Osteogenesis Imperfecta, Multiple Intra-Abdominal Arterial Dissections and a Ruptured Dissecting-Type Intracranial Aneurysm

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Summary

We describe an adult patient with a ruptured dissecting-type superior cerebellar artery aneurysm and known osteogenesis imperfecta. He was successfully treated with coil embolization and intentional parent vessel sacrifice. During his hospital admission, he also suffered from abdominal distension. An incidental note was made of multiple intra-abdominal arterial dissections. These were managed conservatively. We review the rare association of osteogenesis imperfecta and intracranial aneurysms, as well as discuss management implications.

Introduction

The pathogenesis of intracranial aneurysms is unknown, but likely to be multifactorial. Both modifiable (smoking, hypertension, excessive alcohol consumption) and non-modifiable (familial predisposition, female gender) risk factors have been identified¹. More recently, the first intracranial aneurysm genome-wide association studies have uncovered five susceptibility loci harboring candidate genes that ap-

pear to be preferentially involved in cellular proliferation. These candidate genes are yet to be validated, but may suggest a functional, rather than structural, susceptibility of the vessel wall. Rarely, intracranial aneurysms occur in the context of systemic diseases, most prominently, polycystic kidney disease (PKD) and the heritable connective tissue disorders (CTD)⁴. This latter group of disorders is characterized by single gene defects in the structural components of the vessel wall. While the association of saccular aneurysms and PKD is strong⁵, the literature regarding their association with heritable CTDs is surprisingly sparse⁴. Herein, we describe a patient with a ruptured, dissecting-type superior cerebellar artery (SCA) aneurysm and osteogenesis imperfecta (OI). We provide a brief review of the literature and discuss management implications.

Case Report

Clinical Presentation

A 49-year-old man presented with a sudden-onset headache. His medical history was significant for hypertension and OI. In keeping with the latter diagnosis, he had short stature, progressive hearing loss, and multiple bone fractures. He was initially in a comatose state (Glasgow Coma Scale 7, World Federation of Neurological Surgeons grade 4) with a mildly enlarged (4-mm), unreactive right pupil. A head CT showed diffuse subarachnoid hemor-

Abbreviation Key: CT = computed tomography; CTA = computed tomography angiogram; CTD = connective tissue disorder; EVD = external ventricular drain; GCS = Glasgow Coma Scale; MRA = magnetic resonance angiogram; OI = osteogenesis imperfecta; PICA = posterior inferior cerebellar artery; PKD = polycystic kidney disease; SAH = subarachnoid hemorrhage; SCA = superior cerebellar artery; WFNS = World Federation of Neurological Surgeons.

Figure 1 Axial head CT performed at the time of presentation demonstrates thick subarachnoid hemorrhage in the basal cisterns (A). The accompanying CT angiogram (coronal reformats) performed the same day (B) and three days later (C) shows interval growth of a dissecting-type, right superior cerebellar artery aneurysm. A pre-aneurysmal focal luminal narrowing is demonstrated suggestive of dissection (arrow in panel C). In retrospect, a subtle, caliber irregularity was demonstrated on the initial CT angiogram (B).

Figure 2 Conventional digital subtraction angiogram demonstrates the ruptured right superior cerebellar artery aneurysm in working projection before (A) and after (B) coil embolization with intentional parent vessel sacrifice. A follow-up MR angiogram performed 3 months later confirms complete obliteration of the aneurysm.

rhage (SAH) (Fisher grade III) and acute hydrocephalus (Figure 1A). The patient made a remarkable clinical improvement post-insertion of an external ventricular drain (EVD). The computed tomography angiogram (CTA) (Figure 1B) and next-day catheter angiogram were negative.

Three days later, the patient was noted to

have an enlarging right pupil (6-mm). A repeat CTA demonstrated a ruptured dissecting-type right SCA pseudoaneurysm (Figure 1C). In retrospect, a subtle, luminal caliber irregularity was demonstrated on the original CTA (Figure 1B). A decision was made to perform a coil embolization with intentional parent vessel sacrifice.

Intervention

Using a standard microcatheter technique, coils were deposited into the dissecting pseudoaneurysm and allowed to herniate into the parent vessel for intentional parent vessel sacrifice (Figure 2A,B). Control runs demonstrated complete obliteration of the aneurysm and retrograde filling of the distal SCA via leptomeningeal collaterals from the posterior inferior cerebellar artery (PICA).

Post-Intervention

At one-week and three-month follow-up magnetic resonance angiogram (Figure 2C) demonstrated complete obliteration of the aneurysm without evidence of territorial ischemia. At three-month follow-up, he was cognitively intact with only mild right-sided dysmetria and a partial third nerve palsy. The patient's post-operative course was complicated by a distended abdomen. An abdominal CT angiogram revealed dissections of multiple medium-sized abdominal arteries (Figure 3A-C). These were treated with full systemic heparinization, and the patient made an uneventful recovery.

Discussion

Osteogenesis imperfecta (OI) is a heritable CTD whose cardinal manifestation is bone fragility. Other variable clinical features include blue sclera, short stature, dentinogenesis imperfecta, and progressive hearing loss⁶. OI is caused by abnormalities of type I collagen, which is comprised of two $\alpha_1(I)$ and one $\alpha_2(I)$ polypeptide chains encoded by the *COL1A1* and *COL1A2* genes, respectively. Approximately 90% of individuals with OI harbor mutations in one of these two genes⁶. Type I collagen is a major structural component of the blood vessel wall, and it is therefore surprising that cardiovascular complications are not more conspicuous features of the disease. Sporadic case reports and small cases series describe aortic root dilatation and cardiac valvular problems in adult patients with OI. Particularly noteworthy are reports of vascular dissection, for example, aortic⁸, cervical artery⁹, intracranial¹⁰, and coronary artery dissections¹¹. Taken together, vascular dissection may be the dominant pathomechanism of OI-related vascular disease. When interpreted

from this perspective, a previous report of a nine-year-old girl with "OI-related vasculopathic changes" may represent multiple intracranial dissections at variable times in the healing process¹². There are only four prior reports of individuals with intracranial aneurysms and OI¹³⁻¹⁶. Three reports described patients with ruptured saccular anterior communicating artery aneurysms, while the most recent report described a patient with a ruptured saccular aneurysm at a fenestrated basilar artery¹⁴. None were suggestive of, or explicitly implicated, vascular dissection as a possible pathomechanism. Given the prevalence of intracranial aneurysms in the general population (~2%), these aneurysms may represent coincidental associations¹⁷. Alternatively, they may reflect the underlying predisposing weakness of the vessel wall. Notwithstanding, a growing body of evidence suggests that the vascular fragility in OI manifests primarily as vascular dissection. In our patient reported herein, the classical location for a dissecting pseudoaneurysm, i.e., along the free edge of the tentorium cerebelli, characteristic imaging appearance, and concomitant intra-abdominal dissections strongly support vascular dissection as the underlying pathomechanism. While early repeat vascular imaging is always indicated in cases of CTA negative, diffuse SAH¹⁸, a patient's premorbid diagnosis of OI should prompt careful scrutiny of images for subtle signs of intracranial dissection. Clues to this underlying pathomechanism include (i) peripheral location of the aneurysm (distal to bifurcations), (ii) fusiform appearance, and (iii) the presence of a preaneurysmal luminal narrowing¹⁹. With regard to treatment, two principles should be considered. First, the least invasive approach should be prioritized. Second, treatment should include the site of dissection, and not only the aneurysmal dilatation per se¹⁹. Endovascular parent vessel sacrifice is consistent with these management tenets. For example, the potential difficulties with open surgery in patients with OI are highlighted by a 25% acute mortality rate of cardiovascular surgery²⁰. Endovascular parent vessel sacrifice for the treatment of peripheral aneurysms can generally be performed safely, and the decision to proceed must be balanced against the grave natural history of disease²³.

In summary we describe a patient with a ruptured, dissecting-type intracranial aneurysm and OI. He was successfully treated with coil embolization and intentional parent vessel sacrifice. Incidental note was made of concomitant

Figure 3 Abdominal CT angiogram demonstrates dissections of the superior mesenteric artery (sagittal reformats, A and B) and celiac trunk (axial image (C)). The latter is associated with a mural hematoma (*).

intra-abdominal arterial dissections supporting the underlying pathomechanism of disease. Those involved in the diagnosis and treatment of these rare patients should appreciate that their constitutional vascular fragility is often manifested as vascular dissection.

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