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Size matters in gray matter: The neuroanatomy of autism, fragile X and Williams syndromes

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Enlarged head circumference (macrocephaly) and enlarged brain volume (macroencephaly) are the most consistent neurobiological findings in autism. Conversely, genetic, behavioral, and regional specific brain structural/functional neuroimaging findings are widely inconsistent. Finding a consistency within the inconsistencies reasonably warrant further discussion. Indeed, the autism phenotype is associated with an excess of brain volume due in part to decrease pruning during development. Here, we aimed at assessing brain volume early in development to further elucidate previous findings in autism and determine whether this pattern is restricted to idiopathic autism or shared within the autistic phenotype (fragile X syndrome [FXS]). We investigated brain volume in 37 participants, using the fully automated Civet pipeline anatomical magnetic resonance imaging. Three groups with intellectual deficiency: Autism (AUT); its most associated FXS; and its most opposite Williams syndrome (WS) were compared with each other and with normal controls (NC). We report increased total and regional gray and white matter brain volume in AUT and FXS relative to WS and NC. These findings are discussed in light of the possibilities leading for the enlarged brain volume in children with the AUT phenotype. We speculate that this excess suggests reduced regression of neuronal processes "pruning" in cortical and subcortical regions in AUT/FXS, which may be due to a mutation in specific genes involved in pruning and/or a lack of socio-emotional environmental experience during a critical developmental period. The research highlights are as follows: 1. Children with autism show increased brain gray and white matter volume; 2. Children with FXS with the autism behavioral phenotype show increased brain volume; 3. Children with Williams syndrome with no autism display decreased brain volume; 4. Increased brain volume is associated with the autism phenotype in children.

Biography

Cherine Fahim is an Associate Researcher at the Department of Psychiatry, University of Montreal, Canada and a Lecturer at the Department of Biomedical Sciences, University of Fribourg, Switzerland. She is the Founder of Endoxa Neuroscience in Neuchâtel, Switzerland. Her research focuses on Brain Development and Mental Health. She is also very interested in maximizing the impact of neuroscience research. She believes that bridging between neuroscience, the general public, universities, socio-medical institutions, hospitals, schools, museums and the industry is the most effective way to achieve the greatest impact to the advantage of the community and the world.

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