Paternal Sperm DNA Methylation as a Predictive Marker of Autism Risk

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Technology description

Unmet Need:
Autism spectrum disorder (ASD), is a complex neurodevelopmental disorder that refers to a range of conditions characterized by challenges with behavior including social skills, communication, and repetitive behaviors, all of which have ranges and severities that are patient specific. Currently, there are no medical tests such as a scan or in vitro diagnostic blood test that can be used to quickly and easily diagnose ASD. The diagnosis process involves in depth patient observation, interviews, and a series of cognitive tests.
Technical Overview:
JHU researchers have identified specific paternal sperm DNA methylation as a predictive biomarker of Autism risk. In a high-risk autism spectrum disorder (ASD) birth cohort, paternal sperm genome-wide DNA methylation patterns are associated with 12-month ASD-related phenotypes. Observations were validated in a partially independent set of fathers using a second method of DNA methylation assessment. Regions of altered sperm DNA methylation in fathers associated with infant ASD phenotypes included overlapping genes previously associated with Prader-Willi syndrome. A significant subset of sperm DMRs associated with ASD phenotypes showed directionally consistent methylation changes in postmortem cerebellar tissue of ASD patients compared to controls.
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