

When the cause of MDS is unknown it is called idiopathic MDS. A so-called secondary MDS can develop after chemotherapy and radiation treatment for cancer or autoimmune diseases. It is possible that some chemicals (pesticides and benzene), cigarette smoking, and possibly viral infections can predispose to MDS. However, these links are circumstantial and in the majority of individuals developing MDS no obvious connection with environmental hazards can be found. MDS sometimes runs in families, suggesting a genetic link with the disease, but apart from an association with a rare congenital form of anemia (Fanconi aplastic anemia), no definite MDS provoking gene has been found. Myelodysplastic syndromes affect males slightly more often than females. The disorder occurs in any age group, but is far more common in older adults, occurring most often in individuals over 60 years of age. According to one estimate, 22 to 45 per 100,000 people over the age of 70 years have MDS.

Approximately 20,000 new patients are diagnosed each year in the United States. The number of new cases diagnosed each year is increasing, possibly due to better recognition of the disorder combined with an increasing proportion of elderly adults in the general population. Determining the exact frequency of MDS in the general population is difficult because of lack of reporting of patients with mild cytopenias. A diagnosis of myelodysplastic syndrome is made based upon a thorough clinical evaluation, a detailed patient history, and a variety of specialized tests including complete blood counts, examination of the blood smear (often more than one is needed), and bone marrow aspiration and biopsy. A complete blood count measures the number of red and white blood cells and platelets in the body. The blood smear and the small sample of bone marrow removed via a needle (the aspirate) is examined under a microscope for the characteristic features of MDS. Chromosome analysis is helpful for diagnostic and prognostic purposes.