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
-

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*Źródło

orphanet