Form 2029 R2.0: Fanconi Anemia/Constitutional Anemia 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: _____

- If this is a report of a second or subsequent transplant, check here.

Continue with question 177.

Disease Assessment at Diagnosis

Questions: 1 - 141

1 What was the date of diagnosis of Fanconi Anemia? __ __ __ __ - __ __- __ __

2 Was the diagnosis made in utero?

yes no

Specify test(s) performed to identify disease:

3 Amniocentesis

yes no

4 Chorionic villous sampling (CVS)

yes no

5 Fibroblasts

yes no

6 Other test

yes no

7 Specify other test: _____

8 Was the recipient diagnosed with any congenital abnormalities?

yes no

9 Abnormal facies (snub nose, thick upper lip, epicanthic folds, hypertelorism)

yes no

10 Ear abnormalities

yes no

11 Eye abnormalities (strabismus, cataract, microphthalmia)

yes no

12 Other neurologic abnormalities

yes no

13 Specify: _____

14 Microcephaly

yes no

15 Palate or jaw abnormalities (cleft palate and/or lip, Pierre Robin syndrome, small jaw or mouth)

yes no

16 Abnormal neck (short or webbed neck)

yes no

17 Cardiac abnormalities

yes no

18 Exocrine pancreatic deficiency

yes no

19 Gastrointestinal abnormalities

yes no

20 Genital abnormalities (cryptorchism, hypoplasia)

yes no

21 Kidney or urinary tract abnormalities

yes no

22 Thumb abnormalities

yes no

23 Radius abnormalities

yes no

24 Other skeletal abnormalities (syndactyly, chinodactyl, abnormal ribs, metaphyseal dyschondroplasia)

yes no

25 Cafe au lait spots

yes no

26 Other skin abnormalities

yes no

27 Specify: _____

28 Other congenital abnormalities

yes no

29 Specify: _____

30 Specify the date that abnormal blood results were first observed __ __ __ __ - __ __- __ __

31 Specify the presenting hematologic disorder

- acute leukemia **-Complete a corresponding Leukemia insert**
- cytopenia
- myelodysplasia **-Complete a Form 2014-MDS insert**
- other disorder

Specify cytopenia:

32 Anemia (Hb <10 g/dL)

yes no

33 Thrombocytopenia (platelets < 100,000/mm^3)

yes no

34 Neutropenia (ANC <1,000/$mm)^3$

yes no

35 Specify other hematologic disorder _____

36 Was the bone marrow examined at diagnosis?

yes no Unknown

37 Specify date of bone marrow examination __ __ __ __ - __ __- __ __

38 Specify cellularity

Decreased Normal Increased Unknown

39 Were dysplastic features present at diagnosis?

yes no Unknown

40 Blasts in marrow _____ **%**

Cytogenetic Studies/Sensitivity to DNA Cross-Linking Agents (1) — Questions: 41 - 141

41 Was bone marrow karyotyping present at diagnosis?

- Yes
- yes, but no evaluable metaphases
- No
- Unknown

42 Was karyotype normal?

yes no

Specify abnormalities identified:

43 +4

yes no

44 +5

yes no

45 -7

yes no

46 +8

yes no

47 +11
yes no

48 +13
yes no

49 +14
yes no

50 -17
yes no

51 -18
yes no

52 -20
yes no

53 +21
yes no

54 +22
yes no

55 -X
yes no

56 -Y
yes no

57 del(5q)
yes no

58 del(7q)
yes no

59 del(9q)
yes no

60 del(11q)
yes no

61 del(20q)
yes no

62 inv(3) or t(3;3)
yes no

63 inv(16) or t(16;16)
yes no

64 t(1;7)
yes no

65 t(5;7)
yes no

66 t(6;9)

yes no

67 t(8;16)

yes no

68 t(8;21)

yes no

69 t(15;17) and variants

yes no

70 balanced abn(11q23)

yes no

71 abn(12p)

yes no

72 loss of 17p

yes no

73 complex (>=3 distinct abnormalities)

yes no

74 increased breaks

yes no

75 other karyotype abnormality

yes no

76 Specify other abnormality _____

77 Date of karyotyping __ __ __ __ - __ __- __ __

78 Is a copy of the cytogenetic report attached?

yes no

79 Was complementation group testing performed at any time prior to the preparative regimen?

yes no

Specify groups identified:

80 FANCA

yes no

81 FANCB

yes no

82 FANCC

yes no

83 FANCD2

yes no

84 FANCE

yes no

85 FANCF

yes no

86 FANCG

yes no

87 Other group

yes no

88 Specify: _____

89 Is a copy of the complementation group report attached?

yes no

90 Were any genetic mutations identified?

yes no

Specify mutation origin:

91 Maternal

yes no

92 Paternal

yes no

93 Was a mutation analysis of cloned Fanconi Anemia genes performed at any time prior to the preparative regimen?

yes no Unknown

94 Specify the date the analysis was performed __ __ __ __ - __ __- __ __

Specify mutation(s) and specify value:

95 Exon

yes no

96 Specify _____

97 Intron

yes no

98 Specify _____

99 Nucleotide change (e.g, 732G>C)

yes no

100 Specify _____

101 Amino acid changes(e.g., L244F)

yes no

102 Specify _____

103 Specify the mutation type

substitution deletion insertion

104 Is a copy of the mutation analysis report attached?

yes no

Maternity Mutation Analysis:

105 Was a mutation analysis of cloned Fanconi Anemia genes performed at any time prior to the preparative regimen?

yes no Unknown

106 Specify the date the analysis was performed: __ __ __ __ - __ __- __ __

Specify mutation(s) and specify value:

107 Exon

yes no

108 Specify: _____

109 Intron:

yes no

110 Specify: _____

111 Nucleotide change (e.g., 732G>C)

yes no

112 Specify: _____

113 Amino acid changes (e.g., L244F)

yes no

114 Specify: _____

115 Specify mutation type:

substitution deletion insertion

116 Is a copy of the mutation analysis report attached?

yes no

Paternity Mutation Analysis:

117 Was a mutation analysis of cloned Fanconi Anemia genes performed at any time prior to the preparative regimen?

yes no Unknown

118 Specify the date the analysis was performed __ __ __ __ - __ __- __ __

119 Exon

yes no

120 Specify _____

121 Intron

yes no

122 Specify _____

123 Nucleotide change
(e.g, 732G>C)

yes no

124 Specify _____

125 Amino acid changes
(e.g., L244F)

yes no

126 Specify _____

127 Specify the mutation type

substitution deletion insertion

128 Is a copy of the mutation analysis report attached?

yes no

129 Were the recipient's bone marrow cells or peripheral blood mononuclear cells tested for sensitivity to cross-linking agents?

yes no Unknown

130 Specify the date the testing was performed __ __ __ __ - __ __- __ __

131 Specify the type of cross-linking agent used

diepoxybutane mitomycin C other agent

132 Specify other agent _____

133 Were chromatid aberrations present on an unstressed preparation?

Yes No Not evaluable

134 Total number of cells studied _____

135 Number of aberrations per cell _____

136 Number of cells with no aberrations _____

137 Were chromatid aberrations present on a stressed preparation?

Yes No Not evaluable

138 Total number of cells studied _____

139 Number of aberrations per cell _____

140 Number of cells with no aberrations _____

141 Is a copy of the report attached?

yes no

Familial History of Disease

Questions: 142 - 176

142 Were any other genetically related family members affected?

yes no Unknown

Specify family member(s):

143 Sibling

yes no

144 Cousin

yes no

145 Parent

yes no

146 Aunt / uncle

yes no

147 Other relative

yes no

148 Specify relationship _____

149 Is the recipient genetically related to his/her parents?

yes, both mother and father

yes, mother only

yes, father only

no, not genetically related to mother or father

Unknown

150 (Related donors only) Were the donor's blood or bone marrow cells tested for sensitivity to cross-linking agents?

yes no Unknown

151 Specify the date testing was performed __ __ __ __ - __ __- __ __

152 Specify the type of cross-linking agent used

diepoxybutane mitomycin C other agent

153 Specify other agent _____

154 Were chromatid aberrations present on an unstressed preparation?

Yes No Not evaluable

155 Total number of cells studied _____

156 Number of aberrations per cell _____

157 Number of cells with no aberrations _____

158 Were chromatid aberrations present on a stressed preparation?

Yes No Not evaluable

159 Total number of cells studied _____

160 Number of aberrations per cell _____

161 Number of cells with no aberrations _____

162 Is a copy of the report attached?

yes no

163 Was the recipient treated with androgens prior to HSCT?

yes no Unknown

164 Was the recipient treated with corticosteroids prior to HSCT?

yes no Unknown

165 Did the recipient receive growth factors prior to HSCT?

yes no Unknown

If yes, specify cytokine(s) given:

166 Erythropoietin (includes all forms of erythropoietin/darbepoetin)

yes no

167 G-CSF

yes no

168 GM-CSF

yes no

169 IL3

yes no

170 Neulasta

yes no

171 Stem cell factor

yes no

172 Other growth factor

yes no

173 Specify other agent _____

174 Did the recipient receive red blood cell transfusions between diagnosis and the start of the preparative regimen?

yes no

175 Specify the total number of donor exposures (best estimate)

1-5 6-10 11-20 21-30 31-40 41-50 >=51 Unknown

176 Did the recipient receive platelet transfusions in the four weeks prior to the preparative regimen?

yes no

Clinical Features Just Prior to the Preparative Regimen

Questions: 177 - 177

177 What was the recipient's disease status immediately prior to the preparative regimen?

- stable cytopenia, co cytogenetic abnormalities (no MDS)
- stable cytopenia with cytogenetic abnormalities (no MDS)
- progressive cytopenia
- myelodysplasia **-Complete a Form 2014-MDS insert**
- leukemia, untreated **-Complete a corresponding Leukemia insert**
- leukemia, treated **-Complete a corresponding Leukemia insert**

Hematologic Parameters Immediately Prior to the Preparative Regimen

Questions: 178 - 215

178 Was the recipient's bone marrow examined at any time between diagnosis and the preparative regimen?

- Yes
- yes, but no evaluable metaphases
- No
- Unknown

179 Were any karyotype abnormalities identified?

yes no

Specify abnormalities identified:

180 +4

yes no

181 +5

yes no

182 -7

yes no

183 +8

yes no

184 +11

yes no

185 +13

yes no

186 +14

yes no

187 -17

yes no

188 -18

yes no

189 -20

yes no

190 +21

yes no

191 +22

yes no

192 -X

yes no

193 -Y

yes no

194 del(5q)

yes no

195 del(7q)

yes no

196 del(9q)

yes no

197 del(11q)

yes no

198 del(20q)

yes no

199 inv(3) or t(3;3)

yes no

200 inv(16) or t(16;16)

yes no

201 t(1;7)

yes no

202 t(5;7)

yes no

203 t(6;9)

yes no

204 t(8;16)

yes no

205 t(8;21)

yes no

206 t(15;17) and variants

yes no

207 balanced abn(11q23)

yes no

208 abn(12p)

yes no

209 loss of 17p

yes no

210 complex (>=3 distinct abnormalities)

yes no

211 increased breaks

yes no

212 other karyotype abnormality

yes no

213 Specify other abnormality _____

214 Date of karyotyping __ __ __ __ - __ __- __ __

215 Is a copy of the cytogenetic report attached?

yes no

First Name: _____ Last Name: _____

Phone number: _____ Fax number: _____

E-mail address: _____