**2021 Autism Summary**

**January 2021**

**Publication**

Garrido N, Cruz F, Egea RR, Simon C, Sadler-Riggleman I, Beck D, Nilsson E, Ben Maamar M, Skinner MK. Sperm DNA methylation epimutation biomarker for paternal offspring autism susceptibility. Clin Epigenetics. 2021 Jan 7;13(1):6. doi: 10.1186/s13148-020-00995-2.

**Summary**

Autism spectrum disorder (ASD) has increased over tenfold over the past several decades and appears predominantly associated with paternal transmission. Although genetics is anticipated to be a component of ASD etiology, environmental epigenetics is now also thought to be an important factor. Epigenetic alterations, such as DNA methylation, have been correlated with ASD. The current study was designed to identify a DNA methylation signature in sperm as a potential biomarker to identify paternal offspring autism susceptibility. Sperm samples were obtained from fathers that have children with or without autism, and the sperm then assessed for alterations in DNA methylation. A genome-wide analysis (> 90%) for differential DNA methylation regions (DMRs) was used to identify DMRs in the sperm of fathers (n = 13) with autistic children in comparison with those (n = 13) without ASD children. The 805 DMR genomic features such as chromosomal location, CpG density and length of the DMRs were characterized. Genes associated with the DMRs were identified and found to be linked to previously known ASD genes, as well as other neurobiology-related genes. The potential sperm DMR biomarkers/diagnostic was validated with blinded test sets (n = 8-10) of individuals with an approximately 90% accuracy. Observations demonstrate a highly significant set of 805 DMRs in sperm that can potentially act as a biomarker for paternal offspring autism susceptibility. Ancestral or early-life paternal exposures that alter germline epigenetics are anticipated to be a molecular component of ASD etiology.