Diagnosis, Genetics, and Management of Inherited Bone Marrow Failure Syndromes

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Abstract

The inherited bone marrow failure syndromes are traditionally considered to be pediatric disorders, but in fact, many of the patients now are diagnosed as adults, and many diagnosed as children now live to reach adulthood. The most common of these rare disorders include Fanconi anemia, dyskeratosis congenita, Shwachman-Diamond syndrome and amegakaryocytic thrombocytopenia, which often develop aplastic anemia and may evolve into myelodysplastic syndrome and acute myeloid leukemia; and Diamond-Blackfan anemia, severe congenital neutropenia, and thrombocytopenia absent radii, single cytopenias that rarely if ever become aplastic but have increased risks of leukemia. In addition, the first three syndromes have high risks of solid tumors: head and neck and anogenital squamous cell carcinoma in Fanconi anemia and dyskeratosis congenita, and osteogenic sarcoma in Diamond-Blackfan anemia. Diagnosis of a marrow failure syndrome requires recognition of characteristic physical abnormalities when present, and consideration of these disorders in the differential diagnosis of patients who present with

"acquired" aplastic anemia, myelodysplastic syndrome, acute myeloid leukemia, or atypically early cancers of the types seen in the syndromes. Ultimate proof will come from identification of pathogenic mutations in genes associated with each syndrome.

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