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Review

October 1, 2018

Fibromuscular Dysplasia and Its Neurologic Manifestations

A Systematic Review

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Key Points

Question What are the neurologic manifestations of fibromuscular dysplasia?**Findings** This systematic review describes the epidemiologic characteristics, clinical and radiologic manifestations, prognosis, and management of fibromuscular dysplasia involving the cervical or intracranial arteries.**Meaning** Cerebrovascular fibromuscular dysplasia is now considered as frequent as renal fibromuscular dysplasia, and longitudinal, international cohort studies with outcomes and genetic sequencing are needed to further understand its clinical course and enable the development of evidence-based treatments.

Abstract

Importance Data on neurologic manifestations of fibromuscular dysplasia (FMD) are rare, and current knowledge remains limited.**Objectives** To present a comprehensive review of the epidemiologic characteristics, management, and prognosis of the neurologic manifestations associated with
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cerebrovascular FMD (ie, involving cervical or intracranial arteries) and to guide future research priorities.

Evidence Review References were identified through searches of PubMed from inception to December 2017 using both the medical subject headings and text words. Additional sources were also identified by reviewing reference lists of relevant articles and through searches of the authors' personal files. Selected articles described at least 1 clinical or radiologic feature and/or outcome of cerebrovascular FMD. Isolated case reports could be included if they described interesting or noteworthy manifestations of FMD.

Findings A total of 84 relevant references were identified. Diagnosis of cerebrovascular FMD is based on the appearance of alternating arterial dilatation and constriction ("string of beads") or of focal narrowing, with no sign of atherosclerotic or inflammatory lesions. Although the diagnosis is easily apparent on results of radiographic imaging, making a diagnosis can be challenging in children or individuals with atypical phenotypes, such as purely intracranial FMD and arterial diaphragm. Involvement of multiple arteries is common, and there is increased incidence of cervical artery dissection and intracranial aneurysms. A variant in the *PHACTR1* gene has been associated with FMD as well as cervical artery dissection and migraine, although less than 5% of cases of FMD are familial. Headaches, mainly of the migraine type, are observed in up to 70% of patients with FMD. Cerebrovascular FMD is mostly asymptomatic, but the most frequent neurologic manifestations include transient ischemic attack and ischemic stroke, notably in the presence of associated cervical artery dissection. Other conditions associated with FMD include subarachnoid hemorrhage and, rarely, intracranial hemorrhage. Management relies on observational data and expert opinion. Antiplatelet therapy is considered reasonable to prevent thromboembolic complications. Endovascular therapy is typically restricted to cases with symptomatic stenosis despite optimal medical therapy or in those with rupture of an intracranial aneurysm.

Conclusions and Relevance Longitudinal cohort studies of individuals of multiple ethnicities with biosampling are needed to better understand the risk factors, pathophysiological features, and outcomes of FMD. Patient advocacy groups could assist researchers in answering patient-centered questions regarding FMD.

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