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Constitutional Hypoplastic Anemia

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KEY WORDS Constitutional anemia; congenital; inherited; screening

ABSTRACT A large number of patients seen in the hematological practice belong to the broad group of bone marrow failure syndromes (BMFS) - comprising of aplastic anemia (AA) and myelodysplastic syndromes (MDS), AA being much more frequent than MDS. AA is diagnosed in the presence of hypocellular bone marrow and peripheral blood cytopenias. This disorder may be constitutional or acquired. The constitutional disorder may be either congenital (present at birth) or inherited (genetic but not necessarily expressed at birth). Multiple etiologies are implicated (Table 1) and the natural history of AA patients is very diverse (Alter 1993; Alter and Young 1993). According to the western literature, constitutional factors are operative in at least 1/3 of young patients with BMFS (Alter and Young 1993). The definition of exact etiology is important for the obvious reasons of proper manage-ment, family screening and genetic counseling etc.

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