

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:

Q1-5: Leukodystrophies Post-Infusion Data

Q6 – 52: Clinical Status Post-Infusion

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
10/ 29/ 2021	2137: Leukodystrophies Post-Infusion	Add	Version 1 of the Leukodystrophies Post-Infusion Data section of the Forms Instructions Manual released. Version 1 corresponds to revision 3 of the Form 2137.

Last modified: Oct 29, 2021

Q1 – 5: Leukodystrophies Post-Infusion Data

! **Hereditary diffuse leukoencephalopathy (HDLS):** If the primary disease for infusion is HDLS, manually select this option from question 1. 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

Section Updates:

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 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 question 6.

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)
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Last modified: Oct 29, 2021

Q6 – 52: Clinical Status Post-Infusion

Clinical Status Post-Infusion

The Clinical Status Post-Infusion section can only be answered for the six-month and annual reporting periods. This section will be disabled for the Day 100 reporting period.

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 of assessment in. 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 question 24.

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 – 23: 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 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 24: 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 question 26:

- 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 question 26.

Question 25: 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 26 – 28: 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 question 32. 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 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 question 29.

Questions 29 – 31: 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 in question 30.
- **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 question 31.

In the rare case where neither the opening pressure or the total protein was known, leave question 29 blank, override the validation error using the code “unknown,” and continue with question 32.

Questions 32 – 34: 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 recipient's with leukodystrophy.

If an MRI was performed during the current reporting period, select **Yes** and specify if the date 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 question 35.

If an MRI was not performed in the reporting period or it is not known, select **No** or **Unknown**, respectively and continue with question 40. The unknown option should be used sparingly only when no judgment can be made to determine if this assessment was completed.

Question 35: 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 36 – 37: 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 in question 37. 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** and continue with question 38.

Question 38: 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 in question 38, 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 39: 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 40 – 41: 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 of the most recent testing. If NCV testing was not done or it is unknown, report **No** or **Unknown**, respectively and continue with question 44.

Questions 42 – 43: 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 44 – 46: 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 of the most recent assessment. Additionally, specify if documentation of mental development neurocognitive testing was submitted to the CIBMTR. It is highly encouraged to attach this assessment / documentation.

If testing was not done or it is unknown, indicate **No** or **Unknown**, respectively and continue with question 47.

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

Questions 47 – 49: 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** and continue with question 50.

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 question 50.

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 50: 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 51 – 52: 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 acid's. 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) in question 52. 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)
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