

Decreased Microstructural Integrity of White Matter in PKU: a DTI Study

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Abstract

Introduction: Phenylketonuria (PKU) is an autosomal recessive disorder characterized by defective metabolism of the amino acid phenylalanine (Phe) that eventually leads to a deficiency of dopamine. PKU is associated with cognitive deficits and recent studies have identified brain abnormalities in PKU patients, especially in white matter. Diffusion tensor imaging (DTI) allows for the evaluation of the microstructural integrity of white matter tracts in the brain. Two DTI measures are usually reported: the apparent diffusion coefficient (ADC) and fractional anisotropy (FA). ADC is a measure of the rate of water diffusion, while FA is a measure of the asymmetry of water diffusion. The purpose of this study is to investigate the integrity of white matter in the brain of children suffering from PKU as a possible reason for the cognition impairments seen in this disorder. **Methods:** Children with PKU and age- and gender-matched controls were recruited and scanned with a 1.5T scanner. Images acquired include T1-weighted sagittal, magnetization prepared rapid gradient echoes, T2-weighted fast spin echoes, and DTI images. The scans were then registered to the same atlas to allow for comparison. Lastly, the program Analyze was used to calculate the ADC and FA values for 14 different areas of interest in the brain. **Results:** Using rigorous statistical testing, the FA and ADC for each area of interest when compared between the PKU and control groups. No area of interest showed a difference in FA between the PKU and control groups, while ten of the fourteen measured areas showed a decrease of ADC in the PKU subjects. **Conclusion:** The observed results imply that while the water molecules in the brain move along white matter tracts as one would expect, they move much more slowly in the brains of PKU subjects. One hypothesis for this phenomenon is that debris from the inadequate metabolism of Phe collects in the white matter slowing diffusivity. This and other interesting aspects of PKU will be further studied with the continuation of this project.

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