Form 2138 R2.0: Mucopolysaccharidosis and Other Storage Diseases Post-HSCT Data

Center: CRID:

Key Fields

Sequence Number: _____

Date Received: __ __ __ __ - __ __- __ __

CIBMTR Center Number _____

CIBMTR Recipient ID: _____

Today's Date: __ __ __ __ - __ __- __ __

Date of HSCT for which this form is being completed: __ __ __ __ - __ __- __ __

HSCT type: (check all that apply)

☐ Autologous

☐ Allogeneic, unrelated

☐ Allogeneic, related

☐ Syngeneic (identical twin)

Product type: (check all that apply)

☐ Marrow

☐ PBSC

☐ Cord blood

☐ Other product

Specify: _____

Visit:

☐ 100 day ☐ 6 months ☐ 1 year ☐ 2 years ☐ > 2 years,

Specify: _____

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Center: _____   CRID: _____

First 100 days post-HSCT	Questions: 1 - 10

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

- α-L-iduronidase (Hurler / Scheie syndrome - MPS I)
- idurndate sulfatase (Hunter syndrome - MPS II)
- heparan N-sulfatase (Sanfilippo A - MPS IIIA)
- α-N-acetylglucosaminidase (Sanfilippo B -MPS IIIB)
- acetyl-Coα-glucosaminide acetyltransferase (Sanfilippo C - MPS IIIC)
- N-acetylglucosamine 6-sulfatase (Sanfilippo D - MPS IIID)
- N-acetyl-galactosamine-6-sulfatase (Morquio A - MPS IVA)
- β-galactosidase (Morquio B - MPS IVB)
- N-acetlygalactosamine 74-sulfatase (Maroteaux-Lamy syndrome - MPS VI)
- β-glucuronidase (Sly syndrome - MPS VII)
- glucocerebrosidase (Gaucher disease)
- acid sphingomyelinase (Niemann-Pick disease)
- N-acetylglucosamine-1-phosphotransferase (mucolipidosis II or I-cell)
- acid lipase deficiency (Wolman disease)
- α-fucosidase deficeincy (fucosidosis)
- neuronal ceriod lipofuscinosis enzyme - NCL 1 (infantile): PPT-palmitoyl protein thiosterase
- neuronal ceriod lipofuscinosis enzyme - NCL 2 (classic late infantile); transpeptidase
- α-mannosidase B deficiency (α-mannosidosis)
- N-aspartyl-β-glucosaminidase (aspartylglucosaminuria)
- hypoxanthine-guanine phosphoribosyltransferase deficiency (Lesch-Nyhan syndrome)
- Other storage disease

2 Specify: _____

3 Specify the recipient's enzyme activity level: _____

- nmol/hr/mg protein
- pmol/hr/mg protein

4 Date of test: __ __ __ __ - __ __ - __ __

For 100-day follow-up reports, only questions 1-4 are required. Please sign below and submit only this part of the form. For all visits beyond 100 days post-HSCT, go to question 5 and complete the rest of the form.

First Name: _____   Last Name: _____

Phone number: _____   Fax number: _____

E-mail address: _____

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

- yes
- no
- Unknown

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

6 enzyme replacement

- yes
- no

7 gene transfer / gene therapy

☐ yes ☐ no

8 substrate deprevation

☐ yes ☐ no

9 Other

☐ yes ☐ no

    10 Specify _____

Clinical Status Post-HSCT   Questions: 11 - 119

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

☐ yes ☐ no ☐ Unknown

    Specify the results of most recent tests:

12 Date of most recent test:

☐ Known ☐ Not known

    13 Date: __ __ __ __ - __ __- __ __

14 Opening pressure:

☐ Known ☐ Not known

    15 _____ cm H$_2$O

16 Closing pressure:

☐ Known ☐ Not known

    17 _____ cm H$_2$O

18 Total protein:

☐ Known ☐ Not known

    19 Total protein: _____

    ☐ mg/dL ☐ g/L

20 Serum albumin:

☐ Known ☐ Not known

    21 Serum albumin: _____

    ☐ mg/dL ☐ g/L

22 Serum IgG:

☐ Known ☐ Not known

    23 Serum IgG: _____

    ☐ mg/dL ☐ g/L

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

☐ yes ☐ no ☐ Unknown

    25 Date of most recent MRI:

☐ Known ☐ Not known

    26 Date: __ __ __ __ - __ __- __ __

Specify the location of any abnormalities detected on MRI:

27 hydrocephalus

☐ yes   ☐ no   ☐ Unknown

28 odontoid hypoplasia

☐ yes   ☐ no   ☐ Unknown

29 Other

☐ yes   ☐ no   ☐ Unknown

30 Specify: _____

31 Is a copy of the MRI report attached?

☐ yes   ☐ no

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

☐ yes   ☐ no   ☐ Unknown

33 Date of most recent test:

☐ Known   ☐ Not known

34 Date: __ __ __ __ - __ __- __ __

35 Specify the test instrument used:

☐ Bayley Scales of Infant Development

☐ Stanford Binet Intelligence Scale

☐ Wechsler Preschool and Primary Scale of Intelligence (WPPSI - Revised)

☐ Wechsler Intelligence Scale for Children - III (WISC - III)

☐ other test

36 Specify: _____

37 Full scale score (not percentile):

☐ Known   ☐ Not known

38 Score: _____

39 Verbal score (not percentile):

☐ Known   ☐ Not known

40 Score: _____

41 Performance score (not percentile):

☐ Known   ☐ Not known

42 Score: _____

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

☐ yes   ☐ no   ☐ Unknown

44 Date of most recent test:

☐ Known   ☐ Not known

45 Date: __ __ __ __ - __ __- __ __

46 Communication skills score:

☐ Known   ☐ Not known

47 Score: _____

48 Daily living skills score:

     ☐ Known   ☐ Not known

       49 Score: _____

50 Socialization skills score:

     ☐ Known   ☐ Not known

       51 Score: _____

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

     ☐ yes   ☐ no   ☐ Unknown

     53 Date of most recent visual acuity test:

       ☐ Known   ☐ Not known

       54 Date: __ __ __ __ - __ __- __ __

     55 Visual acuity (uncorrected) of right eye (OD):

       ☐ Known   ☐ Not known

       56 _____ / _____

     57 Visual acuity (uncorrected) of left eye (OS):

       ☐ Known   ☐ Not known

       58 _____ / _____

     59 Visual acuity (uncorrected) of both eyes (OU):

       ☐ Known   ☐ Not known

       60 _____ / _____

     61 Was corneal clouding present?

       ☐ yes   ☐ no

62 Did the recipient undergo an opthalmologic exam under anesthesia since the date of the last report?

     ☐ yes   ☐ no   ☐ Unknown

     63 Date of most recent opthalmologic exam:

       ☐ Known   ☐ Not known

       64 Date: __ __ __ __ - __ __- __ __

     65 Specify results:

       ☐ Normal   ☐ abnormal/impaired

     66 Is a copy of the report attached?

       ☐ yes   ☐ no

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

     ☐ yes   ☐ no   ☐ Unknown

     68 Date of most recent audiologic evaluation:

       ☐ Known   ☐ Not known

       69 Date: __ __ __ __ - __ __- __ __

     Specify tympanometry results:

     70 Right ear

       ☐ Normal   ☐ retracted   ☐ flat

71 Left ear

   ☐ Normal   ☐ retracted   ☐ flat

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

☐ yes   ☐ no   ☐ Unknown

   73 Date of most recent evaluation:

     ☐ Known   ☐ Not known

       74 Date: __ __ __ __ - __ __- __ __

   Specify tympanometry results:

   75 Right ear

     ☐ normal/mild = 0-20 dB HL/25-40 dB HL

     ☐ moderate/moderately severe = 45-55 dB HL/60-70 dB HL

     ☐ severe/profound = 75-90 dB HL/>90 dB HL

   76 Left ear

     ☐ normal/mild = 0-20 dB HL/25-40 dB HL

     ☐ moderate/moderately severe = 45-55 dB HL/60-70 dB HL

     ☐ severe/profound = 75-90 dB HL/> 90 dB HL

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

☐ yes   ☐ no   ☐ Unknown

   78 Date of most recent evaluation:

     ☐ Known   ☐ Not known

       79 Date: __ __ __ __ - __ __- __ __

   Specify tympanometry results:

   80 Right ear

     ☐ normal/mild = 0-20 dB HL/25-40 dB HL

     ☐ moderate/moderately severe = 45-55 dB HL/60-70 dB HL

     ☐ severe/profound = 75-90 dB HL/>90 dB HL

   81 Left ear

     ☐ normal/mild = 0-20 dB HL/25-40 dB HL

     ☐ moderate/moderately severe = 45-55 dB HL/60-70 dB HL

     ☐ severe/profound = 75-90 dB HL/> 90 dB HL

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

☐ Yes

☐ stable/
  unchanged

☐ Unknown

   83 Date of most recent evaluation:

     ☐ Known   ☐ Not known

       84 Date: __ __ __ __ - __ __- __ __

85 Specify current neurologic status compared to previous report:

  ☐ Improved   ☐ worsened

86 Is a copy of the physical exam or neurologic exam attached?

  ☐ yes   ☐ no

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

  ☐ yes   ☐ no   ☐ Unknown

88 Date of most recent pulmonary evaluation:

  ☐ Known   ☐ Not known

89 Date: __ __ __ __ - __ __- __ __

90 Specify oxygen saturation on room air: _____ %

91 Specify the results of the most recent pulmonary function test:

  ☐ Normal   ☐ abnormal/impaired

92 Is a copy of the pulmonary evaluation report attached?

  ☐ yes   ☐ no

93 Was an echocardiogram performed since the date of the last report?

  ☐ yes   ☐ no   ☐ Unknown

94 Date of most recent echocardiogram:

  ☐ Known   ☐ Not known

95 Date: __ __ __ __ - __ __- __ __

Specify the findings for valvular insufficiency:

96 Aortic

  ☐ None   ☐ mild or trivial   ☐ moderate or severe   ☐ valve replacement

97 Mitral

  ☐ None   ☐ mild or trivial   ☐ moderate or severe   ☐ valve replacement

98 Pulmonary

  ☐ None   ☐ mild or trivial   ☐ moderate or severe   ☐ valve replacement

99 Tricuspid

  ☐ None   ☐ mild or trivial   ☐ moderate or severe   ☐ valve replacement

100 Was cardiac contractility examined since the date of the last report?

  ☐ yes   ☐ no   ☐ Unknown

101 Date of most recent exam:

  ☐ Known   ☐ Not known

102 Date: __ __ __ __ - __ __- __ __

103 Specify the method used to assess left ventricle performance:

  ☐ ejection fraction   ☐ shortening fraction

104 Specify fraction: _____ %

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

  ☐ yes   ☐ no   ☐ Unknown

106 Date of most recent dobutamine stress echocardiogram:

  ☐ Known   ☐ Not known

107 Date: __ __ __ __ - __ __- __ __

108 Specify the results of the most recent test:

☐ Normal   ☐ abnormal/impaired

109 Is a copy of the report attached?

☐ yes   ☐ no

110 Was orthopedic surgery performed since the date of the last report?

☐ yes   ☐ no   ☐ Unknown

111 Date of most recent orthopedic surgery:

☐ Known   ☐ Not known

112 Date: __ __ __ __ - __ __- __ __

Specify the surgery site(s)

113 Fingers

☐ yes   ☐ no

114 Hips

☐ yes   ☐ no

115 Knees

☐ yes   ☐ no

116 Spine

☐ yes   ☐ no

117 Wrist (carpal tunnel syndrome)

☐ yes   ☐ no

118 Other site:

☐ yes   ☐ no

119 Specify: _____