

Bringing Cerebral Palsy (CP) Into the Genomics Era

Tuesday, January 17

12:30pm

Hybrid: Rosedale Room and Zoom

For Researchers



Speaker:

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Host: Rajiv R. Ratan, M.D., Ph.D.

For more information contact

Darlene White

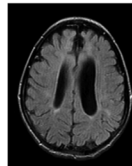
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Abstract

As the most common childhood-onset motor disability, cerebral palsy (CP) affects 764,000 individuals in the United States alone and accounts for lifetime healthcare expenses of \$1.4 million per person. In approximately 20% of cases (or 150,000 individuals), the cause is unknown, raising concern for a genetic disorder, including treatable conditions where a molecular diagnosis may positively alter the trajectory of a child's development. Our research aims to (1) determine the genetic landscape of CP, including potentially treatable conditions (2) develop rules to predict which individuals with CP are likely to have a genetic disorder and (3) contribute to an emerging scientific understanding of the pathophysiology of CP that will impact a large patient population.

• Spastic quadriplegic CP

- Profound ID
- Epilepsy
- Microcephaly
- Short stature
- Failure to thrive
- Cerebral atrophy
- Leukoencephalopathy

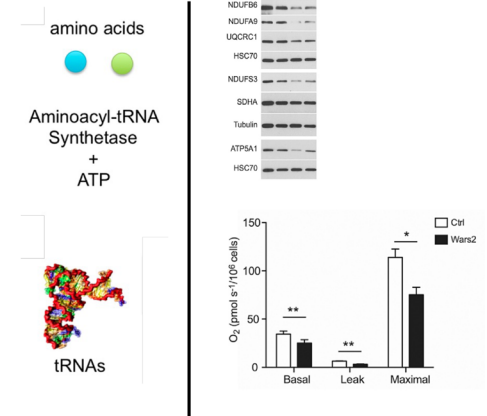


• Perinatal adversity

WARS2 c.938A>T
p.(K313M)

WARS2 c.298_300delCTT
p.(L100del)

Compound heterozygous



1. Chopra M, Gable D, Nichols J, Tsao A, Rockowitz S, Sliz P, Barkoudah E, Bastianelli L, Coulter D, Davidson E, DeGusmao C, Fogelman D, Huth K, Marshall P, Nimec D, Sanders J, Shore B, Snyder B, Stone S, Ubeda A, Watkins C, Berde C, Bolton J, Brownstein C, Costigan M, Fakhari D, Lai A, Luria A, Paciorkowski A, Pinto A, Pugh J, Rodan L, Roe E, Swanson L, Zhang B, Kruer M, Sahin M, Poduri A, Srivastava S. **Mendelian etiologies identified with whole exome sequencing in cerebral palsy.** Ann Clin Transl Neurol. 2022 Feb;9(2):193-205. Pubmed PMID ID: 35076175
2. Srivastava S, Love-Nichols JA, Dies KA, Ledbetter DH, Martin CL, Chung WK, Firth HV, Frazier T, Hansen RL, Prock L, Brunner H, Hoang N, Scherer SW, Sahin M, Miller D. **Meta-analysis and multidisciplinary consensus statement: exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders.** Genetics in medicine : official journal of the American College of Medical Genetics. 2019 Nov; 21(11): 2413-2421. Available from: <https://pubmed.ncbi.nlm.nih.gov/31182824/>. Pubmed PMID: 31182824; PubMed Central PMCID: PMC6831729.
3. Srivastava S, Jo B, Zhang B, Frazier T, Gallagher A, Peck F, Levin A, Mondal S, Li Z, Filip-Dhima R, Geisel G, Dies K, Diplock A, Eng C, Hanna R, Sahin M, Hardan A. **A randomized controlled trial of Everolimus for neurocognitive symptoms in PTEN hamartoma tumor syndrome.** Hum Mol Genet . 2022 May 20. Pubmed PMID: 35594551.

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