Cortical Atrophy Evidenced by Increased Extra-Axial Cerebrospinal Fluid in Individuals with Early-Treated Phenylketonuria: Preliminary Results

B.D. CARMAN, A. BROWN, S.E. CHRIST

Phenylketonuria (PKU) Background

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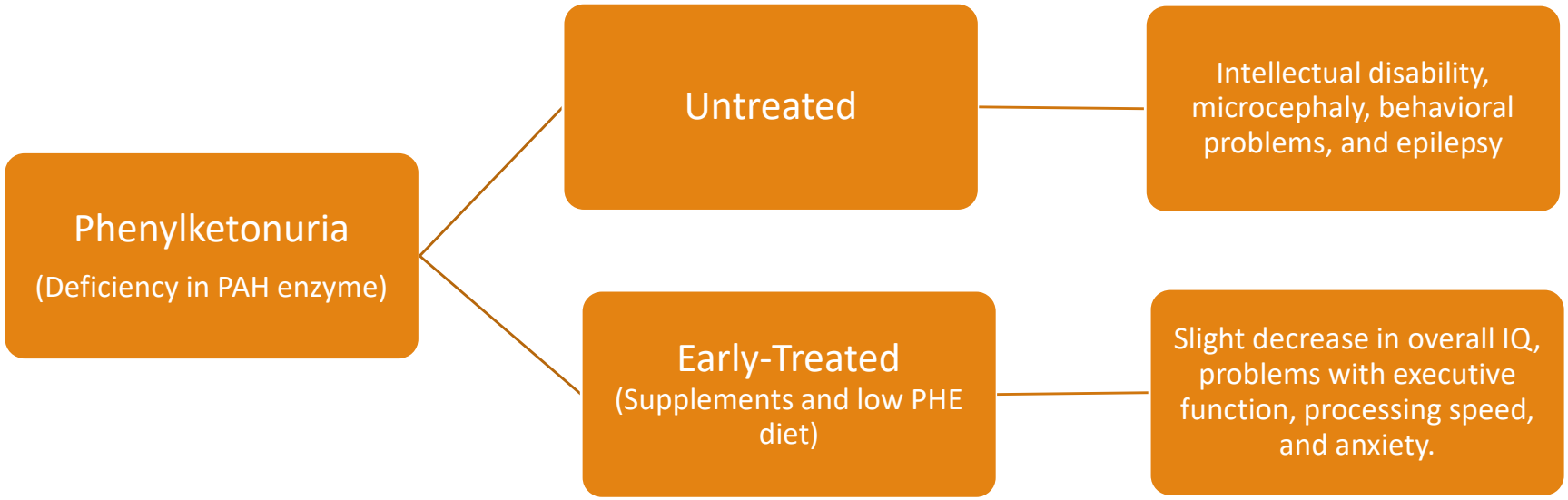


EACSF & cortical atrophy background

Extra-Axial cerebrospinal fluid

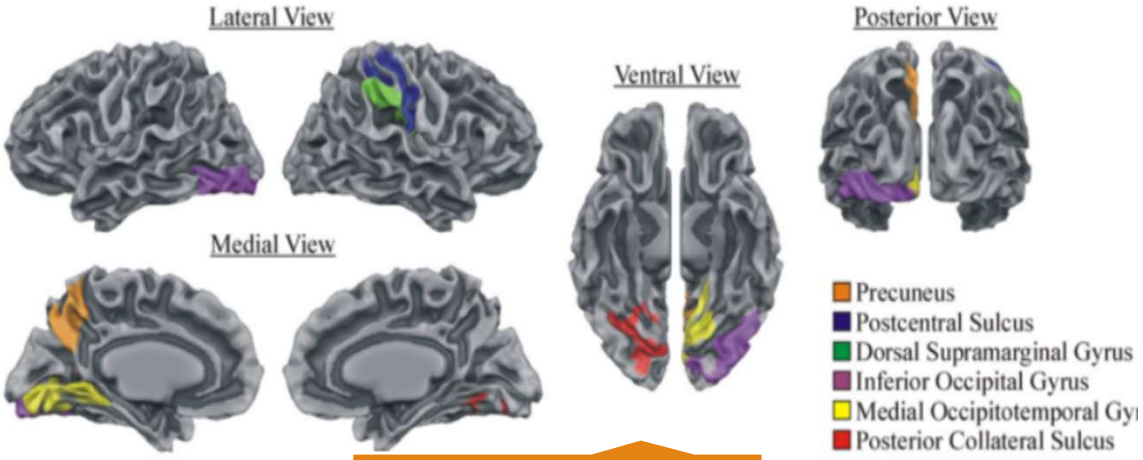


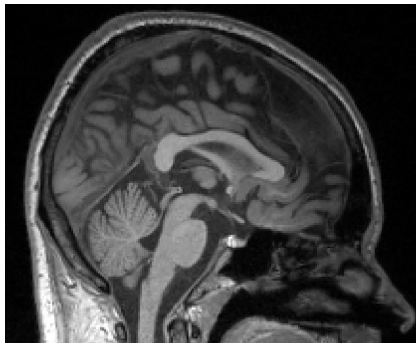
Fig. 3. Illustration of cor ...KU < non-PKU) in GM volume.

Cortical atrophy in ETPKU

Image derived from Christ, S. E., Price, M. H., Bodner, K. E., Saville, C., Moffitt, A. J., & Peck, D. (2016). Morphometric analysis of gray matter integrity in individuals with early-treated phenylketonuria. *Molecular genetics and metabolism*, *118*(1), 3–8. https://doi.org/10.1016/j.ymgme.2016.02.004

Methods

T1 weighted MRI scans → Processed through SPM12 to derive 3D cerebrospinal fluid tissue segmentation → CSF tissue segmentation and the T1 weighted image were aligned at the ACPC line and cropped → Cerebrospinal tissue segmentation were then cropped and manually corrected through FreeSurfer



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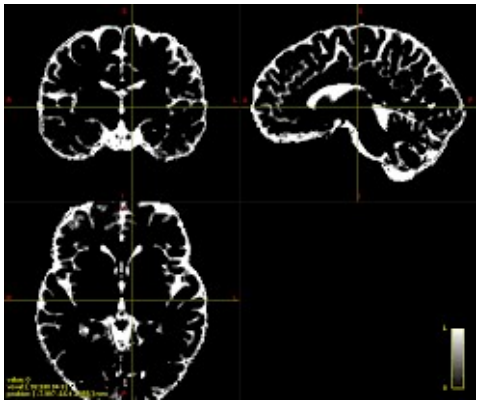

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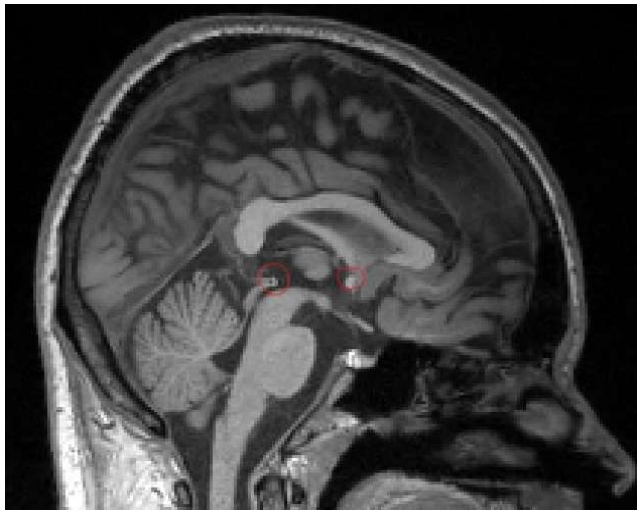

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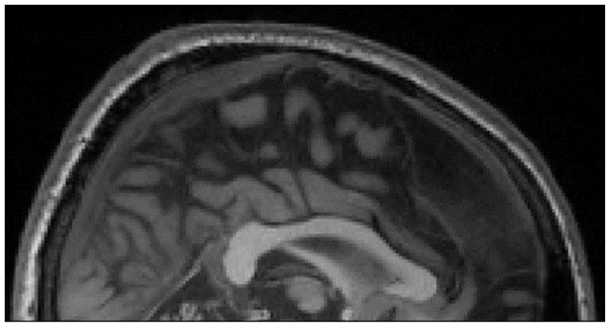

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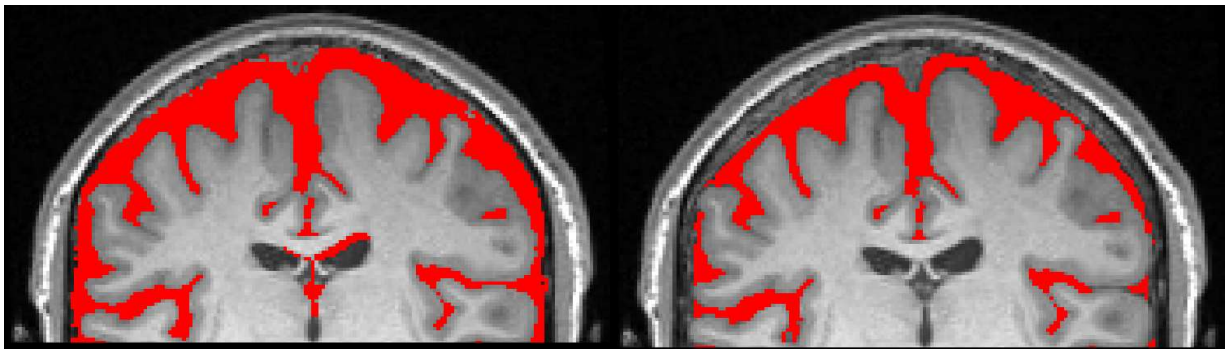

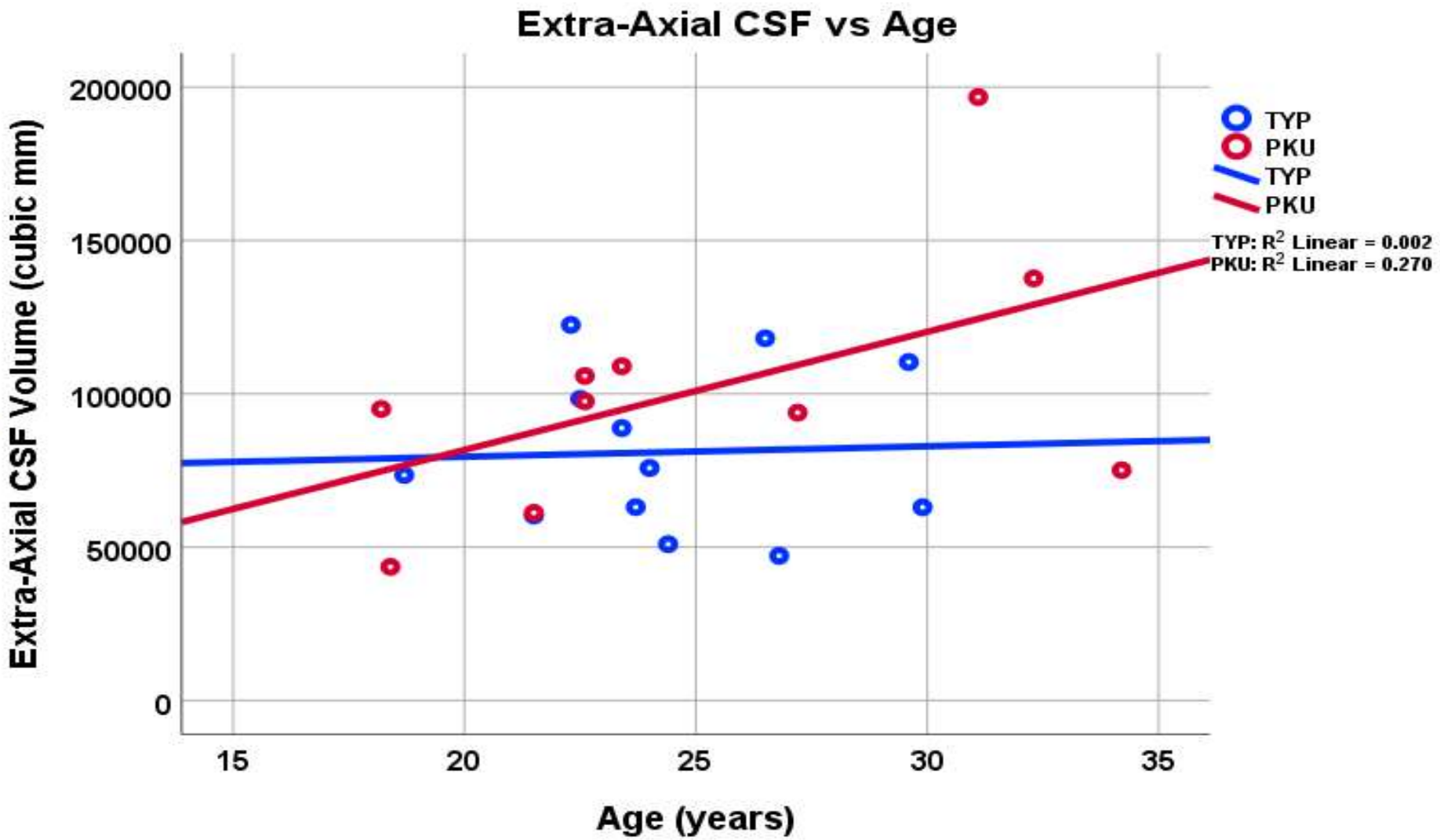
Extra-Axial CSF vs Age
Extra-Axial CSF Volume (cubic mm)
200000
150000
100000
50000
0
15
20
25
30
35
Age (years)
TYP
PKU
TYP
PKU
TYP: R2 Linear = 0.002
PKU: R2 Linear = 0.270

Future directions

Finish processing the current data

Localizing volumetric differences via surface-based analysis

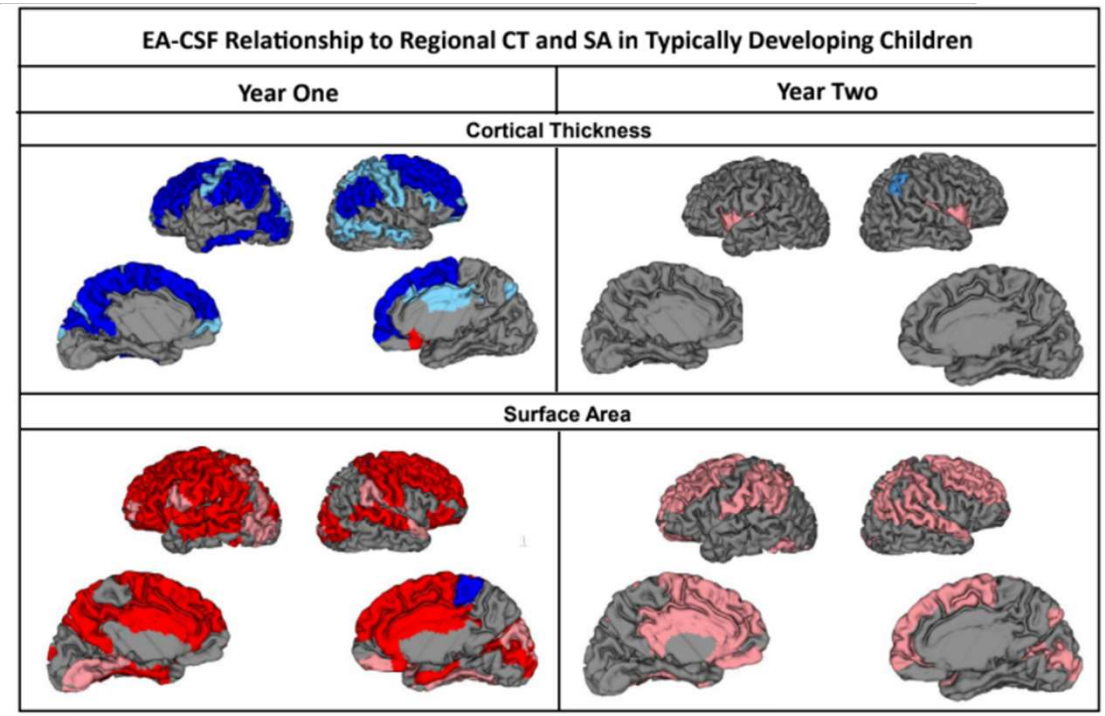

Figure 3. Regional relationships of extra-axial cerebrospinal fluid (EA-CSF) volume to cortical thickness (CT) and surface area (SA) at 1 and 2 years of age. Pink represents a significant positive association, red is an association significant after false discovery rate correction. Light blue represents a significant negative association, dark blue is an association significant after false discovery rate correction.

Image derived from Murphy, V. A., Shen, M. D., Kim, S. H., Cornea, E., Styner, M., & Gilmore, J. H. (2020). Extra-axial Cerebrospinal Fluid Relationships to Infant Brain Structure, Cognitive Development, and Risk for Schizophrenia. *Biological psychiatry. Cognitive neuroscience and neuroimaging*, *5*(7), 651–659. https://doi.org/10.1016/j.bpsc.2020.03.008

Thank You

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