High suspicion for Shwachman Diamond syndrome is essential in young adults with MDS

Shwachman-Diamond Syndrome (SDS) may not be as rare as previously thought, especially in young adults with MDS. SDS is an inherited bone marrow failure syndrome with a high predisposition to myeloid malignancy previously diagnosed predominantly in the pediatric age group, and also commonly associated with pancreatic exocrine dysfunction.

The Shwachman-Diamond syndrome registry has previously shown that almost half of SDS patients do not present with the classic clinical features of neutropenia and steatorrhea (Myers et al., J Pediatr. 2014 Apr;164(4):866-70). MDS may be the first presentation of SDS in adults under 40, even in the absence of other clinical symptoms. Lindsley et al. identified mutations associated with Shwachman-Diamond syndrome in 4% of patients between the ages of 18 and 40 transplanted for MDS in a Center for Blood and Marrow Transplant Research cohort using targeted mutational analysis (N Engl J Med. 2017, Feb 9;376(6):536-547.), most of whom were previously undiagnosed. These adults with MDS and SDS all had also acquired a TP53 mutation and had a very poor prognosis after transplantation.

The diagnosis of SDS was unrecognized in most of these patients with SDS. It is important that SDS be screened for in this young adult MDS population to inform treatment decisions. SDS patients historically have done poorly with myeloablative transplant regimens and there is good data to suggest that reduced intensity regimens to treat bone marrow failure have improved outcomes in SDS (Bhatla et al., Bone Marrow Transplant. 2008 Aug;42(3):159-65). Additionally, these patients may be considered earlier for novel therapies or clinical trials in the setting of advanced MDS/AML.

The SDS Foundation was founded by parents and is driven by the energy of parents and patients trying to improve and advance care of patients with SDS. The Shwachman-Diamond syndrome Foundation continues to be a strong advocate and support system for patients with SDS and their families. The SDS Foundation and the Shwachman-Diamond syndrome registry, directed by Kasiani Myers and Akiko Shimamura, continue to provide resources to clinicians caring for patients with SDS and support for patients and their families. The goal of the research ongoing through the SDS Registry is to improve the health of individuals with SDS and develop better treatments.