

ACTA2

Missense [mutations](#) in the [gene](#) that codes for [smooth muscle actin](#), [ACTA2](#), cause diffuse smooth muscle dysfunction and a distinct [cerebral arteriopathy](#) collectively known as [multisystemic smooth muscle dysfunction syndrome](#) (MSMDS). Until recently, ACTA2 cerebral [arteriopathy](#) was considered to be a variant of [moyamoya disease](#). However, basic science and clinical data have demonstrated that the cerebral arteriopathy caused by mutant ACTA2 exhibits genetic loci, histopathology, neurological sequelae, and radiographic findings unique from moyamoya disease.

Cuoco et al. conducted a literature review to provide insight into the history, clinical significance, and neurosurgical management of this recently described novel cerebral arteriopathy.

They performed a literature search using PubMed with the key words "ACTA2 mutation," "ACTA2 cerebral arteriopathy," and "multisystemic smooth muscle dysfunction syndrome." Case reports with confirmed ACTA2 mutations and cerebral arteriopathy were included in our review. Our literature search revealed 15 articles (58 cases) of confirmed ACTA2 cerebral arteriopathy. Distinctive features of this arteriopathy included an aberrant internal carotid circulation with dilatation of the proximal segments, occlusive disease at the distal segments, and dolichoectasia. As such, mutant ACTA2 predisposed patients to ischemic strokes as children. Direct and indirect cerebral revascularization procedures are the mainstay treatment options with varying degrees of success. Key Messages: ACTA2 cerebral arteriopathy is a recently described novel cerebrovascular disease seen in patients with MSMDS. Patients currently diagnosed with moyamoya disease who also have dysfunction of smooth muscle organs may benefit from reevaluation by a medical geneticist and ACTA2 genotyping

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Cuoco JA, Busch CM, Klein BJ, Benko MJ, Stein R, Nicholson AD, Marvin EA. ACTA2 Cerebral Arteriopathy: Not Just a Puff of Smoke. Cerebrovasc Dis. 2018 Oct 9;46(3-4):159-169. doi: 10.1159/000493863. [Epub ahead of print] Review. PubMed PMID: 30300893.

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