

Mucopolysaccharidosis and Other Storage Diseases Pre-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: / / (2 0)

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

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

This form must be accompanied by Form 2000 – Recipient Baseline Data. All information in the box above, including the date, should be identical with the corresponding Form 2000. Information should come from an actual examination by the Transplant Center physician, or the physician who is following the recipient pre-HSCT, or abstraction of the recipient's medical records.

If this is a report of a second or subsequent transplant, check here and continue with question 11.

1. What was the date of diagnosis of Mucopolysaccharidosis / Other Storage Disease? / /

2. Did the diagnosis include a Sanfilippo-type enzyme deficiency?

- 1 yes →
 2 no

3. Specify enzyme:

- 1 heparan N-sulfatase (Sanfilippo A – MPS IIIA)
 2 α-N-acetylglucosaminidase (Sanfilippo B – MPS IIIB)
 3 acetyl-Coα-glucosaminide acetyltransferase (Sanfilippo C – MPS IIIC)
 4 N-acetylglucosamine 6-sulfatase (Sanfilippo D – MPS IIID)

4. Did the diagnosis include a Morquio-type enzyme deficiency?

- 1 yes →
 2 no

5. Specify enzyme:

- 1 N-acetyl-galactosamine-6-sulfatase (Morquio A – MPS IVA)
 2 β-galactosidase (Morquio B – MPS IVB)

6. Was the diagnosis neuronal ceroid lipofuscinosis (Batten disease)?

- 1 yes →
 2 no

7. Specify the disease subtype:

- 1 neuronal ceroid lipofuscinosis enzyme — NCL 1 (infantile): PPT-palmitoyl protein thioesterase
 2 neuronal ceroid lipofuscinosis enzyme — NCL 2 (classic late infantile): transpeptidase

Specify the leukocyte enzyme activity levels at diagnosis:

8. Date enzyme levels tested: / /

9. Patient enzyme level: • 1 nmol/hr/mg protein 2 pmol/hr/mg protein

10. Donor enzyme level: • 1 nmol/hr/mg protein 2 pmol/hr/mg protein 3 unknown

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11. Was treatment given for the disease between diagnosis and any time prior to the HSCT?

- 1 yes
2 no
3 unknown

Specify treatment(s):

12. 1 yes 2 no 3 unknown Enzyme replacement
13. 1 yes 2 no 3 unknown Gene transfer / gene therapy
14. 1 yes 2 no 3 unknown Substrate deprivation / inhibitor
15. 1 yes 2 no 3 unknown Other treatment

16. If yes, specify:

Clinical Status Prior to HSCT

17. Was cerebrospinal fluid (CSF) testing performed prior to HSCT?

- 1 yes
2 no
3 unknown

18. Date of most recent test prior to HSCT: unknown

Month

Day

Year

Report the results of the most recent test:

19. Opening pressure

- 1 known cm H₂O
2 unknown

20. Total protein

- 1 known . 1 mg/dL 2 g/L
2 unknown

21. Serum albumin

- 1 known . 1 mg/dL 2 g/L
2 unknown

22. Serum IgG

- 1 known . 1 mg/dL 2 g/L
2 unknown

23. Was magnetic resonance imaging (MRI) of the brain / spine performed prior to HSCT?

- 1 yes
2 no
3 unknown

24. Date of most recent MRI prior to HSCT: unknown

Month

Day

Year

Specify the location(s) of the abnormalities:

25. 1 yes 2 no 3 unknown Odontoid hypoplasia
26. 1 yes 2 no 3 unknown Ventricular (hydrocephalus)
27. 1 yes 2 no 3 unknown Other abnormality

28. If yes, specify:

29. Is a copy of the MRI report attached?

- 1 yes
2 no

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30. Was mental development testing performed prior to HSCT?

- 1 yes
- 2 no
- 3 unknown

31. Date of most recent test prior to HSCT: unknown
Month Day Year

32. Specify the test instrument used:

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

33. Specify other test instrument:

34. Full scale score: (not percentile) score unknown

35. Verbal score: (not percentile) score unknown

36. Performance score: (not percentile) score unknown

37. Were the Vineland Adaptive Behavior Scales performed at any time prior to HSCT?

- 1 yes
- 2 no
- 3 unknown

38. Date of most recent test prior to HSCT: unknown:
Month Day Year

39. Communication skills: score unknown

40. Daily living skills: score unknown

41. Socialization skills: score unknown

42. Was an eye exam performed at any time prior to HSCT?

- 1 yes
- 2 no
- 3 unknown

43. Date of most recent exam prior to HSCT: unknown
Month Day Year

Visual acuity (uncorrected vision only):

44. Right eye (OD): / unknown

45. Left eye (OS): / unknown

46. Binocular / both eyes (OU): / unknown

47. Was corneal clouding present?

- 1 yes
- 2 no
- 3 unknown

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48. Was an ophthalmologic exam performed under anesthesia at any time prior to HSCT?

- 1 yes
2 no
3 unknown

49. Date of most recent exam prior to HSCT: unknown
Month Day Year

50. Specify exam results:
1 normal
2 abnormal / impaired
3 unknown

51. Is a copy of the report attached?
1 yes
2 no

52. Was a hearing test performed at any time prior to HSCT?

- 1 yes
2 no
3 unknown

53. Date of most recent test prior to HSCT: unknown
Month Day Year

54. Specify test results:
1 normal
2 abnormal / impaired
3 unknown

55. Is a copy of the report attached?
1 yes
2 no

56. Was pulmonary function evaluated at any time prior to HSCT?

- 1 yes
2 no
3 unknown

57. Date of most recent test prior to HSCT: unknown
Month Day Year

58. Oxygen saturation on room air: % unknown

59. Specify results of pulmonary evaluation:
1 normal
2 abnormal / impaired
3 unknown

60. Is a copy of the report attached?
1 yes
2 no

61. Was an echocardiogram performed at any time prior to HSCT?

- 1 yes
2 no
3 unknown

62. Date of most recent test prior to HSCT: unknown
Month Day Year

Specify valvular insufficiency:

63. Aortic
1 none
2 mild or trivial
3 moderate or severe
4 valve replacement
5 unknown

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- 64. Mitral
 - 1 none
 - 2 mild or trivial
 - 3 moderate or severe
 - 4 valve replacement
 - 5 unknown
- 65. Pulmonary
 - 1 none
 - 2 mild or trivial
 - 3 moderate or severe
 - 4 valve replacement
 - 5 unknown
- 66. Tricuspid
 - 1 none
 - 2 mild or trivial
 - 3 moderate or severe
 - 4 valve replacement
 - 5 unknown

67. Was a cardiac contractility test performed at any time prior to HSCT?

- 1 yes
- 2 no
- 3 unknown

68. Date of most recent test prior to HSCT: unknown
Month Day Year

69. Ejection fraction: % unknown

70. Shortening fraction: % unknown

71. Signed: _____
Person completing form

Please print name: _____

Phone: (_____) _____

Fax: (_____) _____

E-mail address: _____