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Commentary

Variants of Mitochondrial DNA in Autism Spectrum Disorder

Yongcheng Yao*

Department of Psychiatry, Nankai University, Weijin, Nankai, China

ABOUT THE STUDY

Autism Spectrum Disorder (ASD) is a neurodevelopmental disease that is characterised by stereotyped behaviour patterns and the impairment of socio-communicative abilities. The most recent estimates place the frequency of ASD at roughly 1/54 of children in the United States, a significant increase of 10 to 17 percent every year over the past two decades (US). The significant rise in the frequency of ASD has sparked interest in the condition's aetiology on a global scale, which primarily focuses on the influence of environmental variables on genetic predispositions involving both nuclear DNA (nDNA) and mitochondrial DNA (mtDNA). No trustworthy biomarker or distinctive aetiology for ASD has yet been found, despite significant efforts to comprehend its etiological process and biological underpinnings.

Growing evidence from genetic, post-mortem, neuroimaging, and biomarker studies in both ASD subjects and animal models supports Lombard's initial hypothesis that ASD may be a disorder of impaired mitochondrial function, made in 1998. These studies also suggest that mitochondrial physiology may be involved in the pathophysiological pathway of ASD. Although this is still somewhat debatable, accumulating data suggests that oxidative stress and aberrant energy metabolism may both play a role in the aetiology of ASD. This is consistent with some degree of mitochondrial dysfunction. A strong case can be made for looking into the role of mtDNA in the aetiology of ASD given the genetic complexity of the disorder and its link to mitochondrial malfunction.

Energy metabolism in eukaryotic cells is carried out by the double-membraned organelles known as mitochondria, which are regulated by both nuclear DNA (nDNA) and mitochondrial DNA (mtDNA). The brain is the tissue in the human body that requires the most energy, and both the number of mitochondria and the amount of mtDNA in each mitochondria vary depending on the bioenergetic requirements of a particular cell and the levels of oxidative cellular stress.

The human mtDNA is a double-stranded supercoiled ring molecule, which exclusively inherits from the matrilineal line. The 15,000-17,000 base pair human mtDNA codes for 37 genes that produce the 22 tRNAs, 2 rRNAs, and 13 Polypeptides involved in Oxidative Phosphorylation (OXPHOS). In contrast to nDNA, mtDNA exhibits a very high rate of variations because to the absence of protective histones, a less effective system for DNA repair, and a high amount of Reactive Oxygen Species (ROS) formation in this organelle. The amount of oxidative stress and the fidelity of the mtDNA polymerase are the key determinants of the rate of mtDNA variations. Three categories of mtDNA variants-recent harmful mutations, ancient adaptive variants, and *de novo* mutations or somatic mutations-are clinically significant.

The significance of mitochondria and mtDNA in human health and disease was discussed, followed by a systematic review of the evidence pointing to the involvement of mtDNA variants in the emergence of autism spectrum disorder. Finally, we covered possible explanations for these results and the limitations of the available data. mtDNA variations are involved in the underlying pathophysiology of ASD, notwithstanding the ongoing debate over whether or not they actually cause the disorder or are merely an epiphenomenon. The emergence of clinical diagnostic biomarkers and novel therapy options that address the underlying genetic aetiology of this condition will be fueled by the growing awareness of the role of mtDNA variations in the development of ASD.

Correspondence to: Yongcheng Yao, Department of Psychiatry, Nankai University, Weijin, Nankai, China, E-mail: yaoyong@nku.cn

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