Abdominal aortic aneurysm in a patient with occipital horn syndrome

Elina Quiroga, MD, and **Rachel Heneghan, MD,** *Seattle, Wash*

Occipital horn syndrome is a rare X-linked recessive connective tissue disorder caused by deficient copper transport. Our patient presented with abdominal pain, and a computed tomography scan demonstrated a 15-cm infrarenal abdominal aortic aneurysm and bilateral common iliac artery aneurysms. After discussion of surgical management, he wished to proceed with comfort measures only. We report the first known case of an abdominal aortic aneurysm in a patient with occipital horn syndrome. (J Vasc Surg Cases 2015;1:138-40.)

Occipital horn syndrome (OHS), previously known as X-linked cutis laxa or Ehlers-Danlos syndrome type IX, is a rare X-linked recessive connective tissue disorder caused by a mutation in the gene encoding copper-transporting adenosinetriphosphatase, alpha polypeptide (*ATP7A*), with approximately 20 cases reported in the literature.[1-4] OHS and another rare X-linked recessive connective tissue disorder, Menkes syndrome, were proven allelic in 1995, and both disorders are a result of splice-type or frameshift mutations at the *ATP7A* locus, although OHS is characterized by a milder phenotype.[1,5] Clinical features of OHS are related to the deficient activity of lysyl oxidase, a copper-dependent enzyme responsible for collagen and elastin cross-linking, and include hyperelastic skin with easy bruising, hernias, bladder diverticula, varicosities, skeletal abnormalities, and mild mental retardation.[6,7] Occipital exostoses, or occipital horns, from which the disease gets its name, are a result of calcification of the trapezius and sternocleidomastoid origin at the occipital bone.[6-8] Typical life expectancy varies in this disorder, but it is substantially longer than in patients with Menkes syndrome, with reports of patients surviving into their 30s.[3,6] Cause of death, when reported, is most commonly from sequelae of defective copper transport and subsequent toxicity. In addition, we postulate that the severity of clinical manifestations, such as bladder diverticula or rupture, cerebrovascular abnormalities, and bowel diverticula, also has an impact on survival because of the necessary operative intervention or repeated infection. Although cerebral and extremity venous varicosities have been described in the literature, at this time there is no history of case reports or series that document the presence of abdominal aortic aneurysms associated with OHS. Our patient is the first reported case and is particularly interesting as he had marked expansion of his abdominal aneurysm from 2.1 cm to 15 cm in 7 years. Consent for publication was obtained per our institutional policy.

CASE REPORT

A 45-year-old man with known OHS was transferred to our institution with new-onset worsening abdominal pain and fullness. The patient was diagnosed with OHS when he was an infant, and his case was reported by Byers et al[7] in 1980. He had several features of OHS, including bileaflet mitral valve prolapse with mitral regurgitation, bilateral inguinal hernias, hiatal hernia, ptotic right kidney, bladder diverticulum, and large bilateral lower extremity varicosities. He underwent bladder repair for bladder rupture >20 years ago and sigmoidectomy for sigmoid obstruction and perforation 11 years ago; he recovered well from both surgeries. In 2003, he was diagnosed with bilateral common iliac artery aneurysms, with the right aneurysm measuring 3.6 cm and the left 3.2 cm. In 2006, the aneurysms were stable in size, and his infrarenal aorta measured 2.1 cm in its greatest diameter (Fig 1), after which time he was lost to follow-up.

In spring of 2013, he presented with 6 hours of abdominal pain and hypotension with systolic pressures in the 70s mm Hg. The patient stated that during the last 6 months, his quality of life had significantly decreased; he had lost 30 pounds (admission weight, 59 kg; body mass index, 18.6) and was limited to ambulation around his home because of fatigue and shortness of breath. He had also developed left upper quadrant abdominal distention. Physical examination revealed a pale, cachectic man with a very tender, pulsatile abdominal mass occupying most of his abdomen and a palpable thrill over his left upper and lower quadrants. Computed tomography scan demonstrated a 15-cm infrarenal aortic abdominal aneurysm; bilateral 5-cm iliac aneurysms; multiple visceral aneurysms of the left gastric, common hepatic, and splenic arteries; and free fluid in the pelvis measuring 12 Hounsfield units, most probably reactive fluid (Fig 2).

Several discussions were held with the patient and his family regarding goals of care, surgical options for reconstruction (including open and endovascular repair), and complexity of healing due to his known connective tissue disorder. The patient was

From the Division of Vascular Surgery, Department of Surgery, University of Washington.

Author conflict of interest: none.

Reprint requests: Elina Quiroga, MD, Harborview Medical Center, University of Washington, Box 359908, 325 9th Ave, Seattle, WA 98104 (e-mail: elinaq@uw.edu).

The editors and reviewers of this article have no relevant financial relationships to disclose per the Journal policy that requires reviewers to decline review of any manuscript for which they may have a conflict of interest.

2352-667X

http://dx.doi.org/10.1016/j.jvsc.2015.03.012

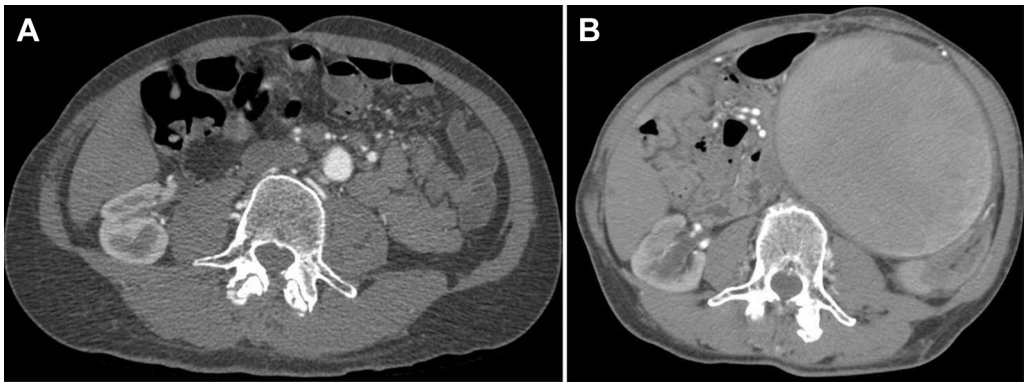

Fig 1. A, Infrarenal aorta in 2006. **B,** Infrarenal aorta in 2013.

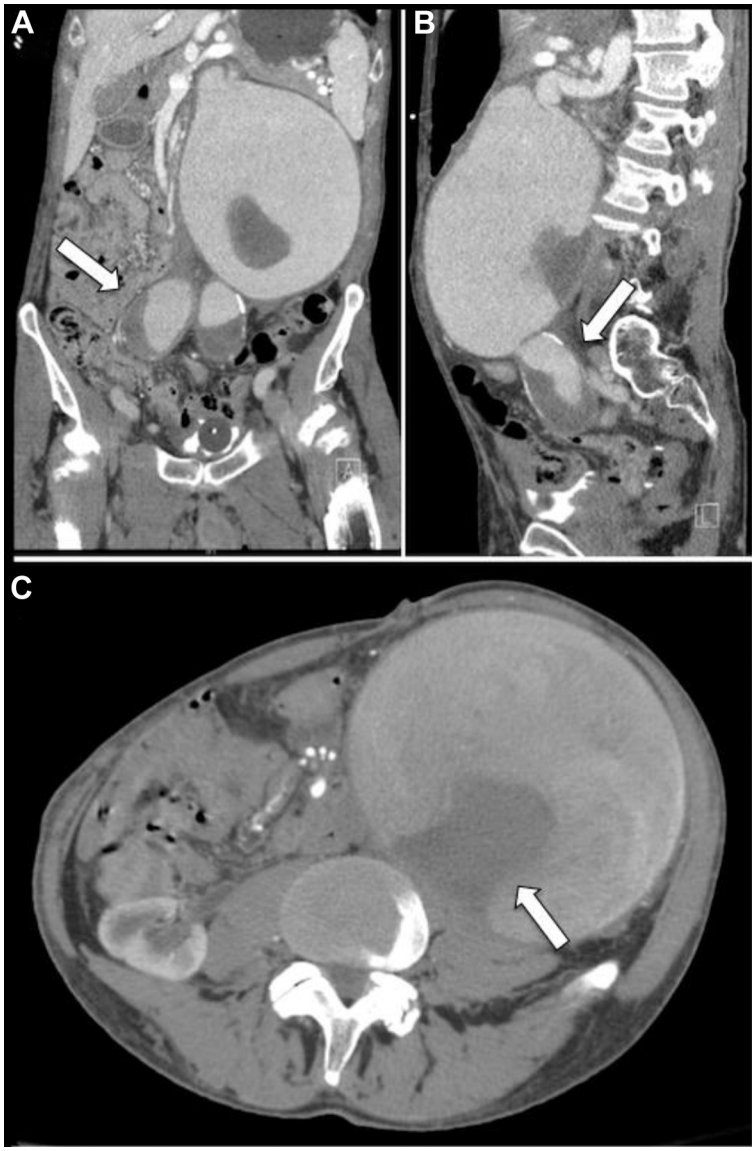

Fig 2. Giant infrarenal aortobi-iliac aneurysm. **A** and **B**, Bilateral 5-cm iliac aneurysms (*arrows*). **C,** Axial view of 15-cm aneurysm, displacing kidney, with a large focus of mural thrombi (*arrow*).

aware of the fact that he had already outlived his life expectancy and refused any surgical intervention. He wished to proceed with comfort care measures with the goal to die at home. He was discharged home on hospital day 3 and died after 4 months, at home.

DISCUSSION

OHS is a rare genetic disorder with multiple clinical features that are sequelae of defective copper transport and decreased collagen and elastin cross-linking. Kinking and elongation of arteries have been described in OHS, and there is a single case report, by Mentzel et al[9] in 1999, that describes an 18-year-old patient with OHS and hepatic and splenic aneurysms. This patient had a known splenic aneurysm on presentation and was found to have a hepatic aneurysm on further workup; the outcome of that patient has not been reported, and we were unable to contact the authors to get more details. There are no previous cases reported of abdominal aortic or iliac artery aneurysms in the literature, and our case is the first one to describe an abdominal aortic aneurysm in a patient with this diagnosis. We contacted specialists in OHS and Menkes syndrome around the world, and none had knowledge of or experience in caring for a patient with OHS after performing any kind of major vascular operation. Although OHS is no longer considered a variant of Ehlers-Danlos syndrome, patients with OHS have decreased collagen and elastin cross-linking, potentially making their connective tissue fragile and operative intervention difficult, in both hemostasis and durability of any type of anastomotic repair. Our patient had previously undergone two intra-abdominal operations and recovered without problems from both but was at much higher risk of operative complications should he have decided on intervention for his aneurysm.

Last, the patient had an aneurysm that grew 13 cm in just 7 years. For unknown reasons, our patient was lost to follow-up, and it is possible that medical or surgical interventions before his presentation in 2013 may have helped prolong or improve the quality of his life.

CONCLUSIONS

We report the first known case of abdominal aortic aneurysm in a patient with OHS. OHS is a connective tissue disorder caused by defective copper transport, leading to decreased collagen and elastin cross-linking.

This is an infrequent and rare disease; special consideration regarding surveillance of vascular disease should be discussed for this very selected patient population.

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Submitted Jan 15, 2015; accepted Mar 15, 2015.