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# Neuroimaging Highlight

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## Cerebral Cavernous Malformations

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A 36-year-old previously healthy right-handed man presented with a 10 day history of left upper extremity sensory seizures. There was no family history of cerebrovascular disease. His interictal neurological examination was entirely normal. CT demonstrated a nonspecific acute hematoma in the right parietal lobe (Figure 1). MR demonstrated multiple other lesions not seen on CT (Figures 2 and 3) and characterized the other lesions as cavernous malformations.

The hematoma was removed and histology confirmed an underlying cavernous malformation.

### DISCUSSION

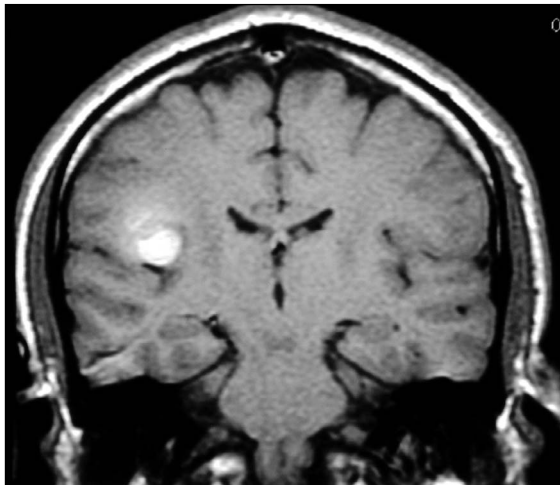
Cerebral cavernous malformations (CM) have previously been referred to as cavernous angiomas and cavernous hemangiomas. Histologically they consist of endothelial-lined blood cavities without intervening brain parenchyma. These vascular malformations present clinically with seizures, hemorrhage, focal neurological deficits or headaches.<sup>1</sup> CMs are angiographically occult. CT demonstrates variable amounts of hyperdensity but frequently small lesions will not be seen. MR

is diagnostic and the MR characteristics of cerebral CMs were first described in detail by Rigamonti et al.<sup>2</sup> The important features include a reticulated core of mixed signal intensity on T2-weighted imaging representing blood-breakdown products of various ages combined with a characteristic surrounding rim of hypointensity representing hemosiderin. Recently, the increased sensitivity of GRE sequences in detecting small lesions has been described<sup>3</sup> and is related to "blooming" of the hypointense hemosiderin rim from susceptibility artifact.

There have been multiple recent advances in the genetic characterization of these lesions, which are thought to be familial in as many as 54% of cases.<sup>4</sup> The familial variants tend to harbour a more aggressive tendency and these patients have an increased risk of *de novo* CM formation.<sup>5</sup> A founder effect has been identified in a Mexican kindred of Hispanic descent. This implies that a single mutation from an ancestor in a recent generation is related to the development of CM in currently affected family members.<sup>6</sup> Moreover, it was shown that in these Mexican kindreds, the mutation is inherited in an autosomal dominant fashion with incomplete penetrance and is related to a



**Figure 1:** Non-enhanced CT scan demonstrates a hematoma in the right parietal lobe.



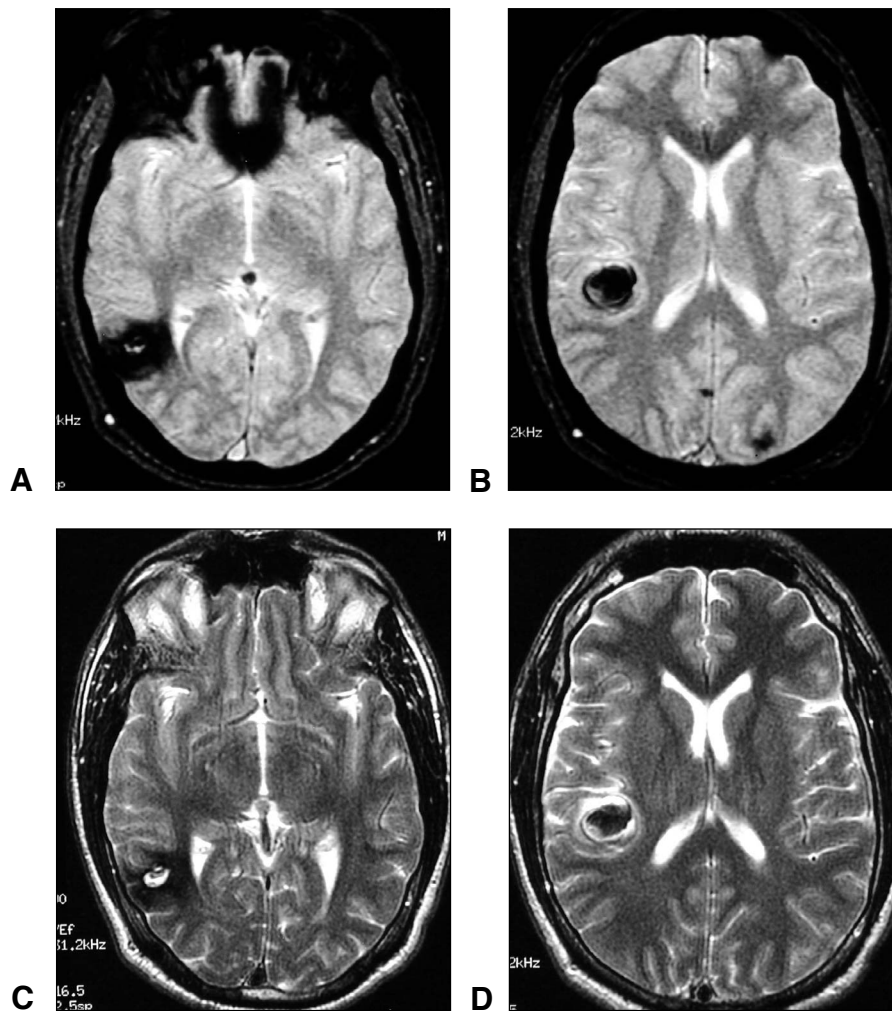
**Figure 2:** Coronal T1 weighted image demonstrates right parietal hematoma.

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**Figure 3:** Axial T2 weighted images demonstrate a right temporal lesion with a reticulated core and a rim of T2 hypointensity (A) as well as the right parietal hematoma that is hypointense on T2 weighted imaging (B). The axial GRE MR demonstrates “blooming” of the hypointense hemosiderin rim and characterizes the right temporal lesion as a cavernous malformation (C). The GRE sequences also identify several other lesions (right and left occipitoparietal regions) that are not seen on CT scan or conventional MR sequences (D).

mutation in a gene (CCM1), located on chromosome 7q.<sup>7</sup> In familial cases of non-Hispanic descent, mutation on chromosomes 7p (CCM2) and 3q (CCM3) have also been identified.<sup>8,9</sup> Finally, “anticipation” has recently been discovered in familial CM with the manifestation of an inherited condition earlier than the condition was manifest in the previous generation.<sup>10</sup>

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