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# AJNR

## **Cerebral Aneurysms in a Patient with Osteogenesis Imperfecta and Exon 28 Polymorphism of *COL1A2***

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## T2 Hypointense Signal of Rathke Cleft Cyst

We read with interest the article by Megdiche-Bazarbacha et al,<sup>1</sup> in which the authors report a case of a giant intrasphenoidal Rathke cleft cyst (RCC). In our recent review of RCCs,<sup>2</sup> we mentioned a comparable case reported by Meyer et al<sup>3</sup> in the *AJNR* of such a giant RCC centered on the sphenoid bone, mimicking a chordoma. A search of PubMed with the keywords “Rathke cyst sphenoid” revealed at least another very similar case recently published in the *Journal of Neurosurgery*.<sup>4</sup> The MR signal intensity characteristics of all these lesions were identical, with T1 and T2 signal intensities slightly greater than that of CSF. It is well known that the signal intensities of RCCs are variable and directly depend on their biochemical content, with most of the reported RCCs demonstrating T2 high signal intensity. It is, however, crucial to point out for practicing radiologists that a homogeneous T2 hypointense signal intensity within a nonenhancing midline sellar cyst is highly suggestive of a RCC.<sup>2</sup> Figure 1 illustrates this feature and also demonstrates how T2-weighted images are more sensitive than T1-weighted images in depicting small cysts located between the anterior and the posterior pituitary lobes.

Finally, the postoperative RCC recurrence rate of only 5% of Megdiche-Bazarbacha et al<sup>1</sup> seems quite low. Although considered as benign lesions, RCCs commonly recur, with reported rates as high as 33%.<sup>5</sup> Independent of the neurosurgeons skill, this recurrence is due to the fact that the most common and safest surgical technique consists of draining the contents of the cyst with only a partial excision or fenestration of the paper-thin wall cyst, leaving in place the source of the secretions that may re-accumulate.



**Fig 1.** A Rathke cleft cyst in a 25-year-old woman with headaches. A, Sagittal T2-weighted image easily demonstrates a homogeneous hypointense-signal-intensity mass perfectly located on the midline between both pituitary lobes. B, Sagittal T1-weighted image barely shows the isointense-to-slightly-hyperintense signal intensity of the lesion.

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## Cerebral Aneurysms in a Patient with Osteogenesis Imperfecta and Exon 28 Polymorphism of COL1A2

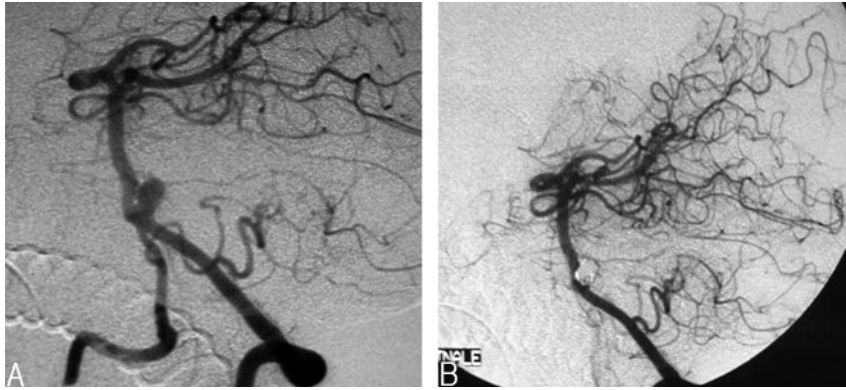
Osteogenesis imperfecta (OI) is a heterogeneous group of heritable disorders characterized by increased bone fragility. Typical extraskel-etal manifestations (hearing loss, blue sclerae, dentinogenesis imperfecta, and hyperlaxity of ligaments and skin) can be variably associated, but no agreed minimum criteria exist.

The neurologic complications mostly concern skeletal disorders, and the most striking finding is basilar invagination. Intracranial hemorrhage caused by mirror trauma can also occur. OI is associated with type I collagen abnormalities: marked decrease in collagen levels can cause vascular complications such as aortic and cervical artery dissection, carotid cavernous fistula, and ulnar and coronary artery aneurysms. Unlike other connective tissue diseases, the cerebrovascular system is less frequently involved in OI.<sup>1</sup> Subarachnoid hemorrhage secondary to ruptured cerebral aneurysm is reported in only 2 cases.<sup>2–3</sup>

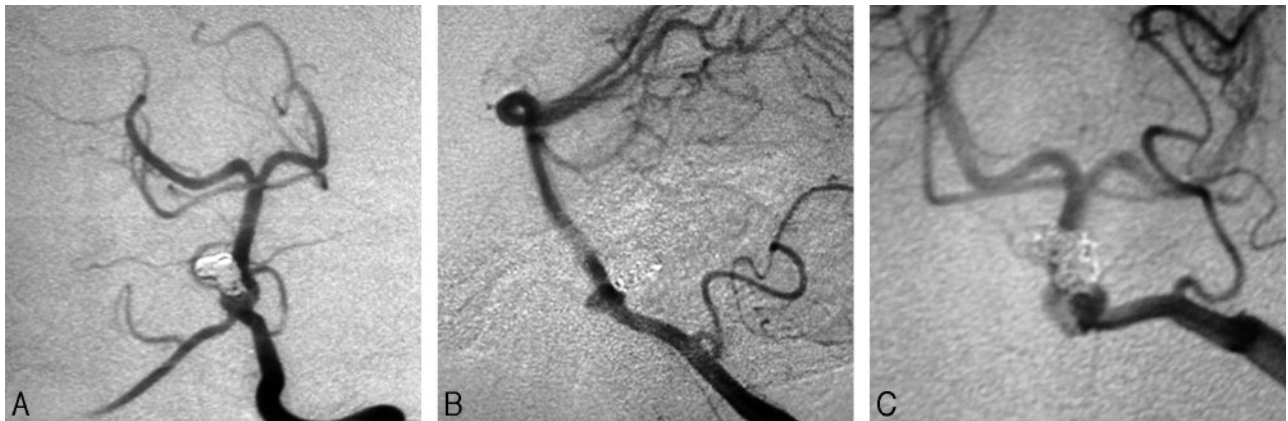
**Case Report.** A 44-year-old man was referred for a sudden loss of consciousness. There was no family history of aneurysm, bone fragility, or congenital musculoskeletal disease. He had left femoral and humeral fractures prior to puberty, caused by mirror traumas, and bilateral mild hearing loss. General physical examination also evidenced dentinogenesis imperfecta. The sclerae were normal. Neurologic examination revealed mild coma (Glasgow Coma Scale score, 13), neck stiffness, and mild right hemiparesis. Brain CT showed subarachnoid hemorrhage. Subsequent CT angiography and cerebral digital subtraction angiography (DSA) disclosed a saccular basilar artery aneurysm and a fenestrated left vertebral artery (Fig 1). Guglielmi detachable coil (GDC) embolization of the aneurysm was successfully performed. His symptoms resolved in 10 days. Four months later, follow-up angiography demonstrated a left vertebral-basilar junction aneurysm (Fig 2). The de novo aneurysm was treated with stent placement (Neuroform, Boston Scientific, Natick, Mass) and GDC coiling. After the procedure, the patient continued to be treated with 75 mg of oral clopidogrel and 100 mg of acetylsalicylic acid. At 10-month follow-up, DSA did not depict any new vascular malformation. Genetic testing for OI was performed, revealing a single nucleotide G/C polymorphism of exon 28 of the gene encoding for alpha-2 polypeptide of collagen I.

Formation of de novo intracranial aneurysm is rare and most commonly appears in the anterior circulation. Acquired changes in vessel walls, resulting from interaction of environmental factors and several genes, may be an explanation. Hereditary factors are often related to some connective-tissue diseases (Ehlers Danlos syndrome type IV, autosomal dominant polycystic kidney disease, pseudoxanthoma elasticum, and Marfan syndrome). At present, the functional candidate genes code for structural proteins of extracellular matrix, like type III and I collagen, elastin, fibrillin 2, lysyl oxidase, and matrix metalloproteinases. Type I collagen is the major extracellular component of the cerebral arterial wall, with a structural role, and is mainly expressed in the adventitia. OI is commonly associated with mutations for type I collagen genes, possibly causing amount reduction or structural variation. Collagen or genetic testing can be performed for further confirmation of the clinical diagnosis of OI, but up to 15% of mildly affected individuals might still have negative findings. OI type I is also characterized by a 10%–30% of bleeding diathesis, because of deficient production of collagen I, an increased vascular fragility, and impaired platelet function.

We did not identify in our patient any known mutation in *COL1A1* and *COL1A2* genes, but the analysis revealed SNP28. This



**Fig 1.** A and B, Pretreatment angiograms demonstrating saccular basilar artery aneurysm.



**Fig 2.** A–C, Posttreatment (with GDC and stent) angiograms of the de novo left vertebral-basilar junction aneurysm.

most significant allelic association, observed in *COL1A2* overexpression, results in an alanine to proline amino acid substitution, determines stability change in collagen I, and may affect the rigidity or elasticity of the vascular wall, causing aneurysm formation. In our opinion, significant vascular fragility may have occurred in our patient with phenotypically mild collagen disease, as confirmed by the association between the functional variant SNP28 and familial IA in a Japanese study.<sup>4</sup>

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## Registry on Matrix Coils: Bias in Inclusion, Exclusion, and Publication

Recently, the first results of a prospective multicenter registry in France using Matrix detachable coils (Boston Scientific, Natick, Mass) were reported in this journal.<sup>1</sup> We have serious concerns about the scientific validity of the reported results because of considerable inclusion and exclusion biases related to patients and aneurysms.

First, the authors presented 2 exclusion criteria for the registry: patients with a Glasgow Coma Scale score of <10 and patients with giant aneurysms “because the final goal of the registry was to evaluate the long-term anatomic results after endovascular treatment with Matrix detachable coils.”

Exclusion of patients with bad clinical scores is likely to increase the angiographic follow-up rate but introduces several biases. For instance, a substantial proportion of patients with bad scores may have an intraparenchymal hematoma. Patients with intraparenchymal hematomas are at risk for early rebleeding after coiling of the ruptured aneurysm. In a study concerning the occurrence of early rebleeding after coiling, patients with a Hunt and Hess (HH) scale grade of III–V had a ninefold increased risk for early rebleeding compared with patients with HH I–II.<sup>2</sup> In the first registry concerning the use of Matrix coils (ACTIVE Study), an unacceptably high proportion of early rebleedings after coiling (7%) was noted.<sup>3, 4</sup> This study has not been published. Therefore, the concern of early rebleeding after coiling, especially with Matrix coils, is not eliminated by this registry, and the finding of the authors that no early rebleedings occurred is without meaning.

The second exclusion criterion is patients with giant aneurysms. If patients with giant aneurysms are excluded from long-term follow-