Revista Información Científica

ISSN 1028-9933
Universidad de Ciencias Médicas de Guantánamo

Volume 102 Year 2023
DOI: 10.5281/zenodo.8104842

LETTER TO EDITOR

VEXAS syndrome: a new hematoinflammatory disease

Síndrome VEXAS: una nueva enfermedad hematoinflamatoria

Síndrome de VEXAS: uma nova doença hematoinflamatória

Roberto Michael Blanco-Alvarez[I]*, Sonia Haila Carbonell-Labadie[I], Margarita Montes de Oca-Carmenaty[II]

[I] Universidad de Ciencias Médicas de Santiago de Cuba. Santiago de Cuba, Cuba.
[II] Hospital Provincial Clínico Quirúrgico Docente "Saturnino Lora Torres". Santiago de Cuba, Cuba.

***Corresponding author:** rmblancoalvarez@gmail.com

Received: 24-05-2023 **Accepted:** 01-06-2023 **Published:**04-07-2023

How to cite this article:
Blanco-Alvarez RM, Carbonell-Labadie SH, Montes de Oca-Carmenaty M.Síndrome VEXAS syndrome: a new hematoinflammatory disease. Rev Inf Cient. 2023; 102:e4264. DOI: https://doi.org/10.5281/zenodo.8104842

Dear Editor:

The last few years have magnified different challenges for public health at international level. One could directly think of the COVID-19 pandemic, however, although it is the most recent and mediatic example, it has not been the only important discovery described in these years. Along with scientific and technical advances and the development of various health science research projects, diseases have been described that could be considered new, and which have increased the clinical-epidemiological characterization of others that were little studied before. A clear example of this is the discovery of VEXAS syndrome.

First reported in December 2020, it is considered a new autoinflammatory condition of acquired etiology. Named VEXAS (Vacuoles, Enzyme E1, X-linked, Autoinflammatory, Somatic), this syndrome is due to mutations of the enzyme ubiquitin 1 (UBA1) and was discovered after whole exome sequencing of 2560 patients. Twenty-five cases were reported and all corresponded to the male sex.[(1)]

According to early research concerning this syndrome, a new disease category described as hematoinflammatory disorder it could be defined. Primarily because somatic mutations occurring in the blood, causes inflammation at the systemic level, affecting multiple organs, especially the bone marrow.

VEXAS syndrome has been shown to have significant morbidity and to be capable of reducing life expectancy. The optimal standard of care for this disease, however, has not been adequately defined.[1]

In a study by Beckel, *et al*.[1], patients diagnosed with VEXAS had anemia with a high rate of macrocytosis and thrombocytopenia (91% of cases). The same research estimated an estimated variant prevalence of 1 in 14 thousand individuals and 1 in 8 thousand patients over 50 years of age.[1]

The pathognomonic finding of VEXAS is the presence of cytoplasmic vacuoles in myeloid and erythroid precursor cells, although it requires an objective differential diagnosis, since this type of vacuoles are present in other conditions such as zinc toxicity, alcoholism and copper deficiency.[1]

VEXAS syndrome is most commonly diagnosed in men over 50 years of age. Clinical manifestations of the disease include skin lesions, ocular symptoms, venous thrombosis, myalgia, chondritis, intermittent fever of unknown origin, and inflammatory processes affecting multiple organs. The most common dermatological findings include periorbital edema, urticaria, erythema nodosum, neutrophilic dermatoses and erythematous papules. Several authors have described significant pulmonary involvement in the disease.[1,2]

A study published by Georgin-Lavaialle, *et al*.[2] succeeded in reporting the different clinical phenotypes of VEXAS syndrome, where clinical-biological profiles and the associated UBA1 mutation were taken into account. However, it raised the need for further prospective research to confirm the data obtained and determine the correct management of the disease, since they failed to know the efficacy of treatments and the limited number of deaths due to VEXAS did not help in formulating accurate conclusions on survival.

With so many questions about this recent and seemingly common condition, science must continue to push for research to expand clinical and epidemiological knowledge about VEXAS syndrome.

Cuba, having an aging population (the disease being common in this age group) cannot postpone studies to determine its incidence in the country and the clinical characteristics of its evolution. Likewise, the professionals of the specialties related to its diagnosis and/or treatment should be updated in this subject, in order to know how to recognize the syndrome and establish an opportune protocol for its management.

REFERENCES

1. Al-Hakim A, Savic S. An update on VEXAS syndrome. Exp Rev Clin Immunol [Internet]. 2023 Feb [citado 7 Feb 2023]; 19(2):203-215. DOI: https://doi.org/10.1080/1744666X.2023.2157262

2. Georgin-Lavaialle S, Terrier B, Guedon AF, Heiblig M, Comont T, Lázaro E, *et al.*Further characterization of clinical and laboratory features in VEXAS syndrome: large-scale analysis of a multicentre case series of 116 French patients. Br J Dermatol [Internet]. 2022 Mar [citado 7 Feb 2023]; 186(3):564-574. DOI: https://doi.org/10.1111/bjd.20805

Conflict of interest:
The authors declare that there are no conflicts of interest.

Funding:
The authors did not receive funding for the development of this article.