Cerebral pseudovasculitic lesion associated with Von Recklinghausen's Neurofibromatosis: A Case Report

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Parole chiave: vasculitis and pseudovasculitis, Neurofibromatosis type 1, MRI and MRA

A young asymptomatic 10 years old girl, affected by NF1, came to our observation for a MRI study in order to evaluate the grade of the central nervous system involvement. Minimum sequences included a T1-weighted, fast spin-echo T2-weighted axial and 3D time-of-flight (TOF) MRA fluid-attenuated inversion recovery sequences were used. Diffusion-weighted imaging (DWI) and intravenous gadopentetate dimeglamine (0.2 mL/kg) enhanced scans were performed as supplementary sequences and these results were recorded. We found a reduction of the right middle cerebral artery's size in its M2 tract and irregularity, without evidence of its distal tracts, revascularized from distal meningeal branches. This radiological finding seems to be suggestive of a pseudovasculitic lesion. The lesion is stabilized and its follow up doesn't need.

Vascular lesions are uncommon in patients with neurofibromatosis 1 (NF1) and CNS involvement seems much rarer. In a series reported by Rosser et al., eight out of 316 children with NF1, who underwent brain MRI, showed cerebrovascular problems, including stenoses or occlusion of the internal carotid and cerebral arteries, aneurysms and moyamoya disease, without evidence of a pseudovasculitic lesion. Its etiological aspects and its clinical features have not been clear. Neurofibromin, the protein product of the NF1 gene, is expressed in endothelial and smooth muscle cells of blood vessels and is likely to be involved in pathogenesis. Loss of neurofibromin function may cause smooth muscles to proliferate, causing vascular stenosis. Our case makes us to understand how this disease can be various and diverse.


-Nicolò Pipitone, Carlo Savarani. Role of imaging in vasculitis and connective tissue diseases, Best Practice and Research Clinical Rheumatology Vol 22 No 6 1075-1091, 2008