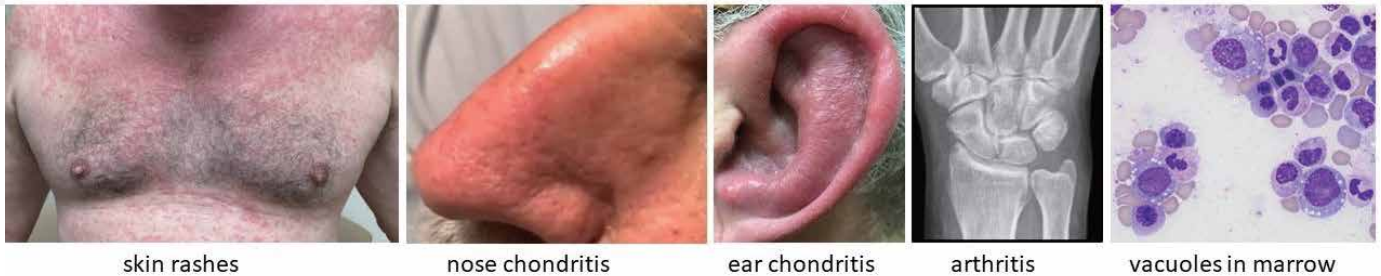
DIACARTA®

QClamp® Plex VEXAS Syndrome Test

Genetic Testing for VEXAS Syndrome

VEXAS syndrome is a chronic, progressive disease that causes severe systemic autoinflammatory and hematological symptoms, including skin rashes that may be painful and swelling, and pain in nose and ears (nose and/or ear chondritis), cough and shortness of breath, pain and swelling in joints, inflammation of blood vessels, and failure of the bone marrow (e.g., macrocytic anemia, myelodysplastic syndrome). Identifying patients with VEXAS syndrome is very challenging, and the diagnosis is often delayed or missed. Clinical vigilance and collaboration among clinicians (Rheumatology, Hematology, Dermatology, etc.) and pathologists are required to diagnose the disease without delay.

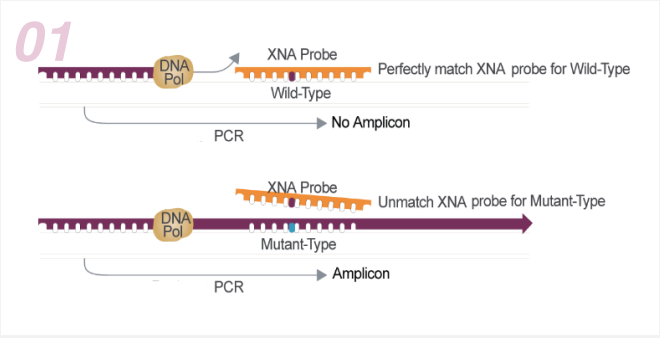
Patients with VEXAS syndrome often have **V**acuoles in their bone marrow precursor cells, and have a mutation in the **E1** ubiquitin activating enzyme, which is encoded by the *UBA1* gene located on the **X** chromosome. VEXAS patients have **A**utoinflammation and carry the **S**omatic *UBA1* mutation, which is acquired at some point later in life and not inherited from parents.



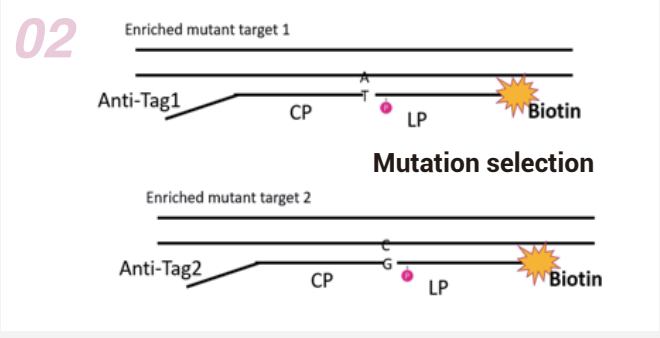
skin rashes | nose chondritis | ear chondritis | arthritis | vacuoles in marrow precursor cells

QClamps® Plex Vexas Syndrome Diagnosis Assay

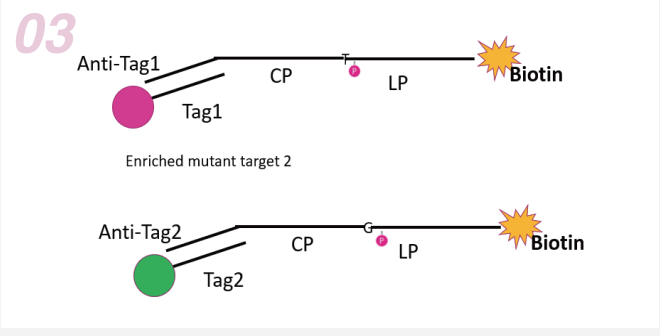
DiaCarta has developed and fully validated a genetic test for VEXAS syndrome, called QClamp® Plex VEXAS Syndrome Test. The assay detects all the known mutations in UBA1 gene that are associated with VEXAS syndrome. The assay uses DiaCarta's proprietary QClamp® Plex technology to increase assay sensitivity and specificity.



Mutant DNA enrichment by XNA



Target DNA Mutation selection by ligation



Hybridization with Luminex beads

Signal reading using Luminex MAGPIX system

Mutations in the *UBA1* gene can be detected on patient's peripheral blood sample. There is no need to obtain bone marrow sample for the QClamp® Plex VEXAS Syndrome Test.

Specimen(s) required

Preferred specimen is 3 mL whole blood in an EDTA (lavender-top) tube (minimum volume 1.0 mL). Alternative specimen is 2-3 mL bone marrow aspirate collected in EDTA tube or extracted DNA sample from a CLIA-certified laboratory.

Storage and transport temperature

Room temperature or refrigerated.

Specimen stability

Room temperature: 7 days; Refrigerated: 7 days; DO NOT freeze specimen.

Turnaround time

3 business days after receiving sample at DiaCarta CLIA-certified laboratory.

Intended use

The test is available as a clinical test, research test, and also a research product.

Ordering Information

Clinical test catalog #: DC-10-1002

Research test catalog #: DC-10-1003

Research product catalog #: DC-10-0001R

References:

1. Beck DB, Ferrada MA, Sikora KA, et al. Somatic mutations in UBA1 and severe adult-onset autoinflammatory disease. N Engl J Med. 2020;383(27):2628-2638.

2. Beck DB, Bodian DL, Shah V, et al. Estimated Prevalence and Clinical Manifestations of UBA1 Variants Associated with VEXAS Syndrome in a Clinical Population. JAMA. 2023;329(4):318-324.

3. Koster MJ, Samec MJ, Warrington KJ. VEXAS syndrome-A review of pathophysiology, presentation, and prognosis. J Clin Rheumatol. 2023;29(6):298-306.