Mohr-Tranebjaerg Syndrome

Deafness-dystonia-optic neuronopathy (DDON) syndrome, also known as Mohr-Tranebjærg syndrome, is characterized by hearing loss that begins early in life, problems with movement, impaired vision, and behavior problems. This condition occurs almost exclusively in males.

Case reports

Coenen et al. from the Department of Stereotactic and Functional Neurosurgery, Department of Neurology and Neurophysiology, Department of Neuroradiology, University Hospital Freiburg and Parkinson-Klinik Wolfach, Germany, reported a 28-year-old man presented with a history of sensorineural deafness since early childhood treated with bilateral cochlear implants (CIs). He showed signs of debilitating dystonia that had been present since puberty. Dystonic symptoms, especially a protrusion of the tongue and bilateral hand tremor, had not responded to botulinum toxin therapy. They diagnosed Mohr-Tranebjaerg syndrome (MTS).

Deep brain stimulation (DBS) of the bilateral globus pallidus internus was performed predominantly with stereotaxic computed tomography angiography guidance under general anesthesia. Electrophysiology was used to identify the target regions and to guide DBS electrode placement.

In the immediate postoperative course and stimulation, the patient showed marked improvement of facial, extremity, and cervical dystonia. More than 2 years after implantation, his dystonic symptoms had dramatically improved by 82%.

The use of DBS for the dystonia in MTS was previously described but not in the presence of bilateral CIs.

DBS in MTS may be a viable option to treat debilitating dystonic symptoms. They describe successful DBS surgery, despite the presence of bilateral CIs, and stimulation therapy over 2 years [1)].

Eggink et al. from the Department of Neurology, Department of Genetics, Department of Rehabilitation, Department of Neurosurgery, University Medical Center Groningen, The Netherlands, reported two patients with dystonia-deafness syndrome due to a beta-actin gene mutation.

They report on disease course, genetic testing, and management of 2 patients, mother and daughter, presenting with dystonia-deafness syndrome.

After exclusion of known dystonia-deafness syndrome causes, whole-exome sequencing revealed a beta-actin gene mutation (p.Arg183Trp) in both patients. Although beta-actin gene mutations are generally associated with developmental Baraitser-Winter syndrome, dystonia-deafness syndrome has been reported once in identical twin brothers. Bilateral GPi-DBS led to a significant decrease of dystonia and regain of independency in our patients.

The p.Arg183Trp mutation in the beta-actin gene is associated with the clinical presentation of dystonia-deafness syndrome, even with only minimal or no developmental abnormalities of Baraitser-Winter syndrome. GPi-DBS should be considered to ameliorate the invalidating dystonia in these

patients. [2].

Cif et al. reported in 2013 the article Progressive dystonia in Mohr-Tranebjaerg syndrome with cochlear implant and deep brain stimulation [3].

References

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