Examining the Frontal Subcortical Brain Vulnerability Hypothesis in Children With Neurofibromatosis Type 1: Are T2-Weighted Hyperintensities Related to Executive Dysfunction?

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Résumé en anglais	Objective: It was hypothesized that neuropsychological impairments in children with neurofibromatosis type I (NF1) are associated with brain areas of increased T2-weighted signal intensity on MRI. Systematic and extensive examination of this hypothesis remains however scarce, particularly regarding executive dysfunction whereas hyperintensities are located preferentially in frontal-sub-cortical networks. In this study, we compared the executive functioning profile with characteristics of brain hyperintensities in children with NF1. Method: A sample of 36 school-age children with NF1 (7-12 years) underwent a detailed examination of executive function, including performance-based tests and child's behavior rating in daily life. Executive function measures were compared with the characteristics of the T2-weighted hyperintensities on parallel MRI scans. The presence, number, and size of hyperintensities in the whole brain were considered as well as their main cerebral locations. Results: Executive dysfunction including traditional cognitive and ecological measures in children with NF1 is not significantly influenced by T2-weighted hyperintensities, in terms of presence or not, number, size, and location, whether in the whole brain or according to involved specific brain areas. Conclusion: T2-weighted hyperintensities, as they are currently measured, cannot be used as a strong indicator of executive dysfunction in children with NF1. Based on the available NF1 cognitive impairment pathogenesis models, a critical discussion on anatomical-functional relationships between hyperintensities and neuropsychological profile is proposed, especially the executive dysfunction. (PsycINFO Database Record (c) 2014 APA, all rights reserved).
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