

2037: Leukodystrophies Pre-Infusion

The Leukodystrophies Pre-Infusion Data Form is one of the Comprehensive Report Forms. This form captures leukodystrophy specific pre-infusion data such as: disease assessments / laboratory studies at diagnosis, pre-infusion treatment and laboratory studies / clinical status prior to the start of the preparative regimen.

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)

Links to sections of the manual:

Q1: Subsequent Transplant or Cellular Therapy

Q2 – 22: Leukodystrophy Diagnosis

Q23 – 63: Clinical Status Prior to Preparative Regimen

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	2037: Leukodystrophies Pre-Infusion	Add	Version 1 of the Leukodystrophies Pre-Infusion Data section of the Forms Instructions Manual released. Version 1 corresponds to revision 3 of the Form 2037.

Last modified: Oct 29, 2021

Q1: Subsequent Transplant or Cellular Therapy

Question 1: Is this the report of a second or subsequent transplant or cellular therapy for the same disease?

Report **No** and go to question 2 in any of the following scenarios:

- This is the first infusion reported to the CIBMTR; or
- This is a second or subsequent infusion for a different disease (i.e., the patient was previously transplanted for a disease other than a leukodystrophy); or
- This is a second or subsequent infusion for the same disease subtype and this baseline disease insert was not completed for the previous transplant (i.e., the patient was on the TED track for the prior infusion, prior infusion was autologous with no consent, etc.).

Report **Yes** and go to question 23 if this is a subsequent infusion for the same disease and the baseline Leukodystrophies disease insert was completed previously.

Section Updates:

Question Number	Date of Change	Add/Remove/Modify	Description	Reasoning (If applicable)
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Last modified: Oct 29, 2021

Q2 – 22: Leukodystrophy Diagnosis

! **Hereditary diffuse leukoencephalopathy (HDLS):** If the primary disease for infusion is HDLS, manually select this option from question 2. This option will not auto populate at this time.

Question 2: Specify the leukodystrophy subtype

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 the 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.

Questions 3 – 4: Specify testing performed to establish the diagnosis (check all that apply)

Specify which testing was performed to establish the diagnosis of the primary disease for infusion, select all that apply.

- **Newborn Screening:** A blood screen which looks for the defective molecules in the circulating blood. In the United States, this blood test is available as part of routine newborn screening. Infants are often diagnosed at birth or in utero.
- **Genetic mutational panel:** A genetic panel is a standard panel of genes known to be associated with hematopoietic abnormalities. The intent of this assessment is to screen for myeloid diseases. This assessment is typically a myeloid mutation panel and is usually labeled as a "Genetic Mutational Panel" within the EMR; however, this varies from institution to institution. If it is unclear if this assessment was performed, seek physician clarification.
- **Laboratory findings (enzyme levels, storage levels, hormone levels):** Laboratory studies such as enzyme levels, storage levels and hormone levels, may be used to identify deficiency's that cause leukodystrophy.
- **Other Testing:** Includes those methods of testing not already listed above. This option will rarely be used; however, a recipient may be diagnosed as a result of another family member's prior leukodystrophy diagnosis or imaging testing characteristic of the disease. If **Other testing** was performed to establish the diagnosis select this option and specify in question 4.

Enzyme / storage activity at diagnosis – recipient

 **At Diagnosis:** Any testing performed between the date of diagnosis and the start of any treatment for Leukodystrophy.

Questions 5 – 6: Was enzyme / storage activity tested?

Indicate if enzyme / storage activity was tested for the recipient at diagnosis. This type of testing performed is specific to the leukodystrophy subtype. Types of testing include enzyme assays, urine testing, etc. If enzyme / storage activity was tested on the recipient select **Yes** and indicate the date of testing. An example of enzyme activity testing is the quantitative measurement of arylsulfatase A enzyme. If the arylsulfatase A enzyme is deficient, it is the gold standard to confirm a diagnosis of metachromatic Leukodystrophy.

Testing is generally only performed once. In the rare scenario testing is performed multiple times, report the date of the most recent assessment and the results prior to the start of treatment.

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

If enzyme / storage activity was not tested for the recipient at diagnosis or it is not known, select **No** or **Unknown**, respectively, and continue with question 9.

Questions 7 – 8: Recipient result

Indicate whether the results of the recipient's enzyme / storage activity testing were **Normal** or **Abnormal** at diagnosis and 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](#).

Enzyme / storage activity at diagnosis – donor

Question 9: Was the donor / CBU a carrier?

Indicate if the donor or CBU was a carrier for a genetic disease / leukodystrophies. If the donor was tested for being a carrier for genetic diseases / leukodystrophies and was negative, select **No** and continue with question 14. If the donor was not tested, or it is unknown if testing was performed, select **Unknown** and continue with question 14.

Questions 10 – 11: Was enzyme activity and / or enzyme substrate tested?

Indicate if enzyme activity and / or enzyme substrate was tested at any time prior to the start of the preparative regimen / infusion. The type of testing performed is specific to leukodystrophy subtype. Types of testing can include enzyme assays, urine testing, etc.

If enzyme / storage activity was tested on the donor select **Yes** and report the date when the donor / CBU was tested. Testing is generally only performed once. In the rare scenario testing is performed multiple times, report the most recent date and results prior to the start of the preparative regimen / infusion.

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 donor or it is unknown, select **No** or **Unknown**, respectively and continue with question 14.

Questions 12 – 13: Donor / CBU testing result

Indicate whether the results of the donor's / CBU's enzyme activity and / or enzyme substrate testing were **Normal** or **Abnormal** and 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](#).

Question 14: Was a genetic mutational panel performed at any time prior to the start of the preparative regimen? (screening for myeloid diseases)

A genetic panel is a standard panel of genes that are known to be associated with hematopoietic abnormalities. The intent of this assessment is to screen for myeloid diseases and is typically a myeloid mutation panel. This report is usually labeled as a "Genetic Mutational Panel" within the EMR; however, this varies from institution to institution. If it is unclear if this assessment was performed, seek physician clarification.

Indicate **Yes** if a genetic panel was performed at any time prior to the start of the preparative regimen / infusion.

If a genetic panel was not performed or it not known if performed at any time prior to the start of the preparative regimen / infusion, select **No** and continue with question 17.

Questions 15 – 16: Specify results

Indicate whether the results of the genetic panel were **Normal** or **Abnormal** and if a copy of the genetic mutational panel was submitted to the CIBMTR.

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

Question 17: Were the recipient's urinary sulfatides elevated at diagnosis? (MLD recipients only)

Indicate if the recipient's urinary sulfatides were elevated at diagnosis. The analysis of urinary sulfatides is determined through a urine test, often performed for recipients with MLD. If the recipient's urinary sulfatides are elevated above the lab's upper limit of normal (ULN), select **Yes**.

If urinary sulfatide analysis was not completed at diagnosis or it is unknown if this analysis was completed, select **No** or **Unknown**, respectively.

Questions 18 – 19: Mean fasting plasma very-long-chain fatty acid (VLCFA) C26:0 level at diagnosis (fasting preferred, but not required) (ALD recipients only)

Indicate if the mean fasting plasma very-long-chain fatty acid (VLCA) level is known at diagnosis, prior to receiving any treatment for ALD. This assessment is typically found within the general lab section of the EMR.

If the mean fasting plasma very-long-chain fatty acid (VLCA) level is **Known**, report the value in µg/mL. If this value is not known, select **Unknown** and continue with question 20.

Question 20: Specify treatment(s) given for adrenal insufficiency with glucocorticoids or mineralocorticoids between diagnosis and infusion (ALD recipients only) (check all that apply)

Select all treatments given for adrenal insufficiency between diagnosis and the start of the preparative regimen / infusion.

- **Glucocorticoids:** Corticosteroids with anti-inflammatory properties used to treat conditions involving inflammation (e.g., cortisol and cortisone).
- **Mineralocorticoids:** Corticosteroids, assist in regulation of sodium and potassium (e.g., aldosterone).

If the recipient did not receive neither **Glucocorticoids** and / or **Mineralocorticoids** at any time prior to the start of the preparative regimen / infusion, select **None**.

Questions 21 – 22: Specify treatment(s) given to lower plasma very-long-chain fatty acids at any time prior to infusion (check all that apply) (ALD recipients only)

Select all treatments given to lower plasma very-long-chain fatty acids at any time prior to the start of the preparative regimen / infusion.

- **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 22. Report the generic name of the agent, not the brand name.

If no therapy was given at any time prior to the start of the preparative regimen / infusion, select **None**.

Question Number	Date of Change	Add/Remove/Modify	Description	Reasoning (If applicable)
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Q23 – 63: Clinical Status Prior to Preparative Regimen

* **Testing Prior to the Start of the Preparative Regimen / Infusion:**
Report all findings within two months prior to the start of the preparative regimen / infusion for the Clinical Status Prior to Preparative Regimen section. If an assessment was performed multiple times, report the most recent results.

* **Enzyme activity / enzyme substrate testing:**
Do not include the recipient's results of enzyme activity / enzyme substrate testing performed at diagnosis.

Questions 23 – 24: Was enzyme activity and/or enzyme substrate tested?

Indicate if the recipient was tested for enzyme activity and / or enzyme substrate prior to the start of the preparative regimen / infusion. The type of testing performed is specific to leukodystrophy subtype. Types of testing can include enzyme assays, urine testing, etc.

If the recipient's enzyme activity and / or enzyme substrate was tested, select **Yes** and report the date of testing. If testing is performed multiple times, report the most recent date and results prior to the start of the preparative regimen / infusion.

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

If the enzyme activity and / or enzyme substrate was not tested prior to the start of the preparative regimen / infusion or it is unknown if testing occurred, select **No** or **Unknown**, respectively and continue with question 27.

Questions 25 – 26: Recipient result

Indicate whether the results of the recipient's enzyme activity and / or enzyme substrate testing were **Normal** or **Abnormal** and 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](#).

Questions 27 – 28: 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 NFS score was obtained prior to the preparative regimen / infusion. 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. If the total NFS was obtained multiple times prior to the preparative regimen / infusion, 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 prior to the preparative regimen / infusion or it is not known if a score was obtained, select **No**, and continue with question 45.



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 note. 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 29: 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 30 – 44: 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.

Question 45: Is there a history of seizures attributed to the underlying disease at any time prior to the preparative regimen?

Indicate **Yes** if there was a history of seizures attributed to the underlying disease at any time prior to the preparative regimen / infusion. If there was no history of seizures attributed to the underlying disease or it is not known, indicate **No**, and continue with question 47.

Question 46: Were any of the seizures considered nonfebrile?

Report **Yes** if any of the seizures prior to the preparative regimen / infusion were considered nonfebrile (spontaneous recurrent seizures unrelated to fever). If all seizures prior to the start of the preparative regimen / infusion were febrile, select **No**.

Questions 47 – 48: Was cerebrospinal fluid (CSF) testing done prior to the preparative regimen?

Indicate if cerebrospinal fluid (CSF) testing was completed prior to the preparative regimen / infusion. CSF is collected via lumbar puncture or spinal tap.

If testing was performed, select **Yes** and report the date of the most recent CSF testing.

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

If CSF testing was not performed prior to the preparative regimen / infusion or it is not known, select **No** or **Unknown**, respectively and continue with question 52.

Questions 49 – 51: Specify known CSF result(s) (check all that apply)

Report the known CSF results.

- **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 50.

- **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 51.

In the rare case where neither the opening pressure or the total protein was known, leave question 49 blank, override the validation error using the code “unknown,” and continue with question 52.

Question 52: Date of most recent MRI prior to the preparative regimen

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

Report the date of the most recent MRI performed prior to the preparative regimen / infusion.

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

Question 53: Specify MRI results

Specify the results of the most recent MRI performed prior to the start of the preparative regimen / infusion. Indicate if the results were **Normal** or **Abnormal** as determined by the radiologist or physician.

Questions 54 – 55: 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.

If gadolinium contrast was used, select **Yes** in question 54. If gadolinium enhancement was reported, select **Yes** in question 55. 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 56.

Question 56: Is a copy of the MRI report attached? (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 57 – 58: Were nerve conduction velocities tested at any time prior to the preparative regimen?

Nerve conduction velocity (NCV) testing measures how quickly an electrical impulse moves through the nerve and can identify nerve damage. This is procedure is typically performed by a neurologist.

Indicate if NCV testing was performed at any time prior to the preparative regimen / infusion. If **Yes**, 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 61.

Questions 59 – 60: Specify Results

Report whether the results of the recipient's NCV testing was **Normal** or **Abnormal**. Additionally, indicate 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 61 – 63: Was a neurocognitive test administered at any time prior to the preparative regimen?

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 at any time prior to the preparative regimen / infusion. 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 and 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 to the signature line.

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

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