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Cortical Atrophy Evidenced by Increased Extra-Axial Cerebrospinal Fluid in individuals with Early-Treated Phenylketonuria

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Main Purpose: Phenylketonuria (PKU) is a rare autosomal recessive disorder characterized by a deficiency in the inability to metabolize the amino acid phenylalanine. Even if treated, individuals with PKU are at risk for neurologic problems (e.g., white matter abnormalities, disruptions in protein synthesis). The majority of past research has focused on children with PKU. Much less is known regarding neurologic outcomes in adults with PKU. In the present study, we examined whether adults with early-treated PKU (ETPKU) experienced cortical atrophy as evidenced by an increased volume of extra axial-cerebrospinal fluid.

Procedure: A sample of 36 individuals with early-treated PKU (18-35 years of age) and a demographically-matched group of 32 individuals without PKU underwent structural MRI scanning in a 3T Siemens Trio Scanner. Data processing is presently underway, and the examiner (B.C.) is blind to the group status of participants. First the MRI data is being processed through SPM12 software in MatLab to acquire 3D masks of cerebrospinal fluid tissue segmentations for each of the individuals. Next, Freesurfer software will be used to manually edit the masks to remove any non-cerebrospinal fluid that SPM12 did not remove. After the editing of the cerebrospinal fluid tissue segmentations is finished, the brains will then be processed through Freesurfer for a statistical analysis on the voxels.

Anticipated Findings: Data processing is still underway. Resulting data will be analyzed using a standard random effects general linear model (GLM) approach. Based on previous findings of cortical atrophy in ETPKU, we anticipate increased volumes of extra-axial cerebrospinal fluid in adults with ETPKU compared to the adults without ETPKU.