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When PHACES hide faces: A Case Report of PHACE Syndrome with Depression

Isaac Arefi[1*], Anish Sharma[1], Jessica Sedlak[1], Andrew Michaelis[1], Navtej Mann[1], Nilan Mani[1], Harmandeep Pannu[2], Tanvir Singhh[3]

[1]Resident, Department of Neurosciences and Psychiatry, 3000 Arlington Avenue, The University of Toledo, Toledo OH 43615

[2]Fellow, Department of Neurosciences and Psychiatry, 3000 Arlington Avenue, The University of Toledo, Toledo OH 43615

[3]Professor, Vice Chair for Clinical Services, Department of Neurosciences and Psychiatry, 3000 Arlington Avenue, The University of Toledo, Toledo OH 43615

Email: isaac.arefi@utoledo.edu

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Posterior fossa malformations, Hemangiomas, Arterial anomalies, Cardiac defects, Eye abnormalities (PHACE Syndrome) is an extremely rare, neurocutaneous syndrome. Its exact incidence and prevalence is currently unknown, but there are around 300 cases that have been reported since its first report in 1996. Majority of the current research focuses on the characteristic symptoms of PHACE Syndrome, however there is increasing evidence that PHACE syndrome is potentially the etiology of a broad list of symptoms. There is sparse literature on the effects and long term outcomes of PHACE syndrome or the psychiatric manifestations of the disease. The goal of this report is to highlight the psychiatric and neurologic history and treatment in a patient with PHACE syndrome.

Patient is a 13-year-old boy with confirmed PHACE syndrome, diagnosed at birth. In November 2024, the patient was admitted to inpatient psychiatry after cutting the sole of the foot with sewing scissors. Past medical and psychiatric history of agenesis of corpus callosum with interhemispheric cyst, esotropia, global development delay, failure to thrive, anxiety, sleep terror disorder, ADHD, and ODD. Mother provided a collateral history of uncontrollable, violent outbursts where he throws objects and punches holes in walls and doors. Patient endorses suicidal and homicidal ideation.

To our knowledge, this is the second report of PHACE syndrome that presents with severe psychiatric and neurologic disease. Much of the current research is focused on the cardinal symptoms of PHACE syndrome and does not follow the psychiatric manifestations of the disease. This case highlights the need for further research regarding auditory, language, neurologic, cognitive, psychotic, and mood symptoms in patients with PHACE syndrome. Establishing a link between psychiatric and physical manifestations of PHACE syndrome may lead to more recognition and proper diagnosis of the disease and allow for further education, treatment, and psychosocial interventions.

Keywords: PHACE Syndrome, Developmental Delay, Neurodevelopmental Disorders, Intellectual Disability, Psychiatric Evaluation

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