

**Table 5. Summary of clinical features in patients with DBA syndrome**

	<b>INTRINSIC (DISEASE RELATED)</b>	<b>THERAPY RELATED</b>
<b>Hematologic</b>	Macrocytic anemia with reticulocytopenia Leukopenia, neutropenia Thrombocytopenia Thrombocytosis (infants) Hypocellular marrow (mostly in adults, sometimes in children)	Alloimmunization with subsequent increased transfusion requirement Agranulocytosis (DFP)
<b>Immunologic</b>	Lymphocytopenia Decreased B-cell numbers Hypogammaglobulinemia Recurrent infections	Port catheter infections Transfusion-related pathogens Viral infections (steroids) Lymphocytopenia (steroids) Infections: campylobacter and other bacteria (associated with iron overload), yersiniosis, mucor mycosis (exacerbated by DFX)
<b>Endocrinologic</b>	Intrauterine growth restriction Failure to thrive Short stature Congenital abnormalities	>50% patients experience endocrine problems related to steroids and iron overload: <ul style="list-style-type: none"> <li>• Adrenal insufficiency</li> <li>• Sex hormone insufficiency</li> <li>• Growth hormone dysfunction</li> <li>• Hypogonadism</li> <li>• Thyroid and parathyroid problems</li> <li>• Pancreatic insufficiency, diabetes mellitus</li> </ul>
<b>Orthopedic</b>	Short stature Congenital abnormalities Osteosarcoma (osteogenic sarcoma)	Osteopenia and bone fractures (steroids)
<b>Hepatic, Urogenital, Cardiac</b>	Congenital abnormalities	Liver toxicity (DFX) Liver cirrhosis (iron) Cardiomyopathy with arrhythmias (iron) Nephrotoxicity, phosphate loss (DFX) Urinary stones (DFX, DFO)
<b>Gastrointestinal</b>	Colitis	Diarrhea, esophagitis, nausea (DFX)
<b>Otolaryngologic, ophthalmologic</b>	Congenital abnormalities	Cataract (steroids, DFX) Ototoxicity (DFO)
<b>Neurologic and psychosocial</b>	Developmental delay	High psychosocial burden from chronic illness
<b>Oncologic</b>	Cancer risk: MDS, osteosarcoma, colorectal cancer, and other cancers	Increased risk after HSCT
<b>Obstetric</b>	High risk pregnancies	Infertility (iron overload, HSCT)

Abbreviations: DFO, deferoxamine; DFP, deferiprone; DFX, deferasirox; HSCT, hematopoietic stem cell transplantation.