Study supports genetic testing for people with cerebral palsy

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A Geisinger meta-analysis of recent research on the genetics of cerebral palsy (CP) provides evidence that genetic testing should be offered as the standard of care for people with the disorder, similar to current

recommendations for individuals with other neurodevelopmental disorders (NDD). The findings were published on March 6 in *JAMA Pediatrics*.

Individual cases of CP—a condition that affects movement, balance and posture—have often been attributed to birth asphyxia, although recent studies show that asphyxia accounts for less than 10% of cases. A growing body of evidence suggests that a significant proportion of CP is caused by genetic changes, as is the case in other NDD like intellectual disability and autism spectrum disorder.

Sequencing of the whole genome—all genetic information in the body—or the exome—the genome's protein-coding regions—is a standard diagnostic test for people with NDD. However, this recommendation does not currently include CP, so people with the disorder may not be offered genetic testing unless they also have a NDD.

The Geisinger research team evaluated 13 studies published between 2013 and 2022, comprising 2,612 people, related to CP and genetic testing. Their analysis found that the genetic diagnostic yield—the proportion of cases with a genetic change that accounts for the disorder—in CP is similar to that of other NDD. This finding supports including CP as one of the NDD clinical diagnoses for which genetic testing is currently recommended as part of clinical care.

"Waiting for a co-occurring diagnosis of intellectual disability or autism in an individual with cerebral palsy to consider genetic testing is a missed opportunity to improve clinical outcomes," said Andres Moreno De Luca, M.D., M.B.A, physician-scientist and clinical neuroradiologist at Geisinger. "Since motor disorders like cerebral palsy can be identified earlier than other neurodevelopmental disorders, genetic testing for people with cerebral palsy may allow for quicker identification of

genetic changes and facilitate early interventions and potential treatment."

More information: Pedro J. Gonzalez-Mantilla et al, Diagnostic Yield of Exome Sequencing in Cerebral Palsy and Implications for Genetic Testing Guidelines, *JAMA Pediatrics* (2023). DOI: 10.1001/jamapediatrics.2023.0008

Clare van Eyk et al, All Patients With a Cerebral Palsy Diagnosis Merit Genomic Sequencing, *JAMA Pediatrics* (2023). DOI: 10.1001/jamapediatrics.2023.0015 , jamanetwork.com/journals/jamap … /fullarticle/2801967

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