

Circulation

JOURNAL OF THE AMERICAN HEART ASSOCIATION



Skin Manifestations, Multiple Aneurysms, and Carotid-Cavernous Fistula in Ehlers-Danlos Syndrome Type IV

Jay-Hyun Koh, Joo Sung Kim, Seung-Chyul Hong, Yeon Hyeon Choe, Young Soo Do, Hong Sik Byun, Won Ro Lee and Duk-Kyung Kim

Circulation 1999, 100:e57-e58

doi: 10.1161/01.CIR.100.13.e57

Circulation is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75214

Copyright © 1999 American Heart Association. All rights reserved. Print ISSN: 0009-7322. Online ISSN: 1524-4539

The online version of this article, along with updated information and services, is located on the World Wide Web at:

<http://circ.ahajournals.org/content/100/13/e57>

Subscriptions: Information about subscribing to *Circulation* is online at
<http://circ.ahajournals.org/subscriptions/>

Permissions: Permissions & Rights Desk, Lippincott Williams & Wilkins, a division of Wolters Kluwer Health, 351 West Camden Street, Baltimore, MD 21202-2436. Phone: 410-528-4050. Fax: 410-528-8550. E-mail:
journalpermissions@lww.com

Reprints: Information about reprints can be found online at
<http://www.lww.com/reprints>

Skin Manifestations, Multiple Aneurysms, and Carotid-Cavernous Fistula in Ehlers-Danlos Syndrome Type IV

Jay-Hyun Koh, MD; Joo Sung Kim, MD; Seung-Chyul Hong, MD, PhD;
Yeon Hyeon Choe, MD, PhD; Young Soo Do, MD, PhD; Hong Sik Byun, MD, PhD;
Won Ro Lee, MD, PhD; Duk-Kyung Kim, MD, PhD

A 21-year-old woman presented with left ocular pain and severe pulsatile headache. She had suffered from easy bruisability since she was young. On physical examination, ecchymoses and bruises were noticed on trauma sites (Figure 1A). The skin was so thin that subcutaneous blood vessels were visible (Figure 1B). Huge hematomas and ecchymoses occurred at the puncture sites after angiography (Figure 1C). There was no significant hyperelasticity of the skin. Mild hypermobility of the joints in the hands was observed.

Angiography showed a large carotid-cavernous fistula and an aneurysm at the cervical portion of the left internal carotid artery (Figure 2A). Abdominal aortography disclosed a large ovoid aneurysm of the left renal artery with delayed nephrogram (Figure 2B). The carotid-cavernous fistula was successfully occluded by detachable balloons.

This case demonstrates characteristic clinical features of type IV Ehlers-Danlos syndrome, an autosomal dominant disorder resulting from mutations in the *COL3A1* gene. The gene defects alter the metabolism of type III collagen, a major constituent of the walls of blood vessels.¹ Type IV Ehlers-Danlos syndrome is life-threatening because of its vascular manifestations, which are characterized by rupture, dissection, or aneurysm formation affecting large or medium-sized arteries.²

References

1. North KN, Whiteman DA, Pepin MG, Byers PH. Cerebrovascular complications in Ehlers-Danlos syndrome type IV. *Ann Neurol*. 1995;38:960-964.
2. Witz M, Lehmann JM. Aneurysmal arterial disease in a patient with Ehlers-Danlos syndrome: case report and literature review. *J Cardiovasc Surg (Torino)*. 1997;38:161-163.

From the Departments of Medicine (J.-H.K., J.S.K., W.R.L., D.K.), Neurosurgery (S.H.), and Radiology (Y.H.C., Y.S.D., H.S.B.), Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Korea.

Correspondence to Duk-Kyung Kim, MD, PhD, Cardiovascular Institute, Samsung Medical Center, Sungkyunkwan University School of Medicine, 50 Ilwon-dong, Kangnam-Ku, Seoul 135-230, Korea. E-mail dkkim@smc.samsung.co.kr

The editor of Images in Cardiovascular Medicine is Hugh A. McAllister, Jr, MD, Chief, Department of Pathology, St Luke's Episcopal Hospital and Texas Heart Institute, and Clinical Professor of Pathology, University of Texas Medical School and Baylor College of Medicine.

Circulation encourages readers to submit cardiovascular images to Dr Hugh A. McAllister, Jr, St Luke's Episcopal Hospital and Texas Heart Institute, 6720 Bertner Ave, MC1-267, Houston, TX 77030.

(*Circulation*. 1999;100:e57-e58.)

© 1999 American Heart Association, Inc.

Circulation is available at <http://www.circulationaha.org>

Downloaded from <http://circ.ahajournals.org/> at RUTGERS UNIVERSITY on April 16, 2012

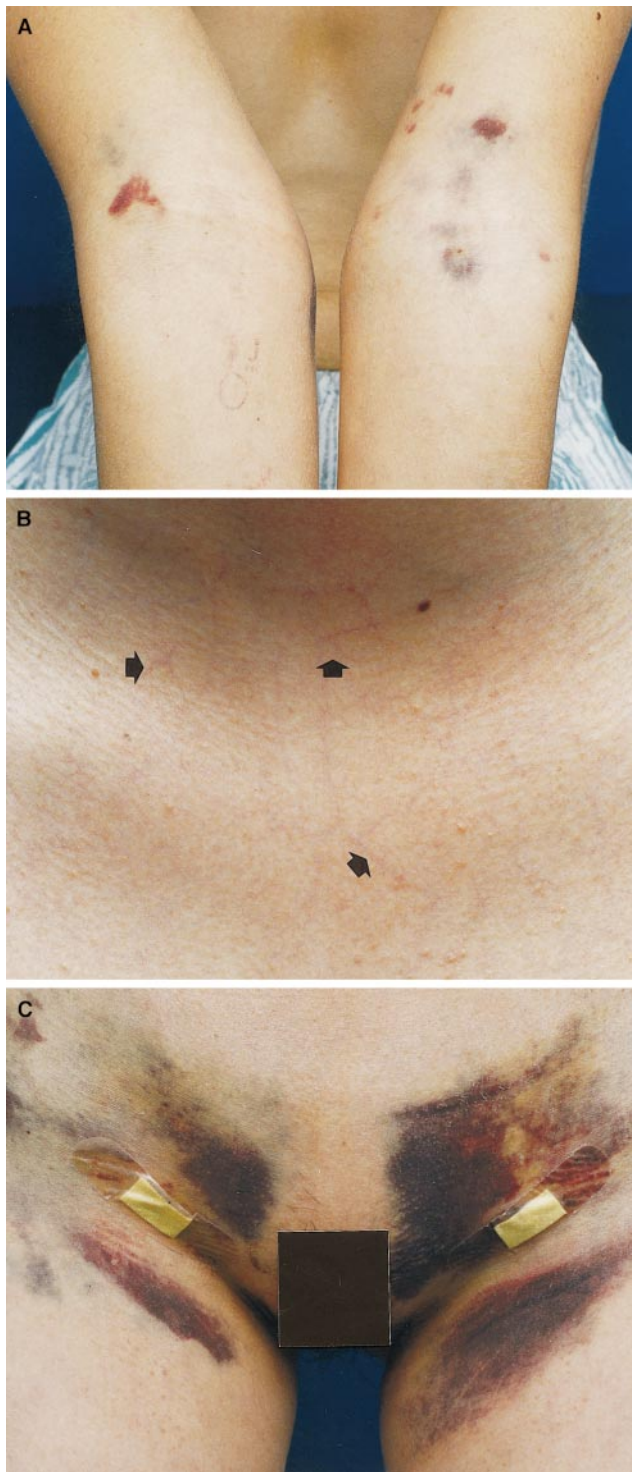


Figure 1. A, Patient's arms showed ecchymoses and bruises at needle puncture sites. B, Patient's skin over upper thorax showed translucence with visible vessels (arrows). C, Severe ecchymoses and hematomas occurred at puncture sites after transfemoral abdominal aortogram despite adequate compression.

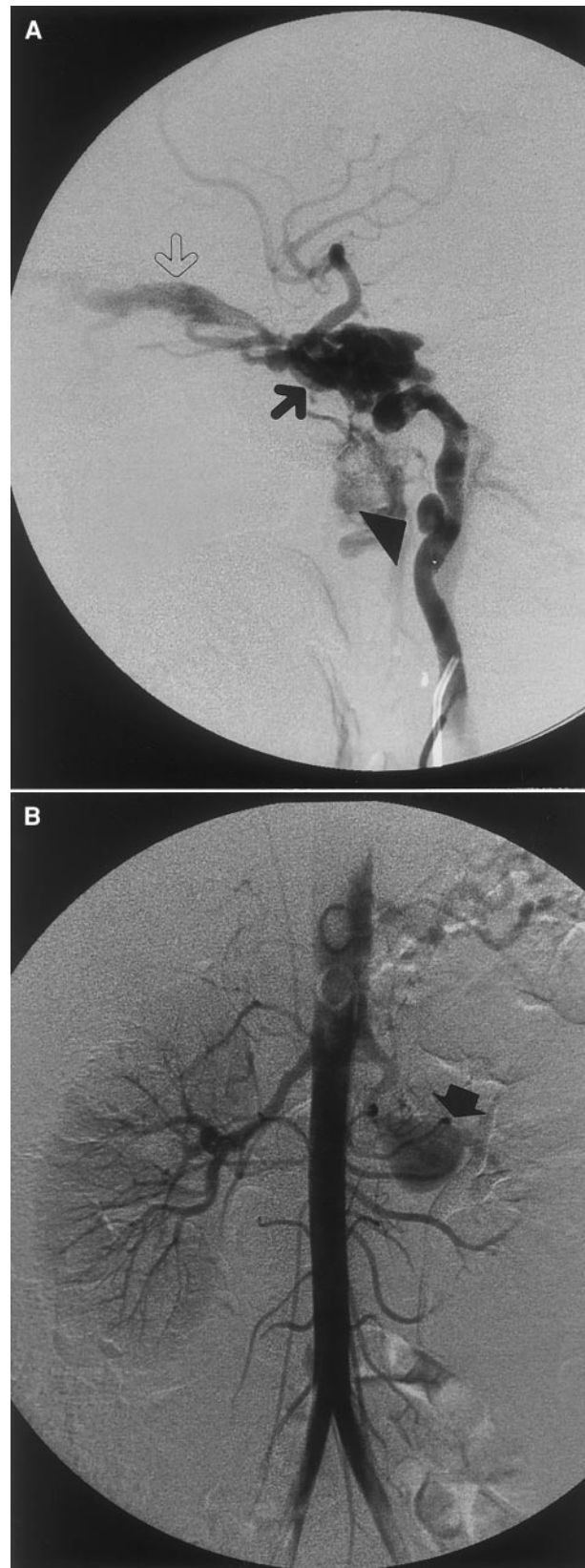


Figure 2. A, Left internal carotid angiogram disclosed a direct carotid-cavernous fistula (solid arrow) with drainage into superior ophthalmic vein (arrowhead). There is a small aneurysm at anterior wall of distal cervical internal carotid artery (open arrow). B, Abdominal aortogram showed large aneurysm (arrow) of left renal artery with delayed nephrogram.