study who have some characteristics in common.

What tests were done in this study?

Ability to walk or walking assessment

Ability to walk was assessed using a test called the **6-minute walking distance (6MWD)**, which measures the distance a person can walk in 6 minutes. When the distance someone can walk in 6 minutes gets shorter, it indicates that their walking ability is getting worse.

The researchers also measured the time it took for people's 6MWD to decline by 10% or 30 metres. This change in 6MWD can be used to predict when a person will lose their ability to walk and start using a wheelchair. Researchers have found that if walking ability gets worse over the year after this test, people are more likely to need a wheelchair 2 to 4 years later.

Physical function

Physical function was assessed using two different types of test.

The North Star Ambulatory Assessment (NSAA) required people to complete 16 tasks that assessed their muscle strength and physical function. Each task was scored on a scale from 0 to 2, with 0 being unable to complete the task and 2 being able to complete the task by themselves without any assistance or adjustments. A higher total score meant a better ability to perform a task. The NSAA test usually has 17 tasks, but this study did not include one task: testing the participant's ability to lift their head. This is because research showed that this task did not work like the others, so it was removed to make the results of the test more accurate.

Timed function tests (TFTs) assessed people's ability to walk or run 10 metres, walk up four stairs and walk down four stairs. A shorter time meant a faster ability to perform the test.

6-minute walking distance (6MWD for short): the distance a person can walk in 6 minutes.

North Star Ambulatory Assessment (NSAA for short): a test with 17 tasks that assesses overall muscle strength and physical function. This study used a revised NSAA test with 16 tasks.

Timed function tests (TFTs for short): tests that assess muscle strength and physical function through the ability to walk or run 10 metres, stand up from the floor and walk up/walk down four stairs.

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Function of the upper limbs, including the shoulders, arms, wrists and hands

Researchers tested the function of upper limbs using the **Performance of Upper Limb (PUL)** test. This test included tasks that assessed the function of the people's shoulders, arms, wrists and hands. Each task was scored on a scale from 0 to 2, with 0 being unable to complete a task and 2 being able to complete a task independently without any assistance or adjustments. A higher score meant a better ability to perform a task.

The PUL test used in this study included 21 tasks related to everyday activities. For example, bringing a hand to the mouth, lifting heavy cans and picking up tokens.

Performance of Upper Limb (PUL) test: a test that assesses how well people's shoulders, arms, wrists and hands work

Safety of ataluren

The researchers also looked at the safety of ataluren and kept track of all the unexpected health issues that people had during the study.

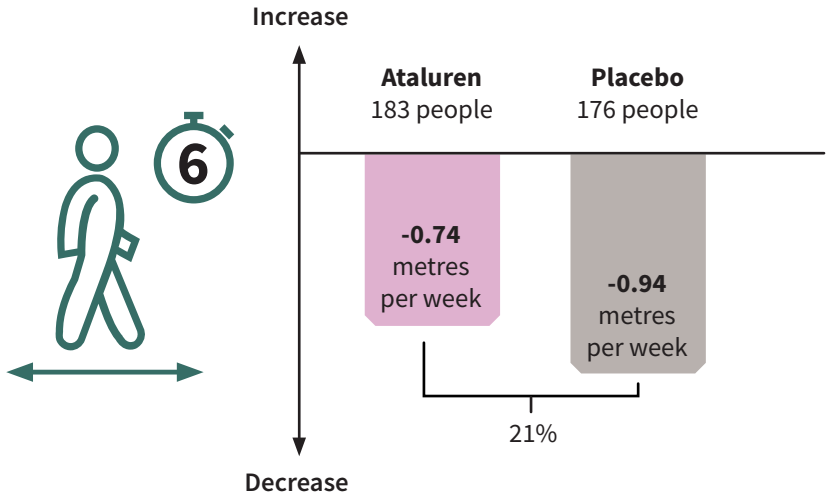
The COVID-19 pandemic overlapped with Study 041, which meant that some tests were delayed and had to be done after 72 weeks in some cases.

What were the results of this study?

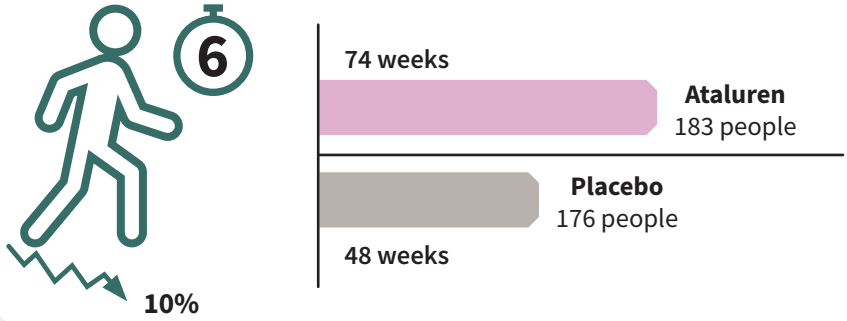
Ataluren slowed people's decline in their ability to walk compared with placebo in the ITT group and subgroup 1

ITT group

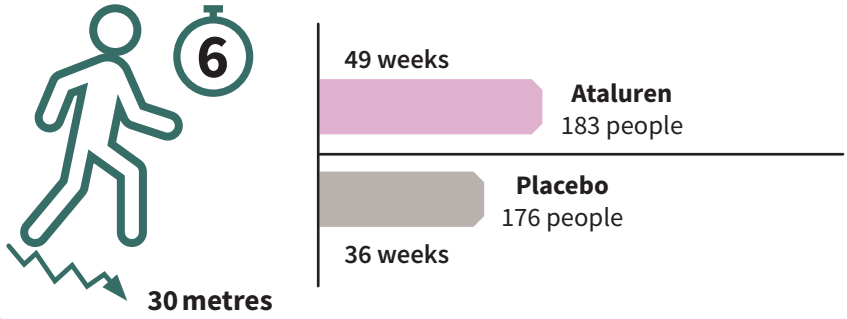
People who received ataluren had a slower decline in 6MWD than those who received placebo. This means that walking ability declined at a slower rate in people taking ataluren than in those taking placebo. These results show that treatment with ataluren may enable people to maintain their ability to walk for longer compared with placebo.



- **Walking ability** (6MWD) in people who received ataluren **declined by 0.74 metres per week** during the 72 weeks of the study
- This was a **21% slower decline** than in people who received placebo



- It took **longer for walking ability** (6MWD) **to decline by 10%** in people who received ataluren than in people who received placebo



- It took **longer for walking ability** (6MWD) **to decline by 30 metres** in people who received ataluren than in people who received placebo

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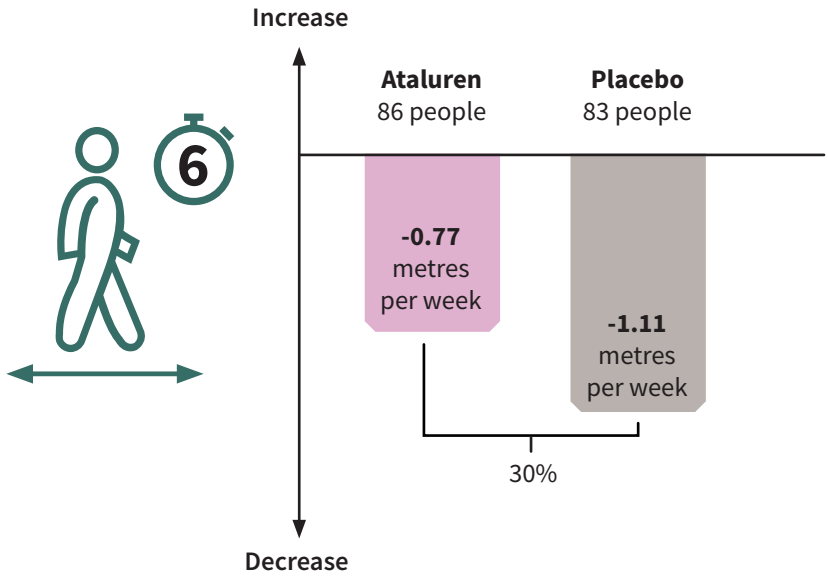
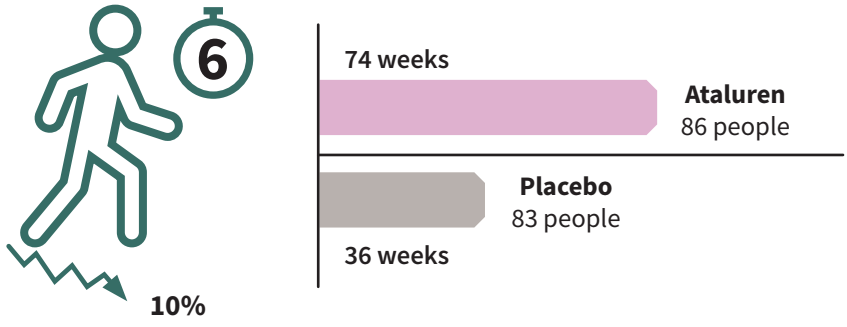
Subgroup 1

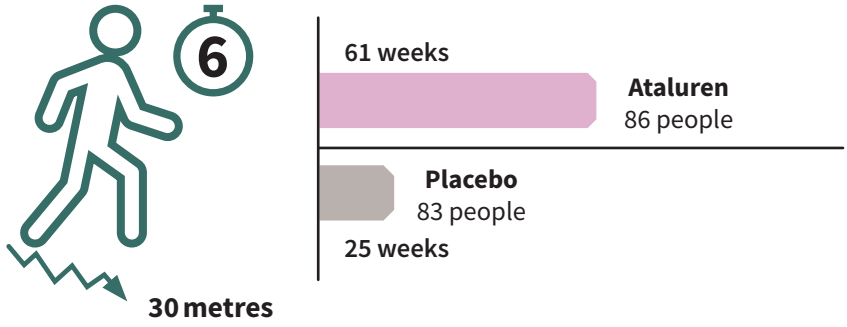
People in subgroup 1 who received ataluren also had a slower decline in 6MWD than those who received placebo. The difference in this subgroup was even greater than in the ITT group, meaning that people in subgroup 1 benefitted more from ataluren treatment than those in the ITT group.



- **Walking ability** (6MWD) in people who received ataluren **declined by 0.77 metres per week** during the 72 weeks of the study
- This was a **30% slower decline** than in people who received placebo



- It took **longer for walking ability** (6MWD) **to decline by 10%** in people who received ataluren than in people who received placebo

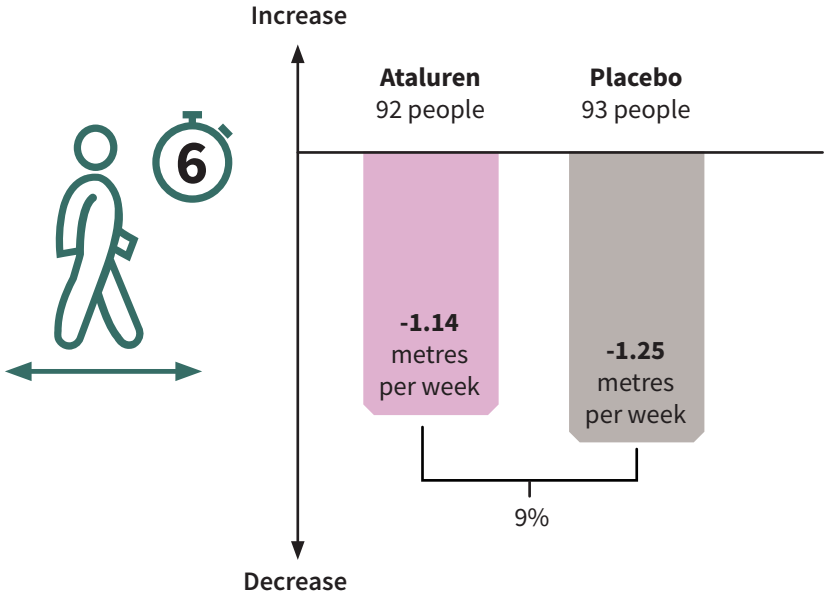


- It took **longer for walking ability** (6MWD) **to decline by 30 metres** in people who received ataluren than in people who received placebo

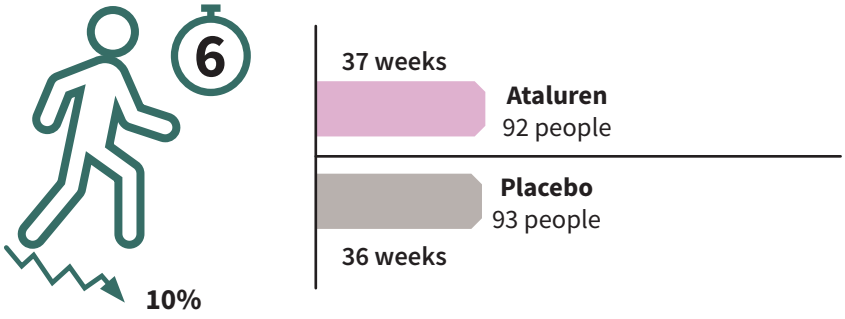
Subgroup 2

The researchers saw small differences in the decline in 6MWD between the people who received ataluren and those who received placebo; however, these differences were too small to conclude that they resulted from treatment with ataluren.

The time taken for people's 6MWD to decline by 30 metres was not measured in this subgroup. This was because the changes seen in the other 6MWD results were not big enough between people who received ataluren and those who received placebo.



- **Walking ability** (6MWD) in people who received ataluren **declined by 1.14 metres per week** during the 72 weeks of the study
- This was a **9% slower decline** than in people who received placebo



- The time taken **for walking ability** (6MWD) **to decline by 10% was similar** in both groups

Ataluren slowed some people's natural decline in physical function

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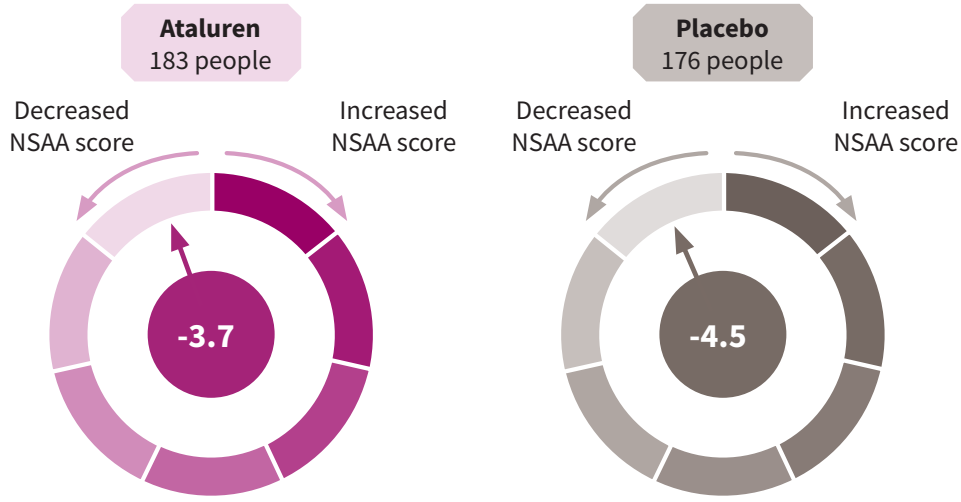
NSAA during the study

ITT group

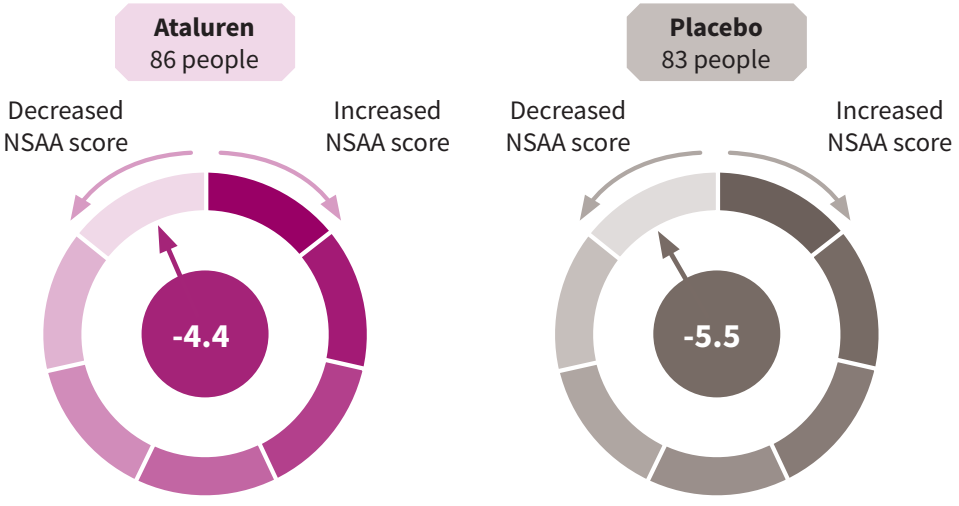
NSAA scores showed that people who received ataluren maintained the ability to perform NSAA functional tasks for longer than people who received placebo. The decline in NSAA total score was about 19% smaller in the people who received ataluren than in those who received placebo.



- People who received ataluren had a **smaller decline in NSAA total score** (-3.7) than people who received placebo during the 72 weeks of the study
- This shows that they **maintained the ability to perform functional tasks** for longer with ataluren than with placebo

Subgroup 1

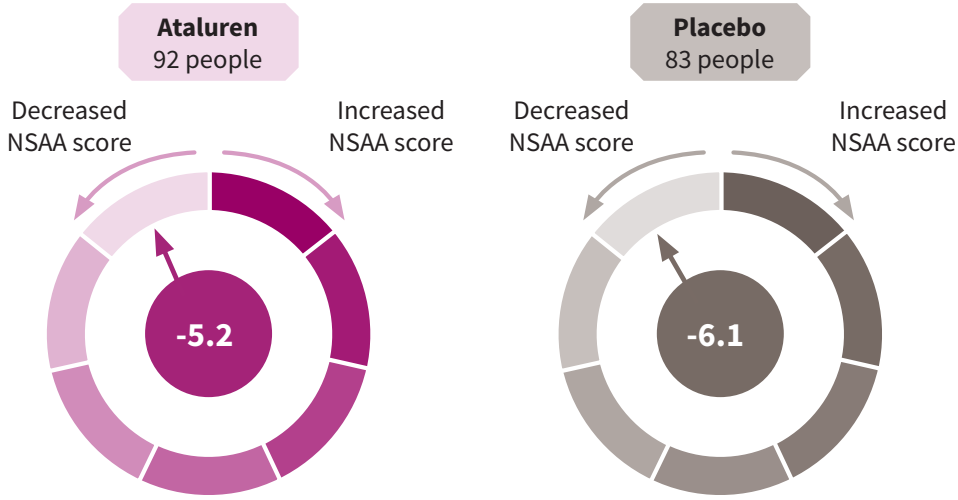
People in subgroup 1 who received ataluren had a smaller decline in their ability to perform NSAA functional tasks than those who received placebo; however, the difference was too small to conclude that it resulted from treatment with ataluren.



- People who received ataluren had a **smaller decline in NSAA total score** (-4.4) than people who received placebo during the 72 weeks of the study
- However, the difference was too small to conclude that it resulted from treatment with ataluren

Subgroup 2

People in this subgroup who received ataluren had a smaller decline in their NSAA total score than those who received placebo; however, the difference in decline was too small to conclude that it resulted from treatment with ataluren.



- People who received ataluren had a **smaller decline in NSAA total score** (-5.2) than people who received placebo during the 72 weeks of the study
- However, the difference was too small to conclude that it resulted from treatment with ataluren

TFTs during the study

ITT group

The time it took to walk or run 10 metres and to walk up four stairs did not increase as much for people who received ataluren as for those who received placebo. This means that physical ability declined more in people receiving placebo than in those receiving ataluren. The difference between ataluren and placebo in the time to walk down four stairs was too small to conclude that it resulted from treatment with ataluren.

	Walk or run 10 metres (average time in seconds)	**Walk up four stairs** (average time in seconds)	**Walk down four stairs** (average time in seconds)
Ataluren 183 people	↑ by 3.04	↑ by 4.98	**Small difference**
Placebo 176 people	↑ by 3.82	↑ by 6.04	**Small difference**

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Subgroup 1

The time it took to walk or run 10 metres and to walk up four stairs did not increase as much in people who received ataluren as in those who received placebo. There were some differences observed between the ataluren and placebo groups in the change in time to walk down four stairs; however, these differences were too small to conclude that they resulted from treatment with ataluren.

	Walk or run 10 metres (average time in seconds)	Walk up four stairs (average time in seconds)	Walk down four stairs (average time in seconds)
Ataluren 86 people	↑ by 2.99	↑ by 5.26	**Small difference**
Placebo 83 people	↑ by 4.28	↑ by 7.55	**Small difference**

Subgroup 2

The time it took to walk up four stairs did not increase as much in people who received ataluren as in those who received placebo. There were some differences between the ataluren and placebo groups in the change in time to walk or run 10 metres and the time to walk down four stairs; however, these differences were too small to conclude that they resulted from treatment with ataluren.

	Walk or run 10 metres (average time in seconds)	Walk up four stairs (average time in seconds)	Walk down four stairs (average time in seconds)
Ataluren 92 people	**Small difference**	↑ by 5.25	**Small difference**
Placebo 93 people	**Small difference**	↑ by 6.98	**Small difference**

Ataluren slowed some people's decline in ability to perform everyday activities that rely on shoulders, arms, wrists and hands

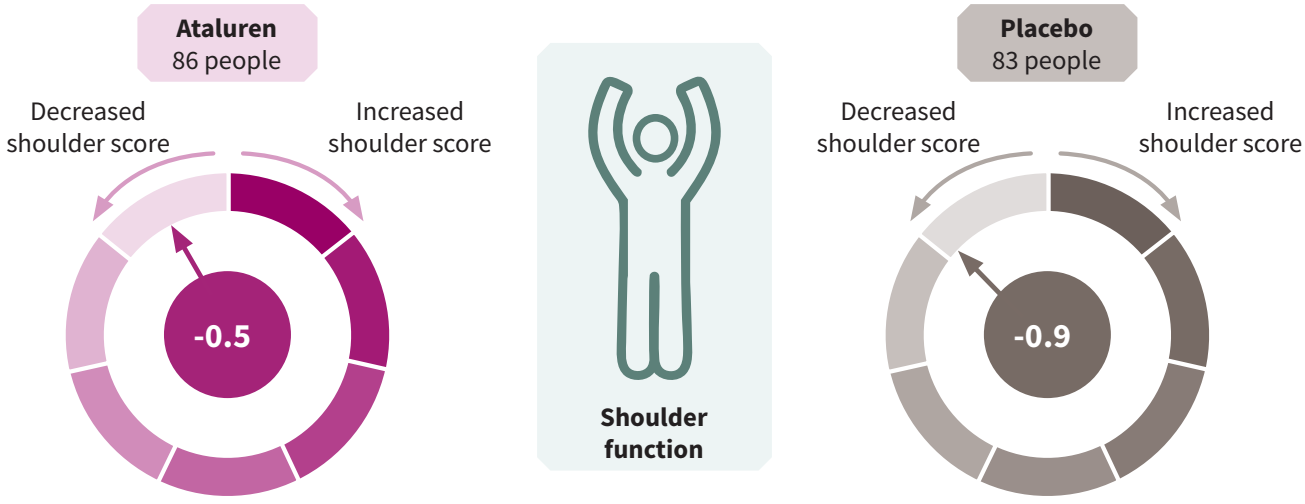
ITT group

Changes in the scores for upper limb tasks (measured by the PUL test) were similar for people who received ataluren and those who received placebo. This means that function of upper limbs declined at around the same rate in both groups.

Subgroup 1

People in this subgroup who received ataluren had a smaller decline in shoulder and elbow function than those who received placebo. This means that people who received ataluren were able to maintain their shoulder and elbow function for longer than those who received placebo.

Decline in shoulder function scores



Decline in elbow function scores

There were some differences observed in activities relating to hand and wrist function between people who received ataluren and those who received placebo; however, these differences were too small to conclude that they resulted from treatment with ataluren.

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Subgroup 2

Changes in upper limb scores were not tested in this subgroup. This is because the differences observed in 6MWD, NSAA and TFT results between people who received ataluren and those who received placebo were not big enough.

The benefits of taking ataluren are thought to outweigh the risks of experiencing any side effects related to the treatment

Unexpected health issues that occur during a clinical study are called **adverse events** and may or may not be caused by the treatments in the study.

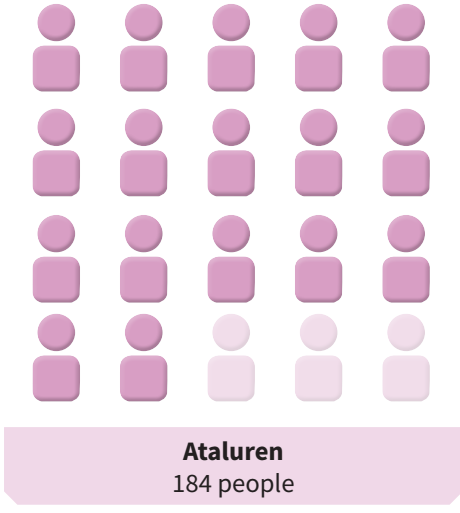
An adverse event is considered **serious** when it is life-threatening, causes lasting problems or requires hospital care. This summary will discuss two types of adverse events that are described below.

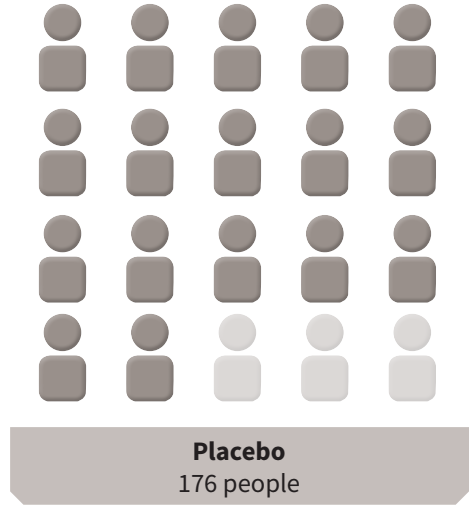
- **Treatment-emergent adverse events:** unexpected health issues that occur after people receive their treatment, but that are not necessarily linked to this treatment.
- **Treatment-related adverse events:** unexpected health issues that people experience that are thought to be linked to the study treatment.

Adverse events: unexpected health issues that people experience that may be associated with a treatment.

Treatment-emergent adverse events

Around **17 out of every 20 people** (85%) in both groups had a treatment-emergent adverse event during the study





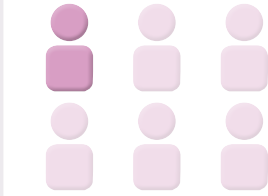
Most common adverse events

The most common adverse events in the study were:

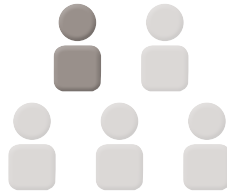
Adverse event	Ataluren 184 people	Placebo 176 people
Vomiting	16%	5%
Common cold	15%	7%
Throat and chest infection	15%	25%
Fall	10%	13%
Headache	10%	9%
Cough	10%	11%
Disease progression	6%	10%

Treatment-related adverse events

Around **1 out of every 6 people** (17%) in the ataluren group and **1 out of every 5 people** (21%) in the placebo group had a treatment-related adverse event during the study



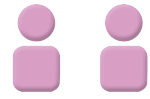
Ataluren 184 people



Placebo 176 people

Serious adverse events

2 people (1%) who took ataluren and nobody (0%) who took placebo had a **serious adverse event** that was **possibly related** to taking study treatment



Ataluren 184 people

Placebo 176 people

No one died during the study

Serious adverse event: an adverse event that is life threatening, causes lasting problems or requires hospital care.

10.57264/cer-2025-0184

What do the results of Study 041 mean?

	Improved ability with ataluren treatment versus placebo					
	Walking ability (6MWD)	Physical function (NSAA total score)	Physical function (TFT: time to walk or run 10 metres)	Physical function (TFT: time to walk up four stairs)	Physical function (TFT: time to walk down four stairs)	Everyday activities that rely on shoulders and elbows (PUL)
ITT group	✓	✓	✓	✓	Small difference	Small difference
Subgroup 1	✓	Small difference	✓	✓	Small difference	✓
Subgroup 2	Small difference	Small difference	Small difference	✓	Small difference	Small difference

Overall, this study confirmed that people who received ataluren were able to maintain their walking ability for longer than those who received placebo. They also showed smaller changes in other tests that measure physical function. This suggests that ataluren may help to slow the worsening of nmDMD. The results also confirmed that ataluren had side effects that people were generally able to manage.

What does this study mean to patients and caregivers?

This section has been authored by a caregiver of a person living with DMD.

When living with DMD, slowing down every aspect of motor function decline for as long as possible is of utmost importance. Walking for longer is meaningful to the boys with DMD and their families because it prolongs the boys' independence and freedom while delaying carer responsibilities. Requiring a lower level of care directly correlates with the boys' ability to access community resources independently, leading to greater inclusion.

Delays in motor function decline also mean that carers have more time to prepare themselves financially for expenses, such as durable medical equipment that is typically not covered by insurance providers, home modification and accessible transportation, all of which help to ease the eventual burden of care.

Delaying wheelchair use may also help to maintain heart and lung function for a longer period of time, which can contribute to the increased lifespan of boys with DMD. Once boys with DMD are in a wheelchair full-time, maintaining upper limb function becomes the primary focus of care. The loss of independent feeding, use of technology, gaming or ability to scratch an itch will all have major impacts on the level of care required and the level of independence. Delaying decline in all areas of motor function contributes greatly to the boys' and carers'/families' quality of life.

Where can I find more information about this study?

The original article discussed in this summary is titled 'Confirmatory long-term efficacy and safety results of ataluren in patients with nmDMD from Study 041, an international, randomized, double-blind, placebo-controlled, phase III trial'. The original article is free to read at: https://becarispublishing.com/doi/full/10.57264/cer-2024-0238

You can read more about this study at: https://www.clinicaltrials.gov/study/NCT03179631

Educational resources

You can find out more about DMD on the websites listed below.

- Parent Project Muscular Dystrophy: https://www.parentprojectmd.org/
- Cure Duchenne: https://cureduchenne.org/

Further reading

1. A publication on the findings of a workshop where doctors, researchers and parent representatives discuss the status of newborn screening for Duchenne muscular dystrophy around the world. Available at: Ellis JA, Vroom E & Muntoni F. 195th ENMC international workshop: newborn screening for Duchenne muscular dystrophy. 14–16th December, 2012, Naarden, The Netherlands. *Neuromuscular Disorders* 23(8), 682–689 (2013).

2. A review looking at how DMD is treated, how it gets worse over time, how severe it becomes, and how it affects people's quality of life. Available at: Ryder S, Leadley RM, Armstrong N *et al.* The burden, epidemiology, costs and treatment for Duchenne muscular dystrophy: an evidence review. *Orphanet Journal of Rare Diseases* 12(1), 1–21 (2017).

3. A review explaining what the dystrophin protein and related proteins normally do in muscles, and describing how these processes are disrupted in people with DMD. Available at: Blake DJ, Weir A, Newey SE & Davies KE. Function and genetics of dystrophin and dystrophin-related proteins in muscle. *Physiological Reviews* 82(2), 291–329 (2002).

4. Information, including a video, about the genetic causes of DMD. Available at: Genetic Causes. Available from: https://www.parentprojectmd.org/about-duchenne/what-is-duchenne/genetic-causes/ (Accessed: November 2025).

5. A review explaining what causes DMD, how it damages muscles, and outlining current and emerging treatments. Available at: Bez Batti Angulski A, Hosny N, Cohen H *et al.* Duchenne muscular dystrophy: disease mechanism and therapeutic strategies. *Frontiers in Physiology* 14, 1183101 (2023).

6. A publication looking at the life expectancy in people with DMD. Available at: Broomfield J, Hill M, Guglieri M, Crowther M & Abrams K. Life expectancy in Duchenne muscular dystrophy: reproduced individual patient data meta-analysis. *Neurology* 97(23), e2304–e2314 (2021).

7. A publication explaining what ataluren, also called PTC124, is. Available at: Welch EM, Barton ER, Zhuo J *et al.* PTC124 targets genetic disorders caused by nonsense mutations. *Nature* 447(7140), 87–91 (2007).

8. An article looking at new treatments for people with DMD, with a focus on how genetic testing can help to guide more personalized treatment approaches. Available at: Bello L & Pegoraro E. Genetic diagnosis as a tool for personalized treatment of Duchenne muscular dystrophy. *Acta Myologica* 35(3), 121–127 (2016).

9. An article looking at new treatments for people with DMD, with a focus on how genetic testing can help to guide more personalized treatment approaches. Available at: Pichavant C, Aartsma-Rus A, Clemens PR *et al.* Current status of pharmaceutical and genetic therapeutic approaches to treat DMD. *Molecular Therapy* 19(5), 830–840 (2011).

10. A publication of one of the 48-week-long studies of ataluren in patients with nmDMD aged 5 years or older. Available at: Bushby K, Finkel R, Wong B *et al.* Ataluren treatment of patients with nonsense mutation dystrophinopathy. *Muscle Nerve* 50(4), 477–487 (2014).

11. A publication of one of the 48-week-long studies of ataluren in patients with nmDMD aged 5 years or older. Available at: McDonald CM, Campbell C, Torricelli RE *et al.* Ataluren in patients with nonsense mutation Duchenne muscular dystrophy (ACT DMD): a multicentre, randomised, double-blind, placebo-controlled, phase 3 trial. *The Lancet* 390(10101), 1489–1498 (2017).

12. A study looking at how changes in walking distance relate to quality of life in boys with DMD. Available at: Henricson E, Abresch R, Han JJ *et al.* The 6-minute walk test and person-reported outcomes in boys with Duchenne muscular dystrophy and typically developing controls: longitudinal comparisons and clinically-meaningful changes over one year. *PLoS Currents,* doi: 10.1371/currents.md.9e17658b007eb79fcd6f723089f79e0 5 (2013).

13. A study looking at whether simple timed movement tests can predict loss of key abilities, such as standing and walking, in boys and young men with DMD. Available at: McDonald CM, Duong T, Henricson E *et al.* P. 2.11 CINRG Duchenne Natural History Study: relationship of longitudinal measures of ambulatory timed function tests and loss of clinical milestones. *Neuromuscular Disorders* 23(9), 752 (2013).

14. An article explaining how the NSAA test is used to assess overall muscle strength and physical function in people with DMD. Available at: Mayhew AG, Cano SJ, Scott E *et al.* North Star Clinical Network for Neuromuscular Disease. Detecting meaningful change using the North Star Ambulatory Assessment in Duchenne muscular dystrophy. *Developmental Medicine and Child Neurology* 55(11), 1046–1052 (2013).

15. A long-term study tracking how walking and physical function change over time in people with DMD. Available at: Arora H, Willcocks RJ, Lott DJ *et al.* Longitudinal timed function tests in Duchenne muscular dystrophy: imagingDMD cohort natural history. *Muscle & Nerve* 58(5), 631–638 (2018).

16. An article explaining how the PUL test works to measure shoulder, arm, wrist and hand function in people with DMD. Available at: Diella E, LoMauro A, Delle Fave M, Cima R & D'Angelo MG. The Performance of Upper Limb (PUL) module in limb-girdle muscular dystrophy. *Acta Myologica* 41(4), 207–211 (2022).

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