Myeloid Neoplasms with Germline DDX41 Mutation

National Cancer Institute

Source

National Cancer Institute. Myeloid Neoplasms with Germline DDX41 Mutation. NCI Thesaurus. Code C151901.

An autosomal dominant familial myelodysplastic syndrome/acute myeloid leukemia syndrome characterized by inherited mutations in the gene on chromosome 5 encoding the DEAD box RNA helicase DDX41. Patients usually present with leukopenia, hypocellular bone marrow with prominent erythroid dysplasia and a normal karyotype, often leading to erythroleukemia. The prognosis is generally poor. (WHO 2017)