MDS Myelodysplastic Syndromes

Chapter 1 Blood Cell Production

Human blood is essential to life. It is important for several transportation processes and circulation. There are red blood cells (erythrocytes), white blood cells (leucocytes), and platelets (thrombocytes). Bone marrow supplies the body with new blood cells. The stem cells (immature cells or blasts) of bone marrow are responsible for cell production.[1] When a patient is diagnosed with Myelodysplastic Syndromes (MDS), the bone marrow does not make normal cells due to dysplasia (abnormal shape), chromosome changes, or molecular changes. This results in too few cells or low blood count (cytopenia) or poor functioning cells. These cytopenias can be anemia (low red blood cells), thrombocytopenia (low platelets used for clotting), leukopenia (low white blood cells), or neutropenia (low neutrophils WBC that fight infection).[2] MDS represent many different conditions, not just a single disease, that are grouped together by several clinical characteristics. One feature of MDS is genetic instability.[3] MDS are a diverse group of disorders. These disorders impair peripheral blood cell production (cytopenias) and impair bone marrow cell production leading to abnormal bone marrow cells.[3,4]

Chapter 2 Genetic Instability

Myelodysplastic Syndromes was once described as “pre-leukemia”, but many patients never develop acute leukemia. Some die of complications from the cytopenias, so mylodysplastic syndromes was coined.[4]

Somatic point mutations (genetic alterations) are common in MDS. Some mutations are predictors of poor overall survival regardless of existing risk factors.[5] Examination of the bone marrow and the peripheral blood reveals morphologic features of the disease and exclude other conditions that can lead to cytopenias. Morphologic features include bone marrow cells with anemia features, atypical megakaryocytes (which produce platelets necessary for normal blood clotting), erythroid hyperplasia (large number of immature red blood cells), defective maturation in the myeloid (bone marrow) series, and increased blasts (immature) or ringed sideroblasts (in some). Ringed sideroblasts are erythroblast (stem cells) with iron-loaded mitochondria that form a ring around the nucleus. Peripheral blood results include monocytosis (increased number of white blood cells), Pelger Heut-like anomaly (unusually shaped white blood cells), circulating immature myeloid (bone marrow) or erythroid (immature) cells, and macrocytosis (enlarged red blood cells).[4]

Chapter 3 MDS Classifications

The World Health Organization (WHO) updated the classification system in 2016. MDS is classified based upon cell shape, presence of blasts, which cell types are involved, and any cytogenetic findings. [2]

MDS with single lineage dysplasia (MDS-SLD) is defined as a low number of 1-2 types of blood cells and 1 type of blood cell looks abnormal (dysplasia) in the bone marrow. At least 10% of the cells look abnormal. Less than 5% of cells in the bone marrow are blasts with no blasts in the bloodstream. [2]