Localization of motor and verbal fluency effects in subthalamic DBS for Parkinson’s disease.

Related Articles

Localization of motor and verbal fluency effects in subthalamic DBS for Parkinson’s disease.

Parkinsonism Relat Disord. 2020 Aug 21;79:55–59

Authors: Mossner JM, Chou KL, Maher AH, Persad CC, Patil PG

Abstract

INTRODUCTION: Subthalamic nucleus deep brain stimulation (STN DBS) improves cardinal motor symptoms of Parkinson’s disease (PD) but can worsen verbal fluency (VF). An optimal site of stimulation for overall motor improvement has been previously identified using an atlas-independent, fully individualized, field-modeling approach. This study examines if cardinal motor components (bradykinesia, tremor, and rigidity) share this identified optimal improvement site and if there is co-localization with a site that worsens VF.

METHODS: An atlas-independent, field-modeling approach was used to identify sites of maximal STN DBS effect on overall and cardinal motor symptoms and VF in 60 patients. Anatomic coordinates were referenced to the STN midpoint. Symptom severity was assessed with the MDS-UPDRS part III and established VF scales.

RESULTS: Sites for improved bradykinesia and rigidity co-localized with each other and the overall part III site (0.09 mm lateral, 0.93 mm posterior, 1.75 mm dorsal). The optimal site for tremor was posterior to this site (0.10 mm lateral, 1.40 mm posterior, 1.93 mm dorsal). Semantic and phonemic VF sites were indistinguishable and co-localized medial to the motor sites (0.32 mm medial, 1.18 mm posterior, 1.74 mm dorsal).

CONCLUSION: This study identifies statistically distinct, maximally effective stimulation sites for tremor improvement, VF worsening, and overall and other cardinal motor improvements in STN DBS. Current electrode sizes and voltage settings stimulate all of these sites simultaneously. However, future targeted lead placement and focused directional stimulation may avoid VF worsening while maintaining motor improvements in STN DBS.

PMID: 32866285 [PubMed — as supplied by publisher]

Comparison of the clinical phenotype and haematological course of siblings with Fanconi anaemia.

Related Articles

Comparison of the clinical phenotype and haematological course of siblings with Fanconi anaemia.

Br J Haematol. 2020 Aug 31;

Authors: Jung M, Mehta PA, Jiang CS, Rosti RO, Usleaman G, Correa da Rosa JM, Lach FP, Goodridge E, Auerbach AD, Davies SM, Smogorzewska A, Boulad F

Abstract

Fanconi anaemia (FA) is a genetic disorder due to mutations in any of the 22 FANC genes (FANCA-FANCW) and has high phenotypic variation. Siblings may have similar clinical outcome because they share the same variants; however, such association has not been reported. We present the detailed phenotype and clinical course of 25 sibling sets with FA from two institutions. Haematological progression significantly correlated between siblings, which was confirmed in an additional 55 sibling pairs from the International Fanconi Anemia Registry. Constitutional abnormalities were not concordant, except for a moderate degree of concordance in kidney abnormalities and microcephaly.

PMID: 32866879 [PubMed — as supplied by publisher]
Donor-derived myelodysplastic syndrome after allogeneic stem cell transplantation in a family with germline GATA2 mutation.

Related Articles

Donor-derived myelodysplastic syndrome after allogeneic stem cell transplantation in a family with germline GATA2 mutation.

Int J Hematol. 2020 Aug 31;:

Abstract
Germline GATA2 heterozygous mutations were identified as complex immunodeficiency and hematological syndromes characterized by cytopenia (monocytes, B-cells, NK-cells), susceptibility to mycobacterium, fungus, or Epstein-Barr virus (EBV) infection, and myelodysplastic syndrome (MDS)/acute myelogenous leukemia (AML) development. Herein, we report a patient with AML who had a fatal infection after allogeneic hematopoietic stem cell transplantation (HSCT) due to impaired immune reconstitution associated with GATA2 mutation. A 15-year-old man was diagnosed with AML with monosomy 7. His family history was negative for immunodeficiency and hematological disorders. He attained complete remission after HSCT from an HLA-identical sister. Post-HSCT examinations performed 15 months later revealed pancytopenia, especially monocytopenia and the absence of B and NK cells, resulting in the occurrence of donor-type MDS. Twenty-one months after HSCT, he developed central nervous system aspergillosis and finally died of the disease. Two months later (24 months after PBSCT), the donor was diagnosed with persistent EBV infection accompanied by MDS with multilineage dysplasia. Genetic analysis of GATA2 revealed a novel heterozygous mutation (c.1023_1026dupCGCC) in both siblings. GATA2 mutations were highly prevalent among adolescent MDS/AML patients with monosomy 7. Therefore, the screening of GATA2 mutations in relatives is necessary when performing HSCT from a relative donor.

PMID: 32865708 [PubMed — as supplied by publisher]

Vascular risk factors, white matter lesions and cognitive impairment in Parkinson’s disease: the PACOS longitudinal study.

Related Articles

Vascular risk factors, white matter lesions and cognitive impairment in Parkinson’s disease: the PACOS longitudinal study.

J Neurol. 2020 Aug 31;:

Abstract
BACKGROUND: Vascular risk factors (VRFs) may be associated with cognitive decline in early Parkinson’s disease (PD) but results are inconclusive. The identification of modifiable risk factors is relevant for prevention and treatment.
METHODS: Parkinson’s disease (PD) patients of the PACOS cohort who underwent a baseline and follow-up neuropsychological evaluation were enrolled in the study. PD with Mild Cognitive Impairment (MCI) and dementia (PDD) were diagnosed according to the MDS criteria. A Baseline 1.5 T brain MRI was used to calculate the white matter lesions (WMLs) burden using the Wahlund visual scale. Laboratory data, presence of hypertension, diabetes and use of anti-hypertensive drugs were collected and the Framingham Risk (FR) score was calculated. VRFs predicting PD-MCI and PDD were evaluated using Cox proportional hazard regression model.
RESULTS: Out of 139 enrolled patients, 84 (60.4%) were classified as normal cognition (NC) and 55 (39.6%) as MCI at baseline. At follow-up 28 (33.3%) PD-NC developed MCI and 4 (4.8%) PDD (follow-up time 23.5 ± 10.3 months). Out of 55 PD-MCI patients at baseline, 14 (25.4%) converted to PDD. At multivariate analysis among PD-NC a systolic blood pressure (SBP) > 140 mmHg was the stronger predictor of MCI (adjHR 4.04; 95% CI 1.41−11.3) while the presence of MCI at baseline (adj HR 7.55; 95% CI 1.76−32.3) and a severe WMLs burden (adj HR 2.80; 95% CI 0.86−9.04) were the strongest predictors of PDD, even if this latter association has a trend towards significance.
CONCLUSION: Hypertension represents the most important modifiable risk factor for PD-MCI in the PACOS cohort, increasing the risk of about four times.

PMID: 32865628 [PubMed — as supplied by publisher]