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Funding Source: Thompson Center Undergraduate Research Opportunity

Cerebellar white matter abnormalities in phenylketonuria (PKU)

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Phenylketonuria (PKU) is a recessive genetic disorder that is characterized by an individual's body being unable to utilize the amino acid phenylalanine because of a dysfunction with the enzyme phenylalanine hydroxylase. Previous studies have shown that the brains of individuals with PKU are decreased in overall size. Additionally, abnormalities of white matter have been previously documented in the cerebrum and periventricular areas. A study of rats with induced hyperphenylalaninemia (i.e., an excess of phenylalanine in the blood) showed cellular abnormalities in the cerebellum (Hogan and Coleman, 1981). We hypothesize that individuals with PKU will show abnormalities in measures of the cerebellum relative to typically developing individuals. The data presented here are from a pilot study examining white matter in the cerebella of individuals with PKU. We collected magnetic resonance images (MRIs) of 1mm³ voxel resolution from four individuals between 12-20 years of age. Overall cerebellar volume and cerebellar white matter volume data were collected using Analyze 8.1 and were then compared between controls and individuals with PKU. The small sample size prohibits statistical analysis so we present trends in our comparative data. Results suggest that overall cerebellar volume and cerebellar white matter volume are decreased in individuals with PKU. Future analysis of larger samples will determine whether these trends in cerebellar abnormalities in individuals with PKU are significant.