Clinical and diagnostic aspects of VEXAS syndrome

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Conflict of interest: none

ABSTRACT. VEXAS syndrome is a rare disorder caused by a mutation in the UBA1 gene, located on the X chromosome. The mutated gene leads to reduced ubiquitination, delayed activation of innate immune pathways, and the development of systemic inflammatory responses. VEXAS is one of the multisystemic disorders with the most common hematological and rheumatological systemic abnormalities. Diagnosing the syndrome is challenging due to the similarity of its clinical symptoms to those of many other diseases. Genetic testing to detect a mutation in the UBA1 gene is a priority in the diagnosis of VEXAS. Treatment involves the administration of high-dose glucocorticoids, Janus kinase inhibitors, and, in some cases, allogeneic hematopoietic stem cell transplantation.

KEY WORDS: VEXAS syndrome, UBA1 gene mutation, ubiquitination, genetic testing, hematological and rheumatological disorders, glucocorticoids, hematopoietic stem cell transplantation.