Subthalamic Deep Brain Stimulation in Westphal variant of Juvenile Huntington's disease

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Introduction

Juvenile-onset Huntington disease (HD) is a rare kind of HD, which affects around 5% of patients with disease manifestations before the age of 21 years

Huntington's disease patients with predominantly choreic symptoms and even Westphal variant of HD may be benefited from bilateral globus pallidus internus (GPi) Deep Brain Stimulation (DBS) to some extent, though the trials give inconsistent results

Methods

We present a rare patient of Westphal variant of Juvenile Huntington's disease who responded very well to STN DBS

Results

A 24-year-old engineer presented with features of Parkinsonism for 7 years and some disturbances in sleep like interrupted sleep and sleep talking for 6 months

Around 5 years back, he was started on levodopa-carbidopa combination but he developed drug-induced dyskinesias within a few months of starting this treatment. Hence, he was referred to our institute for Deep Brain Stimulation

On examination, HMFs were intact, pursuit was broken motor bulk was normal, there was rigidity on the left side more than the right side, cogwheeling in both the upper limbs and truncal and neck rigidity more in the forward direction

His tendon reflexes were brisk, cerebellum could not be assessed, pull test was positive and he had difficulty in initiating gait

His autonomic function tests, neck and spine examination and MRI Head were normal, UPDRS score was 92/199 without levodopa and 50/199 with levodopa

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Results

In view of young-onset Parkinsonism, his whole-exon sequencing was sent, which came out to be normal

Due to some sleep disturbances, for ruling out Westphal variant of Huntington's disease, we sent a repeat analysis for Huntingtin gene and found an upper allele repeat size of 43±2 and a lower allele repeat size of 17±1

There was no significant history for a similar illness in any of the family members and the allele repeats in the parents were within normal limits

On the wishes of the family in view of troubling dyskinesias and good levodopa response, the patient underwent bilateral subthalamic nucleus (STN) Deep Brain Stimulation with rechargeable implantable pulse generator placement and subsequently a pulse width of 60 µs, frequency of 130 Hz and a current of 1.8mA on the right side and that of 1.4 mA on the left side was started

The patient has been under follow up for two years now and is tolerating DBS well without much deterioration in his illness

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Discussion

Most of the previous studies have used GPi DBS in Huntington's disease patients, with a very few researchers using subthalamic nucleus (STN) DBS

Results have been inconsistent with some studies giving good results but not the others

The benefits are mostly observed in chorea predominant cases rather than Parkinsonian symptoms

Ours is probably a very rare, if not the only, case of Westphal variant of Juvenile Huntington's disease, who presented as a young onset Parkinson's disease patient and was benefited using STN DBS rather than GPi DBS

However, we do need to have a longer follow up period in order to observe what happens to the person in the long term

Conclusions

Cases of Westphal variant of Huntington's disease, including the juvenile cases, such as these may be taken up for STN DBS, provided they fulfil all the other criteria for undergoing DBS contrary to the traditional thinking of HD being not very responsive to DBS

References

1. Bakels HS, et al. Mov Disord. 2022;37(1):16-24.
2. Bonomo R, et al. Neurol Sci. 2021;42(11):4447-4457.
3. Kaczynska J, et al. Neurol Neurochir Pol. 2022;56(4):299-307.
4. Yin Z, et al. Brain Stimul. 2020;13(6):1722-1731.
5. Ferrea S, et al. J Neurol. 2018;265(7):1573-1579.
6. Cislaghi G, et al. Neuromodulation. 2014;17(5):502-505.
7. Wojtecki L, et al. Front Neurol. 2015;6:177.
8. Gruber D, et al. J Neural Transm (Vienna). 2014;121(10):1303-1312.