Australian Government
Department of Health and Aged Care

Guidelines for the treatment of infantile-onset and late-onset Pompe disease through the Life Saving Drugs Program

Life Saving Drugs Program

About this program

Through the Life Saving Drugs Program (LSDP), the Australian Government provides subsidised access for eligible patients to expensive life-saving drugs.

Purpose of this document

This document provides guidance for treating physicians with relevant specialist registration who wish to apply for their patients to receive access to subsidised treatment for infantile-onset and late-onset Pompe disease* through the LSDP.

It describes the criteria for general, initial and ongoing eligibility to access subsidised treatment and the administrative requirements associated with the initial application and annual reapplications.

Treatment of Pompe disease through the LSDP

Subsidised treatment is available for eligible patients with a confirmed diagnosis of infantile-onset or late-onset Pompe disease.

Drugs currently available for the treatment of Pompe disease through the LSDP

There are two drugs currently subsidised through the LSDP for the treatment of infantile-onset and late-onset Pompe disease.

The generic names for these drugs are alglucosidase alfa and avalglucosidase alfa. The trade names for these drugs are Myozyme® and Nexviazyme®.

The Therapeutic Goods Administration (TGA) registration and Product Information for alglucosidase alfa (Myozyme®) and avalglucosidase alfa (Nexviazyme®) can be found on the TGA's website.

Choice of treatment

Treating physicians can request the most appropriate drug to treat their patient.

* The classification of Pompe disease is now either infantile-onset or late-onset. Late-onset is inclusive of both juvenile-onset and adult-onset. This change was made following the LSDP Review of Pompe disease.

In line with the Product Information, avalglucosidase alfa can only be used for patients who are one year of age and older.

A treating physician of an existing LSDP patient receiving alglucosidase alfa can request that their patient be switched to avalglucosidase alfa through a clinic letter provided to the LSDP. A full application is not required for these patients. This would also apply to an existing LSDP patient receiving avalglucosidase alfa switching to alglucosidase alfa.

Dosage

The maximum dosage of alglucosidase alfa and avalglucosidase alfa that is subsidised through the LSDP is 20 mg/kg per fortnight.

Home infusion

For patients receiving avalglucosidase alfa, home administration by a trained health care professional may be considered for individual patients after safety and tolerability has been established in the clinical setting.

General eligibility requirements

LSDP funding conditions

A patient must continually meet the LSDP funding conditions in order to be eligible to receive access to Australian Government-subsidised treatment for Pompe disease through the LSDP.

The current LSDP funding conditions can be found on the program's website.

For infantile-onset or late-onset Pompe disease, a patient must:

- satisfy the initial and ongoing eligibility criteria as detailed in these guidelines
- participate in the evaluation of effectiveness of the drug by periodic assessment, as directed by these guidelines, or have an acceptable reason not to participate
- not be suffering from any other medical condition, including complications or sequelae of infantile-onset or late-onset Pompe disease, that might compromise the effectiveness of the drug treatment
- be an Australian citizen or permanent Australian resident who qualifies for Medicare.

In most cases, participation in a clinical trial will not affect a patient's eligibility to access LSDP medicines. However, treating physicians are required to advise the LSDP if their patient is participating in a clinical trial.

Exclusion criteria

The following patients are not eligible for subsidised treatment with alglucosidase alfa or avalglucosidase alfa for the treatment of infantile-onset or late-onset Pompe disease through the LSDP:

- Patients on long-term invasive ventilation for respiratory failure prior to starting enzyme replacement therapy (ERT) which indicates a disease severity that will not benefit from treatment (patients requiring acute ventilation for conditions such as cardiac failure or acute respiratory infection should not be excluded).

- Patients with the presence of another life-threatening or severe disease where the prognosis is unlikely to be influenced by ERT.
- Patients with the presence of another medical condition that might reasonably be expected to compromise a response to ERT.
- Current smokers.

Initial eligibility requirements

Eligible patients

At the time of initial application the patient is aged up to 24 months and has a documented diagnosis of infantile-onset Pompe disease; or

At the time of initial application the patient is aged over 24 months and has a documented diagnosis of late-onset Pompe disease. Patients aged 18 years and over must also present with at least one of the following treatment criteria[†]:

- Respiratory function test: Patients with Forced Vital Capacity (FVC), either supine or erect, less than 80% of predicted value. Both supine and erect FVC should be performed.
- Sleep disordered breathing: Patients with an apnoea/hypopnoea incidence of >5 events/hour of total sleep time or more than two severe episodes of desaturation (oxygen saturation <80%) in an overnight sleep study.
- Significant muscular weakness: Patients with significant muscular weakness as evidenced by Manual Muscle Testing (MMT) (employing the MRC score) of 4 or less in either limb girdle accompanied by a 6 Minute Walk Test (6MWT).

Diagnosis

Diagnosis of infantile-onset or late-onset Pompe disease must have been made using one of the following methods:

- Documented deficiency of acid alpha-glucosidase by prenatal diagnosis using chorionic villus biopsies and/or cultured amniotic cells; or
- At least 2 of the following confirmatory diagnostic tests from a NATA-accredited laboratory:
  - Documented deficiency of acid alpha-glucosidase in dried blood spot or lymphocytes or mixed leukocytes or skin fibroblasts or skeletal muscle.
  - Documented urinary tetrasaccharide testing indicating a diagnostic elevation of glucose tetrasaccharides.
  - Documented molecular genetic testing indicating a disease-causing mutation in the acid alpha-glucosidase gene (GAA gene).

See the initial application form.

Testing is not funded or subsidised through the LSDP, however some tests may be subsidised through Medicare or available through the treating public hospital.

[†] For patients receiving avalglucosidase alfa (Nexviazyme®) before 1 September 2022, treating physicians can liaise with the LSDP for additional information on the application process. Patients receiving alglucosidase alfa on the LSDP and switching to avalglucosidase alfa (Nexviazyme®) do not need to fulfil initial treatment criteria. The treating physician can request the treatment change through the LSDP via a clinic letter.

Ongoing eligibility requirements

The treating physician must submit the separate reapplication form to the LSDP by 1 May every year if they wish their patient to continue to receive subsidised treatment through the LSDP.

The reapplication form must demonstrate clinical improvement in the patient or stabilisation of the patient's condition, and evidence to support ongoing eligibility for the treatment of infantile-onset or late-onset Pompe disease must be provided.

The treating physician must declare that the patient continues to meet the eligibility criteria to receive subsidised treatment through the LSDP in accordance with these guidelines.

Subsidised treatment may continue unless one or more of the following situations apply:

- Failure to comply adequately with treatment or measures.
- Failure to provide data, copies of test results and the Excel spreadsheet for infantile-onset or late-onset Pompe disease, evidencing the effectiveness of the therapy.
- Therapy fails to relieve the symptoms of disease that originally resulted in the patient being approved for subsidised treatment.
- Evidence of disease progression despite regular therapy, including but not limited to the development of the need for 24 hour invasive ventilation, for a period of 14 days or greater, provided that:
    - the cardiorespiratory failure is progressive
    - the requirement for ventilation is not due to a potentially reversible problem such as infection; and
    - muscle tone is so poor that there is no useful movement.
- Development of a life-threatening complication, which would compromise the effectiveness or benefit from continued ERT, including the patient has severe infusion-related adverse reactions or antibody-related reactions which are not preventable or controlled by appropriate pre-medication and/or adjustment of infusion rates.
- The patient develops another life-threatening or severe disease where the long-term prognosis is unlikely to be influenced by ERT.
- The patient develops another medical condition that might reasonably be expected to compromise a response to ERT.
- For late-onset Pompe patients aged over 18 years a decline of 20% or greater versus prior year on measures of 6MWT or a decline of 10% or greater versus prior year on measures of erect FVC. To ensure consistency of testing, the test should be undertaken again within 3 months. Test results of 6MWT and FVC must be provided to LSDP at the first 6 months of initial treatment and thereafter annually. A review of the data to evaluate these parameters will commence 2 years after the initial subsidy was provided.

See the reapplication form for existing patients.

Patients who are applying to recommence treatment following a break should use the reapplication form.