Form 2056 R1.0: Pigmentary Dilution Disorder (PDD) Pre-HCT Data

Center: CRID:

Key Fields

Sequence Number: _____

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

CIBMTR Center Number: _____

CIBMTR Recipient ID: _____

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

HCT Type: (check all that apply)

☐ Autologous

☐ Allogeneic, unrelated

☐ Allogeneic, related

Product type: (check all that apply)

☐ Bone marrow

☐ PBSC

☐ Single cord blood unit

☐ Multiple cord blood units

☐ Other product

Specify: _____

Subsequent Transplant

Is this the report of a second or subsequent transplant for the same disease?

☐ yes ☐ no

Disease Assessment at Diagnosis

Questions: 1 - 7

1 Is this recipient a registered participant in the United States Immunodeficiency Network (USIDNET)?

☐ yes ☐ no

2 USIDNET ID: _____

3 What is the diagnosis?

☐ Chediak-Higashi syndrome

☐ Griscelli syndrome type 2

☐ Hermansky-Pudlak syndrome type 2

☐ Other pigmentary dilution disorder

4 Specify other PDD diagnosis: _____

5 What was the date of diagnosis? __ __ __ __ - __ __- __ __

6 Was genetic testing used to confirm the diagnosis?

☐ yes ☐ no ☐ Unknown

7 Was documentation submitted to the CIBMTR?

☐ yes ☐ no

Assessment of Immunologic Function at Diagnosis

Questions: 8 - 35

8 Cytotoxic T-cell activity

- ☐ Absent (<10% of control)
- ☐ Low (10-30% of control)
- ☐ Normal (>30% of control)
- ☐ Not done

9 Date sample collected

☐ Known ☐ Unknown

10 Date sample collected: __ __ __ __ - __ __- __ __

11 Degranulation of cytolytic lymphocytes (CD107a expression)

- ☐ Absent (<10% of control)
- ☐ Low (10-30% of control)
- ☐ Normal (>30% of control)
- ☐ Not done

12 Date sample collected

☐ Known ☐ Unknown

13 Date sample collected: __ __ __ __ - __ __- __ __

14 Granulocyte chemotaxis

- ☐ Absent (<10% of control)
- ☐ Low (10-30% of control)
- ☐ Normal (>30% of control)
- ☐ Not done

15 Date sample collected

☐ Known ☐ Unknown

16 Date sample collected: __ __ __ __ - __ __- __ __

17 Natural killer cell activity
(against K562 cells)

- ☐ Absent (<10% of control)
- ☐ Low (10-30% of control)
- ☐ Normal (>30% of control)
- ☐ Not done

18 Date sample collected

☐ Known ☐ Unknown

19 Date sample collected: __ __ __ __ - __ __- __ __

20 Phytohemagglutinin (PHA)

- ☐ Absent (<10% of control)
- ☐ Low (10-30% of control)
- ☐ Normal (>30% of control)
- ☐ Not done

21 Date sample collected

☐ Known ☐ Unknown

22 Date sample collected: __ __ __ __ - __ __- __ __

23 Platelet aggregation

- ☐ Absent (<10% of control)
- ☐ Low (10-30% of control)
- ☐ Normal (>30% of control)
- ☐ Not done

24 Date sample collected

☐ Known ☐ Unknown

25 Date sample collected: __ __ __ __ - __ __- __ __

26 Did the recipient receive supplemental intravenous immunoglobulins (IVIG)?

☐ yes ☐ no

27 Was therapy ongoing within three months of immunoglobulin testing?

☐ yes ☐ no

Specify the quantitative immunoglobulins measured at diagnosis:

28 IgG

☐ Known ☐ Unknown

29 _____ ☐ mg/dL ☐ g/dL ☐ g/L

30 IgM

☐ Known ☐ Unknown

31 _____ ☐ mg/dL ☐ g/dL ☐ g/L

32 IgA

☐ Known ☐ Unknown

33 _____ ☐ mg/dL ☐ g/dL ☐ g/L

34 IgE

☐ Known ☐ Unknown

35 _____ IU/mL

Lymphocyte Analysis at Diagnosis

Questions: 36 - 51

36 Were lymphocyte analyses performed?

☐ yes ☐ no

Specify the following lymphocyte analyses performed at diagnosis:

37 Date sample collected: __ __ __ __ - __ __- __ __

Specify the following lymphocyte analyses performed at diagnosis:

38 CD3 (T cells)

☐ Known ☐ Unknown

39 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

40 CD4 (T helper cells)

☐ Known ☐ Unknown

41 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

42 CD8 (cytotoxic T cells)

☐ Known ☐ Unknown

43 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

44 CD20 (B lymphocyte cells)

☐ Known ☐ Unknown

45 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

46 CD56 (natural killer (NK) cells)

☐ Known ☐ Unknown

47 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

48 CD4+ / CD45RA+ (naive T cells)

☐ Known ☐ Unknown

49 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

50 CD4+ / CD45RO+ (memory T cells)

☐ Known ☐ Unknown

51 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

Disease Involvement Between Diagnosis and the Start of the Preparative Regimen(Conditioning) Questions: 52 - 78

Indicate which of the following manifestations of the disease were present between diagnosis and the start of the preparative regimen:

52 Giant leukocyte granules

☐ yes ☐ no ☐ Unknown

53 Neutropenia (ANC < 1.0 x 10^9 /L)

☐ yes ☐ no ☐ Unknown

54 Thrombocytopenia (platelets < 100 x 10^9 /L)

☐ yes ☐ no ☐ Unknown

55 Facial dysmorphisms

(any of the following: epicanthal folds, broad nasal root, posteriorly rotated ears, retrognathia)

☐ yes ☐ no ☐ Unknown

56 Fair hair and/or abnormal accumulation of pigment at microscopic examination of hair

☐ yes ☐ no ☐ Unknown

57 Ocular albinism

☐ yes ☐ no ☐ Unknown

58 Skin albinism / fair skin

☐ yes ☐ no ☐ Unknown

59 Bleeding diathesis

☐ yes ☐ no ☐ Unknown

60 Bleeding from the GI tract

☐ yes ☐ no

61 Easy bruising

☐ yes ☐ no

62 Hematuria

☐ yes ☐ no

63 Oral bleeding

☐ yes ☐ no

64 Recurrent nosebleeds

☐ yes ☐ no

65 Other bleeding

☐ yes ☐ no

66 Specify other bleeding: _____

67 Were there any clinical neurologic abnormalities present?

☐ yes ☐ no ☐ Unknown

Specify neurologic abnormality or abnormalities:

68 Abnormal gait

☐ yes ☐ no

69 Areflexia

☐ yes ☐ no

70 Ataxia and/or other symptoms of cerebellar dysfunction

☐ yes ☐ no

71 Developmental delay

☐ yes ☐ no

72 Was the recipient's IQ tested?

☐ yes ☐ no

73 Date IQ was tested: __ __ __ __ - __ __- __ __

74 IQ test instrument

☐ Kaufman Assessment Battery for Children

☐ Raven's Progressive Matrices

☐ Stanford-Binet

☐ Wechsler Adult Intelligence Scale

☐ Wechsler Intelligence Scale for Children

☐ Woodcock-Johnson Tests of Cognitive Abilities

75 IQ score: _____

76 Seizures

☐ yes ☐ no

77 Other neurologic abnormality

☐ yes ☐ no

78 Specify other neurologic abnormality: _____

Accelerated Phase(AP) Between Diagnosis and the Start of the Preparative Regimen(Conditioning) Questions: 79 - 105

79 Did the recipient develop features of an accelerated phase?

☐ yes ☐ no ☐ Unknown

80 Number of accelerated phases recorded: _____

81 Date first accelerated phase detected

☐ Known ☐ Unknown

82 Date first accelerated phase was detected: __ __ __ __ - __ __- __ __

83 Abnormal CSF
(WBC > 5 cells/µL, elevated protein)
☐ yes ☐ no ☐ Unknown

84 Abnormal liver function
☐ yes ☐ no ☐ Unknown

85 Anemia
(Hgb<10 g/dL)
☐ yes ☐ no ☐ Unknown

86 Cytomegalovirus (CMV)
(associated with accelerated phase)
☐ yes ☐ no ☐ Unknown

87 Epstein-Barr virus (EBV)
(associated with accelerated phase)
☐ yes ☐ no ☐ Unknown

88 Other viral infection associated with accelerated phase
(not CMV or EBV)
☐ yes ☐ no ☐ Unknown

89 Specify other infection: _____

90 Dense bodies (delta granules) on electron micrograph (EM) of platelets
☐ yes ☐ no ☐ Unknown

91 Elevated triglycerides
☐ yes ☐ no ☐ Unknown

92 Fevers
(>38.5° C or > 101.3° F for >7 days)
☐ yes ☐ no ☐ Unknown

93 Hemophagocytosis
☐ yes ☐ no ☐ Unknown

94 Hepatomegaly (liver edge palpable > 3 cm below right costal margin)
☐ yes ☐ no ☐ Unknown

95 Hyperferritinemia
(serum ferritin > 500 ng/mL or 500 µg/dL)
☐ yes ☐ no ☐ Unknown

96 Hypofibrinogenemia
(serum fibrogen <150 mg/dL or <1.5 g/L or <4.4 µmol/L)
☐ yes ☐ no ☐ Unknown

97 Hyponatremia
(serum sodium <135 mg/dL)
☐ yes ☐ no ☐ Unknown

98 Lymphadenopathy

☐ yes ☐ no ☐ Unknown

99 CSF lymphocytosis

☐ yes ☐ no ☐ Unknown

100 Neurologic dysfunction
(e.g. seizures, meningitis signs)

☐ yes ☐ no ☐ Unknown

101 Neutropenia (ANC < 1.0 x 10^9 /L)

☐ yes ☐ no ☐ Unknown

102 Splenomegaly (spleen palpable > 3 cm below left costal margin)

☐ yes ☐ no ☐ Unknown

103 Thrombocytopenia (platelets < 100 x 10^9 /L)

☐ yes ☐ no ☐ Unknown

104 Other feature associated with accelerated phase

☐ yes ☐ no ☐ Unknown

105 Specify other feature: _____

Pre-HCT Therapy

Questions: 106 - 119

106 Was therapy given?

☐ yes ☐ no

Specify therapy given:

107 Acyclovir

☐ yes ☐ no

108 Alemtuzumab (Campath)

☐ yes ☐ no

109 Antithymocyte globulin (ATG)

☐ yes ☐ no

110 Corticosteroids

☐ yes ☐ no

111 Cyclosporine (CSA, Neoral, Sandimmune)

☐ yes ☐ no

112 Etoposide (VP-16, VePesid)

☐ yes ☐ no

113 Ganciclovir (DHPG)

☐ yes ☐ no

114 Intrathecal methotrexate (IT MTX)

☐ yes ☐ no

115 Intravenous immune globulin (IVIG)

☐ yes ☐ no

116 Interferon-α (Intron, Roferon) (includes PEG)

☐ yes ☐ no

117 Rituximab (Rituxan, MabThera)

☐ yes ☐ no

118 Other therapy

☐ yes ☐ no

119 Specify other therapy: _____

Assessment of Immunologic Function at Last Evaluation Prior to the Start of Preparative Regimen

Questions: 120 - 147

120 Cytotoxic T-cell activity

☐ Absent (<10% of control)

☐ Low (10-30% of control)

☐ Normal (>30% of control)

☐ Not done

121 Date sample collected

☐ Known ☐ Unknown

122 Date sample collected: __ __ __ __ - __ __- __ __

123 Degranulation of cytolytic lymphocytes (CD107a expression)

☐ Absent (<10% of control)

☐ Low (10-30% of control)

☐ Normal (>30% of control)

☐ Not done

124 Date sample collected

☐ Known ☐ Unknown

125 Date sample collected: __ __ __ __ - __ __- __ __

126 Granulocyte chemotaxis

☐ Absent (<10% of control)

☐ Low (10-30% of control)

☐ Normal (>30% of control)

☐ Not done

127 Date sample collected

☐ Known ☐ Unknown

128 Date sample collected: __ __ __ __ - __ __- __ __

129 Natural killer cell activity
(against K562 cells)

- ☐ Absent (<10% of control)
- ☐ Low (10-30% of control)
- ☐ Normal (>30% of control)
- ☐ Not done

130 Date sample collected

☐ Known ☐ Unknown

131 Date sample collected: __ __ __ __ - __ __- __ __

132 Phytohemagglutinin (PHA)

- ☐ Absent (<10% of control)
- ☐ Low (10-30% of control)
- ☐ Normal (>30% of control)
- ☐ Not done

133 Date sample collected

☐ Known ☐ Unknown

134 Date sample collected: __ __ __ __ - __ __- __ __

135 Platelet aggregation

- ☐ Absent (<10% of control)
- ☐ Low (10-30% of control)
- ☐ Normal (>30% of control)
- ☐ Not done

136 Date sample collected

☐ Known ☐ Unknown

137 Date sample collected: __ __ __ __ - __ __- __ __

Specify the quantitative immunoglobulins last measured prior to the preparative regimen:

138 Did the recipient receive supplemental intravenous immunoglobulins (IVIG)?

☐ yes ☐ no

139 Was therapy ongoing within three months of immunoglobulin testing?

☐ yes ☐ no

140 IgG

☐ Known ☐ Unknown

141 _____ ☐ mg/dL ☐ g/dL ☐ g/L

142 IgM

☐ Known ☐ Unknown

143 _____ ☐ mg/dL ☐ g/dL ☐ g/L

144 IgA

☐ Known ☐ Unknown

145 _____ ☐ mg/dL ☐ g/dL ☐ g/L

146 IgE

☐ Known ☐ Unknown

147 _____ IU/mL

Lymphocyte Analysis at Last Evaluation Prior to the Start of the Preparative Regimen (Conditioning) Questions: 148 - 163

148 Were lymphocyte analyses performed?

☐ yes ☐ no

149 Date sample collected: __ __ __ __ - __ __ - __ __

150 CD3 (T cells)

☐ Known ☐ Unknown

151 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

152 CD4 (T helper cells)

☐ Known ☐ Unknown

153 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

154 CD8 (cytotoxic T cells)

☐ Known ☐ Unknown

155 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

156 CD20 (B lymphocyte cells)

☐ Known ☐ Unknown

157 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

158 CD56 (natural killer (NK) cells)

☐ Known ☐ Unknown

159 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

160 CD4+ / CD45RA+ (naive T cells)

☐ Known ☐ Unknown

161 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

162 CD4+ / CD45RO+ (memory T cells)

☐ Known ☐ Unknown

163 _____ ☐ x 10^9/L (x 10^3/mm^3)

☐ x 10^6/L

Disease Status at Last Evaluation Prior to the Start of the Preparative Regimen(Conditioning)

Questions: 164 - 165

164 What was the disease status?

☐ No prior accelerated phase

☐ In remission from accelerated phase

☐ In accelerated phase

☐ Unknown

165 Date assessed: __ __ __ __ - __ __- __ __

First Name: _____

Last Name: _____

E-mail address: _____

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