



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

Tom den Heijer, MD, PhD; Agnita J.W. Boon, MD, PhD, Rotterdam, the Netherlands Copyright © 2007 by AAN Enterprises, Inc.

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.

Disclosure: The authors report no conflicts of interest.

Address correspondence and reprint requests to Dr. A.J.W. Boon, Department of Neurology, Erasmus MC, PO Box 2040, 3000 CA, Rotterdam, the Netherlands; e-mail: t.denheijer@erasmusmc.nl or a.j.w.boon@erasmusmc.nl

1. Kim SJ. Blue rubber bleb nevus syndrome with central nervous system involvement. *Pediatr Neurol* 2000;22:410–412.
2. Satya-Murti S, Navada S, Eames F. Central nervous system involvement in blue-rubber-bleb-nevus syndrome. *Arch Neurol* 1986;43:1184–1186.

Neurology[®]

Blue rubber bleb nevus syndrome
Tom den Heijer and Agnita J.W. Boon
Neurology 2007;68;1075
DOI 10.1212/01.wnl.0000258547.31946.a3

This information is current as of March 26, 2007

Updated Information & Services	including high resolution figures, can be found at: http://n.neurology.org/content/68/13/1075.full
References	This article cites 2 articles, 0 of which you can access for free at: http://n.neurology.org/content/68/13/1075.full#ref-list-1
Subspecialty Collections	This article, along with others on similar topics, appears in the following collection(s): MRI http://n.neurology.org/cgi/collection/mri Other neurocutaneous disorders http://n.neurology.org/cgi/collection/other_neurocutaneous_disorders
Permissions & Licensing	Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: http://www.neurology.org/about/about_the_journal#permissions
Reprints	Information about ordering reprints can be found online: http://n.neurology.org/subscribers/advertise

Neurology® is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright . All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.

