Teaching NeuroImages: Huge carotid artery aneurysm in *TSC2/PKD1* contiguous gene syndrome

A 1-day-old infant developed heart murmur with tachypnea. Echocardiography showed multiple biventricular cardiac rhabdomyomas (figure, A). Brain sonography showed multiple cerebral cortical tubers (figure, B) and subependymal nodules (figure, D and E), confirming tuberous sclerosis. Bilateral renal cysts were present (figure, C).

(A) Intra-cardiac rhabdomyomas. (B) Multifocal bilateral hyperechoic cortical tubers. (C) Bilateral renal cysts. (D, E) Subependymal hamartomas along the lateral ventricles and multifocal cortical tubers. (F, G) Huge left internal carotid aneurysm, mainly at the cavernous portion. (H, I) Large heterozygous deletions cause TSC2/PKD1 contiguous gene syndrome.
When the infant was 21 months old, brain MRI showed multifocal subependymal hamartomas, cortical tubers, and a huge left internal carotid artery aneurysm (figure, F and G).

The TSC2 gene lies adjacent to the PKD1 gene on chromosome 16p13.3. Large heterozygous deletions cause TSC2/PKD1 contiguous gene syndrome (figure, H and I), resulting in tuberous sclerosis and polycystic kidney disease\(^1\) simultaneously and contributing the unruptured intracranial aneurysm.\(^2\)

**AUTHOR CONTRIBUTIONS**

Y.-F.T. and Y.-S.T. participated in the neuroimaging interpretations and clinical care of the patient. The manuscript was drafted by B.W. and Y.-S.T.

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**REFERENCES**

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