Table 2. Recommended diagnostic tests in patients with suspected DBA syndrome

Essential diagnostic tests	• CBC (including differential, red cell indices and reticulocyte count) • eADA and HbF [1] • BM morphology/cellularity at initial manifestation or prior to starting steroids • Parvovirus B19 PCR and/or serology in BM or blood • DBA genetic testing (**Table 3**) • Evaluation for congenital abnormalities: physical examination, echocardiography, abdominal ultrasound; additional imaging as indicated[2]
Additional baseline evaluations (all patients)	• Laboratory parameters: ferritin, LDH, bilirubin, transaminases, creatinine, vitamin B12/MMA, folate • DAT (direct Coombs test), blood group antigens, RBC antibodies to guide transfusion management • Immunoglobulin levels (>6 months of age) and lymphocyte immunophenotyping • HLA typing of patient and family members[3]
Further tests in selected patients	• BM cytogenetics and BM biopsy[4] • Suspicion of IBMFS: chromosome breakage (Fanconi anemia), telomere length (dyskeratosis congenita), fecal elastase (Shwachman Diamond syndrome), mitochondrial DNA genetics (Pearson syndrome), ADA2 genetics or enzyme activity (ADA2 deficiency), genetics for other IBMFS[5] • Erythropoietin (EPO) level[6]

[1] Prior to first transfusion or ≥6 weeks (or as far away as possible) after last transfusion, HbF reliably assessed in patients >6 months of age.
[2] Neuro-imaging, hand x-ray and other imaging studies as clinically indicated.
[3] Not required for diagnosis, but essential for long-term therapeutic planning.
[4] In patients with suspicion of MDS or leukemia.
[5] In patients with clinical suspicion of respective syndromes.
[6] In suspected renal dysfunction; note, EPO levels are elevated in patients with DBA syndrome.

Abbreviations: CBC, complete blood counts; ADA, erythrocyte adenosine deaminase; HbF, fetal hemoglobin; BM; bone marrow; MMA, methylmalonic acid; DAT (direct antiglobulin test); IBMFS, inherited BM failure syndromes.