

Intracranial Fibromuscular Dysplasia in a Male Child Presented by Middle Cerebral Artery Dissection and Stroke

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Abstract - Fibromuscular dysplasia (FMD) is a group of nonatherosclerotic, noninflammatory arterial diseases that most commonly involve the renal and carotid arteries it typically occurs in women aged between 30 and 50 years. FMD in children is rare and its most common site of involvement are renal arteries. Stroke in children is generally uncommon and due to FMD is even rarer, which is why it is often unrecognized. We present a case of a boy, with an inconspicuous previous anamnesis and orderly development, who woke up at the age of 9 with weakness of the left side of the body and speech disorders. MR showed an ischemic lesion in the irrigation area of the right middle cerebral artery (arteria cerebri media- ACM). In neurological status mild dysarthria with left central facial nerve lesion was noted as well as mild paresis of the left extremities. Neurosonological testing showed segmental extensions and narrowing of the right ACM with underlying dissection, which speaks in favour of fibromuscular dysplasia. Therapy with acetylsalicylic acid 50 mg was initiated, and with further follow-up of the patient, complete regression of symptoms and normal neurological status were recorded.

Key words: dissection; fibromuscular dysplasia; stroke

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Introduction

Fibromuscular dysplasia (FMD) is a group of nonatherosclerotic, noninflammatory arterial diseases that most commonly involve the renal and carotid arteries. According to numerous studies it seems to be hereditary in 10 % of cases. Angiographic most common classification includes the multifocal type, with

multiple stenoses and the 'string-of-beads' appearance, and tubular and focal types. Also important, histological classification distinguishes 3 types: intimal, medial and perimedial, where medial type is most common among adults. FMD typically occurs in women aged between 30 and 50 years and most common manifestation is renovascular hypertension [1]. Neurological symptoms due to carotid artery involvement are headache, dizziness, pulsatile tinnitus or more severe like TIA and stroke. Changes in blood vessels that can occur, except stenosis, are also aneurysms and dissection. FMD mainly affects the distal extracrani-

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al internal carotid artery [2]. FMD in children is rare and it most commonly affects the renal arteries, or mesenteric arteries and abdominal aorta, while carotid vessels are less frequently involved. Stroke in children is generally uncommon and due to FMD is even rarer, which is why it is often unrecognized [3].

Subjects and Methods

A 9-year-old boy was admitted to the emergency department due to weakness of the left side of the body and speech disorder. The boy woke up with symptoms 2 hours before arriving at the emergency department. According to the parents, upon waking up, they noticed that the boy was speaking more slowly and with difficulty, that he was babbling and that he was walking strangely, dragging both legs. At the time, they did not notice weakness of the one side of the body. The boy did not complain on anything that morning, the parents denied the trauma, and the night before he was speaking and moving properly. The boy did not urinate spontaneously or have a stool. Earlier psychomotor development went well, the boy was born on time, vaginally, without complications, APGAR scores of 10/10, without neonatal jaundice. The mother states that the boy developed properly, had regular paediatric check-ups, was vaccinated regularly, spoke, and walked in accordance with expectations. So far, he has not had any serious illnesses or been hospitalized. Parents also did not notice changes in behaviour, personality, and mood, nor staring or sleepiness. He regularly and successfully performed school duties as well as extracurricular activities. There is also a younger son in the family, aged 4 who, just like our patient, is completely healthy. Upon arrival at the emergency department, the boy was conscious, properly oriented, with mild dysarthria, central left facial nerve lesion and mild paresis of the left extremities with Babinski's sign and normal sensation, National Stroke Scale/Score (NIHSS) of 6. Complete laboratory tests and emergency CT scan of the brain were performed. All the findings, just like the vital parameters (blood pressure 110 / 80 mmHg) were normal and the boy was hospitalized. During hospitalization, an EEG, as well as an EEG after a sleepless night, were performed, but the findings were normal. At that moment, the boy's deficit persisted for 24 hours, showing minor signs of recovery in the form of only discrete dysarthria while the rest of the neurological status remained the same. On the MRI of the brain, acute ischemia was verified in the right parietal lobe, in



Figure 1. Transcranial colour Doppler of the right middle cerebral artery (ACM), which revealed segmental narrowing and expansions

the irrigation area of the right middle cerebral artery (ACM), which officially confirmed the diagnosis of stroke. Neurosonological testing, colour Doppler, was also carried out, which showed a normal extracranial status, but the visualization of the intracranial blood vessels revealed segmental narrowing and expansions in the right ACM with underlying dissection (Figure. 1). The findings of dissection and stenosis were also confirmed by MR-angiography, which speaks in favour of fibromuscular dysplasia. During the hospitalization, extensive tests were carried out such as: heart ultrasound, “bubble” test, ergometry, then hematologic tests as well as genetic tests for hereditary connective tissue diseases. The parents were also involved in the processing, radiological and genetic testing was done, but the results arrived in order. Therapy with acetylsalicylic acid 50 mg was initiated, and at the time of discharge, a discrete facial paresis of the central type on the left with discrete weakness of the left arm persisted (NIHSS 2). The patient is monitored regularly on an outpatient basis and the neurological deficit has completely disappeared with physical therapy.

Discussion

FMD in children has certain specificities. While in adults it is much more common in women, in childhood, according to research, it occurs equally in boys and girls, and some data show an even higher incidence in boys [3]. In childhood, it is assumed that the he-

reditary form of the disease plays a greater role, and recent genetic studies have found a possible link between FMD and the phosphatase and actin regulator 1 (PHACTR1) gene on chromosome six. The protein encoded by this gene is a member of the phosphatase and can bind actin and regulate the reorganization of the actin cytoskeleton. A variant of this gene, rs9349379, has been shown to increase the chances of having FMD by 40 % [4]. In contrast to adults, where multifocal variants are the most common, in children most common forms of FMD are unifocal, most often in the area of the renal arteries, which is why our case is rare [4]. Twenty to thirty percent of adults with FMD develop aneurysms or dissection, the most common sites of dissection are the extracranial carotid, vertebral arteries, renal arteries, and coronary arteries, while aneurysms occur more often extracranial and in the area of the renal arteries [5]. Intracranial changes are rare in adults and most of the recorded cases were discovered precisely in children or young adults [6]. The most common symptoms that occur in children are hypertension, headache, and abdominal bruits, which is typical for involvement of the renal arteries, therefore FMD should always be considered in children with these symptoms. In most studies, stroke was recorded in a very small percentage or not at all, and the most affected blood vessels in children are renal, mesenteric arteries and aorta [3]. Also, one interesting thing about FMD in childhood is that, unlike the adult form, where the tunica media is most

often affected, in children there is a predominant involvement of the tunica intima, which could also explain the more frequent occurrences of dissections and intracranial involvement considering the structure of the blood vessels [7].

In our case, we had a rare presentation of the disease with an intracranial presentation in the form of dissection, and these mentioned changes also caused a complication, a stroke. Even in childhood, adult diseases such as FMD, which can lead to serious consequences, should be considered, especially in children with hypertension or abdominal bruit. Although extra and intracranial blood vessels are less often affected in children suffering from FMD, it is important to do regular checks and keep in mind this form of disease. This case emphasizes on the importance of extensive diagnostic studies and a multidisciplinary approach to these patients for the purpose of prevention and providing the best care and treatment.

Acknowledgements

None.

Conflict of interest

None to declare.

Funding Sources

None.

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