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.

Mucopolysaccharidoses and Other Storage Diseases
Post-HSCT Data

Registry Use Only

Visit: □ 100 day □ 6 month □ year

Today's Date: [ ] [ ] [ ]

Infusion Date: [ ] [ ] [ ]

CIBMTR Recipient ID: [ ] [ ] [ ]

CIBMTR Center Number: [ ] [ ] [ ]

Date of test: [ ] [ ] [ ]

Date of HSCT for which this form is being completed: [ ] [ ] [ ]

HSCT type: □ autologous □ allogeneic □ unrelated □ 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.

Mucopolysaccharidoses
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-CoA-glucosaminide acetyltransferase (Sanfilippo C – MPS IIIC)
6. N-acetylglucosamine 6-sulfatase (Sanfilippo D – MPS IIID)
7. N-acetylglucosamine-1-phosphotransferase
8. β-glucuronidase (Sly syndrome – MPS VII)

Other Storage Diseases
11. glucocerebrosidase (Gaucher disease)
12. acid sphingomyelinase (Niemann-Pick disease)
13. N-acetylgalactosamine-1-phosphotransferase (mucolipidosis 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

3. Specify the recipient’s enzyme activity level: [ ] [ ] [ ]

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

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: ____________________________

Fax this form to your designated campus (Milwaukee 414-805-0714 or Minneapolis 612-627-5895).

Mail this form to your designated campus (Milwaukee or Minneapolis). Retain the original at the transplant center.
### CIBMTR Form 2138 (MUC) v1.0 (2–6) July 2007

**CIBMTR Recipient ID:**

**CIBMTR Center Number:**

**Today's Date:**

**Infusion Date:**

**CIBMTR Center Number:**

**CIBMTR Recipient ID:**

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. [ ] yes  
   1. [ ] enzyme replacement  
8. [ ] yes  
   1. [ ] gene transfer / gene therapy  
9. [ ] yes  
   1. [ ] substrate deprivation / inhibitor  
10. [ ] yes  
    1. [ ] 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

14. **Opening pressure:**

1. [ ] known  
2. [ ] not known

15. **Closing pressure:**

1. [ ] known  
2. [ ] not known

16. **Total protein:**

1. [ ] known  
2. [ ] not known

17. **Serum albumin:**

1. [ ] known  
2. [ ] not known

18. **Serum IgG:**

1. [ ] known  
2. [ ] not known

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

Specify the location of any abnormalities detected on MRI:

20. **Date of most recent MRI:**

1. [ ] known  
2. [ ] not known

21. [ ] yes  
   1. [ ] unknown hydrocephalus

22. [ ] yes  
   1. [ ] unknown odontoid hypoplasia

23. [ ] yes  
   1. [ ] unknown other

24. Specify: ______________________

25. **Is a copy of the MRI report attached?**

1. [ ] yes  
2. [ ] no

Fax this form to your designated campus (Milwaukee 414-805-0714 or Minneapolis 612-627-5895).
### Questionnaire:

<table>
<thead>
<tr>
<th>Question</th>
<th>Options</th>
</tr>
</thead>
<tbody>
<tr>
<td>26. Was a Mental Development test performed since the date of the last report?</td>
<td>1 yes, 2 no, 3 unknown</td>
</tr>
<tr>
<td>27. Date of most recent test:</td>
<td>Month Day Year</td>
</tr>
<tr>
<td>28. Specify the test instrument used:</td>
<td>Bayley Scales of Infant Development, Stanford Binet Intelligence Scale 4th ed, Wechsler Preschool and Primary Scale of Intelligence (WPPSI – Revised), Wechsler Intelligence Scale for Children – III (WISC – III), other test</td>
</tr>
<tr>
<td>29. Specify:</td>
<td></td>
</tr>
<tr>
<td>30. Full scale score (not percentile):</td>
<td></td>
</tr>
<tr>
<td>31. Verbal score (not percentile):</td>
<td></td>
</tr>
<tr>
<td>32. Performance score (not percentile):</td>
<td></td>
</tr>
<tr>
<td>33. Were the Vineland Adaptive Behavior Scales performed since the date of the last report?</td>
<td>1 yes, 2 no, 3 unknown</td>
</tr>
<tr>
<td>34. Date of most recent test:</td>
<td>Month Day Year</td>
</tr>
<tr>
<td>35. Communication skills score:</td>
<td></td>
</tr>
<tr>
<td>36. Daily Living skills score:</td>
<td></td>
</tr>
<tr>
<td>37. Socialization skills score:</td>
<td></td>
</tr>
<tr>
<td>38. Was the recipient's visual acuity tested since the date of the last report?</td>
<td>1 yes, 2 no, 3 unknown</td>
</tr>
<tr>
<td>39. Date of most recent visual acuity test:</td>
<td>Month Day Year</td>
</tr>
<tr>
<td>40. Visual acuity (uncorrected) of right eye (OD):</td>
<td></td>
</tr>
<tr>
<td>41. Visual acuity (uncorrected) of left eye (OS):</td>
<td></td>
</tr>
</tbody>
</table>
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

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

Specify tympanometry results:
50. Right ear
   1. normal
   2. retracted
   3. flat

51. Left ear
   1. normal
   2. retracted
   3. flat

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

Specify tympanometry results:
54. Right ear
   1. normal / mild
   2. moderate / moderately severe
   3. severe / profound

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

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

Specify tympanometry results:
58. Right ear
   1. normal / mild
   2. moderate / moderately severe
   3. severe / profound

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

Degree of Hearing Loss: Pure Tones and Speech Testing
- Normal: 0–20 dB HL
- Mild: 25–40 dB HL
- Moderate: 45–65 dB HL
- Moderately Severe: 60–70 dB HL
- Severe: 75–90 dB HL
- Profound: > 90 dB HL

Fax this form to your designated campus (Milwaukee 414-805-0714 or Minneapolis 612-627-5895).
### CIBMTR Form 2138 (MUC) v1.0 (5–6) July 2007

**For internal use only:** Document F00549 version 1.0   Replaces: n/a

**CIBMTR Recipient ID:**

**CIBMTR Center Number:**

**Today's Date:**

<table>
<thead>
<tr>
<th>Month</th>
<th>Day</th>
<th>Year</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>20</td>
<td></td>
</tr>
</tbody>
</table>

**Infusion Date:**

<table>
<thead>
<tr>
<th>Month</th>
<th>Day</th>
<th>Year</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>20</td>
<td></td>
</tr>
</tbody>
</table>

**CIBMTR Center Number:**

**CIBMTR Recipient ID:**

---

**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.)*

<table>
<thead>
<tr>
<th>1</th>
<th>yes</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>stable / unchanged</td>
</tr>
<tr>
<td>3</td>
<td>unknown</td>
</tr>
</tbody>
</table>

**61. Date of most recent evaluation:**

<table>
<thead>
<tr>
<th>Month</th>
<th>Day</th>
<th>Year</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>20</td>
</tr>
</tbody>
</table>

**62. Specify current neurologic status compared to previous report:**

<table>
<thead>
<tr>
<th>1</th>
<th>improved</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>worsened</td>
</tr>
</tbody>
</table>

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

<table>
<thead>
<tr>
<th>1</th>
<th>yes</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>no</td>
</tr>
</tbody>
</table>

---

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

<table>
<thead>
<tr>
<th>1</th>
<th>yes</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>no</td>
</tr>
<tr>
<td>3</td>
<td>unknown</td>
</tr>
</tbody>
</table>

**65. Date of most recent pulmonary evaluation:**

<table>
<thead>
<tr>
<th>Month</th>
<th>Day</th>
<th>Year</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>20</td>
</tr>
</tbody>
</table>

**66. Specify oxygen saturation on room air:**

| % |

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

<table>
<thead>
<tr>
<th>1</th>
<th>normal</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>abnormal / impaired</td>
</tr>
</tbody>
</table>

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

<table>
<thead>
<tr>
<th>1</th>
<th>yes</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>no</td>
</tr>
</tbody>
</table>

---

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

<table>
<thead>
<tr>
<th>1</th>
<th>yes</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>no</td>
</tr>
<tr>
<td>3</td>
<td>unknown</td>
</tr>
</tbody>
</table>

**70. Date of most recent echocardiogram:**

<table>
<thead>
<tr>
<th>Month</th>
<th>Day</th>
<th>Year</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>20</td>
</tr>
</tbody>
</table>

**Specify the findings for valvular insufficiency:**

<table>
<thead>
<tr>
<th>1</th>
<th>none</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>mild / trivial</td>
</tr>
<tr>
<td>3</td>
<td>moderate / severe</td>
</tr>
<tr>
<td>4</td>
<td>valve replacement Aortic</td>
</tr>
<tr>
<td>71.</td>
<td></td>
</tr>
<tr>
<td>72.</td>
<td></td>
</tr>
<tr>
<td>73.</td>
<td></td>
</tr>
<tr>
<td>74.</td>
<td></td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>1</th>
<th>none</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>mild / trivial</td>
</tr>
<tr>
<td>3</td>
<td>moderate / severe</td>
</tr>
<tr>
<td>4</td>
<td>valve replacement Mitral</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>1</th>
<th>none</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>mild / trivial</td>
</tr>
<tr>
<td>3</td>
<td>moderate / severe</td>
</tr>
<tr>
<td>4</td>
<td>valve replacement Pulmonary</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>1</th>
<th>none</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>mild / trivial</td>
</tr>
<tr>
<td>3</td>
<td>moderate / severe</td>
</tr>
<tr>
<td>4</td>
<td>valve replacement Tricuspid</td>
</tr>
</tbody>
</table>

---

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

<table>
<thead>
<tr>
<th>1</th>
<th>yes</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>no</td>
</tr>
<tr>
<td>3</td>
<td>unknown</td>
</tr>
</tbody>
</table>

**76. Date of most recent exam:**

<table>
<thead>
<tr>
<th>Month</th>
<th>Day</th>
<th>Year</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>20</td>
</tr>
</tbody>
</table>

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

<table>
<thead>
<tr>
<th>1</th>
<th>ejection fraction</th>
</tr>
</thead>
<tbody>
<tr>
<td>2</td>
<td>shortening fraction</td>
</tr>
</tbody>
</table>

**78. Specify fraction:**

| % |

---

Fax this form to your designated campus (Milwaukee 414-805-0714 or Minneapolis 612-627-5895).
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:** __________________________