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Letter to the Editor

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Author for correspondence:
*Maju M. Koola, MD,
Emails: koola-maju@cooperhealth.edu; majumkoola@gmail.com

N.P. and Ar.M. contributed equally to this work.

Hermansky-Pudlak syndrome presenting with paranoid delusion

Nishi Parikh[1], Aravind Menon[2], Aimin Mitwally[3], Janeni Nayagan[3], Gretchen E. Magnani[4], Francis Aguilar[4] and Maju M. Koola[4]*

[1]Cooper Medical School of Rowan University, Camden, NJ, USA, [2]St. George's University School of Medicine, True Blue, Grenada, [3]Department of Psychiatry and Behavioral Health, Cooper University Hospital Care, Camden, NJ, USA and [4]Department of Psychiatry and Behavioral Health, Cooper University Health Care, Cooper Medical School of Rowan University, Camden, NJ, USA

To the Editor,

Hermansky-Pudlak syndrome (HPS) is a rare autosomal recessive disorder with classical presentations of oculocutaneous albinism, a bleeding diathesis due to the absence of dense bodies in platelets, pulmonary fibrosis, and granulomatous colitis.[1–3] These systemic manifestations are due to lysosome-related organelle dysfunction.[1] HPS can present as 1 of 9 subtypes, each due to specific mutation variations, with types 1 and 4 being the most severe.[1] Mutations in the HPS1 gene are responsible for the majority of HPS cases.[1] HPS type 3 classically presents milder than the other 8 subtypes.[1] HPS type 4 has shown an increased susceptibility to schizophrenia.[4,5]

Case

A 19-year-old male with a past medical history of HPS type 3 (how he was diagnosed and age at diagnosis could not be determined from his chart), currently asymptomatic, and no past psychiatric history exhibited new-onset paranoid delusion over a 24-h period in April 2022. The patient expressed a belief that his family members were conspiring to kill him using a knife. The next day, he fled to his uncle's house for safety, where the police were called, and the patient was brought to the emergency department (ED). In the ED, the patient endorsed previous suicidal ideation but denied homicidal ideation, auditory or visual hallucinations, and substance use. Initial laboratory tests were significant for a negative urine drug screen.

Due to new-onset paranoid delusions, the patient was voluntarily admitted to the inpatient psychiatric unit to provide respite from psychosocial stressors, medication management, and therapy. He revealed that he recently voluntarily withdrew from college, citing declining mental health, and now lives with his parents and 3 younger siblings, one of whom is also diagnosed with HPS type 3. There was no family history of psychosis. The patient was started on aripiprazole 5 mg and was gradually increased to 15 mg for psychosis. The patient's hospital course was otherwise unremarkable with the resolution of his paranoia, and he tolerated the medication with no significant adverse reactions. He was discharged home and later admitted to a partial hospitalization program for 2 weeks, where he was switched to olanzapine 5 mg nightly. We do not know why this switch was made. He continues to follow up with outpatient psychiatry and plans to return to college during the next academic year.

Discussion

To the best of our knowledge, this is the first report of HPS type 3 presenting with psychiatric issues—he had paranoid delusion. HPS is a rare, multisystem disorder, and previous studies have shown an increased susceptibility to schizophrenia in patients with HPS type 4.[4] Our patient had HPS type 3 with no past psychiatric history, but he experienced paranoid delusions, suggesting that there may be a genetic predisposition to psychiatric illness in patients with the type 3 variant as well. Further studies should explore this association, as this knowledge may allow for improved awareness of this differential diagnosis (HPS types 3 and 4 with psychosis). When treating patients with HPS, it may be important to obtain a thorough psychiatric history and to make the patient aware of the association of HPS with psychosis.

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