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
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<tr>
<th></th>
<th>Question</th>
<th>Yes</th>
<th>No</th>
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<tbody>
<tr>
<td>11</td>
<td>Eye abnormalities (strabismus, cataract, microphthalmia)</td>
<td>yes</td>
<td>no</td>
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<tr>
<td>12</td>
<td>Other neurologic abnormalities</td>
<td>yes</td>
<td>no</td>
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<td>13</td>
<td>Specify: __________________________________________________________________</td>
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<td>14</td>
<td>Microcephaly</td>
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<td>Palate or jaw abnormalities (cleft palate and/or lip, Pierre Robin syndrome, small jaw or mouth)</td>
<td>yes</td>
<td>no</td>
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<td>16</td>
<td>Abnormal neck (short or webbed neck)</td>
<td>yes</td>
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<td>17</td>
<td>Cardiac abnormalities</td>
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<td>no</td>
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<td>18</td>
<td>Exocrine pancreatic deficiency</td>
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<td>no</td>
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<td>19</td>
<td>Gastrointestinal abnormalities</td>
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<td>no</td>
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<td>20</td>
<td>Genital abnormalities (cryptorchism, hypoplasia)</td>
<td>yes</td>
<td>no</td>
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<td>21</td>
<td>Kidney or urinary tract abnormalities</td>
<td>yes</td>
<td>no</td>
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<td>22</td>
<td>Thumb abnormalities</td>
<td>yes</td>
<td>no</td>
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<td>23</td>
<td>Radius abnormalities</td>
<td>yes</td>
<td>no</td>
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<tr>
<td>24</td>
<td>Other skeletal abnormalities (syndactyly, clinodactyly, abnormal ribs, metaphyseal dyschondroplasia)</td>
<td>yes</td>
<td>no</td>
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<td>25</td>
<td>Cafe au lait spots</td>
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<td>26</td>
<td>Other skin abnormalities</td>
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<td>27</td>
<td>Specify: __________________________________________________________________</td>
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<td>28</td>
<td>Other congenital abnormalities</td>
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<td>29</td>
<td>Specify: __________________________________________________________________</td>
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<tr>
<td>30</td>
<td>Specify the date that abnormal blood results were first observed __ __ __ __ - __ __- __ __</td>
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</table>
### Specify the presenting hematologic disorder

- **Acute leukemia** - Complete a corresponding Leukemia insert
- **Cytopenia**
- **Myelodysplasia** - Complete a Form 2014-MDS insert
- **Other disorder**

### Specify cytopenia:

1. **Anemia** (Hb < 10 g/dL)
   - Yes
   - No

2. **Thrombocytopenia** (platelets < 100,000/mm³)
   - Yes
   - No

3. **Neutropenia** (ANC < 1,000/mm³)
   - Yes
   - No

4. **Specify other hematologic disorder**

5. **Specify date of bone marrow examination** __ __ __ __ - __ __

6. **Specify cellularity**
   - Decreased
   - Normal
   - Increased
   - Unknown

7. **Were dysplastic features present at diagnosis?**
   - Yes
   - No
   - Unknown

8. **Blasts in marrow** __ __ __ __ __ __ __ __ __

### Cytogenetic Studies/Sensitivity to DNA Cross-Linking Agents (1)

- **Was bone marrow karyotyping present at diagnosis?**
  - Yes
  - No
  - Unknown

- **Was karyotype normal?**
  - Yes
  - No

- **Specify abnormalities identified:**
  - +4
    - Yes
    - No
  - +5
    - Yes
    - No
  - -7
    - Yes
    - No
  - +8
    - Yes
    - No
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<td>73</td>
<td>complex (&gt;=3 distinct abnormalities)</td>
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<td>75</td>
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<td>76</td>
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<td>78</td>
<td>Is a copy of the cytogenetic report attached?</td>
<td>yes</td>
<td>no</td>
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<td>79</td>
<td>Was complementation group testing performed at any time prior to the preparative regimen?</td>
<td>yes</td>
<td>no</td>
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Specify groups identified:

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<td>81</td>
<td>FANCB</td>
<td>yes</td>
<td>no</td>
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<td>82</td>
<td>FANCC</td>
<td>yes</td>
<td>no</td>
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<td>83</td>
<td>FANCD2</td>
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<td>no</td>
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<td>84</td>
<td>FANCE</td>
<td>yes</td>
<td>no</td>
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<tr>
<td>85</td>
<td>FANCF</td>
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<td>no</td>
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</table>
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
Specify: ____________________________

Intron:  
yes  no

Specify: ____________________________

Nucleotide change (e.g., 732G>C)  
yes  no

Specify: ____________________________

Amino acid changes (e.g., L244F)  
yes  no

Specify mutation type:  
substitution  deletion  insertion

Is a copy of the mutation analysis report attached?  
yes  no

Paternity Mutation Analysis:
Was a mutation analysis of cloned Fanconi Anemia genes performed at any time prior to the preparative regimen?  
yes  no  Unknown

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

Exon  
yes  no

Specify: ____________________________

Intron  
yes  no

Specify: ____________________________

Nucleotide change (e.g., 732G>C)  
yes  no

Specify: ____________________________

Amino acid changes (e.g., L244F)  
yes  no

Specify the type of cross-linking agent used:  
diepoxybutane  mitomycin C  other agent

Specify other agent: ____________________________

Were the recipient's bone marrow cells or peripheral blood mononuclear cells tested for sensitivity to cross-linking agents?  
yes  no  Unknown

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

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 ___________________
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<th>Question</th>
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<th>No</th>
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<td>Were chromatid aberrations present on an unstressed preparation?</td>
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<tr>
<td>Total number of cells studied</td>
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<td>Number of aberrations per cell</td>
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<td>Number of cells with no aberrations</td>
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<tr>
<td>Were chromatid aberrations present on a stressed preparation?</td>
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<tr>
<td>Total number of cells studied</td>
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<tr>
<td>Number of aberrations per cell</td>
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<td></td>
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<tr>
<td>Number of cells with no aberrations</td>
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<tr>
<td>Is a copy of the report attached?</td>
<td></td>
<td></td>
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</tr>
<tr>
<td>Was the recipient treated with androgens prior to HSCT?</td>
<td></td>
<td></td>
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<tr>
<td>Was the recipient treated with corticosteroids prior to HSCT?</td>
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<tr>
<td>Did the recipient receive growth factors prior to HSCT?</td>
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<tr>
<td>If yes, specify cytokine(s) given:</td>
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<td>Erythropoietin (includes all forms of erythropoietin/darbepoetin)</td>
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<td>Other growth factor</td>
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<tr>
<td>Specify other agent</td>
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<tr>
<td>Did the recipient receive red blood cell transfusions between diagnosis and the start of the preparative regimen?</td>
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<tr>
<td>Specify the total number of donor exposures (best estimate)</td>
<td>1-5</td>
<td>6-10</td>
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<tr>
<td>Did the recipient receive platelet transfusions in the four weeks prior to the preparative regimen?</td>
<td></td>
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</table>
### Clinical Features Just Prior to the Preparative Regimen

**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

**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
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<td>complex (&gt;=3 distinct abnormalities)</td>
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<td>211</td>
<td>increased breaks</td>
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<tr>
<td>212</td>
<td>other karyotype abnormality</td>
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<td>Specify other abnormality</td>
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<td>Date of karyotyping</td>
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<td>215</td>
<td>Is a copy of the cytogenetic report attached?</td>
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