



CLINICAL HISTORY: 6 wk old, term, developmental delay, microcephaly, seizures

FINDINGS:

BRAIN PARENCHYMA: No acute infarct or hemorrhage. Markedly enlarged caudate with absent internal capsule, resulting in fused striata bilaterally.

CORTEX: Bifrontal band heterotopias are seen as well as bioccipital pachygyria.

SULCATION: Delayed for age.

WHITE MATTER: Reduced volume. Myelination is delayed for age.

CORPUS CALLOSUM: Thinned.

VENTRICLES: Enlarged.

CEREBELLUM: Hypoplastic.

VERMIS: Hypoplastic.

BRAINSTEM: Hypoplastic pons.

MRS:

NAA: choline ratio: Normal.

Lactate Peak: Absent.

IMPRESSION:

Combination of abnormal basal ganglia, malformations of cortical development, microcephaly, and a small cerebellum/brainstem are compatible with a tublinopathy.

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