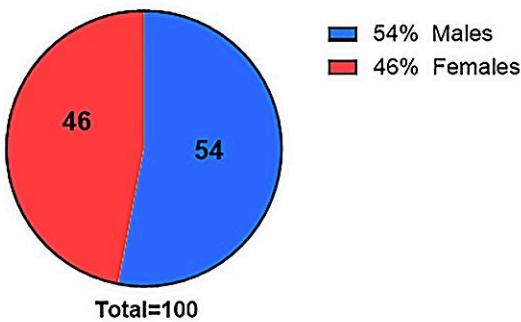


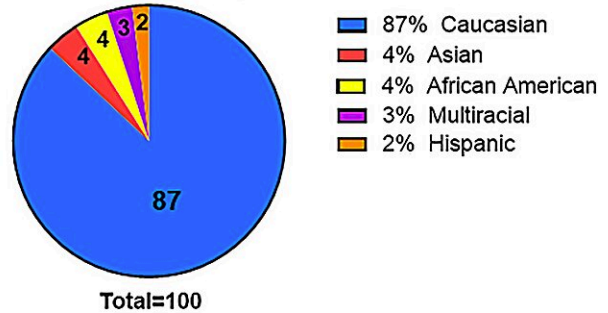
# Prevalence and impact of the KIT M541L variant in patients with mastocytosis

July 24 2024

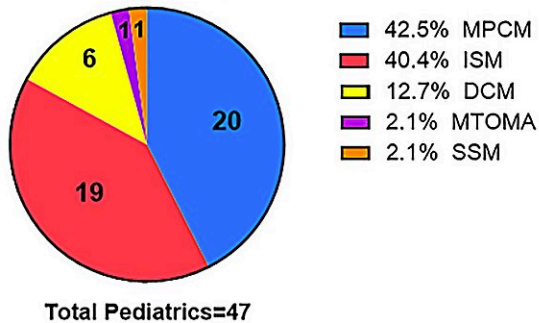
**A Total Population**



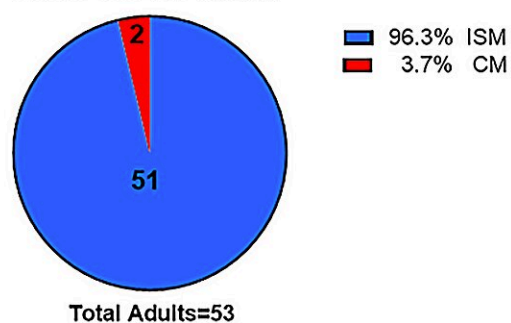
**B Patient Ancestry**



**C Pediatric-Onset Patients**



**D Adult-Onset Patients**



Mastocytosis patient demographics. Credit: 2024 Aldama et al.

A new research paper titled "Prevalence and impact of the KIT M541L variant in patients with mastocytosis" has been [published](#) in *Oncotarget*.

Activating mutations in KIT, particularly D816V, have been associated

with mastocytosis. Additionally, expression of heterozygous KIT M541L has been primarily reported in patients with pediatric mastocytosis.

In this new study, researchers from the National Institutes of Health examined the prevalence of this variant in pediatric and [adult patients](#) with mastocytosis (n = 100) compared to ancestry-matched 1000 Genome Project controls (n = 500) and patients with idiopathic anaphylaxis (n = 23). They then compared clinical symptoms and laboratory data on patients with systemic and cutaneous mastocytosis and bone marrow histopathology in a matched cohort with and without the KIT M541L variant.

"We found a significant association between KIT M541L genotype and the diagnosis of mastocytosis," the team reports.

Overall, the KIT M541L variant was identified in 19 individuals; the majority were diagnosed with systemic mastocytosis (89.4%) with an associated KIT D816V mutation. There were no significant differences in peripheral blood parameters between groups. Patients with mastocytosis carrying the KIT M541L [variant](#) did not demonstrate significant differences in symptomatology compared to a matched reference cohort (n = 13/81) without KIT M541L. In patients with idiopathic anaphylaxis, no significant associations were observed.

"To our knowledge, this is the first case/control study to show a significant genetic association with mastocytosis at the KIT M541L locus," the team adds.

**More information:** Luisa N. Dominguez Aldama et al, Prevalence and impact of the KIT M541L variant in patients with mastocytosis, *Oncotarget* (2024). [DOI: 10.18632/oncotarget.28614](https://doi.org/10.18632/oncotarget.28614)

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