Tuberous Sclerosis - Newborn
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History
Newborn with cardiac rhabdomyomas.

Diagnosis
Tuberous Sclerosis

Discussion
Tuberous sclerosis is a phakomatosis with dysplasias and hamartomas frequently affecting the brain, eyes, kidneys, heart, and skin. It may be transmitted as an autosomal dominant trait with variable penetrance although 60% to 70% of cases occur sporadically. Three different mutations have been associated with the disorder, located on chromosomes 9, 11, and 16. The mean age of diagnosis is 5 years.

Overall, epilepsy and mental deficits are the most common neurologic problems and tend to be more severe if manifested early. Cardiac rhabdomyomas, renal cysts and angiomyelolipomas, sclerotic bone lesions, hamartomas of the retina and optic discs, and aneurysms.

MR imaging reveals hamartomas of tuberous sclerosis are hyperintense relative to immature white matter on T1 (and hypointense on T2) in the newborn period and follow the migratory pathways of neurons and glia. White matter anomalies, subependymal nodules, and SGCA seem to be detected better in neonates probably related to the incomplete myelination at this age. Although detection of cortical tubers is more difficult than it is after myelination is complete, T1 hyperintensity makes tubers that extend into the subcortical white matter quite conspicuous against the hypointense unmyelinated white matter. The signal intensities of gray and white matter start to reverse at approximately 6 months of age.

Findings
MR-Numerous subependymal T1 hyper intensities in a radial ventriculofugal pattern of the white matter.

Reference
Baron Y and Barkovich AJ. MR Imaging of Tuberous Sclerosis in Neonates and Young Infants. AJNR (1999); 20: 907-916.
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