Life Saving Drugs Program (LSDP) updated clinical testing requirements for Pompe disease, Gaucher disease (type 1) and for mucopolysaccharidosis (MPS) types I, II and VI

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Introduction

The LSDP Expert Panel (the Expert Panel) has reviewed the initial and ongoing eligibility requirements for Pompe disease, Gaucher disease (type 1), and mucopolysaccharidosis (MPS) types I, II and VI. These reviews were an outcome of the Expert Panel's LSDP Medicines Review Recommendations, which aimed to ensure the use and performance of LSDP medicines are in line with the recommendations and expectations at the time of listing.

In its review the Expert Panel recommended that the purpose, clinical benefits and frequency of diagnostic/clinical monitoring tests required for LSDP applications and reapplications should be reviewed. The Expert Panel recommended that treating physicians of LSDP patients would be best placed to comment on the appropriateness of the testing required for LSDP eligibility.

Appropriateness of current clinical tests

To further review the appropriateness of the current testing requirements for Pompe disease, Gaucher disease (type 1), and MPS types I, II and VI, the Department of Health and Aged Care (the department) consulted with the specialist treating physicians of LSDP patients. The Expert Panel considered their feedback as well as guidance from the National Referral Laboratory on the appropriateness and accuracy of specific tests alongside contemporary international approaches and the current literature.

The Expert Panel found that some of the tests required were unnecessary to demonstrate ongoing treatment eligibility. The Expert Panel noted that clinical testing imposes a burden on patients requiring LSDP medicines in attending specialist clinics for these tests, and that remote and rural patients are further disadvantaged by accompanying difficulties accessing specialist clinics in the major cities.

The Expert Panel recommended that the clinical tests requested for both the initial and ongoing applications for subsidised treatment through the LSDP should better align with clinical practice and inform the assessment of ongoing eligibility by demonstrating improvement or stabilisation in response to treatment. It further recommended that all data collected should have predefined purpose.

Expert Panel Recommendations

As a result of its reviews the Expert Panel has recommended changes to the testing requirements for Pompe disease, Gaucher disease (type 1), and MPS types I, II, and VI,

which have now been implemented by the department. The revised testing requirements are expected to streamline patient access to the program and reduce the burden of clinical testing to patients and treating physicians without compromising program integrity. The amended eligibility requirements summarised below aim to streamline program administration and enhance future medicines reviews.

Condition Specific Clinical Testing Changes

Details are provided below on the changes to testing requirements, including links to updated application and reapplication documents. If you require clarification or further information on the updated requirements, please contact lsdp@health.gov.au or 02 6289 2336.

Pompe disease

Changes to Infantile-onset Pompe Disease (IOPD) testing requirements

1. The following test requirements have been removed:
    a. Body mass index
    b. Cerebral magnetic resonance imaging (MRI)
    c. Blood tests which are not informative about response to treatment and which are not part of regular clinical practice, including tests for:
        i. Immunoglobulin G
        ii. Immunoglobulin E
        iii. Haemoglobin
        iv. Platelets
        v. Alanine Amino Transferase
        vi. Aspartate Amino Transferase
        vii. Lactate Dehydrogenase
2. The requirement prescribing a specific cardiac test has been amended to requesting a clinically appropriate cardiac test. Tests can include a chest X-ray, echocardiogram, electrocardiogram (ECG), 24-hour ECG and/or paediatric cardiology assessment at the treating physician's discretion.

Changes to Late-onset Pompe disease (LOPD) testing requirements

3. The tests required for ongoing eligibility for LOPD patients are now linked to the criterion/criteria under which they were admitted to the program.
    - For example, if a patient was admitted under the 'significant muscle weakness' criterion, the treating physician is required to provide test results for manual muscle testing and the 6-minute walk test (6MWT) on initial application and every reapplication. The respiratory function test and overnight sleep study test results are only required if the treating physician considers them to be clinically indicated.
4. In keeping with clinical practice for patients aged 18 years and over, both erect and supine FVC tests results are now required in reapplications.
5. Clarification has been provided on the requirement for patients under 18 years of age to provide forced vital capacity (FVC) and 6MWT tests:
    - not required for children between the ages of 2 and 4 years

- should be attempted (if the child is able to cooperate) for children between the ages of 5 to 11 years, and
- should be attempted for children between the ages of 12 and 17 years.

These changes have been reflected in updated patient test results spreadsheets. Consequential amendments have also been made to both the initial application and reapplication forms, which have been further updated to help streamline the application process. Treating physicians are encouraged to familiarise themselves with the new requirements and the Pompe disease Guidelines before submitting a new patient application or reapplication.

Gaucher disease (type 1)

Changes to Gaucher disease (type 1) testing requirements.

1. Chitotriosidase testing has been removed from the requirements with glucosyl sphingosine (lyso-Gb-1) testing being retained as the most appropriate biomarker to determine Gaucher disease severity.
2. The requirement for the ophthalmologic review and neurodevelopmental status in patients under 16 years of age has been removed.
3. Spleen and liver size and volume are only required to be reported until the measure is stable. The treating physician may choose the most appropriate way to assess this, such as palpation or imaging.
4. Skeletal magnetic resonance imaging (MRI) is now only required annually until the measure is stable. Once stable an MRI will only be required once every 2 years.
5. The assessment for pain and/or quality of life (QoL) has been removed. This does not reflect the Expert Panel's opinion of the importance of this measure but rather reflects that eligibility for continued treatment is not dependent on changes to QoL. Treating physicians can undertake these assessments and provide the results to the LSDP if they choose to.

These changes have been reflected in updated patient test results spreadsheet. Consequential amendments have also been made to both the initial application and reapplication forms, which have been further updated to help streamline the application process. Treating physicians are encouraged to familiarise themselves with the new requirements and the Gaucher disease (type 1) Guidelines before submitting a new patient application or reapplication.

MPS type I, II and VI

Changes to MPS I, MPS II and MPS VI testing requirements.

1. The following test requirements have been removed:
    a. Body mass index
    b. Ophthalmological examinations
    c. Skeletal survey assessments
    d. 6-minute walk test
    e. Psychometric testing
    f. Neurological examinations

g. Audiology assessment
h. Urine glycosaminoglycans testing
i. Liver and spleen sizes.

2. The assessment for pain and/or quality of life (QoL) has been removed. This does not reflect the Expert Panel's opinion of the importance of this measure but rather reflects that eligibility for continued treatment is not dependent on changes to QoL. Treating physicians can undertake these assessments and provide the results to the LSDP if they choose to.
3. The tests required for ongoing eligibility are now linked to the criterion/criteria under which the patient was admitted to the program.
    - For example, if a patient was admitted under the 'joint contractures' criterion, the treating physician is required to provide test results for range of movement tests and hand clawing on the initial application and every reapplication until this measure is stable. They only need to provide other tests results if the treating physician considers them to be clinically indicated and if they demonstrate a response to treatment (i.e. stabilisation or improvement).
4. For patients aged 5 years or less not yet demonstrating symptoms consistent with other eligibility criteria, the treating physician should undertake whichever tests are deemed clinically relevant at baseline, and tests for ongoing eligibility should demonstrate whether there has been a response to treatment (i.e. stabilisation or improvement).

These changes have been reflected in updated test result spreadsheets:

- MPS I patient test results spreadsheet
- MPS II patient test results spreadsheet
- MPS VI patient test results spreadsheet.

Consequential amendments have also been made to:

- MPS I initial application and reapplication forms
- MPS II initial application and reapplication forms
- MPS VI initial application and reapplication forms

These forms have been further updated to help streamline the application process. Treating physicians are encouraged to familiarise themselves with the new requirements and the relevant guidelines before submitting a new patient application or reapplication:

- MPS I Guidelines
- MPS II Guidelines
- MPS VI Guidelines