

Zespół monoklonalnej aktywacji mastocytów

Kod Orpha: 529468 Kod OMIM:

Opis choroby *

Definicja

A rare hematologic disease characterized by symptoms of mast cell activation in the absence of cutaneous findings, as well as absence of diagnostic criteria of systemic mastocytosis with tryptase levels of less than 20 ng/ml and normal to low burden of mast cells. Bone marrow biopsy reveals the presence of monoclonal mast cells carrying the *KIT* D816V mutation and/or expressing CD25. Patients present with recurrent episodes of flushing, headache, hypotension, abdominal cramping, nausea, diarrhea, cardiac arrhythmias, bronchoconstriction, and bleeding diathesis, among others.

Dane

Klasyfikacja

Choroba

Synonimy

Monoclonal MCAD

Monoclonal MCAD

Kod ORPHA

529468

Kod OMIM

-

Kod ICD10

C94.3

Kod ICD11

-

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.