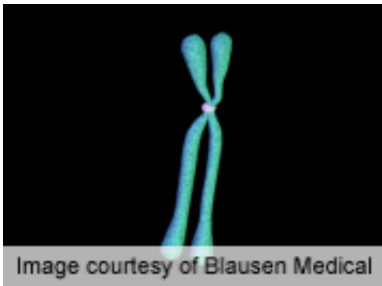


# ASH: CALR mutations ID'd in myeloproliferative neoplasms

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(HealthDay)—Many patients with myeloproliferative neoplasms without mutations in the Janus kinase 2 gene (*JAK2*) or in the thrombopoietin receptor gene (*MPL*) have mutations in the *CALR* gene encoding calreticulin, according to two studies published online Dec. 10 in the *New England Journal of Medicine*. The research was published to coincide with presentation at the annual meeting of the American Society of Hematology, held from Dec. 7 to 10 in New Orleans.

Thorsten Klampfl, Ph.D., from the CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences in Vienna, and colleagues performed whole-exome sequencing to identify [somatic mutations](#) in six patients with primary myelofibrosis without mutations in *JAK2* or *MPL*. The researchers identified somatic insertions or deletions in exon 9 of *CALR* in all patients. Among patients with

essential thrombocythemia and primary myelofibrosis, *CALR* mutations were mutually exclusive with *JAK2* and *MPL* mutations. *CALR* mutations were seen in 67 and 88 percent, respectively, of those with essential thrombocythemia and primary myelofibrosis, with non-mutated *JAK2* or *MPL*.

Jyoti Nangalia, M.B.Chir., from the Cambridge Institute for Medical Research in the United Kingdom, and colleagues performed exome-sequencing of samples from 151 patients with myeloproliferative [neoplasms](#). The researchers identified 1,498 mutations, with somatic *CALR* mutations in 70 to 84 percent of myeloproliferative neoplasms with non-mutated *JAK2*. The mutations were located in exon 9 and generated a +1 base-pair frameshift.

"Somatic mutations in the endoplasmic reticulum chaperone *CALR* were found in a majority of patients with myeloproliferative neoplasms with non-mutated *JAK2*," Nangalia and colleagues write.

Several authors from the Klampfl study reported holding pending patent applications regarding the use of calreticulin gene [mutations](#) for the diagnosis of diseases and targeting for therapy for myeloproliferative neoplasms.

**More information:** [Abstract - Klampfl](#)

[Full Text](#)

[Abstract - Nangalia](#)

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