Leukodystrophies
Pre-Infusion

Registry Use Only
Sequence Number:

Date Received:

CIBMTR Center Number: ___ ___ ___ ___ ___
CIBMTR Research ID: ___ ___ ___ ___ ___ ___ ___ ___ ___ ___
Event date: ___ ___ ___ ___ — ___ ___ — ___ ___
YYYY MM DD

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 23
   □ No – Go to question 2

Leukodystrophy Diagnosis

2. Specify the leukodystrophy subtype
   □ Krabbe Disease (globoid cell leukodystrophy)
   □ Metachromatic leukodystrophy (MLD)
   □ Adrenoleukodystrophy (ALD)
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*
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: _______________________

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)

23. Was enzyme activity and/or enzyme substrate tested?
   □ Yes – Go to question 24
   □ No – Go to question 27
   □ Unknown – Go to question 27

24. Date recipient tested: _______ _______ ______
       YYYY               MM          DD

25. Recipient result
   □ Normal
   □ Abnormal

26. 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

27. Was the total neurologic function scale (NFS) score obtained? (ALD recipients only)
   □ Yes – Go to question 28
   □ No – Go to question 46

28. Specify date of NFS score: _______ _______ ______
       YYYY               MM          DD
29. Specify total neurologic function scale score: ___ ___

30. Select known domain clinical score(s) *(check all that apply)*
- Hearing / auditory processing problems – *Go to question 31*
- Aphasia / apraxia – *Go to question 32*
- Loss of communication – *Go to question 33*
- Vision impairment / fields cut – *Go to question 34*
- Cortical blindness – *Go to question 35*
- Swallowing difficulty or other central nervous system dysfunction – *Go to question 36*
- Tube feeding – *Go to question 37*
- Running difficulties / hyperreflexia – *Go to question 38*
- Walking difficulties / spasticity / spastic gait (no assistance) – *Go to question 39*
- Spastic gait (needs assistance) – *Go to question 40*
- Wheelchair required – *Go to question 41*
- No voluntary movement – *Go to question 42*
- Episodes of urinary or fecal incontinency – *Go to question 43*
- Total urinary or fecal incontinency – *Go to question 44*
- Nonfebrile seizures – *Go to question 45*

31. Hearing / auditory processing problems: ___ ___

32. Aphasia / apraxia: ___ ___

33. Loss of communication: ___ ___

34. Vision impairment / fields cut: ___ ___

35. Cortical blindness: ___ ___

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

37. Tube feeding: ___ ___

38. Running difficulties / hyperreflexia: ___ ___

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

40. Spastic gait (needs assistance): ___ ___
41. Wheelchair required: ___ ___

42. No voluntary movement: ___ ___

43. Episodes of urinary or fecal incontinency: ___ ___

44. Total urinary or fecal incontinency: ___ ___

45. Nonfebrile seizures: ___ ___

46. Is there a history of seizures attributed to the underlying disease at any time prior to the preparative regimen?
   ☐ Yes – Go to question 47
   ☐ No – Go to question 48

47. Were any of the seizures considered nonfebrile?
   ☐ Yes
   ☐ No

48. Was cerebrospinal fluid (CSF) testing done prior to the preparative regimen?
   ☐ Yes - Go to question 49
   ☐ No - Go to question 53
   ☐ Unknown - Go to question 53

49. Date of most recent CSF testing: ___ ___ ___ ___ — ___ ___ — ___ ___
    YYYY                         MM                  DD

50. Specify known CSF result(s) (check all that apply)
    ☐ Opening pressure – Go to question 51
    ☐ Total protein – Go to question 52

51. Opening pressure: ___ ___ ___ ● ___ cm H2O

52. Total protein: ___ ___ ___ ● ___ □ mg/dL
                               □ g/L

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

    ___ ___ ___ ___ — ___ ___ — ___ ___
    YYYY                         MM                  DD

54. Specify MRI results
55. Was gadolinium contrast used for this assessment?
   - Yes – Go to question 56
   - No – Go to question 57

56. Was gadolinium enhancement reported?
   - Yes
   - No

57. Was documentation submitted to the CIBMTR? (CIBMTR recommends attaching the MRI report)
   - Yes
   - No

58. Were nerve conduction velocities tested at any time prior to the preparative regimen?
   - Yes - Go to question 59
   - No - Go to question 62
   - Unknown - Go to question 62

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

   ___ ___ ___ ___ — ___ ___ — ___ ___
   YYYY                        MM                   DD

60. Specify results
   - Normal
   - Abnormal

61. Was documentation submitted to the CIBMTR? (CIBMTR recommends attaching the nerve conduction velocities tests)
   - Yes
   - No

62. Was a neurocognitive test administered at any time prior to the preparative regimen?
   - Yes - Go to question 63
   - No - Go to First Name
   - Unknown - Go to First Name
63. Date of most recent neurocognitive test prior to the preparative regimen:

___ ___ ___ ___ — ___ ___ — ___ ___
YYYY MM DD

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

☐ Yes
☐ No

First Name: _____________________________________________________________________________________

Last Name: _____________________________________________________________________________________

Email address: ___________________________________________________________________________________

Date: ___ ___ ___ ___ — ___ ___ — ___ ___
YYYY MM DD