Leukodystrophies
Pre-Infusion

Registry Use Only

Sequence Number:

Date Received:

CIBMTR Center Number: ___ ___ ___ ___ ___

CIBMTR Research ID: ___ ___ ___ ___ ___ ___ ___ ___ ___ ___

Event date: ___ ___ ___ ___ — ___ ___ — ___ ___

YYYY MM DD

CIBMTR Center Number: ___ ___ ___ ___ ___ CIBMTR Research ID: ___ ___ ___ ___ ___ ___ ___ ___ ___ ___

Subsequent Infusion

If this is a report of a second or subsequent infusion for the same disease subtype and this baseline disease insert has not been completed for the previous infusion (e.g., recipient was on TED track for the prior infusion, prior infusion was autologous with no consent, or prior infusion was not reported to CIBMTR), select "No" to question 1 and continue to question 2.

1. Is this the report of a second or subsequent infusion for the same disease?
   - ☐ Yes – ***Go to question 30***
   - ☐ No – ***Go to question 2***

Leukodystrophy Diagnosis

2. Specify the leukodystrophy subtype
   - ☐ Krabbe Disease (globoid cell leukodystrophy)
   - ☐ Metachromatic leukodystrophy (MLD)
   - ☐ Adrenoleukodystrophy (ALD)
   - ☐ Hereditary diffuse leukoencephalopathy with spheroids (HDLS)

3. Specify testing performed to establish the diagnosis *(check all that apply)*
   - ☐ Newborn screening – ***Go to question 5***
   - ☐ Genetic mutational panel – ***Go to question 5***
   - ☐ Laboratory findings *(enzyme levels, storage levels, hormone levels)* – ***Go to question 5***
   - ☐ Other testing– ***Go to question 4***

4. Specify other testing: _____

Enzyme activity and / or enzyme substrate at diagnosis

Recipient

5. Was enzyme activity and / or enzyme substrate tested?
   - ☐ Yes – **Go to question 6**
   - ☐ No – **Go to question 9**
   - ☐ Unknown – **Go to question 9**

6. Date recipient tested: ___ ___ ___ ___ — ___ ___ — ___ ___
   YYYY MM DD

7. Recipient result
   - ☐ Normal

☐ Abnormal

8. Was documentation submitted to the CIBMTR? *(e.g., enzyme activity and / or enzyme substrate testing) (CIBMTR recommends attaching the enzyme activity and / or enzyme substrate testing)*

☐ Yes

☐ No

Donor

9. Was the donor / CBU a carrier?

☐ Yes – ***Go to question 10***

☐ No ***– Go to question 14***

☐ Unknown – ***Go to question 14***

10. Was enzyme activity and / or enzyme substrate tested?

☐ Yes – ***Go to question 11***

☐ No ***– Go to question 14***

☐ Unknown ***– Go to question 14***

11. Date donor / CBU tested: ___ ___ ___ ___ — ___ ___ — ___ ___
YYYY MM DD

12. Donor / CBU testing result

☐ Normal

☐ Abnormal

13. Was documentation submitted to the CIBMTR? *(e.g., enzyme activity and / or enzyme substrate testing) (CIBMTR recommends attaching the enzyme activity and / or enzyme substrate testing)*

☐ Yes

☐ No

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

☐ Yes – ***Go to question 15***

☐ No – ***Go to question 17***

15. Specify results

☐ Normal

☐ Abnormal

16. Was documentation submitted to the CIBMTR? *(CIBMTR recommends attaching the genetic mutational panel)*

☐ Yes

☐ No

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

☐ Yes

☐ No

☐ Unknown

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

☐ Known – ***Go to question 19***

☐ Unknown – ***Go to question 20***

19. VLCFA C26:0 level: ___ • ___ ___ ___ µg/mL

20. Specify therapy given for adrenal insufficiency with glucocorticoids or mineralocorticoids between diagnosis and infusion *(check all that apply) (ALD recipients only)*

☐ Glucocorticoids

☐ Mineralocorticoids

☐ None

21. Specify therapy given to lower plasma very-long-chain fatty acids at any time prior to infusion *(check all that apply) (ALD recipients only)*

☐ N-acetyl-L-cysteine (NAC) **– *Go to question 23***

☐ GTE:GTO oil (Lorenzo's oil) **– *Go to question 23***

☐ Other therapy – ***Go to question 22***

☐ None **– *Go to question 23***

22. Specify other therapy: _____

Disease Modifying Therapies

23. Were disease modifying therapies given? *(excludes blood transfusions)*

☐ Yes – ***Go to question 24***

☐ No - ***Go to question 30***

☐ Unknown - ***Go to question 30***

If there is more than one therapy given copy questions 24-29 for each therapy.

24. Specify the disease modifying therapy *(check all that apply)*

☐ Leriglitazone – ***Go to question 26***

☐ Other therapy – ***Go to question 25***

25. Specify other therapy: _____

26. Date therapy started

☐ Known - ***Go to question 27***

☐ Unknown – ***Go to question 28***

27. Date therapy started: ___ ___ ___ ___ -___ ___ - ___ ___ ☐ Date estimated
YYYY MM DD

28. Date therapy stopped

☐ Known – ***Go to question 29***

☐ Unknown - ***Go to question 30***

☐ Not applicable *(still receiving therapy)* - ***Go to question 30***

29. Date therapy stopped: ___ ___ ___ ___ -___ ___ - ___ ___ ☐ Date estimated
YYYY MM DD

Clinical Status Prior to Preparative Regimen

Recipient enzyme activity and / or enzyme substrate prior to preparative regimen *(do not include diagnostic testing in questions 5-8)*

30. Was enzyme activity and / or enzyme substrate tested?

☐ Yes – **Go to question 31**

☐ No – **Go to question 34**

☐ Unknown – **Go to question 34**

31. Date recipient tested: ___ ___ ___ ___ — ___ ___ — ___ ___
YYYY MM DD

32. Recipient result

☐ Normal

☐ Abnormal

33. Was documentation submitted to the CIBMTR? *(e.g., enzyme activity and / or enzyme substrate testing) (CIBMTR recommends attaching the enzyme activity and / or enzyme substrate testing)*

☐ Yes

☐ No

34. Was the total neurologic function scale (NFS) score obtained? *(ALD recipients only)*

☐ Yes – ***Go to question 35***

☐ No – ***Go to question 53***

35. Specify date of NFS score: ___ ___ ___ ___ — ___ ___ — ___ ___
YYYY MM DD

36. Specify total neurologic function scale score: ___ ___

37. Select known domain clinical score(s) *(check all that apply)*

☐ Hearing / auditory processing problems – ***Go to question 38***

☐ Aphasia / apraxia – ***Go to question 39***

☐ Loss of communication – ***Go to question 40***

☐ Vision impairment / fields cut – ***Go to question 41***

☐ Cortical blindness – ***Go to question 42***

☐ Swallowing difficulty or other central nervous system dysfunction – ***Go to question 43***

☐ Tube feeding – ***Go to question 44***

☐ Running difficulties / hyperreflexia – ***Go to question 45***

☐ Walking difficulties / spasticity / spastic gait (no assistance) – ***Go to question 46***

☐ Spastic gait (needs assistance) – ***Go to question 47***

☐ Wheelchair required – ***Go to question 48***

☐ No voluntary movement – ***Go to question 49***

☐ Episodes of urinary or fecal incontinency – ***Go to question 50***

☐ Total urinary or fecal incontinency – ***Go to question 51***

☐ Nonfebrile seizures– ***Go to question 52***

38. Hearing / auditory processing problems: ___ ___

39. Aphasia / apraxia: ___ ___

40. Loss of communication: ___ ___

41. Vision impairment / fields cut: ___ ___

42. Cortical blindness: ___ ___

43. Swallowing difficulty or other central nervous system dysfunction: ___ ___

44. Tube feeding: ___ ___

45. Running difficulties / hyperreflexia: ___ ___

46. Walking difficulties / spasticity / spastic gait (no assistance): ___ ___

47. Spastic gait (needs assistance): ___ ___

48. Wheelchair required: ___ ___

49. No voluntary movement: ___ ___

50. Episodes of urinary or fecal incontinency: ___ ___

51. Total urinary or fecal incontinency: ___ ___

52. Nonfebrile seizures: ___ ___

53. Is there a history of seizures attributed to the underlying disease at any time prior to the preparative regimen?

☐ Yes – ***Go to question 54***

☐ No – ***Go to question 55***

54. Were any of the seizures considered nonfebrile?

☐ Yes

☐ No

55. Was cerebrospinal fluid (CSF) testing done prior to the preparative regimen?

☐ Yes - ***Go to question 56***

☐ No - ***Go to question 60***

☐ Unknown - ***Go to question 60***

56. Date of most recent CSF testing: ___ ___ ___ ___ — ___ ___ — ___ ___

YYYY MM DD

57. Specify known CSF result(s) *(check all that apply)*

☐ Opening pressure – ***Go to question 58***

☐ Total protein – ***Go to question 59***

58. Opening pressure: ___ ___ ___ • ___ cm H_2O

59. Total protein: ___ ___ ___ • ___ ___ ☐ mg/dL
☐ g/L

60. Date of most recent magnetic resonance imaging (MRI) prior to the preparative regimen:

___ ___ ___ ___ – ___ ___ – ___ ___
YYYY MM DD

61. Specify MRI results
☐ Normal
☐ Abnormal

62. Was gadolinium contrast used for this assessment?
☐ Yes – ***Go to question 63***
☐ No – ***Go to question 64***

63. Was gadolinium enhancement reported?
☐ Yes
☐ No

64. Was documentation submitted to the CIBMTR? *(CIBMTR recommends attaching the MRI report)*
☐ Yes
☐ No

65. Were nerve conduction velocities tested at any time prior to the preparative regimen?
☐ Yes - ***Go to question 66***
☐ No - ***Go to question 69***
☐ Unknown - ***Go to question 69***

66. Date of most recent nerve conduction velocities test prior to the preparative regimen:

___ ___ ___ ___ – ___ ___ – ___ ___
YYYY MM DD

67. Specify results
☐ Normal

☐ Abnormal

68. Was documentation submitted to the CIBMTR? *(CIBMTR recommends attaching the nerve conduction velocities tests)*

☐ Yes

☐ No

69. Was a neurocognitive test administered at any time prior to the preparative regimen?

☐ Yes - **Also complete Neurocognitive Assessment Form 3503 - *Go to question 70***

☐ No - ***Go to question 72***

☐ Unknown - ***Go to question 72***

70. Date of most recent neurocognitive test prior to the preparative regimen:

___ ___ ___ ___ – ___ ___ – ___ ___
YYYY MM DD

71. Was documentation submitted to the CIBMTR? *(CIBMTR recommends attaching the neurocognitive testing report)*

☐ Yes

☐ No

Marrow Evaluation

Complete question 72 for gene therapy infusions only.

72. Was a marrow aspirate and / or biopsy performed?

☐ Yes - **Also complete Laboratory Studies Form 3502 and Marrow Surveillance Form 3506**

☐ No

☐ Unknown