CIBMTR®
CENTER FOR INTERNATIONAL BLOOD & MARROW TRANSPLANT RESEARCH

Fanconi Anemia / Constitutional Anemia Pre-HSCT Data

Registry Use Only

Sequence Number:

Date Received:

CIBMTR Center Number:

CIBMTR Recipient ID:

Today's Date: Month Day Year (20__)

Date of HSCT for which this form is being completed: Month Day Year (20__)

HSCT type: ☐ autologous ☐ allogeneic, unrelated ☐ allogeneic, related ☐ syngeneic (identical twin)

Product type: ☐ marrow ☐ PBSC ☐ cord blood ☐ other product, specify: _____

This form must be accompanied by Form 2000 – Recipient Baseline Data. All information in the box above, including the date, should be identical with the corresponding Form 2000. Information should come from an actual examination by the Transplant Center physician, or the physician who is following the recipient pre-HSCT, or abstraction of the recipient's medical records.

If this is a report of a second or subsequent transplant, check here ☐ and continue with question 153.

1. What was the date of diagnosis of Fanconi Anemia? Month Day Year

2. Was the diagnosis made in utero?
   1 ☐ yes → Specify test(s) performed to identify disease:
   2 ☐ no

3. 1 ☐ yes 2 ☐ no Amniocentesis
4. 1 ☐ yes 2 ☐ no Chorionic villous sampling (CVS)
5. 1 ☐ yes 2 ☐ no Fibroblasts
6. 1 ☐ yes 2 ☐ no Other test → 7. If yes, specify other test: _____

8. Was the recipient diagnosed with any congenital abnormalities?
   1 ☐ yes → Specify test(s) performed to identify disease:
   2 ☐ no

9. 1 ☐ yes 2 ☐ no Abnormal facies (snub nose, thick upper lip, epicanthic folds, hypertelorism)
10. 1 ☐ yes 2 ☐ no Ear abnormalities
11. 1 ☐ yes 2 ☐ no Eye abnormalities (strabismus, cataract, micropthalmia)
12. 1 ☐ yes 2 ☐ no Other neurologic abnormalities → 13. If yes, specify: _____
14. 1 ☐ yes 2 ☐ no Microcephaly
15. 1 ☐ yes 2 ☐ no Palate or jaw abnormalities (cleft palate and/or lip, Pierre Robin syndrome, small jaw or mouth)
16. 1 ☐ yes 2 ☐ no Abnormal neck (short or webbed neck)
17. 1 ☐ yes 2 ☐ no Cardiac abnormalities
18. 1 ☐ yes 2 ☐ no Exocrine pancreatic deficiency
19. 1 ☐ yes 2 ☐ no Gastrointestinal abnormalities
20. 1 ☐ yes 2 ☐ no Genital abnormalities (cryptorchism, hypoplasia)
21. 1 ☐ yes 2 ☐ no Kidney or urinary tract abnormalities
22. 1 ☐ yes 2 ☐ no Thumb abnormalities
23. 1 ☐ yes 2 ☐ no Radius abnormalities

For internal use only: Document F00530 version 1.0 Replaces: n/a

Mail this form to your designated campus (Milwaukee or Minneapolis). Retain the original at the transplant center.

CIBMTR Center Number: ☐☐☐☐☐ CIBMTR Recipient ID: ☐☐☐☐☐☐☐☐☐☐

24. 1 ☐ yes 2 ☐ no Other skeletal abnormalities (syndactyly, clinodactyly, abnormal ribs, metaphyseal dyschondroplasia)
25. 1 ☐ yes 2 ☐ no Cafe au lait spots
26. 1 ☐ yes 2 ☐ no Other skin abnormalities → 27. If yes, specify: _____
28. 1 ☐ yes 2 ☐ no Other congenital abnormalities → 29. If yes, specify: _____

30. Specify the date that abnormal blood results were first observed: ☐☐ Month ☐☐ Day ☐☐☐☐ Year

31. Specify the presenting hematologic disorder:

1 ☐ acute leukemia → **Complete a corresponding Leukemia insert**

2 ☐ cytopenia →
Specify cytopenia:
32. 1 ☐ yes 2 ☐ no Anemia (Hb < 10 g/dL)
33. 1 ☐ yes 2 ☐ no Thrombocytopenia (platelets < 100,000/mm^3)
34. 1 ☐ yes 2 ☐ no neutropenia (ANC < 1,000.mm^3)

3 ☐ myelodysplasia → **Complete a Form 2014 – MDS insert**

4 ☐ other disorder → 35. Specify other hematologic disorder: _____

36. Was the bone marrow examined at diagnosis?

1 ☐ yes →
37. Specify date of bone marrow examination: ☐☐ Month ☐☐ Day ☐☐☐☐ Year
38. Specify cellularity:
1 ☐ decreased
2 ☐ normal
3 ☐ increased
4 ☐ unknown
39. Were dysplastic features present at diagnosis?
1 ☐ yes
2 ☐ no
3 ☐ unknown
40. Blasts in marrow: ☐☐☐ %

2 ☐ no
3 ☐ unknown

Cytogenetic Studies / Sensitivity to DNA Cross-Linking Agents

This section requests information about bone marrow karyotype and examination of bone marrow and blood cells for non-specific chromatid abnormalities. If more than one investigation was carried out, please copy this section and report each event separately.

41. Was bone marrow karyotyping performed at diagnosis?

1 ☐ yes →
42. Was karyotype normal?
1 ☐ yes
2 ☐ no →
Specify abnormalities identified:
43. 1 ☐ yes 2 ☐ no +4
44. 1 ☐ yes 2 ☐ no +5
45. 1 ☐ yes 2 ☐ no –7
46. 1 ☐ yes 2 ☐ no +8
47. 1 ☐ yes 2 ☐ no +11

2 ☐ yes, but no evaluable metaphases
3 ☐ no
4 ☐ unknown

CIBMTR Center Number: | | | | | | CIBMTR Recipient ID: | | | | | | | | | | |

48.	1 ☐ yes 2 ☐ no	+13
49.	1 ☐ yes 2 ☐ no	+14
50.	1 ☐ yes 2 ☐ no	–17
51.	1 ☐ yes 2 ☐ no	–18
52.	1 ☐ yes 2 ☐ no	–20
53.	1 ☐ yes 2 ☐ no	+21
54.	1 ☐ yes 2 ☐ no	+22
55.	1 ☐ yes 2 ☐ no	–X
56.	1 ☐ yes 2 ☐ no	–Y
57.	1 ☐ yes 2 ☐ no	del(5q)
58.	1 ☐ yes 2 ☐ no	del(7q)
59.	1 ☐ yes 2 ☐ no	del(9q)
60.	1 ☐ yes 2 ☐ no	del(11q)
61.	1 ☐ yes 2 ☐ no	del(20q)
62.	1 ☐ yes 2 ☐ no	inv(3) or t(3;3)
63.	1 ☐ yes 2 ☐ no	inv(16) or t(16;16)
64.	1 ☐ yes 2 ☐ no	t(1;7)
65.	1 ☐ yes 2 ☐ no	t(5;7)
66.	1 ☐ yes 2 ☐ no	t(6;9)
67.	1 ☐ yes 2 ☐ no	t(8;16)
68.	1 ☐ yes 2 ☐ no	t(8;21)
69.	1 ☐ yes 2 ☐ no	t(15;17) and variants
70.	1 ☐ yes 2 ☐ no	balanced abn(11q23)
71.	1 ☐ yes 2 ☐ no	abn(12p)
72.	1 ☐ yes 2 ☐ no	loss of 17p
73.	1 ☐ yes 2 ☐ no	complex (≥ 3 distinct abnormalities)
74.	1 ☐ yes 2 ☐ no	increased breaks
75.	1 ☐ yes 2 ☐ no	other karyotype abnormality

76. Specify other abnormality: _____

77. Date of karyotyping: | | | | | | | | |
Month Day Year

78. Is a copy of the cytogenetic report attached?
1 ☐ yes
2 ☐ no

CIBMTR Center Number: | | | | | | CIBMTR Recipient ID: | | | | | | | | | | |

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

1 ☐ yes → Specify groups identified:
2 ☐ no

80. 1 ☐ yes 2 ☐ no FANCA
81. 1 ☐ yes 2 ☐ no FANCB
82. 1 ☐ yes 2 ☐ no FANCC
83. 1 ☐ yes 2 ☐ no FANCD2
84. 1 ☐ yes 2 ☐ no FANCE
85. 1 ☐ yes 2 ☐ no FANCF
86. 1 ☐ yes 2 ☐ no FANCG
87. 1 ☐ yes 2 ☐ no Other group → 88. If yes, specify: _____

89. Is a copy of the complementation group report attached?
1 ☐ yes
2 ☐ no

90. Were any genetic mutations identified?

1 ☐ yes → Specify mutation origin:
2 ☐ no

91. 1 ☐ yes 2 ☐ no maternal → **Copy questions 93–104 and complete for mother**
92. 1 ☐ yes 2 ☐ no paternal → **Copy questions 93–104 and complete for father**

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

1 ☐ yes →
2 ☐ no
3 ☐ unknown

94. Specify the date the analysis was performed: | | Month | | | Day | | | | Year

Specify mutation(s): Specify value:

95. 1 ☐ yes 2 ☐ no Exon → 96. Specify: | |
97. 1 ☐ yes 2 ☐ no Intron → 98. Specify: | |
99. 1 ☐ yes 2 ☐ no Nucleotide change (e.g., 732G>C) → 100. Specify: | | | | | | | | | | |
101. 1 ☐ yes 2 ☐ no Amino acid changes (e.g., L244F) → 102. Specify: | | | | | | | | | | |

103. Specify the mutation type:
1 ☐ substitution
2 ☐ deletion
3 ☐ insertion

104. Is a copy of the mutation analysis report attached?
1 ☐ yes
2 ☐ no

For internal use only: Document F00530 version 1.0 Replaces: n/a

CIBMTR Center Number: | | | | | | CIBMTR Recipient ID: | | | | | | | | | | |

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

1 ☐ yes →
2 ☐ no
3 ☐ unknown

106. Specify the date the testing was performed: | | Month | | | Day | | | | Year

107. Specify the type of cross-linking agent used:

1 ☐ diepoxybutane (DEB)
2 ☐ mitomycin C
3 ☐ other agent → 108. Specify other agent: _____

109. Were chromatid aberrations present on an *unstressed* preparation?

1 ☐ yes →
2 ☐ no
3 ☐ not evaluable

110. Total number of cells studied: | |

111. Number of aberrations per cell: | | |

112. Number of cells with no aberrations: | |

113. Were chromatid aberrations present on a *stressed* preparation?

1 ☐ yes →
2 ☐ no
3 ☐ not evaluable

114. Total number of cells studied: | |

115. Number of aberrations per cell: | | |

116. Number of cells with no aberrations: | |

117. Is a copy of the report attached?

1 ☐ yes
2 ☐ no

Familial History of Disease

118. Were any other genetically related family members affected?

1 ☐ yes →
2 ☐ no
3 ☐ unknown

Specify family member(s):

119. 1 ☐ yes 2 ☐ no Sibling
120. 1 ☐ yes 2 ☐ no Cousin
121. 1 ☐ yes 2 ☐ no Parent
122. 1 ☐ yes 2 ☐ no Aunt / uncle
123. 1 ☐ yes 2 ☐ no Other relative → 124. Specify relationship: _____

125. Is the recipient genetically related to his / her parents?

1 ☐ yes, both mother and father
2 ☐ yes, mother only
3 ☐ yes, father only
4 ☐ no, not genetically related to mother or father
5 ☐ unknown

CIBMTR Center Number: | | | | | | CIBMTR Recipient ID: | | | | | | | | | | |

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

1 ☐ yes →
2 ☐ no
3 ☐ unknown

127. Specify the date testing was performed: | | Month | | | Day | | | | Year

128. Specify the type of cross-linking agent used:

1 ☐ diepoxybutane (DEB)
2 ☐ mitomycin C
3 ☐ other agent → 129. Specify other agent: _____

130. Were chromatid aberrations present on an *unstressed* preparation?

1 ☐ yes →
2 ☐ no
3 ☐ not evaluable

131. Total number of cells studied: | |

132. Number of aberrations per cell: | | |

133. Number of cells with no aberrations: | |

134. Were chromatid aberrations present on a stressed preparation?

1 ☐ yes →
2 ☐ no
3 ☐ not evaluable

135. Total number of cells studied: | |

136. Number of aberrations per cell: | | |

137. Number of cells with no aberrations: | |

138. Is a copy of the report attached?

1 ☐ yes
2 ☐ no

139. Was the recipient treated with androgens prior to the HSCT?

1 ☐ yes
2 ☐ no
3 ☐ unknown

140. Was the recipient treated with corticosteroids prior to the HSCT?

1 ☐ yes
2 ☐ no
3 ☐ unknown

141. Did the recipient receive growth factors prior to the HSCT?

1 ☐ yes →
2 ☐ no
3 ☐ unknown

If yes, specify cytokine(s) given:

142. 1 ☐ yes 2 ☐ no Erythropoietin (includes all forms of erythropoietin / darbepoetin)
143. 1 ☐ yes 2 ☐ no G-CSF
144. 1 ☐ yes 2 ☐ no GM-CSF
145. 1 ☐ yes 2 ☐ no IL3
146. 1 ☐ yes 2 ☐ no Neulasta
147. 1 ☐ yes 2 ☐ no Stem cell factor
148. 1 ☐ yes 2 ☐ no Other growth factor → 149. Specify other agent: _____

CIBMTR Center Number: | | | | | | CIBMTR Recipient ID: | | | | | | | | | | |

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

1 ☐ yes → 151. Specify the total number of donor exposures (best estimate):
- 1 ☐ 1–5
- 2 ☐ 6–10
- 3 ☐ 11–20
- 4 ☐ 21–30
- 5 ☐ 31–40
- 6 ☐ 41–50
- 7 ☐ ≥ 51
- 8 ☐ unknown

2 ☐ no

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

1 ☐ yes
2 ☐ no

Clinical Features Just Prior to the Preparative Regimen

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

1 ☐ stable cytopenia, no cytogenetic abnormalities (no MDS)
2 ☐ stable cytopenia with cytogenetic abnormalities (no MDS)
3 ☐ progressive cytopenia
4 ☐ myelodysplasia → **Complete a Form 2014 – MDS insert**
5 ☐ leukemia, untreated → **Complete a corresponding Leukemia insert**
6 ☐ leukemia, treated → **Complete a corresponding Leukemia insert**

Hematologic Parameters Immediately Prior to the Preparative Regimen

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

1 ☐ yes → 155. Were any karyotype abnormalities identified?
- 1 ☐ yes → Specify abnormalities identified:

156.	1 ☐ yes	2 ☐ no	+4
157.	1 ☐ yes	2 ☐ no	+5
158.	1 ☐ yes	2 ☐ no	–7
159.	1 ☐ yes	2 ☐ no	+8
160.	1 ☐ yes	2 ☐ no	+11
161.	1 ☐ yes	2 ☐ no	+13
162.	1 ☐ yes	2 ☐ no	+14
163.	1 ☐ yes	2 ☐ no	–17
164.	1 ☐ yes	2 ☐ no	–18
165.	1 ☐ yes	2 ☐ no	–20
166.	1 ☐ yes	2 ☐ no	+21
167.	1 ☐ yes	2 ☐ no	+22
168.	1 ☐ yes	2 ☐ no	–X
169.	1 ☐ yes	2 ☐ no	–Y
170.	1 ☐ yes	2 ☐ no	del(5q)
171.	1 ☐ yes	2 ☐ no	del(7q)
172.	1 ☐ yes	2 ☐ no	del(9q)
173.	1 ☐ yes	2 ☐ no	del(11q)
174.	1 ☐ yes	2 ☐ no	del(20q)
175.	1 ☐ yes	2 ☐ no	inv(3) or t(3;3)
176.	1 ☐ yes	2 ☐ no	inv(16) or t(16;16)

- 2 ☐ no
- 3 ☐ yes, but no evaluable metaphases
- 4 ☐ no
- 5 ☐ unknown

CIBMTR Center Number: | | | | | | CIBMTR Recipient ID: | | | | | | | | | | |

177. 1 ☐ yes 2 ☐ no t(1;7)
178. 1 ☐ yes 2 ☐ no t(5;7)
179. 1 ☐ yes 2 ☐ no t(6;9)
180. 1 ☐ yes 2 ☐ no t(8;16)
181. 1 ☐ yes 2 ☐ no t(8;21)
182. 1 ☐ yes 2 ☐ no t(15;17) and variants
183. 1 ☐ yes 2 ☐ no balanced abn(11q23)
184. 1 ☐ yes 2 ☐ no abn(12p)
185. 1 ☐ yes 2 ☐ no loss of 17p
186. 1 ☐ yes 2 ☐ no complex (≥ 3 distinct abnormalities)
187. 1 ☐ yes 2 ☐ no increased breaks
188. 1 ☐ yes 2 ☐ no other karyotype abnormality
189. Specify other abnormality: _____

190. Date of karyotyping: | | Month | | | Day | | | | Year

191. Is a copy of the cytogenetic report attached?
1 ☐ yes
2 ☐ no

192. Signed: _____
Person completing form

Please print name: _____

Phone: (_____) _____

Fax: (_____) _____

E-mail address: _____