Main Purpose: While the effects of early-treated phenylketonuria (ETPKU) on white matter have been heavily researched, the impact of this metabolic disorder on gray matter structures of the brain, such as the basal ganglia, remains largely unaddressed. A past study by Bodner (2012) found significantly larger putamen volumes for a sample of individuals with ETPKU as compared to a non-PKU group. They also found a significant correlation between the degree of enlargement in the putamen, blood phenylalanine levels, and full scale IQ in the ETPKU group. The study aims to extend this research and examine potential volumetric differences of the basal ganglia (i.e., caudate nucleus, putamen, and nucleus accumbens) in a much larger sample of individuals with and without PKU.

Procedure: The study used a 3T Siemens Trio MRI scanner with a standard 8-channel head coil to obtain high-resolution (1 mm3) T-1 weighted structural images of the brain. The structural MRI data for each participant was rotated into AC–PC coordinates, and the skull and dural tissue were removed from the images using a semi-automated object extraction procedure in Free Surfer. Whole brain volume (WBV) for each participant was calculated, and the gray matter structures of interest (i.e., caudate nucleus, putamen, and nucleus accumbens) were manually segmented in a standardized fashion to obtain precise volumetric measurements.

Results and Conclusions: Data for the study was processed and analyzed using a hierarchical linear regression model. This method of analysis would allow for evaluation of potential group-related differences in basal ganglia volume while controlling for individual differences in WBV and age. For the regression model, age and whole brain volume were entered in the first step followed by group (PKU and non-PKU) in the second step. Interaction terms (e.g., age x group) were included in the third and final step of the model.