

2137: Leukodystrophies Post-Infusion

The Leukodystrophies Post-Infusion Data Form is one of the Comprehensive Report Forms. This form captures Leukodystrophies post-HCT data for the reporting period.

This form must be completed for all recipients whose primary disease is reported on the Pre-TED Disease Classification (2402) Form as a leukodystrophy and specified as one of the following:

- Krabbe Disease (globoid cell leukodystrophy)
- Metachromatic leukodystrophy (MLD)
- Adrenoleukodystrophy (ALD)
- Hereditary diffuse leukoencephalopathy with spheroids (HDLS)

The Post-Infusion Leukodystrophies (2137) Form must be completed in conjunction with each Post-Infusion Follow-up (2100) Form. This form is designed to capture specific data occurring within the timeframe of each reporting period (i.e., between day 0 and day 100; between day 100 and the six-month date of contact for six-month follow-up; and between the date of contact for the six-month follow-up and the date of contact for the one-year follow-up, etc.).

Links to sections of the manual:

Manual Updates:

Sections of the Forms Instruction Manual are frequently updated. The most recent updates to the manual can be found below. For additional information, select the manual section and review the updated text.

If you need to reference the historical Manual Change History for this form, please reference the retired manual section on the [Retired Forms Manuals](#) webpage.

Date	Manual Section	Add/Remove/Modify	Description
7/26/2024	2137: Leukodystrophies Post-Infusion	Add	Version 2 of the Leukodystrophies Post-Infusion Data section of the Forms Instructions Manual released. Version 2 corresponds to revision 4 of the Form 2137.

Q1 – 5: Leukodystrophies Post-Infusion Data

If the primary disease for infusion is HDLS, manually select this option from *Specify the leukodystrophy subtype*. This option will not auto populate at this time.

Question 1: For which type of leukodystrophy was the infusion performed

Indicate the recipient's disease subtype:

- **Krabbe Disease (globoid cell leukodystrophy):** A rare lysosomal storage disorder affecting the nervous system, most typically occurring in infants.
- **Metachromatic leukodystrophy (MLD):** A rare genetic disorder in which there is an accumulation of sulfatides, causing damage to the myelin sheath within the nervous system. The three types of MLD are based on age symptoms occur: late-infantile MLD, juvenile MLD, and adult MLD.
- **Adrenoleukodystrophy (ALD):** A rare genetic disorder causing buildup of very long-chain fatty acids (VLCFAs) and in response, damages myelin sheath within the nervous system. The most common form of ALD is X-linked ALD, more commonly affecting males.
- **Hereditary diffuse leukoencephalopathy with spheroids (HDLS):** A rare hereditary disorder affecting adults. The disease is associated with leukoencephalopathy and spheroids in the axons of the brain.

Enzyme activity / enzyme substrate – recipient

Questions 2 – 3: Was enzyme activity and/or enzyme substrate tested

Indicate if the recipient's enzyme activity and / or enzyme substrate was tested during the current reporting period. The type of testing performed is specific to leukodystrophy subtype. Types of testing can include enzyme assays, urine testing, etc. An example of enzyme activity testing is the quantitative measurement of arylsulfatase A enzyme.

If enzyme activity / enzyme substrate was tested on the recipient select **Yes** and report the date (YYYY-MM-DD) of testing. If testing is performed multiple times in the reporting period, report the most recent date and results.

If the exact date is not known, use the process described in [General Instructions, Guidelines for Completing Forms](#).

If enzyme activity / enzyme substrate was not tested for the recipient within the reporting period or it is unknown, select **No** or **Unknown**, respectively and continue with *Was the total neurologic function scale (NFS) score obtained*.

Questions 4 – 5: Recipient result

Indicate whether the results of the recipient's enzyme activity and / or enzyme substrate testing were **Normal** or **Abnormal**. Additionally, specify if documentation was submitted to CIBMTR (CIBMTR recommends attaching the enzyme / storage activity testing).

For further instructions on how to attach documents in FormsNet3SM, refer to the [Training Guide](#).

Section Updates:

Question Number	Date of Change	Add/Remove/Modify	Description	Reasoning (If applicable)
.

DRAFT

Q6 – 53: Clinical Status Post-Infusion

Questions 6 – 7: Was the total neurologic function scale (NFS) score obtained? (ALD recipients only)

The total neurologic function scale (NFS) is a 25-point scale used to assess severity of neurological dysfunction and is done by scoring 15 disabilities.

Indicate **Yes** or **No** if a neurologic function scale (NFS) score was obtained during the current reporting period. This information will be documented within a physician's note.

If a total NFS score was obtained, select **Yes**, and report the date (YYYY-MM-DD) of assessment. If the total NFS score was obtained multiple times prior in the reporting period, report the date and results of the most recent assessment.

If the exact date is not known, use the process described in [General Instructions, Guidelines for Completing Forms](#).

If the total NFS score was not obtained within the reporting period or it is not known if a score was obtained, indicate **No**, and continue with *Did post-infusion seizures attributed to the underlying disease occur*.

Total Neurologic Function Scale (NFS) Score and Domain Clinical Scores

When reporting the total NFS and domain clinical scores, report the scores based on documentation within the physician's progress notes. If the total NFS score is documented but the domain clinical scores are not provided, only report the total NFS score, and leave the data fields for the domain clinical scores blank. Similarly, if only the domain clinical scores are known, report the domain clinical scores and leave the total NFS score blank.

Question 8: Specify total neurologic function scale score

Report the total neurologic function scale (NFS) score as documented by the physician. The total NFS score will be a value between 1-25.

If the NFS score is not known and only the domain clinical scores are documented, leave this data field blank.

Questions 9 – 24: Select known domain clinical score(s) (check all that apply)

Select the known domain clinical score(s) and report the score(s) as documented by the physician.

- **Hearing / auditory processing problems:** Trouble with hearing.
- **Aphasia / apraxia:** Speech disorder / impairment.
- **Loss of communication:** Loss of the ability to communicate.
- **Vision impairment / fields cut:** Reduced field of vision.
- **Cortical blindness:** Total or partial loss of vision due to damage to the occipital cortex.
- **Swallowing difficulty or other central nervous system dysfunction:** Dysphagia due to damage of the nervous system.

- **Tube feeding:** Percutaneous endoscopic gastrostomy (PEG), esophagogastroduodenoscopy (EGD), or G-tube insertion.
- **Running difficulties / hyperreflexia:** Muscles are less responsive to stimuli. Causing trouble holding, running, driving etc.
- **Walking difficulties / spasticity / spastic gait (no assistance):** Trouble walking, and / or abnormal muscle tightness due to prolonged muscle contractions.
- **Spastic gait (needs assistance) wheelchair required:** Stiff, often foot dragging walk, due to prolonged muscle contractions on one side. Assistance with wheelchair required.
- **No voluntary movement:** Absence of voluntary movements such as moving fingers, toes, sitting upright, etc.
- **Episodes or urinary or fecal incontinency:** Loss of bladder and / or bowel control with episodes of any urinary or bowel incontinence.
- **Total urinary or fecal incontinency:** Total loss of bladder and / or bowel control. This will require continuous use of a catheter.
- **Nonfebrile seizures:** A single, uncontrolled electrical activity in the brain, which may produce a physical convulsion, minor physical signs, thought disturbances or a combination of symptoms. Nonfebrile seizures are characterized as spontaneous recurrent seizures unrelated to fever.

If only the total NFS score is known and not the domain clinical scores, leave these questions blank.

Seizures

If the recipient developed post-infusion seizures unrelated to disease and there was not a prior diagnosis of seizures either prior to infusion or in a prior reporting period, report the seizures in the Organ Function section of the Post-Infusion Follow-Up (2100) form.

Question 25: Did post-infusion seizures attributed to the underlying disease occur since the date of the last report?

Indicate if the recipient had post-infusion seizures attributed to the underlying disease at any time during the current reporting period.

Unless otherwise noted by the physician, it can be assumed seizures were due to primary disease.

Report **No** in the following scenarios and continue with *Was cerebrospinal fluid (CSF) testing performed:*

- The recipient did not have any seizures during the current reporting period
- The recipient experienced seizures during the current reporting period but they were not attributed to the underlying disease, as noted by the physician

If it is not known if the recipient had any seizures during the current reporting period or if the seizures were related to the underlying disease, select **Unknown** and continue with *Was cerebrospinal fluid (CSF) testing performed.*

Question 26: Were any of the seizures considered nonfebrile?

Specify if any of the seizures in the current reporting period were considered nonfebrile (spontaneous recurrent seizures unrelated to fever). If at least one seizure in the reporting period was nonfebrile, select

Yes. If all seizures within the reporting period were febrile, or it is not known if all seizures were febrile, indicate **No** or **Unknown**, respectively.

Questions 27 – 29: Was cerebrospinal fluid (CSF) testing performed?

Indicate if cerebrospinal fluid (CSF) testing was completed within the current reporting period. CSF is collected via lumbar puncture or spinal tap. If CSF testing was not performed within the reporting period or it is not known, select No or Unknown, respectively and continue with *Was magnetic resonance imaging (MRI) performed*. The unknown option should be used sparingly and only when no judgment can be made to determine if this assessment was completed.

If CSF testing was performed select **Yes** and specify if the date (YYYY-MM-DD) of the most recent CSF testing is **Known** or **Unknown**. If **Known**, report the date of the most recent CSF assessment. If date is **Unknown**, continue with *Specify known CSF result(s)*.

Questions 30 – 32: Specify known CSF result(s) (check all that apply)

Report the known CSF results, select all that apply.

- **Opening Pressure:** Opening pressure is measured during the lumbar puncture; this is a measurement of intracranial pressure. If the opening pressure is known, select this option and report the opening pressure value.
- **Total Protein:** Total protein is a measurement used to determine the levels of protein in cerebrospinal fluid. If the total protein is known, select this option and report the CSF total protein value.

In the rare case where neither the opening pressure or the total protein was known, leave *Specify known CSF result(s)* blank, override the validation error using the code “unknown,” and continue with *Was magnetic resonance imaging (MRI) performed*.

Questions 33 – 35: Was Magnetic Resonance Imaging (MRI) performed

Magnetic resonance imaging (MRI) is an imaging technique used to form pictures of the anatomy and the physiological processes of the body. MRIs are used to assess recipients with leukodystrophy.

If an MRI was performed during the current reporting period, select **Yes** and specify if the date (YYYY-MM-DD) of the most recent MRI is **Known** or **Unknown**. If **Known**, report the most recent MRI's date. If the date is **Unknown**, continue with *Specify MRI results*.

If an MRI was not performed in the reporting period or it is not known, select **No** or **Unknown**, respectively and continue with *Were nerve conduction velocities tested*. The unknown option should be used sparingly only when no judgment can be made to determine if this assessment was completed.

Question 36: Specify MRI results

Specify the results of the most recent MRI performed in the reporting period. Indicate if the results were **Normal** or **Abnormal** as determined by the radiologist or physician.

Questions 37 – 38: Was gadolinium contrast used for this assessment?

Gadolinium contrast is often used in MRI assessments to enhance imaging, improving the visibility of inflammation, blood vessels and blood supply. Report **Yes** or **No** if gadolinium contrast was used in the MRI assessment.

If gadolinium contrast was used, select **Yes**, and indicate if gadolinium enhancement was reported. If gadolinium contrast was used, gadolinium enhancement will be noted in the MRI report and can be suggestive of abnormalities.

If gadolinium contrast was not used in the MRI assessment, select **No**.

Question 39: Loes composite score (ALD recipients only)

The Loes composite score is often used to assess disease / progression for recipients with ALD. The Loes composite score is a rating from 0-34, this signifies the severity of abnormalities detected in the brain after evaluation of MRI. This score should be listed within the MRI or within progress notes. Report the Loes composite score, if the score is unknown, check with a transplant physician to determine this value.

In the event where the transplant physician cannot determine the Loes composite score, leave the data field blank and override the FormsNet3SM error as “not documented.”

Question 40: Was documentation submitted to CIBMTR? (CIBMTR recommends attaching the MRI report)

Report **Yes** or **No** if a copy of the MRI report is attached. For instructions on how to attach documents in FormsNet3SM, refer to the [Training Guide](#).

Questions 41 – 42: Were nerve conduction velocities tested

Indicate if nerve conduction velocities were tested at any time within the current reporting period. Nerve conduction velocity (NCV) testing measures how quickly an electrical impulse moves through the nerve and can identify nerve damage. This procedure is typically performed by a neurologist.

Indicate **Yes**, if NCV testing was performed prior within the reporting period and report the date (YYYY-MM-DD) of the most recent testing. If NCV testing was not done or it is unknown, report **No** or **Unknown**, respectively and continue with *Was a neurocognitive test performed*.

Questions 43 – 44: Specify Results

Indicate whether the results of the recipient's NCV testing was **Normal** or **Abnormal** and if documentation was submitted to CIBMTR (CIBMTR recommends attaching the nerve conduction velocities tests).

The results of the NCV test may be found in the procedure / results report or in the neurologist's note.

For instructions on how to attach documents in FormsNet3SM, refer to the [Training Guide](#).

Questions 45 – 47: Was a neurocognitive test performed

A neurocognitive test is an assessment completed by a neuropsychologist, used to assess the cognitive function of a recipient. Indicate if a neurocognitive test was administered within the reporting period. This information will be found within a progress note by the neuropsychologist.

If neurocognitive testing was performed, indicate **Yes**, and report the date (YYYY-MM-DD) of the most recent assessment. Specify if documentation of mental development neurocognitive testing was submitted to the CIBMTR. It is highly encouraged to attach this assessment / documentation. Additionally, complete the Neurocognitive Assessment (3503) Form.

If testing was not done or it is unknown, indicate **No** or **Unknown**, respectively and continue with *Has there been a change in the recipient's neurologic status*.

For instructions on how to attach documents in FormsNet3SM, refer to the [Training Guide](#).

Questions 48 – 50: Has there been a change in the recipient's neurologic status (Report clinical status, not neuropsychological status.)

Indicate if there has been a change in the recipient's neurologic status since the previous reporting period. If **Yes**, specify if the current neurologic status has **Improved** or **Worsened**, when compared to the previous report. Additionally, specify if documentation of the physical or neurologic exam was submitted to the CIBMTR. It is highly encouraged to attach documentation of the exam from the neurologist or transplant physician.

If the recipient's neurologic status is stable and / or unchanged since the previous reporting period, select **Stable / unchanged**.

If it is not known if the recipient's neurologic status has changed (i.e., the physician's progress notes are not clear if there has been a change in the neurologic status), select **Unknown** and continue with *Specify global improvement*.

Clinical Global Impression (CGI) (neurologic assessment)

Clinical Global Impression (CGI) (neurologic assessment)

When reporting the CGI, report the scores based on neurologist's or transplant physicians' progress notes. If the scores are not clearly documented, select Unknown.

Question 51: Specify global improvement (select one)

Report the recipient's global improvement within the current reporting period. The Clinical Global Impression (CGI) is a 7-point scale, measuring the illness severity (CGIS), global improvement and / or change (CGIC) and therapeutic response. This information may not be documented within a routine follow-up progress note and will likely be found within a progress note by the neurologist or transplant physician.

If the recipient's global improvement is not documented for the current reporting period, select **Unknown**.

Questions 52 – 53: Specify leukodystrophy-specific therapy given (check all that apply)

Select all leukodystrophy-specific treatment given during the current reporting period.

- **N-acetyl-L-cysteine (NAC):** (Acetylcysteine) is a supplement of cysteine (amino acid). Cysteine is an antioxidant agent that assists with respiratory conditions, fertility, and brain health.
- **GTE:GTO oil (Lorenzo's oil):** Composed of erucic acid and oleic acids. Known to decrease / lower very long chain fatty acids (VLCA) levels.
- **Other treatment:** Includes those treatments not already listed above. If the recipient received treatments not listed, such as experimental clinical trials, select this option and specify the other treatment(s). Report the generic name of the agent, not the brand name.

If no therapy was given during the current reporting period, select **None** and go to the signature line.

Section Updates:

Question Number	Date of Change	Add/Remove/Modify	Description	Reasoning (If applicable)
.

DRAFT

Q54 – 61 Disease Modifying Therapies

Question 54: Were disease modifying therapies given? (excludes blood transfusions)

Indicate if the recipient received disease modifying therapies (review the question below for a list of common disease modifying therapies) during the current reporting period, excluding blood transfusion(s).

If the recipient did not receive disease modifying therapies or if no information is available to determine if the recipient received disease modifying therapies, select **No** or **Unknown**, respectively and submit the form.

Reporting Multiple Disease Modifying Therapies

FormsNet3SM application: Complete questions 55 – 61 for each disease modifying therapy administered by adding an additional instance in FormsNet3SM application.

Paper form submission: Copy questions 55 – 61 and complete for each disease modifying therapy administered.

Same Therapy Restarted

If the same therapy was started and stopped multiple times during the current reporting period, only one instance needs to be reported. In this case, report the therapy start date as the date therapy first began in the reporting period.

Questions 55 – 56: Specify the disease modifying therapy (check all that apply)

Select the disease modifying therapy administered as part of the line of therapy being reported.

- **Leriglitazone:** a novel selective peroxisome proliferator-activated receptor gamma agonist.

If the recipient received a therapy which is not listed, select **Other** and specify the treatment. Report the generic name of the agent, not the brand name.

Question 57: Was the date therapy started previously reported?

Specify if the therapy start date was previously reported. If the therapy was started in a prior reporting period and continued into the current reporting period, select **Yes**, and continue with *Date Therapy Started*.

The **Yes** option is not applicable for the Day 100 reporting period.

Questions 58 – 59: Date therapy started

Indicate if the therapy start date is known. If **Known**, report the first date (YYYY-MM-DD) the recipient began this line of therapy.

If the exact date is not known report an estimated date and check the **Date estimated** box. Refer to [General Instructions, General Guidelines for Completing Forms](#) for information about reporting estimated dates.

Questions 60 – 61: Date therapy stopped

Indicate if the stop date is known. If the therapy stop date is **Known**, report the date (YYYY-MM-DD) when the therapy end. If the therapy is being given in cycles, report the end date as the date when the recipient started the last cycle for this line of therapy. Otherwise, report the final administration date for the therapy being reported.

If the exact date is not known report an estimated date and check the **Date estimated** box. Refer to [General Instructions, General Guidelines for Completing Forms](#) for information about reporting estimated dates.

Report **Not applicable** if the recipient is still receiving therapy on the contact date.

Section Updates:

Question Number	Date of Change	Add/Remove/Modify	Description	Reasoning (If applicable)
.

Q62: Marrow Evaluation

Marrow evaluation

This section is only completed for gene therapy infusions.

Question 62: Was a marrow aspirate and / or biopsy performed?

Indicate Yes or No if a marrow aspirate and or biopsy was performed in this reporting period. Additionally, complete the Laboratory Studies (3502) Form and Marrow Surveillance (3506) Form. The intent is to screen for and/or identify changes in the marrow such as dysplasia, MDS, or new hematologic malignancy.

Report **Unknown** if not documented.

Section Updates:

Question Number	Date of Change	Add/Remove/Modify	Description	Reasoning (If applicable)
.