24th Annual Resident Poster Day

October 12, 2021



Case of Acute Promyelocytic Leukemia with Basophilic Differentiation and an ETV6 Mutation

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The translocation between chromosomes 15 and 17 t(15;17) at the promyelocytic leukemia (PML) and retinoic acid receptor alpha (RARA) is thought to be specific for acute promyelocytic leukemia (APL). We present a case of acute leukemia with evidence of t(15;17) by banding karyotype, FISH, and PCR, with rare hypogranular promyelocytes and significant basophilia detected on marrow aspirate, and an ETV6 missense mutation by molecular diagnostic testing. The patient underwent treatment with isotretinoin (ATRA) and arsenic trioxide (ATO) with attainment of a morphologic remission at the end of induction. There was no evidence of coagulopathy or basophil granule release with therapy. To our knowledge, this is the first report of the co-occurrence of APL with marrow basophilia in conjunction with an ETV6 mutation. The prognostic impact of an ETV6 mutation in this setting is unclear.