Diagnosis and Treatment Spectrum for Diamond-Blackfan Anemia: A Single Center Experience

Erin Goode[1], Paulo Pena[2], Ashnaa Rao[3], and Sanjay Shah[1]

[1]Phoenix Children's Hospital Center for Cancer and Blood Disorders
[2]The University of Arizona College of Medicine Phoenix
[3]Phoenix Children's Hospital General Pediatrics

August 15, 2022

Abstract

Diamond-Blackfan Anemia (DBA) is a rare congenital bone marrow failure syndrome typically diagnosed during infancy and is characterized by macrocytic anemia, congenital malformations, and predisposition to cancer. There were 32 patients treated for DBA at a single tertiary care referral center from 2000-2022. A retrospective chart review characterized patient presentation at diagnosis, the clinical course of each patient, and the various treatment strategies with correlation to patient outcome. Our study emphasizes the importance of screening for DBA in patients who present with macrocytic anemia regardless of age at presentation as a substantial proportion of patients presented after infancy.

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Characteristics	N=32
Sex	
Female	19 (59.4)
Male	13 (40.6)
Age at Diagnosis	
<1 year	21 (65.6)
1-5 years	6 (18.8)
6-10 years	2 (6.3)
>10 years	3 (9.4)
Family History	
Yes	6 (18.8)
No	26 (81.3)
Hemoglobin at Diagnosis*	
<5 gm/dL	14 (60.9)
6-10 gm/dL	5 (21.7)
>10 gm/dL	4 (17.4)
Reticulocyte Count at Diagnosis!	
<1%	7 (31.8)
1-3%	11 (50.0)
>3%	4 (18.2)
Positive Genetic Testing#	
Yes	16 (72.7)
No	6 (27.3)
Illness at Diagnosis	
Yes	6 (18.8)
No	26 (81.2)
Congenital Anomalies	
Yes	26 (81.3)
No	6 (18.8)