Bourneville-Pringle Syndrome Detected at the Age of Two Days

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A case of tuberous sclerosis in a two-day-old infant is described. A retinal tumor of the left eye was detected postnatally by ophthalmoscopy and verified by echography. Clinical examinations revealed hamartomas of both eyes, brain and heart, pointing to the diagnosis of a tuberous sclerosis. Retinal phakomas were grey and translucent, with an internal hyalinization. No changes were observed during a one year period of the follow-up. Due to brain hamartomas, epileptic seizures occurred on several occasions, and a continuous antiepileptic therapy was prescribed. This case of tuberous sclerosis detected in a two-day-old newborn is the earliest one reported so far. The occurrence of tuberous sclerosis at such a young age suggests that retinal hamartomas may be present during intrauterine life.

Key words: hamartoma; newborn; tuberous sclerosis

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