

INTRACRANIAL HYPERTENSION MIMICKING PSEUDOTUMOR CEREBRI AS THE PRIMARY MANIFESTATION OF JAK2-POSITIVE MYELOPROLIFERATIVE NEOPLASMS: A TWO-CASE SERIES

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Background. Idiopathic intracranial hypertension (IIH) is a rare condition (0.9-2/100.000 persons) that affects mainly obese young women. Secondary intracranial hypertension due to cerebral venous sinus thrombosis (CVST) is far rarer condition (0.5/100,000 persons). However, chronic, oligosymptomatic CVST can perfectly mimic the clinical phenotype of IIH. In young adults carrying the JAK2V617F mutation, these clinical presentation and condition can represent the primary herald manifestation for an underlying myeloproliferative neoplasm (MPN). We present the cases of two women both born in 1983 affected from severe intracranial hypertension driven by this mechanism.

Case Presentations.• Case 1: A normal-weight female developed in 2018 a clinical picture characterized by tinnitus, negative scotomas, and severe headache. Patient was hospitalized elsewhere and bilateral papilledema was ascertained. Also MRI was performed and reported empty sella, and optic hydrops; colleagues started acezolamide without clinical changes. We evaluated patients few weeks later since persistence of headache condition and we performed a second MRI/MRV study confirming previous reports, but also showing extensive chronic parcellar thrombosis in the right transverse/sigmoid and superior sagittal sinuses. Patient affected from JAK2-positive PMF was treated with acetazolamide and warfarin. Visual symptoms resolved by November 2019, but a high frequency/chronic migraine-like headache persisted. The introduction of ruxolitinib in 2022 marked a turning point, relieving pain symptoms and allowing acetazolamide discontinuation by 2023, though sporadic perimenstrual headache episodes persist. Follow-up imaging up to 2025 demonstrated a stable, unresolved, chronic thrombotic framework.• Case 2: A female with class I obesity presented in late 2025 with recent abrupt onset headache, bilateral peripheral distorted vision, marked bilateral papilledema, and an elevated opening pressure of 350 mmH2O. MRI/MRV confirmed optic hydrops and a less extensive parcellar thrombotic defect in the right transverse-sigmoid sinus. Subsequent workup revealed a JAK2V617F-positive unclassifiable MPN. Driven by acetazolamide, rapid weight loss via semaglutide, and edoxaban, she achieved rapid clinical resolution, with mid-2026 neuroimaging confirming near-complete sinus recanalization.

Conclusions. While sparse literature reports link between intracranial hypertension conditions and essential thrombocythemia or polycythemia vera, cases secondary to primary myelofibrosis (PMF) or unclassifiable MPNs remain exceptionally rare. In our cases, lower initial thrombotic burden early managed with edoxaban favored complete radiologic resolution, whereas extensive organized and more lasting thrombosis condition resulted in a chronic headache phenotype where targeted JAK1/2 inhibition, rather than standard pressure-lowering agents or anticoagulant, resulted essential for long-term clinical compense.