



Mucopolysaccharidosis and Other Storage Diseases Post-HSCT Data

Registry Use Only

Sequence Number:

Date Received:

CIBMTR Center Number:

CIBMTR Recipient ID:

Today's Date: / / (2 0)

Date of HSCT for which this form is being completed: / /

HSCT type: autologous allogeneic, unrelated allogeneic, related syngeneic (identical twin)

Product type: marrow PBSC cord blood other product, specify: _____

Visit: 100 day 6 month 1 year 2 years > 2 years, specify:

To be completed in conjunction with a Form 2100 – 100 Days Post-HSCT Data, Form 2200 – Six Months to Two Years Post-HSCT Data, or Form 2300 – Yearly Follow-Up for Greater Than Two Years Post-HSCT Data. Information reported here should reflect the date of last contact as reported in the post-HSCT data collection form, or immediately prior to death.

1. Indicate the enzyme that was evaluated for activity level in the first 100 days post-HSCT. The enzyme reported on this form must correspond to the enzyme found deficient at diagnosis.

Mucopolysaccharidosis

- 1 α -L-iduronidase (Hurler / Scheie syndrome – MPS I)
- 2 iduronate sulfatase (Hunter syndrome – MPS II)
- 3 heparan N-sulfatase (Sanfilippo A – MPS IIIA)
- 4 α -N-acetylglucosaminidase (Sanfilippo B – MPS IIIB)
- 5 acetyl-Co α -glucosaminide acetyltransferase (Sanfilippo C – MPS IIIC)
- 6 N-acetylglucosamine 6-sulfatase (Sanfilippo D – MPS IIID)
- 7 N-acetylgalactosamine 6-sulfatase (Morquio A – MPS IVA)
- 8 β -galactosidase (Morquio B – MPS IVB)
- 9 N-acetylgalactosamine 4-sulfatase (Maroteaux-Lamy syndrome – MPS VI)
- 10 β -glucuronidase (Sly syndrome – MPS VII)

Other Storage Diseases

- 11 glucocerebrosidase (Gaucher disease)
- 12 acid sphingomyelinase (Niemann-Pick disease)
- 13 N-acetylglucosamine-1-phosphotransferase (mucopolipidosis II or I-cell)
- 14 acid lipase deficiency (Wolman disease)
- 15 α -fucosidase deficiency (fucosidosis)
- 16 neuronal ceroid lipofuscinosis enzyme — NCL 1 (infantile): PPT-palmitoyl protein thioesterase
- 17 neuronal ceroid lipofuscinosis enzyme — NCL 2 (classic late infantile): transpeptidase
- 18 α -mannosidase B deficiency (α -mannosidosis)
- 19 N-aspartyl- β -glucosaminidase (aspartylglucosaminuria)
- 20 hypoxanthine-guanine phosphoribosyltransferase deficiency (Lesch-Nyhan syndrome)
- 21 other storage disease \longrightarrow 2. Specify: _____

3. Specify the recipient's enzyme activity level: .

- 1 nmol/hr/mg protein
- 2 pmol/hr/mg protein

4. Date of test: / / (2 0)

For 100-day follow-up reports, only questions 1–5 are required. Please sign below and submit only page 1 of this form. For all visits beyond 100 days post-HSCT, also continue with question 6.

5. Signed: (person completing form) _____

Please print name: _____

Phone number: (_____) _____ Fax number: (_____) _____

E-mail address: _____

Mail this form to your designated campus (Milwaukee or Minneapolis). Retain the original at the transplant center.

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6. Was any treatment given for the disease since the date of the last report?

- 1 yes
- 2 no
- 3 unknown

Specify disease treatment given since the date of the last report:

- 7. 1 yes 2 no enzyme replacement
- 8. 1 yes 2 no gene transfer / gene therapy
- 9. 1 yes 2 no substrate deprivation / inhibitor
- 10. 1 yes 2 no other

11. If yes, specify:

Clinical Status Post-HSCT

12. Was cerebrospinal fluid (CSF) testing performed since the date of the last report?

- 1 yes
- 2 no
- 3 unknown

Specify the results of most recent tests:

13. Date of most recent test:

- 1 known
 - 2 not known
- Month: Day: Year: 2 0

14. Opening pressure:

- 1 known
 - 2 not known
- cm H₂O

15. Closing pressure:

- 1 known
 - 2 not known
- cm H₂O

16. Total protein:

- 1 known
 - 2 not known
- . 1 mg/dL
2 g/L

17. Serum albumin:

- 1 known
 - 2 not known
- . 1 mg/dL
2 g/L

18. Serum IgG:

- 1 known
 - 2 not known
- . 1 mg/dL
2 g/L

19. Was Magnetic Resonance Imaging (MRI) of the brain and/or spine performed since the date of the last report?

- 1 yes
- 2 no
- 3 unknown

20. Date of most recent MRI:

- 1 known
 - 2 not known
- Month: Day: Year: 2 0

Specify the location of any abnormalities detected on MRI:

- 21. 1 yes 2 no 3 unknown hydrocephalus
- 22. 1 yes 2 no 3 unknown odontoid hypoplasia
- 23. 1 yes 2 no 3 unknown other

24. Specify:

25. Is a copy of the MRI report attached?

- 1 yes
- 2 no

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26. Was a Mental Development test performed since the date of the last report?

- 1 yes
- 2 no
- 3 unknown

27. Date of most recent test:

1 known →
2 not known

Month Day Year

28. Specify the test instrument used:

- 1 Bayley Scales of Infant Development
- 2 Stanford Binet Intelligence Scale 4th ed
- 3 Wechsler Preschool and Primary Scale of Intelligence (WPPSI – Revised)
- 4 Wechsler Intelligence Scale for Children – III (WISC – III)
- 5 other test →

30. Full scale score (not percentile):

1 known →
2 not known

31. Verbal score (not percentile):

1 known →
2 not known

32. Performance score (not percentile):

1 known →
2 not known

33. Were the Vineland Adaptive Behavior Scales performed since the date of the last report?

- 1 yes
- 2 no
- 3 unknown

34. Date of most recent test:

1 known →
2 not known

Month Day Year

35. Communication skills score:

1 known →
2 not known

36. Daily Living skills score:

1 known →
2 not known

37. Socialization skills score:

1 known →
2 not known

38. Was the recipient's visual acuity tested since the date of the last report?

- 1 yes
- 2 no
- 3 unknown

39. Date of most recent visual acuity test:

1 known →
2 not known

Month Day Year

40. Visual acuity (uncorrected) of right eye (OD):

1 known → /
2 not known

41. Visual acuity (uncorrected) of left eye (OS):

1 known → /
2 not known

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42. Visual acuity (uncorrected) of both eyes (OU):

1 known → /
2 not known

43. Was corneal clouding present?

1 yes
2 no

44. Did the recipient undergo an ophthalmologic exam under anesthesia since the date of the last report?

1 yes →
2 no
3 unknown

45. Date of most recent ophthalmologic exam:

1 known → /
2 not known
Month Day Year

46. Specify results:

1 normal
2 abnormal / impaired

47. Is a copy of the report attached?

1 yes
2 no

48. Was an audiologic evaluation (auditory brain stem or conditioned response) performed since the date of the last report?

1 yes →
2 no
3 unknown

49. Date of most recent audiologic evaluation:

1 known → /
2 not known
Month Day Year

Specify tympanometry results:

50. 1 normal 2 retracted 3 flat Right ear

51. 1 normal 2 retracted 3 flat Left ear

52. Was the hearing loss (HL) in decibels (dB) assessed at the speech threshold for 500 hertz (HZ) since the date of the last report?

1 yes →
2 no
3 unknown

53. Date of most recent evaluation:

1 known → /
2 not known
Month Day Year

Specify tympanometry results: (See *Degree of Hearing Loss chart below for scale ranges.*)

54. 1 normal / mild 2 moderate / moderately severe 3 severe / profound Right ear

55. 1 normal / mild 2 moderate / moderately severe 3 severe / profound Left ear

56. Was the hearing loss (HL) in decibels (dB) assessed at the speech threshold for 2000 hertz (HZ) since the last report?

1 yes →
2 no
3 unknown

57. Date of most recent evaluation:

1 known → /
2 not known
Month Day Year

Specify tympanometry results: (See *Degree of Hearing Loss chart below for scale ranges.*)

58. 1 normal / mild 2 moderate / moderately severe 3 severe / profound Right ear

59. 1 normal / mild 2 moderate / moderately severe 3 severe / profound Left ear

Degree of Hearing Loss: Pure Tones and Speech Testing

Normal:	0–20 dB HL	Moderate:	45–55 dB HL	Severe:	75–90 dB HL
Mild:	25–40 dB HL	Moderately Severe:	60–70 dB HL	Profound:	> 90 dB HL

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60. Has there been a change in the recipient's neurologic status since the date of the last report?
(Report clinical status, not neuropsychological status.)

- 1 yes
- 2 stable / unchanged
- 3 unknown

61. Date of most recent evaluation:

- 1 known
 - 2 not known
- Month: Day: Year: 2 0

62. Specify current neurologic status compared to previous report:

- 1 improved
- 2 worsened

63. Is a copy of the physical exam or neurologic exam attached?

- 1 yes
- 2 no

64. Was a pulmonary evaluation performed since the date of the last report?

- 1 yes
- 2 no
- 3 unknown

65. Date of most recent pulmonary evaluation:

- 1 known
 - 2 not known
- Month: Day: Year: 2 0

66. Specify oxygen saturation on room air: %

67. Specify the results of the most recent pulmonary function test:

- 1 normal
- 2 abnormal / impaired

68. Is a copy of the pulmonary evaluation report attached?

- 1 yes
- 2 no

69. Was an echocardiogram performed since the date of the last report?

- 1 yes
- 2 no
- 3 unknown

70. Date of most recent echocardiogram:

- 1 known
 - 2 not known
- Month: Day: Year: 2 0

Specify the findings for valvular insufficiency:

- 71. 1 none 2 mild / trivial 3 moderate / severe 4 valve replacement Aortic
- 72. 1 none 2 mild / trivial 3 moderate / severe 4 valve replacement Mitral
- 73. 1 none 2 mild / trivial 3 moderate / severe 4 valve replacement Pulmonary
- 74. 1 none 2 mild / trivial 3 moderate / severe 4 valve replacement Tricuspid

75. Was cardiac contractility examined since the date of the last report?

- 1 yes
- 2 no
- 3 unknown

76. Date of most recent exam:

- 1 known
 - 2 not known
- Month: Day: Year: 2 0

77. Specify the method used to assess left ventricle performance:

- 1 ejection fraction
- 2 shortening fraction

78. Specify fraction: %

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79. Was a dobutamine stress echocardiogram performed since the date of the last report?

- 1 yes →
- 2 no
- 3 unknown

80. Date of most recent dobutamine stress echocardiogram:

- 1 known →
- 2 not known

Month Day Year

81. Specify the results of the most recent test:

- 1 normal
- 2 abnormal / impaired

82. Is a copy of the report attached?

- 1 yes
- 2 no

83. Was orthopedic surgery performed since the date of the last report?

- 1 yes →
- 2 no
- 3 unknown

84. Date of most recent orthopedic surgery:

- 1 known →
- 2 not known

Month Day Year

Specify the surgery site(s):

85. 1 yes 2 no Fingers

86. 1 yes 2 no Hips

87. 1 yes 2 no Knees

88. 1 yes 2 no Spine

89. 1 yes 2 no Wrist (carpal tunnel syndrome)

90. 1 yes 2 no Other site →

91. Specify: _____