Figure. (A) Neurologic examination after the seizure was unrevealing; physical examinations revealed several venous malformations on the face and on the trunk. These were bluish nipplelike lesions, which could be easily compressed. (B) Axial T2 gradient echo MRI scan of the brain demonstrating venous malformations.

Blue rubber bleb nevus syndrome

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A 54-year-old man presented with a first generalized tonic-clonic seizure due to intracerebral venous malformations in blue rubber bleb nevus syndrome (BRBNS) (figure). Differential diagnosis included hereditary hemorrhagic telangiectasia (punctiform—typically subungual—angioma, epistaxis, and affected first-

degree relative) and Maffucci syndrome (presence of enchondromatosis). Besides skin malformations, patients with BRBNS have frequent malformations in the gastrointestinal system, which can bleed. Few BRBNS cases with CNS involvement have been described. Patients had focal seizures¹ or neurologic symptoms due to compression.² Due to its rarity, it is unclear whether cerebral venous malformations lead to hemorrhage. We treated our patient with carbamazepine; he remained seizure free after 1 year of follow-up.

BRBNS cases are mostly sporadic; familial cases have been described with linkage to chromosome 9p. The receptor tyrosine kinase TIE2 lies on this gene, which is implied in vascular morphogenesis.

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