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

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Petruzzellis et al.1 present an interesting case of a patient with osteogenesis imperfecta and a ruptured aneurysm at the fenestrated basilar artery. However, they misidentify the fenestration as a vertebral fenestration and, as such, do not seem to relate the fenestration to the basilar aneurysm. Figures 1A and 2A beautifully show well-known features of the basilar fenestration just above the vertebral junction2–3: the joining of both vertebral arteries into 1 artery. With the basilar diameter beyond, and rejoining of the fenestrated basilar artery. However, they misidentify the fenestration as a vertebral fenestration and, as such, do not seem to relate the fenestration to the basilar aneurysm. A substantial series by Campos et al.2 of 59 aneurysms of the basilar trunk found 35.5% in association with osteogenesis imperfecta. In this patient with osteogenesis imperfecta and exon 28 polymorphism of COL1A2, the authors claim that the aneurysm seen 4 months after coiling is the same aneurysm as the treated aneurysm, just at the left side of the proximal split of the basilar fenestration. We can compare Fig 1A with Fig 2C for the closest possible orientation, and this comparison gives strong suggestion of the same aneurysm with a refilled neck after coiling, a common enough finding. It seems, then, that this aneurysm is not a new, newly developed one but another occurrence of lack of attention to the details of the case.

Many reports describing coiling of aneurysms at the basilar fenestration are in the literature.5 Perhaps this is the first reported case in a patient with osteogenesis imperfecta, but the discussion in this case avoids this main theme through oversight of important findings and claims others that are dubious. The American Journal of Neuroradiology has an educational responsibility to show readers exemplary neuroradiating cases and interpretations, in addition to rigorous scientific reports and interesting musings of authors in discussion.


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Reply:

We thank Dr. Fox for his comments concerning our previously published letter to the editor entitled, “Cerebral Aneurysms in a Patient with Osteogenesis Imperfecta and Exon 28 Polymorphism Of COL1A2.”1,3 He strongly informs us of a misunderstanding regarding the de novo aneurysm that developed after 4 months. Dr. Fox points out some potential errors in our diagnosis, claiming that the de novo aneurysm is actually a refilling of the previously treated one. We believe that indeed at 4 months, our images revealed that a new aneurysm had developed in front of the previous one (Fig 2B). Further evidence of this was found in the posttreatment un-subtracted images (not published due to space constraints), in which it was possible to appreciate the stent crossing the vertebrobasilar junction and the coils occluding 2 different and clearly separable aneurysms.

Finally, we are grateful to Dr. Fox for bringing to our attention some bibliographic references that may help us achieve a better understanding of the anatomic features of this particular case.