POMPE
SUPPORT NETWORK

Pompe Disease Treatments

January 2026 Edition

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Contents

Disclaimer:

The information in this booklet serves as a guide to help people with Pompe disease and parents or carers of children with Pompe disease make informed decisions about treatment and prepare for specialist appointments.

This guide does not provide medical advice; always consult your specialist doctor with any medical concerns.

Links and QR codes to external sites are provided for convenience and information purposes. These links do not imply endorsement, and the Pompe Support Network has no control over external content. For questions about external site content, please contact the respective site directly.

Current treatments

The treatments currently available for Infantile Onset Pompe disease (IOPD) and Late Onset Pompe disease (LOPD) are enzyme replacement therapy (ERT).[1]

Trade name	Myozyme® [2]	Nexviadyme® [3]	Pombiliti® [4] plus Opfolda® [5]
Other name	alglucosidase alfa	avalglucosidase alfa	cipaglucosidase alfa plus miglustat
Year approved	2006	2022	2023
Type of Pompe disease	All types	All types	LOPD only
Age of patient	All ages	All ages	18 years or older

How are they given?

ERTs are given as an **intravenous infusion** (through a drip into a vein) **once every 2 weeks**. These infusions take around 3-6 hours.[1]

The dose of ERT you will receive depends on your weight. The standard dose for Myozyme®, Nexviadyme® and Pombiliti® is 20mg of ERT for every kilogram you weigh.[1]

Dose example

If the recommended dose is **20mg per kg of body weight**, then a patient who weighs **50kg** is going to receive **1000mg** of ERT at each infusion.

Higher dose Nexviadyme®

A higher dose (**40mg** per kg every two weeks) may be considered in **Infantile Onset Pompe Disease** patients who are not showing improvement on the standard dose.[1]

Opfolda®

Opfolda® capsules are taken one hour before the Pombiliti® infusion.[5] The dose is:

- Three 65mg capsules, if you weigh between 40kg and 50kg
- Four 65mg capsules, if you weigh 50kg or more.

Eating must be avoided for 2 hours before and after taking Opfolda®, but you can drink water, skimmed cow's milk and tea or coffee without cream, sugars or sweeteners.

How do they work?

People with Pompe disease do not have enough of an enzyme called acid alpha-glucosidase (GAA). This enzyme helps to break down glycogen, a type of sugar that your body stores in muscles to use as energy when needed. Without enough GAA, glycogen builds up and damages the muscles. ERTs work like GAA to help reduce the build up of glycogen.[1]

What is the difference between the ERTs?

- Myozyme® was the **first ERT for Pompe disease.** Two newer, second generation ERTs are also available.
- Nexviadyme® has modifications that improve how well it is absorbed by muscle cells and therefore its ability to break down glycogen.[6]
- Pombiliti® has been designed to improve how well it is absorbed by muscle cells. It is used alongside another medication called Opfolda® that protects Pombiliti® in the blood so that more can reach the muscle cells to break down glycogen.[4,5,7]

What are the possible side effects?

Like all medicines, Myozyme®, Nexviadyme®, Pombiliti® and Opfolda® can cause side effects, though not everyone will have them. The most common ones are mild or moderate and usually occur during or soon after the infusion.[2-5]

Because serious infusion or allergic reactions can happen, your first ERT infusions are given in hospital so staff can monitor you closely.[1]

Side effects can occur at any time, not just on treatment days. Known side effects are listed in each medicine's *Patient Information Leaflet* (see links below), but there is a small chance you might experience a side effect that has not been reported before and is not listed in the leaflet.

If you experience any side effects, contact your doctor. If the side effects are severe, call 999 or go to the nearest emergency department immediately.

Information on the safety of medicines is continually collected and reviewed. The list of known side effects is listed on a medicine's *Patient Information Leaflet* and updated if needed as more people use the medicine over time.

Where can I find more information?

Talk to your Pompe specialist about available treatments. Our booklets *My Child's Pompe Treatment* and *My Pompe Treatment* (at www.pompe.uk) include useful questions to ask. You can also view *Patient Information Leaflets* below.

Myozyme® Nexviadyme® Pombiliti® Opfolda®

If you are unable to scan the QR codes, links are provided on page 8.

How do these treatments compare?

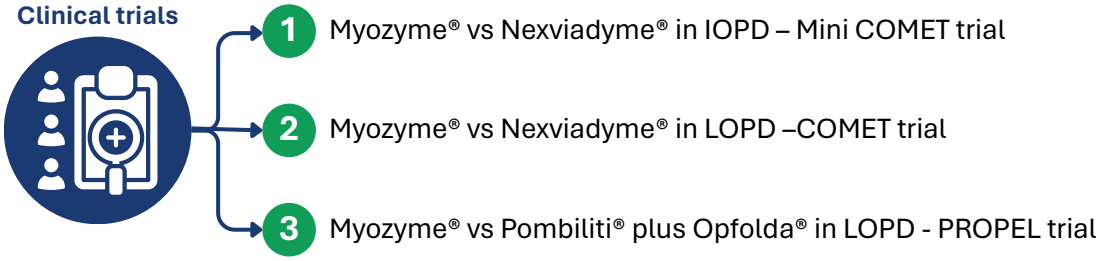
Myozyme® can help with breathing, walking, muscle strength, and reducing fatigue. It can also help people with Pompe disease to stay more active, lower the chance of needing a wheelchair and reduce the risk of death.[8-14]

The newer treatments have been studied in comparison to Myozyme® in three clinical trials.[6,7,15,16] They have not yet been directly compared to each other. Instead, researchers have combined results from separate trials in people with LOPD to see how they might compare.[17,18]

Current evidence does not suggest than one ERT is better than another in any particular situation. Your specialist will discuss the different options with you and help decide the most suitable treatment, based on the latest UK guidelines and your individual circumstances.

Comparing treatments in clinical trials

Three clinical trials have compared different treatments for Pompe disease.[6,7,15,16]



Understanding the results of clinical trials?

Clinical trials are designed to answer a question about a treatment - for example, how safe it is, how well it works, the best dose to use or how it compares to existing treatments. To do this, researchers choose a main measure called the **primary endpoint**. The trial is planned carefully so that the results can be tested to see if they are statistically significant—meaning the results are likely to be real and not just the result of chance.

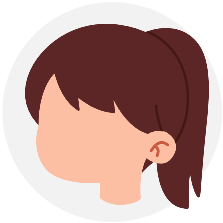
Trials may also include additional endpoints that provide more insight into how the treatment works. These are not the main focus, so their results may not always be tested for statistical significance.

One example of an endpoint in Pompe disease clinical trials is how much farther a person can walk, measured by the 6-minute walk test.

Myozyme® vs Nexviadyme®

Mini-COMET trial

The mini-COMET trial: Summary



This trial included 22 **children with IOPD** aged between 1-12 years who had been treated with Myozyme® for at least 6 months. They were divided into 4 treatment groups and received an infusion every 2 weeks for 6 months.[15]

11 children whose condition had **worsened** on Myozyme®		11 children who were **not doing as well as expected** on Myozyme®	
GROUP 1 **Nexviadyme®** **20mg/kg** 6 children	GROUP 2 **Nexviadyme®** **40mg/kg** 5 children	GROUP 3 **Nexviadyme®** **40mg/kg** 5 children	GROUP 4 **Myozyme®** **20-40mg/kg** 6 children

Primary endpoint and result

The primary endpoint was safety of Nexviadyme®. **Similar safety results** were seen between treatment groups, and no serious or severe **adverse events** were reported.

Additional endpoints and results

Any differences between treatments were not proven as they were not tested for statistical significance.

Motor function: Children in groups 1 and 2 stabilised or improved on Nexviadyme®. Those in groups 3 and 4, mostly stabilised or improved, with little difference between the two treatments.

6-minute walk test: Some children (aged 6+ and able to walk at the start of the trial) on Nexviadyme® 40mg/kg improved, those on Nexviadyme® 20mg/kg or Myozyme® stayed the same or did worse.

Breathing: No child needed to start invasive ventilation during the trial.

Heart size: Stayed normal or improved for all.

Number with droopy eyelids (ptosis): Increased in group 1 but was stable or reduced in the other groups.

Biomarkers: CK levels tended to fall with either dose of Nexviadyme® and were stable with Myozyme®. **Hex4** levels dropped most with 40mg/kg Nexviadyme® and were more variable with the lower dose or Myozyme®.

Adverse event: Any unwanted or harmful effect that happens while taking a medicine. It might not always be caused by the medicine.

Motor function: The ability to move and control your muscles.

Biomarkers: Substances measured in the blood and urine, that indicate how well a treatment is working.

CK: Lower levels of creatine kinase (CK) mean there is less muscle damage and better muscle function.

Hex4: Lower levels mean the body is breaking down glycogen better, helping to improve muscle function.

The researchers concluded that Nexviadyme® (40 mg/kg) may improve outcomes for children who have declined or not benefitted as expected on Myozyme® treatment.

Children in the Myozyme® group were on a mixture of different doses, making comparisons between treatments difficult.

Myozyme® vs Nexviadyme®

COMET trial

The COMET trial: Summary

This trial included 100 people with **LOPD** who had never received ERT for their Pompe disease and included one child. All the other participants were 18 years of age or older. They were treated for 49 weeks by infusion every 2 weeks.[6,16]

49 people received Myozyme® 20mg/kg

51 people received Nexviadyme® 20mg/kg

Primary endpoint and result

The study was designed to see if Nexviadyme® was no worse than Myozyme® in treating LOPD, by testing for changes in breathing muscle function over the 49 weeks of the trial. **Nexviadyme® was not less effective than Myozyme®.**

Another statistical test, known as superiority testing, was done to see if Nexviadyme® was better than Myozyme®. **Nexviadyme® was not** shown to be **more effective than Myozyme®** in this test.

Additional endpoints and results

Any differences between treatments for these additional endpoints were not proven as they were not tested for statistical significance.

Breathing, muscle strength and endurance: Those treated with Nexviadyme® tended to show more improvement than those taking Myozyme® in:

- Breathing muscle strength
- Arm and leg muscle strength
- 6-minute walk test

Motor function: On average, those treated with Nexviadyme® did better on motor function tests than those treated with Myozyme®.

Quality of life: Those receiving Nexviadyme® tended to report feeling better overall compared with those receiving Myozyme®.

Biomarkers: Levels reduced with both treatments with a trend towards more reduction with Nexviadyme®.

Safety: No new safety signals for Nexviadyme® were reported.

Forced Vital Capacity: The total amount of air a person can breathe out forcefully after taking a deep breath.

The researchers concluded that Nexviadyme® provided improvements in breathing, motor function, and endurance compared to Myozyme®, with no new safety signals.

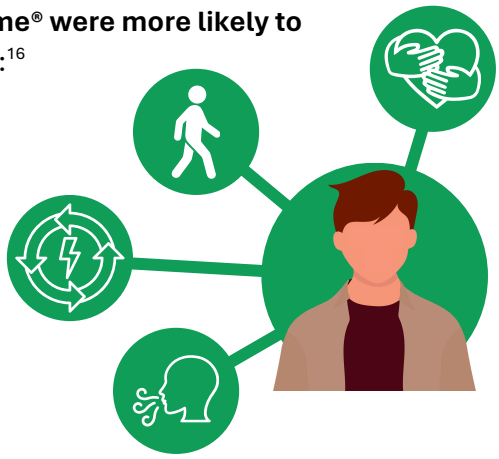
Further studies may be needed to confirm any differences between the treatments.

A review of this trial concluded: Nexviadyme® likely improves the 6-minute walk test compared with Myozyme®. There is probably little or no difference between the two in lung function (measured by **Forced Vital Capacity**), quality of life, the risk of allergic reactions, and number of side effects.[19]

More on quality of life from the COMET trial

In a separate analysis of the results, **people taking Nexviadyme® were more likely to notice meaningful improvements in** several areas, including:[16]

- **Breathing** – feeling less short of breath
- **Energy and comfort** – less tiredness and pain
- **Morning headaches** – fewer or less severe headaches
- **Movement and independence** – improved mobility and ability to manage daily activities
- **Overall wellbeing** – feeling better about general health and Pompe symptoms



These results were statistically significant.

My treatment notes

Myozyme® versus Pombiliti® plus Opfolda®

PROPEL trial

The PROPEL trial: Summary

This trial included 125 **adults with LOPD** who had never received ERT for their Pompe disease or had been treated with Myozyme® for at least 2 years. They were split into 2 groups and both received an infusion and a capsule every 2 weeks for one year.[7]

40 Myozyme® 20mg/kg plus a placebo capsule

85 Pombiliti® 20mg/kg plus Opfolda® capsule

Primary endpoint and result

The study was designed to see if Pombiliti® plus Opfolda® was better than Myozyme® in treating LOPD, by testing for changes in the 6-minute walk test. Although these results improved more for those treated with Pombiliti® plus Opfolda®, the **difference was not statistically significant.**

Additional endpoints and results

Any differences between treatments for these additional endpoints were not proven as they were not tested for statistical significance.

Breathing: Those receiving Pombiliti® plus Opfolda® tended to show more improvement in lung function, compared to Myozyme®, in five tests. In one test, patients on Myozyme® tended to show the most improvement.

Muscle strength: There was a trend to more improvement with Pombiliti® plus Opfolda® in all but one measure, where better results were achieved with Myozyme®.

Motor function: People treated with Pombiliti® plus Opfolda® appeared to show more improvement than those taking Myozyme® in all the tests apart from one.

Quality of life: Improvements in physical function and fatigue tended to be greater for those receiving Pombiliti® plus Opfolda®.

Biomarkers: A greater reduction of CK and Hex4 was recorded with Pombiliti® plus Opfolda®.

Safety: The number of adverse events were similar in both groups.

Patients who had been treated with Myozyme® before the trial. Results were broadly similar to those in the total trial population.

Patients who had not received ERT before the trial. Results were varied with more tending to favour Myozyme® compared to the total population. This included the 6-minute walk test, 2 out of 6 tests for lung function, most of the muscle strength and motor function tests and both of the quality of life measures.

The researchers felt that Pombiliti® plus Opfolda® may have potential benefits over Myozyme® in supporting lung function and in patients who had received more than two years of ERT.

More research is needed to explore these potential benefits.

A review of the PROPEL trial concluded: Pombiliti® plus Opfolda® may not improve walking distance but probably improves lung function, when compared to Myozyme® plus placebo. There appears to be little or no difference between the two treatments in quality of life, risk of allergic reactions, or number of side effects.[19]

Nexviadyme® versus Pombiliti® plus Opfolda®

Summary

No clinical trials have directly compared Nexviadyme® to Pombiliti® plus Opfolda®. To date the only way these treatments have been compared is by using statistical methods, comparing published information from clinical trials of Nexviadyme® and Pombiliti® plus Opfolda® in LOPD. Two of these statistical comparisons have been conducted with differing results.[17,18]

It is important to remember that comparing results across different trials can be difficult as each trial may have been designed differently and involved different groups of patients.

As experience with these treatments grows, a clearer understanding of their relative benefits should develop.

Future treatments for Pompe disease

Researchers are still working on new and better treatments for Pompe disease. If you would like to stay up to date you can check trusted patient organization websites:

- Pompe Support Network: www.pompe.uk
- Association for Glycogen Storage Disease (AGSD) UK: www.agsd.org.uk

If you are **interested in taking part in a clinical trial, you can talk to your Pompe specialist** about this. You can also find out more about clinical trials on the following websites:

NHS search tool to find out about clinical trials in the UK

- www.bepartofresearch.nihr.ac.uk

International clinical trial registers

- www.clinicaltrials.gov
- www.clinicaltrialsregister.eu/ctr-search/search
- www.trialsearch.who.int

QR code links

The safety of medicines:
www.nhs.uk/tests-and-treatments/medicines-information/#:~:text=Safety%20of%20medicines

Patient Information Leaflets for each treatment:
Myozyme®: www.medicines.org.uk/emc/product/263/pil#about-medicine
Nexviadyme®: www.medicines.org.uk/emc/product/14562/pil#about-medicine
Pombiliti®: www.medicines.org.uk/emc/product/14898/pil#about-medicine
Opfolda®: www.medicines.org.uk/emc/product/14904/smpc#gref

Pregnancy and breastfeeding

Anyone who is pregnant or breastfeeding, think they may be pregnant, or are planning to have a baby, should ask their doctor for advice before using these medicines.

Information regarding pregnancy and breastfeeding from the *Patient Information Leaflet* for each treatment is summarised below. This information can change as experience of using these medicines grows. It is therefore important to speak to your doctor for the most up to date information and advice that's right for you and your situation.

Myozyme®

There is limited experience of the use of Myozyme® in pregnancy or when breastfeeding.[2]

- You should not be using Myozyme® during pregnancy unless clearly necessary.
- There is limited experience suggesting that Myozyme® passes into human breast milk in very small amounts. No effect on the breastfeed infant is expected.
- Breastfeeding during the treatment may be considered. However, you can discuss with your doctor whether to interrupt breastfeeding as a precautionary measure for the first 24 hours after each dose of Myozyme®.
- Anyone who is pregnant or breastfeeding, think they may be pregnant, or are planning to have a baby, should ask their doctor or pharmacist for advice before using this medicine.

Nexviadyme®

There is no information about use in pregnancy.[3]

- You must not receive Nexviadyme® during pregnancy unless your doctor specifically recommends it.
- You and your doctor should decide if you can use Nexviadyme® if you are breastfeeding.
- Anyone who is pregnant or breastfeeding, think they may be pregnant, or are planning to have a baby, should ask their doctor or pharmacist for advice before using this medicine.

Pombiliti® plus Opfolda®

There is no experience of use in pregnancy.[4,5]

- You should not receive Pombiliti® and / or take Opfolda® if pregnant.
- Tell your doctor immediately if you are pregnant or breastfeeding, think that you may be pregnant, or if you are planning to become pregnant. There may be risks to the unborn baby.
- Pombiliti® in combination with Opfolda® should not be given to women who are breastfeeding. A decision will need to be made whether to stop treatment or to stop breastfeeding.

Medical references

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POMPE
SUPPORT NETWORK

Based in the United Kingdom, we are a network of individuals, families, scientists and healthcare professionals who aim to improve the lives of all people living and working with Pompe disease. The network is run by members of the Pompe community, for the benefit of the Pompe community.

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