Form 2038 R2.0: Mucopolysaccharidosis and Other Storage Diseases Pre-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: _____

Mucopolysaccharidosis and Other Storage Diseases   Questions: 1 - 16

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

1 What ws the date of diagnosis of Mucopolysaccharidosis / Other Storage Disease? __ __ __ __ - __ __- __ __

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

☐ yes   ☐ no

     3 Specify enzyme:

         ☐ 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)

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

☐ yes   ☐ no

     5 Specify enzyme:

         ☐ N-acetyl-galactosamine-6-sulfatase (Morquio A - MPS IVA)

         ☐ β-galactosidase (Morquio B - MPS IVB)

6 Was the diagnosis neuronal ceriod lipofuscinosis (Battan disease)?

☐ yes   ☐ no

     7 Specify the disease subtype:

         ☐ neuronal ceriod lipofuscinosis enzyme - NCL 1 (infantile): PPT-palmitoyl protein thiosterase

         ☐ neuronal ceriod lipofuscinosis enzyme - NCL 2 (classic late infantile); transpeptidase

Center: CRID:

Specify the leukocyte enzyme activity levels at diagnosis:

8   Date enzyme levels tested: __ __ __ __ - __ __- __ __

9   Patient enzyme level: _____   Unit: ☐ nmol/hr/mg protein   ☐ pmol/hr/mg protein

10   Donor enzyme level: _____   Unit: ☐ nmol/hr/mg protein   ☐ pmol/hr/mg protein   ☐ Unknown

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

    ☐ yes   ☐ no   ☐ Unknown

       Specify treatment(s):

    12   Enzyme replacement

       ☐ yes   ☐ no   ☐ Unknown

    13   Gene transfer / gene therapy

       ☐ yes   ☐ no   ☐ Unknown

    14   Substrate deprivation / inhibitor

       ☐ yes   ☐ no   ☐ Unknown

    15   Other treatment

       ☐ yes   ☐ no   ☐ Unknown

       16   Specify: _____

Clinical Status Prior to HSCT   Questions: 17 - 74

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

    ☐ yes   ☐ no   ☐ Unknown

    18   Date of most recent test prior to HSCT: __ __ __ __ - __ __- __ __   ☐ Unknown

       Report the results of the most recent test:

    19   Opening pressure

       ☐ Known   ☐ Unknown

       20   _____ cm H_2O

    21   Total protein

       ☐ Known   ☐ Unknown

       22   _____   ☐ mg/dL   ☐ g/L

    23   Serum albumin

       ☐ Known   ☐ Unknown

       24   _____   ☐ mg/dL   ☐ g/L

    25   Serum IgG

       ☐ Known   ☐ Unknown

       26   _____   ☐ mg/dL   ☐ g/L

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

    ☐ yes   ☐ no   ☐ Unknown

    28   Date of most recent MRI prior to HSCT: __ __ __ __ - __ __- __ __   ☐ Unknown

Specify the location(s) of the abnormalities:

29 Odontoid hypoplasia

☐ yes ☐ no ☐ Unknown

30 Ventricular (hydrocephalus)

☐ yes ☐ no ☐ Unknown

31 Other abnornmality

☐ yes ☐ no ☐ Unknown

32 Specify: _____

33 Is a copy of the MRI report attached?

☐ yes ☐ no

34 Was mental development testing performed prior to the HSCT?

☐ yes ☐ no ☐ Unknown

35 Date of most recent test prior to HSCT: __ __ __ __ - __ __- __ __ ☐ Unknown

36 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

37 Specify other test instrument: _____

38 Full scale score *(not percentile)* _____ ☐ Unknown

39 Verbal score: *(not percentile)* _____ ☐ Unknown

40 Performance score: *(not percentile)* _____ ☐ Unknown

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

☐ yes ☐ no ☐ Unknown

42 Date of most recent test prior to HSCT: __ __ __ __ - __ __- __ __ ☐ Unknown

43 Communication skills: _____ ☐ score unknown

44 Daily living skills: _____ ☐ score unknown

45 Socialization skills: _____ ☐ score unknown

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

☐ yes ☐ no ☐ Unknown

47 Date of most recent exam prior to HSCT: __ __ __ __ - __ __- __ __ ☐ Unknown

Visual acuity (uncorrected vision only):

48 Right eye (OD): _____ / _____

☐ Unknown

49 Left eye (OS): _____ / _____

☐ Unknown

50 Binocular / both eyes(OU): _____ / _____

☐ Unknown

51   Was corneal clouding present?

☐ yes   ☐ no   ☐ Unknown

52   Was an opthalmologic exam performed under anesthesia at any time prior to HSCT?

☐ yes   ☐ no   ☐ Unknown

53   Date of most recent exam prior to HSCT: __ __ __ __ - __ __- __ __   ☐ Unknown

54   Specify exam results:

☐ Normal   ☐ abnormal/impaired   ☐ Not known

55   Is a copy of the report attached?

☐ yes   ☐ no

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

☐ yes   ☐ no   ☐ Unknown

57   Date of most recent test prior to HSCT: __ __ __ __ - __ __- __ __   ☐ Unknown

58   Specify test results:

☐ Normal   ☐ abnormal/impaired   ☐ Not known

59   Is a copy of the report attached?

☐ yes   ☐ no

60   Was pulmonary function evaluated at any time prior to HSCT?

☐ yes   ☐ no   ☐ Unknown

61   Date of most recent test prior to HSCT: __ __ __ __ - __ __- __ __   ☐ Unknown

62   Oxygen saturation on room air: _____ %   ☐ Unknown

63   Specify results of pulmonary evaluation:

☐ Normal   ☐ abnormal/impaired   ☐ Not known

64   Is a copy of the report attached?

☐ yes   ☐ no

65   Was an echocardiogram performed at any time prior to HSCT?

☐ yes   ☐ no   ☐ Unknown

66   Date of most recent test prior to HSCT: __ __ __ __ - __ __- __ __   ☐ Unknown

Specify valvular insufficiency:

67   Aortic

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

68   Mitral

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

69   Pulmonary

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

70   Tricuspid

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

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

☐ yes   ☐ no   ☐ Unknown

72   Date of most recent test prior to HSCT: __ __ __ __ - __ __- __ __   ☐ Unknown

73 Ejection fraction: _____ %   ☐ Unknown

74 Shortening fraction: _____ %   ☐ Unknown

First Name: _____   Last Name: _____

Phone number: _____   Fax number: _____

E-mail address: _____