Disseminated Juvenile Xanthogranuloma (JXG) is a rare, non-Langerhans histiocytic disorder that usually affects children and infants. The most common locations of JXGs are the skin of the scalp and face (2). Visceral involvements of the eyes and testes have also been reported (3). Adult-onset JXG is very rare, and consists of solitary lesions that do not regress spontaneously.

Patient History

We present a case of a 22 year old male with history of B-ALL and immune thrombocytopenic purpura (ITP), who completed chemotherapy in 01/16 with negative bone marrow biopsy result. He was admitted for two days history of fever, fatigue, abdominal pain, right thigh pain, and erythema. During the hospital course, he was diagnosed with fungal sepsis leading to septic shock.

Materials and Methods

Peripheral blood and bone marrow aspirate smears were reviewed in conjunction with bone marrow biopsy and clot sections. A right elbow skin shave biopsy was also submitted for evaluation. Additional material were submitted for flow cytometry, karyotype analysis, and fluorescence in situ hybridization (FISH) as well as bacterial, fungal, and acid-fast bacilli cultures.

PeripheraBlood and Bone Marrow: The peripheral blood showed pancytopenia with no circulating blasts seen. The bone marrow was normocellular for age (70-80%). There is marked nodular proliferation composed of histiocytes and Touton-type multinucleated giant cells with 40% involvement (A). These nodules stain strongly positive for CD68 (B) and CD163. They are negative for CD1a and EBER.

Autopsy: JXG involving skin, spine (C), and spleen.

Skin biopsy: Right elbow skin biopsy revealed a dense, diffuse infiltrate composed predominately of histiocytes, consistent with JXG (D).

Flow cytometry: Blasts were approximately 0.2% of total events. No significant immunophenotypic abnormalities detected.

Genetic Testing: Normal karyotype (46, XY), with no BRAF gene mutation.

Cultures: Fungal organisms positive in blood and skin culture.

Discussion

Disseminated JXG is easily confused with Erdheim-Chester Disease (ECD), which is also a rare, non-Langerhans histiocytic proliferation disease most commonly characterized by multifocal osteosclerotic lesions, preferably in the long bones (in 95% of retrospective cases) (4). Interestingly, PET-CT of our patient also revealed numerous small foci of increased activity throughout the axial and appendicular skeleton. However, the X-rays of our patient’s lower extremities were unremarkable. Furthermore, an activating point mutation of BRAF V600E is also identified in approximately 50% of ECD cases. It is noteworthy because clinical response to BRAF inhibitor, Vemurafenib, has been reported. However, our patient does not have a BRAF V600E mutation, which also points us away from the diagnosis of ECD.

Conclusion

To the best of our knowledge, only 1 case of adult-onset JXG associated with B-ALL was reported (1). Here, we describe a unique case of adult-onset JXG involving skin, spine, and spleen, with association of recently resolved B-ALL. Our patient also has diffuse petechiae and erythema of the skin. Together with positive blood and skin cultures, it further complicated the association of JXG with oncohematologic disease and infection, creating a potential diagnostic dilemma.

References