

Fibromuscular dysplasia as a rare cause of severe ischemic stroke in a young man

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Fibromuscular dysplasia (FD) is defined as an idiopathic, segmental, non inflammatory, non atherosclerotic disease of the arterial walls, leading to stenosis of small and medium-sized arteries. FD is an uncommon vascular disorder that occurs in young to middle-aged individuals affecting more often women than men¹.

CASE

A 43-years-old man was admitted to our Neuro-rehabilitation Unit with a severe aphasia and a moderate to severe right hemiparesis mainly involving the upper limb. CT scan showed a deep lesion in the left fronto-temporal region.

Complete hematological examination and cardiac ultrasounds were negative. A carotid Doppler scan showed an occlusion of left internal carotid artery (ICA), immediately after its origin. Cerebral catheter angiography showed an “irregular profile of ICA in the extracranial tract with fibroplastic phenomena and severe stenosis of its intracranial tract and of the proximal tract of left MCA” (Figure).

Diagnosis of “major stroke due to mean cerebral artery dissection in patient with fibromuscular dysplasia” was supposed and the patient continued oral anticoagulation treatment for six months. At one year-follow-up the patient did not present any ischemic recurrences with a slow but significant improvement of his neurological condition.

DISCUSSION

FD is a rare vascular disease often overlooked or misdiagnosed. Cervical FD is asymptomatic in the 70% of cases; thus diagnosis can be posed with angiography examination or autopsy after death for unspecified stroke or complicated hypertension². The pathogenesis of FD is unclear. It is likely that FD may start as minor lesion of congenital origin, which predisposes to an abnormal fibro-proliferative response to mechanical or circulatory stimuli (i.e. hypertension, smoke and frequent microtrauma)³.

Spontaneous cervical artery dissection is a common cause of stroke in young adults and is associated with FD in 15% of cases. A typical patient with carotid artery dissection presents the classic triad with pain on one side of the head, face or neck accompanied by a



Figure. Angiogram of the left ICA shows the irregularity of the artery profile.

partial Horner’s syndrome and followed hours or days later by cerebral or retinal ischemia. The presence of any two elements of this triad should strongly suggest the diagnosis^{4,5}.

In our patient diagnosis was difficult since personal history and symptomatology were not typical. Firstly, the patient was a male, and FD is more common in female; secondly, the clinical presentation of the artery dissection was not so usual since the patient did not refer any kind of neck or head pain and did not present Horner’s syndrome signs.

Nevertheless, vasculitides and collagenopathies can lead to a misdiagnosis². Since our patient did not present any clinical or laboratory sign of the main collagen or autoimmune diseases, FD diagnosis may have been hypothesized.

The poor knowledge of the natural history and the lack of randomized trials that compared the different treatment options do not allow any satisfactory judgment to be made regarding the need for or the efficacy of any treatment, including surgery and oral anticoagulation.

With the present case we want to underline the importance of a specific and accurate diagnostic screening in young patients affected by cryptogenic stroke. Indeed, rare causes of stroke, such as FD, have to be suspected in young people even when clinical and/or laboratory tests are not significant.

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DISPLASIA FIBROMUSCULAR COMO CAUSA RARA DE ACIDENTE VASCULAR ISQUÊMICO EM JOVEM

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SCA2 presenting as an ataxia-parkinsonism-motor neuron disease syndrome

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The procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national). All subjects were provided with the approved informed consent.

Spinocerebellar ataxia type 2 (SCA2) is characterized by progressive cerebellar ataxia, slow saccadic eye movements and peripheral neuropathy. Atypical SCA2 phenotypes with prominent dementia, an amyotrophic lateral sclerosis-like presentation, and levodopa-responsive parkinsonism are also encountered¹. The definite diagnosis of SCA2 is based on clinical symptoms and molecular genetic testing to detect an abnormal CAG trinucleotide repeat expansion of the *ATXN2* gene on chromosome 12q¹. The protein synthesized by *ATXN2* is known as ataxin-2 that is involved in RNA metabolism and translation regulation¹. Herein we report an unusual case of SCA2 presenting as an ataxia-parkinsonism-motor neuron disease (MND) syndrome.

CASE

A 46-year-old Brazilian man of Italian ancestry, presented for consultation because of progressive gait instability and muscle cramps that started 18 months before. He also developed rapidly progressive muscle weakness few months before his first appointment. His family history revealed affected individuals (ataxia) within first and second generations. On neurological examination, there were mild dysarthria, slow saccades, mild limb and gait ataxia, parkinsonism, brisk reflexes and bilateral Babinski sign. He also presented muscle weakness in upper limbs, diffuse fasciculations and atrophy involving upper limbs, chest and face. Genetic testing confirmed the diagnosis

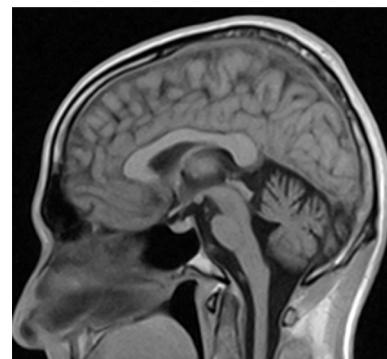


Figure. Sagittal T₁ brain MRI disclosing cerebellar and brainstem atrophy. This imaging finding is frequently seen in SCA2.

of SCA2 with 40 CAG repeats. Brain magnetic resonance imaging (MRI) disclosed cerebellar and brainstem atrophy (Figure). Electroneuromyography showed denervation activity at rest, with fasciculation, fibrillation and positive sharp wave potentials in the bulbar, cervical, thoracic and lumbosacral regions. Nerve conduction studies showed normal sensory and motor nerve conduction velocities and reduced amplitudes of the motor potentials. We introduced riluzole 50 mg b.i.d. as treatment for amyotrophic lateral sclerosis (ALS).

DISCUSSION

Few previous studies have described the association of SCA2 with MND. Infante et al.² reported a case of a 61 years-old woman with SCA2 diagnosis who developed a levodopa-responsive parkinsonism after 6 months of the ataxia-onset of symptoms, but later on disease course