

Duke Faculty and Team Publications about Alternating Hemiplegia of Childhood

- **Gene Therapy of AHC:** Hunanyan AS, et al. Adeno-Associated Virus-Mediated Gene Therapy in the Mashloul, *Atp1a3^{Mashl/+}*, Mouse Model of Alternating Hemiplegia of Childhood. Hum Gene Ther. 2021. doi: 10.1089/hum.2020.191. Epub ahead of print. PMID: [33577387](#).
- **Clinical Manifestations and Management of AHC:** Moya-Mendez ME. Early onset severe ATP1A2 epileptic encephalopathy: Clinical characteristics and underlying mutations. Epilepsy Behav. 2021;116:107732. doi: 10.1016/j.yebeh.2020.107732. Epub ahead of print. PMID: [33493807](#).
- **Gene Therapy in General:** Uchitel J, Kantor B, Smith EC, Mikati MA. Viral-Mediated Gene Replacement Therapy in the Developing Central Nervous System: Current Status and Future Directions. Pediatr Neurol. 2020 Sep;110:5-19. doi: 10.1016/j.pediatrneurol.2020.04.010. Epub 2020 Apr 24. PMID: [32684374](#).
- **Clinical Manifestations and Management of AHC:** Wallace K. Characterization of Severe and Extreme Behavioral Problems in Patients With Alternating Hemiplegia of Childhood. Pediatr Neurol. 2020;111:5-12. Doi: 10.1016/j.pediatrneurol.2020.06.012. Epub 2020 Jun 27. PMID: [32951661](#).
- **Heart in AHC:** Balestrini S, et al. Cardiac phenotype in *ATP1A3*-related syndromes: A multicenter cohort study. Neurology. 2020;95(21):e2866-e2879. doi: 10.1212/WNL.00000000000010794. Epub 2020 Sep 10. PMID: [32913013](#).
- **Clinical Manifestations and Management of AHC:** Pratt M, et al. Alternating Hemiplegia of Childhood: gastrointestinal manifestations and correlation with neurological impairments. Orphanet J Rare Dis. 2020 Sep 3;15(1):231. Doi: 10.1186/s13023-020-01474-w. PMID: [32883312](#).
- **Non-AHC ATP1A3 disease:** Prange L, et al. D-DEMØ, a distinct phenotype caused by *ATP1A3* mutations. Neurol Genet. 2020 Aug 4;6(5):e466. Doi: 10.1212/NXG.0000000000000466. PMID: [32802951](#).
- **MRI in AHC:** Ghusayni R, et al. Magnetic resonance imaging volumetric analysis in patients with Alternating hemiplegia of childhood: A pilot study. Eur J Paediatr Neurol. 2020;26:15-19. doi: 10.1016/j.ejpn.2020.02.001. Epub 2020 Feb 13. PMID: [32115366](#).
- **Clinical Manifestations and Management of AHC:** Tran L, et al. Epileptic encephalopathy with features of rapid-onset dystonia Parkinsonism and alternating hemiplegia of childhood: a novel combination phenotype associated with *ATP1A3* mutation. Epileptic Disord. 2020;22(1):103-109. doi: 10.1684/epd.2020.1127. PMID: [32043468](#).
- **Clinical Manifestations and Management of AHC:** Uchitel J, et al. Social impairments in alternating hemiplegia of childhood. Dev Med Child Neurol. 2020;62(7):820-826. doi: 10.1111/dmcn.14473. Epub 2020 Feb 7. PMID: [32031250](#).
- **Clinical Manifestations and Management of AHC:** Uchitel J, et al. The epileptology of alternating hemiplegia of childhood. Neurology. 2019 Sep 24;93(13):e1248-e1259. Doi: 10.1212/WNL.00000000000008159. Epub 2019 Sep 4. Erratum in: Neurology. 2020;95(15):708. PMID: [31484714](#).

- **Sleep in AHC:** Kansagra S, et al. Polysomnography Findings and Sleep Disorders in Children With Alternating Hemiplegia of Childhood. *J Clin Sleep Med*. 2019;15(1):65-70. doi: 10.5664/jcsm.7572. PMID: [30621840](#).
- **Clinical Manifestations and Management of AHC:** Jasien JM, et al. Cognitive, adaptive, and behavioral profiles and management of alternating hemiplegia of childhood. *Dev Med Child Neurol*. 2019;61(5):547-554. Doi: 10.1111/dmcn.14077. Epub 2018 Oct 26. PMID: [30362107](#).
- **E815K (Matoub) Mouse Model and Effect of Flunarizine:** Helseth AR, et al. Novel E815K knock-in mouse model of alternating hemiplegia of childhood. *Neurobiol Dis*. 2018;119:100-112. doi: 10.1016/j.nbd.2018.07.028. Epub 2018 Jul 30. PMID: [30071271](#).
- **D801N (Mashlool) Mouse Model:** Hunanyan AS, et al. Mechanisms of increased hippocampal excitability in the Mashl^{+/-} mouse model of Na⁺/K⁺-ATPase dysfunction. *Epilepsia*. 2018 Jul;59(7):1455-1468. doi: 10.1111/epi.14441. Epub 2018 Jun 11. PMID: [29889309](#).
- **Clinical Manifestations and Management of AHC:** Masoud M, et al. Motor function domains in alternating hemiplegia of childhood. *Dev Med Child Neurol*. 2017;59(8):822-828. Doi: 10.1111/dmcn.13443. Epub 2017 May 25. PMID: [28543714](#).
- **Overview of AHC:** Masoud M, et al. Diagnosis and Treatment of Alternating Hemiplegia of Childhood. *Curr Treat Options Neurol*. 2017;19(2):8. doi: 10.1007/s11940-017-0444-7. PMID: [28337648](#).
- **Correlation Between Mutation and Clinical Manifestations:** Panagiotakaki E, et al. Clinical profile of patients with ATP1A3 mutations in Alternating Hemiplegia of Childhood-a study of 155 patients. *Orphanet J Rare Dis*. 2015;10:123. doi: 10.1186/s13023-015-0335-5. PMID: [26410222](#).
- **Heart and AHC:** Jaffer F, et al. Faulty cardiac repolarization reserve in alternating hemiplegia of childhood broadens the phenotype. *Brain*. 2015;138(Pt 10):2859-74. doi: 10.1093/brain/awv243. Epub 2015 Aug 21. PMID: [26297560](#).
- **D801N (Mashlool) Mouse Model:** Hunanyan AS, et al. Knock-in mouse model of alternating hemiplegia of childhood: behavioral and electrophysiologic characterization. *Epilepsia*. 2015;56(1):82-93. doi: 10.1111/epi.12878. Epub 2014 Dec 19. PMID: [25523819](#).
- **Overview of ATP1A3 Related Disorders and their Biology:** Heinzen EL, et al. Distinct neurological disorders with ATP1A3 mutations. *Lancet Neurol*. 2014 May;13(5):503-14. doi: 10.1016/S1474-4422(14)70011-0. PMID: [24739246](#).
- **Overview of AHC:** Kansagra S, et al. Alternating hemiplegia of childhood. *Handb Clin Neurol*. 2013;112:821-6. doi: 10.1016/B978-0-444-52910-7.00001-5. PMID: [23622289](#).
- **Gene Discovery Article:** Heinzen EL, et al. De novo mutations in ATP1A3 cause alternating hemiplegia of childhood. *Nat Genet*. 2012;44(9):1030-4. doi: 10.1038/ng.2358. Epub 2012 Jul 29. PMID: [22842232](#).
- **Large Series of Clinical Manifestations and Management of AHC:** Mikati MA, et al. Alternating hemiplegia of childhood: clinical manifestations and long-term outcome. *Pediatr Neurol*. 2000;23(2):134-41. doi: 10.1016/s0887-8994(00)00157-0. PMID: [11020638](#).

- **PET Scan Changes in Cerebellum in AHC:** Mikati M. Alternating hemiplegia of childhood. *Pediatr Neurol.* 1999;21(4):764. doi: 10.1016/s0887-8994(99)00090-9. PMID: [10580894](#).
- **Clinical Manifestations and Management of AHC:** Dangond F, et al. Focal brain dysfunction in a 41-year old man with familial alternating hemiplegia. *Eur Arch Psychiatry Clin Neurosci.* 1997;247(1):35-41. doi: 10.1007/BF02916251. PMID: [9088804](#).
- **The First Article to Show that AHC is a Genetic Disease by Studying the First Reported Family with AHC:** Mikati MA, et al. A syndrome of autosomal dominant alternating hemiplegia: clinical presentation mimicking intractable epilepsy; chromosomal studies; and physiologic investigations. *Neurology.* 1992; 42(12):2251-7. doi: 10.1212/wnl.42.12.2251. PMID: [1361034](#).