CASE REPORT/*OPIS PRZYPADKU*

Successful bilateral pallidal stimulation in a patient with isolated lower limb dystonia coexistent with Langerhans cell histiocytosis and coeliac disease

Skuteczna obustronna stymulacja części wewnętrznych gałek bladych u chorej na izolowaną dystonię kończyn dolnych ze współistniejącą histiocytozą i celiakią

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Abstract

The authors report a case of bilateral globus pallidus internus (GPi) stimulation for treatment of medically intractable isolated lower limb dystonia. The 14-year-old girl developed dystonic movements in her left lower limb. At the age of 17, the patient was handicapped by dystonic movements in her lower limbs, and became wheelchair-bound. Pharmacological therapy and botulinum toxin injection resulted in transient and modest benefit. Moreover, the patient was diagnosed with histologically proven coeliac disease and Langerhans cell histiocytosis. Genetic testing revealed the presence of DYT-1 mutation. The 17-year-old girl underwent bilateral implantation of deep brain stimulation leads. Bilateral GPi stimulation resulted in remarkable improvement of phasic dystonic movements, and dystonic posture of lower limbs. Over 2 years postoperative follow-up, the patient is able to walk independently. Bilateral GPi stimulation appears to be an effective treatment modality for isolated lower limb dystonia.

Key words: movement disorders, lower limb dystonia, pallidal stimulation, functional neurosurgery.

Streszczenie

Autorzy przedstawiają przypadek chorej poddanej obustronnej stymulacji części wewnętrznej gałki bladej z powodu izolowanej dystonii kończyn dolnych. U 14-letniej dziewczynki zaobserwowano ruchy dystoniczne obejmujące początkowo lewą kończynę dolną. Stopniowo ruchy dystoniczne objęły również prawą kończynę dolną. W wieku 17 lat dystonia w znacznym stopniu upośledzała życie chorej, która poruszała się wyłącznie na wózku inwalidzkim. Farmakoterapia i wstrzyknięcia toksyny botulinowej przyniosły niewielką i przejściową poprawę. Badanie genetyczne wykazało obecność mutacji DYT-1. Chorą skierowano do leczenia operacyjnego. W wieku 17 lat przeszła operację obustronnej implantacji elektrod do głębokiej stymulacji mózgu. Obustronna stymulacja GPi przyczyniła się do znacznego zmniejszenia ruchów dystonicznych i wymuszonego dystonicznego ustawienia kończyn dolnych. Podczas dwuletniej obserwacji pacjentka zaczęła chodzić samodzielnie. Obustronna stymulacja GPi wydaje się skuteczną metodą leczenia izolowanej dystonii kończyn dolnych.

Słowa kluczowe: choroby ruchu, dystonia kończyn dolnych, stymulacja gałki bladej, neurochirurgia czynnościowa.

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Introduction

Pallidal stimulation is now a well-established treatment modality for primary generalized dystonia (PGD) [1-4]. Among patients with PGD, most benefit has been observed in those harbouring DYT-1 mutation [1-3]. Furthermore, striking improvement has also been noted in patients with PGD without DYT-1 mutation [5]. A patient with secondary generalized dystonia can also gain functional benefit under chronic bilateral GPi stimulation, although the outcome is less predictable than in patients with PGD. Recent reports have indicated that patients with medically intractable cervical dystonia or segmental dystonia (Meige's syndrome) can improve under chronic bilateral GPi stimulation [6-8].

Pallidal stimulation is also very effective in ameliorating dystonic features in advanced stages of Parkinson's disease [9]. Early morning foot dystonia responds very favourably to GPi stimulation [10].

Foot or lower limb dystonia is very often the initial site in young-onset dystonia. It is usually primary and spreads to other body parts. Primary generalized dystonia in early childhood is associated with DYT-1 mutation. The symptoms usually appear in childhood or early adulthood. The mean age of onset is approximately 12 years. Symptom onset involving a lower limb is also associated with an increased likelihood that the condition will quickly evolve to generalized dystonia.

The management of patients with PGD is challenging. The pharmacological treatment has a very limited influence on dystonic movements and dystonic posturing. Botulinum toxin injections can be applied only locally. The dystonic movements produce musculoskeletal deformities involving the foot or the cervical or lumbar spine. To avoid such complications in patients with PGD, an early surgical approach is warranted.

In this article, we present a young patient with isolated lower limb dystonia without dystonic movements in other body parts. The isolated dystonia confined only to both feet resulted in total inability to walk. After three and a half years of her illness she was physically handicapped only by lower limb dystonia. Genetic testing revealed the presence of DYT-1 mutation. This clinical picture of the presented positive DYT-1 case with onset in a lower limb is rather unusual. There was no spread of dystonia to other body parts and both feet were equally affected by dystonic movements. Moreover, the patient suffered from histiocytosis and coeliac disease. These two entities can also be associated with various movement disorders [11,12]. In the presented case, genetically proven for DYT-1 mutation, the dystonic features have a rather genetic background. This case demonstrates that dystonia confined only to feet can severely handicap the individual without the spread of dystonic movements to other body parts.

Case report

This 17-year-old right-handed woman with negative family history of movement disorders and no other neurological disease was referred to our neurosurgical department for consideration of pallidal stimulation due to medically intractable young-onset lower limb dystonia. The first symptoms were noticed at the age of 14 as increased tension in the left foot only after walking. The first admission to a paediatric ward was made to elucidate muscle spasm involving only the left foot, with small foot inversion, and curling of the big toe of the left foot. The neurological examination was unremarkable besides inappropriate dystonic posturing of the left foot. The laboratory tests indicated only increased inflammatory markers. She was seen by a neurologist who prescribed hydroxizinum. This treatment improved the patient's symptoms. A computed tomography (CT) scan and magnetic resonance imaging (MRI) of the brain were normal. An EEG was also unremarkable. She was diagnosed as having vegetative dystonia. To exclude spinal pathology of atypical left foot posturing, MRI of the entire spine was done, which was unremarkable. After 10 months, the right foot started to be affected by dystonic movements. During the second admission the right foot was inverted and all the toes were curled. The right foot was more affected than the left. The patient experienced walking difficulty for the first time. The electromyography and electroneurography examinations were normal. A battery of laboratory tests including mitochondrial antibodies, smooth muscle antibodies, gastric cell antibodies, and antinuclear antibodies was performed. All antibody values were within the normal range. Combi-test was negative. The psychological testing and psychiatric consultations were unremarkable. The diagnosis of young-onset dystonia involving only the feet was made. The patient was put on levodopa + benserazide (62.5 mg) with the daily dose titrated up to 4 tablets a day. The levodopa treatment brought only transient and modest benefit lasting 2 months. Benzodiazepine was initiated but also with slight effect. Laboratory examinations were within the normal range beside visibly increased inflammatory markers. To exclude an active inflammatory process, tests